

Genexus™ Assay Development Software 6.6

USER GUIDE

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B.0	17 January 2022	Updated with minor typographical and style changes throughout. In addition, updates made to the following sections. <ul style="list-style-type: none">• The Key Variant matrix• Upload results files to another Genexus™ Software server• About assays in Genexus™ Software• System administration and management (manager/administrator)• Available versions of software update packages• Disk space usage and backup settings (administrator)• System-installed sample attributes The following sections are new for revision B. <ul style="list-style-type: none">• Linked accounts in Genexus™ Software (manager/administrator) — replaces Accounts for use with Connect and Ion Reporter™ Software (manager/administrator)• Create a Connect account• Link Genexus™ Software accounts (manager/administrator) — replaces Configure Connect accounts in Genexus™ Software (manager/administrator)
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Introduction to Genexus™ Software

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Software overview

Ion Torrent™ Genexus™ Software supports the Ion Torrent™ Genexus™ Integrated Sequencer workflow for research use purposes from sample preparation through library preparation, template preparation, and sequencing. During and after sequencing, the software generates base calls, trims reads, and determines quality values (primary analysis), then aligns reads, calls variants, and generates reports (secondary analysis). The software also supports the Ion Torrent™ Genexus™ Purification System for nucleic acid isolation, followed by sequencing on the Genexus™ Integrated Sequencer.

This documentation provides instructions for using the software to perform the following functions:

- Create and manage samples, libraries, assays, and runs in the software.
- Monitor the status of runs on the Genexus™ Purification System and the Genexus™ Integrated Sequencer, which provides all-in-one library preparation, chip loading, template preparation, and sequencing.
- View data, results, and quality control (QC) reports after a run.
- Download data files.
- Export and print audit records.
- Create and manage annotation sets, genome references, report templates, baseline files, primer sets, plugins, and reference files.
- Create and manage user accounts.

Software compatibility and requirements

The procedures in this guide are designed for use with Genexus™ Software 6.6 or later. Version-specific information is provided in the software release notes for each version of the software. An administrator-level user can view the software version in the ⚙️ **(Settings) / Software Updates** screen.

Genexus™ Software is supported on Google™ Chrome™ browser version 90 and later and is best viewed with 1440 × 900 screen resolution. Google™ Chrome™ browser is recommended for use with the software.

The operating system of the sequencer is Ubuntu™ 18.04.1 LTS.

Software anomalies

For Genexus™ Software anomalies, see the release notes for your version of the software. You can also download the release notes, as well as the *Genexus™ Assay Development Software 6.6 User Guide* (Pub. No. MAN0024923), at <http://www.thermofisher.com/ngsssoftwaresupport>.

Antivirus software

Thermo Fisher Scientific has tested Genexus™ Software with the following antivirus software products and found them compatible as antivirus solutions:

- Bitdefender GravityZone™ Business Security
- Kaspersky™ Endpoint Security for Linux

Note: Antivirus Software definition files are updated frequently, sometimes daily. Definition file updates for antivirus software can bring additional settings or updates to the system which may affect the function of Genexus™ Software.

Network and password security requirements

Network configuration and security

The network configuration and security settings (for example, firewalls, antivirus software, network passwords) of your laboratory or facility are the sole responsibility of your facility administrators and IT and security personnel. Genexus™ Software does not provide any network or security configuration files, utilities, or instructions.

If external or network drives are connected to the sequencer, it is the responsibility of your IT personnel to ensure that such drives are configured and secured correctly to prevent data corruption or loss.

Note: If a LIMS system is configured to retrieve files from the sequencer, manual FTP setup or drive mapping is required. This configuration is not provided as part of sequencer installation, and must be set up by your LIMS system integration or IT group.

Password security

Best practice is to maintain unique passwords for all accounts in use in Genexus™ Software. All passwords must be reset the first time a user signs into the software. Change passwords according to your organization's password policy.

It is the sole responsibility of your IT personnel to develop and enforce secure use of passwords.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

Installation and verification run

IMPORTANT! The Genexus™ Integrated Sequencer and the Genexus™ Software are locked until a Field Service Engineer (FSE) has installed and performed a performance qualification verification run, signed the report, and provided a basic system overview.

Notifications are sent when the PQ run is expired. Contact your Field Support Engineer (FSE) or Technical Support if you have questions about an expired PQ status.



Get started with Genexus™ Software

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About Genexus™ Software

Genexus™ Software provides menus to help you add, select, and manage samples, libraries, runs, and assays. You can also view and manage sequencing results, monitor Genexus™ Integrated Sequencer runs in progress, and manage software settings.

The screenshot shows the Genexus Assay Development software interface. At the top, there is a navigation menu with eight items: Dashboard, Samples, Runs, Results, Assays, Notifications, Settings, and Profile. Each item is numbered 1 through 8. Below the navigation menu, the main content area displays a table of samples. The table has columns for Sample Name, Tags, Created On, Application Category, Cancer Stage, and Sample Type. Three samples are listed: LibSample384, LibSample383 (selected), and LibSample382. Above the table, there are filters and action buttons like 'Filter Samples by...', 'Manage Tags', 'Create Sample', and 'Columns'. Below the table, there are action buttons for 'Clear', 'Edit', 'Copy', 'Delete', 'Export', 'Assign Tags', and 'Prepare Library Batch'.

- ① **Dashboard:** View recent run history, and current purification or sequencing run status.
- ② **Samples:** Add new samples, import samples, prepare library batches, import library batches and manage attributes.
- ③ **Runs:** Plan a run starting from a sample, a nucleic acid sample, a BAM sample, or a library. View, edit, and manage runs. Sample to result runs are for nucleic acid isolation on a Genexus™ Purification Instrument followed by sequencing on a Genexus™ Integrated Sequencer.
- ④ **Results:** View sample results, run results, and verification results.
- ⑤ **Assays:** Manage, create, and import assays. Manage assay preset parameters and panels.
- ⑥ **Notifications:** Receive alerts and messages regarding completion of runs and other processes.
- ⑦ **Settings:** Access audit records and run logs, configure network settings, manage backup settings, restore runs, manage gene lists, link to Connect user accounts and Ion Reporter™ Software accounts, check for software updates, and manage data archiving, disk space, and users. Field Service Engineers access verification templates during sequencer installation.
- ⑧ **Profile:** Access the Help system, manage and edit user profile settings, configure an SSH key (system administrator only), and sign out.

Access End User License Agreement (EULA)

A link to the End User License Agreement (EULA) is located at the bottom of the screen.

1. Scroll to the bottom of the screen.
2. Click the **EULA** link to open a PDF file of the EULA.

Contact Support

You can access Genexus™ Software technical support from the software.

1. Scroll to the bottom of the screen.
2. Click **Support** to open <https://www.thermofisher.com/contactus>, where you can request help and technical support.

User-access levels

Users at this level	Can perform these actions
Operator	<ul style="list-style-type: none"> • Add, import, and export sample files • Edit sample files • Prepare library batches • Plan, save, audit, and delete runs • Monitor runs • View results and reports • Upload sample results files to Ion Reporter™ Software • View the audit trail for samples, library batches, runs, and assays • Reanalyze runs and run plugins
Manager	<p>Operator functions, plus:</p> <ul style="list-style-type: none"> • Create, edit, and obsolete sample attributes • Create and import assays • Create presets for annotation sets, filter chains, copy number baselines, sequence variant baselines, exon tile assay baselines, and report templates • Edit and delete sample files • Add or edit Thermo Fisher accounts • Manage gene lists • Restart a stalled or failed run • Manage reference sequences and panel, hotspot, and other sequence files • Access services information

(continued)

Users at this level	Can perform these actions
Administrator	Operator and manager functions, plus: <ul style="list-style-type: none">• View, export, and print audit records• Configure network settings• View and manage software updates• Install and manage plugins• Configure backup settings and restore runs• Manage sequencer and software log files• Add and manage user accounts

System tracking

The system tracks and checks auditable user activities for samples, assays, QC metrics and information related to reagents.

If the software detects an error at any step—for example, a scanned barcode is inconsistent with the information given for the run—the software alerts the user and does not proceed with the run.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.



Reagent management

Put in place a reagent management system to inventory and track reagent use.

Proper use of a reagent management system ensures optimal use of kits, chips, and reagents.

View notifications

You can view notifications for files that you upload and import in Genexus™ Software.

1. In the menu bar, click  **(Notifications)**.
If you have any unread notifications, the number of unread notifications is shown to the upper right of the notification button.
2. View the list of notifications in the dialog box that appears.
3. To delete a notification, click  **(Remove)** in the section of the notification.

Request and sign in to a new user account

Operator-level users must request a new user account from the manager- or administrator-level user before they can sign in to Genexus™ Software.

Administrator-level users receive the request and create the user account.

Your user name and password must be unique and not shared with other users.

- To request a new account, contact your administrator-level user.
After the administrator creates a new account, the software sends an email to the new user with a user name and temporary password.
- To sign in to a new account for the first time:
 - a. Obtain your user name and temporary password that is supplied by your administrator in an email message.
 - b. Browse to the Genexus™ Software sign in page (for example, <https://<your server address>>) enter your user name and the temporary password supplied by your administrator.
 - c. Press **Enter**, or click **Sign In**.
 - d. In the End User License Agreement that appears at first sign on, read the agreement, then click **Accept**.
 - e. In the **Change Password** screen, enter your temporary password supplied by your administrator.
 - f. Type a new password, confirm the password, then click **Change**.
 - Passwords must be between 8 and 80 characters.
 - Passwords must contain only alphanumeric characters (numbers 0 to 9 and letters A to Z), no spaces or special characters.
 - Passwords must contain at least one alphabetic character (a–z, A–Z).
 - Passwords must contain at least one numeric character (0–9).
 - Passwords are case-sensitive.

Genexus™ Software opens to the **Dashboard** screen.

Sign in

Before you can sign in to Genexus™ Software, you must have a registered account. For more information, see “Request and sign in to a new user account” on page 18.


1. Go to the Genexus™ Software sign in page.
2. Click **Switch Language**, then select the preferred language from the dropdown list that appears, if desired.
3. Enter your user name and password, then press **Enter** or click **Sign In**.

Note: If a message appears that indicates that there is a problem with the security token, refresh the browser and enter the user name and password again.

The software opens to the **Dashboard** screen.

Edit a user profile

After your user profile has been established, you can view user profile information, change a password, or change email contact information.

1. In the menu bar, click  **(Profile) ▶ My Profile**.
2. In the **Profile** screen, view the profile information.

To	Do this
Change a password	<ol style="list-style-type: none"> 1. Click Change Password. 2. In the Change Password dialog box, enter the current password and a new password. Confirm the password, then click Change.
Change an email address	<ol style="list-style-type: none"> 1. Click Change Email. 2. In the Set Email dialog box, enter a new email address, and a password, then click Save.
Edit security questions and answers	<ol style="list-style-type: none"> 1. Click Edit Security questions and answers. 2. In the Edit Security Questions dialog box, select a security question from the drop-down list, then enter an answer. 3. Enter the password for the account that is associated with the profile. 4. Click Submit.

The changes are applied immediately. After the password is changed, you must sign in with the new password.

Genexus™ Software concepts

The following terms are frequently used in Genexus™ Software.

For more definitions, see “Genexus™ Software definitions” on page 376.

Term or concept	Description	Example
Sample	The information that describes the material from a single source. Each sample is defined by a set of attributes, which are categories of information that describe important aspects of each sample, such as date of sample collection, gender, or a sample type. In the software, a sample can be isolated nucleic acid or a sample can be a specimen that requires integrated nucleic acid purification before sequencing.	Data from a biopsy or blood draw.
Sample name	A unique identifier for a particular sample.	Sample1
Assay	A collection of settings and parameters for library preparation, templating, sequencing, and sequencing data analysis.	Oncomine™ Precision Assay GX
Library	A sample associated with an assay, including barcode information and sample input quantity. Libraries are associated with an assay during library batch creation.	DNA from a non-small cell lung cancer specimen that is sequenced with the Oncomine™ Precision Assay GX.
Library batch	A collection of libraries that are prepared and sequenced in the same run under the same conditions on the Genexus™ Integrated Sequencer.	<ul style="list-style-type: none"> • A library batch that contains libraries that are different pools from a single sample. • A library batch that consists of one library of sample DNA and another library of sample RNA (from the same sample).
Library name	A unique identifier for a library.	Library1
Library batch ID	A unique identifier for a library batch.	DNA_B1_Lib123456_Samples123

Workflows

Type of workflow	More information
Sample to Result	“Sample to Result workflow” on page 22
Nucleic Acid to Result Library to Result	“Nucleic Acid to Result or Library to Result workflow” on page 24
BAM to Result	“BAM to Result workflow” on page 26

Sample to Result workflow

This is a workflow for a **Sample to Result** purification and sequencing run using the Genexus™ Purification Instrument and the Genexus™ Integrated Sequencer.

Workflow for a Sample to Result purification and sequencing run using the Genexus™ Purification Instrument and the Genexus™ Integrated Sequencer

Create and manage assays (manager/administrator) (page 190)

System-installed assays that are specifically configured for each sample type are available in Genexus™ Software. You can use the system-installed assays in a run without change. To modify any assay settings, copy the system-installed assay that best represents the experiment, then edit the assay settings if needed.

Enter samples and libraries (page 27)

Enter samples in Genexus™ Software to assign sample names and provide other information such as sample collection date, gender, type, and application category.



Plan a Sample to Result run (page 65)

Runs created in Genexus™ Software contain the settings that are used in purification, library preparation, templating, sequencing, and analysis, including sample information and plate location, assays, and barcodes.

Prepare samples and load the instrument (see the *Genexus™ Purification System User Guide* MAN0018475)

Load reagents on to the instrument for purification of DNA, then start the run.



Review samples for Sample to Result runs (page 70)

You can review samples before sequencing.



Workflow for a Sample to Result purification and sequencing run using the Genexus™ Purification Instrument and the Genexus™ Integrated Sequencer

Load the sequencer and start a run (see the instrument or assay user guide)

Follow the step-by-step instructions in the sequencer touch screen to load the sample plate and consumables in the Genexus™ Integrated Sequencer.



Genexus™ Software Dashboard (page 86)

Monitor the run in Genexus™ Software in real time.

Review data and results (page 92)

Review data and results in Genexus™ Software.

You can also analyze data in Ion Reporter™ Software using an Ion Reporter™ analysis workflow.



Nucleic Acid to Result or Library to Result workflow

Workflow for a Nucleic Acid to Result or Library to Result sequencing run using the Genexus™ Integrated Sequencer

Create and manage assays (manager/administrator) (page 190)

System-installed assays that are specifically configured for each sample type are available in Genexus™ Software. You can use the system-installed assays in a run without change. To modify any assay settings, copy the system-installed assay that best represents the experiment, then edit the assay settings if needed.

Enter samples and libraries (page 27)

Enter samples in Genexus™ Software to assign sample names and provide other information such as sample collection date, gender, type, and application category.



Plan a Nucleic Acid to Result run (page 71)

OR

Plan a Library to Result run (page 75)

Runs created in Genexus™ Software contain the settings that are used in library preparation, templating, sequencing, and analysis, including sample information and plate location, assays, and barcodes.

Dilute samples and load the sample plate (see the instrument or assay user guide)

Quantify and dilute the nucleic acid samples, then load the sample plate.



Load the sequencer and start a run (see the instrument or assay user guide)

Follow the step-by-step instructions in the sequencer touch screen to load the sample plate and consumables in the Genexus™ Integrated Sequencer.



Workflow for a Nucleic Acid to Result or Library to Result sequencing run using the Genexus™ Integrated Sequencer

Genexus™ Software Dashboard (page 86)

Monitor the run in Genexus™ Software in real time.

Review data and results (page 92)

Review data and results in Genexus™ Software.

You can also analyze data in Ion Reporter™ Software using an Ion Reporter™ analysis workflow.



BAM to Result workflow

Workflow for a Bam to Result run

Create and manage assays (manager/administrator) (page 190)

System-installed assays that are specifically configured for each sample type are available in Genexus™ Software. You can use the system-installed assays in a run without change. To modify any assay settings, copy the system-installed assay that best represents the experiment, then edit the assay settings if needed.

Upload a BAM file to create a sample or samples (page 35)

Enter samples in Genexus™ Software to assign sample names and provide other information such as sample collection date, gender, type, and application category.



Plan a BAM to Result run (page 78)

Plan a BAM to Result run, then save and launch it in Genexus™ Software. Runs created in the software contain the settings that are used for the analysis of BAM samples.

Review data and results (page 92)

Review data and results in Genexus™ Software or analyze data in Ion Reporter™ Software using an Ion Reporter™ analysis workflow.



3

Enter samples and libraries

- About samples and library batches 27
- Enter and manage samples 27
- Enter and manage libraries 54

Before you plan a run in Genexus™ Software, you must first enter sample information in the software to assign sample names and provide other information.

From the **Samples** menu, you can add samples in three ways. You can enter sample information for individual samples, you can import sample information from a file to create multiple samples, or you can upload a BAM file to create samples.

About samples and library batches

In Genexus™ Software, the data and attributes that characterize genomic data are called samples. A sample can be isolated nucleic acid, a specimen that requires nucleic acid isolation, or the sequencing data created from a BAM file that contains sample reads. Before you can plan a run to sequence or analyze a sample, you must add the information that characterizes each sample in the software.

A library batch is a group of samples that are sequenced in a Library to Result run. You can create library batches from samples you have previously added or uploaded to the software. During library batch preparation, you identify the barcode adapters that were used to prepare the libraries. After you create a library batch, you can plan and start a run to sequence and analyze the samples in the library.

Enter and manage samples

There are two types of samples in Genexus™ Software.

Sample	Description
Sample	A sample of isolated nucleic acid that is ready for library preparation, sequencing, and analysis or a specimen sample that requires nucleic acid isolation on a connected integrated purification system before sequencing.
BAM sample	A sample created from a BAM file that contains sample reads. BAM samples can be uploaded and analyzed by the software. BAM samples are denoted with BAM after the sample name in the list of samples.

You can add samples to the software in three ways.

- Enter information for a single sample or multiple samples into the software.
- Create a file of information for a group of samples and import that file.
- Enter information and import a BAM or BAM files for a single sample or multiple samples.

If you enter the information for samples in the software, you can select from system-installed sample attributes that are available in the software. For more information, see “Create a new sample” on page 33 and “System-installed sample attributes” on page 368.

System-installed and custom sample attributes are also available in the example file that you can use for importing samples. See “Download a template file for sample creation” on page 38 for more information.

Samples are listed in the **Manage Samples** screen. For more information, see “Manage samples” on page 49 and “Customize the samples table” on page 28.

Customize the samples table

The samples table is organized into columns of sample attributes. You can customize the table with table preferences to determine which columns appear. When you save the table customizations, the changes you make are saved for your user profile only.

1. In the menu bar, click **Samples ▶ Manage Samples**.
2. Click **Columns**.
 - Select the checkbox next to each attribute you would like to display.
 - Deselect the checkbox next to each attribute you would like to remove from the table.
3. Scroll to the bottom of the bottom of the list.
 - Click **Save** to save your new selections. Your selections persist when you reopen the table.
 - Click **Restore default** to restore the table to the default column settings.

Find samples

You can sort, search, and filter information to refine and narrow the samples that are listed in the **Samples / Manage Samples** screen in Genexus™ Software. The sort, search, and filter tools help you organize samples and find a samples of interest.

For information to search for a sample by a unique identifier, such as the sample name, see “Search for samples” on page 29.

For information to filter samples by a common trait, such as the date the sample was created, see “Filter the list of samples by the date of sample creation” on page 31.



Search for samples

Samples are listed in the **Samples / Manage Samples** screen in Genexus™ Software. The most recently created sample is at the top of the list.

You can search for samples with keywords or text that are included in the sample names, tags, or by the application category for the sample.

IMPORTANT! Search terms must adhere to the following rules.

- An asterisk (*) is not allowed in the search field for use as a wildcard.
 - Searches are case-insensitive, that is, both upper-case and lower-cases letters are found regardless the case of search term letters.
 - Searches find every occurrence of a continuous string. For example, a search for *demo* in assay names returns a list of all assays with a name that includes *demo*. For example, demo1, demo2, and so on.
 - Spaces are removed during searches, and are therefore not recommended. For example, a search for *demo 1* returns results only for data that includes the string *demo1*.
-

1. In the menu bar, click **Samples ▶ Manage Samples**.
2. In the **Manage Samples** screen, in the column header of interest, click  **(Filter)**.
3. In the search field, enter a full or partial name or ID of at least 3 characters.
When you have entered at least 3 characters, the filter dialog box limits the list in the filter dialog box based on the entry.
4. In the filter dialog box, select from the limited list.
 - Select a checkbox next to the name or ID to select one or more items.
 - Click **Select All** to select all of the items listed in the filter dialog box.
 - Click **Clear** to close the filter dialog box.
5. Click **Filter**.
In the column header,  changes color to indicate that a filter is applied.

The filtered list of samples is shown in the **Manage Samples** screen.

Click  **Clear Filters** to remove all filters and view the full list of samples.

Sort the list of samples

You can sort the list of samples to make it easier to find a sample in Genexus™ Software.

By default, samples are listed in the **Samples / Manage Samples** screen by the date the sample was created. The most recently created sample is at the top of the list.

You can change the order that the samples are listed in the table.

1. In the menu bar, click **Samples ▶ Manage Samples**.
2. In the **Manage Samples** screen, click a column header of interest.
The list of samples is sorted by the sample attribute, in alphabetical or chronological order of the attribute, depending on the header.
3. Click the column header again to reverse the order.



Filter the list of samples by the date of sample creation


You can apply filters to narrow search results, or shorten the list of samples when you view the list of samples in Genexus™ Software.

In addition to the filter tools in the column headers, you can also filter the list of samples with a dropdown list and search by the sample name or other column headers. For more information, see “Refine the list of samples” on page 32 and “Search for samples” on page 29.

1. In the menu bar, click **Samples ▶ Manage Samples**.
2. In the **Manage Samples** screen, select one or more filters.

Option	Description
List only samples created on an exact date	<ol style="list-style-type: none"> 1. In the Created On column header, click ▼ (Filter). 2. In the first dropdown list, select Is equal to. 3. In the calendar that appears, select the date of interest. 4. Click Filter.
List samples created within a date range	<ol style="list-style-type: none"> 1. In the Created On column header, click ▼ (Filter). 2. In the first dropdown list, select Is after or equal to. 3. In the calendar that appears, select the earliest date that the samples of interest were created. 4. In the Boolean operator dropdown list, select And. 5. In the second dropdown list, select Is before or equal to. 6. In the calendar that appears, select the latest date that the samples of interest were created. 7. Click Filter. <p>Records are listed for samples created on dates that meet both conditions.</p>
List samples according to a date that meets either one condition or a second condition	<ol style="list-style-type: none"> 1. In the Created On column header, click ▼ (Filter). 2. In the first dropdown list, select Is after or equal to. 3. In the calendar that appears, select the earliest date that the samples of interest were created. 4. In the Boolean operator dropdown list, select Or. 5. In the second dropdown list, select Is before or equal to. 6. In the calendar that appears, select the latest date that the samples of interest were created. 7. Click Filter. <p>Records are listed for samples created on dates that meet at least one of the conditions.</p>

Option	Description
List only samples that were created before a date that you indicate	<ol style="list-style-type: none"> 1. In the Created On column header, click  (Filter). 2. In the first dropdown list, select Is before or equal to. 3. In the calendar that appears, select the date of interest. 4. Click Filter.
List only samples that were created after a date that you indicate	<ol style="list-style-type: none"> 1. In the Created On column header, click  (Filter). 2. In the first dropdown list, select Is after or equal to. 3. In the calendar that appears, select the date of interest. 4. Click Filter.
Remove all selections and close the filter dialog box.	In the filter dialog box, click Clear .

3. In the filter dialog box, click **Filter** to apply the filtered selections.
In the column header,  changes color to indicate that a filter is applied.

The list of samples is filtered according to the selections.

Click  **Clear Filters** to remove all filters and view the full list of samples.

Refine the list of samples

You can shorten the list of samples that are shown in Genexus™ Software.

1. In the menu bar, click **Samples ▶ Manage Samples**.
2. In the **Manage Samples** screen, in the **Filter Samples by** dropdown list, select the type of sample you want to view.

Option	Description
List only the samples that have not been prepared as a library.	Select To Be Prepared .
List samples created from BAM files.	Select BAM Samples .
List all samples.	Select All .

Only the selected type of samples are listed.

Create a new sample

You can enter information individually for each sample that you add in the software. Complete this procedure to make a sample available for use in a run, or to add the sample to a library batch. If you have many samples to add, see instead “Create multiple samples” on page 33 and “Import samples” on page 34.

1. In the menu bar, click **Samples ▶ Create Sample**.
2. In the **Samples/Create Sample** screen, enter a name for the sample, select an **Application Category**, then complete the required fields, and optional fields if needed.
 - In the **Tags** field, enter a full or partial tag name of at least 3 characters, then select one or more tags of interest to assign a tag to the sample.
 - If the tag that you want to assign to the sample does not exist in the software, a manager-or administrator-level user can enter a new tag name in the **Tags** field to create a new tag and assign it to the sample.

The fields for sample attributes change based on the **Application Category**. Attributes identified with a red asterisk (*) are required.

If you select **Other** as the **Sample Type**, you must enter a name for the custom sample type in the text box that is shown. For example, enter *Nasopharyngeal Swab*.

Sample Type *

Other ✓ ↕	Nasopharyngeal Swab ✓
-----------	-----------------------

3. Click **Save**.
The new sample is listed in the **Manage Samples** screen and is available to use in a run plan.

Create multiple samples

There are two ways you can create multiple new samples in Genexus™ Software.

- You can enter sample information for multiple samples in a table format in the software.
 - You can import a file that contains sample data. For more information, see instead “Import samples” on page 34.
1. In the menu bar, click **Samples ▶ Create Sample**.
 2. In the **Create Sample** screen, click the **Multiple (.xls, .csv, .tsv)** tab.
 3. In **Application Category**, select the application category for the samples.
A table with columns that are specific for the selected application category appears.

4. (Optional) Click **Columns** in the upper right corner of the screen to customize the optional attributes for the samples that you want to create.
 - Select the checkbox for a sample attribute to add a column to the table. Click the row to view fields into which you can enter attribute information.
 - Deselect the checkbox for a sample attribute to remove the attribute from the table.
5. Enter the information for a sample.
Attributes identified with a red asterisk (*) are required.
6. Click **Add Row**, then enter the information in the new row for each new sample.
7. Repeat step 6 for each new sample.
8. Select the checkbox in the row for each sample in the list that you want to create. To select all samples, select the checkbox in the column header row.

IMPORTANT! The information for samples that are not selected is not retained by the software. Ensure that you select the checkbox for every sample that you will create.

9. Click **Save**.
The new samples are listed in the **Manage Samples** screen and are available to use in a run plan.

Import samples

You can enter sample information for multiple samples directly in Genexus™ Software. When you want to create more than a few samples, an easy and fast way to add multiple samples in the software is to create a file of information for a group of samples and import that file.

Sample data files can be used to capture, manage, and edit sample data. You can import sample data files in TSV, XLS, XLSX, or CSV file formats. For a list of the sample attributes that are included in the import file, see “System-installed sample attributes” on page 368. For ease of use, you can download a Microsoft™ Excel™ template file to create an import file.

You must create custom attributes before importing sample and run information for the attributes to be propagated through to output files. All attributes that are included in the file that you use to import samples must be either system-installed attributes or custom attributes that exist in the software. Other file content is not transferred with the sample.

You can use a file to import sample information into the software. You can create the file, or use a file that is exported from external LIMS software. Before you import a file from LIMS software, you must first map the sample attribute names that are named differently in the LIMS file to the attribute used in Genexus™ Software. For more information, see “Map a sample attribute (manager/administrator)” on page 43.

1. In the menu bar, click **Samples ▶ Create Sample**.
2. In the **Create Sample** screen, click the **Multiple (.xls, .csv, .tsv)** tab.
3. In the **Application Category** dropdown list, select the application category for the samples.

Tip: Use the search field to search for the application category of interest.

4. Choose one of two options.

Option	Description
Download a template file, then edit it to create a new file.	For more information, see “Download a template file for sample creation” on page 38. Upload the edited file using the Browse button. Note: When you select Other as the Application Category , you must enter text in the Application Category text box, then click anywhere in the screen in order for the template file to be available.
Upload the sample data from an existing file.	1. Click Browse . 2. Navigate to the file, then click Open . The data contained in the file populates the table in the screen.

5. Edit the sample table and data, if needed.

Option	Description
Remove an attribute column	Click Columns , then deselect the column name.
Add an attribute column to the table	Click Columns , then select the column name. The column names that are listed are Application Category -specific.
Edit sample data	Click the sample row of interest, then edit the text field or dropdown list for the sample.
Add more samples to the table	Click Add Row , then enter the sample data.
Remove a sample from the table	Select the checkbox for the row, then click Remove Row .

6. Select the checkbox in the row for each sample to create. To select all samples, select the checkbox in the column header row.

IMPORTANT! The information for samples that are not selected is not retained by the software. Ensure that you select the checkbox for every sample you intend to create.

7. Click **Save**.

You can place the pointer over  (**alert**) to view more information if needed.

The new samples are listed in the **Manage Samples** screen and are available to assign to a run plan.



Upload a BAM file to create a sample or samples

You can upload BAM files to create samples in Genexus™ Software. You can also redefine existing BAM files as new samples. That is, you can upload a BAM file and create a new sample name for the BAM file and enter the same or different values for the sample attributes that are associated with the BAM file.

If the BAM file is not on the Genexus™ Integrated Sequencer, you can upload a BAM file from the computer that is used to access Genexus™ Software. You can upload the BAM file directly from the computer or from a USB drive connected to the computer.

BAM files can be analyzed in the software. For more information, see “Plan a BAM to Result run” on page 78.

1. In the menu bar, click **Samples ▶ Create Sample**.
2. In the **Create Sample** screen, click the **From Sequence Files (.bam)** tab.
3. In **Application Category**, select the application category for the samples.
4. Click **Browse**, then navigate to the file.

Option	Description
Find the file on the instrument.	Click  Server , then navigate to the file.
Find the file on the computer used to access the software.	Click  My Computer , then in the Browse dialog box that appears, navigate to the file.

5. Navigate to the file, then click **Select**.
The file is now available to select and assign to a sample. The BAM file name is visible in the dropdown lists in the **DNA BAM File**, **RNA BAM File**, and **TNA BAM File** columns.
6. Enter the information for the sample.
Attributes identified with a red asterisk (*) are required.
7. In one or more of the **DNA BAM File**, **RNA BAM File**, and **TNA BAM File** dropdown lists for the sample, select the BAM file that you want to assign to the sample.
8. To add more samples, click **Add Row**.
9. Repeat step 4 to step 8 for each extra BAM sample you want to create.
10. Select the checkbox in the row for each sample that you want to create. To select all samples, select the checkbox in the column header row.
11. Click **Save**.

The new samples are listed in the **Manage Samples** screen, are denoted with **BAM** after the sample name, and are available to assign to a BAM to Result run plan.

View sample summary

You can view a summary of the details for the attributes of a sample. The sample summary lists the information for sample attributes for the sample and the date the sample was created in the software. For a list of the information that is shown in the sample summary, see “Sample summary” on page 37.

For more information about sample attributes, see “System-installed sample attributes” on page 368.

To view the sample summary:

1. In the menu bar, click **Samples ▶ Manage Samples**.
2. In the **Sample Name** column, click the sample name of interest.
Details for the sample are shown in the sample summary pane.
3. When you are finished viewing the sample details, click another sample name to view details for another sample, or click **✕ (Close)** to return to the **Manage Samples** screen.

If needed, you can click **⋮ (More Options)** to perform more actions, such as copy, edit, assign tags, add notes, or audit the sample. The options that are available depend on whether the sample is assigned to a run plan. The actions for each sample are also available in the **Manage Samples** screen when you place the pointer over the row of a sample.

Sample summary

You can view details for its attributes when you select a single sample that is listed in the **Samples/Manage Samples** screen in Genexus™ Software. The sample summary lists details that describe sample attributes, such as gender and the date that the sample was created in the software.

Notes for a sample are not shown in the sample summary pane. For more information, see “View notes or add a note to a sample” on page 51.

The sample attributes that are shown in the sample summary pane depend on the application category for the sample. The following table lists the system-installed sample attributes for a sample that has an application category of cancer. Custom sample attributes are also shown in the sample summary pane, but are not listed in the following table.

Sample attribute	Description
Sample Name	A unique identifier representing the sample.
Collection Date	The date that the sample was collected.
Created On	The date the sample was imported or created in the software.
Application Category	The application type of the sample.
Cancer Type	The type of cancer that is represented by the sample.
Sample Type	A term that describes the sample, for example, RNA.
Gender	The biological sex of the sample: Female , Male , or Unknown .
%Cellularity	The percentage of tumor cells over normal cells in the sample. The %Cellularity attribute is applicable to FFPE samples only.

(continued)

Sample attribute	Description
%Necrosis	The percentage of cellular necrosis in the sample.
Cancer Stage	The stage of the cancer from which the sample was collected. Stage 0–IV, or Primary, Unknown, or Other.
BAM File Path ^[1]	The path and file name for the BAM files associated with the sample. Files are listed for samples created from BAM files only.
Metadata ^[1]	The data for the BAM sample is shown. Metadata can include the device ID and the type of instrument that generated the BAM file. Other information, such as the assay, analysis pipeline version, chip type, chip ID, sequencing kit, library kit, templating kit, and name of the run results from the BAM file source instrument are shown.

^[1] This information is only shown for BAM samples.

Download a template file for sample creation

You can download a template file, add sample information to it, then use the file to import the sample data for multiple samples.

Template files contain two tabs. The **Instruction** tab in the spreadsheet lists and indicates required and optional attributes, which are the column headers in the **Sample Details** tab. Use the **Sample Details** tab to enter sample information.

Template files contain both the system-installed and the custom sample attributes as column headers.

1. In the menu bar, click **Samples ▶ Create Sample**.
2. In the **Create Sample** screen, click the **Multiple (.xls, .csv, .tsv)** tab.
3. In **Application Category**, select the application category for the samples.
Template files are specific for each application category.
4. Click **Download Template** to download the Microsoft™ Excel™ template file.
The template file contains default sample attributes as columns. If custom sample attributes have been configured in the software, the custom attributes are added to the template file.
5. Save the file to the computer, then open the file, enter sample data in the **Sample Details** tab.
6. When you are finished adding sample information to the file, save the file.

You can now import the file to Genexus™ Software. For more information, see “Import samples” on page 34.

Sample attributes (manager/administrator)

Samples contain attributes that are relevant to genetic research, such as information about gender, tumor, or a sample type such as RNA or DNA. In Genexus™ Software, manager- and administrator-level users can add custom attributes and add or change values of those attributes. For more information, see “Create a new sample” on page 33, “Create multiple samples” on page 33, “Import samples” on page 34, and “Edit a sample” on page 51.

Custom and predefined sample attributes can be viewed in the Samples table. Each column in the Samples table represents a single sample attribute.

For more information, see:

- “System-installed sample attributes” on page 368
- “Create a custom sample attribute (manager/administrator)” on page 39

Create a custom sample attribute (manager/administrator)

A manager- or administrator-level user can create custom sample attributes. Sample attributes, such as the date of collection and **Application Category**, characterize and define a nucleic acid sample. A sample attribute can be required, in which case you are required to enter the attribute information for each new sample. Custom attributes can be used to further characterize samples.

Sample attributes can be assigned to an application category, such as cancer or infectious disease, because some attributes are not applicable to all application categories. The default application category for new custom attributes is **ALL**, which means that the custom attribute is available for all samples. When you restrict custom attributes to one or more application types, the field for the custom attribute is only visible for samples for the application types that you designate.

Custom samples attributes cannot be edited or obsoleted after the samples that use the attributes are added to a library. To remove a custom sample attribute from use, see “Obsolete a sample attribute (manager/administrator)” on page 42.

Genexus™ Software supports following types of sample data:

- **Text**—Accepted values: general text.
- **Date**—Accepted values: provides a calendar tool to select a date.
- **Integer**—Accepted values: integer numbers. Examples: 0, 9, -44.
- **Decimal**—Accepted values: decimal numbers Examples: 0.1, 9.0, -44.3.
- **List**—Creates a list of these choices that you define with the preset terms.
- **Boolean**—Creates a list of these two choices: True, False.

1. In the menu bar, click **Samples ▶ Manage Attributes**.
2. In the **Manage Attributes** screen, click **+ Add New Attribute**.
3. In the **Add New Attribute** dialog box, in **Attribute Name**, enter a name for the attribute.
Attribute names are limited to ≤50 alphanumeric characters (0–9 and Aa–Zz), period (.), underscore (_), or hyphen (-).


4. In **Application Category**, select an applicable application category or application categories to change the application category from **All**.

The default setting **All** makes the attribute available for all samples when you create new samples and when you edit existing samples. You can refine the availability of the attribute to selected application categories when you select an application category.

5. In **Data Type**, specify the data type to create.
6. Click the toggle switch to designate the attribute as **Required** to require users to select the new attribute when adding or importing samples, if needed.
7. Click the toggle switch to designate the attribute as **Unique**, if needed.

When enabled, the **Unique** setting allows you to require a unique attribute value for samples. For example, you can create a unique attribute called **Sample number**. Then, when each new sample is created, you enter a unique number for each sample in the custom **Sample number** attribute field.

8. For **List** and **Boolean** data types, complete the following options.

Data type	Steps
List	<ol style="list-style-type: none"> 1. In Vocabulary List, enter a list of values separated by commas, then click Add. The first option that you enter is designated as the default option. 2. Change the default option, if needed. Deselect Use default if you want the new attribute to be shown without a default preset. Select a different option to change the value that is shown as the default option. Click  (Delete) to remove an attribute list value.
Boolean	Select True or False to designate one option as the default value for the attribute.

9. Click **Save**.

The new sample attribute is listed in the **Attribute Name** column and is available when you add a new sample.

You can now add information for the custom sample attribute to samples when you create new samples. In addition, you can enter information for the attribute to existing samples that were created before the custom attribute was created. For more information, see “Enter and manage samples” on page 27 and “Edit a sample” on page 51.

Example custom attribute

When you create a custom attribute, you can add metadata pertinent to that attribute when you create or edit samples in the software. Attributes that are created with the **List** data type allow you to restrict the information to a list of terms.

This example shows the creation of the options **High** and **Low** for a custom attribute of **Grade** for **Cancer** samples. **Low** is set as the default value in this example. Consider the convenience and consequences of assigning a default value to any custom attribute. Unless the default value is changed during sample creation, all samples will be associated with the default value.

Add New Attribute

✕

Attribute Name *

Data Type *

Application Category *
 Cancer
 Cancer (Germline)

Vocabulary List

Use default

High

Low

After the new attribute has been saved, it is available for sample creation in the software. When you create a new cancer sample in this example, **Low** appears as the default **Grade** value. You can select **High** from the **Grade** dropdown list to change the value.

Sample Name *

Sample Type *

Cancer Stage *

% Cellularity

Collection Date *

Phenotype

Tumor Margin

Histology Type

Notes

Application Category *

Cancer Type *

% Necrosis

Gender * Male Female Unknown

Cell Line Sample True False

Race

Tumor Size in mm

Grade


High

Edit a custom sample attribute (manager/administrator)

A manager- or administrator-level user can edit custom sample attributes. Some attribute values, such as the **Attribute name**, **Data Type**, and whether or not the attribute is required, cannot be edited.

Custom attributes created in Genexus™ Software version 6.2 are set to **All** application categories. If desired, you can edit the custom attributes to associate the application categories with the applicable applications.

1. In the menu bar, click **Samples ▶ Manage Attributes**.
2. In the **Manage Attributes** table, place the pointer over the row of a custom sample attribute that you want to change, then click **Edit**.
3. In the **Edit Attribute** dialog box:

Option	Description
Add an application category or categories to the sample attribute	In Application Category , select the checkbox for the application category.
Remove an application category or categories from the sample attribute	In Application Category , deselect the checkbox for the application category.
Add a new option for List type of sample attributes only	<ol style="list-style-type: none"> 1. In the Vocabulary List text box, enter the new option, then click Add. 2. Change the default option, if needed. Deselect Use default if you want the attribute to be shown without a default preset. Select a different option to change the value that is shown as the default option. Click  (Delete) to remove an attribute list value.

4. Click **Save**.

Obsolete a sample attribute (manager/administrator)

Manager- and administrator-level users can obsolete custom sample attributes from use in the software. You cannot designate as obsolete system-installed sample attributes.

When you obsolete an attribute, no change is made to the samples that already have an assigned value for the attribute, but the attribute is no longer available when you create new samples.

1. In the menu bar, click **Samples ▶ Manage Attributes**.
The **Manage Attributes** screen opens to the **Manage Attributes** tab.
2. Place the pointer over the row of the custom sample attribute that you want to make obsolete, then click **Obsolete**.
3. In the confirmation dialog box that appears, click **Yes** to confirm that you want to obsolete the sample attribute.

The sample attribute that has been made obsolete is listed in the **Manage Attributes** screen, but is no longer available when you create a sample.

Custom sample attributes that have been made obsolete can be reactivated. For more information, see “Reactivate a sample attribute (manager/administrator)” on page 43.

Reactivate a sample attribute (manager/administrator)

Manager- and administrator-level users can reactivate custom sample attributes that have been made obsolete.

1. In the menu bar, click **Samples ▶ Manage Attributes**.
The **Manage Attributes** screen opens to the **Manage Attributes** tab.
2. In the **Manage Attributes** table, place the pointer over the row of a custom sample attribute that you want to reactivate, then click **Reactivate**.
3. In the confirmation dialog box that appears, click **Yes** to confirm that you want to reactivate the sample attribute.


The sample attribute is available when you create a sample relevant to the attribute.

Map a sample attribute (manager/administrator)

You can import a file that you have exported from external LIMS software to add samples in Genexus™ Software. Each column header in the file that you want to import to create samples in the software is a name for a sample attribute. Before you import samples with the file, a manager- or administrator-level user must map the names of sample attributes that are used by the LIMS software to the names of the sample attributes recognized by Genexus™ Software.

For example, in Genexus™ Software, the sample attribute for the name of a sample is **Sample Name**. In the file that you want to import, however, the same attribute can be called *Name*. You can map the sample attribute *Name* to **Sample Name** so that the software recognizes *Name* as the name of a sample.

If the attribute values in the file contain vocabulary that is not recognized in the software, you can map the associated vocabulary so that you can seamlessly import the file to create samples. For more information, see “Map the name of a value for a sample attribute (manager/administrator)” on page 44.

1. In the menu bar, click **Samples ▶ Manage Attributes**.
2. In the **Manage Attributes** screen, click the **Sample Attribute Mapping** tab.
If any attributes have been mapped, a list of sample attributes and any associated attribute names for all application categories is shown. Attributes identified with a red asterisk (*) are the attributes that are required when you create a sample.
3. Click  **Edit Mapping**.
The names of the sample attribute are case-sensitive.

Option	Description
Populate attribute name options from a file, then select the attribute names to add the names in the software.	<ol style="list-style-type: none"> 1. Click Click here, then find and upload the file that you want to import. The attribute names are listed in the right pane. 2. In the Associated Attribute Name field for the attribute of interest, select the attribute name from the dropdown list that appears. For sample attributes that include a date, select the proper date format for the LIMS software from the dropdown list. 3. Select the checkbox of an associated attribute name to map the name to an Attribute Name.
Enter an associated attribute name to add the name in the software.	<ol style="list-style-type: none"> 1. In the Associated Attribute Name field for the attribute of interest, enter the attribute name used in the file that you want to import. For sample attributes that include a date, select the proper date format for the LIMS software from the dropdown list. 2. Select the checkbox of an associated attribute name to map the name to an Attribute Name.
Remove an associated attribute name.	In the Associated Attribute Name field for the attribute of interest, in the tile for the name that you want to remove, click X (Remove) .

4. Click **Save**.


The external attribute names are mapped to the attribute names used in Genexus™ Software and are listed as associated attribute names in the **Manage Attribute Mapping** list.

You can then import the LIMS software file to create samples with the sample data. For more information, see “Import samples” on page 34.

Map the name of a value for a sample attribute (manager/administrator)

You can import a file that you have exported from external LIMS software to add samples in Genexus™ Software. Each column cell in the file to create samples in the software can contain alternate vocabulary for a sample attribute that are not recognized in the software. Before you import samples with the file, a manager- or administrator-level user must map the vocabulary for the sample attributes that is used by the LIMS software to the names of the sample attributes recognized by Genexus™ Software.

For example, in Genexus™ Software, one of the sample attribute values for **Sample Type** is **Blood (Whole)**. In the file that you want to import, however, the same attribute value can be called *whole blood*. You can map the sample attribute value, *whole blood*, to **Blood (Whole)** so that the software recognizes *whole blood* as **Blood (Whole)**.

1. In the menu bar, click **Samples ▶ Manage Attributes**.
2. In the **Manage Attributes** screen, click the **Sample Attribute Mapping** tab.
If any attributes have been mapped, a list of sample attributes and any associated attribute names for all application categories is shown. Attributes identified with a red asterisk (*) are the attributes that are required when you create a sample.
3. Click **Manage Vocabulary Mapping**.
By default, sample attributes that are a list type, such as **Sample Type** and **Cancer Stage**, and have vocabulary values defined in the software, are shown in the pane on the left. Any sample attributes that have associated vocabulary mapped to the attribute are also listed.
 - Click an attribute name in the left pane to view the vocabulary value and associated vocabulary value in the pane on the right.
4. Click  **Edit Mapping**.
5. In the **Associated Vocabulary Value** field for the attribute value of interest, enter the term of the attribute value used in the file that you want to import, then click **Save**.
 - To remove a term that you have added, in the **Associated Vocabulary Value** field for the attribute of interest, delete the text.
6. Click **Save**.


The name for the external attribute value is mapped to the attribute vocabulary value recognized in Genexus™ Software and is listed as an associated vocabulary value in the **Manage Vocabulary Mapping** list.

You can then import the LIMS software file to create samples with the sample data. For more information, see “Import samples” on page 34.



Review sample attribute mapping history (manager/administrator)

A manager- or administrator-level user can review and export the history of sample attribute mapping in Genexus™ Software.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

1. In the menu bar, click **Samples ▶ Manage Attributes**.
2. In the **Manage Attributes** screen, click the **Sample Attribute Mapping** tab.
A list a sample attributes and any associated attribute names appears.
3. Click  **Audit**.

4. In the **Audit Details** screen:

Option	Description
Review the audit trail details.	Each action that was performed for the sample attribute mapping, such as creation of mapped attributes, or edits, is listed.
Export the audit record.	Click  Download to generate a PDF file. Depending on the browser settings, the software automatically downloads the file or prompts you to open or save the file.
Close the Audit Details screen.	Click  Back to return to the list of sample attributes.

Sample tags

A sample tag is a label that you add to a sample to help you group and track samples in Genexus™ Software. For example, when you have a project or a study that involves a group of samples, you can assign a tag to each sample in the study or project so that you can easily track and find that group of samples in the software. A manager-or administrator-level user firsts creates a tag, then any user can assign that tag to a single sample or a group of samples. You can assign multiple tags to each sample.

You can assign a tag when you create a sample or you can add, edit, or remove the tags that are assigned to a sample later. You can apply sample tags to multiple samples at one time. Later, you can filter the table of samples with that tag to list only the samples that have the tag. The tags that you assign to samples are also shown in the list of sample results so sample tags can be used to group and find sample results that are part of a study or project.

If you edit a sample and make a change to the tags associated with the sample, the changes you make are tracked in the software and can be reviewed. For more information, see “Edit a sample” on page 51 and “Review sample edit history” on page 53.

Tags are preserved with the sample when you archive a run that includes a sample with one or more assigned tags.

You can also add tags to sample results. For more information, see “Assign sample tags to a sample result” on page 100 and “Assign sample tags to multiple sample results” on page 100.

Create a sample tag (manager/administrator)

A manager-or administrator-level user can create sample tags in Genexus™ Software that can be used to label and group samples.

You can create parent tags to nest the labels if desired. For example, if a large project is divided into sections, you can create a parent tag called **Master project** and a child tag called **New variant of concern**. You can create multiple layers of nested tags to help sort projects and sections within projects. When you assign nested tags to a sample, the parent tag and all child tags are listed as tags

for the sample. Later, when you search or filter for nested tags, the parent tags are also shown to help you group and track the samples.

1. In the menu bar, click **Samples ▶ Manage Samples**.
Alternatively, you can create a tag when you create a sample. For more information, see “Create a new sample” on page 33 and “Import samples” on page 34.
2. In the **Manage Samples** screen, click **Manage Tags**.
The **Manage Tags** screen opens and lists tags, if any, that have been created in the software.
3. Click **+ Create Tag**.
4. In the **Create Tag** dialog box, enter a name for the tag.
 - Select a **Parent Tag** from the list to create a hierarchical tag that is nested below the parent tag, if desired.
5. Click **Save**.

The tag is available to assign to samples.

Assign sample tags

You can assign tags to samples in Genexus™ Software to label and group samples.

1. In the menu bar, click **Samples ▶ Manage Sample**.
2. In the **Manage Samples** screen, place the pointer over the row of the sample that you want to tag, then click **Assign Tags**.
3. In the **Assign Tags** dialog box, search for or scroll to a tag.
If the tag that you want to assign to a sample does not exist in the software, a manager-or administrator-level user can click **+ Create Tag** to create a new sample tag.
4. Select one or more tags that you want to assign to the sample, then click **Save**.

The tag is assigned to the sample and is listed in the row for the sample in the samples table.

Assign a sample tag to multiple samples

You can assign tags to multiple samples at once in Genexus™ Software.

1. In the menu bar, click **Samples ▶ Manage Sample**.
2. In the **Manage Samples** screen, click the checkbox to the left of each sample of interest, then click **Assign Tags** above the list of samples.
Assign Tags is not shown until you select at least one sample.

3. In the **Assign Tags** dialog box, search for or scroll to a tag.
If the tag that you want to assign to a sample does not exist in the software, a manager-or administrator-level user can click **+ Create Tag** to create a new sample tag.
4. Select one or more tags that you want to assign to the samples, then click **Save**.

The tag is assigned to the selected samples and is listed in the row for each sample in the samples table.



Change or remove the tags that are assigned to a sample

You can change or remove the tags that are assigned to a sample in Genexus™ Software.

1. In the menu bar, click **Samples ▶ Manage Sample**.
2. In the **Manage Samples** screen, place the pointer over the row of the sample of interest, then click **Assign Tags**.
3. In the **Assign Tags** dialog box:
 - Deselect the checkbox next to each tag that you want to remove from the sample.
 - Select the checkbox next to each tag that you want to add to the sample.
4. Click **Save**.

Edit a sample tag (manager/administrator)


A manager-or administrator-level user can rename a tag and change the parent tag associated with a tag in Genexus™ Software to change the hierarchy of the tags. When a tag is edited, the change is applied to all samples assigned with the tag.

1. In the menu bar, click **Samples ▶ Manage Sample**.
2. In the **Manage Samples** screen, click  **Manage Tags**.
The **Manage Tags** screen opens and lists tags, if any, that have been created in the software.
3. In the **Manage Tags** dialog box, click  **(Edit)**.
4. Change the name of the tag and parent tag assignment if needed, then click **Save**.
5. In the confirmation dialog box that appears, click **Yes** to confirm that you want to edit the sample tag.

All samples that have the tag that you edit are updated with the change. If the name of the tag is changed, the new name is shown for all samples and sample results with the tag. If the hierarchy of the tag is changed, that change is also applied to all samples and sample results with the tag.

Delete a sample tag (manager/administrator)

A manager-or administrator-level user can delete sample tags in Genexus™ Software. When a tag is deleted, the tag is also removed from any samples assigned with the tag. Additionally, when you delete a parent tag, all tags that are nested under the parent tag are also deleted.

1. In the menu bar, click **Samples ▶ Manage Sample**.
2. In the **Manage Samples** screen, click **Manage Tags**.
The **Manage Tags** screen opens and lists tags, if any, that have been created in the software.
3. In the **Manage Tags** dialog box, click  **Delete**.
4. In the **Confirm** dialog box, click **Yes** to confirm the changes.

The tag is no longer listed in the **Manage Tags** list and is removed from all samples and sample results.

Manage samples

You can find tools for creating, searching, sorting, editing, deleting, and exporting samples, and for viewing the sample history in the **Samples / Manage Samples** screen.

Copy a sample


You can copy a sample and change the information for the sample, if needed, to create a new sample in Genexus™ Software.

1. In the menu bar, click **Samples ▶ Manage Samples**.
2. In the **Manage Samples** screen, place the pointer over the row of a sample that you want to copy, then click **Copy**.
The **Copy Sample** screen opens. The sample information for the source sample is shown. The **Sample Name** for the new sample is the sample name of the sample that was copied, appended with **_Copy**.
3. In the **Copy Sample** screen, edit the sample information and sample tags, and change the sample name, if needed.
4. Click **Save**.

The new sample is listed in the **Manage Samples** table and is available to assign to a run plan.

Copy multiple samples


You can copy multiple samples at once in Genexus™ Software. You can only copy multiple samples that have not been assigned to a run plan. To copy samples that have been assigned to a run plan, you must copy one sample at a time. For more information, see “Copy a sample” on page 49.

1. In the menu bar, click **Samples ▶ Manage Samples**.
2. In the **Manage Samples** screen, select two or more samples.
The actions that you can apply to multiple samples appear.
3. Click  **Copy**.
The **Copy Sample** screen opens. The sample information for each source sample is shown. The **Sample Name** for each new sample is the sample name of the sample that was copied, appended with **_Copy**.
4. In the **Copy Sample** screen, edit the sample information and sample tags, and change the sample names, if needed.
5. Click **Save**.

The new samples are listed in the **Manage Samples** table and are available to assign to a run plan.

Export samples

The **Export** function generates an XLS file that contains details about the selected sample.

1. In the menu bar, click **Samples ▶ Manage Samples**.
2. In the **Manage Samples** screen, select the checkbox in the row of each sample that you want to export. To select all samples, select the checkbox in the column header row.
3. Click  **Export**.
An XLS file is created that contains the details of the selected samples. Depending on your browser settings, the software automatically downloads the file or prompts you to open or save the file.
4. Open the XLS file in an appropriate viewer to review or print.

View notes or add a note to a sample


You can view existing notes or add new notes to a sample in Genexus™ Software. Use notes to capture time-stamped written comments and observations for a sample. A sample can contain multiple notes from different users.

1. In the menu bar, click **Samples ▶ Manage Samples**.
2. In the **Manage Samples** screen, place the pointer over the row of the sample of interest, then click **Notes**.

Action	Description
View existing notes for a sample.	In the Add Notes dialog box, notes that are associated with the sample are listed by user, date, and time.
Add a new note to a sample.	<ol style="list-style-type: none"> 1. In the Add Notes dialog box, click + Add Note. 2. Enter the note, then click Save. <p>The note is added to the sample and includes the username for the account of the person who entered the note, and a timestamp that records the note entry date and time.</p>

Delete a sample note

You can delete notes that have been added to a sample in Genexus™ Software.

1. In the menu bar, click **Samples ▶ Manage Samples**.
2. In the **Manage Samples** screen, place the pointer over the row of the sample of interest, then click **Notes**.
3. In the **Add Notes** dialog box, in the row of the note that you want to delete, click  **(Delete)**.

The note is no longer listed in the **Add Notes** dialog box and the note is removed from the software.

Click **Close** to return to the **Manage Samples** screen.

Edit a sample

You can edit the information that describes a sample in Genexus™ Software. When you edit a sample, you can change the application category, update sample data, and remove data that are not required.

For samples created from BAM files, you can add or remove the associated BAM data files.

You can edit a sample until a run that uses that sample is started. You cannot edit a sample during a run. After a run is complete, a manager- or administrator-level user can edit a sample for 30 days. However, when a manager- or administrator-level user edits a sample during the 30-day period after the run is complete, the sample status for the result is changed to obsolete and the report and other files associated with the sample are no longer available.

If a manager- or administrator-level user edits a sample within 30 days after the run and the sample result is obsolete, you must reanalyze the sample to generate sample results and a new variant report for the sample. For more information, see “Reanalysis” on page 165.

To add a custom sample attribute to a sample, see “Create a custom sample attribute (manager/administrator)” on page 39.

1. In the menu bar, click **Samples ▶ Manage Samples**.
2. In the **Manage Samples** screen, place the pointer over the row of a sample that you want to edit, then click **Edit**.
3. In the **Edit Sample** screen, modify the sample information.
See “System-installed sample attributes” on page 368 for more information about system-installed sample attributes.
4. For samples created from BAM files, change the associated BAM file if desired.
 - a. Click the dropdown list in **DNA BAM File**, **RNA BAM File**, or **TNA BAM file**.
 - b. Click **Browse**, then click **Server** or **My Computer**.
 - c. Navigate to the file, then click **Select**.
5. Click **Save**.


Edit multiple samples

You can edit multiple samples at once in Genexus™ Software.

You can edit samples until a run that uses that sample is started. You cannot edit samples during a run. After a run is complete, a manager- or administrator-level user can edit samples for 30 days. However, when a manager- or administrator-level user edits samples during the 30-day period after the run is complete, the status for the sample results is changed to obsolete and the report and other files associated with the samples are no longer available.

If a manager- or administrator-level user edits samples within 30 days after the run and the sample results are obsolete, you must reanalyze the samples to generate sample results and a new variant report for the samples. For more information, see “Reanalysis” on page 165.

Only samples that have the same sample application category can be edited together.

1. In the menu bar, click **Samples ▶ Manage Samples**.
2. In the **Manage Samples** screen, select two or more samples.
The options that you can apply to multiple samples appear.
3. Click  **Edit**.
4. In the **Edit Sample** screen, modify the sample information.
For more information about edit options, see “Create multiple samples” on page 33.
5. Click **Save**.

The changes to the samples are saved and the edited samples are available to assign to run plans.

You can view a history of changes that have been made to each sample. For more information, see “Review sample edit history” on page 53.



Review sample edit history

The entire history of defining a sample is available to review and export in Genexus™ Software. The history shows the original sample values and any new values or activities. The following actions generate an audit record:

- Create a new sample
- Edit a sample, including changes to sample tags
- Delete a sample


This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

1. In the menu bar, click **Samples ▶ Manage Samples**.
2. In the **Manage Samples** screen, place the pointer over the row of a sample of interest, then click **Audit**.
3. In the **Audit Details** screen:

Option	Description
Review the audit trail details.	Each action that was performed for the sample, such as sample creation or edits, is listed.
Export the audit record.	Click  Download to generate a PDF file. Depending on the browser settings, the software automatically downloads the file or prompts you to open or save the file.
Close the Audit Details screen.	Click  Back to return to the list of samples.

Delete samples

You can delete samples that have not been assigned to a run. Samples that are assigned to a run or library batch cannot be deleted.

1. In the menu bar, click **Samples ▶ Manage Samples**.
2. In the **Manage Samples** screen, select the checkbox in the row of each sample that you want to delete.
The options that you can apply to multiple samples appear.
3. Click  **Delete**.
The **Delete Sample** dialog box opens with the message *Are you sure that you want to delete selected sample(s)?*
4. Click **Yes** to delete the selected samples.

The samples are removed from the software.

Enter and manage libraries

You can prepare a library batch manually or import library batch information from a file. To change sample or library batch information, you can edit the information in the samples, update library batches, or delete samples or library batches.

Prepare or import a library batch

A library batch is a group of prepared libraries that are sequenced in the same Library to Result run. If you are planning a run starting from libraries that you have already prepared manually, you must first create a library batch in Genexus™ Software from samples. You can enter samples in the software or import a sample file. For more information see, “Enter and manage samples” on page 27.

Select the library batch when you plan the run. If you are planning a run starting from nucleic acid samples, skip this step and proceed to “Plan a Nucleic Acid to Result run” on page 71.

Note:

- Each library in a library batch must have a unique library name.
 - Fields identified with a red asterisk (*) are required.
-

No-template controls

You can add a no-template control (NTC) when you assign samples in run planning, or when you create a library batch in Genexus™ Software. No-template controls do not contain nucleic acid but are processed in a sequencing run to monitor contamination or primer-dimer formations that could produce false-positive results. When you plan a run that contains an NTC, data generated from the NTC are shown in the QC results. For more information, see “QC results” on page 119.

Prepare a library batch

A library batch is a group of prepared libraries that are sequenced in the same library run. If you are planning a run starting from libraries that you have already prepared manually, you must first create a library batch in Genexus™ Software from samples that you have added. If you are planning a run starting from nucleic acid samples, skip this step and proceed to “Plan a Nucleic Acid to Result run” on page 71.

1. In the menu bar, click **Samples ▶ Manage Samples**.
2. In the **Manage Samples** screen, in the **Filter Samples by** dropdown menu, apply the **To Be Prepared** filter to limit the displayed samples to those samples that have not been placed in a library batch.
For more information about how to filter the list of samples, see “Find samples” on page 28.
3. Select samples in the list by clicking the checkbox to the left of each sample, then click **+ Prepare Library Batch**.

4. In the **Create Library batch** screen, select the assay that you want to use in the run plan from the **Select Assay** dropdown menu.

The assay determines parameter selections for the run, including any required controls and post-run data analysis settings. If you select an assay that includes one library prep type, the library preparation type for the assay listed in the **Library Prep Type**: automatically fills for the nucleic acid type specified by the assay you selected: **DNA, RNA, DNA+RNA, or TNA**.

- a. For assays that include more than one library prep type, select the library prep type, such as **DNA, RNA, DNA+RNA, or TNA** for the library batch.
5. In the expanded screen, in **Library Batch ID**, enter a unique identifier for the library batch.

Note: Library Batch IDs can contain only alphanumeric characters (0–9, Aa–Zz), period (.), underscore (_), and hyphen (-). Required fields are indicated with a red asterisk (*).

6. (Optional) Enter the **Library Kit Barcode** and the **Panel Kit Barcode**.
7. Select the **Include NTC** checkbox to add no template control sample processing and reporting to the library batch.
8. Enter a unique library name for each DNA and/or RNA library in the appropriate field.
 - Library names can contain only alphanumeric characters (0–9, Aa–Zz), period (.), underscore (_), and hyphen (-).
 - If your assay requires specific controls, they are automatically listed in the dialog box. These controls each require a unique barcode ID within the library batch, but do not require library names.
9. Select the barcode ID of the adapter used to prepare each library. If appropriate, swap the default barcodes in the dialog box between DNA, RNA, and Fusions by clicking the **Swap Barcodes** swap image.

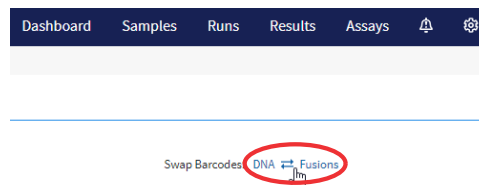
For example, click [DNA](#) ⇄ [Fusions](#) to swap barcodes.

Each library in a library batch must have a different barcode ID. When preparing the physical libraries, best practice is to swap barcodes between DNA and RNA libraries in consecutive sequencing runs to prevent carryover contamination. The barcodes that are listed in the **DNA Barcode** or **RNA Barcode** dropdown list belong to the barcode set that was selected when the assay was created.

IMPORTANT! Ensure that the barcodes that you used to create the libraries match the barcodes that you enter in the **Create Library batch** screen.


10. Enter the **DNA, RNA, DNA+RNA, or TNA Input Quantity** for each library.
11. In the **DNA, RNA, or TNA Kit Barcodes** columns, enter the extraction kit barcode for each sample and no template control, if applicable.
12. Click **Submit** to save and submit your selections.

The **Samples / Manage Libraries** screen opens, listing the library batch that you created. Libraries that are prepared in the same batch have the same **Library Batch ID**.



Import a library batch

You can import library batch information in the form of an .XLS or .XLSX file. The import file must include all required library and kit information.

1. In the menu bar, click **Samples ▶ Manage Libraries**.
2. In the **Manage Libraries** screen, click  **Import Library Batch**.
3. In the **Import Library Batch** dialog box, click **Click here** to select an assay for which the libraries are prepared, and to download an example file for import.
If the assay includes more than one library preparation type, a library preparation type dropdown list appears.
 - a. Select the library preparation type, such as **DNA, RNA, DNA+RNA, or TNA** for the library batch.
4. Select an assay from the list, then click **Download**.
The assay name is auto-populated in the Microsoft™ Excel™ template file that downloads to your drive.
5. In the template file, enter or confirm the library batch information.

Template item	Description
Reagents tab	
Assay Name	Auto-populated when assay is selected in step 4 (required)
Extraction Method Type	Auto-populated when assay and library prep type, if applicable, is selected in step 4 (required)
Library Batch ID	Must be alphanumeric (0–9, Aa–Zz), period (.), underscore (_), and hyphen (-) (required)
Library Kit Barcode	For example, Genexus™ Library Strips 1 and 2-AS barcode (optional)
Panel Kit Barcode	For example, OncoPrint™ Comprehensive Assay v3 DNA GX barcode (optional)
Libraries tab	
Sample Name	Must be alphanumeric (0–9, Aa–Zz), period (.), underscore (_), and hyphen (-) (required)
Library Name	Must be alphanumeric (0–9, Aa–Zz), period (.), underscore (_), and hyphen (-) (required)
Barcode	Barcodes used for each sample and control library preparation (required)
Nucleic Acid Type	DNA, RNA, or TNA (required)
Input Quantity	Library input quantity (optional)

(continued)

Template item	Description
No Template Control	To include a no template control, add a row with Sample Name as NTC , Library Name as NA , and Barcode and Nucleic Acid Type similar to sample rows (optional)
Control Kit Barcode	The barcode for the kit used for nucleic acid extraction of no template controls (optional)
Extraction Kit Barcode	The barcode for the kit used for nucleic acid extraction of a sample (optional)

IMPORTANT! For DNA and Fusions assays, the DNA library and RNA libraries must be listed in sequential order per pool for each sample. For example, for a 1-pool DNA and Fusions assay, order should be DNA, RNA for sample 1, DNA, RNA for sample 2. For a 2-pool DNA and Fusions assay, library order should be DNA, RNA (pool 1), DNA, RNA (pool 2) for sample 1, then DNA, RNA (pool 1), DNA, RNA (pool 2) for sample 2.

6. Save the file.
7. In the **Import Library Batch** dialog box, click **Select Library batch file**, navigate to the saved file, then select it.
8. Click **Upload**.
A progress bar followed by an import report displays. If the import process fails, an error message indicates the reason for failure (for example, an invalid character was used). For additional troubleshooting support, see “Library batch import fails” on page 372.

Find libraries

You can sort, search, and filter information to narrow the libraries that are listed in the **Samples / Manage Libraries** screen in Genexus™ Software. The sort, search, and filter tools help you organize the libraries and find a specific library.

For information to search for a library by a unique identifier, such as the library batch ID or the library name, see “Search for libraries” on page 58.

For information to filter libraries by a common trait, such as the type of nucleic acid in the library, see “Filter the list of libraries” on page 59.

Search for libraries



Libraries are listed in the **Samples / Manage Libraries** screen in Genexus™ Software. The most recently created library is at the top of the list.

You can search for libraries with keywords or text that are included in names that are associated with the library.


- **Library Batch ID**
- **Assay Full Name**
- **Sample Name**
- **Library Name**
- **Barcode**

IMPORTANT! Search terms must adhere to the following rules.

- An asterisk (*) is not allowed in the search field for use as a wildcard.
- Searches are case-insensitive, that is, both upper-case and lower-cases letters are found regardless the case of search term letters.
- Searches find every occurrence of a continuous string. For example, a search for *demo* in assay names returns a list of all assays with a name that includes *demo*. For example, demo1, demo2, and so on.
- Spaces are removed during searches, and are therefore not recommended. For example, a search for *demo 1* returns results only for data that includes the string *demo1*.

-
1. In the menu bar, click **Samples ▶ Manage Libraries**.
 2. In the **Manage Libraries** screen, in the column header of interest, click  **(Filter)**.
 3. In the search field, enter a full or partial name or ID of at least 3 characters.
When you have entered at least 3 characters, the filter dialog box limits the list in the filter dialog box based on the entry.
 4. In the filter dialog box, select from the limited list.
 - Select a checkbox next to the name or ID to select one or more items.
 - Click **Select All** to select all of the items listed in the filter dialog box.
 - Click **Clear** to close the filter dialog box.
 5. Click **Filter**.
In the column header,  changes color to indicate that a filter is applied.

The filtered list of libraries is shown in the **Manage Libraries** screen.

Click  **Clear Filters** to remove all filters and view the full list of libraries.

Sort the list of libraries

You can sort the list of libraries to make it easier to find a library in Genexus™ Software.

By default, libraries are listed in the **Samples / Manage Libraries** screen by the date the library was created. The most recently created library is at the top of the list.

You can change the order that the libraries are listed and sort the libraries alphabetically or reverse alphabetically by the **Library Batch ID**, **Assay Full Name**, or by **Sample Name**.

1. In the menu bar, click **Samples ▶ Manage Libraries**.
2. In the **Manage Libraries** screen, click the column header of interest.
The list of libraries is sorted alphabetically.
3. Click the column header again to reverse the order.


Filter the list of libraries


You can apply filters to narrow search results, or shorten the list of libraries when you view the list of libraries in Genexus™ Software.

You can also search for libraries with keywords or text. For more information, see “Search for libraries” on page 58.


In addition to the filter tools in the column headers, you can also filter the list of libraries with a dropdown list. For more information, see “Manage libraries and library batches” on page 60.

1. In the menu bar, click **Samples ▶ Manage Libraries**.
2. In the **Manage Libraries** screen, select one or more filters.

Option	Description
Filter and search by the Library Batch ID or other column headers.	For more information, see “Search for libraries” on page 58.
Filter by the type of nucleic acid in the library.	In the Nucleic Acid Type column header, click  (Filter) then use the options to filter the list in the filter dialog box.
Remove all selections and close the filter dialog box.	In the filter dialog box, click Clear .

3. In the filter dialog box, click **Filter** to apply the filtered selections.
In the column header,  changes color to indicate that a filter is applied.

The list of libraries is filtered according to the selections.




Click  **Clear Filters** to remove all filters and view the full list of libraries.

Manage libraries and library batches

You can view library information, edit, delete, add notes and review the history of changes to libraries and library batches in Genexus™ Software.

You can also search, sort, and filter the list of libraries. For more information, see “Find libraries” on page 57.

1. In the menu bar, click **Samples ▶ Manage Libraries**.
2. In the **Manage Libraries** screen, perform one or more actions.

Option	Description
View more library information	Click a library name in the library list.
View all libraries	In the Filter Libraries by dropdown menu, select All .
View libraries that are not in a run plan	In the Filter Libraries by dropdown menu, select To Be Planned .
View multiple pages of libraries	Click the page numbers and arrows at the bottom of the screen.
Delete a library batch	Select the checkbox next to one or more library batch IDs, then click  Delete .
Edit a library batch	<ol style="list-style-type: none"> 1. Place the pointer over the row of a library that you want to edit, then click Edit. 2. Modify the information in the Edit Library batch screen, then click Submit. <p>You can edit a library batch only if none of the libraries in the library batch have been assigned to a run.</p>
View or export the audit trail of a library batch	<ol style="list-style-type: none"> 1. Place the pointer over the row of a library that you want to audit, then click Audit. The Audit Details screen opens with time-stamped audit records that include details and changes for each action performed on the library batch. 2. Click the time stamp and action listed in the left pane of the screen to view audit details for other actions. 3. Click  Download in the upper right corner of the screen to download the record as a PDF file.
View, add, or delete notes for a library batch	<ol style="list-style-type: none"> 1. Place the pointer over the row of a library of interest, then click Notes. The Add Notes dialog box opens and notes that are associated with the library are listed by user, date, and time. 2. In the Add Notes dialog box, click + Add Note. 3. Enter the note, then click Save. 4. If needed, in the Add Notes dialog box, in the row of the note that you want to delete, click  (Delete).



Plan and manage runs

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Runs created in Genexus™ Software contain the settings that are used in library preparation, templating, sequencing, and analysis, including sample information and plate location, assays, and barcodes. Runs are used to track samples, consumables, and chips throughout the library preparation, templating, sequencing, and data analysis workflow.

There are four types of runs that you can plan in Genexus™ Software. The type of run that you plan depends on the sample type. For more information about samples, see “About samples and library batches” on page 27.

Types of runs in Genexus™ Software

Run type	Description
Sample to Result	An integrated run for sequential and automated nucleic acid purification and sequencing. This run type requires the Genexus™ Purification Instrument.
Nucleic Acid to Result	A sequencing run that uses nucleic acid samples as input.
Library to Result	A sequencing run that uses libraries that you have previously prepared as input.
BAM to Result	A run that analyzes BAM samples.

Genexus™ Software guides you step-by-step through the process to set up a run. The software prompts you to select required information and consumables, then provides a printed run setup guide to help you load consumables on the Genexus™ Integrated Sequencer and the Genexus™ Purification Instrument, if applicable.

You can plan sequencing runs that use nucleic acid samples with a **Nucleic Acid to Result** run or that use manually prepared libraries with a **Library to Result** run.

You can plan runs for sequential and automated nucleic acid purification and sequencing with a **Sample to Result** run. You must connect at least one Genexus™ Purification Instrument in order to execute a **Sample to Result** run. For more information, see “Integrated nucleic acid isolation in Sample to Result runs” on page 65.

Samples that are imported from BAM files can be analyzed in the software. For more information, see “Plan a BAM to Result run” on page 78.

Before you plan a run

The following conditions must be met before you plan a run in Genexus™ Software.

- Sample information is accurate in the software and the name assigned to each sample is unique. For more information, see “Enter and manage samples” on page 27.
- A Genexus™ Purification Instrument is integrated for use with the system before you plan a **Sample to Result** run.
- Ensure that the following conditions are met for a **Library to Result** run.
 - Library batches are prepared and each batch uses a unique library batch ID.
 - Each sample library in a library batch is prepared with and assigned a unique barcode or barcode pair.

For more information, see “Enter and manage libraries” on page 54.

The software returns an error message when any of the conditions are not met.

Find a run plan

You can sort, search, and filter information to narrow the run plans that are listed in the **Runs / Manage Runs** screen. The sort, search, and filter tools help you organize runs and find a specific run plan in Genexus™ Software. You can also apply the search, sort, and filter tools in the **Assays** step when you plan a run. Use the tools to limit the assays that are listed so that you can easily identify one or more assays to include in the run plan.

For information to search for a run plan by a unique identifier, such as the name of the run plan or the assay applied to the run plan, see “Search for a run plan” on page 63.

For information to filter run plans by a common trait, such as the run plans that include purification, see “Filter the list of run plans” on page 64.



Search for a run plan

Run plans are listed in the **Runs / Manage Runs** screen in Genexus™ Software. The most recently created run plan is on the top of the list.

You can search for run plans with keywords or text that are included in the name of the run plan or the name of an assay applied to the run plan.

IMPORTANT! Search terms must adhere to the following rules.

- An asterisk (*) is not allowed in the search field for use as a wildcard.
 - Searches are case-insensitive, that is, both upper-case and lower-cases letters are found regardless the case of search term letters.
 - Searches find every occurrence of a continuous string. For example, a search for *demo* in assay names returns a list of all assays with a name that includes *demo*. For example, demo1, demo2, and so on.
 - Spaces are removed during searches, and are therefore not recommended. For example, a search for *demo 1* returns results only for data that includes the string *demo1*.
-

1. In the menu bar, click **Runs ▶ Manage Runs**.
2. In the **Manage Runs** screen, in the **Run Name** column header or **Assay Name** column header, click  (**Filter**).
3. In the search field, enter a full or partial assay or run name of at least 3 characters.
When you have entered at least 3 characters, the filter dialog box limits the list in the filter dialog box based on the entry.
4. In the filter dialog box, select from the limited list.
 - Select a checkbox next to a run or assay name to select one or more items.
 - Click **Select All** to select all items listed in the filter dialog box.
 - Click **Clear** to close the filter dialog box.
5. Click **Filter**.
In the column header,  (**Filter**) changes color to indicate that a filter is applied.

The filtered list of run plans is shown in the **Manage Runs** screen.

Click  **Clear Filters** to remove all filters and view the full list of run plans.

Sort the list of run plans

You can sort the list of run plans to make it easier to find run plans in Genexus™ Software.

By default, run plans are listed in the **Runs / Manage Runs** screen by the date the run plan was created. The most recently created run plan is at the top of the list.



You can change the order that the run plans are listed and sort the run plans alphabetically or reverse alphabetically.


1. In the menu bar, click **Runs ▶ Manage Runs**.
2. In the **Manage Runs** screen, click the **Run Name** heading.
The list of runs is sorted alphabetically.
3. Click the column header again to reverse the order.

Filter the list of run plans

You can apply filters to narrow search results, or shorten the list of run plans when you view the list of run plans in Genexus™ Software. You can also apply filters in the **Assays** step when you plan a run to help find one or more assays to include in the run plan.

1. In the menu bar, click **Runs ▶ Manage Runs**.
2. In the **Manage Runs** screen, select one or more filters.

Option	Description
Filter and search by the run name or the assay selected in the run.	For more information, see “Search for a run plan” on page 63.
Filter by the number of samples in the run.	In the Total Samples column header, click  (Filter) then use the options to filter the list in the filter dialog box based on the number or span of numbers of samples in the run.
List runs based on whether the run includes purification.	In the Run Status column header, click  (Filter) then select a status. <ul style="list-style-type: none"> • Select Planned to list all Nucleic Acid to Result, Library to Result, and BAM to Result planned runs. • Select Purification Planned to list planned Sample to Result runs only.
Remove all selections and close the filter dialog box.	In the filter dialog box, click Clear .

3. In the filter dialog box, click **Filter** to apply the filtered selections.
In the column header,  (**Filter**) changes color to indicate that a filter is applied.

The list of run plans is filtered according to the selections.

Click  **Clear Filters** to remove all filters and view the full list of run plans.

Integrated nucleic acid isolation in Sample to Result runs

A **Sample to Result** run is an integrated run for sequential and automated nucleic acid purification and sequencing. You can create **Sample to Result** runs for the following sample types:

- Blood (buffy coat)
- Blood (plasma)
- Blood (whole)
- Bone marrow
- Cell line
- Core needle biopsy (CNB)
- Formalin-fixed, paraffin-embedded (FFPE) tissue
- Fine needle aspiration (FNA)
- Fresh frozen tissue

When you plan a **Sample to Result** run with multiple assays, the nucleic acid isolation is performed in a separate batch for each assay. Nucleic acid isolation is also performed in a separate batch for each sample type, except for FFPE tissue and FNA, which are grouped together in a single batch for each assay. The Genexus™ Purification Instrument can run one nucleic acid isolation batch at a time. You can run different nucleic acid isolation batches simultaneously on multiple purification instruments or sequentially on a single instrument.

You can select which samples are sequenced after nucleic acid isolation. The options are as follows.

- Sequence all samples.
- Sequence only the samples that have a concentration within a specified threshold.
- Review the samples that do not have a concentration within a specified threshold, then select samples to sequence on a per sample-basis.

For more information, see the *Genexus™ Purification System User Guide* MAN0018475.

Plan a Sample to Result run

This procedure applies only if a Genexus™ Purification Instrument is used with the Genexus™ Integrated Sequencer.

Ensure that the following prerequisites are complete before you plan a **Sample to Result** run.

- Integrate a Genexus™ Purification Instrument with the system.
- Enter sample information into Genexus™ Software. For more information, see “Create a new sample” on page 33, or “Import samples” on page 34.

Planning a **Sample to Result** run is organized into five steps: **Setup**, **Assays**, **Samples**, **Purification**, and **Review**. Progress through the steps is tracked in the upper left corner of the **Runs / Sample to Result** screen.

1. In the menu bar, click **Runs** ▶ **Sample to Result**.

Note: You can also click + **Sample to Result** in the **Runs / Manage Runs** screen.

2. In the **Setup** step, enter or make the following selections.
 - a. In the **Plan** section, enter a unique name.
The name is limited to 50 characters and no spaces are allowed.
 - b. (Optional) In the **Reporting (Optional)** section, enable **Generate Report** to generate a variant report that uses the default report template.
 - c. (Optional) In the **Reporting (Optional)** section, enable **Upload BAM files to Server** to upload BAM files to another server.

Option	Description
Upload BAM files to another Genexus™ Software server.	In the dropdown lists that appear, select the target Genexus™ Software server and version. To configure a Genexus™ Software account that you can use for data uploads, click Set up Account . For more information, see “Link Genexus™ Software accounts (manager/administrator)” on page 253.
Automatically upload data for further analysis with Ion Reporter™ Software.	In the dropdown lists that appear, select the Ion Reporter™ Software account and Ion Reporter™ Software version. To configure an Ion Reporter™ Server account that you can use for data uploads, click Set up Account . For more information, see “Configure an Ion Reporter™ Server account (manager/administrator)” on page 254.

Alternatively, you can upload BAM files to a server after a run. For more information, see “Upload results files to another Genexus™ Software server” on page 173 and “Upload results files to Ion Reporter™ Software” on page 171.

- d. Click **Next**.
3. In the **Assays** step, select one or more assays that you want to include in the run.
Only assays that are configured for **Sample to Result** runs are listed. If a chip is installed in the sequencer, the **Chip View** graphic in the lower left corner indicates the lanes that are available for sequencing.
 - a. Use the ▼ (**Filter**) tools in table column headers to find assays of interest, if desired.
For more information, see “Find the assay that you want to use in a run plan” on page 79.
 - b. In the **Application Type** column for the assay of interest, select one or more application types, such as **DNA and Fusions**, to include each selected application type for the assay in the run plan.
After selecting an assay, the list is filtered to show compatible assays that can be selected and run at the same time. Assays must have the same **Analysis Version**, **Chip Type**, and **Library Chemistry** to be compatible with the selected assay.

Note: To create a new assay, see Chapter 8, “Create and manage assays (manager/administrator)”.

 - c. If more assays are included in the run, repeat substep 3b for each extra assay.

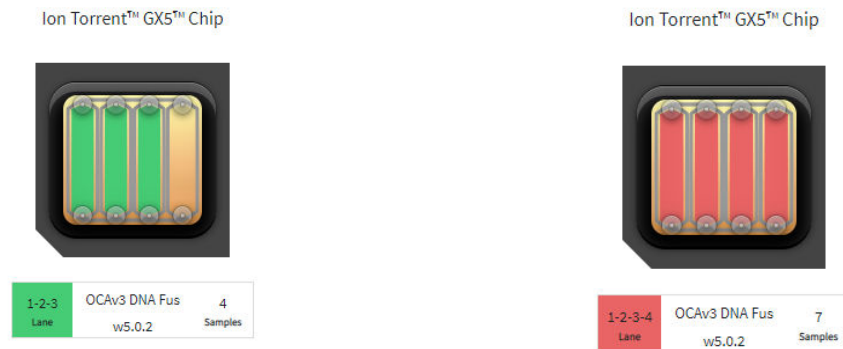
- d. Click **Next**.
4. In the **Samples** step, select the samples that you want to run with each application type of each assay.
- a. Select the checkbox next to each sample that you want to assign to the application type of an assay, then in the **Selected Assays** pane, for the assay and application type that you want to use for the selected samples, click **Assign**.

The **Chip View** updates to show the lanes used in the run. Lane usage is calculated based on the number of samples (including a no template control, if selected) and the minimum read counts per sample for the assay. Green denotes a chip lane in the run containing assigned samples within lane capacity.

If the

$$\text{minimum reads per sample} \times \text{number of samples}$$

exceeds the chip or lane well capacity, a dialog box appears after you click **Assign** asking you to confirm that you want to continue. After confirmation, the **Chip View** updates and shows the lane color as red instead of green. The run is allowed if the lane capacity is exceeded, but you may not achieve the required reads per sample to pass QC metrics.



Green lane color denotes lane usage and sample assignment within lane capacity.

Red lane color denotes sample assignment that exceeds lane capacity.

- b. If you selected more than one application type or assay, repeat substep 4a for each application type for each assay in the run plan.
- c. If needed, edit samples in one of the following ways.
- Click **View & Remove**, make the selections, then click **Update**.
 - Click **Remove All**, to remove all sample assignments for all assays.

- d. If you selected the **Upload BAM files to Server** reporting option in the **Setup** step, make the following selections from the **Genexus Workflow** or **Ion Reporter Workflow** dropdown list for each assay that you selected.
- Select **Upload Only** to upload sample data to the selected server automatically after run completion.
 - Select the desired Genexus™ Software assay or Ion Reporter™ Software analysis workflow to upload sample data *and* perform an analysis in the target software automatically after run completion.
-
- Note:** In order for the Ion Reporter™ Software analysis workflow to appear in the list, you must tag the analysis workflow for use with the IonReporterUploader plugin. For more information, see “Tag an Ion Reporter™ Software analysis workflow for use with the IonReporterUploader plugin” on page 172.
-
- e. If desired, for each application type of each assay in the run plan, select **NTC** to include a no template control and select **Control Sample** to include a positive control.
The **Chip View** updates to show the lanes used in the run for the included controls.
- f. Click **Next**.

5. In the **Purification** step, review and edit selections.

- a. Review and edit purification selections.

Option	Description
Protocol Selection	Select a protocol from the dropdown list for each purification kit.
Elution Vol. (µL)	Modify the elution volume within the allowable range, if needed.
Review?	<ul style="list-style-type: none"> • Select the checkbox in the row of a purification kit to review the run after purification, before sequencing, for the kit. This allows you to review samples when the quantity of a nucleic acid is below the quality threshold. You can then decide on a per sample-basis whether or not to sequence the samples. • Deselect the Review? checkbox in the row of a purification kit to enable the sequencer to bypass sequencing samples with concentrations below the threshold for the kit. This option does not require you to review samples during the run. <p>Note: This selection, whether or not it is selected, overrides the Review? setting of the assay.</p>

- b. Review and edit the information for control samples, if applicable.

Option	Description
Conc. (ng/µL)	Review and edit the concentration of control samples, if needed.
Kit Barcodes	Enter the extraction kit barcode for each control sample, if needed.

- c. Click **Next**.

6. In the **Review** step, review the run plan summary, then click **Save**.

Note: A run can include multiple purification batches.

For more information, see “Integrated nucleic acid isolation in Sample to Result runs” on page 65.

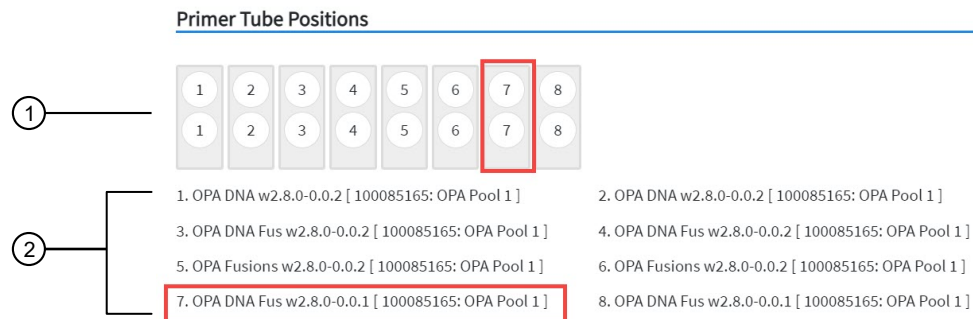
The run plan summary lists the following details:

- the consumables that are required for purification
- the consumables that are required for sequencing
- how much volume to load
- details about the assay or assays.
- where to load samples

Tip: Click **Sequencing** to expand the sequencing section of the run plan summary.

IMPORTANT! The consumables required for sequencing can change. After purification, review the run plan in the **Runs / Manage Runs** screen for updated sequencing consumable information. For more information, see “Print a run plan” on page 82.

Note: If you are using an assay with Ion AmpliSeq™ HD library chemistry, the primer pool positions show that HD primer pools occupy both rows.



① The position of the primer tubes.

② The legend for the image.

In the example outlined in red, primer tube 7, which contains OPA Pool 1 for the OPA DNA Fusion assay, is in the seventh position from the left.

The run plan appears in the **Manage Runs** screen with the name that you specified.

After you select the run and load the Genexus™ Purification Instrument, the run is started on the purification instrument screen.

When purification and sequencing are complete, you can recover the leftover volume from each library that was prepared in the run. For more information to find, recover, and purify the leftover library preparations, see the *Genexus™ Integrated Sequencer User Guide* (Pub. No. MAN0017910).

Review samples for Sample to Result runs

A **Sample to Result** run integrates nucleic acid purification and sequencing. When you create a **Sample to Result** run plan, you can include an option that allows samples to be reviewed before library preparation starts. Samples that do not meet a specified concentration threshold after purification have a run status of **Purification Review Required**.

For more information about **Sample to Result** runs, see “Integrated nucleic acid isolation in Sample to Result runs” on page 65.

You can also apply this option when you create an assay. For more information, see “Custom assays for Sample to Result runs (manager/administrator)” on page 208.

1. In the menu bar, click **Results ▶ Run Results**.

Note: **Sample to Result** runs that require review after purification have a **Run Status** of **Purification Review Required**.

2. In the **Run Results** screen, place the pointer over the row of the run plan of interest, then click **Review**.
The **Purification Samples** screen opens.
3. Review the concentration for each sample, then select the checkbox in the row for each sample you want to sequence.
4. Click **Submit**.

The samples are ready for sequencing. For more information, see the *Genexus™ Purification System User Guide* (Pub. No. MAN0018475).

Abort a Sample to Result run

When you create a **Sample to Result** run plan, you can include an option that allows samples to be reviewed before library preparation starts. When you enable the review of samples after purification, you can end the run before sequencing if needed.

1. In the menu bar, click **Results ▶ Run Results**.
2. In the **Run Results** screen, place the pointer over the row of the run that you want to abort, then click **Abort**.
3. In the confirmation dialog box that appears, click **Yes** to confirm that you want to abort the run.

In the Run Summary, you can view information about the purification run. The status of the run in the Run Summary is Aborted. For more information, see “View the run summary” on page 108.

Plan a Nucleic Acid to Result run

You can plan a run for sequencing that starts with nucleic acid samples. In Genexus™ Software 6.6 and later, a run that starts with nucleic acid samples is called a **Nucleic Acid to Result** run.

Planning a **Nucleic Acid to Result** run is organized into steps: **Setup, Assays, Samples, Sample Plate**, and **Review**. You can view progress through the steps in the upper left corner of the **Runs / Nucleic Acid to Result** screen.

Ensure that the following prerequisites are complete before you plan a **Nucleic Acid to Result** run.

- Enter sample information into Genexus™ Software. For more information, see “Create a new sample” on page 33, or “Import samples” on page 34.
- Ensure that the assay that you want to use in the run exists in the software. For more information, see “About assays in Genexus™ Software” on page 190.
- Quantify sample concentration, if you do not intend to dilute sample concentrations manually to the target concentration of the assay. For more information, see the *Genexus™ Integrated Sequencer User Guide* (Pub. No. MAN0017910).

1. In the menu bar, click **Runs ▶ Nucleic Acid to Result**.

Note: Alternatively, you can click **+ Nucleic Acid to Result** in the **Runs / Manage Runs** screen.

2. In the **Setup** step, enter or make the following selections.

- a. In the **Plan** section, enter a unique name.
The name is limited to 50 characters and no spaces are allowed.
- b. *(Optional)* In the **Reporting (Optional)** section, ensure that **Generate Report** is enabled to generate a variant report using the default report template.
- c. *(Optional)* In the **Reporting (Optional)** section, enable **Upload BAM files to Server** to upload BAM files to another server.

Option	Description
Upload BAM files to another Genexus™ Software server.	In the dropdown lists that appear, select the target Genexus™ Software server and version. To configure a Genexus™ Software account that you can use for data uploads, click Set up Account . For more information, see “Link Genexus™ Software accounts (manager/administrator)” on page 253.
Automatically upload data for further analysis with Ion Reporter™ Software.	In the dropdown lists that appear, select the Ion Reporter™ Software account and Ion Reporter™ Software version. To configure an Ion Reporter™ Server account that you can use for data uploads, click Set up Account .

Alternatively, you can upload BAM files to a server after a run. For more information, see “Upload results files to another Genexus™ Software server” on page 173 and “Upload results files to Ion Reporter™ Software” on page 171.

- d. Click **Next**.

3. In the **Assays** step, select one or more assays that you want to include in the run.
 - a. Use the **Filter** tools in table column headers to find assays of interest, if desired.
For more information, see “Find the assay that you want to use in a run plan” on page 79.
 - b. In the **Application Type** column for the assay of interest, select one or more application types, such as **DNA and Fusions** or **DNA**, to include each selected application type for the assay in the run plan.
After selecting an assay and the research application for the assay, the list is filtered to show compatible assays that can be selected and run at the same time.

Note: To create a new assay, see Chapter 8, “Create and manage assays (manager/administrator)”.

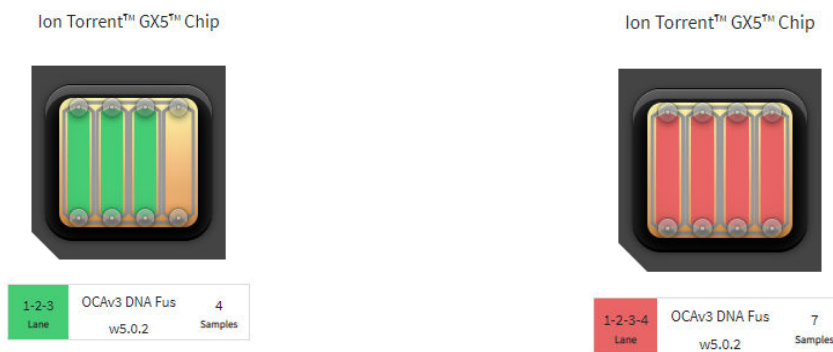
- c. If more assays are included in the run, repeat substep 3b for each extra assay.
 - d. Click **Next**.
4. In the **Samples** step, select the samples that you want to run with each application type of each assay.
 - a. Select the checkbox next to each sample that you want to assign to the application type of an assay, then in the **Selected Assays** pane, for the assay and application type that you want to use for the selected samples, click **Assign**.

The **Chip View** updates to show the lanes used in the run. Lane usage is calculated based on the number of samples (including a no template control, if selected) and the minimum read counts per sample for the assay. Green denotes a chip lane in the run containing assigned samples within lane capacity.

If the

$$\text{minimum reads per sample} \times \text{number of samples}$$

exceeds the chip or lane well capacity, a dialog box appears after you click **Assign** asking you to confirm that you want to continue. After confirmation, the **Chip View** updates and shows the lane color as red instead of green. The run is allowed if the lane capacity is exceeded, but you may not achieve the required reads per sample to pass QC metrics.



Green lane color denotes lane usage and sample assignment within lane capacity.

Red lane color denotes sample assignment that exceeds lane capacity.

- b. If you selected more than one application type or assay, repeat substep 4a for each application type for each assay in the run plan.
- c. If needed, edit samples in one of the following ways.
 - Click **View & Remove**, make the selections, then click **Update**.
 - Click **Remove All**, to remove all sample assignments for all assays.
- d. If you selected the **Upload BAM files to Server** reporting option in the **Setup** step, make the following selections from the **Genexus Workflow** or **Ion Reporter Workflow** dropdown list for each assay that you selected.
 - Select **Upload Only** to upload sample data to the selected server automatically after run completion.
 - Select the desired Genexus™ Software assay or Ion Reporter™ Software analysis workflow to upload sample data *and* perform an analysis in the target software automatically after run completion.

Note: In order for the Ion Reporter™ Software analysis workflow to appear in the list, you must tag the analysis workflow for use with the IonReporterUploader plugin. For more information, see the *Ion Reporter™ Software 5.18 User Guide* (Pub. No. MAN0024776).

- e. If desired, for each application type of each assay in the run plan, select **NTC** to include a no template control.

The **Chip View** updates to show the lanes used in the run for the included no template controls.
 - f. Click **Next**.
5. In the **Sample Plate** step, review position assignments in the sample plate. Drag-and-drop samples and no template controls to edit the location of samples and controls, if applicable.
- a. If desired, enter the extraction kit barcode for one or more samples or controls. For a single sample, in the row of the sample of interest, in the **Kit Barcodes** column, enter the extraction kit barcode or control kit barcode, if applicable. For multiple samples or controls, select the samples and controls, then click [✎](#) **Assign Kit Barcodes**. In the **Assign Kit Barcodes** dialog box, enter the extraction kit barcode for the samples, and if applicable, enter the barcode for the no template control.
 - b. Modify the concentration of samples, if needed. For a single sample, in the row of the sample of interest, in the **Conc. (ng/μl)** column, edit the concentration.

To modify the concentration of multiple samples, select the samples of interest, then click [✎](#) **Edit Concentration**. In the **Bulk Edit** dialog box, enter the concentration for all selected samples, then click **Submit**. The concentration for each selected sample is updated to the new value.

If a sample concentration is $\leq 1,024X$ of the target concentration for the assay, which is displayed as a default value for each sample in the **Sample Plate** screen, the sequencer automatically dilutes the sample to the target concentration during the run. If a sample concentration is greater than this value, you must manually dilute the sample to the target

concentration, or to a value within range for automated dilution before loading on the sample plate.

Note: The sample volume that is required for library preparation is not adjustable. The volume depends on the number of primer pools in the assay, sample type, and library chemistry.

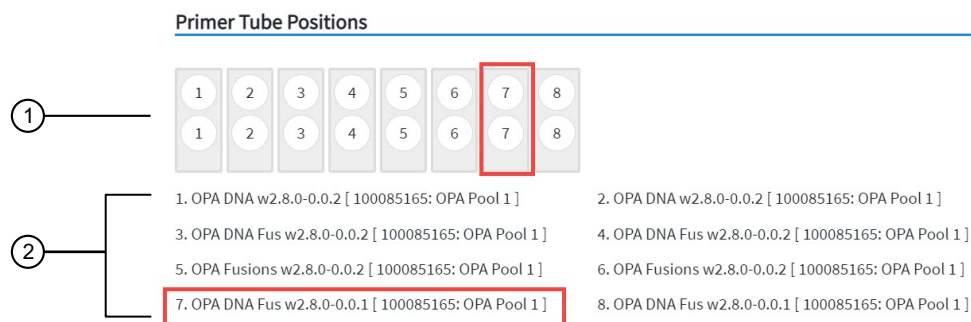
- c. Ensure that sample plate information is correct, then click **Next**.
6. In the **Review** step, review the run plan summary, then click **Save & Print** to print the run setup guide, if desired. Click **Save** to save the run without printing.

Note: To print the run setup guide, you must allow pop-ups in the browser.

The run plan summary lists the following details.

- the consumables that are required for this run
- where to load samples and primer pool tubes
- how much sample volume to load
- details about the assay.

Note: If you are using an assay with Ion AmpliSeq™ HD library chemistry, the primer pool positions show that HD primer pools occupy both rows:



① The position of the primer tubes.

② The legend for the image.

In the example outlined in red, primer tube 7, which contains OPA Pool 1 for the OPA DNA Fusion assay, is in the seventh position from the left.

After saving, the run appears in the **Manage Runs** screen in the run list with the name you specified.

After you select the run and load the sequencer, the run is started on the sequencer screen.

When the sequencing run is complete, you can recover the leftover volume from each library that was prepared in the run. For more information to find, recover, and purify the leftover library preparations, see the *Genexus™ Integrated Sequencer User Guide* (Pub. No. MAN0017910).

Plan a Library to Result run

You can plan runs for sequencing that start with libraries. In Genexus™ Software 6.6 and later, this is called a **Library to Result** run. Before you can plan a **Library to Result** run, you must first enter sample information and prepare a library batch that associates an assay with the library batch. The library batch must be created with the assay to be used in the run. The assay specifies the barcode set that was used to prepare the sample libraries.

For more information, see “Enter and manage libraries” on page 54.

If the sample libraries were prepared using a barcode set that is not specified in an assay you want to use in the run, you must do the following:

- Create a new assay, or copy an existing assay, and specify the new barcode set in assay setup.
- Prepare a library batch that selects the new assay.

For more information, see “Create an assay (manager/administrator)” on page 194, “Copy an assay (manager/administrator)” on page 196, and “Prepare a library batch” on page 54.

Genexus™ Software guides you through the four steps of planning a **Library to Result** run: **Setup**, **Assays**, **Library Batches**, and **Review**. You can view progress through the steps in the upper left corner of the **Runs / Library to Result** screen.

1. In the menu bar, click **Runs** ▶ **Library to Result**.

Note: You can also click + **Library to Result** in the **Runs / Manage Runs** screen.

2. In the **Setup** step, enter a name for the run, then configure the reporting options.
 - a. In **Run Name**, enter a unique name.
The name is limited to 50 characters and no spaces are allowed.
 - b. *(Optional)* In the **Reporting (Optional)** section, ensure that **Generate Report** is enabled to generate a variant report using the default report template.

- c. (Optional) In the **Reporting (Optional)** section, enable **Upload BAM files to Server** to upload BAM files to another server.

Option	Description
Upload BAM files to another Genexus™ Software server.	In the dropdown lists that appear, select the target Genexus™ Software server and version. To configure a Genexus™ Software account that you can use for data uploads, click Set up Account . For more information, see “Link Genexus™ Software accounts (manager/administrator)” on page 253.
Automatically upload data for further analysis with Ion Reporter™ Software.	In the dropdown lists that appear, select the Ion Reporter™ Software account and Ion Reporter™ Software version. To configure an Ion Reporter™ Server account that you can use for data uploads, click Set up Account .

Alternatively, you can upload BAM files to a server after a run. For more information, see “Upload results files to another Genexus™ Software server” on page 173 and “Upload results files to Ion Reporter™ Software” on page 171.

- d. Click **Next**.

If a chip is installed in the sequencer, the **Chip View** graphic in the lower left corner indicates the lanes that are available for sequencing.

3. In the **Assays** step, select one or more assays that you want to include in the run.

For the assay to be selectable at this step, you must have prepared a library batch that assigns the assay to the batch. The assay specifies the barcode set that was used to prepare the sample libraries.

- a. Use the ▼ (**Filter**) tools in table column headers to find assays of interest, if desired.
For more information, see “Find the assay that you want to use in a run plan” on page 79.
- b. In the **Application Type** column for the assay of interest, select one or more application types, such as **DNA and Fusions**, to include each selected application type for the assay in the run plan.

After selecting an assay and the research application for the assay, the list is filtered to show compatible assays that can be selected and run at the same time.

Note: To create a new assay, see Chapter 8, “Create and manage assays (manager/administrator)”.

- c. If more assays are included in the run, repeat substep 3b for each extra assay.
- d. Click **Next**.

4. In the **Library Batches** step, select the library batch that you want to use in the run, then click **Next**.

Note: Only one library batch can be selected per assay. However, you can plan a library run that uses multiple assays if you select multiple compatible assays in the **Assays** step.

- a. If you selected the **Upload BAM files to Server** reporting option in the **Setup** step, make the following selections from the **Genexus Workflow** or **Ion Reporter Workflow** dropdown list.
- Select **Upload Only** to upload sample data to the selected server automatically after run completion.
 - Select the desired Genexus™ Software assay or Ion Reporter™ Software analysis workflow to upload sample data *and* perform an analysis in the target software automatically after run completion.

Note: In order for the Ion Reporter™ Software analysis workflow to appear in the list, you must tag the analysis workflow for use with the IonReporterUploader plugin. For more information, see the *Ion Reporter™ Software 5.18 User Guide* (Pub. No. MAN0024776).

The **Chip View** updates to show the lanes to be used in the run as green. Lane usage is calculated based on the number of samples and minimum reads per sample entered at assay setup.



Green lane color denotes lane usage and sample assignment within lane capacity.

Red lane color denotes sample assignment that exceeds lane capacity.

If the

$$\text{minimum reads per sample} \times \text{number of samples}$$

exceeds the chip or lane well capacity, a dialog box appears after you click **Next** asking you to confirm that you want to continue. After clicking **Yes**, the **Chip View** updates and shows the lane color as red instead of green. In the example shown at right, seven samples were included in a library batch instead of four. The run is allowed if the lane capacity is exceeded, but you may not achieve the required reads per sample to pass QC metrics.

5. In the **Review** step, review the run plan summary.
- Click **Save and Print** to print the run setup guide.
 - Click **Save** to save the run without printing.

Note: To print the run setup guide, you must allow pop-ups in the browser.

The run plan summary lists the consumables that are required for the run, where to load the library batch on the sample plate, and how much library volume to load.

The run appears in the run list on the **Manage Runs** screen with the name you specified.

After you select the run and load the sequencer, the run is started on the sequencer screen.

Plan a BAM to Result run

Before you can plan a **BAM to Result** run, you must enter BAM samples into Genexus™ Software. For more information, see “Upload a BAM file to create a sample or samples” on page 35.

Planning a run to analyze BAM samples is organized into steps: **Assays**, **Samples**, and **Review**. You can view progress through the steps in the upper left corner of the **Runs / BAM to Result** screen.

1. In the menu bar, click **Runs** ▶ **BAM to Result**.

Note: You can also click + **BAM to Result** in the **Runs / Manage Runs** screen.

2. In the **Assays** step, enter or make the following selections.
 - a. Enter a unique name.
The name is limited to 50 characters and no spaces are allowed.
 - b. Use the ▼ (**Filter**) tools in table column headers to find assays of interest, if desired.
For more information, see “Find the assay that you want to use in a run plan” on page 79.
 - c. In the **Application Type** column for the assay of interest, select the application type, such as **DNA and Fusions** or **DNA**, to include the selected application type for the assay in the run plan.
You can select only one application type of an assay per run.
 - d. Click **Next**.
3. In the **Samples** step, select the BAM samples from the list that you want to analyze, then click **Assign**.

Note: Only BAM samples that are compatible with the selected assay are listed. Compatibility is determined by the sample type, such as DNA or DNA and Fusion.

- a. If needed, click ⊗ in an assigned sample tile to remove the sample from the plan.
 - b. Click **Next**.
4. In the **Review** step, review the samples in the run plan summary, then click **Save and Launch**.
The run is listed in the **Results / Run Results** screen. The run is launched or the run is queued if other runs are in progress.

Find the assay that you want to use in a run plan

You can search and filter the list of assays when you plan a run to help you find the assay that you want to use in a run plan in Genexus™ Software.

For information to search for an assay by a unique identifier, such as the name of the assay, see “Search for an assay to use in a run plan” on page 79.



For information to filter assays by a common trait, such as the analysis version, see “Filter the list of assays shown when you plan a run” on page 80.

Search for an assay to use in a run plan


When you plan a run, you can search for an assay in Genexus™ Software with keywords or text that are included in the name or full name of an assay to help find the assay that you want to use in the run plan.

IMPORTANT! Search terms must adhere to the following rules.

- An asterisk (*) is not allowed in the search field for use as a wildcard.
- Searches are case-insensitive, that is, both upper-case and lower-cases letters are found regardless the case of search term letters.
- Searches find every occurrence of a continuous string. For example, a search for *demo* in assay names returns a list of all assays with a name that includes *demo*. For example, demo1, demo2, and so on.
- Spaces are removed during searches, and are therefore not recommended. For example, a search for *demo 1* returns results only for data that includes the string *demo1*.

-
1. In the menu bar, click **Runs**, then click **Sample to Result**, **Nucleic Acid to Result**, **Library to Result**, or **BAM to Result**, based on the type of run that you want to plan.
 2. Enter and make the selections in the **Setup** step, if applicable, then click **Next**.
 3. In the **Assays** step, in the **Assay Full Name** column header, click  (**Filter**).
 4. In the search field, enter a full or partial assay name of at least 3 characters. When you have entered at least 3 characters, the filter dialog box limits the list in the filter dialog box based on the entry.
 5. In the filter dialog box, select assays from the limited list.
 - Select a checkbox next to an assay name to select one or more assays.
 - Click **Select All** to select all of the assays in the filter dialog box.
 - Click **Clear** to close the filter dialog box.
 6. Click **Filter**.
In the column header,  (**Filter**) changes color to indicate that a filter is applied.




The filtered list of assays is shown in the **Assays** step. You can select the assay of interest from the limited list and proceed through the remaining steps for the run plan. see “Plan a Sample to Result run” on page 65, “Plan a Nucleic Acid to Result run” on page 71, “Plan a Library to Result run” on page 75, or “Plan a BAM to Result run” on page 78.


Click  **Clear Filters** to remove all filters and view the full list of assays.

Filter the list of assays shown when you plan a run

When you plan a run in Genexus™ Software, you can apply filters to narrow the list of assays that are shown to help find the assay that you want to use in the run plan.

1. In the menu bar, click **Runs**, then click **Sample to Result, Nucleic Acid to Result, Library to Result, or BAM to Result**, based on the type of run that you want to plan.
2. Enter and make the selections in the **Setup** step, if applicable, then click **Next**.
3. Select from the filter and search options in the column header of interest.

Option	Description
List assays according to the analysis version	<ol style="list-style-type: none"> 1. Click  (Filter) in the Analysis Version column. 2. Select one or more of the versions that are shown, then click Filter.
List assays according to chip type	<ol style="list-style-type: none"> 1. Click  (Filter) in the Chip Type column. 2. Select Ion Torrent™ GX5™ Chip or Ion Torrent™ GX7™ Chip then click Filter.
List assays according to library chemistry	<ol style="list-style-type: none"> 1. Click  (Filter) in the Library Chemistry column. 2. Select the library chemistry: <ul style="list-style-type: none"> • AmpliSeq – The standard library preparation chemistry for targeted next-generation sequencing. • AmpliSeq HD – High sensitivity library preparation chemistry for targeted next-generation sequencing with unique molecular tags to reduce the number of false positives and improve confidence in variant calling. 3. Click Filter.

4. (Optional) Click  **Clear Filters** to return to the entire list of assays.
5. In the shortened list of assays, select one or more checkboxes in the **Research Application** column for each assay that you want to add to the run, then click **Next**.
6. Click the **Filter Assays By** dropdown menu, if applicable, then select **Assigned** to list only the assays that you have selected for the run plan.
 - Click **Filter Assays By** ▶ **All** dropdown menu, to show all assays again, if desired.

You can proceed through the remaining steps for the run plan. For more information, see “Plan a Sample to Result run” on page 65, “Plan a Nucleic Acid to Result run” on page 71, “Plan a Library to Result run” on page 75, or “Plan a BAM to Result run” on page 78.

Manage runs


Run plans are listed in the **Runs / Manage Runs** screen. The most recently created run is on the top of the list. Only runs that have not started are listed.

In this screen, you can:

- Plan a run.
- Sort and search the list of run plans.
- Review and print a run plan.
- View the audit trail of a run plan.
- Edit a run plan.
- Delete a run plan.
- View, add, or delete the notes for a run plan.

View a run plan

You can view detailed run plan information in Genexus™ Software.

1. In the menu bar, click **Runs ▶ Manage Runs**.
2. In the **Manage Runs** screen, click the run name of interest.
Run plan details are shown in the run plan summary pane. For more information, see “Run plan details” on page 81.
3. When you are finished viewing the run plan details, click another run name to view details for another run plan, or click **✕ (Close)** to return to the **Manage Runs** screen.
4. (Optional) Click  **Print** to print the information about the run.
To print the run plan, you must allow pop-ups in the browser.

You can click **⋮ (More Options)** to perform more actions, such as edit, add notes, or audit the run plan. The actions for each run plan are also available in the **Manage Runs** screen when you place the pointer over the row of a run name. For more information, see “Edit a run” on page 84, “Add a note to a run” on page 84, “Review and download a run history” on page 83.

Run plan details

Detailed run plan information is shown in the run plan summary pane.

Run plan detail	Description
Generate Report	Indicates if a report is automatically generated for the run.
Server	The Ion Reporter™ Software account or the target Genexus™ Software server and version, if any, that was selected for uploading BAM files when the run is complete.
Run Status	The run status. For example, Planned or Purification Planned .
Run Start Date	This information is available when the run is started.
Run Completion Date	This information is available when the run is completed.

(continued)

Run plan detail	Description
Sequencing^[1]	
Consumables Required For This Run	List of the required consumables. For example, the chip and barcodes. The quantity of each consumable is shown in parentheses before the name of the consumable.
Consumables Installed Ready To Use	List of consumables installed on the sequencer and available for the run.
Primer Tube Positions	An image that shows the locations of the primer pool tubes.
Sample Locations	Detailed sample information and an image that shows sample locations on the 96-well plate. For more information, see “The run summary” on page 109.
Assays	
Assay	Detailed assay information for each assay in the run. For more information, see “The run summary” on page 109.

^[1] The Sequencing section is collapsible and expandable.

Additional information for Sample to Result runs


Run plan detail	Description
Purification^[1,2]	
Consumables Required For Purification Batch x	List of the required consumables. For example, the Quantitation Plate and Quantitation Tubes . The quantity of each consumable is shown in parentheses before the name of the consumable. Information for each purification batch is listed in a separate table.
Input Sample Locations For Purification Batch x	Detailed sample information and an image that shows sample locations on the 96-well plate. Information for each purification batch is listed in a separate table.

^[1] The Purification section is collapsible and expandable.

^[2] For information about purification batches, see “Integrated nucleic acid isolation in Sample to Result runs” on page 65.

Print a run plan

You can print detailed run plan information in Genexus™ Software.

1. In the menu bar, click **Runs** ▶ **Manage Runs**.
2. In the **Manage Runs** screen, click a run name in the list.
3. Click  **Print** to print the information about the run plan.
To print the run plan, you must allow pop-ups in the browser.

Run plan details are shown in the run plan summary pane. You can review the information that was entered or generated when you created the run. For more information, see “Run plan details” on page 81.

Review and download a run history


The entire history of a run plan is available to review, export, or print in Genexus™ Software. The history shows the original run plan values and any new values or activities.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

1. In the menu bar, click **Runs** ▶ **Manage Runs**.
2. In the **Manage Runs** screen, place the pointer over the row of the run plan of interest, then click **Audit**.
The **Audit Details** screen opens with time-stamped audit records that include run plan details and changes for each action performed on the run plan. Details for the most recent action are shown.
3. Click the time stamp and action listed in the left pane of the screen to view audit details for other actions.
4. Review the audit record.

Details in the audit record

Item	Description
User name	The name of the person who was signed in when the action was performed.
Run Plan	The name of the run plan.
Data Object Name	The data object that is affected by the action.
Action Performed	Actions performed on the run plan include: <ul style="list-style-type: none"> • Create • Edit
Time Stamp	The time and date of the action.
ID	An identifier to represent a specific action in the audit record. Each action in the audit record includes a unique identifier.
New Values	A list of all values in the run plan. If a value changed as a result of the action, the new value is shown.
Old Values	A list of the previous values in the run plan.

5. Click  **Download** to generate a PDF file, if needed.
Depending on the browser settings, the software automatically downloads the file or prompts you to open or save the file.

Click  **Back** to return to the list of runs.

Edit a run

You can edit runs that have a run status of **Planned** or **Purification Planned**. Parameters that can be edited include reporting options, assays used in the run, samples included in the run, chip assignment, and sample concentration.


1. In the menu bar, click **Runs** ▶ **Manage Runs**.
2. In the **Manage Runs** screen, place the pointer over the row of the run plan of interest, then click **Edit**.
3. In the **Edit Sample Run** workflow, change the desired settings by navigating through the steps clicking **Next**, then click **Save**.

For more information about the settings, see “Plan a Sample to Result run” on page 65, “Plan a Nucleic Acid to Result run” on page 71 and “Plan a Library to Result run” on page 75.

Delete a run

You can delete runs from the software.

You must select at least one run in the **Manage Runs** screen before the option to delete a run appears.

1. In the menu bar, click **Runs** ▶ **Manage Runs**.
2. In the **Manage Runs** screen, select one or more runs to delete by selecting the checkbox next to the run name.
Select all of the runs in the screen by selecting the checkbox above the column.
If needed, click **Clear** to deselect all of the runs.
3. Click  **Delete**.
4. In the **Delete Plan** message box, click **Yes** to delete the selected run.

Add a note to a run

You can add a note or notes to **Nucleic Acid to Result** and **Library to Result** runs that have a run status of **Planned**, or **Sample to Result** runs that have a run status of **Purification Planned** in Genexus™ Software. A note is any written comment or observation that you want to add to the run.

1. In the menu bar, click **Runs** ▶ **Manage Runs**.
2. In the **Manage Runs** screen, place the pointer over the row of the run plan of interest, then click **Notes**.
3. In the **Add Notes** pane, click **+ Add Note**.
4. In the **Add Notes** dialog box, enter the note.
5. Click **Save**.

The note appears in the **Add Notes** pane of a selected run in the list of runs.

View a run note

You can view the notes that have been added to a run. Run notes are time-stamped written notes to capture comments or observations for a run that is planned or completed. A run can contain multiple notes from different users.

You cannot edit run notes. However, you can add multiple notes to a run.


1. In the menu bar, click **Runs ▶ Manage Runs**.
2. In the **Manage Runs** screen, place the pointer over the row of the run plan of interest, then click **Notes**.

Review the note or notes information.

Delete a run note

You can delete a run note in Genexus™ Software.

Notes record any written comments or observations for a run plan.

1. In the menu bar, click **Runs ▶ Manage Runs**.
2. In the **Manage Runs** screen, place the pointer over the row of the run plan of interest, then click **Notes**.
3. In the **Add Notes** pane, in the row of the note you want to delete, click  (**Delete**).

The note is no longer listed in the **Add Notes** dialog box and the note is removed from the software.

Click **Close** to return to the **Manage Samples** screen.



Genexus™ Software Dashboard

- View run progress on the instrument 88
- View the status of an instrument 90
- Information about connected instruments on the Dashboard 91

In the **Dashboard** screen you can view instrument status and details for recent and current runs.

The screenshot displays the Genexus Assay Development Software Dashboard. At the top, there is a navigation bar with 'Dashboard', 'Samples', 'Runs', 'Results', and 'Assays'. Below this, the 'Recent Runs' section (1) lists several runs, with 'Cpy_OCAv3_DnF_47' selected. The 'Instruments' section (2) shows two systems: 'GNXP-0017' and 'System1'. The 'System1' details (5) show a run 'Re_GenericHDRNA_assayPage' with a progress bar for 'Templating', 'Sequencing', 'Analysis', and 'Plugins'. The 'Results' section (4) shows assay metrics for 'CpyOCAv3DnF4516433' and 'SRTestSample01' through 'SRTestSample05'. A 'QC Status' indicator shows '5 Pass'.

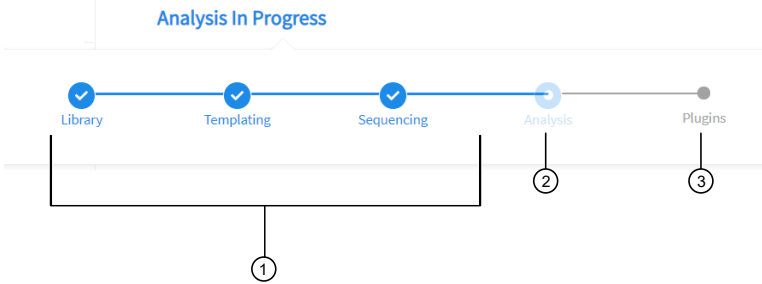
- ① The five most recent runs are listed in the left pane.
 - You can click the row of a run of interest to view details for the run in the right pane.
 - You can click the name of the run to leave the dashboard and view the results for the run.
- ② Instruments that are integrated with the software are shown.
- ③ In the details pane on the right, you can place the pointer over the status for the run to view the progress of the run.
- ④ For the recent run that is selected in the left pane, each assay for the run is shown. You can click the name of an assay to leave the dashboard and open the results for the run and view assay metrics.
- ⑤ For the recent run that is selected in the left pane, each sample in the run is shown. You can click a sample name to leave the dashboard and open the sample results.

View run progress on the instrument

You can view details for the five most recent and current runs in the **Dashboard** in Genexus™ Software. Runs that are reanalyzed are listed with ↻ (**Reanalysis**) after the run name. For more information, see “Reanalyze a run” on page 165.

1. In the menu bar, click **Dashboard**.
2. In the **Dashboard** screen, in the **Recent Runs** section, view information for recent runs.

Option	Description
View detailed run result information for completed runs.	In the left side of the screen, click the run name of interest. The Run Summary tab opens. For more information, see “The run summary” on page 109. In the menu bar, click Dashboard to return to the Dashboard .
View the quality control information.	In the left side of the screen, click the row of the run of interest. The QC status for the run and for the samples is shown in the details pane in the right side of the screen. For more information, see “QC results” on page 119.
View assay-level metrics and results for the run.	<ol style="list-style-type: none"> 1. In the left side of the screen, click the row of the run of interest. 2. In the run details pane in the right side of the screen, click the assay name of interest. <p>The Assay Metrics tab opens. For more information, see “Assay metrics and the run report” on page 114.</p> <p>In the menu bar, click Dashboard to return to the Dashboard.</p>
View the sample results.	<ol style="list-style-type: none"> 1. In the left side of the screen, click the row of the run of interest. 2. In the run details pane in the right side of the screen, click the sample name of interest. <p>The Key Findings tab opens. For more information, see “Key Findings” on page 123.</p> <p>In the menu bar, click Dashboard to return to the Dashboard.</p>



Option	Description
View the progress of the run.	<p>1. In the left side of the screen, click the row of the run of interest.</p> <p>2. In the run details pane in the right side of the screen, place the pointer over the status for the run.</p> <p>A progress bar for the run is shown. Steps in the run that are complete are listed with a check mark. Steps that are in progress are shown with a flashing circle. Steps that remain to be completed are listed in grey. If any failures occur, failed steps are also listed in grey.</p>  <p>① Completed steps are shown with a check mark. In this example, Library preparation, Templating, and Sequencing are complete.</p> <p>② Steps that are in progress are shown with a flashing circle. In this example, Analysis is in progress.</p> <p>③ Steps that remain to be completed are shown in grey. In this example, the Plugins step remains to be started after Analysis is complete.</p>

Tip: If the recent run includes many samples, limit the samples that are listed. For more information, see “Filter the list of samples shown in the dashboard” on page 90.


Filter the list of samples shown in the dashboard

You can apply filters to shorten the list of samples shown in the **Dashboard** in Genexus™ Software.

1. In the menu bar, click **Dashboard**.
2. In the **Dashboard** screen, in the **Recent Runs** section, click the row of the run of interest. Assays and samples in the run are listed in the details pane in the right side of the screen. In addition, the QC status is shown.
3. In the details pane, filter the list of samples.


Option	Description
Shorten the list of samples.	Click  (Filter), select the samples of interest, scroll to the bottom of the filter dialog box, then click Filter .  (Filter) changes color to indicate that a filter is applied.
Refine the samples shown by the QC status of the sample result.	In the details pane, in the QC Status section, click Pass to list only samples that passed QC metrics or click Fail to list only samples that failed QC metrics.

The list of samples is filtered according to the selections.


Click  **Clear Filters** to remove all filters and view the full list of samples.

View the status of an instrument

You can view the status of the Genexus™ Integrated Sequencer and any Genexus™ Purification Instrument that is integrated with Genexus™ Software.

1. In the menu bar, click **Dashboard**.
2. In the **Dashboard** screen, in the **Instruments** section, view the status of an instrument. The following information is provided for all integrated instruments.
 - instrument name
 - run status of the current or most recent run
 - PQ status
 -  (**Alarms**)– instrument alarms, if any
If the instrument displays one or more alarms, the number of alarms is shown in parentheses. You can place the pointer over **Alarms** to view more information about an alarm.
3. In the **Instruments** section, in an instrument pane, click **Details** to view the current or most recent run name, serial number, the operator, run start and completion times, and instrument alarms, if any.

Tip: Check the status of a purification instrument for **Run completed waiting user input** to know when you must perform an action to proceed through the run.

4. Click  **Refresh** to update the information.

Information about connected instruments on the Dashboard

The following information is available in the **Dashboard** screen. You can expand the **Details** section for each instrument to view more information.

Item	Description
Run	The name of the run that is in progress or the last completed run on the instrument.
Serial	The serial number of the instrument.
Operator	The name of the operator who started the run on the instrument.
Started	The date and time that the run started on the instrument.
Last Init	The last initialization completion date and time.
PQ Status	Indicates whether the performance qualification (PQ) status of the instrument is Verified or Expired .
Next PQ Due	The expiration date of the current PQ verification run. Expiration of the PQ status prevents the instrument from performing a non-PQ run until a valid PQ run has been performed and signed off.



Review data and results

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Use the **Results** menu to review results and data analysis and perform data management tasks. You can view results sorted by sample or by run.

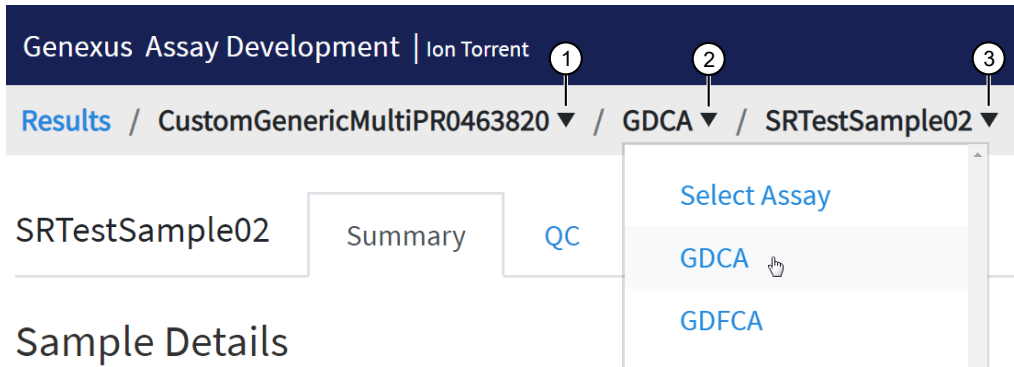
Selection	Description
Click Results ▶ Sample Results	Select this option to review completed sample results and reports.
Click Results ▶ Run Results	Select this option to review completed run results and reports by assay.
Click Results ▶ Verification Results	Select this option to review data from completed verification runs that were performed during sequencer installation or performance qualification.

Results navigation bar overview

After you select a run result or a sample result, a results navigation bar appears and allows you to easily toggle between different results screens for each run. You can quickly switch between run-level results, assay-level results, and sample-level results for the run.

Assay-level results include assay metrics, such as final read data. Assay metrics are also shown in the **Run Report** tab. For more information, see “Assay metrics and the run report” on page 114.

Samples that are run with the same assay in the same run share the same run report. All other results are specific to the sample and are shown in the sample results.










Callout	Dropdown menu	Description
1	Run name	The run name is listed. Multiple runs are listed only if the run has been reanalyzed. For more information, see “Reanalyze a run” on page 165.
2	Select Assay	<p>You can click the assay name of interest to view assay metrics for the run.</p> <p>You can toggle between different assays used in the run. If only one assay is associated with the run, only one assay name is listed.</p> <p>Some results from a plugin that is included in the run, such as the customer support archive plugin, apply to all of the samples for an assay in a run. In this case, the results from assay-level plugin are shown in the Plugins tab when you select an assay but do not select any samples for the run. For more information about plugins, see Chapter 10, “Plugins for data analysis”.</p> <p>You can select Select Assay from the dropdown menu to remove the assay selection. Removing the assay selection opens the Run Summary tab for the run selected in the Run Results dropdown menu.</p>
3	Select Sample	<p>You can click a sample name to view the sequencing results for the sample. You can click a different sample name to view other sample results for other samples in the run for the assay that is selected in Select Assay.</p> <p>For Sample to Result runs that include a positive control, results for the positive control are shown when you select Control Sample.</p> <p>You can select Select Sample from the dropdown menu to remove the sample selection. Removing the sample selection opens the Run Report tab for the assay selected in the Select Assay dropdown menu.</p>

Review sample results



In the **Results / Sample Results** screen, samples that have been sequenced are listed by sample name.

See “Find sample results” on page 95 for information about how to find sample results of interest.

The following information appears in the **Sample Results** screen.

Column	Description
Sample Name	The unique identifier created when the sample was entered into the software. Click the Sample Name to open the Sample Details screen for the sample. Use the tabs above the Sample Details to view the run summary, the run report, quality control, detailed variant results, and results for plugins that are associated with the selected assay, if any.
Sample Name followed by  (Signed) or  (Locked)	Manager- and administrator-level users can provide their electronic signature on sample results for completed runs. A sample name followed by  (Signed) indicates that the report for the run has been signed electronically. The signature information appears in the variant report PDF file. A sample name followed by  (Locked) indicates that the report for the run is locked. For more information, see “Sign off on the run results” on page 178.
Assay Name	The name of the assay used in the run plan.
Run Name	The unique name of the run given when it was created in the software.
Sample Status	The status of the run or sample (for example: Completed, Running, Failed, Terminated, Pending, Stalled).
QC Status	The QC status of a completed run. <ul style="list-style-type: none">  (Passed) indicates the sample passed all QC metrics.  (Failed) indicates the sample failed a QC metric.  (Not Calculated) indicates a sample did not undergo QC analysis.
Started On	The date and time when the run was started.
Updated On	The date and time when the last action was completed on the run.
Tags	Tags that are assigned to the sample are listed.

You can also perform the following actions in the **Sample Results** screen.

Option	Description
Actions	<p>Available action links for a sample are shown when you place the pointer over the row of a sample.</p> <ul style="list-style-type: none"> • Report—Download the report (available only for samples with a sample status of completed). For more information, see “Variant report” on page 175. • Assign Tags—Add or remove the tags assigned to a sample result. For more information, see “Sample tags” on page 46. • Audit—View the audit trail for the run. For more information, see “Review and download a run history” on page 83. • Notes—View, add, or delete notes for sample result. • CSA—Download customer support archive (CSA) log files for the run to help with troubleshooting. For more information, see “Download a customer support archive” on page 371. • Reanalyze—Reanalyze a sample from the alignment step when a sample or reanalysis successfully completes the basecalling step. For more information, see “Reanalyze a sample” on page 167. • Variant Audit—View the history of variant classifications.
Actions that you can perform when you select samples	<p>More actions are shown when you select one or more samples.</p> <ul style="list-style-type: none"> • Assign Tags—Assign a tag or tags to selected sample results in one action. The  Assign Tags command appears above the Sample Results table after you select one or more samples. For more information, see “Assign sample tags to multiple sample results” on page 100. • Compare—Compare variant results between multiple samples, or from a single sample over time. The  Compare command is not shown until you select at least two samples. For more information, see “Compare sample results” on page 153.

Find sample results

You can sort, search, and filter information to narrow the sample results that are listed in the **Results / Sample Results** screen. The sort, search, and filter tools help you organize sample results and find a specific sample result in Genexus™ Software.

For information to search for a sample result by a unique identifier, such as the name of a run or the name of a sample, see “Search for sample results” on page 96.

For information to filter sample results by a common trait, such as QC status, see “Filter the list of sample results” on page 98.



Search for sample results

Sample results are listed in the **Results / Sample Results** screen in Genexus™ Software. The most recent sample result is on the top of the list.

You can search for sample results with keywords or text that are included in the name of the sample, run name, or the name of an assay applied to the run.

IMPORTANT! Search terms must adhere to the following rules.

- An asterisk (*) is not allowed in the search field for use as a wildcard.
- Searches are case-insensitive, that is, both upper-case and lower-cases letters are found regardless the case of search term letters.
- Searches find every occurrence of a continuous string. For example, a search for *demo* in assay names returns a list of all assays with a name that includes *demo*. For example, demo1, demo2, and so on.
- Spaces are removed during searches, and are therefore not recommended. For example, a search for *demo 1* returns results only for data that includes the string *demo1*.

-
1. In the menu bar, click **Results ▶ Sample Results**.
 2. In the **Sample Results** screen, in the **Sample Name**, **Run Name**, or **Assay Name** column header, click  (**Filter**).
 - a. In the search field, enter a full or partial assay, sample, or run name of at least 3 characters. When you have entered at least 3 characters, the filter dialog box limits the list in the filter dialog box based on the entry.
 - b. In the filter dialog box, select from the limited list.
 - Select a checkbox next to a sample, run, or assay name to select one or more items.
 - Click **Select All** to select all items listed in the filter dialog box.
-
- Note:** If you click **Clear**, all selections are removed and the filter dialog box closes.
-
- c. In the filter dialog box, click **Filter**. In the column header,  (**Filter**) changes color to indicate that a filter is applied.

The filtered list of sample results is shown in the **Sample Results** screen.

Click  **Clear Filters** to remove all filters and view the full list of sample results.

Sort the list of sample results

You can sort the list of sample results to make it easier to find sample results in Genexus™ Software.

By default, sample results are listed in the **Results / Sample Results** screen by date. The most recent sample result is at the top of the list.

You can change the order that the sample results are listed in the table.





1. In the menu bar, click **Results ▶ Sample Results**.
2. In the **Sample Results** screen, click a column header of interest.
The list of sample results is sorted in alphabetical or chronological order depending on the entries in the column.
3. Click the column header again to reverse the order.

Filter the list of sample results

You can apply filters to narrow search results, or shorten the list of sample results when you view the list of sample results in Genexus™ Software.

1. In the menu bar, click **Results** ▶ **Sample Results**.
2. In the **Sample Results** screen, select one or more filters.

Option	Description
Filter and search by the name of the sample, the run name or the assay selected in the run.	For more information, see “Search for sample results” on page 96.
Filter by the status of the sample result.	<ol style="list-style-type: none"> 1. In the Sample Status column header, click ▼ (Filter) then select at least one status. <ul style="list-style-type: none"> • Completed • Running • Failed • Terminated • Stalled • Aborted • Pending 2. Click Filter. 3. Records are listed for sample results that meet the sample status criteria.
Filter by the QC status.	<ol style="list-style-type: none"> 1. In the QC Status column header, click ▼ (Filter) then select at least one QC status. <ul style="list-style-type: none"> • Failed • Passed • Not Calculated 2. Click Filter. <p>Records are listed for sample results that meet the QC status criteria.</p>
List only sample results from runs that were started on or updated on an exact date.	<ol style="list-style-type: none"> 1. In the Started On or Updated On column header, click ▼ (Filter). 2. In the first dropdown list, select Is equal to. 3. In the calendar that appears, select the date of interest. 4. Click Filter. <p>Records are listed for sample results from runs that were started on or updated on the date entered.</p>

Option	Description
<p>List only sample results from runs that were started on or updated within a date range.</p>	<ol style="list-style-type: none"> 1. In the Started On or Updated On column header, click  (Filter). 2. In the first dropdown list, select Is after or equal to. 3. In the calendar that appears, select the earliest date of the range. 4. In the Boolean operator dropdown list, select And. 5. In the second dropdown list, select Is before or equal to. 6. In the calendar that appears, select the latest date of the range. 7. Click Filter. <p>Records are listed for sample results from runs started on or updated on dates that meet both conditions.</p>
<p>List sample results according to a date that meets either one condition or a second condition</p>	<ol style="list-style-type: none"> 1. In the Started On or Updated On column header, click  (Filter). 2. In the first dropdown list, select Is after or equal to. 3. In the calendar that appears, select the earliest date for the sample results of interest. 4. In the Boolean operator dropdown list, select Or. 5. In the second dropdown list, select Is before or equal to. 6. In the calendar that appears, select the latest date for the sample results of interest. 7. Click Filter. <p>Records are listed for sample results from runs that were started on or updated on dates that meet at least one of the conditions.</p>
<p>List only sample results for runs that were started on or before or updated on or before a date that you indicate.</p>	<ol style="list-style-type: none"> 1. In the Started On or Updated On column header, click  (Filter). 2. In the first dropdown list, select Is before or equal to. 3. In the calendar that appears, select the date of interest. 4. Click Filter. <p>Records are listed for sample results from runs that were started on or before the date entered.</p>
<p>List only sample results for runs that were started on or after or updated on or after a date that you indicate.</p>	<ol style="list-style-type: none"> 1. In the Started On or Updated On column header, click  (Filter). 2. In the first dropdown list, select Is after or equal to. 3. In the calendar that appears, select the date of interest. 4. Click Filter. <p>Records are listed for sample results from runs that were started on or after, or updated on or after, the date entered.</p>

In the column header,  **(Filter)** changes color to indicate that a filter is applied.

Click  **Clear Filters** to remove all filters and view the full list of sample results.

Assign sample tags to a sample result



You can assign tags to sample results in Genexus™ Software to label and group sample results. For more information, see “Sample tags” on page 46.

1. In the menu bar, click **Results ▶ Sample Results**.
2. In the **Sample Results** screen, place the pointer over the row of the sample result that you want to tag, then click **Assign Tags**.
3. In the **Assign Tags** dialog box, search for or scroll to a tag.
If the tag that you want to assign to a sample does not exist in the software, a manager-or administrator-level user can create a new sample tag. For more information, see “Create a sample tag (manager/administrator)” on page 46.
4. Select one or more tags that you want to assign to the sample result, then click **Save**.

The tag or tags are assigned to the sample result and are listed in the row for the sample result in the list of sample results.

Assign sample tags to multiple sample results

You can assign tags to multiple sample results at once in Genexus™ Software. For more information about sample tags, see “Sample tags” on page 46.

1. In the menu bar, click **Results ▶ Sample Results**.
2. In the **Sample Results** screen, click the checkbox to the left of each sample result of interest, then click  **Assign Tags** above the list of sample results.
 **Assign Tags** is not shown until you select at least one sample result.
3. In the **Assign Tags** dialog box, search for or scroll to a tag.
If the tag that you want to assign to a sample result does not exist in the software, a manager-or administrator-level user can create a new sample tag. For more information, see “Create a sample tag (manager/administrator)” on page 46.
4. Select one or more tags that you want to assign to the sample results, then click **Save**.

The tag or tags are assigned to the selected sample results and are listed in the row for each sample result in the list of sample results.

Review run results

In the **Results / Run Results** screen, runs that are pending, in progress, or completed are listed. Runs with a status of failed, aborted, or stalled are also listed.

See “Find a run result” on page 102 for information about how to find run results of interest.

The following run information appears in the **Results / Run Results** screen.

Column	Description
Run Name	The unique name of the run given when it was created in the software. Click a run name to open the Run Summary . For more information, see “The run summary” on page 109. Runs that are reanalyzed are listed with ↻ (Reanalysis) after the run name. For more information, see “Reanalyze a run” on page 165.
Assay Name	The name of the assay selected for the run. You can view the Assay Name and corresponding Assay Full Name for all assays in the Assays ▶ Manage Assays screen.
Run Status	The status of the run. For example, Analysis In Progress , Executing Plugins , Analysis Completed , Terminated , Archival: In Progress , Purification In Progress , or Purification Completed . For purification runs that have a status of Purification In Progress , Purification Complete , or for failed or aborted purification runs, you can place the pointer over the shaded number, in the Status column to view the status of each purification batch for the run. The shaded number, such as 1 or 2 , represents a purification batch for the run. For example, a status can be 1 Completed , 2 Started , and 3 Planned , which indicates that the first batch is complete, the second batch has started, and the third purification batch has not yet started, but is planned. For more information about purification batches, see “Integrated nucleic acid isolation in Sample to Result runs” on page 65.
Total Samples	The total number of samples in a run.
PCR Plate Number	A unique identifier for the 96-well plate used for library preparation and templating. For more information, see “Assign PCR Plate” on page 113.
Started On	The date and time when the run was started.
Updated On	The date and time when the last action was completed on the run.

You can also perform the following actions in the **Results / Run Results** screen.

Option	Description
Actions ^[1, 2]	<p>Available action links for a run are shown when you place the pointer over the row of a run.</p> <ul style="list-style-type: none"> • Delete—Delete the run. • BAM Uploader—Upload run information to another Genexus™ Software server or to Ion Reporter™ Software for further analysis. For more information, see “Upload results files to another Genexus™ Software server” on page 173 and “Upload results files to Ion Reporter™ Software” on page 171. • Audit—View the audit trail for the run. For more information, see “Review and download a run history” on page 83. • CSA—Download customer support archive (CSA) log files for the run to help with troubleshooting. For more information, see “Download a customer support archive” on page 371. • Assign PCR Plate—Enter a unique identifier for the 96-well plate used for library preparation and templating. For more information, see “Assign PCR Plate” on page 113.
Actions ^[3]	<ul style="list-style-type: none"> • View Plan—View detailed run plan information. For more information, see “Run plan details” on page 81. • Review—Review samples that do not have a concentration within a specified threshold after purification, but before library preparation. For more information, see “Review samples for Sample to Result runs” on page 70. • Abort—Allows you to abort a run after purification, but before sequencing. This action is available when the run status is Purification Review Required, or when the run status is Purification Completed and some purification samples have been excluded from sequencing.

^[1] The actions that are available depend on the type of run.

^[2] **BAM Uploader** is not available for BAM run results or for archived runs in which BAM files have been removed.

^[3] Actions that are available for Sample to Result runs only.

Find a run result

You can sort, search, and filter information to narrow the run results that are listed in the **Results / Run Results** screen. The sort, search, and filter tools help you organize run results and find a specific run result in Genexus™ Software.

For information to search for a run result by a unique identifier, such as the name of a run, see “Search for run results” on page 103.

For information to filter run results by a common trait, such as run results that have a status of analysis completed, see “Filter the list of run results” on page 105.


Search for run results

Run results are listed in the **Results / Run Results** screen in Genexus™ Software. The most recent run result is on the top of the list.

You can search for run results with keywords or text that are included in the name of the run name, the name of an assay applied to the run, or by the PCR Plate number. For more information about how to use the PCR Plate number to track libraries, see “Assign PCR Plate” on page 113.

IMPORTANT! Search terms must adhere to the following rules.

- An asterisk (*) is not allowed in the search field for use as a wildcard.
- Searches are case-insensitive, that is, both upper-case and lower-cases letters are found regardless the case of search term letters.
- Searches find every occurrence of a continuous string. For example, a search for *demo* in assay names returns a list of all assays with a name that includes *demo*. For example, demo1, demo2, and so on.
- Spaces are removed during searches, and are therefore not recommended. For example, a search for *demo 1* returns results only for data that includes the string *demo1*.

-
1. In the menu bar, click **Results ▶ Run Results**.
 2. In the **Run Results** screen, in the **Run Name**, **Assay Name**, or **PCR Plate Number** column header, click  (**Filter**).
 - a. In the search field, enter a full or partial assay or run name or PCR plate number of at least 3 characters.

When you have entered at least 3 characters, the filter dialog box limits the list in the filter dialog box based on the entry.
 - b. In the filter dialog box, select from the limited list.
 - Select a checkbox next to a run or assay name to select one or more items.
 - Click **Select All** to select all items listed in the filter dialog box.

Note: If you click **Clear**, all selections are removed and the filter dialog box closes.

- c. In the filter dialog box, click **Filter**.

In the column header,  (**Filter**) changes color to indicate that a filter is applied.

The filtered list of run results is shown in the **Run Results** screen.

Click  **Clear Filters** to remove all filters and view the full list of run results.

Sort the list of run results

You can sort the list of run results to make it easier to find a run result of interest in Genexus™ Software.

By default, run results are listed in the **Runs / Run Results** screen by date. The most recent run result is at the top of the list.

You can change the order that the run results are listed and sort the run results alphabetically or reverse alphabetically.

1. In the menu bar, click **Runs ▶ Run Results**.
2. In the **Run Results** screen, click the **Run Name** heading.
The list of run results is sorted alphabetically.
3. Click the column header again to reverse the order.

Filter the list of run results

You can apply filters to narrow search results, or shorten the list of run results when you view the list of run results in Genexus™ Software.

1. In the menu bar, click **Runs ▶ Run Results**.
2. In the **Run Results** screen, select one or more filters.

Option	Description
Filter and search by the run name or the assay selected in the run.	For more information, see “Search for run results” on page 103.
Filter by the status of the run result.	<ol style="list-style-type: none"> 1. In the Run Status column header, click ▼ (Filter) then select at least one status. <ul style="list-style-type: none"> • Analysis Completed • Analysis In Progress • Analysis Failed • Analysis Aborted • Analysis Stalled • User Aborted • Run Aborted • Purification Completed • Purification Run Aborted • Purification Failed • Purification Review Required • Purification In Progress • Selected on Purification Instrument • Backup In Progress • Executing Plugins • Library Preparation Aborted • Library Preparation Completed • Library Preparation Started • Selected on Instrument • Sequencing Completed • Sequencing Started • Sequencing Aborted • Templating Aborted • Templating Completed • Templating Started • Kit Barcodes Required 2. Click Filter.

Option	Description
	Records are listed for run results that meet the run status criteria.
Filter by the number of samples in the run.	In the Total Samples column header, click ▼ (Filter) then use the options to filter the list in the filter dialog box based on the number or span of numbers of samples in the run.
List only run results from runs that were started on or updated on an exact date.	<ol style="list-style-type: none"> 1. In the Started On or Updated On column header, click ▼ (Filter). 2. In the first dropdown list, select Is equal to. 3. In the calendar that appears, select the date of interest. 4. Click Filter. <p>Records are listed for run results that were started on or updated on the date entered.</p>
List only run results from runs that were started on or updated within a date range.	<ol style="list-style-type: none"> 1. In the Started On or Updated On column header, click ▼ (Filter). 2. In the first dropdown list, select Is after or equal to. 3. In the calendar that appears, select the earliest date of the range. 4. In the Boolean operator dropdown list, select And. 5. In the second dropdown list, select Is before or equal to. 6. In the calendar that appears, select the latest date of the range. 7. Click Filter. <p>Records are listed for sample results started on or updated on dates that meet both conditions.</p>
List run results according to a date that meets either one condition or a second condition.	<ol style="list-style-type: none"> 1. In the Started On or Updated On column header, click ▼ (Filter). 2. In the first dropdown list, select Is after or equal to. 3. In the calendar that appears, select the earliest date for the sample results of interest. 4. In the Boolean operator dropdown list, select Or. 5. In the second dropdown list, select Is before or equal to. 6. In the calendar that appears, select the latest date for the sample results of interest. 7. Click Filter. <p>Records are listed for sample results for runs that were started on or updated on dates that meet at least one of the conditions.</p>
List only run results for runs that were started on or before or updated on or before a date that you indicate.	<ol style="list-style-type: none"> 1. In the Started On or Updated On column header, click ▼ (Filter). 2. In the first dropdown list, select Is before or equal to. 3. In the calendar that appears, select the date of interest. 4. Click Filter. <p>Records are listed for run results that were started on or before the date entered.</p>
List only run results for runs that were started on or after, or updated on	<ol style="list-style-type: none"> 1. In the Started On or Updated On column header, click ▼ (Filter). 2. In the first dropdown list, select Is after or equal to.

Option	Description
or after, a date that you indicate.	<p>3. In the calendar that appears, select the date of interest.</p> <p>4. Click Filter.</p> <p>Records are listed for runs that were started on or after, or updated on or after, the date entered.</p>

In the column header,  (**Filter**) changes color to indicate that a filter is applied.

Click  **Clear Filters** to remove all filters and view the full list of run results.

View the run summary

The run summary provides an overview of the run. The information that is displayed includes the name of the assay used in the run, sample locations, information about the reagents used in the run, primer tube positions, and instrument information. Metrics from sample purification are also provided, if applicable.

1. In the menu bar, click **Results ▶ Run Results**.
2. In the **Run Name** column, click the run name of interest.
The **Run Summary** tab opens.
3. Review the run summary.

Action	Procedure
View the assay metrics.	In the Assays section, click the assay name of interest.
View the sample locations in an image of a 96-well sample plate.	In the Sample Locations section, click PCR Plate View .
Reanalyze a run with a new assay.	In the upper right corner of the screen, click ⋮ (More Options) ▶ Reanalyze . For more information, see “Reanalyze a run” on page 165.
Run plugins on the sequencing data after a sequencing run is complete.	In the upper right corner of the screen, click ⋮ (More Options) ▶ Run Plugin . For more information, see “Run plugins after a sequencing run is complete” on page 165.
Download customer support archive (CSA) log files for the run to help with troubleshooting.	In the upper right corner of the screen, click ⋮ (More Options) ▶ CSA . For more information, see “Customer Support Archive (CSA)” on page 371.
Upload results to another Genexus™ Software server or to Ion Reporter™ Software for further analysis.	In the upper right corner of the screen, click ⋮ (More Options) ▶ BAM Uploader . For more information, see “Upload results files to another Genexus™ Software server” on page 173 and “Upload results files to Ion Reporter™ Software” on page 171.
View the history of variant classifications. ^[1]	In the upper right corner of the screen, click ⋮ (More Options) ▶ Variant Audit .
View the run report.	Click the Run Report tab.

^[1] This information appears only for results that include variant classifications.

The run summary

The run summary displays run information, assays used in the run, sample locations, metrics from sample purification, information about the reagents used in the run, primer tube positions, and instrument information.

Depending on the type of the run and the selections made in the run plan, the run information can include the following items.

Run information

Item	Description
Run Name	The name of the run.
Started On	The date and time when the run was started.
Completed On	The date and time when the run was completed.
Run Status	The status of the run.
Report Template	If the option to generate a report was enabled in the run plan, the report template used is listed.
Chip Type	The semiconductor sequencing chip used in the run, such as the Ion Torrent™ GX5™ Chip.
Library Chemistry	The type of library chemistry used in the run, such as Ion AmpliSeq™ HD.
Server	The Ion Reporter™ Software account or the target Genexus™ Software server and version, if any, that was selected for uploading BAM files when the run is complete.

Sequencing instrument information

Item	Description
Instrument Name	The name of the sequencer.
Instrument Serial Number	The serial number of the sequencer.
Status	The status of the sequencing portion of the run.
Operator	The name of the person who was signed in the Genexus™ Integrated Sequencer when the sequencing run was performed.
Start Date	The date and time when the run was started on the sequencer.
Completion Date	The date and time when the run was completed on the sequencer.
Sequencing Flows	The number of flows performed by the sequencer.

Purification instrument information

Item	Description
Purif. Instrument	The name of the purification instrument.
Instrument Serial Number	The serial number of the purification instrument.
Status	The status of the purification portion of the run for the batch.
Operator	The name of the person who was signed in the Genexus™ Purification Instrument when purification was performed.
Start Date	The date and time when the batch was started on the purification instrument.
Completion Date	The date and time when the batch was completed on the purification instrument.
Purif. Batch	The nucleic acid isolation batch.

Assays table

Item	Description
Assay Full Name	The name of the assay used in the run. Click the assay name to display the details of the assay.
Assay Name	An abbreviated name of the assay or assays used in the run.
Analysis Version	The version of the assay used for analysis.
Research Application	The research application for the assay, such as DNA or DNA and Fusions .
Lane	The chip lane or lanes used in the sequencing run for the assay.
Total Samples	The total number of samples sequenced for the assay. A single sample can correlate with multiple wells on the plate for some assays when multiple nucleic acid types are contained within the sample.

Purification Samples table

Item	Description
Batch ID	When the run includes more than one purification batch, the Batch ID is listed. The Batch ID , such as 1 or 2, indicates the purification group for each sample.
Sample Name	The unique identifier created when the sample was entered into the software.
Sample Type	A term that describes the sample, for example, FFPE or Blood (Plasma).
Nucleic Acid Type	DNA, RNA, or DNA+RNA.

Purification Samples table (continued)

Item	Description
Conc. (ng/μl)	The concentration of the sample measured by the purification instrument. In some instances, such as when purification is in progress or when the sample is a no-template control, the values is listed as N/A. If the concentration is not within the QC concentration range specified by the assay, a Quantity Not Sufficient (QNS) alert is displayed.
QC Conc. Range (ng/μl)	The QC concentration range for the assay.
Batch Status	The run status of a purification batch. The status can be N/A, Planned, Review, Started, Aborted, or Completed.
Archive Position	The sample positions in the archive plate. Sample positions can be in rows A to D and columns 1 to 8.
Library Prep	An indicator of whether the sample is selected for sequencing. <ul style="list-style-type: none"> ✔ – indicates that the sample is selected for sequencing. ⊘ – indicates that the sample is not selected for sequencing. N/A – indicates that purification has not started or is in progress.

Sample Locations table

Item	Description
PCR Plate View	Click to view the sample locations in an image of a 96-well sample plate
Well Pos.	The well position indicates the location of the sample on the plate.
Sample Name	The unique identifier created when the sample was entered into the software.
Nucleic Acid Type	The sample nucleic acid type, such as DNA, RNA, or TNA.
Vol. (μl)	The volume of the sample.
Conc. (ng/μl)	The concentration of the sample.
Dilution Factor	The dilution factor of the sample.
Kit Barcodes	The barcode for the kit used for nucleic acid extraction, if applicable.
Barcode	The name of the barcode adapter or adapters that are associated with the sample, for example, IonHDdual_0101.
Assay Name	The assay that was used to sequence the sample for the indicated well position.
Library Position	The plate location of libraries. For completed runs that were planned using samples, the Library Position indicates the location of unused libraries that can be sequenced by planning and initiating a library run.

Reagents table

Item	Description
Consumable	Consumables used in the run, such as the PanelKit and LibraryKit , are listed.
Barcode	The consumable barcode, if applicable.
Part #	The consumable part number, if applicable.
Expiration Date	The consumable expiration date, if applicable.
Lot #	The consumable lot number, if applicable.

The **Primer Tube Positions** table shows an image that indicates the locations of the primer pool tubes.

Retain run results

You can retain run results in Genexus™ Software. When you retain a run result, run data, including the run result, .wells files, and intermediate files, are retained and cannot be deleted. Users with any level of permissions can retain run results. When a run result is retained, it overrides the automatic deletion settings in the software. For more information, see “Set a schedule to permanently delete files (administrator)” on page 277.

1. In the menu bar, click **Results ▶ Run Results**.
2. In the **Run Results** screen, select the checkbox in the row of one or more run results that you want to retain.
3. Click **Retain Data**.

Run results that have been retained are denoted with after the run name in the **Run Results** screen. The run result is retained in the system and cannot be deleted. For information to revert the run to a run that is not retained, see “Release run results” on page 112.

Release run results

Only run results that have been retained in Genexus™ Software can be released. For more information, see “Retain run results” on page 112.

When you release run results, you can delete the run results. The run results, the associated .wells files, and intermediate files are available to be auto-deleted according to system settings. For more information, see “Set a schedule to permanently delete files (administrator)” on page 277.

1. In the menu bar, click **Results ▶ Run Results**.
2. In the **Run Results** screen, click **Release Data** in the row of the run of interest.
Run results that have been retained are denoted with (retained) after the run name.
3. Click **Yes** to confirm that you want to proceed with the data release.

The run result is released. The run result can be deleted or auto-deleted.

Assign PCR Plate

Genexus™ Software allows you to track and associate a run with the PCR plate used in the run. The PCR plate is the 96-well plate that is used for library preparation and templating. You can assign a unique identifier, a **PCR Plate Number**, to runs that have a status of **Library Preparation Completed**, **Sequencing Completed**, or **Run Completed**. The PCR plate number that you enter is shown in the **Run Results** screen and if needed, can help you track libraries and troubleshoot. Later, if you sequence the remaining libraries in a different sequencing run and assign the PCR plate number to the run, you can easily search for and find all run results associated with the libraries in the PCR plate.

For more information to find a run with a search of the PCR Plate Number, see “Search for run results” on page 103.

1. In the menu bar, click **Results ▶ Run Results**.
2. In the **Run Results** screen, place the pointer over the row of a run of interest, then click **Assign PCR Plate number**.
3. In the **Assign PCR Plate Number** dialog box, confirm, edit, or enter the **PCR Plate Number**.
The PCR plate number must be between 1 and 10 characters. Only alphanumeric characters (numbers 0 to 9 and letters A to Z), period (.), underscore (_), or hyphen (-) are allowed. Spaces are not allowed.
4. Click **Submit** to associate the PCR plate with the run.

View assay metrics and the run report

The run report provides detailed information about a run, such as the **Total Bases** and **Final Reads**. For runs with multiple assays, metrics are provided for each assay in the run. The run report contains the assay metrics for all assays in the run.

For a runs with only one assay, the run report and assay metrics show the same information for assay metrics. In addition to the metrics for the assay, the run report shows information about the run and the instruments used in the run.

1. Click **Results ▶ Run Results** to view sequencing results, including variant and fusion calls, for a particular sample.
2. In the **Run Name** column, click the run name of interest.

Action	Procedure
View the run report	Click the Run Report tab.
View the assay metrics for a single assay	In the Select Assay dropdown list at the top of the screen, select the assay of interest.

For more information, see “Assay metrics and the run report” on page 114.

Assay metrics and the run report

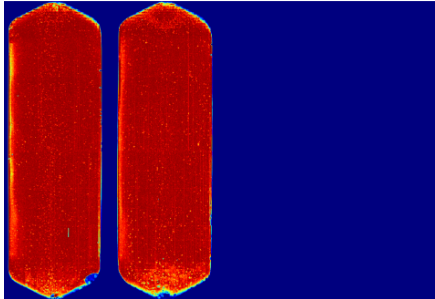
The run report provides detailed information, such as various chip metrics for the run, and well and Ion Sphere™ Particles (ISPs) statistics. For runs with multiple assays, metrics are provided for each assay in the run. Sequencing metrics are shown at the top of the screen, followed by sample-specific metrics in the **Run Samples** table. Read data for individual samples for the assay are listed in the **Run Samples** table.

Barcode-specific metrics for barcodes that are included in the run are listed in the **Barcodes With Reads Reported** table, which follows the **Run Samples** table. The CSA file for the run contains information for barcodes that are not assigned to samples in the run. Information in the CSA file can help you troubleshoot results, if needed. For more information, see “Download a customer support archive” on page 371.

Run Report


Metric	Description
Loading	The number and percentage of total addressable wells on the chip that contain an ISP.
Enrichment	The number and percentage of wells ISPs that contain live ISPs.
Library	The number and percentage of wells with live ISPs that contain Library ISPs.
Final Reads	Library reads passing all filters that are recorded in the output BAM files. This value can be different from the total number of reads due to technicalities associated with read trimming beyond a minimal requirement.
Total Bases	The number of filtered and trimmed base pairs that are reported in the output BAM file.
Raw Read Accuracy	The raw read accuracy across each individual base position in a read calculated as, $(1 - [\text{total errors in the sequenced reads}] / [\text{total bases sequenced}]) \times 100$. Raw read accuracy is measured at each base across the length of the read and is based on 1x sequencing coverage; raw read accuracy is <i>not</i> based on consensus accuracy across multiple reads for the same base position.
Wells with ISPs	The number of wells that contain an ISP.
Unenriched	The number and percentage of wells with ISPs that do not contain live ISPs.
Total Addressable Wells	Wells on the chip that can be physically reached by a library.
Empty Wells	The percentage of total addressable wells on the chip that do not contain an ISP.
Wells with Live ISPs	Loaded wells with ISPs with a signal of sufficient strength and composition to be associated with the library or control fragment key.
Wells with Library ISPs	Loaded wells with live ISPs with a key signal that is identical to the library key signal.
Control ISPs	Loaded wells with live ISPs with a key signal that is identical to the control fragment key signal.
Filtered out	The total percentage of filtered reads, or the sum of the percentages of polyclonal, low quality, and adapter dimer reads.

Run Report (continued)

Metric	Description
Polyclonal	<p>Wells with a live ISP that carries clones from two or more templates.</p> <p>To view polyclonal metrics, mouse over the first low quality portion (gray) of the Final Reads bar plot.</p> <p>Final Reads 58.39% </p> <p>30,735,120</p> <p>Filtered out 41.61%</p> <p>Final Reads(30,735,120) / Wells with Library ISPs(52,635,124)</p> <p>Polyclonal: 15935106 (30.27%)</p>
Low Quality	<p>Loaded wells with a low or unrecognizable signal.</p> <p>To view polyclonal metrics, mouse over the second low quality portion (gray) of the Final Reads bar plot.</p> <p>Final Reads 58.39% </p> <p>30,735,120</p> <p>Filtered out 41.61%</p> <p>Final Reads(30,735,120) / Wells with Library ISPs(52,635,124)</p> <p>Low Quality: 3208258 (6.1%)</p>
Adapter Dimer	<p>Loaded wells with a library template of an insert size less than 8 bases.</p> <p>To view adapter dimer metrics, mouse over the lightest gray portion of the Final Reads bar plot.</p> <p>Final Reads 43.57% </p> <p>52,494,934</p> <p>Filtered out 56.44%</p> <p>Final Reads(52,494,934) / Wells with Library ISPs(120,492,758)</p> <p>Adapter Dimer: 10862794 (9.02%)</p> <p>Note: In assays using Ion AmpliSeq™ HD library chemistry, adapter dimer reads represent a small proportion of total reads and can be seen by hovering the pointer over the right end of the Final Reads bar.</p>
Loading Density	<p>A visual representation of chip loading. Red color indicates areas of higher density of loading. Blue color indicates areas of lower density of loading. The following example shows a sequencing experiment where two lanes on the chip are uniformly loaded with ISPs.</p> <p>Loading Density</p> 


Run Samples

The Run Samples table lists read data for each individual sample in the assay.

Column	Description
Sample Name	The unique identifier created when the sample was entered in the software.
Nucleic Acid Type	The sample nucleic acid type, such as DNA, RNA, or TNA.
Barcode	The unique identifiers of the dual barcode pair assigned to the DNA and/or RNA library for a sample.
Total Reads	The total number of filtered and trimmed reads with the listed dual barcodes assigned to the sample.
Mean Read Length	The average length, in base pairs, of usable library reads for each sample.
≥Q20 Bases	The total number of called bases that have ≥99% accuracy (or less than 1% error rate) aligned to the reference for the sample.
Mapped Reads	The number of reads that are mapped to the reference file.
On Target Reads	The percentage of sequencing reads mapped to any target region of the reference.
Mean Depth	The average number of reads of all targeted reference bases.
Uniformity	The percentage of bases in all targeted regions (or whole genome) with a depth of coverage ≥20% of the mean read coverage.
Read Length Histogram	A histogram that presents all filtered and trimmed library reads that are reported in the output BAM file (Y-axis) and the mean read length in base pairs (X-axis). 


Barcodes with Reads Reported

The Barcodes with Reads Reported table lists barcode-specific metrics

Column	Description
Barcode	The unique identifiers of the dual barcode pair assigned to the DNA and/or RNA library for a sample.
Total Reads	The total number of filtered and trimmed reads with the listed dual barcodes assigned to the sample. The reads are independent of length reported in the output BAM file.
Mean Read Length	The average length, in base pairs, of usable library reads for each sample.
≥Q20 Bases	The total number of called bases that have ≥99% accuracy (or less than 1% error rate) aligned to the reference for the sample.
Read Length Histogram	A histogram that presents all filtered and trimmed library reads that are reported in the output BAM file (Y-axis) and the mean read length in base pairs (X-axis). 

Download information about run samples

Information about each sample in a run, such as the barcode for the sample and the total reads, are shown in the run samples table. You can download the information shown in the run samples table.


1. In the menu bar, click **Results ▶ Run Results**.
2. In the **Run Results** screen, in the **Run Name** column, click the run name of interest.
3. Click the **Run Report** tab, then scroll to the **Run Samples** table.
4. In the upper right corner above the **Run Samples** table, click  **Export to Excel**.

An XLSX file that contains the information shown in the run samples table is downloaded.

Read length histograms are not included in the file.

Download reads reported by barcodes

Metrics for the barcodes that are included in a run are listed in the barcodes with reads reported table. You can download the information shown in the barcodes with reads reported table.

1. In the menu bar, click **Results ▶ Run Results**.
2. In the **Run Results** screen, in the **Run Name** column, click the run name of interest.
3. Click the **Run Report** tab, then scroll to the **Barcodes with Reads Reported** table.
4. In the upper right corner above the **Barcodes with Reads Reported** table, click  **Export to Excel**.

An XLSX file that contains the information shown in the barcodes with reads reported table is downloaded.

Read length histograms are not included in the file.

View sequencing results

1. Click **Results** ▶ **Sample Results** to view sequencing results, including variant and fusion calls, for a particular sample.
2. In the **Sample Name** column, click a sample name.
3. In the **Results** screen, click the tabs to view the different types of sample-specific results and data.

Tab	Description
QC	The quality metrics for the sample sequenced in the run. For more information, see “QC results” on page 119.
Key Findings	An overview of the results for the sample, including Sample Details , Key Metrics , Genes , and Coverage graphs. For more information, see “Key Findings” on page 123.
Variants	Detailed variant results for SNVs/Indels , Fusions , and CNVs . For more information, see “View variant results” on page 128.
Plugins	Results generated from the plugins associated with the assay used to analyze the sequenced sample. For more information, see Chapter 10, “Plugins for data analysis”.
Reports	You can download and generate summaries of run results. There are two types of reports: <ul style="list-style-type: none"> • Run reports – include assay metrics and the record of reagents that were used in a run. • Variant reports – include the variant results for each sample in a sequencing run, reagents used, and QC evaluation metrics. For more information, see Chapter 7, “Create, view, and sign reports”.

View the QC results

You can view the quality control (QC) metrics for each sample that was sequenced in a run in Genexus™ Software.




1. In the menu bar, click **Results** ▶ **Sample Results**.
2. In the **Sample Name** column, click the sample name of interest.
3. Click the **QC** tab.
The QC metrics are shown.

Review the QC data. For more information, see “QC results” on page 119.

QC results

The quality control (QC) metrics for each sample that was sequenced in a run are displayed in Genexus™ Software.

If a sample fails a single test metric, the sample fails that QC test. A sample must meet all QC parameter thresholds of a particular QC test in order to pass. The QC status is divided into the following categories.

-  (Passed) indicates the sample passed all QC metrics.
-  (Failed) indicates the sample failed a QC metric.
-  (Not Calculated) indicates a sample did not undergo QC analysis.

Note: If a sample fails to meet one or more QC parameters, you can reanalyze a run after adjusting the parameter or parameters. For more information, see “Reanalyze a run” on page 165.


The data displayed in the screen depend on the assay that was used in the run.

Metric ^[1]	Description
Purification QC	Quality control information for nucleic acid extraction that is performed on a Genexus™ Purification Instrument.
Sample Concentration RNA	The concentration of extracted RNA after purification.
Sample Concentration DNA	The concentration of extracted DNA after purification.
Sample Concentration TNA	The concentration of extracted TNA after purification.
Run QC	General run quality control information.
Key Signal	The average signal after software processing for library ISPs that identically match the library key (TCAG).
Percent Loading	The number of wells with ISPs divided by the number of the total addressable wells in a run.
Raw Read Accuracy	The average raw accuracy across each individual base position in a read, where raw read accuracy is calculated as $100 * (1 - (\text{sum}(\text{per base error})/\text{sum}(\text{per base depth})))$.
Templating QC—CF-1 Control	Sequencing quality metrics of the control fragment. These metrics indicate templating success.
Average Reads Per Lane	The number of CF-1 reads divided by the number of chip lanes used in the run.
Base Call Accuracy	The probability that a given base is called correctly. $1 - (\text{total number of errors for all positions in CF-1}) / (\text{total number of CF-1 base reads})$.
Mean AQ20 Read Length (bp)	Average length, in base pairs, at which the accuracy rate is $\geq 99\%$ for CF-1 reads.

(continued)

Metric ^[1]	Description
Sample QC – DNA	Sequencing quality metrics of the sample DNA library.
MAPD	MAPD (Median of the Absolute values of all Pairwise Differences) is a quality metric that estimates coverage variability between adjacent amplicons in CNV analyses. A MAPD value of less than or equal to 0.4 indicates an acceptable level of coverage variability. High MAPD value typically translates to a lower coverage uniformity. Lower coverage uniformity can result in missed or erroneous CNV calls. If the MAPD QC threshold is not met, CNVs are not called. The MAPD metric does not affect SNVs/INDEL calls.
Mapped Reads	The total valid mapped reads.
Mean AQ20 Read Length (bp)	The average length, in base pairs, at which the accuracy rate is $\geq 99\%$ for all aligned reads of a library.
Mean Read Cov	The number of average reads per amplicon. This metric is generated by the CoverageAnalysis plugin.
Molecular Uniformity	Uniformity of molecular coverage for all amplicons. This metric is generated from the MolecularCoverageAnalysis plugin.
Mean Read Length (bp)	The average length, in base pairs, of final library reads for the sample.
Uniformity of Amplicon Coverage	The percentage of amplicons that had at least 20 percent of the average number of reads per amplicon. Cumulative coverage is linearly interpolated between nearest integer read depth counts.
Median Mol Cov	The median number of functional molecule reads per amplicon calculated over all amplicons in the assay. This metric is applicable to Ion AmpliSeq™ HD library chemistry only.
Uniformity of Base Coverage	The percentage of reads with a depth of coverage $\geq 20\%$ of the mean read coverage at each position. This metric is applicable to Ion AmpliSeq™ HD library chemistry only.
Read Length Histogram	The histogram presents all filtered and trimmed DNA library reads that are reported in the output BAM file (Y-axis) and the mean read length in base pairs (X-axis). The shape of the histogram should closely resemble the library size distribution trace without the adapter sequences.

(continued)

Metric ^[1]	Description
<p>Read Ratio for Inline Controls (Endogenous to Spike-ins)</p>	<p>The ratio of genomic sample reads to control reads for each inline control amplicon.</p>  <p>Note: The Read Ratio for each inline control amplicon is expected to be approximately 3 with 10 ng DNA input for both Ion AmpliSeq™ and Ion AmpliSeq™ HD chemistries.</p>
<p>Sample QC – RNA</p>	<p>Sequencing quality metrics of the sample RNA library.</p>
<p>Mapped Reads</p>	<p>The total valid mapped reads. That is, the mapped reads that pass the thresholds and filters across all of the RNA targets in the reference file. This number is a subset of the mapped reads shown in the Run Samples table in the Assay Metrics.</p>
<p>Mean Read Length (bp)</p>	<p>The average length, in base pairs, of the final library reads for the sample.</p>
<p>RNA Expression Ctrl's Detected</p>	<p>The number of expression control genes detected for the sample. This metric measures the RNA input integrity, input amount, and the fidelity of the reverse transcriptase that was used in library preparation.</p> <p>Fusion panels include primer pairs that cover control housekeeping genes.</p>
<p>Mean AQ20 Read Length</p>	<p>The average length, in base pairs, at which the accuracy rate is $\geq 99\%$ for all aligned reads of a library.</p>
<p>Read Length Histogram</p>	<p>The histogram presents all filtered and trimmed library reads that are reported in the output BAM file (Y-axis) and the mean read length in base pairs (X-axis).</p>
<p>Read Ratio for Inline Controls (Endogenous to Spike-ins)</p>	<p>The ratio of genomic sample reads to control reads for each inline control amplicon. You can use this metric to measure the level of contaminating genomic DNA in RNA libraries when you select the RNA checkbox for Include Inline Controls in assay set up. Using inline controls in RNA assays will reduce the total number of panel reads.</p> <p>Note: The Read Ratio for each inline control amplicon is expected to be approximately 3 with 10 ng DNA input for both Ion AmpliSeq™ and Ion AmpliSeq™ HD chemistries.</p>
<p>NTC QC – DNA</p>	<p>DNA Sequencing quality metrics of the no-template control.</p>
<p>Average Base Coverage Depth</p>	<p>The average number of DNA reads of all targeted reference bases.</p>

(continued)

Metric ^[1]	Description
Mean Read Length (bp)	The average length, in base pairs, of final DNA library reads for the no template control.
Read Length Histogram	The histogram presents all filtered and trimmed reads for the no template control that are reported in the output BAM file (Y-axis) and the mean read length in base pairs (X-axis).
Read Ratio for Inline Controls (Endogenous to Spike-ins)	The ratio of no template control sample reads to control reads for each DNA inline control amplicon.
NTC QC – RNA	RNA Sequencing quality metrics of the no template control.
Mapped Reads	The total number of valid mapped reads for the NTC.
Mean Read Length (bp)	The average length, in base pairs, of final RNA library reads for the no template control.
Read Length Histogram	The histogram presents all filtered and trimmed reads for the no template control that are reported in the output BAM file (Y-axis) and the mean read length in base pairs (X-axis).
Read Ratio for Inline Controls (Endogenous to Spike-ins)	The ratio of no template control sample reads to control reads for each RNA inline control amplicon.

^[1] For BAM to Result runs, only the Sample QC—DNA or Sample QC—RNA metrics are shown.

MAPD copy number QC metric

The Median of the Absolute values of all Pairwise Differences (MAPD) score is reported on Aneuploidy run results and other runs that detect CNVs.

MAPD is one of the metrics that we define to assess whether the panel data is useful for copy number variation (CNV) run results.

MAPD is defined as the Median of the Absolute values of all Pairwise Differences between log₂ ratios per tile for a given run. Tiles roughly correspond to amplicons in an Ion AmpliSeq™ assay. Each pair is defined as adjacent in terms of genomic distance. Tiles corresponding to copy number amplicons and other amplicons are being treated equally as no differences in variability are seen between these types. Then, any two log₂ ratios that are adjacent on the genome are a pair. Except at the beginning and the end of a chromosome, every log₂ ratio belongs to two pairs.

Formally, if x_i is the log₂ ratio at position i , with i ordered by genomic position:

$$\text{MAPD} = \text{median} (| x(i-1) - x(i) |)$$

MAPD is a per-sequencing run estimate of copy number variability, similar to standard deviation (SD). If one assumes the log₂ ratios are distributed normally with mean 0 and a constant SD, then MAPD/0.95 is approximately equal to SD. However, unlike SD, using MAPD is robust against high biological variability in log₂ ratios induced by known conditions such as cancer.

Regardless of the source of the variability, increased variability decreases the quality of CNV calls.

View Key Findings

You can view **Sample Details**, a summary of the metrics for the run, the **Key Variants** matrix, and **Coverage Graphs** for genes assayed in the run in Genexus™ Software.

1. In the menu bar, click **Results ▶ Sample Results**.
2. In the **Sample Name** column, click the sample name of interest.

Review the key findings. For more information, see “Key Findings” on page 123.

Key Findings

The **Key Findings** table shows details about the sample, a summary of key metrics for the run, and coverage plots for genes assayed in the run.

Note: The **Key Findings** table is by default the first view that is first shown for sample results.

The information that is displayed depends on the assay that was used in the run.

Sample Details

Section	Description
Sample Name	A unique identifier representing the sample.
Collection Date	The date that the sample was collected.
Gender	The biological sex of the sample: Female , Male , or Unknown .
Sample Type	A term that describes the sample, for example, FFPE, DNA, DNA & RNA.
Application Category	The application type of the sample.
Cancer Type	The type of cancer that is represented by the sample.
Cancer Stage	The stage of the cancer from which the sample was collected.
%Cellularity ^[1]	The percentage of tumor cellularity in the sample. This is a whole number between 1 and 100. The % Cellularity attribute is entered when a sample is created. The attribute is applicable to FFPE samples only.

^[1] Metric is shown only for some assays.

Key Metrics

Section	Description
Target Coverage ^[1]	
Target base coverage at Nx	The percentage of reference genome bases covered by at least <i>N</i> reads.
Amplicon Summary	
Average Base Coverage Depth	The average number of reads of all targeted reference bases. This is the total number of base reads on target divided by the number of targeted bases, and therefore includes any bases that had no coverage.

Key Metrics (continued)

Section	Description
Uniformity Of Base Coverage	The percentage of bases in all targeted regions (or whole genome) with a depth of coverage $\geq 20\%$ of the mean read coverage. Cumulative coverage is linearly interpolated between the nearest integer base read depths.
Percent Reads On Target	The percentage of filtered reads that are mapped to any targeted region relative to all reads mapped to the reference. A read is considered on target if at least one aligned base overlaps at least one target region. If no target regions (file) was specified, this value will be the percentage of reads passing uniquely mapped and/or non-duplicate filters, or 100% if no filters were specified.

^[1] Metrics are shown only for analyses that run the `coverageAnalysis` plugin.

The Key Variants matrix

The **Key Variants** matrix provides a color-coded visual representation of the variant results. When the application category of the sample is **Solid Tumor**, **Cancer (Germline)** or **Hematologic Cancer**, the genes that are shown in the **Key Variants** matrix are determined by the filter chain and the gene list applied to the results. You can change the filters that are applied to expand or restrict the genes that are shown. For more information, see “View the Key Variants matrix” on page 125.

Each tile in the **Key Variants** matrix represents one gene and indicates whether a variant is detected or not. Note that a single gene is represented with multiple tiles when there are multiple mutations detected in that gene.

View the Key Variants matrix

You can view a color-coded visual summary of variant results in Genexus™ Software.

1. In the menu bar, click **Results** ▶ **Sample Results**.
2. Click a sample name.
The **Results** screen opens to the **Key Findings** tab, which shows the **Sample Details**, **Key Metrics**, the **Key Variants**, and the **Coverage Graphs**.
3. Refine the genes that are shown in the **Key Variants** matrix.

Action	Procedure
Hide the genes that are not assayed.	Deselect the Not Assayed checkbox.
Refine the genes that are shown in the Key Variants matrix.	<ol style="list-style-type: none"> 1. Click Edit Filters. 2. To refine or expand the genes, change one or both of the filters. <ul style="list-style-type: none"> • Select a different filter chain or no filter chain. For more information, see “Filter results” on page 150. • Select a different gene list or no gene list. For more information, see “Gene lists (manager/administrator)” on page 244. 3. Click Save.

Here is an example **Key Variants** matrix.

SRTTestSample01 [QC](#) [Key Findings](#) [Variants](#) [Plugins](#) [Reports](#) ...

Sample Details

Sample Name	Collection Date	Gender	Sample Type	Application Category	Cancer Type	Cancer Stage	%Cellularity
SRTTestSample01	29 APR 2018	Male	FFPE	Cancer	Non-Small Cell Lung Cancer	Primary	10

Key Metrics

Amplicon Summary

Average Base Coverage Depth	Uniformity Of Base Coverage	Percent Reads on Target
1705	94.14%	97.05%

Key Variants

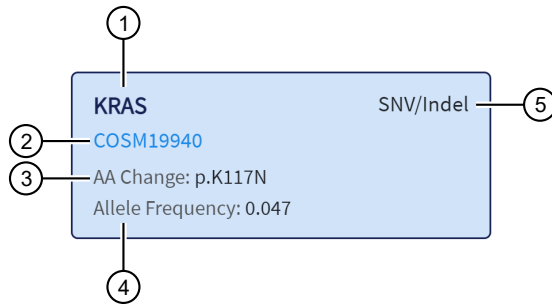
1 Key Variants Detected 2 Other Variants Detected 3 None Detected 4 Not Assayed 5 Not Assayed (4) **Oncomine Variants (5.14)** filter chain and **Non-Small Cell Lung Cancer** gene list applied 6 [Edit Filters](#)

7 KRAS SNV/Indel KRAS G13D AA Change: p.Gly13Asp Allele Frequency: 0.39	ALK	BRAF	EGFR
MET	NTRK2	NTRK3	ERBB2
NTRK1	RET	ROS1	

- ① **Key Variants Detected** – A gene is present in the gene list, variants are called by the Oncomine™ Variant Annotator, and the variants are **Key Variants**. Genes that are called are listed in the Variants tab with **Present** in the **Call** column. **Key Variants** are listed in the **Variants** tab, with a value of **Yes** in the **Key Variant** column. For more information about gene lists, see “Gene lists (manager/administrator)” on page 244.
- ② **Other Variants Detected** – A gene is present in the gene list and variants are called by the Oncomine™ Variant Annotator, but the variants are not **Key Variants**. Genes that are called are listed in the Variants tab with **Present** in the **Call** column. Variants that are not **Key Variants** are listed in the **Variants** tab, with a value of **No** in the **Key Variant** column. For more information about gene lists, see “Gene lists (manager/administrator)” on page 244.
- ③ **None Detected** – A gene is present in the gene list but no variants are called by the Oncomine™ Variant Annotator.
- ④ **Not assayed** – A gene is present in the gene list, but is not included in the panel used in the assay.
- ⑤ Checkbox to hide or show genes that are not assayed.
- ⑥ **Edit filters** allows you to select an available filter chain and gene list. Changes of the filter chain or gene list change the genes that appear in the **Key Variants** matrix.
- ⑦ **Variant ID** – Specific for a particular mutation. The **Variant ID** is a hyperlink to the pileup for the variant in the **Variants** tab.

Gene tiles in the Key Variants matrix

Each blue tile in the **Key Variants** matrix represents a variant and summarizes information for the variant.






- ① The gene name.
- ② The **Variant ID** – link to the variant details in the **Variants** tab.
- ③ The amino acid change using Human Genome Variation Society (HGVS) nomenclature.
- ④ The number of variant read counts divided by the total number of read counts for the sample.
- ⑤ The variant type.

View the amplicon coverage

You can view DNA exon amplicon coverage in Genexus™ Software to help you determine whether the sequencing reads across a gene are uniform and sufficient. The default genes that are shown are determined by the gene list that is designated as default for the cancer type of the sample. You can also view amplicon coverage graphs for more genes. These amplicon coverage graphs provide a high-level overview of coverage. More detailed coverage information is also available in the software. For more information, see “coverageAnalysis plugin in Genexus™ Software” on page 297.

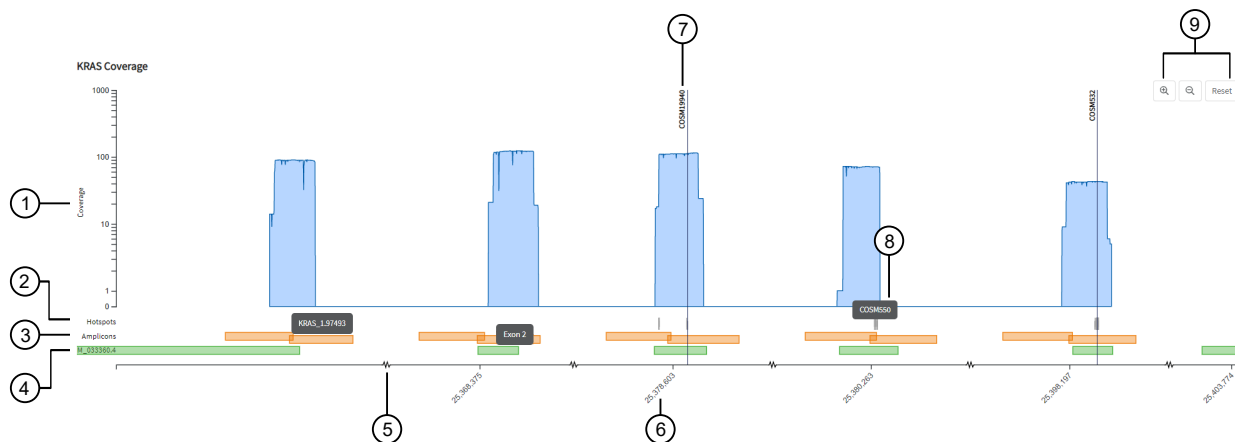
1. In the menu bar, click **Results ▶ Sample Results**.
2. Click a sample name to open the **Key Findings** tab in the **Results** screen.
3. Scroll to the amplicon **Coverage Graphs**.
4. Review the coverage graphs.
5. To show coverage graphs for more genes, scroll to the bottom of the screen. Click **Show Coverage for Gene**, then select the gene of interest.
6. You can adjust the amplicon coverage graph with the pointer and the buttons in the upper right corner of each graph.

Action	Procedure
Zoom in on a region of interest	Click  one or multiple times.
Zoom out for an expanded view	Click  one or multiple times.
Revert to the default view	<ul style="list-style-type: none"> • Click  one or multiple times. • Click Reset.
Move the image left or right in the screen	After you zoom in, click-drag at any position in the image.

The coverage graphs change to show coverage for the selected gene.

Example amplicon coverage graph

Here is an example of an amplicon coverage graph for the *KRAS* gene.



- ① Base coverage is shown on the y-axis on a logarithmic scale.
- ② The location of the known hotspots are denoted with gray bars.
- ③ The location and span of the amplicons are represented with orange bars. You can hover the mouse over an orange bar to view the amplicon name.
- ④ The location and span of the transcript track is denoted with green bars. You can hover the mouse over the green bar to view the exon number.
- ⑤ The line along the x-axis is broken to indicate that the graph is discontinuous to show exons.
- ⑥ The genome coordinate position is shown on the x-axis.
- ⑦ Called variants are indicated with a vertical line above the blue coverage plot. The variants noted in the graph depend on the filter chain applied to the results in the **Key Variants** matrix. For more information, see “View the Key Variants matrix” on page 125.
- ⑧ You can place the pointer over the hotspots that are represented with gray lines to view the hotspot name.
- ⑨ You can adjust the amplicon coverage graph. For more information, see “View the amplicon coverage” on page 127.

View variant results

You can view detailed variant results in the **Variants** table. In the default view, all variants are listed. You can also limit the types of variants that are shown and toggle between different variant results: SNVs/Indels, Fusions, and CNVs.

Variant Tables

Variant type	Description
SNVs/Indels	<ul style="list-style-type: none"> • SNVs: missense and nonsense single nucleotide variants. Multi-nucleotide variants are also included. • Indels: insertion and deletion variants. <p>For more information, see “View SNV/INDEL results” on page 131.</p>

Variant Tables (continued)

Variant type	Description
Fusions	Translocations of genetic material. For more information, see “View Fusion results” on page 134.
CNVs	Copy number variations (CNVs) are variations of the number of copies of a given gene. For more information, see “View CNV results” on page 140.

Show gene list variants

You can refine the variants that are shown in the **Variants** tab in Genexus™ Software:

- Filter the variants. For more information, see “Filter results” on page 150.
- Show only the gene list variants. For more information, see “Gene lists (manager/administrator)” on page 244.

1. In the menu bar, click **Results** ▶ **Sample Results**.
2. Click a sample name to open the **Results** screen.
3. Click the **Variants** tab.

Option	Description
Only show the variants for the genes listed in the gene list	Click the Gene List Only checkbox. Changes to the Gene List Only checkbox, whether the gene list is shown or not shown, persist for the selected result only. When you select a new sample in the same run, the selection persists. When you open a new run result or a sample result from a different run, the checkbox is deselected by default.
Change the gene list that is applied to the results	<ol style="list-style-type: none"> 1. Click Edit Filters, then select a different gene list. 2. Click Save. <p>The changed gene list is saved for the selected sample result only. The saved change applies only to a single sample, even if other samples are present in the same run.</p>

Note: The **Gene List Only** checkbox is only shown when a gene list is applied to the results. When **No Gene List** is selected, the **Gene List Only** checkbox is not shown in the results screen.

Customize the view of the variants table

The **Variants** table includes detailed variant results in Genexus™ Software. You can customize the table to reflect the information that is most relevant to your research.

Variants table column options

When you view the **Variants** table in Genexus™ Software, a default set of columns is included. The table columns are based on various factors, including the samples that are sequenced and the assay used for sequencing.

Add or remove columns in the variants table

You can add columns to the **Variants** table that you view in Genexus™ Software.

1. In the menu bar, click **Results** ▶ **Sample Results**.
2. Click a sample name to view results for the sample.
3. Click **Columns**, then select the columns that you want to add to the **Variants** table, or deselect the columns that you want to remove from the table.
The selected columns are added to the **Variants** table until you close the table.
4. To save the custom table so that the changes to the columns persist the next time the table is viewed, click **Save**.

Change the width of columns in the variants table

You can change width of columns in the **Variants** table in Genexus™ Software. Not all columns are visible on the screen. You can use the scrollbar under the table to view more columns.

1. In the menu bar, click **Results** ▶ **Sample Results**.
2. Click a sample name to open the **Results** screen.
3. Change the width of each column, as desired.

Restore default settings for the variants table

You can restore default column headers in the **Variants** table in Genexus™ Software.

1. In the menu bar, click **Results** ▶ **Sample Results**.
2. Click a sample name to open the **Results** screen.
3. Click **Columns** ▶ **Restore default**.

The default columns shown in the table are restored.

View SNV/INDEL results

The **SNVs/Indels** table lists the calls and other information for the SNV and INDEL variants that are analyzed in each sample in a run.

1. In the menu bar, click **Results** ▶ **Sample Results**.
2. Click a sample name to open the **Results** screen.
3. Click the **Variants** tab.
4. Click **SNVs/Indels** to view the **SNVs/Indels** table.
For more information about the data displayed in the table, see “SNVs/Indels table” on page 131.
5. (Optional) Click **Export** in the upper right corner of the screen to export the results data in tabular format.

SNVs/Indels table

The data displayed in the table depend on the assay that was used in the run.

You can filter the results list in the table using filtering tools and filter chains. For more information, see “Filter results” on page 150.

Column	Description
User Classifications	User-defined classification to selected from the list. For more information, see “Create and assign variant classifications” on page 149.
Variant ID	The name of the hotspot as defined in the Browser Extensible Data (BED) file. Click the link to view more annotation information. For more information, see “View more annotations and annotation sources” on page 143.
Key Variant	Indicates whether or not the variant is a key variant. Possible values are Yes or No . This information is available if the Apply Oncomine Variant Annotations checkbox is selected in the assay used in the run.
Locus	The chromosome and position of the detected variant.
Oncomine Variant Class	The type of SNV or INDEL at the locus based on Oncomine™ annotations. This information is available if the Apply Oncomine Variant Annotations checkbox is selected in the assay used in the run.

(continued)

Column	Description
Oncomine Gene Class	<p>The change in molecular function of the altered gene product due to the mutation, based on Oncomine™ annotations:</p> <ul style="list-style-type: none"> • Gain-of-function—the altered gene product has a new molecular function or pattern of gene expression compared to the wild-type gene • Loss-of-function—the altered gene product lacks the molecular function of the wild-type gene <p>This information is available if the Apply Oncomine Variant Annotations checkbox is selected in the assay used in the run.</p>
Gene	The gene name.
AA Change	Identification of the amino acid change using Human Genome Variation Society (HGVS) nomenclature.
Ref	The reference base or bases at that locus.
Alt	The alternate base or bases at that locus.
Type	<p>The type of variant that is detected.</p> <ul style="list-style-type: none"> • snp (single nucleotide polymorphism) • mnp (multi-nucleotide polymorphism) • ins (insertion) • del (deletion) • complex
Call	<p>Indicates the presence or absence of an SNV/Indel variant. When the default filter chain is applied, only the variant calls that are designated with PRESENT (HOMOZYGOUS) or PRESENT (HETEROZYGOUS) are displayed in the results table. To view all calls, including calls that do not pass the required filter thresholds, apply the No Filter option or download the Variants (VCF) file (see “Results files” on page 169).</p> <ul style="list-style-type: none"> • PRESENT (HOMOZYGOUS) or PRESENT (HETEROZYGOUS) – indicates a high confidence call that passes all filter thresholds at a given variant position. <p>Note:</p> <ul style="list-style-type: none"> • When the default filter chain is applied, PRESENT (HOMOZYGOUS) or PRESENT (HETEROZYGOUS) indicates the presence of the ALT (alternative) allele. • When the No Filter option is applied or when viewing the Variants (VCF) file, Present does <i>not</i> imply the presence of the ALT (alternative) allele. To infer the presence of the ALT allele, see the Alt column. • NO CALL – although some evidence for the presence of a variant exists, the call does not pass one or more filters that are required for a high confidence variant call. • ABSENT – indicates the presence of a variant at a given position that is not targeted by the assay and is not the reference allele.
Call Details	The reason why a variant is reported as NO CALL .

(continued)

Column	Description
Phred QUAL Score	The relative probability of either the "reference" hypothesis interval [0,cutoff], or the "variant" hypothesis interval [cutoff,1], Phred-scaled ($-10 \cdot \log_{10}$). A higher score means more evidence for the variant call.
Raw Read Depth	Total read coverage across amplicon containing SNV/INDEL hotspot locations. Count of chip-level reads aligned at this locus that participate in variant calling.
Effective Read Depth	The number of reads covering the position.
Alt Allele Read Counts	The number of reads containing the alternate allele.
Coverage	The number of total reads at a locus, wild-type + alt allele.
Nuc Change	The position and identity of the nucleic acid change.
Allele Frequency	The number of variant read counts divided by the total number of read counts for the sample.
Allele Frequency (%)	The allele frequency, represented as a percentage.
Allele Ratio	The relative frequency of each allele.
Zygosity	Describes whether the variant is homozygous (0) or heterozygous (1).
Filtered Read Coverage	Coverage at the position considering only filtered reads.
Allele read Count	The number of reads detected for the allele.
Raw Alt Allele Read Counts	Number of unfiltered reads containing the alternate allele.
PPA	Possible Polyploidy Alleles (PPA). A value of Yes indicates variants that are PPA alleles. No indicates variants that are not PPA alleles.
P-Value	Probability value for the detection of variant calls.
Mol Depth ^[1]	Reports number of interrogated DNA molecules containing target. This metric defines the limit of detection at a hotspot position in a particular run and sample. For reference calls, molecular depth provides a measurable metric that serves as confirmation of variant absence among a large number of interrogated molecules. For instance, if molecular depth is $\geq 1,500$, you can have high confidence that no variant is present at $\geq 0.2\%$ variant allele frequency. If molecular depth is $\geq 2,500$, you can have high confidence that no variant is present at $\geq 0.1\%$ variant allele frequency.
WT Mol Counts ^[1]	Number of detected molecules containing the wildtype allele.
Alt Allele Mol Counts ^[1]	Number of detected molecules containing the alternate allele.

(continued)

Column	Description
Mol Freq % ^[1]	Molecular frequency percentage. The percentage of alternate allele reads over total reads at the locus.
% LOD ^[1]	Limit of detection (LOD) of a variant allele expressed as a percentage of the WT allele. LOD is the lowest possible variant frequency in the sample that can be detected by the system with a true positive rate greater than 98% for FFPE samples or 95% for cfTNA samples. LOD is dependent on the molecular read depth at the locus. %LOD is reported when there are no variant calls for the gene.

^[1] Column appears in analyses of Ion AmpliSeq™ HD sequencing data only.

View Fusion results

You can view calls and other information for the fusions that are analyzed within each sample in a run.

1. In the menu bar, click **Results** ▶ **Sample Results**.
2. Click a sample name to open the **Results** screen.
3. Click the **Variants** tab.
4. Click **Fusions** to view the **Fusions** table.
For more information about the data displayed in the table, see “Fusions table” on page 134.
5. (Optional) Click **Export** in the upper right corner of the screen to export the results data in tabular format.

Fusions table

The data displayed in the table depend on the assay that was used in the run.

You can filter the results list in the table using filtering tools and filter chains. For more information, see “Filter results” on page 150.

Column	Description
User Classifications	A user-defined classification selected from the list. For more information, see “Create and assign variant classifications” on page 149.
Variant ID	The name of the fusion target as defined in the BED file.
Key Variant	Indicates whether or not the variant is a key variant. Possible values are Yes or No . This information is available if the Apply OncoPrint Variant Annotations checkbox is selected in the assay used in the run.
Locus	The chromosome positions in the reference genome that define the fusion junction.

(continued)

Column	Description
Oncomine Variant Class	Oncomine variant class annotation that indicates fusion type based on Oncomine™ annotations. This information is available if the Apply Oncomine Variant Annotations checkbox is selected in the assay that is used in the run.
Oncomine Gene Class	The change in molecular function of the altered gene product due to the mutation, based on Oncomine™ annotations: <ul style="list-style-type: none"> • Gain-of-function—the altered gene product has a new molecular function or pattern of gene expression compared to the wild-type gene • Loss-of-function—the altered gene product lacks the molecular function of the wild-type gene <p>This information is available if the Apply Oncomine Variant Annotations checkbox is selected in the assay used in the run.</p>
Genes (Exon)	The name of fusion target and representative acceptor and donor exons.
Read Counts	The frequency that the fusion was detected in the sample.
Type	Assay type (for example, Fusion, RNA exon variant (exon skipping), RNAExon Tile, Proc Control).
Call	Indicates the presence or absence of a fusion or RNA exon variant. When the default filter chain is applied, only the fusion/RNA exon variant calls that are designated with PRESENT are displayed in the results table. To view all calls, including calls that do not pass the required filter thresholds, apply the No Filter option or download the Variants (VCF) file (see “Results files” on page 169). <ul style="list-style-type: none"> • PRESENT – indicates a high confidence call that passes all filter thresholds at a given variant position. • ABSENT – indicates the absence of a fusion due to a variant call that falls below thresholds. • NO CALL – although some evidence for the presence of a fusion exists, the call does not pass one or more filters that are required for a high confidence fusion call.
Call Details	The reason for reporting a fusion as NO CALL .
Failed Reason	The reason for reporting a fusion as Absent .
Read Counts Per Million	The number of fusion read counts detected per million total reads.
Oncomine Driver Gene	The gene believed to be associated with increased oncogenic properties. The gene is inappropriately activated by the fusion.
Gene Isoform	The detected fusion isoform.
Mol Cov. Mutant^[1]	The median molecular coverage across a fusion amplicon.

(continued)

Column	Description
Imbalance Score ^[2]	<p>Each fusion gene exhibits a characteristic Imbalance Score threshold. Scores that exceed the threshold value indicate a high likelihood of the presence of the fusion in the sample. Imbalance score calculation starts with the normalization of reads in the exon-tiling amplicons of the gene, followed by correction with a baseline that represents expression values in normal samples. Reads for observed imbalance scores come from samples. Baseline scores come from normal, fusion-negative samples and are stored in the exon tile assay baseline file for the assay.</p> $\text{Imbalance score} = \frac{\text{Observed imbalance (samples)}}{\text{Expected imbalance (baseline)}}$ <p>where</p> <ul style="list-style-type: none"> Observed imbalance = $\frac{3' \text{ of breakpoint}}{5' \text{ of breakpoint (in samples)}}$ <ul style="list-style-type: none"> 3' of breakpoint is the sum of the normalized reads of amplicons 3' of a predicted breakpoint for the gene in the sample. 5' of breakpoint is the total normalized reads of the amplicons for the gene in the sample. Expected imbalance = $\frac{3' \text{ of breakpoint}}{5' \text{ of breakpoint (in the baseline)}}$ <ul style="list-style-type: none"> 3' of breakpoint is the sum of normalized baseline values of amplicons 3' of the breakpoint for the gene. 5' of breakpoint is the total normalized baseline values of the amplicons for the gene.
Imbalance P-Value ^[2]	The statistical significance of measure of imbalance relative to a control gene.
Predicted Break-point Range ^[2]	The exonic range for predicted fusion break point in exon tiling assays.
Ratio To Wild Type ^[1]	The molecular ratio for exon skipping assays relative to wild type control amplicons.
Norm Count Within Gene ^[1]	(<i>Lung panel only</i>) Exon skipping assay coverage normalized to molecular coverage of wild type (WT) MET control amplicons.

^[1] Column appears in analyses of Ion AmpliSeq™ HD sequencing data only.^[2] Column appears in analyses that use the exon tiling fusion detection method.

View RNA Exon Variants

The RNA Exon Variant data view displays a bar graph summary of intragenic exon rearrangements or fusions. The displayed RNA exon variants are defined in the BED file that is associated with an assay. The RNA Exon Variant data view is available for all RNA and Fusion assays.

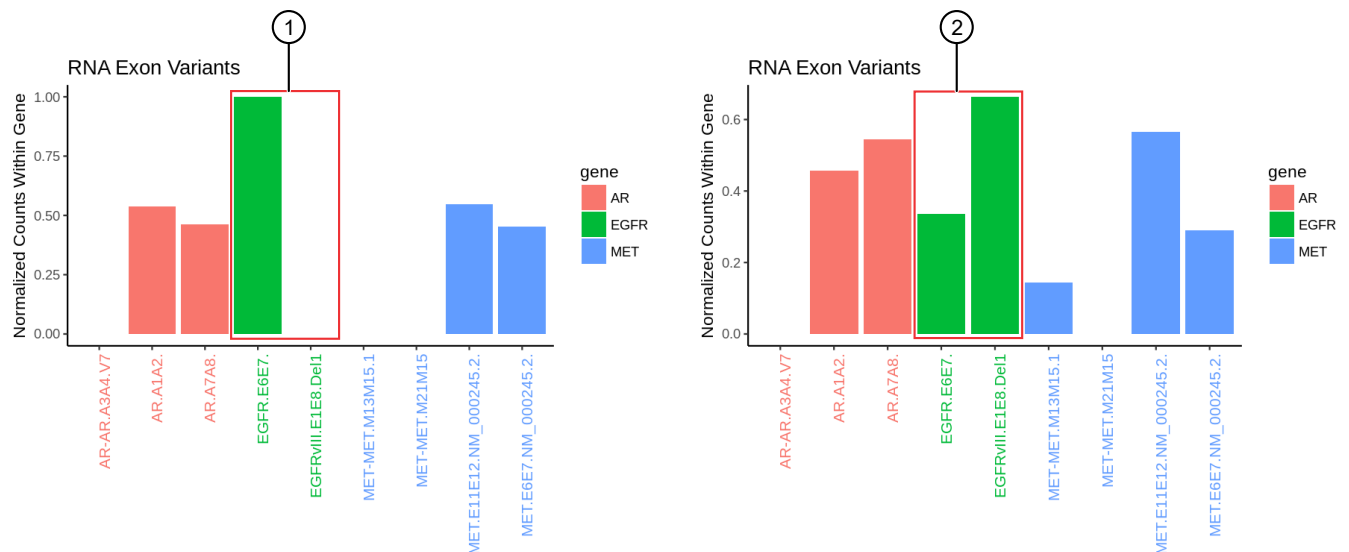
1. In the menu bar, click **Results** ▶ **Sample Results**.
2. In the **Sample name** column, click the name of the sample of interest.

3. Click the **Variants** tab, then click **Fusions**.

The **Fusions** table opens to display fusions results. For more information, see “Fusions table” on page 134.

4. In the top right corner of the screen, click **Visualization** ▶ **RNA Exon Variant**, then review the **RNA Exon Variants** plot.

Representative RNA Exon Variant plots



The X-axis represents specific exon variants, where each variant is labeled with a gene ID followed by a sequence of adjacent exons. The Y-axis measures the read counts for each variant, normalized to the wild type.

- ① Example result where only the wild type EFGR (EFGR.E6E7) was detected.
- ② Example result where RNA exon 2–7 deletion occurred in the EFGR gene. The deletion of exons 2–7 resulted in an increase of normalized read counts for the EFGR variant that contains the intragenic fusion of exon 1 and exon 8 (EFGR.E1E8.Del1) and a decrease of normalized read counts for the wild type EFGR (EFGR.E6E7).

To return to the table view of fusions, click **X (Remove)** next to the **Visualization** dropdown menu.

View RNA exon tile fusion imbalance

The **RNA Exon Tile Fusion Imbalance** data view provides a visual representation of the RNA fusion imbalance analyses.

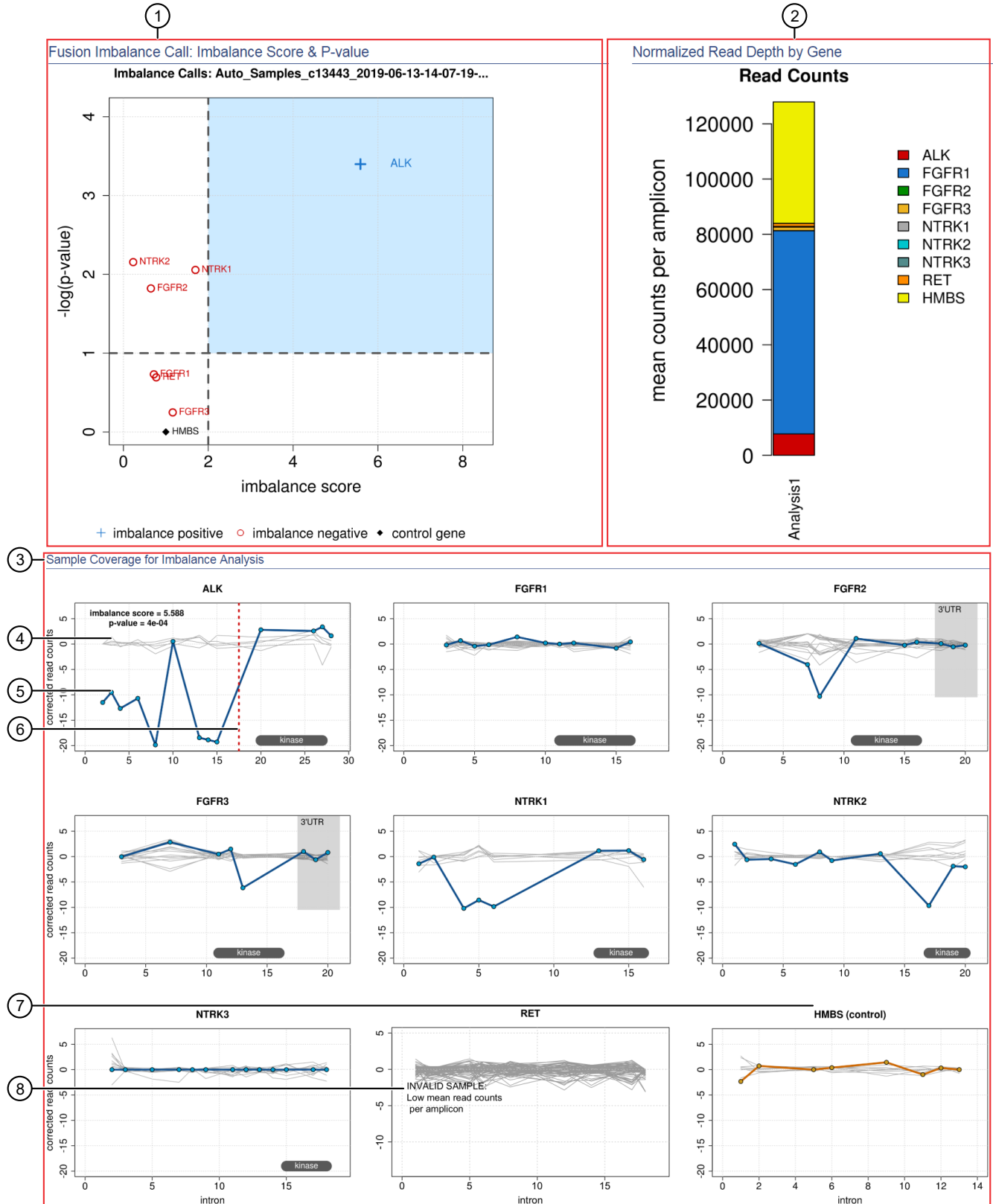
Driver genes, for example ALK, RET, NTRK1, have amplicons that span exon-exon junctions to probe the expression difference between the 3' and the 5' regions of the gene.

An imbalance score is calculated for each driver gene in a sample to quantify the magnitude of the expression imbalance change between the two parts of the gene. For example, the 3':5' ratio in ALK; and the 5':3' ratio in FGFR2.

Note: The exon tiling method for fusion detection and the RNA Exon Tile Fusion Imbalance data view are specific to the OncoPrint™ Precision Assay GX. For more information, see the *OncoPrint™ Precision Assay GX User Guide* (Pub. No. MAN0018508).

1. In the menu bar, click **Results** ▶ **Sample Results**.
2. In the **Sample name** column, click the name of the sample of interest.
3. Click the **Variants** tab, then click **Fusions**.
The **Fusions** table opens to display fusions results. For more information, see “Fusions table” on page 134.
4. In the top right corner of the screen, click **Visualization** ▶ **RNA Exon Tile Fusion Imbalance**, then review the RNA Exon Tile Fusion Imbalance plots.

Representative RNA exon tile fusion imbalance plots



1 The Fusion Imbalance Call: Imbalance Score & P-value plot shows the imbalance scores and p-values for all the genes in

the selected sample. The dashed gray lines mark the threshold for an imbalance call, which is applied to all genes across all samples. Points that fall within the blue shaded area of the plot represent fusion-positive genes (+). All other points that are outside of the blue shaded area represent fusion-negative genes (○). Control genes are marked with ◆.

- ② The **Normalized Read Depth by Gene** plot shows the mean read counts of each gene that is captured on the chip for the selected sample. For each gene, the read counts are normalized to the number of amplicons.
- ③ The **Sample Coverage for Imbalance Analysis** plots show the expression profile for each exon-exon tiling amplicon for each gene. The Y-axis represents the corrected molecular counts. The X-axis represents individual exon-exon junctions, which are listed from 5' to 3'. The **imbalance score** and **p-value** are listed in the panel of each gene that was called positive for fusion.
- ④ Baseline (a cluster of gray lines), generated from a fusion-negative sample.
- ⑤ Test sample corrected read coverage (blue line), normalized to the baseline. Each point on the line represents a unique exon-exon junction that was covered by the assay and normalized to the baseline.
- ⑥ Predicted range for the fusion break point for a fusion-positive gene (dashed red line).
- ⑦ Sample coverage profile for the control gene (orange line).
- ⑧ If the collected data are insufficient to determine an imbalance score, the **INVALID SAMPLE** message appears in the panel for that gene.

To return to the table view of fusions, click **X (Remove)** next to the **Visualization** dropdown menu.

View CNV results

The **CNVs** table lists the calls and other information for the copy number variants (CNVs) analyzed in each sample in a run.

1. In the menu bar, click **Results ▶ Sample Results**.
2. Click a sample name to open the **Results** screen.
3. Click the **Variants** tab.
4. Click **CNVs** to view the **CNVs** table.

For more information about the data displayed in the table, see “CNVs table” on page 141.

5. (Optional) Click **Export** in the upper right corner of the screen to export the results data in tabular format.

CNVs table

The data displayed in the table depend on the assay that was used in the run.

You can filter the results list in the table using filtering tools and filter chains. For more information, see “Filter results” on page 150.

IMPORTANT! (*FFPE samples only*) If the %**Cellularity** value for a sample is set to <100, then the magnitude of copy number gain or loss can be decreased. For more information, see “System-installed sample attributes” on page 368.

Column	Description
User Classifications	A user-defined classification selected from the list. For more information, see “Create and assign variant classifications” on page 149.
Variant ID	The identifier of the CNV variant. Click the link to view more annotation information. For more information, see “View more annotations and annotation sources” on page 143.
Key Variant	Indicates whether or not the variant is a key variant. Possible values are Yes or No . This information is available if the Apply OncoPrint Variant Annotations checkbox is selected in the assay used in the run.
Locus	The starting position of the first amplicon covering the CNV gene.
OncoPrint Variant Class	Annotation that indicates whether CNV is an amplification or deletion. This information is available if the Apply OncoPrint Variant Annotations checkbox is selected in the assay used in the run.
OncoPrint Gene Class	The change in molecular function of the altered gene product due to the mutation, based on OncoPrint™ annotations: <ul style="list-style-type: none"> Gain-of-function—the altered gene product has a new molecular function or pattern of gene expression, compared to the wild-type gene Loss-of-function—the altered gene product lacks the molecular function of the wild-type gene This information is available if the Apply OncoPrint Variant Annotations checkbox is selected in the assay used in the run.
Gene	The gene name.
Copy Number	The copy number of a CNV gene locus per genome. This column is available when a positive call is made.

(continued)

Column	Description
Call	<p>Indicates the presence or absence of a CNV. When the default filter chain is applied, only the CNV-positive calls that are designated with PRESENT are displayed in the results table. To view all calls, including calls that do not pass the required filter thresholds, apply the No Filter option or download the Variants (VCF) file (see “Results files” on page 169).</p> <ul style="list-style-type: none"> • PRESENT – indicates a high confidence call that passes all filter thresholds. • PRESENT (GAIN) – a CNV-positive call that indicates gene amplification; a high confidence variant call that passes all filter thresholds. • PRESENT (LOSS) – a CNV-positive call that indicates gene deletion; a high confidence variant call that passes all filter thresholds. • ABSENT – the absence of a variant; result is below the detection threshold for a CNV call. • NO CALL– although some evidence for the presence of a variant exists, the call does not pass one or more filters that are required for a high confidence variant call.
P-Value	The statistical significance of the CNV ratio measurement.
Call Details	The reason for reporting a CNV as NO CALL.
CNV Confidence	The CNV confidence interval associated with the call. The 5% lower confidence bound value is the ploidy estimate, where there is a 5% chance that the true ploidy is below that value. The 95% upper confidence bound is the ploidy estimate, where it is 95% certain that the true ploidy is below that value.
CNV Ratio	The ratio of measured CNV gene locus coverage relative to coverage of non-CNV loci.
Med Read Cov Gene	The median read coverage of targeted CNV gene.
Med Read Cov Ref	The median read coverage of non-CNV reference loci.
Valid CNV Amplicons	The number of amplicons spanning the CNV call.
Type	<p>The type of variant that is detected.</p> <ul style="list-style-type: none"> • CNV– copy number variant • LOH– loss of heterozygosity
Subtype	<p>The CNV subtype.</p> <ul style="list-style-type: none"> • BigDel– deletion of at least one exon. • BigDup– duplication of at least one exon. • GeneCNV– whole BRCA1/BRCA2 gene deletion or duplication. • NOCALL– read count differs from baseline by non-integer amount; evidence for a BigDel or BigDup call is weak. • REF– read count matches reference baseline. • ARM– aneuploidy of a chromosome arm.

View more annotations and annotation sources

In addition to the annotations that are included with Genexus™ Software, you can view annotations and annotation source information from public websites for each variant.

1. In the menu bar, click **Results ▶ Sample Results**.
2. Click the sample name in the **Sample Name** column in the row of a sample of interest.
3. Click the **Variants** tab.
4. In the **Variants** table, click the **Variant ID** of the variant of interest.

Note: You can click the row even if a **Variant ID** is not listed.

The **Pileup** view is shown below the variants table. For more information, see “View the variant pileup” on page 143.

5. Click the **Annotations** tab.
6. Review annotation information in the **Annotations** tab, and if available, click a link in the row of an annotation to open a website with information about the annotation.

View the variant pileup

You can view variant tracks with the pileup view in Genexus™ Software. The pileup is a figure generated in the software that shows each BAM track represented by a single bar, with the sequence variation or variations denoted in each track. The BAM tracks are aligned against a reference track. The variant pileup can be used to identify potential variants.

1. In the menu bar, click **Results ▶ Sample Results**.
2. Click a sample name to open the **Results** screen.
3. Click the **Variants** tab.
4. In the variants table, in the **Variant ID** column, click the identifier of the variant of interest.

The **Pileup** view is shown below the variants table.

Change the view of the variant pileup

You can zoom and pan the variant pileup and add or remove guides and track labels to more easily understand the data. You can also adjust the view of the individual pileup tracks. For more information, see “Adjust pileup tracks” on page 144.

1. In the menu bar, click **Results ▶ Sample Results**.
2. Click a sample name to open the **Results** screen.
3. Click the **Variants** tab.

- In the variants table, in the **Variant ID** column, click the identifier of the variant of interest.

Action	Procedure
Track the position as you place the pointer over the variant pileup	Click Cursor Guide .
Mark the center of the variant pileup	Click Center Line .
Remove the track labels	Click Track Labels .
Zoom in	Click + one or multiple times or double-click the image.
Zoom out	Click - one or multiple times.
Pan across the image	Click and drag the pointer over the image.

Adjust pileup tracks

You can adjust the BAM read coverage track, the reference track, hotspot tracks, the target region track, and annotation tracks in Genexus™ Software to make it easier to view the data. When you adjust a track, it changes the amount of data that is shown in the plot. For example, you can see more tracks when you use the **Squish** setting.

Note: For best results, use the **Expanded** option to click and view hover help with details about variants and base calls that are shown in each read coverage track


In the **Variants** tab, in the **Pileup** subtab, click **⚙️ Actions** next to a track, then select an option to adjust the view of the track.

Option	Description
Expanded	Select this option to view the tracks with the maximum visible height.
Squish	Select this option to view the tracks with the minimal height for each track.
Collapse	Select this option to view the tracks with overlapping transcripts shown along a single line. When Collapse is selected for BAM tracks, the BAM tracks are not shown; only the coverage density is shown.

Adjust the view of the reference track

You can adjust the view of the reference track in Genexus™ Software to view or hide the three-frame translation of the reference track sequence. You can also view the forward or reverse reference track sequence.


- In the menu bar, click **Results ▶ Sample Results**.
- Click a sample name to open the **Results** screen.
- Click the **Variants** tab.
- In the variants table, in the **Variant ID** column, click the identifier of the variant of interest.

5. In the **Variants** tab, in the **Pileup** subtab, click  **Actions** next to the reference track.

Option	Description
View the reverse strand	Click Reverse . This option is only available when the forward strand is shown.
View the forward strand	Click Forward . This option is only available when the reverse strand is shown.
View the reference sequence translation	Click Three-Frame Translate . This option is only available when the amino acid sequence is not shown.
Hide the reference sequence translation	Click Close Translation . This option is only available when the amino acid sequence is shown.

Adjust the view of the BAM tracks

You can adjust the view of the BAM tracks in Genexus™ Software to dynamically customize the view of the data.

1. In the menu bar, click **Results** ▶ **Sample Results**.
2. Click a sample name to open the **Results** screen.
3. Click the **Variants** tab.
4. In the variants table, in the **Variation ID** column, click the identifier of the variant of interest.
5. In the **Variants** tab, in the **Pileup** subtab, click  **Actions** next to the BAM tracks.

Option	Description
View all BAM tracks in monochrome gray	In Color By , deselect the checkmark next to Read Strand . This option is only available when the BAM tracks are colored by read strand.
Distinguish between the direction of the BAM tracks with color	In Color By , click Read Strand . This option is only available when the BAM tracks are not colored by read strand.
Expand, squish or collapse the BAM tracks	For more information, see “Adjust pileup tracks” on page 144.
Show all the bases in the BAM tracks	Click Show all bases . This option is only available when all bases in the BAM tracks are not shown.
Change the minimum threshold for the visibility of BAM tracks	<ol style="list-style-type: none"> 1. Click Set visibility window. 2. In the Set visibility window dialog box, enter the minimum number of bases needed to show the BAM tracks. <p>When you zoom in and out to view the variant pileup, the scale of the tracks shown in the screen changes. For more information, see “Change the view of the variant pileup” on page 143. You can adjust the visibility window according to the scale shown in the screen.</p>

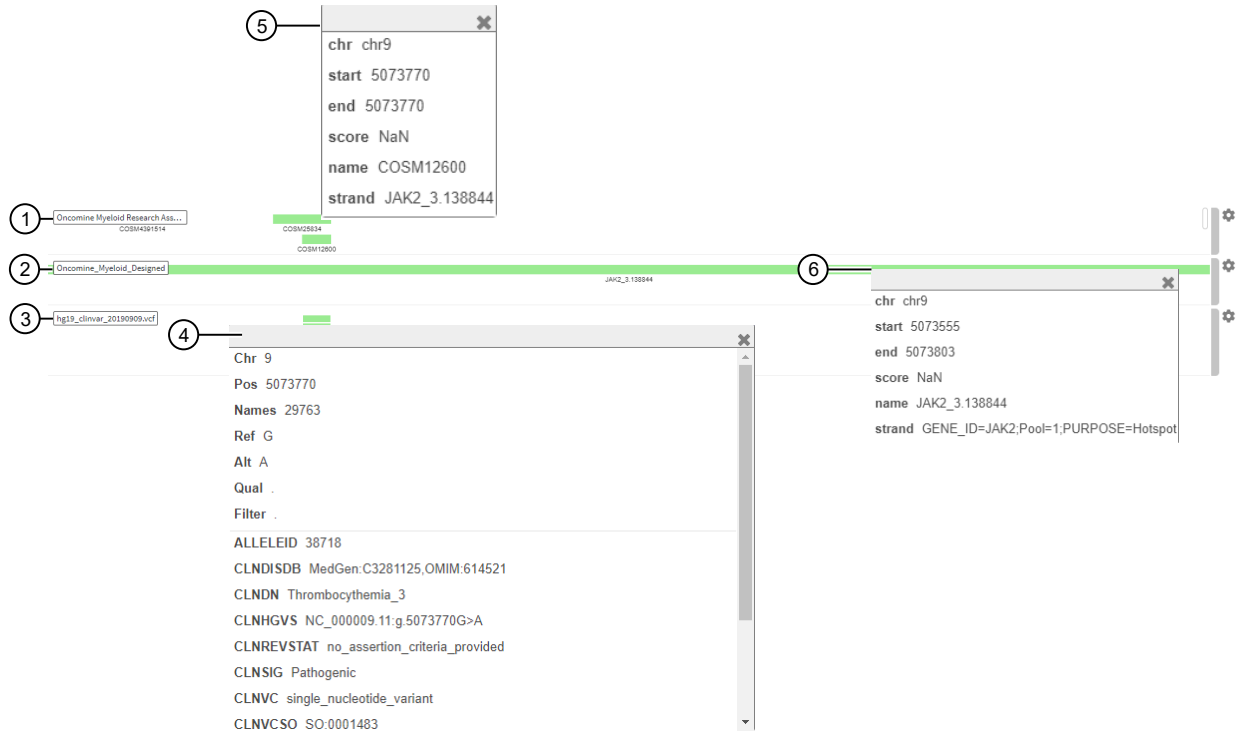
Examples of a variant pileup

This is an example of a variant pileup, shown in the **Gene Browser** in the **Variants** tab. The variant pileup graphically represents the sequencing tracks, the reference track, and the annotation tracks and can help you understand the variant data and analyze results. The reference allele and the alternate base for the selected variant are indicated in the image, both in the variant table and in the pileup. You can customize the order of the tracks shown in the pileup view by clicking on a gray bar on the right edge of a track, then dragging the track to a different location. You can also select and hold the mouse at any position in the image to move the image left or right in the screen. The way the tracks are displayed can be adjusted. For more information, see “Adjust pileup tracks” on page 144.

The screenshot displays the Gene Browser interface. At the top, there is a table of variants. The table includes columns for User Classification, Variant ID, Driver, Type, Oncomine Gene Class, Oncomine Variant Class, Gene, Locus, AA Change, Ref, Alt, Call, and Oncomine Driver Gene. Below the table, the Gene Browser view shows a reference track (1), a three-frame translation track (2), a coverage track (3), a BAM track (4), and a detailed read information popup (6). The popup shows read name, alignment start, read strand, cigar string, mapping quality, and secondary status. Numbered callouts 1-10 point to specific features in the Gene Browser view.

- ① The reference track. When zoomed out, the nucleotide sequence is no longer displayed, but the sequence is represented by colored bars.
- ② The three-frame translation is shown in this example (not by default). For more information, see “Adjust the view of the reference track” on page 144.
- ③ The coverage track.
- ④ The BAM track.
- ⑤ Click on the coverage track to see detailed information, such as the total count, the number of reads, and the number of molecules.
- ⑥ Click on a .bam track to see detailed information, such as the **Mapping Quality** and **Base Quality**.
- ⑦ Use the sliders to view more tracks.
- ⑧ Deletion.
- ⑨ Insertion.
- ⑩ Alternate base in the BAM read coverage track.

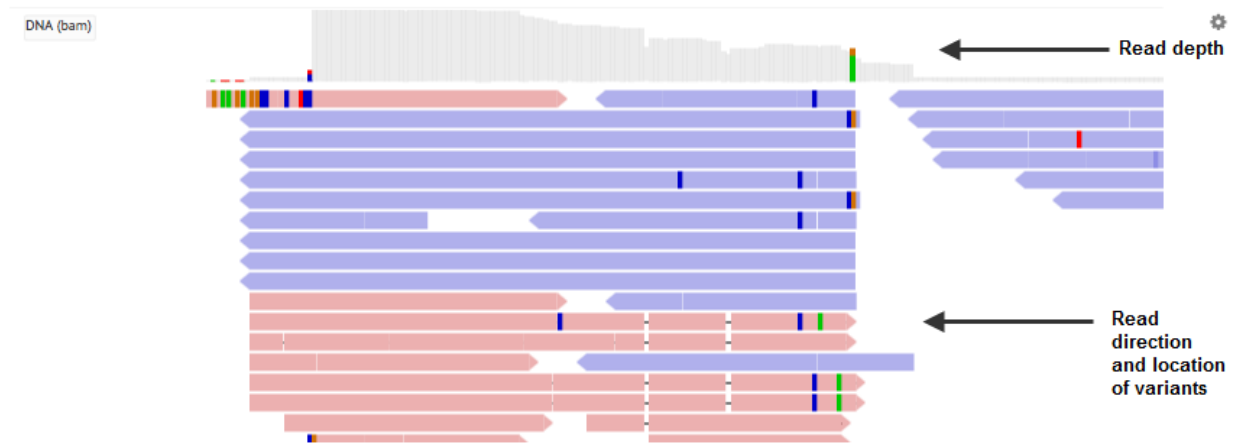
This is an example of the reference tracks and annotation tracks shown beneath the .bam tracks in the **Gene Browser**.



- ① The hotspot track.
- ② The target region track.
- ③ Annotation tracks (COSMIC and ClinVar). In this example, only ClinVar is shown.
- ④ Click on the annotation track for additional details; for example the CLNHGVS (the top-level genomic HGVS expression for the variant).
- ⑤ Click on a hotspot track for additional details, such as the start and end positions.
- ⑥ Click on the target region track for additional details, such as the start and end positions.

Coverage histogram

When you zoom in on the pileup, you see the coverage histogram and reads from the .bam track. The histogram in gray shows read depth at that location and reads from the .bam track show read direction and location of variants.



View the variants from an Ion AmpliSeq™ HD-Library Chemistry run

You can view a summary of data about the identified variants, and toggle to other views that provide more details about the same variants.

1. In the menu bar, click **Results** ▶ **Sample Results**.
2. Click a sample name to open the **Results** screen.
3. Click the **Variants** tab.
4. In the variants table, in the **Variant ID** column, click the identifier of the variant of interest.
5. Review detailed variant data and read coverage tracks.

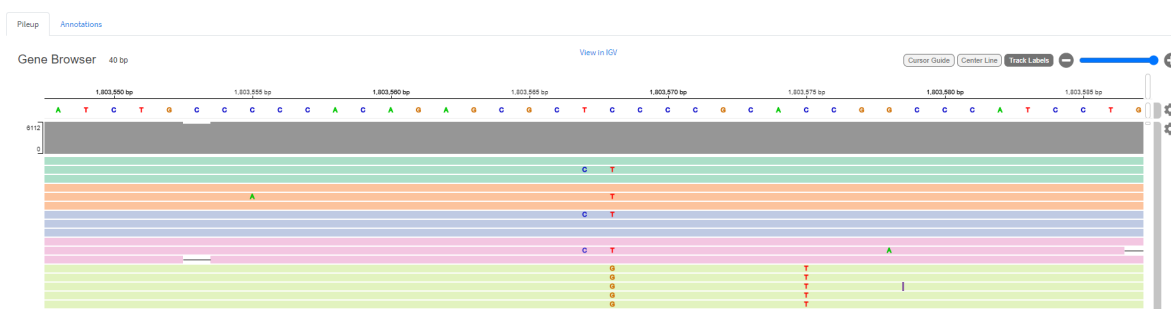


Figure 1 Example visualization

Ion AmpliSeq™ HD sequencing runs group consensus reads into families. A family is a group of reads that are associated with the same DNA molecule before library amplification. Each family is identified using the molecular tags, and consensus reads with the same molecular tags are grouped into the same family. The color of the consensus reads is used to indicate a family.

Within each read track, each nucleotide variant is indicated by a different color. T, A, C, and G are red, green, blue, and orange, respectively. An "I" denotes insertion, and white color with a dash indicates deletion.

6. You can sort, adjust, and view details about variants and base calls that are visualized in each read coverage track.

Option	Description
View the read coverage tracks sorted by family	Click ⚙️ (Actions) next to the read coverage track, then select Color ▶ Molecule .
Adjust the BAM tracks	Click ⚙️ (Actions) next to the read coverage track, then select an option to adjust the view of the track. For more information, see "Adjust pileup tracks" on page 144.
Review detailed data about a BAM read	Single-click the read track to get information such as the mapping quality, the strand, and the read base.
Review distribution of base calls at a selected position	Click the density plot (the gray bar at the top of the read coverage tracks) to view information about the total count, total reads, and total number of molecules, the distribution of single nucleotides at that position, and the number of insertions and deletions.

View variants with IGV

You can view data from Genexus™ Software with the Broad Institute Integrative Genomics Viewer (IGV). The viewer is available at the Broad Institute website: <http://software.broadinstitute.org/software/igv/home>.

1. In the menu bar, click **Results** ▶ **Sample Results**.
2. Click a sample name to open the **Results** screen.
3. Click the **Variants** tab.
4. In the variants table, in the **Variation ID** column, click the identifier of the variant of interest. The **Pileup** view is shown below the table.
5. Click **View in IGV** to download a JNPL file that can be opened in the Integrative Genomics Viewer (IGV) from the Broad Institute (see software.broadinstitute.org/software/igv).

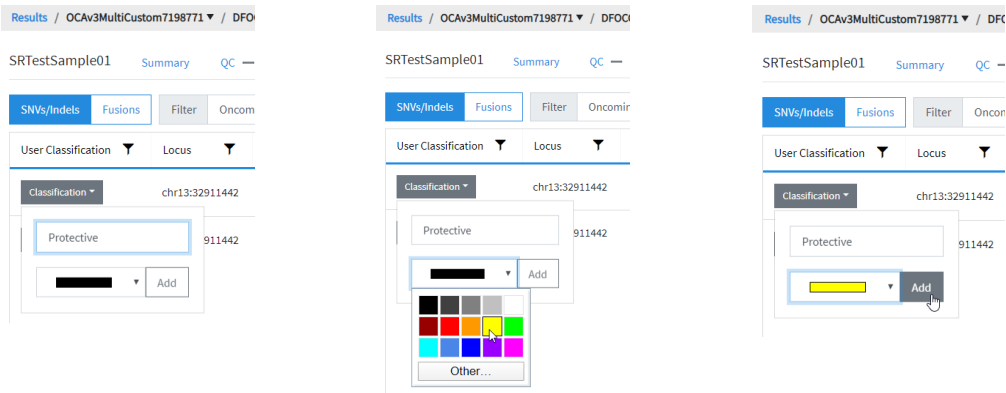
Create and assign variant classifications

You can create and assign user-defined variant classifications in the **SNVs/Indels**, **Fusions**, and **CNVs** tables in the **Variants** tab of the **Results** screen.

1. In the menu bar, click **Results** ▶ **Sample Results**.
2. Click a sample name in the **Sample Name** column in the row of a sample of interest to open the **Results** screen for the sample.
3. Click the **Variants** tab.
4. To refine the list of variants shown in the table, select a variant type to display: **SNVs/Indels**, **Fusions**, or **CNVs**.
5. In the **Variants** table, in the **User Classification** column, perform any of the following actions.
 - To assign an existing classification to a variant, select it from the list. The **Classification** menu changes to the name of the classification, as shown in this example.

The image displays three sequential screenshots of the Genexus software interface, illustrating the process of assigning a classification to a variant. The interface shows a table with columns for 'User Classification' and 'Locus'. The first screenshot shows the 'Classification' dropdown menu open, displaying a list of classification options: Benign, Likely Benign, Likely Pathogenic, Pathogenic, and Uncertain Significance. The second screenshot shows the 'Likely Pathogenic' classification selected, and the third screenshot shows the 'Classification' dropdown menu updated to 'Likely Pathogenic'.

- To create a new classification, enter a name for the classification in the text box, select a color for the new classification, then click **Add**.



- To remove a classification from a variant, click **X (Remove)**.
- To delete a classification from the list, click **Delete** next to the classification name. The classification will be removed from all variants in all results.

Filter results


You can filter results in the SNVs/Indels, Fusions, and CNV tables in the **Variants** tab of the **Results** screen in two ways. You can apply filters to columns of information that appear in the screen. The filters, available at the top of each column, immediately narrow the list of information in any columns to which filters are applied.

You can also apply a filter chain, a set of filters that Genexus™ Software uses to narrow the list of variants that are included in results. A manager-or administrator-level user creates filter chains from system-installed filters.


Search and filter the list of variants

You can search and filter to immediately narrow the list of results that are shown in the variant tables. You can apply filters to columns of information that appear in the screen. The filters, available at the top of each column, immediately narrow the list of information in any columns to which filters are applied.

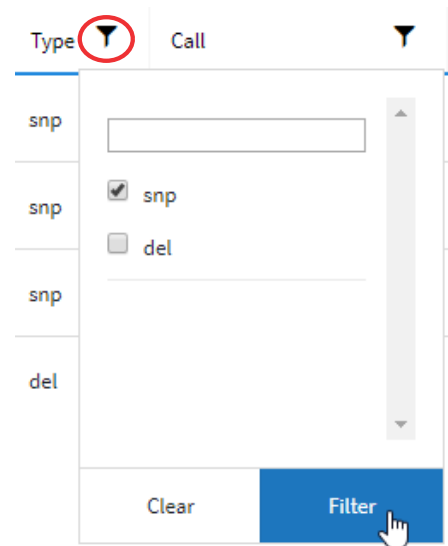
- In the menu bar, click **Results** ▶ **Sample Results**.
- Click a sample name in the **Sample Name** column.
- In the **Results** screen, click the **Variants** tab.
- Select the variant class to display the results: **SNVs/Indels**, **Fusions**, or **CNVs**.

- In the table of variants, in the column header of interest, click  (**Filter**).
 - In the search field, enter at least 3 characters, then click **Filter**.
 - Select the checkbox in the row of each filter that you want to apply, then click **Filter**.

The options that are available depend on the column and variant class. For example, you can filter data in the **Type** column to show one specific variant type.

- Click  **Clear Filters** to remove all filters and view the full list of run results.

The column or columns to which you applied a filter change to reflect the filter and selected options.



Filter results using a filter chain

You can filter the results that are listed in the **Results** screen with a system-installed or custom filter chain. A filter chain is a set of filters that Genexus™ Software uses to narrow the list of variants that are included in results.

Select a filter chain to change the list of variants that are included in the results. You can apply the filter chain temporarily, then review the results before you decide whether to save the updated results, or discard the changes.

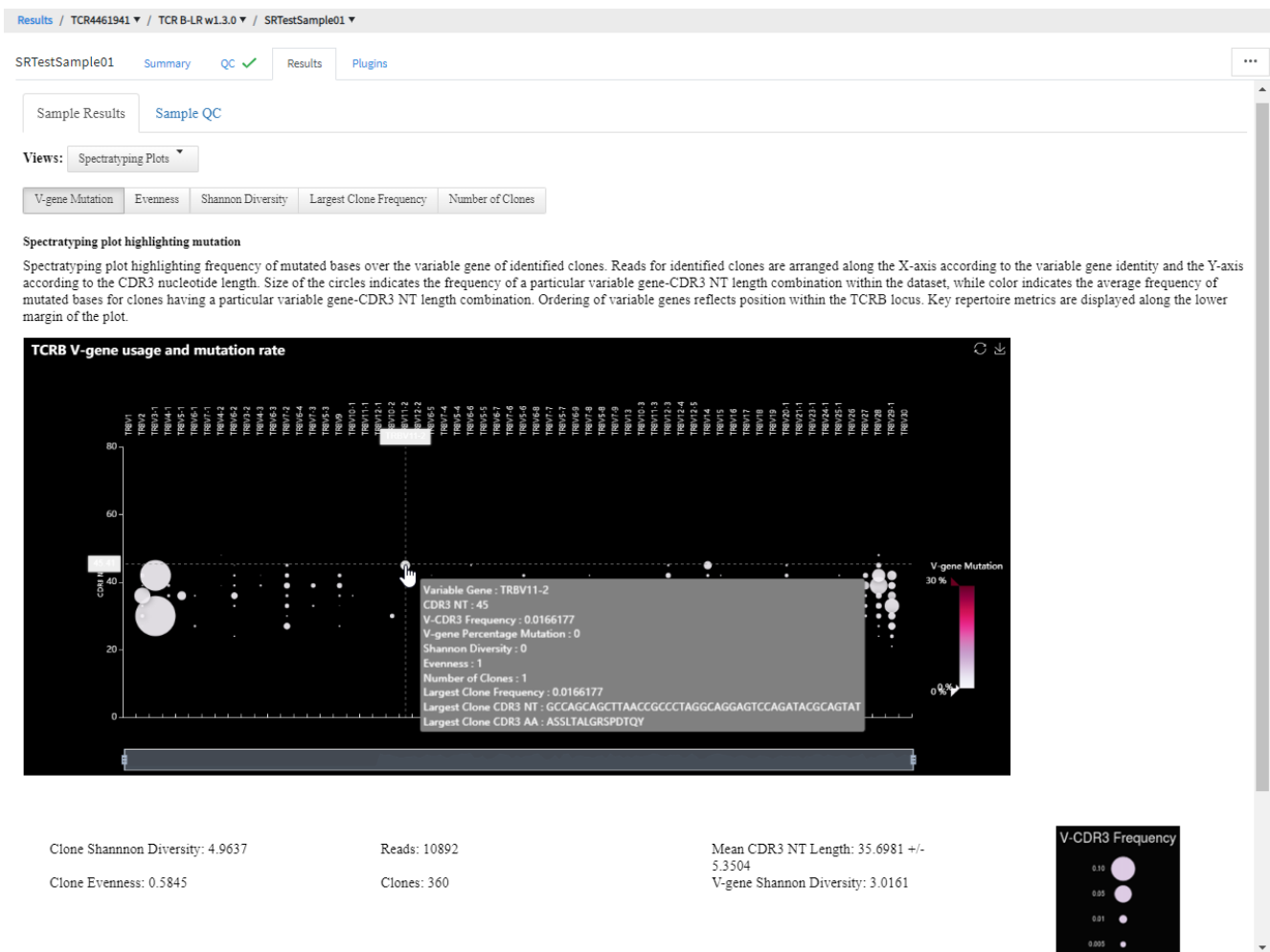
If you save the filter chain to a result, the variants that are included reflect the filtered results when the results are later opened.

For information about system-installed filter chains, see “System-installed filter chains” on page 351. Manager- and administrator-level users can create filter chains. For more information see, “Create a custom filter chain (manager/administrator)” on page 210.

- In the menu bar, click **Results** ▶ **Sample Results**.
- In the **Sample Results** screen, in the **Sample Name** column, select a sample of interest.
- In the **Results** screen, click the **Variants** tab.
- To refine the list of variants shown in the table, select a variant type to display: **SNVs/Indels**, **Fusions**, or **CNVs**.
- Above the variant table in the **Results** screen, click **Edit Filters**, then select a filter chain. The list of results changes to reflect the selected filter chain.
- (Optional) Click **Save** if you want the filter chain to be applied to the results when the results are later opened. The filter chain is selected and applied to the results when the results are reopened.

View OncoPrint™ TCR Beta-LR Assay GX results

- To view the OncoPrint™ TCR Beta-LR Assay GX expression data, in the **Results / Sample Results** screen, click the sample name in the **Sample Name** column, then click the **Results** tab. Results are viewed in two tabs: **Sample Results** and **Sample QC**.
- In the **Sample Results** tab, select from the following options in the **Views** dropdown menu.
 - Spectratyping Plots**
 - V-gene usage**
 - VJ-gene usage**
 - Clones detected per variable gene**
 - CDR3 histogram**
 - Clone Summary**



- In the **Sample QC** tab, select from the following options in the **Views** dropdown menu.
 - Read classification**
 - Proportion of full length, quality-trimmed, and reads lacking P1key, by read classification**
 - Base composition of overcalled and undercalled homopolymers**
 - Downsampling analysis**
 - QC metrics**

For detailed information on viewing and analyzing OncoPrint™ TCR Beta-LR Assay GX results, see the *OncoPrint™ TCR Beta-LR Assay GX User Guide* (Pub. No. MAN0018513).

Compare sample results

You can compare variant results across samples with Genexus™ Software. Comparisons between various results can be made.

- For information about the comparison of results from immune repertoire assays, see “Compare immune repertoire results” on page 159.
- For more information about the comparisons of variant results across all other assays, see “Compare variant results” on page 153.

Results generated using the system-installed OncoPrint™ TCR Beta-LR Assay GX or any other immune repertoire assay can only be compared to sample results from runs that use the same assay.

Compare variant results

You can compare variant results and create comparison plots from multiple sample results with Genexus™ Software. A description of the comparison of results from immune repertoire is not included here. For more information, see “Compare sample results” on page 153. Comparisons across samples or runs can be used to analyze results for a hotspot of interest. You can also view the trends of a reported variant over time.

The following comparisons can be made in Genexus™ Software:

- Comparisons of multiple samples, sequenced and analyzed with the same or different assays
- Comparisons of multiple samples over time, sequenced and analyzed with the same or different assays
- Comparisons of the results from one sample, sequenced and analyzed with different assays


If samples from different assays are compared, the research application (such as DNA or DNA and Fusions) must be comparable. That is, results generated from DNA-only assays cannot be compared to results generated from RNA-only or fusion-only assays. However, if the results from a research application include the type of results from another research application, the comparison can be made. That is, you can compare variant results generated from a DNA and Fusion assay to the variant results generated from a DNA-only assay.


View variant result comparisons

You can view variant comparisons in tabular format in **Fusions**, **SNVs/Indels**, and **CNV** comparison tables in the **Compare** screen.

1. In the menu bar, click **Results** ▶ **Sample Results**.
2. Click the checkboxes next to the sample names of interest.

Tip: In the **Sample Status** column header, click ▼ (**Filter**), then select **Completed** to limit the entries that are displayed on the **Sample Results** screen.

- Above the list of sample results, click  **Compare**.

Note:  **Compare** is not enabled until at least two samples are selected.

The **Compare** screen opens to the **Compare Samples** tables. The **OncoPrint™ Extended** filter is the default filter for the data displayed in the tables.

- (Optional) In the **Compare** screen, in the upper right corner, select a different **Filter Chain** to apply a different filter chain to the variants displayed in the tables. For more information, see “Filter results” on page 150.
- (Optional) Click **Export** to download an XLSX spreadsheet file of the variant comparison table.

Results / Compare

Compare Samples

⑤ SNVs/Indels

① Export

② ✓ SRTTestSample01 | 2021-03-26

③ ✓ SRTTestSample03 | 2021-03-26

④

⑥ Variant ID	⑦ Gene	⑧ AA Change	⑨ Call	Call
<input type="checkbox"/> COSM5673	CTNNB1	p.S33Y	PRESENT (HETEROZYGOUS)	PRESENT (HETEROZYGOUS)
<input type="checkbox"/> COSM6128	CTNNB1	p.S45del	PRESENT (HETEROZYGOUS)	PRESENT (HETEROZYGOUS)
<input type="checkbox"/> COSM763	PIK3CA	p.E545K	PRESENT (HETEROZYGOUS)	PRESENT (HETEROZYGOUS)
<input type="checkbox"/> COSM775	PIK3CA	p.H1047R	PRESENT (HETEROZYGOUS)	PRESENT (HETEROZYGOUS)
<input type="checkbox"/> COSM6252	EGFR	p.G719S	PRESENT (HETEROZYGOUS)	PRESENT (HETEROZYGOUS)

⑩

- Export:** Download a Microsoft™ Excel™ file of the sample comparison **Fusions** table.
- The QC status of the sample.
- The sample name.
- The date the sample was analyzed.
- The variant type. In this example: **SNVs/Indels**.
- Variant ID:** The name of the hotspot as defined in the Browser Extensible Data (BED) file.
- Gene:** The gene name.
- AA Change:** Identification of the amino acid change using Human Genome Variation Society (HGVS) nomenclature.
- Call:** Indicates the presence or absence of an SNV/Indel variant.
- Table slider. Use to extend table to view more samples.

The following table lists and describes the default information shown in the comparison tables.

Column	Description
Fusions comparison tables	
Variant ID	The name of the hotspot as defined in the BED file.
Oncomine Driver Gene	For fusion class variants, the 5' gene that drives proliferation of expression.
Read Counts	The frequency that the fusion was detected in the sample.
Call	The presence or absence of a fusion for the specific hotspot.
Read counts per million	The number of fusion read counts detected per number of total reads (in millions).
SNV/Indels comparison tables	
Variant ID	The name of the hotspot as defined in the BED file.
Gene	The gene name.
AA Change	Identification of the amino acid change using HGVS nomenclature.
Call	Indicates the presence or absence of an SNV/Indel variant. See SNV/Indel section for more information.
CNVs comparison tables	
Variant ID	The name of the hotspot as defined in the BED file.
Gene	The gene name.
Copy Number	The frequency that the fusion was detected in the sample. Note: Copy Number is not displayed for TNA CNV result comparisons.
Call	Indicates the presence or absence of a CNV.
CNV Ratio	Ratio of CNV amplicons after outlier removal.


Create time series graphs

You can create line graphs to view changes and trends in variant results over time.

1. In the menu bar, click **Results ▶ Sample Results**.
2. Click the checkboxes next to the sample names of interest.

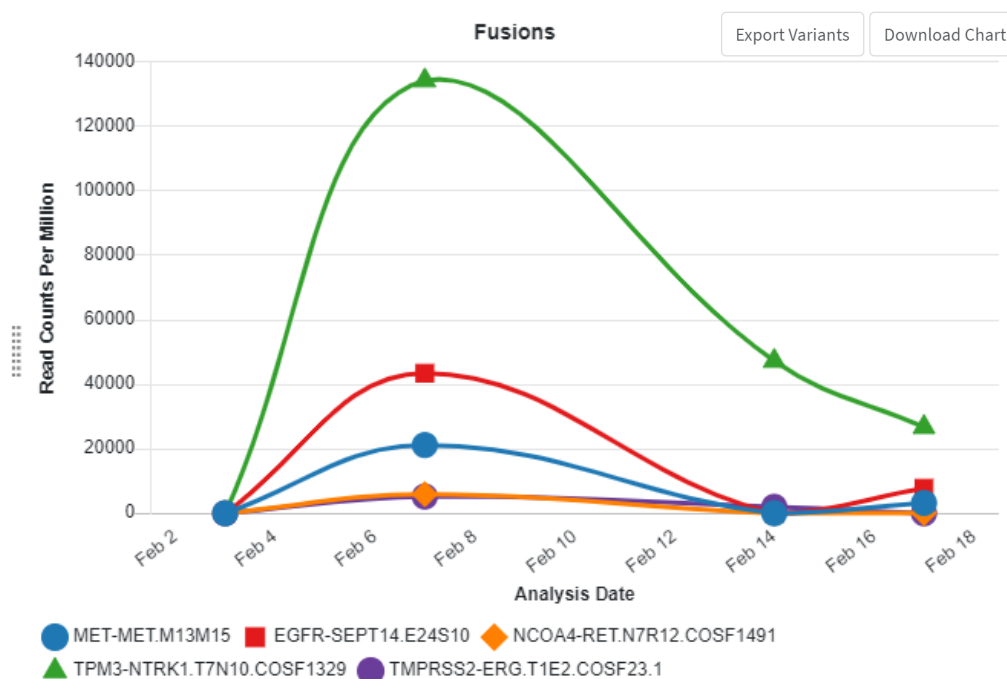
Note: Select **Filter Runs by ▶ Sample Status ▶ Completed** to limit the entries that appear in the **Sample Results** screen.

3. Above the list of sample results, click  **Compare**.

Note:  **Compare** is not enabled until at least two samples are selected.

The **Compare** screen opens to the **Compare Samples** tables. **Oncomine™ Extended** is the default filter for the data that appear in the tables.


- (Optional) In the **Compare** screen, in the upper right corner, select a different **Filter Chain** to apply a different filter chain to the variants that are shown in the tables. For more information, see “Filter results” on page 150.
- In the **Compare Samples** table, select the checkboxes next to the **Variant IDs** that you want to view in a plot.
The datapoints are added to the line graph with a live preview that is shown to the right of the **Compare Samples** table.
- Download a file of the line graph and the information in the variant table that was used to create the graph, if needed.
 - Click **Export Variants** to download in XLSX format, the information in the **Compare Samples** variant table for the selected variants.
 - Click **Download Chart** to download a PNG file of the time series line graph.




Create and customize comparison plots

- In the menu bar, click **Results** ▶ **Sample Results**.
- Click the checkboxes next to the sample names of interest.

Note: Select **Filter Runs by** ▶ **Sample Status** ▶ **Completed** to limit the entries that appear in the **Sample Results** screen.

- In the upper right corner of the screen, click  **Compare**.

Note:  **Compare** is not enabled until at least two samples are selected.

The **Compare** screen opens to the **Compare Samples** tables. **OncoPrint™ Extended** is the default filter for the data that appear in the tables.

4. (Optional) In the **Compare** screen, in the upper right corner, select a different **Filter Chain** to apply a different filter chain to the variants that are shown in the tables. For more information, see “Filter results” on page 150.
5. In the **Compare Samples** table, select the checkboxes next to the **Variant IDs** you want to view in a plot.
The datapoints are added to the line graph with a live preview that is shown to the right of the Compare Samples table.
6. (Optional) To customize the chart, click **Chart Options** in the upper right corner of the screen.
7. In the **Preferences** dialog box, change the parameters to customize the plot.

- **X-Axis Options**

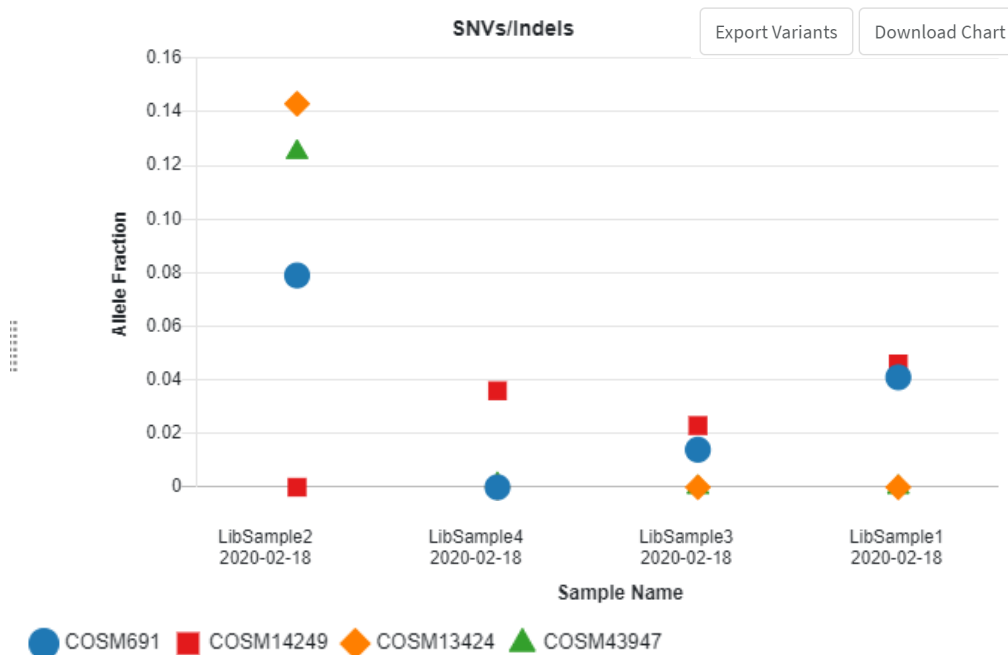
Option	Description
Sample Name	The unique identifier for a sample.
Analysis Date	The date of the sequencing run. The analysis date or run completion date is typically included in the Sequencing run details of the variant report.
Collection Date	The date that the sample was collected.

- **Y-Axis Options**

Option	Description
SNVs/Indels	See “View SNV/INDEL results” on page 131.
Fusions	See “View Fusion results” on page 134.
CNVs	See “View CNV results” on page 140.

Note: Y-Axis options depend on the assay or assays used to analyze the samples.


8. Click **Apply** to update the chart with the new selections.
9. Download a file of the image and the information in the variant table that was used to create the graph, if needed.
 - Click **Export Variants** to download in XLSX format, the information in the **Compare Samples** variant table for the selected variants.
The columns that are selected in **Table Columns** determine the headers that appear in the spreadsheet. If you change the column selections, you can download a new spreadsheet with updated headers.
 - Click **Download Chart** to download a PNG file of the comparison plot or time series line graph.




View and customize comparison tables

1. In the menu bar, click **Results** ▶ **Sample Results**.
2. Click the checkboxes next to the sample names of interest.

Note: Select **Filter Runs by** ▶ **Sample Status** ▶ **Completed** to limit the entries that appear in the **Sample Results** table.

3. In the upper right corner of the screen, click  **Compare**.

Note:  **Compare** is not enabled until at least two samples are selected.

The **Compare** screen opens to the **Compare Samples** tables. **OncoPrint™ Extended** is the default filter for the data that appear in the tables.

4. (Optional) In the **Compare** screen, in the upper right corner, select a different **Filter Chain** to apply a different chain to the variants that are shown in the tables. For more information, see “Filter results” on page 150.

5. (Optional) To customize the table or tables, click **Table Columns** in the upper right corner of the screen then use the checkboxes next to a table column option to add (select) or remove (deselect) the column to the table.

The available options depend on the assay or assays used to analyze the samples. For descriptions of options, see the following sections.

Option	Description
SNVs/Indels	See “View SNV/INDEL results” on page 131.
Fusions	See “View Fusion results” on page 134.
CNVs	See “View CNV results” on page 140.

Selected options are added as column headers to the **Fusions, SNVs/Indels, and CNV** comparison table with live preview.


6. Click **Table Columns** again to close the dialog window.
7. Click **Export Variants** to download an XLSX spreadsheet file of the Fusions, SNVs/Indels, or CNVs variant comparison table if needed.


Compare immune repertoire results

You can compare immune repertoire variant results with Genexus™ Software. A description of the comparison of results generated from all other assays is not included here. For more information, see “Compare sample results” on page 153.

Compare the immune repertoire between samples

Genexus™ Software can perform multi-sample (or cross-sample) analyses to compare the immune repertoire between samples.

1. In the menu bar, click **Results ▶ Sample Results**.
2. In the **Sample Results** screen, click the checkboxes next to the sample names of interest. Select **Filter Runs by ▶ Sample Status ▶ Completed** to limit the entries that are displayed on the **Sample Results** screen.
The immune repertoire comparison is only applicable to samples sequenced with the system-installed OncoPrint™ TCR Beta LR - GX5 - RNA assay (or any other custom or system immunology assay).
3. In the upper right corner of the screen, click  **Compare**.

Note:  **Compare** is not enabled until at least two samples are selected.

The **Overview of Repertoire Sequencing Results for Selected Samples** table is displayed.

Results / Compare

Search Go Join Type: CDR3 NT [Compare Samples](#)

Overview of Repertoire Sequencing Results for Selected Samples

Metrics indicate the total number of reads reported after data processing, the number of clones detected, and the Shannon Diversity and Evenness of detected clones. Click on sample name to view detailed report. Data visualizations, clone summary and repertoire metrics are available via Download button. To track clones across samples, use the checkbox to select two or more samples, then click Compare Samples.

<input type="checkbox"/>	Analysis	Sample	Barcode	Project	Reads	Clones	Shannon Diversity	Evenness	Sequenced On	Actions
<input type="checkbox"/>	R_2019_05_28_16_19_10_sierr... 12-VB12_TCR- Beta_Smin_extension_283May20...	JP_Sample3	IonDual_0122	R_2019_05_28_16_19_10_sierr... 12-VB12_TCR- Beta_Smin_extension_283May20...	253601	13682	9.4818	0.6901	May 29 2019 06:26 AM	Download
<input type="checkbox"/>	R_2019_05_28_16_19_10_sierr... 12-VB12_TCR- Beta_Smin_extension_283May20...	JP_Sample1	IonDual_0121	R_2019_05_28_16_19_10_sierr... 12-VB12_TCR- Beta_Smin_extension_283May20...	212790	11612	9.2409	0.6843	May 29 2019 06:26 AM	Download
<input type="checkbox"/>	R_2019_05_28_16_19_10_sierr... 12-VB12_TCR- Beta_Smin_extension_283May20...	JP_Sample2	IonDual_0124	R_2019_05_28_16_19_10_sierr... 12-VB12_TCR- Beta_Smin_extension_283May20...	266390	14113	9.3596	0.679	May 29 2019 06:26 AM	Download
<input type="checkbox"/>	R_2019_05_28_16_19_10_sierr... 12-VB12_TCR- Beta_Smin_extension_283May20...	JP_Sample4	IonDual_0123	R_2019_05_28_16_19_10_sierr... 12-VB12_TCR- Beta_Smin_extension_283May20...	241379	13160	9.4192	0.6883	May 29 2019 06:26 AM	Download

1 10 items per page 1 - 4 of 4 items

- (Optional) Click the hyperlinked Sample name to open the Immune Repertoire Results for that sample. See “View Oncomine™ TCR Beta-LR Assay GX results” on page 152 for more information.
- Click the checkboxes next to the analyses you would like to compare.
- In the dropdown menu at the top of the screen, select how you would like to compare clones across the sample. The options that are available depend on the immune repertoire assay.

Options	Description
CDR3 NT	Compare the clones based on the CDR3 nucleotide sequence. If the nucleotide sequence is shared across the samples, those are considered identical clones. This is the least stringent comparison option.
Variable Gene + CDR3 NT	Compare the clones based on the rearrangement of the variable gene plus the CDR3 nucleotide sequence.
Variable Gene + CDR3 NT + Variable Gene SHM Locations	Compare the clones based on the rearrangement of the variable gene plus the CDR3 nucleotide sequence plus somatic hypermutation (SHM) locations.
Variable Gene + CDR3 NT + Variable Gene SHM Locations + Isotype	Compare the clones based on the rearrangement of the variable gene plus the CDR3 nucleotide sequence plus somatic hypermutation (SHM) locations plus isotype. This is the most stringent comparison option.

- Click **Compare Samples** to generate a comparison table and plots.

Note: **Compare Samples** is not enabled until at least two samples are selected.

8. View variable comparison results in the **Comparison Table** tab.

Results / Compare

Comparison Table Comparison Plots

Search Go ②

Variable	Joining	CDR3 AA	CDR3 NT	R_2019_05_28_16_19_10_si...	R_2019_05_28_16_19_10_si...	R_2019_05_28_16_19_10_si...	R_2019_05_28_16_19_10_si...
① TRBV3-1	TRBJ2-1	ASSQLGEQF	GCCAGCAGCCAACAATTA...	0.1652714	0.1639175	0.1765607	0.1731592
TRBV3-1	TRBJ2-3	ASSQDGGQNTDTQY	GCCAGCAGCCAAGATGG...	0.0976337	0.111556	0.1141034	0.1087709
TRBV28	TRBJ1-2	ASSLHHSNYGYT	GCCAGCAGTTTACATCAC...	0.0263682	0.0240425	0.0236533	0.0267215
TRBV29-1	TRBJ2-2	SIHQNTGELF	AGCATCATAATCAGAAC...	0.0230953	0.0178862	0.0158377	0.0145415
TRBV7-9	TRBJ2-7	ASSRGPAYEQY	GCCAGCAGTCGAGGGCC...	0.0135291	0.0139386	0.0064417	0.0074447
TRBV28	TRBJ2-7	ASSFLGAGGLYEQY	GCCAGCAGTTTCCTTGGT...	0.0135291	0.0133606	0.0124404	0.0117367
TRBV29-1	TRBJ2-1	SVEDSLASGSGEQF	AGCGTTGAAGAITCTTTG...	0.0103825	0.0085906	0.0070686	0.0068109
TRBV2	TRBJ1-2	ASTEWSGNYGYT	GCCAGCACTGAATGGTCC...	0.0089116	0.0142535	0.0111303	0.0106844
TRBV11-2	TRBJ2-3	ASSLTALGRSPDTQY	GCCAGCAGCTTAACCGCC...	0.0084069	0.0069364	0.0080709	0.0085592
TRBV5-1	TRBJ2-2	ASSLGFNTGELF	GCCAGCAGCTTGGGATT...	0.0073344	0.009789	0.0081009	0.0089362

1 2 3 4 5 6 7 8 9 10 ... 10 items per page 1 - 10 of 43498 items

- ① Each row represents a clone.
- ② The frequency of the clone for the listed sample.

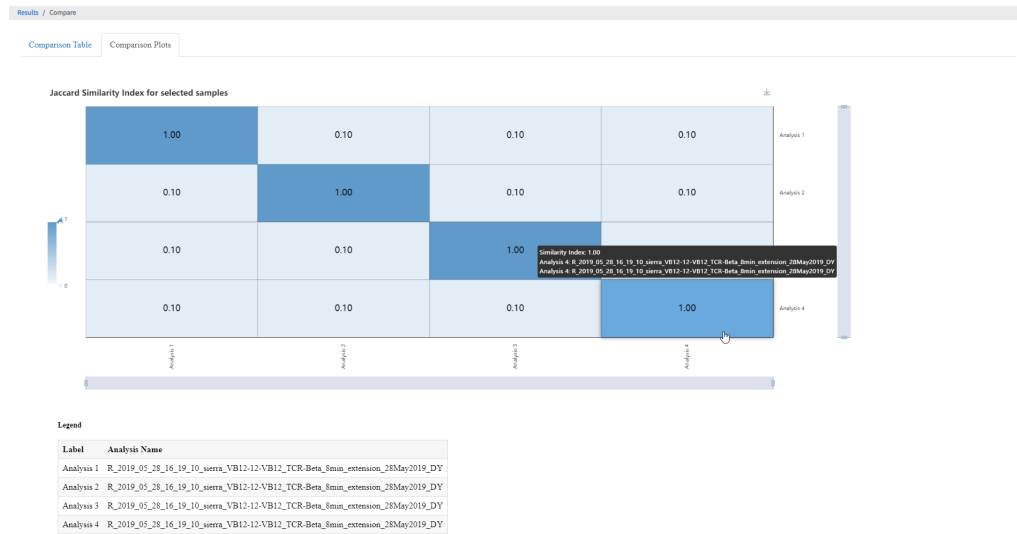
9. View comparison plots in the **Comparison Plots** tab.

Options	Description
Comparisons between 2 analyses.	An interactive scatter plot for the selected analyses is shown in the Comparison Plots tab.
Comparisons between more than 2 analyses.	A Jaccard Similarity Index for the selected analyses is shown in the Comparison Plots tab.

- The interactive scatter plot indicates the frequency of clones across two analyses. For more information, see “Example correlation and proportion of shared clones” on page 162.
- The Jaccard similarity index is determined for each pairwise comparison and is displayed in heat map format. For more information, see “Example Jaccard Similarity Index for selected samples” on page 162.

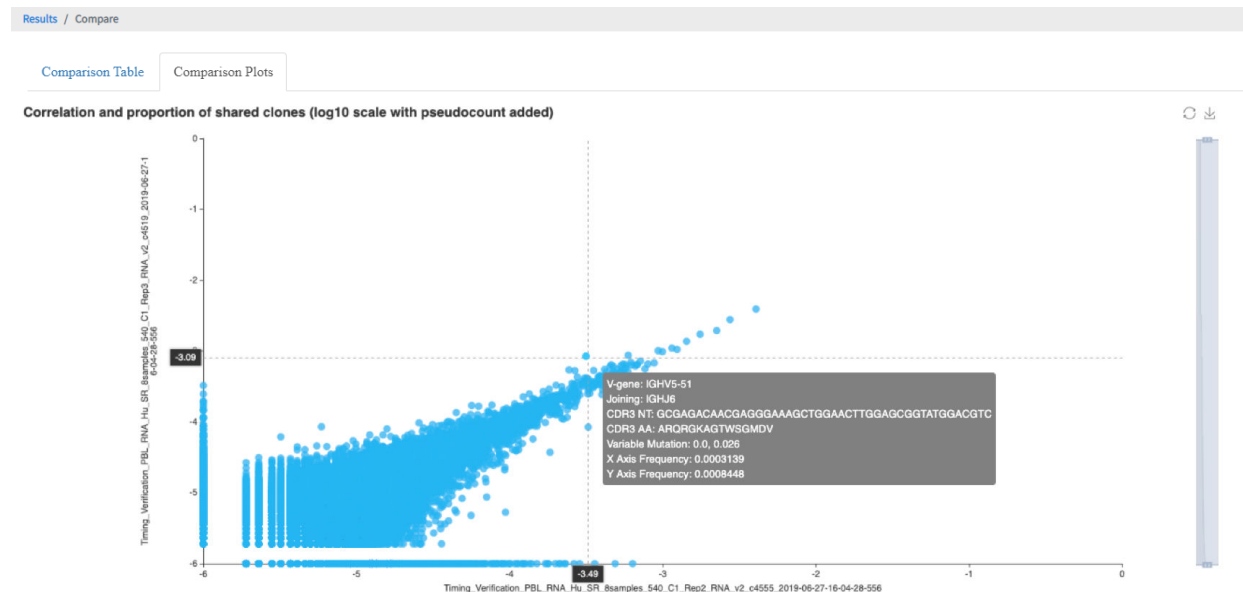
Example Jaccard Similarity Index for selected samples

This is an example of a **Comparison Plots** tab showing the Jaccard Similarity Index for selected samples. The Jaccard similarity index is determined for each pairwise comparison and displayed in heatmap form. The Jaccard Similarity Index ranges from 0 to 1 and is calculated as the total number of shared clones divided by the total number of distinct clones across two samples.



Example correlation and proportion of shared clones

This is an example of a **Comparison Plots** tab scatterplot. The illustration indicates the frequency of clones across two samples. Frequency values are log10 transformed with a pseudocount frequency of 1E-6 added to each value. Hovering over a point reveals the CDR3NT and AA sequence of a clone, the variable mutation, and the frequency in either sample.



View verification run results

Manager- and administrator-level users can view verification run details, including the run summary, QC summary, and the reagents used in the run. Verification runs are performed during installation to validate the performance of the instrument or instruments. Sequencing verification runs are performed to validate the Genexus™ Integrated Sequencer.

If a Genexus™ Purification Instrument is also connected, a verification run that validates both the purification instrument and sequencer is performed.

Note: Field Service Engineers use system-installed verification templates to validate the installation of the Genexus™ Integrated Sequencer. Field Service Engineers can view the template information but cannot modify it.

1. In the menu bar, click **Results ▶ Verification Results**.
2. In the **Verification Results** screen, view or download information for the verification run.

Option	Description
View verification results details	In the Run Name column, click the verification run name of interest to open the Verification Results Details screen. In the QC section of the Verification Results Details screen, click the > symbol to the left of the Lane column in the lane of interest to expand the results details pane.
Download a verification report that has been signed	Place the pointer over the row of the verification run of interest, then click Report .

3. You can perform the following actions for **Verification Results**. Action links are available when you place the pointer over the row of a verification result.

Option	Description
View Plan	If the run has not started on an instrument, you can view the run plan for the verification run.
Audit	Displays the list of users who created and edited the run. You can export and print information from the list from the Audit Trail dialog box.
CSA	CSA —Customer Support Archive. Click this link to download the sequencer log files. Log files contained within the CSA can be useful when troubleshooting problems with the sequencer. For more information, see “CustomerSupportArchive plugin” on page 316.
Report	Download a verification report that has been signed.

Verification runs


The following information is available in the **Results / Verification Results** screen.

Column	Description
Run Name	The name of the run, created when the run was planned. Click the name to open the Verification Results Details screen.
Assay Full Name	The name of the assay used in the run.
Field Engineer Name	The name of the support specialist who performed the run.
Instrument Name	The name of the sequencer that was verified.
Run Status	The current status of the full sequencing run, including analysis.
QC Status	Indicates whether a sequencing run passed or failed, based on the sequencing QC metrics selected for the assay.
Started On	The date and time that the run was started.
Updated On	The date and time that the run was last updated.

Sign verification run reports (manager/administrator)

Manager- and administrator-level users can sign results reports for verification runs. However, we recommend that only qualified support specialists sign performance qualification (PQ) reports.

Verification reports can be electronically signed only when all samples pass QC analyses.

1. Click **Results** ▶ **Verification Results**.
2. In the **Verification Results** screen, click the name of the verification run of interest.
3. In the **Verification Results Details** screen, click  **Sign Off**.
4. In the **Sign Off Report** dialog box, enter or make the following selections.
 - a. Enter the password.
 - b. In **Electronic Signature**, select **Approval**.
 - c. In **Sign off Comments**, enter a comment.
 - d. In **Laboratory Comments** and **Footer Field**, enter optional information.
5. Click **Sign Off**.

The verification run report is electronically signed.

You can view, print, or download the report in a PDF file. For more information, see “Verification runs” on page 164.

Run plugins after a sequencing run is complete

You can run plugins on your sequencing data after a sequencing run is complete using the **Run Plugin** function in the **Results** screen for the sample. Use this command to run one or more plugins on the data for the first time, or to rerun any plugin that has been run before (for example, if a plugin has been updated or failed during analysis).

1. In the menu bar, click **Results** ▶ **Sample Results**.
2. Click a sample name in the **Sample Results** screen.
3. Click ⋮ (**More Options**).
4. Click **Run Plugin**.
5. In the **Run Plugin** dialog box, select one or more assays in the run, then select the plugins to run on those assays.
6. Click **Run**.
While plugins are running, the **Run Plugin** link is unavailable for that run.
7. When analysis is complete, you can review the results. For more information, see Chapter 10, “Plugins for data analysis”.

Reanalysis

If a run fails to meet one or more QC parameters defined by the assay, you can adjust the assay parameters and reanalyze a run or a sample from the run. For more information to adjust assay parameters, see “Edit the QC parameters of a locked assay (manager/administrator)” on page 243 and Chapter 8, “Create and manage assays (manager/administrator)”.

Reanalysis of runs can start from the alignment, basecalling, or signal processing steps. A sample can be reanalyzed starting only from the alignment step.

For more information, see “Reanalyze a sample” on page 167 and “Reanalyze a run” on page 165.

Reanalyze a run

Runs can be reanalyzed using the same assay in Genexus™ Software. Reanalysis of runs can start from the alignment, basecalling, or signal processing steps. When you reanalyze a run, the reanalysis is applied to all samples in the assay.

Reanalysis from the	Description
Signal processing step	<p>The option to reanalyze a run from the signal processing step is only available when the following conditions are met.</p> <ul style="list-style-type: none"> • When a run fails during the basecalling step or earlier. • When the run has not already been successfully reanalyzed. <p>Reanalysis at the signal processing step uses DAT files.</p>

(continued)

Reanalysis from the	Description
Basecalling step	The option to reanalyze a run from the basecalling step is only available when a run or reanalysis successfully completes the signal processing step. Reanalysis at the basecalling step uses .wells files.
Alignment step	The option to reanalyze a run from the alignment step is only available when a run or reanalysis successfully completes the basecalling step. Reanalysis at the alignment step uses BAM files.

Note:

- Manager- and administrator-level users can reanalyze a sequencing run only if the run completed without any critical alarms or errors. If the run aborted or produced major alarms or errors, the run cannot be reanalyzed.
- QC parameters at the limits of stringency cannot be adjusted further. If samples fail QC and you cannot adjust the QC parameters further, you can sequence the sample library again. For information to find, recover, and purify the leftover library preparations, see the *Genexus™ Integrated Sequencer User Guide* (Pub. No. MAN0017910). Alternatively, you can prepare a new library for sequencing.

The files needed for the stage of reanalysis that you select must be present in the software. For example, if the .wells files for the run have been removed from the software, you cannot reanalyze from basecalling. Instead, reanalyze the BAM files at alignment. Administrator-level users can manage the settings and schedule to backup and delete files and data. For more information, see “Manage backup settings and restore runs (administrator)” on page 268.

You can reanalyze a run with any compatible assay that exists in the software. Alternatively, you can create a new assay or copy the original assay that was used in a run and modify assay parameters if needed. For more information, see Chapter 8, “Create and manage assays (manager/administrator)”.

1. In the menu bar, click **Results ▶ Run Results**.
2. In the **Results / Run Results** screen, in the **Run Name** column, click the run name of interest.
3. In the upper right corner of the screen, click **⋮ (More Options) ▶ Reanalyze**.
4. In the **Reanalyze** dialog box, enter or select the following information.
 - a. In **Reanalysis Run Name** field, enter a reanalysis run name.
 - b. In **Start Reanalysis from** dropdown list, select **Alignment, Basecalling** or **Signal Processing**.

- c. In the **Current Assay in Run** column, select the checkbox in the row of each assay that you want to reanalyze, then in the **Choose Assay to Reanalyze** column, select an assay that you want to use for each reanalysis from the dropdown list.

Reanalyze Sample

Reanalysis Run Name

Reanalysis Run 1

Start Reanalysis from

Alignment

Current Assay in Run

Choose Assay to Reanalyze

BRCA_Somatic_Custom

Oncomine BRCA - GX5 - Somatic - DNA - w3.6.0

Cancel Reanalyze

5. Click **Reanalyze**.

Follow the progress of the reanalysis in the **Results / Run Results** screen in the **Run Status** column and in the **Results / Sample Results** screen in the **Sample Status** screen. When reanalysis is complete, the new results can be viewed by clicking the run name corresponding to the reanalysis assay in the **Results / Run Results** screen. Runs that have been reanalyzed are appended with ↻ after the run name.

Reanalyze a sample

A sample can be reanalyzed starting only from the alignment step. Reanalysis at the alignment step uses BAM files. You can reanalyze all samples in a run from the basecalling or signal processing steps. For more information, see “Reanalyze a run” on page 165.

Note: Manager- and administrator-level users can reanalyze a sample only if the run completed without any critical alarms or errors. If the run aborted or produced major alarms or errors, the sample cannot be reanalyzed.

The option to reanalyze a sample from the alignment step is only available when a sample or reanalysis successfully completes the basecalling step.

You can reanalyze a sample with any compatible assay that exists in the software. Alternatively, you can create a new assay or copy the original assay that was used in a run and modify assay parameters if needed. For more information, see Chapter 8, “Create and manage assays (manager/administrator)”.

1. In the menu bar, click **Results ▶ Sample Results**.
2. In the **Sample Results** screen, place the pointer over the row of a sample of interest, then click **Reanalyze**.

Alternatively, you can reanalyze a sample when you click a sample name, then click **⋮ (More Options) ▶ Reanalyze**.

3. In the **Reanalyze** dialog box, enter or select the following information.
 - a. In **Reanalysis Run Name** field, enter a reanalysis run name.
 - b. In the **Current Assay in Run** column, select the checkbox in the row of each assay that you want to reanalyze, then in the **Choose Assay to Reanalyze** column, select an assay that you want to use for each reanalysis from the dropdown list.
4. Click **Reanalyze**.

Follow the progress of the reanalysis in the **Results / Sample Results** screen in the **Sample Status** screen. When reanalysis is complete, the new results can be viewed by clicking the sample name corresponding to the reanalysis assay in the **Results / Sample Results** screen. Samples that have been reanalyzed are listed in the table of sample results with the run name appended with ↻.

Download results files

You can download results files in Genexus™ Software.

1. In the menu bar, click **Results ▶ Sample Results**.
2. In the **Sample Name** column, click the sample name of interest.
3. Click **⋮ (More Options) ▶ Download Files**.
4. In the **Download Files** dialog box, select the files to download, then click **Download**.

The selected results files are downloaded in one ZIP folder. For information about the files, see “Results files” on page 169.

Results files

The following files can be downloaded from the **View Results** screen for each sample. The files that are available for download vary depending on the assay used. Results files include the sequencing data, results from the analyses, such as variant files, and audit and log files.

For a list and descriptions of plugin output files, see “Output files generated by the coverageAnalysis plugin” on page 305 and “Output files generated by the molecularCoverageAnalysis plugin” on page 312.

For instructions to download results files, see “Download results files” on page 168.

Option	File name	Description
Variants		
Filtered Variants (.vcf) ^[1]	<filter_name>_filtered.vcf	Summary of filtered variant results in variant call format (VCF).
All Variants (.vcf) ^[1]	Oncomine_<LibPrepID>_<analysisID>.vcf	Summary of variant results in variant call format (VCF).
Variant Summary (.tsv)	Summary.tsv	File that lists SNV/INDEL, copy number, and fusion results in tab-separated value format (TSV).
Snvindel (.tsv)	Snvindel.tsv	File that lists SNV/INDEL variant results in tab-separated value format (TSV).
Fusion (.tsv)	Fusion.tsv	File that lists fusion results in tab-separated value format (TSV).
CNV (.tsv)	Cnv.tsv	File that lists copy number variant results in tab-separated value format (TSV).
Sequencing Results		
DNA Unmapped Bam File (.bam)	<barcode>_rawlib.basecaller.bam	Unmapped DNA barcode BAM file; output after mapping reads to reference.
DNA mapped bam file (.bam)	merged.bam	Mapped BAM file of combined barcode reads.
DNA Mapped Bam Index File (.bai)	merged.bam.bai	Mapped BAM Index file.
DNA Basecaller FASTAQ File (.fastq)	<barcode>_rawlib.basecaller.fastq	FASTQ file of the DNA barcodes used.
DNA Processed Bam File	merged.bam.ptrim.bam	Mapped BAM file of combined barcode reads.
DNA Processed Bam Index	merged.bam.ptrim.bam.bai	Mapped BAM index file.
RNA Unmapped Bam File (.bam)	<barcode>_rawlib.basecaller.bam	Unmapped RNA BAM file; output of base calling, contains unmapped reads.

(continued)

Option	File name	Description
RNA Mapped Bam File (.bam)	<barcode>_rawlib. basecaller_alignments .bam	Mapped BAM file of combined barcode reads.
RNA Mapped Bam Index File (.bai)	<barcode>_rawlib. basecaller_alignments .bam.bai	Mapped BAM index file.
RNA Basecaller FASTAQ File (.fastq)	<barcode>_rawlib. basecaller.fastq	FASTQ file generated from the unmapped BAM file of the RNA barcodes used.
Test Fragment Basecaller FASTAQ File (.fastq)	rawtf.basecaller.fastq	FASTQ file for the test fragment.
Audit and Log		
Analysis Log File	analysis.log	Analysis log file.
Run Summary^[2]	Info.csv	Contains information about the run and analysis, including software version details, sample details, library details, run details, assay details, reagent and consumable information, run and sample QC metrics, and instrument summary.
Run Audit	PlannedRun-AuditTrail.pdf	Contains all audit records pertaining to the run.
Reports		
Report	<language>_ <sampleName>_<mode> _<templateName>_ <assayName>_<date>.pdf	A PDF report that contains sample-specific results. For more information, see “Variant report” on page 175.
Sample Summary^[3]	Info.csv	Contains information about the run and analysis, including software version details, sample details, library details, run details, assay details, reagent and consumable information, run and sample QC metrics, and instrument summary.
Troubleshooting Files		
Log Files^[4]	analysis.log	Analysis log file.
	summary.<timestamp>.log	Start and end time for each time an assay module is executed for the analysis.
	various.err various.out	Analysis pipeline logs used by field service engineers for troubleshooting.

(continued)

Option	File name	Description
Log Files ^[4]	Info.csv	Contains information about the run and analysis, including software version details, sample details, library details, run details, assay details, reagent and consumable information, run and sample QC metrics, and instrument summary.
	PlannedRun-AuditTrail.pdf	Contains the audit trail of the run plan in PDF format.
Other ^[4]	analysis.ini analysisSamples.json	Analysis configuration files, including secondary and tertiary INI files.
VCF Files	analysis.vcf	Summary of variant results in variant call format (VCF).

^[1] You can view the extracted files individually, or upload a VCF file to a software application that accepts VCF files, such as Ion Torrent™ OncoPrint™ Reporter software.

^[2] Files are available only for assays from Genexus™ Software version 6.2 and earlier.

^[3] Files are available for both custom and system installed assays in Genexus™ Software version 6.6.

^[4] Separate folders are generated for each sample. If included in the run, separate folders are also generated for an NTC and positive control.

Upload results files to Ion Reporter™ Software

When a run completes successfully in Genexus™ Software, and the results were not automatically uploaded to Ion Reporter™ Software, you can upload the results (BAM files) to Ion Reporter™ Software for further analysis.

You can also upload the samples to Ion Reporter™ Software, then use a custom Ion Reporter™ Software analysis workflow to view analysis results. Or, you can upload sample results files to run assays that are not yet available on the Genexus™ Software Integrated Sequencer.

Note: To automatically upload BAM files when a run is complete, select the **Upload BAM files to Ion Reporter™ Software** checkbox in the **Setup** step of planning a run.

Before you can upload results files to Ion Reporter™ Software, you must configure an Ion Reporter™ Software account.

- To configure an Ion Reporter™ Server account, see “Configure an Ion Reporter™ Server account (manager/administrator)” on page 254.
- To configure an Ion Reporter™ Software on Connect account, see “Configure an Ion Reporter™ Software on Connect account (manager/administrator)” on page 253.

1. In the menu bar, click **Results ▶ Run Results**.
2. In the **Run Results** screen, place the pointer over the row of a run of interest, then click **BAM Uploader**.
3. In the **Upload Samples to Server** dialog box, make the following selections.
 - a. From the **Ion Reporter™ Software Account** list, select the Ion Reporter™ Server account.


- b. In the **Select** column, select one or more assays from the list to upload the results files from the selected assays.
- c. In the **Ion Reporter Workflow** column, do one of the following:
 - Select **Upload Only** to upload the sample results (BAM files) Ion Reporter™ Software. You must sign in to Ion Reporter™ Software to launch an analysis.
 - Select an appropriate Ion Reporter™ Software analysis workflow to upload the sample results (BAM files) and automatically launch the selected analysis workflow in Ion Reporter™ Software. This results in an analysis in Ion Reporter™ Software.
- d. Click **Upload**.

The results are uploaded to Ion Reporter™ Software.

Tag an Ion Reporter™ Software analysis workflow for use with the IonReporterUploader plugin

If you configure Genexus™ Software for use with Ion Reporter™ Software, you can select an Ion Reporter™ Software analysis workflow to transfer sample results files to and launch an analysis automatically in Ion Reporter™ Software. To limit the number of analysis workflows that are listed as available in the Genexus™ Software, you can tag workflows for use with the IonReporterUploader plugin.

IMPORTANT! If you use the **Tag for IRU** option to tag analysis workflows for use with the IonReporterUploader plugin, you must tag *all* analysis workflows that you want to be available in Genexus™ Software. After you use the tag for the first time, only analysis workflows that include the tag are visible.

1. Sign in to Ion Reporter™ Software.
2. In the **Workflows** tab, click the row of the workflow of interest.
3. In the **Details** pane, click  **(Actions)** ▶ **Tag for IRU**.

Note: To remove a previously tagged workflow from the list of workflows that are available for selection in the Genexus™ Software, click  **Actions** ▶ **Untag from IRU**.

The analysis workflows are available for selection when you perform one of the following actions.

Action	Description
Plan a run	<ol style="list-style-type: none"> 1. In the Setup step of run planning, select Upload BAM files to Server, then select an Ion Reporter™ Software account and software version. 2. In the Samples step, the workflow that was tagged for use with the IonReporterUploader plugin is available for selection from the Ion Reporter Workflow dropdown list.
Upload sample results files to Ion Reporter™ Software after the sequencing run is complete	<ol style="list-style-type: none"> 1. In the Results / Run Results screen, place the pointer over the row of a run of interest, then click BAM Uploader. 2. In the Upload Samples to Server dialog box, the workflow that was tagged for use with the IonReporterUploader plugin is available for selection from the Ion Reporter™ Software Account dropdown list.

Upload results files to another Genexus™ Software server

When a run completes successfully in Genexus™ Software, you can upload the results (BAM files) to another Genexus™ Software server.

Tip: To automatically upload BAM files when a run is complete, select the **Upload BAM files to Server** checkbox in the **Setup** step of planning a run.

Before you can upload results files to Genexus™ Software, you must link a Genexus™ Software account that you can use for data uploads. For more information, see “Link Genexus™ Software accounts (manager/administrator)” on page 253.

1. In the menu bar, click **Results ▶ Run Results**.
2. In the **Run Results** screen, place the pointer over the row of a run of interest, then click **BAM Uploader**.
3. In the **Upload Samples to Server** dialog box, make the following selections.
 - a. From the **Configured Account** list, select the target Genexus™ Software server and version.
 - b. In the **Select** column, select one or more assays from the list to upload the results files from the selected assays.
 - c. In the **Genexus Workflow** column, select whether you want analysis to start on the target server immediately.
 - Select **Upload Only** to upload the sample results (BAM files) to Genexus™ Software.
 - Select an assay of interest to upload the sample results (BAM files) and automatically start analysis with the selected assay in the target server.
 - d. Click **Upload**.

The results are uploaded to another Genexus™ Software server.

Data files and flow

During a Genexus™ Integrated Sequencer run, raw data (DAT) files from the first flows are available. The Genexus™ Software starts processing the data, producing a 1.wells file. Basecalling is performed on the 1.wells file data, producing an unmapped BAM (uBAM) file. Subsequent analysis produces mapped reads (BAM) and variant calls (VCF) files.

The following table shows the flow of data and typical file size from runs that use a GX5™ Chip as Genexus™ Software processes data.

IMPORTANT! Data from approximately 200 sequencing runs can be accommodated on the sequencer before disk space becomes a limiting factor and data archiving is required. You can perform approximately 45–50 whole chip runs without deleting any wells files on the system. For more information about data archival of the analysis files, see “Manage backup settings and restore runs (administrator)” on page 268. Note that the 1.wells file is not archived during the archival process.

Process	File type	File size
Sequencing ^[1]	DAT	2 TB
▼		
Signal Processing ^[2]	Wells	250 GB
▼		
Base Calls (reads)	uBAM	15 GB
▼		
Mapped Reads	BAM	15–20 GB
▼		
Variant Calls	VCF	120 Kb

^[1] The sequencing raw data (DAT) files are deleted from the sequencer after run completion.

^[2] A disk space cleanup process is executed nightly to free up space for future runs and to ensure a 2TB hard disk space is maintained. If the system memory available for storing run analysis files reaches the 2TB threshold, the system auto-deletes 1.wells files for the oldest completed runs. If the system approaches the 2TB threshold, an alert notifies you when you sign in. If desired, you can back up 1.wells files to an external drive.

7

Create, view, and sign reports

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Genexus™ Software provides two types of reports.

- Variant report - A PDF report of the results for a sample in a run. You can customize the layout and contents of a variant report, or you can use a system-installed report template. Multiple users can sign the Variant report and one user can be designated to lock the report.
- Run report - A PDF report of the information that is shown in the **Run Report** tab. The run report includes assay metrics and the record of reagents that were used in a run. For more information, see “Assay metrics and the run report” on page 114.

To save or distribute a variant or run report, you can download the report.

Variant report



The variant report is a PDF report of the results for each sample in a sequencing run. You can use a system-installed report template. For system-installed report templates, the assay used in the run determines the data that are included in the report.

Alternatively, you can customize the layout and contents of a variant report.

To automatically generate a variant report for each sample during data analysis of a run, enable **Generate Report** in the **Setup** step when you plan the run (for more information, see Chapter 4, “Plan and manage runs”). To generate a variant report for each sample after a run is complete, see “Generate a variant report” on page 177.

When a variant report has been generated for a sample, it is available for download in two places:

- A link is available in the **Results / Sample Results** screen when you place the pointer over the row for that sample. Click the link to download the PDF.
- A **Download Report** button is available in the **Variant Report** pane in the **Reports** tab.

Variant reports can be electronically or manually signed by users. Electronically signed reports have  (**Sign off**) after the sample name in the **Sample Results** screen. The electronic signature is included in the footer of the report. If included, electronic signatures also appear in the Electronic Signature section of the report. A sample name followed by  indicates that the variant report is locked. For more information, see “Sign off on the run results” on page 178.


For information about the sections that can be included in a variant report, see “Sections of a variant report” on page 177.

View a variant report

You can view a variant report in Genexus™ Software. A variant report includes details of sample results.

You can download variant reports as PDF files. For more information, see “Download a variant report” on page 176.

A variant report must be generated before you can view the report. One or more variant reports are generated after a run completes successfully when a report template is set as a default template for the assay associated with the run. For more information, see “Create a report template (manager/administrator)” on page 182. Alternatively, you can generate one or more variant reports after a run completes successfully. For more information, see “Generate a variant report” on page 177.

1. In the menu bar, click **Results** ▶ **Sample Results**.
2. In the **Sample Results** screen, in the **Sample Name** column, click the sample name of interest.
3. Click the **Reports** tab.
A pane for the run report and each variant report that has been generated is shown. The name of the report is listed at the top of the pane. If a variant report has been signed at least once, the signature information is listed in the pane for the variant report. If a variant report is locked,  (**Lock**) is shown in the upper right corner of the pane.
4. In the pane of the report of interest, click **View Report**.

Download a variant report

You can download a variant report for a sample result of interest from the **Results / Sample Results** screen.

Note: The variant report is also available for download as part of the results files in the **Results** screen for a specific sample. For more information, see “Results files” on page 169.

1. In the menu bar, click **Results** ▶ **Sample Results**.
2. In the **Sample Results** screen, place the pointer over the row of the sample of interest, then click **Report**.
A ZIP file that contains the PDF report downloads automatically.
3. Extract the downloaded files, then open the PDF file in an appropriate viewer.

Generate a variant report

When generating a customized report, you can update any report template selections. For more information about results files, see “Results files” on page 169.

1. In the menu bar, click **Results** ▶ **Sample Results**.
2. In the **Results / Sample Results** screen, click the sample of interest in the **Sample Name** column.
3. Click the **Reports** tab.
4. In the **Variant Report** pane, click **Generate Report**.
5. In the **Generate Report** dialog box, change the name of the report that is generated by the software, if needed, enter an optional description, and select the report template and language of the report.
 - a. If the report template includes an image from the results, select the images to include in the report.
 - b. If the custom text that was designated as **Editable on Report Generation** when the report template was created, enter text if needed.

The **Report Template** list includes the report templates that are associated with the assay that was used in the run. For more information, see “Create a report template (manager/administrator)” on page 182.

If you have previously generated a variant report that has not been locked and then you select the same report template that was used to generate the variant report, the new report and any new selections you make override the previous variant report.

6. Click **Generate**.
A ZIP file that contains all the selected reports and other files is downloaded.

Reports that have been generated are available for download in the **Reports** tab and in the **Sample Results** screen.

Sections of a variant report

Manager- and administrator-level users can create custom report templates that can include various sections. Each section contains details about a result, so you can include information to meet laboratory requirements.

A variant report can contain the following sections and information, depending on the assay used.



Section	Description
Header	Information that repeats on each page of the variant report. Includes the title of the report, the organization address, an optional logo, and other optional fields about the sample.
Sample Details	The sample information that is entered into the software. You can customize the format of the Sample Details section when you create a new report template. To create a new report template, click Assays ▶ Manage Presets , then in the Report Templates tab, click + Add New .

(continued)

Section	Description
Variant details	Details about variants.
QC^[1]	<ul style="list-style-type: none"> • Run QC—a summary of the run quality control metrics • NTC QC – DNA—a summary of the DNA no template control quality control metrics • NTC QC – RNA—a summary of the RNA no template control quality control metrics • Sample QC – RNA—a summary of RNA sample quality control metrics • Sample QC – DNA—a summary of DNA sample quality control metrics • Templating Control QC – CF-1—a summary of the CF-1 quality control metrics
Sequencing Run Details^[1]	<p>Contains the following subsections:</p> <ul style="list-style-type: none"> • Assay Name—the assay used in the run • Panel—the panel used in the run • Started On—the date and time that the run was started • Completed On—the date and time when the run was completed • Operator—the user who sent the run to the instrument • Instrument—the name of the instrument • Serial Number—the serial number of the instrument • Filter Chain—the filter chain applied to results • Gene List—the gene list applied to the results
Reagents^[1]	Details about run consumables, including part numbers, expiration dates, and lot numbers.
Electronic Signatures^[1]	Includes information about one or more report signatures, including the name of the user who signed the report, the meaning the signature, the date, and comments.
Image	A section for an image or images. If the report template is associated with an assay, you can include in the variant report the images that are shown for the results in the software.
Footer	The footer is repeated on each page of the report.
Custom Text	A section for custom text. You can allow the text to be edited when the report is generated if desired.
Manual Signatures	A section for a physical signature of the printed report.

^[1] This section is locked and cannot be customized.

Sign off on the run results

Multiple users can sign variant reports. In the **Results / Sample Results** screen, a sample name followed by  indicates that the sample results are signed off by at least one user. A sample name followed by  indicates that the variant report is locked. Once a variant report is locked, the report is no longer available to be signed by any other user. The signature information appears in the variant report PDF file. For more information, see “Variant report” on page 175, and “Create a report template (manager/administrator)” on page 182.

Multilanguage support for PDF report generation is provided.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

You must have the permission to sign reports enabled in order to sign reports. An administrator-level user manages electronic signature permissions for user accounts. For more information, see “Add a new user account (administrator)” on page 257 or “Edit user details (administrator)” on page 258.

1. In the menu bar, click **Results** ▶ **Sample Results**.
2. In the **Sample Results** screen, in the **Sample Name** column, click the sample result of interest.
3. In the upper right corner of the screen, click ⋮ (**More Options**) ▶ **Sign Off**.
The **Sign Off Report** dialog box opens.
4. In **Password**, enter the password.
5. In the **Electronic Signature** dropdown list, select the meaning of the signature.
Administrator-level users configure the meaning of electronic signatures. One type of signature is designated to lock a variant report when the report is signed.
For more information, see “Manage electronic signatures (administrator)” on page 188.
6. In the **Report Template** dropdown list, select the report template that you want to use.
The option to select the report template is available only before the variant report has been signed by any user.
7. In **Sign Off Comments**, enter a comment.
8. In **Report Name**, change the name of the report if needed.
The option to change the report name is available only before the variant report has been signed by any user.
9. In **Select Your Language**, change the language of the report if needed.
The option to change the language is available only before the variant report has been signed by any user.
10. In **Footer Field**, enter any text that you want to appear in the footer of the PDF report pages.
If you entered footer information in the **Footer Field** when you created a report template, the same footer information appears in the **Electronic Signature** dialog box. You can enter new footer information to override the report template.
11. Click **Sign Off** to confirm the electronic signature.

The report is signed. If the signature is designated to lock the report, the variant report is signed and locked.


For information to download the signed report, see “Download a variant report” on page 176.

Send a notification to another user



You can send notifications to other users in Genexus™ Software. For example, you can notify another user when you have signed a variant report.

To receive email notifications, the recipient must have an email address that is configured in the software. For more information, see “Configure email notifications (manager/administrator)” on page 263.

1. In the menu bar, click **Results** ▶ **Sample Results**.
2. In the **Sample Results** screen, in the **Sample Name** column, click the sample result of interest.
3. In the upper right corner of the screen, click ⋮ **(More Options)** ▶ **Send Notification**.
The **Send Notification** dialog box opens.
4. In **Send Notification to**, select one or more administrator-level users of interest.
5. In **Comments** enter the message.
6. Click **Send** to send the message.

An email message with the text that you entered is sent to the users that you selected. In addition, when a recipient signs in to the software, an unread notification,  (Notifications), is shown. For more information, see “View notifications” on page 17.

Amend a locked variant report

If you have permission to sign reports, you can amend a variant report within 30 days after the report is locked. A sample that has a signed-off variant report is listed with the `sample name` followed by  **(Sign off)** and  **(Lock)** in the **Sample Name** column in the **Sample Results** screen. When you amend a locked report, you unlock the report and remove all signatures. You can then make changes to the report and start the signature and lock cycle for the variant report again.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

You must have the permission to sign reports enabled in order to sign reports. Any user with permission to sign a report can amend a report. An administrator-level user manages electronic signature permissions for user accounts. For more information, see “Add a new user account (administrator)” on page 257 or “Edit user details (administrator)” on page 258.

1. In the menu bar, click **Results** ▶ **Sample Results**.
2. In the **Sample Results** screen, click the signed off sample name of the sample of interest.
3. In the upper right corner of the screen, click ⋮ **(More Options)**, then click **Amend Report**.
4. In the **Sign Off Report** dialog box, enter the password.

5. In the **Electronic Signature** dropdown list, select the meaning of the signature.
6. In **Sign Off Comments**, enter a comment, then click **Amend Report**.

The variant report is no longer locked or signed.

Report templates (manager/administrator)

Manager- and administrator-level users create and manage custom report templates that are used to generate PDF reports after a run. Users select a report template when they plan a run.

View a report template (manager/administrator)

Manager- and administrator-level users can view custom and system-installed report templates in Genexus™ Software.

For more information about system-installed report templates, see “System-installed report templates” on page 368.

1. In the menu bar, click **Assays ▶ Manage Presets**.
2. In the **Manage Presets** screen, click the **Report Templates** tab.
Existing report templates are listed in this tab, including system-installed report templates. For more information, see “The report templates table” on page 181.
3. In the **Report Templates** table, place the pointer over the row of the report template that you want to view, then click **Preview**.
The **Report Template Preview** opens.
4. In the **Report Template Preview** screen, click **Open** to download a PDF file of the report template.
 - a. Click **Close** to close the **Report Template Preview** and return to the list of report templates.

You can place the pointer of the row of a report template to perform more actions, such as lock, edit, delete, or add notes to a report template. The options that are available depend on whether the report template is a draft or is locked.

The report templates table

You can view details about report templates in Genexus™ Software.

Details about report templates

Item	Description
Name	The name of the template. System-installed report templates are included with the software. Custom report templates are named by the person who creates the template.
Application Category	The application category that is associated with the report template, such as Cancer or All.

Details about report templates *(continued)*

Item	Description
Assay Name	If the report template is associated with an assay, the assay is listed. If no assay is listed, the report template is available to select when you generate a report for any result for samples with the same application category of the report template.
Status	Status of the report template. <ul style="list-style-type: none"> • Locked • Draft
Default	Indicates if the report template is designated as the default report template.
Created On	Date on which the report template was created.

Create a report template (manager/administrator)

Manager- and administrator-level users create custom report templates that are used to generate PDF reports after a run. As you create the report template, you can reorder sections, and add or remove sections. For more information, see “Sections of a variant report” on page 177.

To easily create a new report template, you can start from an existing report template and make changes to the layout and content of the sections if needed.

IMPORTANT! For a report template to be used in a run, it must first be locked.

1. In the menu bar, click **Assays ▶ Manage Presets**.
2. In the **Manage Presets** screen, click the **Report Templates** tab.
Existing report templates are listed in this tab, including system-installed templates.
3. Click **+ Add New**.
4. In the **Report Template** screen, enter a unique report template name, then complete the remaining sections.
 - a. In **Application Category**, select the application category for the report template.
The application category that you select determines the sample attributes that are available to add to the report template.

- b. In **Select Assay**, select the assay that you want to associate with the report.

Note: The application category and assay that you associate with the report template allows you to later select the report template when you generate a report from any result that is processed with the assay and application category to which your sample belongs.

If you select **None** for the assay, the report template is available to select when you generate a report from a result that was processed with any assay. That is, a report template that is associated with **Select Assay** of **None** is available to select for all results when you generate a report.

- c. In **Start From Existing Report Template**, select a report template to start from an existing report template if desired.
Only report templates that have the same application category that you selected in substep 4a are listed.

- d. In **Languages Setting**, select the language for the report.



- e. To set the new report template as default, keep the **Set as Default** toggle switch enabled.
When you designate a report template as default, a report is generated from the report template each time you complete a run with an assay that is associated with the report template. Each assay can be associated with 3 default report templates in various languages. If you do not designate a report template as default, you can generate reports from the report template after a run is complete in the **Reports** tab. For more information, see “Generate a variant report” on page 177.

Report templates that are not associated with an assay and are designated as default are used to generate a report each time a run completes that includes samples with the application category that you selected in substep 4a.

5. Add information to the **Header** section.

The Header and Footer are required sections. All other sections are optional. For more information about the sections that can be included in a report, see “Sections of a variant report” on page 177.

- a. Enter a report title and the organization address.
 - b. To include a logo on the report, in the **Logo** field, click **Select File** to browse to the logo file. Acceptable formats are PNG, GIF, and JPG.
 - c. In **Header Fields**, click the field, then in the list that appears, select a term to add it to the Header.
 - d. Enable or disable the **Report Date** and **Assay Name** checkboxes if needed.
6. In the **Configuration** pane on the left, you can add new sections, then move or delete sections from the template if needed.
 - a. In **Add Section**, select the section, then click **+ (Add)**.
The new section is added at the bottom of the list.
 - b. Use the following options to move or delete sections from the template.

Option	Description
Add multiple sections	<ol style="list-style-type: none"> 1. Click Bulk Add. 2. In the Bulk Add dialog box, in the Available Sections pane on the left, for each section to include, click the section name of interest, then click  (Add). Repeat for each section to include in the report template. 3. Click Apply.
Remove a section	In the box for the section, click ✕ (Remove) .
Remove multiple sections	<ol style="list-style-type: none"> 1. Click Bulk Add. 2. In the Bulk Add dialog box, in the Configured Sections pane on the right, click the section name of interest, then click  (Remove). Repeat for each section to remove from the report template. 3. Click Apply.
Add all sections	<ol style="list-style-type: none"> 1. Click Bulk Add. 2. In the Bulk Add dialog box, in the Available Sections pane on the left, click Select All. 3. Click Apply.
Remove all sections	<ol style="list-style-type: none"> 1. Click Bulk Add. 2. In the Bulk Add dialog box, in the Configured Sections pane on the right, click Select All. 3. Click Apply.
Rearrange sections of the report template	Click-drag the section boxes to rearrange the order.
Apply the gene list to the report template	Select Apply Gene List .


You can add the following items and sections to a report template multiple times. Each section, except for a page break, can be uniquely customized.

- **Variant Details**
- **Image**
If you select an assay in substep 4b, you can select from the images that are shown in the results for each assay that is used in the run.
- **Custom Text**
You can add text that can be edited when you generate a report. For more information, see “Generate a variant report” on page 177.
- **Page Break**

The following items and sections can appear in the report template in one place.

- **Manual Signatures**
- **Custom Text**
- **Sample Details**
- **QC**
- **Sequencing Run Details**
- **Reagents**
- **Electronic Signatures**

7. Enter and customize the information for each section of the report template.
 - a. For each unlocked section of the template, in the **Configuration** pane on the left, click the section name of interest.
 - b. In the **Section** pane on the right, add or remove information for the section.
You can click any customizable field to populate a list of available options for the field.

Sections that are locked and cannot be customized are designated with  (locked).

8. To preview the report layout, click **Preview**.
You must enter information in the required fields before you can preview a report template.
9. When you are finished, click **Save**.
The new report template appears in the **Report Templates** list.

Lock a draft report template (manager/administrator)

Manager- and administrator-level users use the tools in the **Assays / Manage Presets** screen under the **Report Templates** tab to manage report templates.

When a manager or administrator first creates an report template, the status of the report template is **Draft**. After a report template is locked, it cannot be edited or deleted.

1. In the menu bar, click **Assays ▶ Manage Presets**
2. In the **Manage Presets** screen, click the **Report Templates** tab.
3. In the **Report Templates** table, place the pointer over the row of the report template that you want to lock, then click **Lock**.

The report template is now locked and cannot be edited or deleted.

Edit a draft report template (manager/administrator)

When a manager- and administrator-level user first creates a report template, the status of the report template is **Draft**. While the report template is in draft status, it can be edited. A locked report template cannot be edited.

1. In the menu bar, click **Assays ▶ Manage Presets**
2. In the **Manage Presets** screen, click the **Report Templates** tab.
3. In the **Report Templates** table, place the pointer over the row of the report template that you want to edit, then click **Edit**.
4. Edit the sections of the report template as desired, then click **Save**.

The report template remains a draft until you lock the report template. For more information, see “Lock a draft report template (manager/administrator)” on page 185.

Delete a draft report template (manager/administrator)

When a report template is in draft status, it can be deleted from the software. Only draft report templates can be deleted.


1. In the menu bar, click **Assays ▶ Manage Presets**
2. In the **Manage Presets** screen, click the **Report Templates** tab.
3. In the **Report Templates** table, place the pointer over the row of the report template that you want to delete, then click **Delete**.
4. In the **Delete Report Template** dialog box, click **Yes** to confirm the deletion.

The draft report template is removed from the software.

View or add notes to a report template (manager/administrator)

Manager- and administrator-level can view existing notes or add new notes to draft or locked report templates in Genexus™ Software.

1. In the menu bar, click **Assays ▶ Manage Presets**
2. In the **Manage Presets** screen, click the **Report Templates** tab.
3. In the **Report Templates** table, place the pointer over the row of the report template of interest, then click **Notes**.

Action	Description
View existing notes for a report template.	In the Add Notes dialog box, notes that are associated with the report template are listed by user, date, and time.
Add a new note to a report template.	<ol style="list-style-type: none"> 1. In the Add Notes dialog box, click + Add Note. 2. Enter the note, then click Save. <p>The note is added to the report template and includes the username for the account of the person who entered the note, and a timestamp that records the note entry date and time.</p>
Delete a note from a report template	In the Add Notes dialog box, in the row of the note that you want to delete, click  (Delete).

Click **Close** to return to the list of report templates.

Configure electronic signatures (administrator)








Administrator-level users configure electronic signatures in Genexus™ Software. For more information about electronic signatures, see “Sign off on the run results” on page 178.

Manage electronic signatures (administrator)

Administrator-level users can add new types of electronic signatures and define the meaning of different types of electronic signatures Genexus™ Software. The signatures that are managed in this section are available in the **Meaning of Signature** list during report sign-off. For more information, see “Sign off on the run results” on page 178.

For example, to require three users to sign off on a report, you can add three different types of signatures, such as review, approve, and lock. Then, when manager- and administrator-level users provide their electronic signature on a variant report, they select from the types of signatures that you have created. Designate one of the signatures to lock the report during sign-off.

1. Click  **(Settings)** ▶ **Configuration**.
2. In the **Configuration** screen, in the **Manage Electronic Signature** section, manage signatures.

Option	Description
Add new types of electronic signatures.	In the Bulk add list of comma delimited meaning of signature field, enter new types of signatures, separated by commas. For example, enter <i>Review,Inconclusive</i> . Signatures that are added can then be selected in the Electronic Signature list during report sign-off. For more information, see “Sign off on the run results” on page 178.
Rename a type of signature.	<ol style="list-style-type: none"> 1. Click  (edit), then enter a new name for the signature. 2. Click  (save) to update the name of the signature.
Obsolete a type of signature.	Click  (obsolete) to remove the signature as an option during variant report sign-off.
Associate locking a report with a type of signature so that the signature indicates that a report is locked when that type of signature is selected during report sign-off.	Select the circular checkbox next to the type of signature that you want to use to lock a report during report sign-off.
Restore a type of signature.	Click  (restore) to restore a signature that has been made obsolete.
Review the changes made to a type of signature.	<ol style="list-style-type: none"> 1. Click  Audit Trail. 2. In the Audit Details screen: 3. <ul style="list-style-type: none"> • Review the audit trail details for the most recent change. • In the Audit Record Details pane, click a time stamp of interest to view the details for other changes that have been made to the selected signature. • Click  Download to export the record of the single change to a PDF file. • Click  Back to return to the Configuration screen.

Run report

The run report includes assay metrics and the record of reagents that were used in a run. For information about the contents of the run report, see “Assay metrics and the run report” on page 114. If you entered extraction kit barcodes for samples or control samples when you prepared library batches or when you planned the run, the extraction kit barcodes are listed in the run report.

View a run report

You can view or download a run report in Genexus™ Software.

For information to download a run report, see “Download a run report” on page 189.

1. In the menu bar, click **Results ▶ Sample Results**.
2. In the **Sample Results** screen, in the **Sample Name** column, click the sample name of interest.
3. Click the **Reports** tab.
A pane for the run report and each variant report that has been generated is shown. The name of the report is listed at the top of the pane.
4. In the **Run Report** pane, click **View Report**.

The run report opens in a new browser tab.

Download a run report

You can download a run report summary in PDF format.

1. In the menu bar, click **Results ▶ Sample Results**.
2. In the **Sample Results** screen, in the **Sample Name** column, click the sample name of interest.
3. Click the **Reports** tab.
Multiple panes including a **Run Report** pane, a **Variant Report** pane, and any panes for customized reports that have been generated are shown.
4. In the **Run Report** pane, click **Download Report** to download a run report summary in PDF format.



Create and manage assays (manager/administrator)

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About assays in Genexus™ Software

Assays contain the settings and parameters for library preparation, templating, controlling the sequencing run, analyzing, and reporting the results. Assays also define the panels, kits, and chips that are used in a run, and specify the reference files and threshold values for quality control and variant detection. The software files that contain the assay settings and parameters are packaged in a ZIP file called an assay definition file (ADF).

An assay is a reusable experimental design that contains predefined settings appropriate for use with common types of research applications. An assay can be used to plan many runs and plays an important role in enabling rapid throughput across the sequencing instrument. Assays help reduce the chance of errors, because information is stored and then applied to runs instead of entered manually for each run.

Custom assays can be created from system-installed templates, and you can copy a locked assay to create an assay, or copy and edit custom assays. You can also create custom versions of the system-installed presets that you can add to assays, including annotation sets, and copy number baselines, sequence variant baselines, exon tile assay baselines, and report templates.

Before you can create a custom assay, you must add a panel file, and hotspot and copy number baseline files (if needed for the assay), to the software. Custom assays are for advanced users. For help, contact a Field Service Engineer.

Find an assay

You can sort, search and filter information to narrow the list of assays shown in the **Assays / Manage Assays** screen, then find a specific assay in Genexus™ Software.

For information to search for an assay by a unique identifier, such as the name of the assay, see “Search for assays” on page 191.


For information to filter assays by a common trait, such as the name of the creator of the assay, see “Filter and search in the list of assays” on page 192.

Search for assays


You can search for assays in Genexus™ Software with keywords or text that are included in the name or full name of an assay.

IMPORTANT! Search terms must adhere to the following rules.


- An asterisk (*) is not allowed in the search field for use as a wildcard.
- Searches are case-insensitive, that is, both upper-case and lower-cases letters are found regardless the case of search term letters.
- Searches find every occurrence of a continuous string. For example, a search for *demo* in assay names returns a list of all assays with a name that includes *demo*. For example, demo1, demo2, and so on.
- Spaces are removed during searches, and are therefore not recommended. For example, a search for *demo 1* returns results only for data that includes the string *demo1*.

-
1. Click **Assays ▶ Manage Assays**.
 2. In the **Manage Assays** screen, in the **Assay Full Name** column header or **Assay Name** column header, click  (**Filter**).
 - a. In the search field, enter a full or partial assay name of at least 3 characters.
When you have entered at least 3 characters, the filter dialog box limits the list in the filter dialog box based on the entry.
 - b. In the filter dialog box, select from the limited list.
 - Select a checkbox next to the name of an assay to select one or more assays.
 - Click **Select All** to select all assays listed in the filter dialog box.

Note: If you click **Clear**, all selections are removed and the filter dialog box closes.

- c. In the filter dialog box, click **Filter**.
In the column header,  (**Filter**) changes color to indicate that a filter is applied.

The filtered list of assays is shown in the **Manage Assays** screen.

Click  **Clear Filters** to remove all filters and view the full list of assays.

Sort the list of assays

You can sort the list of assays to make it easier to find assays in Genexus™ Software.

1. Click **Assays** ▶ **Manage Assays** to open the **Manage Assays** screen.
2. Click a column header in the list of assays.
If the column is sortable, the contents of the column is sorted alphabetically, or chronologically.
3. Click the column header again to reverse the order.

Filter and search in the list of assays

You can apply filters to narrow search results, or shorten the list of assays when you view the list of assays in Genexus™ Software.

You can also search for assays in Genexus™ Software with keywords or text that are included in an assay name.

IMPORTANT! Search terms must adhere to the following rules.

- An asterisk (*) is not allowed in the search field for use as a wildcard.
 - Searches are case-insensitive, that is, both upper-case and lower-cases letters are found regardless the case of search term letters.
 - Searches find every occurrence of a continuous string. For example, a search for *demo* in assay names returns a list of all assays with a name that includes *demo*. For example, demo1, demo2, and so on.
 - Spaces are removed during searches, and are therefore not recommended. For example, a search for *demo 1* would return results only for data that includes the string *demo1*.
-

1. Click **Assays** ▶ **Manage Assays** to open the **Manage Assays** screen.
2. Click the **Filter Assays By** dropdown menu, then apply the filters.
 - Select **IRU Shortlist** to limit the assays shown to the assays that are tagged for use with the IonReporterUploader plugin.
 - Select **Status** to expand the list of options for status, then select a status:
 - Locked & Draft
 - Obsolete
 - Locked
 - Draft
 - Select **All** to return to the complete list of assays.

3. Select from the filter and search options in the column header of interest.

Option	Description
Filter and search by name of an object	<p>Click Filter in the column header of interest, then select one or more of the search options.</p> <ul style="list-style-type: none"> Enter at least 3 characters to search by the full or partial name for the object. Select one or more matches from the filter dialog box. To select all matches, click Select All. Next, click Filter. The filtered list of assays is shown in the Manage Assays screen.
Filter and search by research application	<p>Click Filter in the Research Application column, then select one or more of the search options.</p> <ul style="list-style-type: none"> Enter at least 3 characters to search by the full or partial research application name. Select one or more matches from the filter dialog box. To select all matches, click Select All. Next, click Filter. The filtered list of assays is shown in the Manage Assays screen. Select from the research applications shown, then click Filter.
Filter and search by the creator of the assay	<p>Click Filter in the Created By column, then select one or more of the search options.</p> <ul style="list-style-type: none"> Enter at least 3 characters to search by the full or partial assay name. Select one or more matches listed in the filter dialog box. To select all matches, click Select All. Next, click Filter. The filtered list of assays is shown in the Manage Assays screen. Select from the user names that are shown, then click Filter.
Filter by the exact or approximate date on which assays were created	<p>Click Filter in the Created On column, then use the options to filter by a date or range of dates to filter the list based on the assay creation date.</p>

4. (Optional) Click  **Clear Filters** to return to the entire list of assays.

The list of assays is filtered according to the selections.

Assay creation overview

You can view the assay parameters and other components of the assays in Genexus™ Software. For more information, see “View assay details” on page 238 and “Assay details” on page 238. Manager- and administrator-level users can create custom assays if system-installed assays do not meet needs.

To create a custom assay:

- Copy an existing system-installed assay or a locked custom assay, then edit the settings of the assay to create a new custom assay. For more information, see “Copy an assay (manager/administrator)” on page 196.
- Use a template to create a custom assay that contains preconfigured settings. The template includes settings for various genetic research analysis applications. For more information, see “Create an assay (manager/administrator)” on page 194.

After you create an assay, you can optimize the assay configuration if needed. You can plan a run with a draft assay, view the results, then change the configuration and plan new runs before you lock the draft assay. For more information, see “Lock a draft assay (manager/administrator)” on page 242.

Create an assay (manager/administrator)

Manager- and administrator-level users can create a new assay from a system-installed template in Genexus™ Software. Each template allows you to select kits, chips, primer panels, and adjustable Quality Control (QC) thresholds and other parameters that are appropriate for the assay type. The assay is then available to select when you plan a run.

IMPORTANT! Use the default settings for system-installed assays unless you are an advanced user. Contact your Field Bioinformatics Specialist (FBS) for assistance.

1. In the menu bar, click **Assays ▶ Create Assay**.
The **Create Assay** screen opens.
2. Click the assay type for the template that provides the appropriate kits, chips, primer panels, and adjustable QC thresholds and read mapping parameters.

Assay type	Description
Generic Sequencing Application	Use this template to create a generic sequencing assay with no application-specific post-run analysis. The assay generates BAM files.
Non-Human Reference	Use this template to create an assay that detects germline variants. Annotations are not included in the results.
TNA	Use this template to create an assay that detects and annotates DNA and Fusion variants in total nucleic acid (TNA) samples
Fusions	Use this template to create an assay that detects and analyzes variants in fusion samples.
DNA Germline	Use this template to create an assay that detects and annotates germline variants in DNA samples.
DNA and Fusions	Use this template to create an assay that detects and annotates somatic DNA variants and RNA fusion targets in a sample. The DNA and Fusions template creates a DNA-only, a Fusion-only, and a DNA and Fusions application type for the assay. Then, when you plan a run, you can select one or more application types of the assay to use in the run.
DNA Somatic	Use this template to create an assay that detects and annotates somatic variants in DNA samples.

3. In the **Panel** step, select a panel and other options for the assay. For descriptions of the panel information appropriate for the assay, see “Panel step assay options” on page 197.
4. Click **Next**.

5. In the **Reagent** step, select the kits and chips that you are using. Ensure that the reagents are appropriate for the primer panel that you are using.
For descriptions of the reagent information that is available for the assay, see “Reagent step assay options” on page 199.
6. Click **Next**.
7. In the **Parameters** step, accept the recommended default values, or adjust the values within the allowed ranges, then click **Next**.
You can set the parameters by uploading an advanced parameters configuration file that overrides the default values in the template.
 - a. At the bottom of the **Parameters** step, click **Download** to download a ZIP file of the JSON template that is appropriate for the assay.
 - b. Edit the file with custom settings, then save the file to the computer.
 - c. At the bottom of the **Parameters** step, click **Upload**.
 - d. Click **Select files**, navigate to the file on the computer, then click **Open**. Click **Upload** to upload the file and override the default values in the template.

For more information about parameters, see “Parameters step assay options” on page 201.

8. In the **QC** step, enter QC metric values, then click **Next**.
For descriptions of the QC information for the assay, see “QC step assay options” on page 203.
9. In the **Plugins** step, select one or more plugins to run with this assay, then click **Next**.
For more information, see Chapter 10, “Plugins for data analysis”.
10. In the **Save** step, enter an assay name, an assay full name, and an optional description, then click **Save**.
The types of runs that the assay is compatible with are listed in the **Save** step.

Assays / Create Assay

- ✓ Panel
- ✓ Reagent
- ✓ Parameters
- ✓ QC
- ✓ Plugins
- Save

Save

Assay Name *

 ✓

Assay Full Name *

 ✓

Description

Run Workflows: Library to Result, Nucleic Acid to Result

Exit Previous Save

The new assay is listed in the **Manage Assays** screen. The **Assay Full Name** is followed by **Draft** to indicate that the assay is in draft status. You can plan a run with the draft assay and the assay is available to lock, edit, or delete, and audit. You can also download parameters from assays in **Draft** or **Locked** statuses.

Copy an assay (manager/administrator)

Manager- and administrator-level users can create a new assay by copying an existing system-installed assay or other custom assay and modifying it if needed. Only locked assays can be copied.

1. In the menu bar, click **Assays ▶ Manage Assays**.
2. In the **Manage Assays** screen, place the pointer over the row of the assay that you want to copy, then click **Copy**.
The **Copy Assay** screen opens to the **Panel** step. The assay settings can be modified for the new assay.
3. Proceed through the workflow steps, and modify assay settings if needed. For more information, see “Assay options” on page 197.
4. In the **Save** step, enter a new assay name, an assay full name, and an optional description, then click **Save**.

The newly created assay is added to the list of assays in the **Assays / Manage Assays** screen. The **Assay Full Name** is followed by **Draft** to indicate that the assay is in draft status. You can plan a run with the draft assay. The assay remains in draft status until it is locked. You can lock the draft assay to ensure that the assay cannot be edited. For more information, see “Lock a draft assay (manager/administrator)” on page 242.

Assay options

Panel step assay options

Manager- and administrator-level users create assays in the Genexus™ Software.

In the **Panel** step, you can select the appropriate reference files to use in an assay.

The following table lists options that are available in the **Panel** step. The available options depend on the type of assay

IMPORTANT! Use the default settings for system-installed assays unless you are an advanced user. Contact your Field Bioinformatics Specialist (FBS) for assistance.

Option	Description
Panel	<p>The panel to use in the assay. The remaining options are appropriately populated based on the selected panel.</p> <p>The panel is a tab-delimited BED file that defines the coordinates of the regions of interest to be amplified with the selected primer set.</p>
Hotspots	<p>The hotspots file to use in the assay. The hotspots file is a tab-delimited BED file that indicates the positions where a DNA variant is expected to occur. It also annotates these positions with the reference allele, variant allele, and anchor base.</p>
Library Chemistry	<p>The type of library chemistry to use in the assay.</p>
Extraction Method Type	<p>The method used to extract the DNA, RNA, or DNA and RNA from the sample, or as a single library TNA from the sample.</p> <p>Available options depend on the panel that is selected.</p>
Minimum Read Count Per Sample	<p>The minimum number of reads per sample that are needed to achieve the coverage depth required in an assay.</p> <p>This value, usually associated with the panel, determines the number of samples that can be run on the lanes that are available on a chip. The system uses this value to calculate how many lanes a run uses and assigns samples accordingly. For example, a minimum read count per sample of 1,000,000 allows twice as many samples on the same lane as that of 2,000,000.</p>
Target Regions	<p>The target regions BED file that is appropriate for the sample. Depending on the application, you may need to select a separate DNA and Fusions Target Regions file.</p> <p>Check with a Field Service Engineer for updates to ensure that the most current files are being used.</p>

(continued)

Option	Description
Genome Reference	<p>The genome reference file for the panel that is selected is displayed. When you create or upload a panel, you select the genome reference. For more information, see “Add new panel files (manager/administrator)” on page 284.</p> <p>The hg19 and GRCh38 and genome references are installed with the system.</p>
Run CNV	<p>When Run CNV is enabled, the software analyzes CNV results based on the selected CNV baseline file. You must select a baseline file from the dropdown list to detect copy number variants (CNVs). If the default setting, Do Not Run CNV Algorithm, is selected in the dropdown list, the software does not run the CNV algorithm even if Run CNV is enabled.</p>
Copy Number Baseline for CNV Calling	<p>You must select a baseline file from the dropdown list to detect copy number variants (CNVs).</p> <p>The copy number baseline is a set of control samples that are used to create a baseline for the detection of copy number variants (CNVs). You can use system-installed baselines, import or create a baseline, or copy and edit an existing baseline. You can view the settings that are used for the baseline in the Assays/Manage Presets/Copy Number Baselines tab. For more information, see “Copy number baselines (manager/administrator)” on page 218.</p>
CNV Calling method^[1]	<p>Select either Mosaic, Full Ploidy, or both.</p> <p>When mosaicism detection is enabled, the software can make non-integer ploidy calls. CNV events are reported as a decimal ploidy value instead of an integer value.</p> <p>CNV calling with Full Ploidy includes read coverage correction for GC bias. Corrected coverage is compared to a baseline coverage from control samples of regions with known expected normal ploidy. That is, 2 on autosomes and X in females, and 1 on sex chromosomes in males.</p>
Adjust Sample Copy Number by Cellularity^[2]	<p>The % cellularity that is the minimum value that the assay requires of a sample. The assay analysis adjusts all copy numbers based on the % cellularity that is selected.</p> <p>Click Adjust Copy Number by Sample Cellularity to enable the adjustment in the software. When the adjustment is enabled, the toggle switch is shown in blue.</p> <p>You must select the adjustment percentage in the dropdown list.</p> <p>If the default setting, Do Not Adjust CN and Do Not Reject Samples, is selected in the dropdown list, the software does not adjust the sample copy number even if Adjust Copy Number by Sample Cellularity is enabled.</p>
Fusion Reference	<p>The tab-delimited BED file to use in the assay that indicates the positions where an RNA fusion target is expected to occur. It also annotates these positions.</p>

(continued)

Option	Description
Fusion Panel	The tab-delimited BED file to use in the assay that defines the coordinates of the regions of interest to be amplified with the selected primer set.
Fusion Baseline	The fusion baseline used in the assay.
Primer Pools	The number of primer pools used in the assay.
Annotation Sets	The annotation set to use in the assay. Some annotation sets are system-installed. Custom annotation sets can be created as required. For more information, see “Create a new annotation set (manager/administrator)” on page 215.
Apply OncoPrint Variant Annotations ^[3]	When you select the Apply OncoPrint™ Variant Annotations option in the Panel step to create or edit an assay, analysis results data from more than 24,000 exomes across solid tumor and hematological cancer types is used as a reference to annotate variants that are relevant to cancer. The annotations are shown in results in the OncoPrint™ Gene class and OncoPrint™ Variant class columns. For more information, see “Annotation criteria for OncoPrint™ Variant Annotator” on page 340 or contact a support representative, Field Bioinformatics Specialist (FBS), or Clinical Account Consultant (CAC).

^[1] This option is only available for **Aneuploidy** assays.

^[2] This option is only available for assays that use a **DNA and Fusions** panel or a **DNA** panel.

^[3] This option is available only for assays that use custom Ion AmpliSeq™ HD and OncoPrint™ panels.

Reagent step assay options

Manager- and administrator-level users create assays in Genexus™ Software.

In the **Reagent** step, you select the kits and chips to use in an assay. Ensure that the kits and chips that you select are appropriate for the primer panel that you are using.

The following tables list options that can be available in the **Reagent** step. The available options depend on the type of assay.

IMPORTANT! Use the default settings for system-installed assays unless you are an advanced user. Contact your Field Bioinformatics Specialist (FBS) for assistance.

Purification

Option	Description
Purification Kit	The purification kit that is used for nucleic acid purification.

Library Preparation

Option	Description
Library Kit	The library kit that is used to prepare the library.
Barcode Kit	The barcode kit to use in the assay.
Include Inline controls	<p>You can select DNA or RNA, or both DNA and RNA, depending on the assay template that is used. For example, you might want to ensure that both DNA and RNA are selected if you use the DNA/RNA template.</p> <p>By default, only DNA is enabled and RNA is disabled. RNA is available for selection only if the assay template includes it.</p> <p>TNA is an option for TNA assays.</p>

Templating

Option	Description
Sequencing Chip ^[1]	The sequencing chip to use in the assay.
Templating Kit ^[2]	The templating kit to use in the assay.
Template Size (bp)	<p>The template size, measured in base pairs, to use in the assay.</p> <ul style="list-style-type: none"> • 200 • 400

^[1] When you change the Sequencing Chip, other assay parameters are changed to the default settings that are associated with the chip. Review the settings in the Parameters step after you make this change.

^[2] When you change the Templating Kit, other assay parameters are changed to the default settings that are associated with the templating kit. Review the runparameter.json after you make this change.

Sequencing

Option	Description
Sequencing Kit	The sequencing kit to use in the assay.
Sequencing Flows	The number of flows performed by the sequencer during the sequencing stage of the run.

Parameters step assay options

Manager- and administrator-level users create assays in Genexus™ Software.

IMPORTANT! Use the default parameter settings for assays unless you are an advanced user. Contact your Field Bioinformatics Specialist (FBS) for assistance.

When you click the field of a parameter, the software shows the default value and the range that is allowed for the parameter. You can see a description of most parameters when you place the pointer over [?](#) (**Learn More**) next to a parameter.

The available options depend on the type of assay and the selections made in the **Reagent** step:

- **Purification**
- **Library Prep & Templating**
- **Primary Analysis**
Advanced Command Line Arguments in the **Primary Analysis** section can only be adjusted when you click **Enable customization**.
- **Annotation**
- **MSI** (Microsatellite Instability)
- **CNV Finding**
- **Fusions**
- **Read Mapping**
- **Variant Finding**

In the **Parameters** step, you can accept the default settings for the assay parameters, or adjust the settings if needed.

Edit assay parameters in a file

Manager- and administrator-level users create assays in Genexus™ Software.

You can edit assay parameters in the **Parameters** step. Not all parameters appear in the software. You can download a JSON file that contains the parameters that appear in the software and more parameters that do not appear in the software. If you are advanced and understand how the changes can affect results, you can edit the parameters file and then upload the edited file to create an assay that captures that values of the JSON file.

1. In the menu bar, click **Assays ▶ Create Assays**.
2. Select the type of assay you want to create.
3. Select a panel and other options in the **Panel** step, then click **Next**.
4. Select the kits and chips in the **Reagent** step, then click **Next**.

5. In the **Parameters** step:
 - a. In the **Filter Parameters by** dropdown list at the bottom of the screen, select the section for the parameters you want to change or select **All** to download a file that contains all parameters.
 - b. Click **Download** to download a JSON file for the parameters.
6. Edit the parameter values in the file and save the file.
7. Click **Upload**.
 - a. Click **Select File**.
 - b. Navigate to the edited file.
 - c. Click **Open**.
 - d. In the **Upload Parameters** dialog box, click **Upload** to upload the file.

The parameter values in the uploaded file are added to the assay.

Resequencing

When you copy and edit or create an assay in Genexus™ Software, the custom assay that you create can include an option to sequence and resequence samples in the same run. The option to resequence samples is available only in assays that specify a 200-bp template size. Runs that include the resequencing option have longer run times. For more information about resequencing samples, see the sequencing instrument or assay user guides.

IMPORTANT! When you create an assay, keep the **Resequencing** parameter set to **False** unless you have spoken with a Field Bioinformatics Specialist (FBS) and understand how the change can affect results.

Resequencing settings are available in the **Primary Analysis** section of the **Parameters** step when you create an assay. Template size is specified in the **Templating** section of the **Reagent** step.

QC step assay options

Manager- and administrator-level users create assays in Genexus™ Software.

You can specify Quality Control (QC) metric values to use in an assay in the **QC** step.

The following tables list options that are available in the **QC** step. The available options depend on the type of assay.

CF-1 Control

Option	Description
Min Average Reads Per Lane	The number of average reads per chip lane. You can specify the minimum threshold value required for the CF-1 (templating Control Fragment-1) to pass the QC metric.
Min Base Call Accuracy	The total number of errors for all positions in CF-1/ total number of CF-1 base reads. You can specify the minimum threshold value required for CF-1 to pass the QC metric.
Min Mean AQ20 Read Length (bp)	The average length, in base pairs, at which the accuracy rate is $\geq 99\%$ for reads of a library. You can specify the minimum threshold value required for CF-1 to pass the QC metric.

NTC QC - DNA

Option	Description
Max Average Base Coverage Depth	The average number of reads of all targeted reference bases. You can specify the maximum threshold value required for the DNA no template control to pass the QC metric.
Max Mean Read Length (bp)	The average length, in base pairs, of called reads. You can specify the maximum threshold value required for the DNA no template control to pass the QC metric.

NTC QC - RNA

Option	Description
Max Mapped Reads	<p>The total number of reads that are mapped to the reference genome in Genexus™ Software.</p> <p>You can specify the maximum threshold value required for the RNA no template control to pass the QC metric.</p> <p>Mapped Reads can be configured in the QC step or in the Parameters step in the Fusions section as Minimum Total Valid mapped reads. The values configured in the Parameters step overwrite those configured in the QC step.</p>
Max Mean Read Length (bp)	<p>The average length, in base pairs, of called reads.</p> <p>You can specify the maximum threshold value required for the RNA no template control to pass the QC metric.</p> <p>Mapped Reads and Mean Read Length can be configured in the QC step or in the Parameters step in the Fusions section as Minimum Total Valid mapped reads and Minimum mean read length for valid sample QC. The values configured in the Parameters step overwrite those configured in the QC step.</p>

Purification QC – DNA

Option	Description
Min Sample Concentration DNA (ng/μl)	The minimum threshold value for the sample concentration of purified DNA is shown. This value cannot be changed in the QC step, but can be changed in the Parameters step. For more information, see “Create an assay for a Sample to Result run (manager/administrator)” on page 208.
Max Sample Concentration DNA (ng/μl)	The maximum threshold value for the sample concentration of purified DNA is shown. This value cannot be changed in the QC step, but can be changed in the Parameters step. For more information, see “Create an assay for a Sample to Result run (manager/administrator)” on page 208.

Purification QC – RNA

Option	Description
Min Sample Concentration RNA (ng/μl)	The minimum threshold value for the sample concentration of purified RNA is shown. This value cannot be changed in the QC step, but can be changed in the Parameters step. For more information, see “Create an assay for a Sample to Result run (manager/administrator)” on page 208.
Max Sample Concentration RNA (ng/μl)	The maximum threshold value for the sample concentration of purified RNA is shown. This value cannot be changed in the QC step, but can be changed in the Parameters step. For more information, see “Create an assay for a Sample to Result run (manager/administrator)” on page 208.

Purification QC – TNA

Option	Description
Min Sample Concentration TNA (ng/μl)	The minimum threshold value for the sample concentration of purified TNA is shown. This value cannot be changed in the QC step, but can be changed in the Parameters step. For more information, see “Create an assay for a Sample to Result run (manager/administrator)” on page 208.
Max Sample Concentration TNA (ng/μl)	The maximum threshold value for the sample concentration of purified TNA is shown. This value cannot be changed in the QC step, but can be changed in the Parameters step. For more information, see “Create an assay for a Sample to Result run (manager/administrator)” on page 208.

Run QC

Option	Description
Min Key Signal	The average signal after software processing for all library ISPs that identically match the library key (TCAG). This value is a measure of the efficiency of template amplification. You can specify the minimum threshold value required for a run to pass QC for the key signal metric.
Min Percent Loading	The percentage of addressable wells on a chip lane that are loaded with an ISP. The percentage is derived from the number of wells with ISPs divided by the number of the total addressable wells in a run. You can specify the minimum threshold value required for a run to pass QC for the percent loading metric.
Min Raw Read Accuracy	The average raw accuracy across each individual base position in a read, where raw read accuracy is calculated as $100 * (1 - (\text{sum(per base error)}/\text{sum(per base depth)}))$. You can specify the minimum threshold value required for a run to pass QC for the raw read accuracy metric.

Sample QC - DNA

Options	Description
Min Deamination score	Deamination is reported as the estimated SNP proportion consistent with deamination (low allele frequency C:G>T:A SNVs). The deamination score can be used to determine the quality of the FFPE sample. You can specify the minimum threshold value required for a DNA sample to pass the QC metric.
Min Mapped Reads	The number of reads aligned to the reference genome. You can specify the minimum threshold value required for a DNA sample to pass the QC metric.

Sample QC - DNA (continued)

Options	Description
Min Mean AQ20 Read Length (bp)	<p>The average length, in base pairs, at which the accuracy rate is $\geq 99\%$ for reads of a library.</p> <p>You can specify the minimum threshold value required for a DNA sample to pass the QC metric.</p>
Min Mean Read Length (bp)	<p>The average length, in base pairs, of called reads.</p> <p>You can specify the minimum threshold value required for a DNA sample to pass the QC metric.</p>
Min Number Of Bases Used In Calculating TMB	<p>The number of exonic bases or all of the genomic bases that are covered by the panel. Only bases with sufficient base coverage are used in the calculation, as defined in the workflow parameters. By default, the TMB calculation uses total exonic bases with ≥ 60 bp coverage. In the TMB parameter settings, you can select between exonic bases or all of the genomic bases. You can also modify the sufficient coverage value.</p> <p>You can specify the minimum threshold value required for a DNA sample to pass the QC metric.</p>
Min Mean Red Cov	<p>The mean molecular coverage of targeted CNV gene.</p> <p>You can specify the minimum threshold value required for a DNA sample to pass the QC metric.</p>
Min Median Mol Cov	<p>The median molecular coverage of non-CNV reference loci.</p> <p>You can specify the minimum threshold value required for a DNA sample to pass the QC metric.</p>
Min Number of variant calls	<p>The number of somatic variants that are identified in the sample. This value is reported in the statistic.txt file as Total Somatic Filtered Variants Count (numerator for TMB calculation) and Variant Count.</p> <p>You can specify the minimum threshold value required for a DNA sample to pass the QC metric.</p>
Min Uniformity of Base Coverage	<p>The percentage of reads showing a depth of coverage $\geq 20\%$ of the mean base coverage.</p> <p>You can specify the minimum threshold value required for a DNA sample to pass the QC metric.</p>

Sample QC - RNA

Option	Description
Min MAPD ^[1]	<p>MAPD is a quality metric that estimates coverage variability between adjacent amplicons in CNV analyses. A MAPD value of 0.4 indicates an acceptable level of coverage variability. High MAPD value typically translates to a lower coverage uniformity. Lower coverage uniformity can result in missed or erroneous CNV calls. If the MAPD QC threshold is not met, CNVs do not get called. The MAPD metric does not affect SNVs/INDEL calls.</p> <p>You can specify the minimum threshold value required for an RNA sample to pass the QC metric.</p>
Max MAPD [1]	<p>You can specify the maximum threshold value required for an RNA sample to pass the QC metric.</p>
Min Mapped Reads	<p>The number of reads aligned to the reference genome.</p> <p>You can specify the minimum threshold value required for an RNA sample to pass the QC metric.</p> <p>The minimum allowed value for mapped reads. Mapped Reads and Mean Read Length can be configured in the QC step or in the Parameters step in the Fusions section as Minimum Total Valid mapped reads and Minimum mean read length for valid sample QC. The values configured in the Parameters step overwrite those configured in the QC step.</p> <p>The best practice to achieve consistent results is to use the same values in both the QC step and the Parameters step.</p>
Min Mean AQ20 Read Length (bp)	<p>The average length, in base pairs, at which the accuracy rate is $\geq 99\%$ for reads of a library.</p> <p>You can specify the minimum threshold value required for an RNA sample to pass the QC metric.</p>
Min Mean Read Length (bp)	<p>The average length, in base pairs, of called reads.</p> <p>You can specify the minimum threshold value required for an RNA sample to pass the QC metric.</p>
Min RNA Expression Ctrls Detected ^[2]	<p>The number of expression control genes detected for the sample. This metric measures the RNA input integrity, input amount, and the fidelity of the reverse transcriptase that was used in library preparation. Fusion panels include primer pairs that cover seven control housekeeping genes. For OncoPrint™ Precision Assay GX to pass QC, cTNA samples require 2 out of 7 control genes to be detected and FFPE RNA samples require 5 out of 7 control genes to be detected.</p> <p>You can specify the minimum threshold value required for an RNA sample to pass the QC metric.</p>

^[1] This option is only available when a Copy Number Baseline for CNV Calling is selected in the panel step.

^[2] This option is available for cTNA panels only.

Custom assays for Sample to Result runs (manager/administrator)

Manager- and administrator-level users can create a new assay or copy and edit an existing assay for a **Sample to Result** run. For more information, see “Create an assay (manager/administrator)” on page 194 and “Copy an assay (manager/administrator)” on page 196.

Sample to Result runs integrate sequential nucleic acid purification and sequencing. The concentration of purified nucleic acid for each sample is calculated after purification, before sequencing. You can choose to review samples after purification or proceed directly to sequencing.

Option	Description
Sequence all nucleic acid samples, regardless of concentration	All samples in the run are sequenced.
Sequence only samples that have a nucleic acid concentration within a set range	All samples that have a concentration within a specified range are sequenced. Samples with a concentration outside the specified range are not sequenced.
Review samples after purification	You can review the concentration of all samples and determine which samples to sequence.

Create an assay for a Sample to Result run (manager/administrator)

1. In the menu bar, click **Assays** ▶ **Create Assay**.
2. In the **Create Assay** screen, click the assay type for the template that provides the appropriate kits, chips, primer panels, and adjustable QC thresholds and read mapping parameters. For more information about selections, see “Create an assay (manager/administrator)” on page 194.
3. In the **Panel** step, select a panel and other options for the assay. For descriptions of the panel information appropriate for the assay, see “Panel step assay options” on page 197.
4. Click **Next**.
5. In the **Reagent** step, in **Purification Kit**, select the appropriate kit.

Note: If you select **Other**, the assay cannot be used in a **Sample to Result** run.

6. Select the kits and chips that you are using. Ensure that the reagents are appropriate for the primer panel that you are using, then click **Next**.
For descriptions of the reagent information appropriate for the assay, see “Reagent step assay options” on page 199.

7. In the **Parameters** step, accept the recommended default values, or adjust the values within the allowed ranges, then click **Next**.

You can set the parameter values with an advanced configuration file. For more information, see “Edit assay parameters in a file” on page 201.

Purification Parameters

Option	Description
Sample Type	The types of samples that are compatible with the selected purification kit are listed.
Review Require	<ul style="list-style-type: none"> Select True to review the concentration of all samples after purification and select which samples to sequence. Select False to enable automated sequencing of all samples that are within the specified concentration after purification. <p>Note: You can override this assay setting when you plan a purification run. For more information, see “Plan a Sample to Result run” on page 65.</p>
QC Concentration Minimum	QC concentration minimum thresholds in ng/μl for DNA or RNA.
QC Concentration Maximum	QC concentration maximum thresholds in ng/μl for DNA or RNA.

For more information about parameters, see “Parameters step assay options” on page 201.

8. In the **QC** step, enter QC metric values, then click **Next**.
For descriptions of the QC information appropriate for the assay, see “QC step assay options” on page 203.
9. In the **Plugins** step, select one or more plugins to run with this assay, then click **Next**.
For more information, see Chapter 10, “Plugins for data analysis”.
10. In the **Save** step, enter an assay name, an assay full name, and an optional description, then click **Save**.

The new assay is listed in the **Manage Assays** screen. The **Assay Full Name** is followed by **Draft** to indicate that the assay is in draft status. You can plan a run with the draft assay and the assay is available to lock, edit, or delete, and audit. You can also download parameters from assays in **Draft** or **Locked** statuses.

Filters and filter chains

A filter chain is a set of filters that Genexus™ Software uses to narrow the list of variants that are included in results. There are two ways to use filter chains. You can apply the filter chain to results to dynamically change the view of the variants, then optionally save the filter chain to the results. See “Filter results using a filter chain” on page 151.

Genexus™ Software includes predefined filter chains that you can apply to run results. Most system-installed assays include default filter chains. For information about system-installed filter chains, see “System-installed filter chains” on page 351.

Manager- and administrator-level users can create custom filter chains that are built from system-installed filter chains, or from one or more system-installed filters. For more information, see “Create a custom filter chain (manager/administrator)” on page 210. The system-installed filters are based on public and proprietary annotation sources and data types that are included in the software. For more information, see “System-installed filters” on page 352.

Review filter chain details

A filter chain is a set of filters that Genexus™ Software uses to narrow the list of variants that are included in results. Administrator-level users create custom filter chains from system-installed filters to meet specific variant filtering needs.

After a run, you can review the details of the filter chains that were applied to the run. If the filter chain does not meet your needs, you can adjust the settings or apply a different filter chain.

1. In the menu bar, click **Assays ▶ Manage Presets**.
2. In the **Manage Presets** screen, click the **Filter Chains** tab.
3. Click a filter chain.

Review the following details:

- The version of the analysis pipeline used for the filter chain.
- Whether the filter chain is system-installed.
- The date that the filter chain was created.
- The filters that are included in the filter chain.
- The order in which the filters and/or filter chains contained within are applied.

Create a custom filter chain (manager/administrator)

Manager- and administrator-level users can create and manage filter chains. A filter chain is a set of filters that you apply to the variants identified in the results. Filter chains are used to narrow the results to only those variants of interest.

1. In the menu bar, click **Assays ▶ Manage Presets**.
2. In the **Manage Presets** screen, in the **Filter Chains** tab, click **+ Add New**.
3. In the **Create Filter Chains** screen, enter a name for the new filter chain.
4. *(Optional)* Add a description for the filter chain.
5. From the **Choose Filter** list, select the filter that you want to use, then click **Set**.

The selected filters are shown on the right side of the window. If the list does not populate with the filters available in the software, you may need to reboot the sequencer.

6. Repeat step 5 for each filter that you want to add to the filter chain.
7. Click **Save**.
A confirmation message appears, and the new filter chain is listed in the **Filter Chains** tab and is available for selection when you view results. For more information, see “Filter results using a filter chain” on page 151.

Custom filter chain examples (manager/administrator)

Manager- and administrator-level users can create custom filter chains for use in Genexus™ Software.

Create a Deamination, dbsnp, and COSMIC(85) filter chain (manager/administrator)

Manager- and administrator-level users can create a custom filter chain with multiple filters.

1. In the menu bar, click **Assays ▶ Manage Presets**.
2. In the **Assays/Manage Presets** screen, click the **Filter Chains** tab.
3. Click **+ Add New**.
4. In the **Create Filter Chains** screen, enter a name and optional description, then in the **Choose Filter** list, then click **Set** to add each filter and create a filter chain.
To include unannotated variants in the results, select **Include Unannotated Variants** for each filter selection.
 - a. Select **Deamination**, set its value to **In**, then click **Set**.
The filter name appears on the right side of the **Create Filter Chains** screen.
 - b. Select a **dbsnp** filter, set its value to **In**, then click **Set**.
The second filter name appears on the right side of the **Selected Filter Chains** list.
 - c. Select **COSMIC(85)**, set its value to **In**, then click **Set**.
The third filter name appears on the right side of the **Create Filter Chains** screen.
5. (Optional) If you want to change the order in which your filters are applied, click **🗑 Delete** to remove a filter, then reselect the filter and click **Set** to reinsert the filter in the **Selected Filter Chains** list.
6. Click **Save**.

A confirmation message appears, and the new filter chain is listed in the **Filter Chains** tab and is available for selection when you view results. For more information, see “Filter results using a filter chain” on page 151.

Create a COSMIC ClinVar and Variant Impact filter chain (manager/administrator)

Manager- and administrator-level users can create a custom filter chain that includes both annotation source filters and variant data type filters. This procedure shows how to create a COSMIC, ClinVar, Allele Fraction, and Variant Effect filter chain.

1. In the menu bar, click **Assays** ▶ **Manage Presets**.
2. In the **Manage Presets** screen, click the **Filter Chains** tab.
3. Click **+ Add New**.
4. In the **Create Filter Chains** screen, enter a name and optional description.
5. Select the filters to add to the filter chain.
To include unannotated variants in the results, select **Include Unannotated Variants** for each filter selection.
 - a. In the **Choose Filter** list, choose a version of the **COSMIC** filter.
 - b. Select the annotations that you want to include in the filter.
 - c. Click **Set**.
 - d. In the **Choose Filter** list, select a version of **clinvar**.
 - e. Select the annotations that you want to include in the filter.
 - f. Click **Set**.
 - g. In the **Choose Filter** list, select **Allele Fraction**.
 - h. Enter a range value, then click **Set**.
 - i. In the **Choose Filter** list, select **Variant Effect**.
 - j. Select the value that you want to include in the filter to detect the effect of variants on coding sequences.
 - k. Click **Set**.
6. Click **Save**.

Your filter chain can now be applied to results. For more information, see “Filter results using a filter chain” on page 151.

Create a PValue/dbSNP/Variant Type filter chain (manager/administrator)

Manager- and administrator-level users can create a custom filter chain with multiple filters. This example shows how to create a filter chain with PValue, dbSNP, and Variant Effect filters.

1. In the menu bar, click **Assays ▶ Manage Presets**.
2. In the **Manage Presets** screen, click the **Filter Chains** tab.
3. Click **+ Add New**.
4. In the **Create Filter Chains** screen, enter a name and optional description.
To include unannotated variants in the results, select **Include Unannotated Variants** for each filter selection.
 - a. Select the **PValue** filter.
 - b. Enter a range
 - c. Click **Set**.
5. In the **Choose Filter** list, select a version of **dbSNP**.
 - a. Set **Filter value** to **In**.
 - b. Click **Set**.
6. In the **Choose Filter** list, select **Variant Effect**.
 - a. Select the value that you want to include in the filter to detect the effect of variants on coding sequences.
 - b. Click **Set**.
7. Click **Save**.

Your filter chain can now be applied to results. For more information, see “Filter results using a filter chain” on page 151.

Obsolete a custom filter chain

Manager- and administrator-level users can make filter chains unavailable in the software. You cannot designate as obsolete system-installed filter chains.

1. Click **Assays ▶ Manage Presets**.
2. In the **Filter Chains** tab, place the pointer over the row of the filter chain that you want to make unavailable, then click **Obsolete**.

The filter chain status is **Obsolete**.

Create a gene symbol filter (manager/administrator)

Manager- and administrator-level users can create a custom filter that is based on gene symbols. This filter can be added to a filter chain that narrows results to only specific genes. If you have numerous gene symbols to filter, you can use a text file to create the gene symbol filter chain. For more information, see “Gene Symbol filter” on page 355.

1. In the menu bar, click **Assays ▶ Manage Presets**.
2. In the **Manage Presets** screen, click the **Filter Chains** tab.
3. Click **+ Add New**.
4. In the **Create Filter Chains** screen, enter a name and optional description, then in the **Choose Filter** list, choose the **Gene Symbol** filter.
5. Add genes of interest to the filter chain. Select a checkbox for the gene symbol of the gene in the list that you want to add.
To find a gene symbol in the list, enter the gene symbol into the search field, then click **Q (Search)**.
6. After you select all of the gene symbols that you want to include, click **Set**.
Your genes appear in the **Selected Filter Chains** list.
7. Click **Save**.

Your filter chain can now be added to an assay, or applied to results. For more information, see “Filter results using a filter chain” on page 151.

Annotation sets (manager/administrator)

Manager- and administrator-level users use the tools for creating and managing annotation sets that are available in the **Assays / Manage Presets** screen in the **Annotation Sets** tab.

View annotation set details

You can view details about an annotation set in Genexus™ Software.

1. Click **Assays ▶ Manage Presets**.
2. In the **Assays / Manage Presets** screen, in the **Annotation Sets** tab, click the annotation set name.

The following details for the selected annotation set are shown.

Annotation Set Details

Item	Description
Version	The analysis pipeline version that is used by the annotation set.
Last Modified By	For custom annotation sets, the sign-in name of the person who created or modified a custom annotation set.
Last Modified On	Date on which a custom annotation set was last modified.
Created By	Custom annotation sets show the sign-in name of the person who created the annotation set. Annotation sets that are included with the software are labeled as System Installed .
Created On	Date on which a custom annotation set was created.
Status	Status of the annotation set, including: <ul style="list-style-type: none"> • Locked • Draft • Obsolete
Reference	Genome reference that is available in the annotation set. <ul style="list-style-type: none"> • hg19 • GRCh38
Annotation Versions	A list of the annotations included in the annotation, and the version of each annotation.

Create a new annotation set (manager/administrator)

Manager- and administrator-level users can create custom annotation sets in Genexus™ Software. The custom annotation sets are available as an option when you create or copy an assay.

1. Click **Assays ▶ Manage Presets**.
2. In the **Manage Presets** screen, in the **Annotation Sets** tab, click **+ Add New**.
The **Create Annotation Set** screen opens and you can view the current **Application Version**.
3. Enter a name for the new annotation set in **Name**, and an optional description in **Description**.
4. In **Genome Reference**, select a reference genome from the dropdown list.

Note: The default reference is **hg19**.

5. In the **Available Annotation Sources** dropdown list, select the annotation source.

Source	Description
5000 Exomes	Population frequency information from the 5000 Exomes project. Source: https://evs.gs.washington.edu/EVS/
ClinVar	An assessment of impact of the variants observed from NCBI ClinVar database. Source: http://www.ncbi.nlm.nih.gov/clinvar
COSMIC	A catalog of known somatic mutations in cancer. Source: https://cancer.sanger.ac.uk/cosmic
dbSNP	A database of genomic variants. Source: http://www.ncbi.nlm.nih.gov/projects/SNP
DGV	A curated database of human genomic structural variation. Source: http://dgv.tcag.ca/dgv/app/home
DRA	Disease Research Area; a hierarchically organized database of disease categories. https://meshb.nlm.nih.gov
DrugBank	A database from which names of therapies that are used for researching variants are collated. Source: https://www.drugbank.ca
ExAC	Exome Aggregation Consortium—A database catalog of variant frequencies. Source: https://gnomad.broadinstitute.org
Gene Model	A set of genes that the variant overlaps. Sources: https://www.ncbi.nlm.nih.gov/refseq and http://www.ensembl.org
Gene Ontology	Standardized ontology for gene and gene projects (for example, functional role or localization). Source: http://geneontology.org
Gene Set (Custom)	Select this option to import a TXT file that contains a custom set of variants in a specific set of genes.
Genomic Regions (Custom)	Select this option to import BED file that contains a custom set of variants in specific gene regions.
OMIM	Online Mendelian Inheritance of Man. Source: https://www.ncbi.nlm.nih.gov/omim
Pfam	A database of protein families that includes their annotations and multiple sequence alignments using hidden Markov models. Source: http://pfam.xfam.org
PhyloP Scores	A measure of conservation of the protein across a wide range of organisms. Source: http://compgen.cshl.edu/phast
SIFT / PolyPhen	A prediction of the functional effect of a variant on a protein. Sources: https://sift.bii.a-star.edu.sg and http://genetics.bwh.harvard.edu/pph2 and

Source	Description
Transcript Set (Custom)	Select this option to import a TSV file that contains a custom set of transcript annotations.
Variant DB (Custom)	Select this option to import a VCF file that contains custom set of variant annotations.

The annotation sources that you select are listed in the **Available Annotation Sources** list.

6. Add annotation sources to the annotation set.

Options	Description
Use an existing source	Select the checkbox next to each annotation source in Available Annotation Sources list that you want to include in the annotation set.
Import a new custom source file in a TXT file ^[1]	Click + Add New Source below the list, enter a name and version for the source, browse to and select the source file, then click Upload . The new sources are added to Available Annotation Sources list.

^[1] You must have selected one of the following **Available Annotation Sources:Gene Set (Custom), Genomic Regions (Custom), Transcript Set (Custom), VariantDB (Custom)**.

- After all of the annotation sources that you want to add to the annotation set are selected in the **Available Annotation Sources** list, click **Add >** to move the annotation sources to the **Selected Annotation Sources** list.
- Review the list of selected annotation sources, then add or remove sources, if needed. To remove a source from the **Selected Annotation Sources** list, select the checkbox, then click **Remove X**.
- Click **Save** to save the annotation set.

The annotation set is listed in the **Annotation Sets** tab and is available when you create an assay.

Make custom annotation sets unavailable

Manager- and administrator-level users can make custom annotation sets unavailable in the software.

You cannot designate as obsolete system-installed annotation sets.

- In the menu bar, click **Assays > Manage Presets**.
- In the **Annotation Sets** tab, place the pointer over the row of the custom annotation set that you want to make unavailable, then click **Obsolete**.

The annotation set status is **Obsolete** in the list of annotation sets.

Copy number baselines (manager/administrator)

A copy number baseline is a set of control samples that are used to create a baseline for the detection of copy number variants (CNVs). A copy number baseline is associated with a panel file in the software. CNV analysis is based on reads counts for each amplicon.

Known sources of variability include pool imbalance (when an assay has more than one pool of amplicons), total number of reads, per-amplicon attributes of GC proportion, and length of the amplicon insert. In practice, we observe other variability that does not associate with known attributes yet is systematic.

Copy number estimates are made using a proprietary algorithm.

Genexus™ Software includes predefined copy number baselines.

Manager- and administrator-level users can perform the following functions related to copy number baselines.

Option	See the following section for more information
View details for system-installed, or custom, copy number baselines.	“View copy number baseline details (manager/administrator)” on page 219
View the panel associated with a copy number baseline.	“Panel files (manager/administrator)” on page 283
Create a copy number baseline.	“Create a copy number baseline (manager/administrator)” on page 220
Create a copy number baseline that uses an existing copy number baseline as the base	“Copy a copy number baseline (manager/administrator)” on page 222
View or add notes to a copy number baseline	“View, add, or remove a note in a copy number baseline (manager/administrator)” on page 223
Augment a copy number baseline.	“Augment a copy number baseline with more samples (manager/administrator)” on page 221
Import a copy number baseline.	“Import a copy number baseline (manager/administrator)” on page 223
Export a copy number baseline that has been created in the software or imported into the software	“Export a custom copy number baseline (manager/administrator)” on page 225
Delete a copy number baseline that fails during job creation	“Delete a copy number baseline (manager/administrator)” on page 224
Download log files	“Download the log files for copy number baseline creation (manager/administrator)” on page 224
Remove copy number baselines that are not system-installed.	“Remove a copy number baseline (manager/administrator)” on page 225

View copy number baseline details (manager/administrator)

Manager- and administrator-level users can view details for the system-installed copy number baselines, or for custom copy number baselines.

1. In the menu bar, click **Assays ▶ Manage Presets**.
2. In the **Assays / Manage Presets** screen, click the **Copy Number Baselines** tab.
The **Copy Number Baselines** table lists both system-installed and custom copy number baselines.
3. Click the name of a copy number baseline.

Review the details in the **Copy Number Baseline Details** pane.

Copy Number Baseline Details

Item ^[1]	Description
Created By	Sign-in name of the person who created the baseline. System-installed baselines are included with the software.
Created On	Date on which the baseline was created.
Number of Normal Samples	The number of normal samples that are included in the baseline.
Gender	The biological sex of the sample: Female, Male, or Unknown.
Number of Samples	The total number samples that are included in the baseline.
Amplification Type	Library amplification technology that is used in the baseline (Ion AmpliSeq™ or Ion AmpliSeq™ HD).
Panel Name	Name of the panel that is included in the baseline.
Reference	The reference genome for the baseline.
Reference File Name	The name of the reference genome file that is used in the baseline.

^[1] The details that are shown depend on the setup of the copy number baseline. Parameters used to create copy number baselines are also shown, but are not listed in this table.

You can click **⋮ (More Options)** to perform more actions, such as obsolete, export, copy, edit, or audit the copy number baseline. The options that are available depend on whether the copy number baseline is system-installed or is a custom copy number baseline.

The actions for each baseline are also available in the **Copy Number Baselines** table when you place the pointer over the row of a copy number baseline.

Create a copy number baseline (manager/administrator)

Manager- and administrator-level users can create a copy number baseline.

IMPORTANT! Use the default parameter settings unless you are an advanced user. Contact a Field Bioinformatics Specialist (FBS) for help.

1. In the menu bar, click **Assays ▶ Manage Presets**.
2. In the **Assays / Manage Presets** screen, click the **Copy Number Baselines** tab.
3. Click **+ Add New**.
4. Select the type of library for the copy number baseline.
 - **AmpliSeq** – Create a copy number baseline for Ion AmpliSeq™ libraries.
 - **AmpliSeq HD** – Create a copy number baseline for Ion AmpliSeq™ HD libraries.

The baseline creation tool opens in the **Panel** step.


Use the **Next** and **Previous** buttons to navigate the screens to configure the baseline.

5. Complete the **Panel** step.
 - a. Enter a name in the **Copy Number Baseline Name** field.
The name is limited to 50 characters and no spaces are allowed.
 - b. Leave **Augment an Existing Copy Number Baseline as Base** unselected.
For information to **Augment an Existing Copy Number Baseline as Base**, see “Augment a copy number baseline with more samples (manager/administrator)” on page 221.
 - c. Select the panel.
 - d. Select **CNV VCIB** in **Algorithm Type**.
 - e. *(Optional)* Enter information about the baseline into the **Description** field.
 - f. Click **Next**.
6. In the **Parameters** step, accept the default settings or adjust the appropriate parameters, then click **Next**.
7. In the **Samples** step, select samples for the baseline.

All of the samples that have been run with the selected panel are listed.

If you do not select the minimum number of samples that are required for the baseline, an error message appears and you cannot proceed. You can adjust the **Minimum number of samples required to add to an existing baseline** parameter in the **CNV Baseline Creation** section of the **Parameters** step.

 - a. Select the checkbox next to each sample to use for the baseline.

- b. Click  in the **Normal Sample** column to designate a selected sample as "Normal" in the baseline.

The tube icon changes to green. A **Normal Sample** is designated with .

If you do not designate the minimum number of normal samples that are required for the baseline, an error message appears and you cannot proceed.

- c. Click **Next**.

8. In the **Review** step, review the selections, then click **Save**.

The new copy number baseline is added to the **Copy Number Baselines** tab.

Copy number baselines that have a **State** of **Completed** are available for selection when an assay is created or copied and edited. Copy number baselines can only be added to **DNA and Fusions**, **DNA Somatic** and **DNA Germline** assays.

If the **State** of a copy number baseline is **Failed**, place the pointer over the row of the copy number baseline and click **View Log** to open the log in a separate browser tab. The log file can help with troubleshooting if the baseline creation job fails.

Augment a copy number baseline with more samples (manager/administrator)

Manager and administrator-level users can augment an existing copy number baseline. The newly created copy number baseline is saved with a new name and is listed as a new copy number baseline; the original copy number baseline used as a base remains unchanged in the software.

When a baseline is augmented with the addition of samples, new samples are run, the size of each systematic affect encoded in the baseline is estimated, and a correction is applied to remove the effect. Any added samples cannot be designated as a **Normal Sample** and should be diverse so as to capture likely systematic variation.

IMPORTANT! Use the default parameter settings unless you are an advanced user. Contact a Field Bioinformatics Specialist (FBS) for help.

1. In the menu bar, click **Assays ▶ Manage Presets**.
2. In the **Assays / Manage Presets** screen, click the **Copy Number Baselines** tab.
3. Click **+ Add New**.
4. Select the type of library for the copy number baseline.
 - **AmpliSeq** – Create a copy number baseline for Ion AmpliSeq™ libraries.
 - **AmpliSeq HD** – Create a copy number baseline for Ion AmpliSeq™ HD libraries.

The baseline creation tool opens in the **Panel** step.

Use the **Next** and **Previous** buttons to navigate the screens to configure the baseline.

5. Complete the **Panel** step.
 - a. Enter a name in the **Copy Number Baseline Name** field.

The name is limited to 50 characters and no spaces are allowed.

- b. Select the panel.
 - c. Select **Augment an Existing Copy Number Baseline as Base**, then select the baseline you would like to edit or augment.
 - d. Select **CNV VCIB** in **Algorithm Type**.
 - e. *(Optional)* Enter information about the baseline into the **Description** field.
 - f. Click **Next**.
6. In the **Parameters** step, accept the default settings or adjust the appropriate parameters, then click **Next**.
7. In the **Samples** step, select samples for the baseline.

All of the samples that have been run with the selected panel are listed.

If you do not select the minimum number of samples that are required for the baseline, an error message is displayed and you cannot proceed. You can adjust the **Minimum number of samples required to add to an existing baseline** parameter in the **CNV Baseline Creation** section of the **Parameters** step.

 - a. Select the checkbox next to each sample to use for the baseline.
 - b. Click **Next**.
8. In the **Review** step, review the selections, then click **Save**.

The new copy number baseline is added to the **Copy Number Baselines** tab.

Augmented copy number baselines that have a **State** of **Completed** are available for selection when an assay is created or copied and edited. Copy number baselines can only be added to **DNA and Fusions**, **DNA Somatic** and **DNA Germline** assays.

If the **State** of a copy number baseline is **Failed**, place the pointer over the row of the copy number baseline and click **View Log** to open the log in a separate browser tab. The log file can help with troubleshooting if the baseline creation job fails.

Copy a copy number baseline (manager/administrator)

Manager- and administrator-level users can copy then edit a copy number baseline.

1. In the menu bar, click **Assays** ▶ **Manage Assays**.
2. In the **Assays / Manage Presets** screen, click the **Copy Number Baselines** tab.
3. Place the pointer over the row of the copy number baseline that you want to copy, then click **Copy**.

The **Copy Copy Number Baseline** screen opens to the **Panel** step. The settings and samples that are included can be modified for the new baseline.


4. Change the software-generated name of the baseline, proceed through the workflow steps, and modify settings, if needed. For more information, see “Augment a copy number baseline with more samples (manager/administrator)” on page 221.
5. In the **Review** step, review the selections, then click **Save**.
 The new copy number baseline is added to the **Copy Number Baselines** tab.

The copy number baseline is available for selection when an assay is created or copied and edited. Copy number baselines can only be added to **DNA and Fusions**, **DNA Somatic** and **DNA Germline** assays.

View, add, or remove a note in a copy number baseline (manager/administrator)

Manager- and administrator-level users can add notes to a copy number baseline or view existing notes. Use **Notes** to capture time-stamped written comments and observations for a copy number baseline. A copy number baseline can contain multiple notes from different users.

1. In the menu bar, click **Assays ▶ Manage Presets**.
2. In the **Assays / Manage Presets** screen, click the **Copy Number Baselines** tab.
3. Place the pointer over the row of the copy number baseline of interest, click **Notes**, then review existing notes or add a new note.

Action	Description
View existing notes for a copy number baseline.	In the Add Notes dialog box, notes that are associated with the copy number baseline are listed by user, date, and time.
Add a new note to a copy number baseline.	<ol style="list-style-type: none"> 1. In the Add Notes dialog box, click + Add Note. 2. Enter the note, then click Save. <p>The note is added to the copy number baseline and includes the username for the account of the person who entered the note, and a timestamp that records the note entry date and time.</p>
Delete a copy number baseline note	In the Add Notes dialog box, in the row of the note that you want to delete, click  (Delete) .


Click **Close** to return to the **Manage Presets** screen.

Import a copy number baseline (manager/administrator)

Manager- and administrator-level users can import a copy number baseline. Two types of files are supported:

- H5
- ZIP

For ZIP files, the panel must be available in the software before you import the copy number baseline. For more information, see “Panel files (manager/administrator)” on page 283.

1. In the menu bar, click **Assays ▶ Manage Presets**.
2. In the **Assays / Manage Presets** screen, click the **Copy Number Baselines** tab.
3. Click  **Import Copy Number Baseline** in the top right of the screen.

Option	Description
Import a ZIP file	<ol style="list-style-type: none"> 1. In File Type, select ZIP. 2. Click Select Copy Number Baseline file. 3. Browse for and select the file. 4. Click Upload.
Import an H5 file	<ol style="list-style-type: none"> 1. In File Type, select H5. 2. In the Panel list, select the panel. 3. Click Select Copy Number Baseline file. 4. Browse for and select the file. 5. Click Upload.

A message confirms that the file import was successful when the file import is complete.

Imported copy number baselines are available for selection when an assay is created or copied and edited. Copy number baselines can only be added to **DNA and Fusions**, **DNA Somatic** and **DNA Germline** assays.

Download the log files for copy number baseline creation (manager/administrator)

Manager- and administrator-level users can download and view log files that show the details for copy number baseline creation. The log files are available after the copy number baseline creation job is complete.

1. In the menu bar, click **Assays ▶ Manage Presets**.
2. In the **Assays / Manage Presets** screen, click the **Copy Number Baselines** tab.
3. Place the pointer over the row of the custom copy number baseline of interest, then click **Download Log**.

Delete a copy number baseline (manager/administrator)

If a copy number baseline fails during job creation, it can be deleted from the software by a manager- and administrator-level user.

1. In the menu bar, click **Assays ▶ Manage Presets**.
2. In the **Assays / Manage Presets** screen, click the **Copy Number Baselines** tab.

3. Place the pointer over the row of a failed copy number baseline, then click **Delete**.
The **Delete Copy Number Baseline** dialog box opens with the message *Are you sure that you want to delete this Copy Number Baseline?*
4. Click **Yes** to delete the copy number baseline.

The failed copy number baseline is removed from the software and is no longer listed in the **Copy Number Baselines** table.

Export a custom copy number baseline (manager/administrator)

Manager- and administrator-level users can export copy number baselines that have been created, copied and edited, or imported in Genexus™ Software. Only custom copy number baselines can be exported. System installed copy number baselines cannot be exported.

A copy number baseline can be exported, for example to use that copy number baseline in another Genexus™ Integrated Sequencer.

1. In the menu bar, click **Assays ▶ Manage Presets**.
2. In the **Assays / Manage Presets** screen, click the **Copy Number Baselines** tab.
3. Place the pointer over the row of the copy number baseline that you want to export, then click **Export**.

The copy number baseline files are downloaded as a ZIP file and are available for import to another sequencer.

Remove a copy number baseline (manager/administrator)

Manager- and administrator-level users can make copy number baselines unavailable in the software. You cannot designate as obsolete system-installed copy number baselines.

1. In the menu bar, click **Assays ▶ Manage Presets**.
2. In the **Assays / Manage Presets** screen, click the **Copy Number Baselines** tab.
3. Place the pointer over the row of the copy number baseline that you want to make unavailable, then click **Obsolete**.

The copy number baseline is **Obsolete** in the list of copy number baselines.

Sequence variant baselines

A sequence variant baseline is used to reduce the number of false positives for panels that detect somatic variants.

Variant calling filters in Genexus™ Software suppress and remove most sequencing errors. However, it is possible that some errors can pass through the filters and be reported as false positives. In somatic panels, sequencing errors that survive filtering are reported as false positives that require investigation.

A sequence variant baseline is a BED file that contains a list of alleles that are known sequencing errors. The baseline file includes detailed properties for each sequencing error. The errors and properties are based on known germline normal samples. When added to an assay in Genexus™ Software, the baseline detects and filters out the errors in sequencing that could lead to false positive variant calls.

Genexus™ Software includes predefined sequence variant baselines.

You can also create a custom sequence variant baseline.

View sequence variant baseline details (manager/administrator)

Manager- and administrator-level users can view details for custom sequence variant baselines. You can view the name of a system installed sequence variant baseline file for an assay in the **Variant Finding** section of the **Parameters** step when you copy an assay.

1. In the menu bar, click **Assays ▶ Manage Presets**.
2. In the **Assays / Manage Presets** screen, click the **Sequence Variant Baselines** tab. The **Sequence Variant Baselines** table lists custom sequence variant baselines.
3. Click the name of a sequence variant baseline.

Review the details in the **Sequence Variant Baselines** pane.

Sequence variant baseline details

Item ^[1]	Description
Created By	Sign-in name of the person who created the baseline.
Created On	The date the baseline was created.
Number of Samples	The number of samples that are included in the baseline.
Amplification Type	Library amplification technology that is used in the baseline (Ion AmpliSeq™ or Ion AmpliSeq™ HD).
Panel Name	The name of the panel that is included in the baseline.
Chip Type	The semiconductor sequencing chip for the panel and baseline.
Reference	The reference genome for the panel and baseline.

^[1] The details that are shown depend on the setup of the sequence variant baseline. Parameters used to create sequence variant baselines are also shown, but are not listed in this table.

You can click ⋮ (**More Options**) to perform more actions, such as obsolete, export, copy, edit, or audit the sequence variant baseline.

The actions for each baseline are also available in the list of **Sequence Variant Baselines** when you place the pointer over the row of a sequence variant baseline.

Create a sequence variant baseline (manager/administrator)

Manager- and administrator-level users can create a sequence variant baseline (SVB) if system-installed sequence variant baselines do not meet research needs. You can add the custom baseline to an assay to use as a baseline for a run.

IMPORTANT! Use the default parameter settings unless you are an advanced user. Contact a Field Bioinformatics Specialist (FBS) for help.

1. In the menu bar, click **Assays ▶ Manage Presets**.
2. In the **Assays / Manage Presets** screen, click the **Sequence Variant Baselines** tab.
3. Click **+ Add New**.
4. Complete the **Panel** step.
 - a. Enter a name in the **Sequence Variant Baseline Name** field.
The name is limited to 50 characters and no spaces are allowed.
 - b. Select or leave **Augment an Existing Sequence Variant Baseline as Base** unselected.
 - Select **Augment an Existing Sequence Variant Baseline as Base**, then select an SVB from the dropdown list to use the existing sequence variant baseline as a starting point for the new baseline.
 - Leave **Augment an Existing Sequence Variant Baseline as Base** unselected to create a new baseline.
 - c. Select the panel.
 - d. *(Optional)* Enter information about the baseline into the **Description** field.
 - e. Click **Next**.
5. In the **Parameters** step, accept the default settings or adjust the appropriate parameters, then click **Next**.
6. In the **Samples** step, select at least 40 DNA, DNA & RNA, or TNA samples for the baseline.
The samples that you select must have the same library chemistry, chip type, and flow order.
7. In the **Review** step, review the selections, then click **Launch**.
The new sequence variant baseline is added to the **Sequence Variant Baselines** list.

Sequence variant baselines with a **State** of **Completed** are available for selection when an assay is created or copied and edited. For assays that are compatible with the sequence variant baseline, you can select the sequence variant baseline file in the **Variant Finding** section of the **Parameters** step, in the **Analysis SVB File** dropdown list.

If the **State** of a sequence variant baseline is **Failed**, place the pointer over the row of the sequence variant baseline and click **View Log** to open the log in a separate browser tab. The log file can help with troubleshooting if the baseline creation job fails.

Copy a sequence variant baseline (manager/administrator)

Manager- and administrator-level users can copy then edit a sequence variant baseline. You cannot copy sequence variant baselines that have been imported.

1. In the menu bar, click **Assays ▶ Manage Assays**.
2. In the **Manage Assays** screen, place the pointer over the row of the sequence variant baseline that you want to copy, then click **Copy**.
The **Copy Sequence Variant Baseline** screen opens to the **Panel** step with the panel and samples predefined. The settings and samples that are included can be modified for the new baseline.
3. Change the software-generated name of the baseline if needed, proceed through the workflow steps, and modify settings. For more information, see “Create a sequence variant baseline (manager/administrator)” on page 227.
4. In the **Review** step, review the selections, then click **Launch**.


The new sequence variant baseline is added to the **Sequence Variant Baseline** list. When the **State** of the SVB is **Completed** and the **Status** is **Locked**, the sequence variant baseline is available for selection when an assay is created or copied and edited.

For assays that are compatible with the sequence variant baseline, you can select the sequence variant baseline file in the **Variant Finding** section of the **Parameters** step, in the **Analysis SVB File** dropdown list. For more information, see “Create an assay (manager/administrator)” on page 194.

Import a sequence variant baseline (manager/administrator)

Manager- and administrator-level users can import a sequence variant baseline. Two types of files are supported:

- ZIP
- BED

1. In the menu bar, click **Assays ▶ Manage Presets**.
2. In the **Assays / Manage Presets** screen, click the **Sequence Variant Baselines** tab.
3. Click  **Import Sequence Variant Baseline** in the top right of the screen.

Option	Description
Import a ZIP file	<ol style="list-style-type: none"> 1. In File Type, select ZIP. 2. Click Select Sequence Variant Baseline file. 3. Browse for and select the file. 4. Click Upload.
Import a BED file	<ol style="list-style-type: none"> 1. In File Type, select BED. 2. Enter a name for the baseline. 3. In the Panel list, select the panel. 4. Click Select Sequence Variant Baseline file. 5. Browse for and select the file. 6. Click Upload.

A message confirms that the file import was successful when the file import is complete.


The imported sequence variant baseline is added to the **Sequence Variant Baseline** list. When the **State** of the SVB is **Completed** and the **Status** is **Locked**, the sequence variant baseline is available for selection when an assay is created or copied and edited.

For assays that are compatible with the sequence variant baseline, you can select the imported sequence variant baseline file in the **Variant Finding** section of the **Parameters** step, in the **Analysis SVB File** dropdown list. For more information, see “Create an assay (manager/administrator)” on page 194.

View, add, or remove a note in a sequence variant baseline (manager/administrator)

Manager- and administrator-level users can view existing notes or add notes to a sequence variant baseline. Use notes to capture time-stamped written comments and observations for a sequence variant baseline. A sequence variant baseline can contain multiple notes from different users.

1. In the menu bar, click **Assays ▶ Manage Presets**.
2. In the **Assays / Manage Presets** screen, click the **Sequence Variant Baselines** tab.
3. Place the pointer over the row of the sequence variant baseline of interest, click **Notes**, then review existing notes or add a new note.

Action	Description
View existing notes for a sequence variant baseline.	In the Add Notes dialog box, notes that are associated with the sequence variant baseline are listed by user, date, and time.
Add a new note to a sequence variant baseline.	<ol style="list-style-type: none"> 1. In the Add Notes dialog box, click + Add Note. 2. Enter the note, then click Save. <p>The note is added to the sequence variant baseline and includes the username for the account of the person who entered the note, and a timestamp that records the note entry date and time.</p>
Delete a sequence variant baseline note	In the Add Notes dialog box, in the row of the note that you want to delete, click  (Delete).

Click **Close** to return to the **Manage Presets** screen.

Download the log files for sequence variant baseline creation (manager/administrator)

Manager- and administrator-level users can download and view log files that show the details for sequence variant baseline creation. Log files include ERR, TXT and JSON files. The log files are available after the copy number baseline creation job is complete.

1. In the menu bar, click **Assays ▶ Manage Presets**.
2. In the **Assays / Manage Presets** screen, click the **Sequence Variant Baselines** tab.
3. Place the pointer over the row of the custom sequence variant baseline of interest, then click **Download Log**.

Delete a sequence variant baseline (manager/administrator)

If a custom sequence variant baseline fails or is terminated during job creation, the sequence variant baseline can be deleted from the software by a manager- or administrator-level user.

1. In the menu bar, click **Assays ▶ Manage Presets**.
2. In the **Assays / Manage Presets** screen, click the **Sequence Variant Baselines** tab.
3. Place the pointer over the row of a failed sequence variant baseline, then click **Delete**.
4. In the **Delete Sequence Variant Baseline** dialog box, click **Yes** to confirm the deletion.

The failed or terminated sequence variant baseline is removed from the software and is no longer included in the **Sequence Variant Baselines** table.

Export a custom sequence variant baseline (manager/administrator)

Manager- and administrator-level users can export sequence variant baselines that have been created, copied and edited, or imported in Genexus™ Software. Only custom sequence variant baselines can be exported.

A sequence variant baseline can be exported, for example to use that sequence variant baseline in another Genexus™ Integrated Sequencer.

1. In the menu bar, click **Assays ▶ Manage Presets**.
2. In the **Assays / Manage Presets** screen, click the **Sequence Variant Baselines** tab.
3. Place the pointer over the row of the sequence variant baseline that you want to export, then click **Export**.

The sequence variant baseline files are downloaded as a ZIP file and are available for import to another sequencer.

Remove a sequence variant baseline (manager/administrator)

Manager- and administrator-level users can make custom sequence variant baselines unavailable in the software.

1. In the menu bar, click **Assays ▶ Manage Presets**.
2. In the **Assays / Manage Presets** screen, click the **Sequence Variant Baselines** tab.
3. Place the pointer over the row of the sequence variant baseline that you want to make unavailable, then click **Obsolete**.

The status of the sequence variant baseline is **Obsolete** in the list of sequence variant baselines and is unavailable for selection during assay creation.

Exon tile assay baselines

An exon tile assay baseline is used to model the amplicon expression variation within a gene. When added to an assay in Genexus™ Software, the exon tile fusion baseline normalizes a test sample, corrects the coverage in exon-tile amplicons, and identifies the presence or absence of expression imbalance in a gene. Exon tile assay baselines that you have created can only be incorporated into some assays. Contact a Field Bioinformatics Specialist (FBS) for guidance.

An exon tile assay baseline is a TXT file that contains the following items.

- The normalized expression values of exon-tile amplicons for genes with imbalance assays.
- The median normalized expression for each amplicon.

Exon tile assay baselines are associated with a panel BED file in the software.

Genexus™ Software includes predefined exon tile assay baselines.

You can also create an exon tile assay baseline in the software. You can later add the exon tile fusion baseline to an assay and use it as a baseline control for analyses.

An exon tile assay baseline must include at least 40 RNA, DNA & RNA, or TNA samples. Various parameters and thresholds used in the baseline computation determine the inclusion or exclusion of samples in the baseline for each of the genes, for example, ALK, RET, NTRK1.

View exon tile assay baseline details (manager/administrator)

Manager- and administrator-level users can view details for the system-installed exon tile assay baselines, or for custom exon tile assay baselines.

1. In the menu bar, click **Assays ▶ Manage Presets**.
2. In the **Assays / Manage Presets** screen, click the **Exon Tile Assay Baselines** tab.
The **Exon Tile Assay Baselines** table lists both system-installed and custom exon tile assay baselines.
3. Click the name of an exon tile assay baseline.

Review the details in the **Exon Tile Assay Baselines** pane.

Exon Tile Assay Baselines

Item ^[1]	Description
Created By	Sign-in name of the person who created the baseline. System-installed baselines are included with the software.
Created On	Date on which the baseline was created.
Number of Samples	The total number samples that are included in the baseline.
Amplification Type	Library amplification technology that is used in the baseline. Note: This applies only to Ion AmpliSeq™ or Ion AmpliSeq™ HD.

Exon Tile Assay Baselines (continued)

Item ^[1]	Description
Panel Name	Name of the panel that is included in the baseline.
Reference	The file name of the fusion reference for the baseline.

^[1] The details that are shown depend on the setup of the exon tile assay baseline. Parameters used to create exon tile assay baselines are also shown, but are not listed in this table.

You can click **⋮ (More Options)** to perform more actions, such as obsolete, export, copy, edit, or audit the exon tile assay baseline. The options that are available depend on whether the exon tile assay baseline is system-installed or is a custom exon tile assay baseline.

The actions for each baseline are also available in the list of **Exon Tile Assay Baselines** when you place the pointer over the row of a exon tile assay baseline.

Create an exon tile assay baseline (manager/administrator)

Manager- and administrator-level users can create custom exon tile assay baselines for some assays. In some cases, custom exon tile baselines can be added only to the latest version of an assay. For more information about assay versions, see “Available versions of software update packages” on page 265. Contact a Field Bioinformatics Specialist (FBS) for guidance.

By default, you must use at least 40 RNA, DNA & RNA, or TNA samples to create an exon tile assay baseline.

1. In the menu bar, click **Assays ▶ Manage Presets**.
2. In the **Assays / Manage Presets** screen, click the **Exon Tile Assay Baselines** tab.
The **Exon Tile Assay Baselines** table lists both system-installed and custom exon tile assay baselines.
3. Click **+ Add New**.
4. In the **Create Exon Tile Assay Baseline** screen, select the library chemistry for the panel that you want to use for baseline creation.
 - **AmpliSeq**
 - **AmpliSeq HD**
5. In the **Panel** step, enter a name for the baseline, select the panel that you want to use, and enter an optional description for the baseline.
Only panels that are compatible with the selected library chemistry and exon tile assay baseline creation are listed. In some cases, custom exon tile assay baselines can be created only for the latest version of an assay panel.
6. In the **Parameters** step, do not change parameters from the default settings unless you understand how the change can affect the results.
Gene-specific parameters can be enabled with a TXT file.
 - a. Click **Download Example**.

- b. Open the file with a text editor, enter new values if needed, and save the file with a new file name.
 - c. Click **Upload**, navigate to the file, then click **Open**.
 - d. In the **Gene specific threshold** file dropdown list, select the file to enable gene specific thresholds.
 - e. Click **Next**.
7. In the **Samples** step, select the samples to be used in the baseline creation, then click **Next**.
You can add BAM samples or RNA, DNA & RNA, or TNA samples that have been sequenced or a combination of both.
8. In the **Review** step, review the information.
9. Click **Launch**.


The software creates the exon tile assay baseline with a **Status** of **Locked**. The new exon tile assay baseline is listed in the **Exon Tile Assay Baselines** table with a **State** of **Creating**. If there are other active jobs in the software, the **State** is **Pending** until the software starts exon tile assay baseline creation.

When the software completes the process of exon tile assay baseline creation, the **State** changes to **Completed** and the exon tile assay baseline is available to add to an assay. The new exon tile assay baseline is available in the **Fusion Baseline** dropdown list in the **Panel** step when you create an assay that contains a fusion panel that is associated with the new exon tile assay baseline. For more information, see “Panel step assay options” on page 197.

View, add, or remove a note in an exon tile assay baseline (manager/administrator)

Manager- and administrator-level users can view existing notes or add notes to an exon tile assay baseline. Use **Notes** to capture time-stamped written comments and observations for an exon tile assay baseline. An exon tile assay baseline can contain multiple notes from different users.


1. In the menu bar, click **Assays ▶ Manage Presets**.
2. In the **Assays / Manage Presets** screen, click the **Exon Tile Assay Baselines** tab.
3. Place the pointer over the row of the exon tile assay baseline of interest, click **Notes**, then review existing notes or add a new note.

Action	Description
View existing notes for an exon tile assay baseline.	In the Add Notes dialog box, notes that are associated with the exon tile assay baseline are listed by user, date, and time.
Add a new note to an exon tile assay baseline.	<ol style="list-style-type: none"> 1. In the Add Notes dialog box, click + Add Note. 2. Enter the note, then click Save. <p>The note is added to the exon tile assay baseline and includes the username for the account of the person who entered the note, and a timestamp that records the note entry date and time.</p>
Delete an exon tile assay baseline note	In the Add Notes dialog box, in the row of the note that you want to delete, click  (Delete) .

Click **Close** to return to the **Manage Presets** screen.

Import an exon tile assay baseline (manager/administrator)

Manager- and administrator-level users can import an exon tile assay baseline. You can import baselines in ZIP format, and in some instances in TXT format. ZIP-format files contain a fusion panel file and a fusion FASTA file. For more information about the option to import an exon tile assay baseline in TXT format, contact a Field Bioinformatics Specialist (FBS).

1. In the menu bar, click **Assays ▶ Manage Presets**.
2. In the **Assays / Manage Presets** screen, click the **Exon Tile Assay Baselines** tab.
3. Click  **Import Exon Tile Assay Baseline** in the top right of the screen.

Option	Description
Import a ZIP file	<ol style="list-style-type: none"> 1. In File Type, select ZIP. 2. Click Select Exon Tile Assay Baseline file. 3. Browse for and select the file. 4. Click Upload.
Import a TXT file	<ol style="list-style-type: none"> 1. In File Type, select TXT. 2. In the Panel list, select the panel. 3. Click Select Exon Tile Assay Baseline file. 4. Browse for and select the file. The Name field is autopopulated based on the file name. 5. Enter a name for the baseline to change the name from the software-generated name. 6. Click Upload.

A message confirms that the file import was successful when the file import is complete. Imported exon tile assay baselines are locked by the software.

The imported exon tile assay baseline is available in the **Fusion Baseline** dropdown list in the **Panel** step when you create an assay that contains a fusion panel that is associated with the imported exon tile assay baseline. For more information, see “Panel step assay options” on page 197.

Download the log file for custom exon tile assay baselines (manager/administrator)

Manager- and administrator-level users can download and view a log file that shows the details for exon tile assay baselines creation. The log file is available after the exon tile assay baseline creation job is complete. The log file can help with troubleshooting if the exon tile assay baseline creation fails.

1. In the menu bar, click **Assays ▶ Manage Presets**.
2. In the **Assays / Manage Presets** screen, click the **Exon Tile Assay Baselines** tab.
3. Place the pointer over the row of the custom exon tile assay baseline of interest, then click **Download Log**.

Delete an exon tile assay baseline (manager/administrator)

If a custom exon tile assay baseline fails or is terminated during job creation, the exon tile assay baseline can be deleted from the software by a manager- or administrator-level user.

1. In the menu bar, click **Assays ▶ Manage Presets**.
2. In the **Assays / Manage Presets** screen, click the **Exon Tile Assay Baselines** tab.
The **Exon Tile Assay Baselines** table lists both system-installed and custom exon tile assay baselines.
3. Place the pointer over the row of the draft exon tile assay baseline that you want to delete, then click **Delete**.
4. In the **Delete Exon Tile Assay Baseline** dialog box, click **Yes** to confirm the deletion.

The failed or terminated exon tile assay baseline is removed from the software and is no longer included in the **Exon Tile Assay Baselines** table.

Export an exon tile assay baseline (manager/administrator)

Manager- and administrator-level users can export exon tile assay baselines that have been created in Genexus™ Software. System installed exon tile assay baselines cannot be exported.

An exon tile assay baseline can be exported, for example to add the exon tile assay baseline to an assay for use on a different Genexus™ Integrated Sequencer.

1. In the menu bar, click **Assays ▶ Manage Presets**.
2. In the **Assays / Manage Presets** screen, click the **Exon Tile Assay Baselines** tab.
3. Place the pointer over the row of the locked exon tile assay baseline that you want to export, then click **Export**.

The exon tile assay baseline files are downloaded as a ZIP file and are available for import for use with another Genexus™ Integrated Sequencer.

Remove an exon tile assay baseline (manager/administrator)

Manager- and administrator-level users can make exon tile assay baselines unavailable in the software. You cannot designate as obsolete system-installed exon tile assay baselines.

1. In the menu bar, click **Assays ▶ Manage Presets**.
2. In the **Assays / Manage Presets** screen, click the **Exon Tile Assay Baselines** tab.
3. Place the pointer over the row of the exon tile assay baseline that you want to make unavailable, then click **Obsolete**.

The **Status** of the exon tile assay baseline is **Obsolete** in the list of exon tile assay baselines. Exon tile assay baselines that have been made obsolete cannot be added to an assay when an assay is created or copied and edited.

Manage assays

You can view assay details, view the audit trail of an assay, and download assay parameters in Genexus™ Software.

In addition, manager- and administrator-level users can import an assay, export a locked assay, edit, lock, or delete a draft assay, edit the QC parameters of a locked assay, and obsolete a locked assay in the software.

View assay details

You can view assay details in Genexus™ Software.

1. In the menu bar, click **Assays ▶ Manage Assays**.
2. In the **Manage Assays** screen, click the assay name of interest.

The assay details are shown.

If needed, you can click ⋮ (**More Options**) to perform more actions, such as export, copy, edit, lock, delete, obsolete, edit QC, download parameters, or audit the assay. The options that are available depend on whether the assay is system-installed or is a custom assay. For custom assays, the options that are available also depend on whether or not the assay is locked.

The actions for each assay are also available in the **Manage Assays** screen when you place the pointer over the row of an assay. For more information, see “Export a custom, locked assay (manager/administrator)” on page 240, “Copy an assay (manager/administrator)” on page 196, “Edit a draft assay (manager/administrator)” on page 241, “Lock a draft assay (manager/administrator)” on page 242, “Delete a draft assay (manager/administrator)” on page 242, “Remove access to an assay (manager/administrator)” on page 243, “Edit the QC parameters of a locked assay (manager/administrator)” on page 243, “Download assay parameters” on page 243, and “Review and export an assay history” on page 240.

Assay details

When you view assay details in Genexus™ Software, you can view information about the components of the assay. The information that is shown depends on the type of assay.

You can install and use multiple versions of an assay in the software. When there is a new version of an assay that is available to download, the available assay definition file is listed in the ⚙️ (**Settings**) / **Software Updates** screen, in the **Software Updates** table. In some cases, the most recent version (wx.x.x) of an assay allows you to perform actions in the software that you cannot perform with previous versions of the assay. For example, you can only create custom exon tile assay baselines for some versions of some assays. For more information about software updates, see “Software updates” on page 265.


Note: For system-installed assays or assays that you install in the software, the end of the assay name, for example w4.1.2 or w5.0.0, is the version number. The assay version number is not the same as the **Version** shown in the assay details. See the following table for more information about **Version**.

Assay detail ^[1]	Description
Assay creation details	<ul style="list-style-type: none"> • Research Application – The research application for the assay, such as DNA or DNA and Fusions. • Created By – The sign-in name of the person who created the assay. Assays that are included with the software are labeled as System Installed. • Analysis Pipeline Version – The version of the analysis pipeline used for the assay. • Created On – The date on which the assay was created. • Assay Name – The name and version of the assay. • Version – The revision number tracks the number of times that changes that have been made to the assay in the software. For example, when you edit a custom assay for the first time, the version changes from 1 to 2.
Panel	<p>In addition to other components, the panel section lists the reference files used in an assay.</p> <p>One item that is listed is the Minimum Read Count Per Sample, which is the minimum number of reads per sample that are needed to achieve the coverage depth required for the assay. The minimum read counts per sample is used to calculate the number of samples that are allowed in a chip lane when you plan a run.</p> <p>For more information and a complete list of components in the panel section, see “Panel step assay options” on page 197.</p>
Reagents	<p>In addition to other components, the reagents section lists the kits and chips for the assay.</p> <p>For more information and a complete list of components in the reagents section, see “Reagent step assay options” on page 199.</p>
<ul style="list-style-type: none"> • NTC QC – RNA • CF-1 Control • Purification QC • NTC QC – DNA • Run QC • Sample QC 	<p>The QC sections list the Quality Control (QC) metric values for the assay.</p> <p>For more information and a complete list of components in the reagents section, see “QC step assay options” on page 203.</p>
Plugins	<p>The plugins sections lists the plugins that are selected to run with the assay.</p> <p>For more information about plugins, see Chapter 10, “Plugins for data analysis”.</p>
Description	A short description of the assay, similar to the assay full name.

^[1] The details that are shown depend on the assay.

Import an assay (manager/administrator)

A manager- or administrator-level user can import an assay from another Genexus™ Integrated Sequencer if the assay has been first exported to a local drive.

1. In the menu bar, click **Assays ▶ Manage Assays**.
2. In the **Manage Assays** screen, click  **Import Assay**.
3. In the **Import Assay** dialog box, click **Select Assay file**, then go to the folder on the computer that contains the exported assay and select the ZIP file for the assay.
4. Click **Upload**.

The assay appears in the list of assays in the **Assays ▶ Manage Assays** screen.

Export a custom, locked assay (manager/administrator)

An assay can be exported, for example to use that assay in another Genexus™ Integrated Sequencer. You cannot export system-installed assays. Only locked, custom assays can be exported.

1. In the menu bar, click **Assays ▶ Manage Assays**.
2. In the **Assays ▶ Manage Assays** screen, place the pointer over the row of a locked, custom assay, then click **Export**.

The assay parameter files are downloaded to the computer as a ZIP file and are available for import to another sequencer.

Panel reference files are not included in the exported file.

Review and export an assay history

You can review, export, or print the entire history of changes for an assay. The history shows the original values and any new values or activities.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

1. In the menu bar, click **Assays ▶ Manage Assays**.
2. In the **Manage Assays** screen, place the cursor in the row for the assay of interest, then click **Audit**.

The **Audit Details** screen opens with time-stamped audit records that include assay details and changes for each action performed on the assay.

3. (Optional) Review the audit record.

Details in the audit record

Item	Description
User name	The name of the person who was signed in when the action was performed.
Assay Full Name	The name of the assay.
Data Object Name	The data object that is affected by the action.
Action Performed	Actions performed on the assay include: <ul style="list-style-type: none">• Create• Edit• Delete• Obsolete
Time Stamp	The time and date of the action.
ID	An identifier to represent a specific action in the audit record. Each action in the audit record includes a unique identifier.
New Values	A list of all values in the assay. If a value changed as a result of the action, the new value is shown. If no change was made, the value is listed as NA . For example, Plan Executed Status=false indicates a change; Template Kit Barcode=NA indicates that the value did not change.
Old Values	A list of the previous values in the assay. Values are listed for each change. For example, State = Draft indicates that a change was made to the state of the assay. The new value is listed in the New Values section.

4. (Optional) Click **Download** to download a print-ready PDF file of the audit record.

You can open the PDF file in an appropriate viewer, then print the record from within the open document.

5. Click **Back** to return to the **Manage Assays** screen.

Edit a draft assay (manager/administrator)

When a manager- and administrator-level user first creates an assay, the assay name is followed by **Draft**. While the assay is in draft status, it can be edited. A locked assay cannot be edited. Each time you edit a draft assay, the software creates a new version of the draft assay. The version number of the assay is shown in the assay details. For more information, see “View assay details” on page 238. You can view the history of changes made to a draft assay. For more information, see “Review and export an assay history” on page 240.

You can optimize the configuration of an assay when you edit an assay. You can plan a run with a draft assay, view the results, then change the configuration and plan new runs before you lock the draft assay.

IMPORTANT! Use the default parameter settings for assays unless you are an advanced user. Contact your Field Bioinformatics Specialist (FBS) for assistance.

1. In the menu bar, click **Assays ▶ Manage Assays**.
2. In the **Manage Assays** screen, place the pointer over the row of a draft assay, then click **Edit**. The **Edit Assay** workflow reopens.
3. Edit the options on each assay step as desired, then click **Save**.

The assay remains a draft until you lock the assay. For more information, see “Lock a draft assay (manager/administrator)” on page 242.

Lock a draft assay (manager/administrator)

When a manager or administrator first creates an assay, the status of the assay is **Draft**. After an assay is locked, it cannot be edited or deleted.

1. In the menu bar, click **Assays ▶ Manage Assays** to open the **Manage Assays** screen. An assay full name followed by **Draft** indicates that the assay is in draft status.
2. In the **Manage Assays** screen, place the pointer over the row of a draft assay, then click **Lock**.

The assay is now locked and cannot be edited or deleted. **Draft** is removed from the assay full name in the **Assays ▶ Manage Assays** screen.

Delete a draft assay (manager/administrator)

When an assay is in draft status, it can be deleted from the software. Only draft assays can be deleted. Draft assays are indicated by **Draft** next to the **Assay Full Name**.

1. In the menu bar, click **Assays ▶ Manage Assays**.
2. In the **Manage Assays** screen, place the pointer over the row of a draft assay, then click **Delete**.
3. In the **Delete Assay** dialog box, click **Yes** to confirm the deletion.

The draft assay is removed from the software and is no longer available in the **Assays / Manage Assays** screen.

Edit the QC parameters of a locked assay (manager/administrator)

Manager- and administrator-level users can edit the QC parameters of a locked assay. The QC parameters can be adjusted according to research needs that can change over time. You can edit the QC parameters of any locked assay, including system-installed assays, if needed. When the QC parameters are changed, the edited QC parameters are applied to any new run that uses the assay. Edits to QC parameters do not affect runs that use the assay and are completed before edits are made.

1. In the menu bar, click **Assays ▶ Manage Assays**.
2. In the **Manage Assays** screen, place the pointer over the row of a locked assay, then click **Edit QC**.
The **Edit QC** dialog window opens.
3. Edit the QC parameters as desired.
To reset the QC parameters to the original values in a subsection, such as **Run QC** or **Sample QC – DNA**, click **Reset** in the upper right corner of the subsection.

4. Click **Save**.

The new QC parameter values are saved and applied in any run that uses the assay after the changes are saved.

Remove access to an assay (manager/administrator)

A manager- or administrator-level user can remove a locked, custom assay from use in the software by designating it obsolete. The assay is not deleted and a record of it is maintained in the audit trail. The results for any runs already performed with the assay remain available in the software. System-installed assays cannot be made obsolete.

1. In the menu bar, click **Assays ▶ Manage Assays**.
2. In the **Manage Assays** screen, place the pointer over the row of a locked assay, then click **Obsolete**.
3. Click **Yes** to confirm the operation.
The assay is no longer available for use in a run.

Download assay parameters

Assay parameters can be downloaded and exported, for example to use that assay in another Genexus™ Integrated Sequencer.

Panel reference files are not included in the export.

1. In the menu bar, click **Assays ▶ Manage Assays**.
2. In the **Manage Assays** screen, place the pointer over the row of a locked assay, then click **Download Parameters**.

The assay parameter files are downloaded to the computer as a ZIP file that contains JSON files.

The assay parameter files are available for import to another sequencer.

Gene lists (manager/administrator)

A gene list is a collection of genes relevant to a particular cancer type. In Genexus™ Software, there are system-installed gene lists. Manager- and administrator-level users can also create custom gene lists that are relevant to their research. Each gene list is specific for a particular cancer type. You can create multiple gene lists for each cancer type, but only one gene list can be designated as the default for that cancer type.

When a gene list is designated as the default gene list for a particular cancer type, that gene list is applied, by default, to the sample results. You can change the gene list that is applied to the Key Variants matrix in the sample results. For more information, see “View the Key Variants matrix” on page 125.

Although you cannot directly edit system installed gene lists, you can indirectly edit the lists. When you create a new gene list in the software for a cancer type that has a system installed gene list, the software generates a list of draft genes to include in the gene list as a starting point. The genes initially included in the gene list are populated with the system installed genes for that cancer type. Manager- and administrator-level users can then add or remove genes to create a custom gene list that is derived from the genes included in a system installed gene list.

A record of how each gene list is used, removed, or changed, and by whom, is maintained in the audit trail of a gene list.

View a gene list (manager/administrator)

Manager- and administrator-level users can view the genes included in a gene list in Genexus™ Software. For each type of cancer, a gene list can be designated as the default gene list. Default gene lists are designated with a ☆ after the gene list name.

1. Click **Assays ▶ Manage Gene Lists** to open the **Manage Gene Lists** screen.
2. In the **Name** column, click the gene list name of interest.
Details for the gene list are shown. For more information, see “The View Gene List pane (manager/administrator)” on page 245.
3. When you are finished viewing the gene list details, click **X** to return to the **Manage Gene Lists** screen.

The View Gene List pane (manager/administrator)

Manager- and administrator-level users can select a gene list in the **Manage Gene Lists** screen in Genexus™ Software to view details for the gene list in the **View Gene List** pane.


Detail	Description
Name	The name of the gene list.
Cancer Type	Cancer type for the gene list
Content Version	The version of the gene list.
Default for Cancer Type	Indicates if the gene list is designated as the default gene list. Each cancer type can have only one default gene list.
Created On	The date the gene list was created in the software.
Created By	Sign-in name of the person who created the gene list. System-installed gene lists are created by System Installed .
Updated On	The date the gene list was updated in the software.
Updated By	Sign-in name of the person who last updated the gene list.
Relevant Genes	The genes in the gene list.

Create a gene list (manager/administrator)

Manager- and administrator-level users can create a new gene list in Genexus™ Software. If a gene list does not already exist in the software for the cancer type that is associated with the gene list when a gene list is created, the new gene list is set as the default gene list for that cancer type.

When a gene list is designated as the default gene list for a specific cancer type, that gene list is applied, by default, to the sample results. You can change the gene list that is applied to the **Key Variants** matrix in the sample results. For more information, see “The Key Variants matrix” on page 124.

1. Click **Assays ▶ Manage Gene Lists** to open the **Manage Gene Lists** screen.
2. Click **+ Add New**.
3. In the **Create Gene List** screen, enter a unique gene list name, then complete the remaining sections.
 - a. In **Cancer Type**, select the cancer type for the new gene list.
If a default gene list for the selected cancer type already exists in the software, the relevant genes are populated by the software. If a default gene list for the selected cancer type does not already exist in the software, the new gene list is designated as the default gene list for the cancer type.
 - b. To set the new gene list as default, enable the **Set as Default** toggle switch.
 - c. In the **Relevant Genes** field:

Option	Description
Remove a gene from the gene list	<p>Click X in the gene box for the gene you want to remove.</p> 
Add a gene to the gene list.	<ol style="list-style-type: none"> 1. Enter the name of partial name of a gene to autopopulate a master gene list. 2. Select the gene from the populated list. <p>In this example, typing MAGE in the Relevant Genes field allows you to select and add the MAGEA3 gene from the master gene list.</p> <p>Create Gene List</p> <p>Name * <input type="text" value="test"/> ✓ Cancer Type * <input type="text" value="Melanoma"/> ✓ ↕</p> <p>Relevant Genes *</p> <p><input type="text" value="BRAF"/> X <input type="text" value="KIT"/> X <input type="text" value="NTRK1"/> X <input type="text" value="NTRK2"/> X <input type="text" value="NTRK3"/> X <input type="text" value="mage"/></p> <p>MAGEA10-MAGEA5 MAGEA11 MAGEA12 MAGEA2 MAGEA2B MAGEA3 MAGEA4</p>

4. Click **Save**.

The new gene list is included in the **Manage Gene Lists** screen and can be applied to filter the genes displayed in the **Key Variants** matrix. For more information, see “The Key Variants matrix” on page 124.

Edit a gene list (manager/administrator)

Manager- and administrator-level users can edit the genes in custom gene lists in Genexus™ Software. Only custom gene lists that are not locked can be edited. System-installed gene lists are always locked and cannot be edited.

1. Click **Assays** ▶ **Manage Gene Lists**.
2. In the **Manage Gene Lists** screen, place the pointer over the row of the gene list that you want to edit, then click **Edit**.

3. In the **Edit Gene List** screen, in the **Relevant Genes** field:

Option	Description
Remove a gene from the gene list	Click X .
Add a gene to the gene list.	<ol style="list-style-type: none"> 1. Enter the name of partial name of a gene to autopopulate a master gene list. 2. Select the gene from the populated list.

4. Click **Save**.

The updated gene list can be applied to filter the genes that appear in the **Key Variants** matrix. For more information, see “The Key Variants matrix” on page 124.

Change the default gene list (manager/administrator)

Manager- and administrator-level users can change the default gene list for a cancer type. When a gene list is designated as the default gene list for a particular cancer type, that gene list is applied to the sample results. For more information, see “The Key Variants matrix” on page 124.

You must have more than one gene list for the cancer type to change the default gene list for that cancer type. When there is only one gene list for a cancer type, that gene list is the default gene list. For more information about default gene lists, see “Gene lists (manager/administrator)” on page 244.

1. Click **Assays** ▶ **Manage Gene Lists**.
2. In the **Manage Gene Lists** screen, place the pointer over the row of the gene list that you want to designate as default, then click **Set as default**.
3. Click **Yes** to confirm that you want to change the default gene list.

The gene list is noted as default in the **Manage Gene Lists** screen and is applied to the **Key Variants** matrix in the sample results for the cancer type. Default gene lists include a ☆ after the gene list name.

Delete a gene list (manager/administrator)

Manager- and administrator-level users can delete custom gene lists that have a draft status in Genexus™ Software. Only custom gene lists that are not locked can be deleted. System installed gene lists are always locked and cannot be deleted.

1. Click **Assays** ▶ **Manage Gene Lists**.
2. In the **Manage Gene Lists** screen, place the pointer over the row of the gene list that you want to delete, then click **Delete**.
The **Confirm** dialog box opens with the message *Are you sure that you want to delete the Gene List?*
3. In the **Confirm** dialog box, click **Yes** to delete the gene list.

The gene list is removed from the software and is no longer listed in the **Manage Gene Lists** screen.

Lock a gene list (manager/administrator)

Manager- and administrator-level users can lock custom gene lists in Genexus™ Software. Custom gene lists that are locked cannot be edited or deleted. System installed gene lists are always locked.

1. Click **Assays** ▶ **Manage Gene Lists**.
2. In the **Manage Gene Lists** screen, place the pointer over the row of the gene list that you want to lock, then click **Lock**.
The **Confirm** dialog box opens with the message *Are you sure that you want to lock the Gene List?*
3. In the **Confirm** dialog box, click **Yes** to lock the selected gene list.

The gene list is now locked and cannot be edited or deleted.

Remove a gene list (manager/administrator)

Manager and administrator-level users can remove custom gene lists that have been locked from use in Genexus™ Software. You cannot designate as obsolete system-installed gene lists. Obsolete gene lists are not available to apply to sample results, but are listed in the **Manage Gene Lists** screen with a **Status of Obsolete**. You can view the gene list and the audit trail for gene lists that are obsolete. You cannot reactivate gene lists that are obsolete.

1. Click **Assays** ▶ **Manage Gene Lists**.
2. In the **Manage Gene Lists** screen, place the pointer over the row of the gene list that you want to remove, then click **Obsolete**.
The **Confirm** dialog box opens with the message *Are you sure that you want to obsolete the Gene List?*
3. In the **Confirm** dialog box, click **Yes** to obsolete the gene list.

The gene list is listed in the **Manage Gene Lists** screen with the status of **Obsolete**.



Review a gene list history (manager/administrator)

Manager- and administrator-level users can review and export the entire history of a gene list in Genexus™ Software. The history shows the original gene list values and any new values or activities. The following actions generate an audit record:

- Create a new gene list.
- Edit a gene list.
- Lock a gene list.
- Delete a gene list.
- Obsolete a gene list.
- Content version updates that change the genes in system installed gene lists.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

1. Click **Assays** ▶ **Manage Gene Lists**.
2. In the **Manage Gene Lists** screen, place the pointer over the row of the gene list that you want to review, then click **Audit**.
The **Audit Details** screen opens with time-stamped audit records that include changes for each action performed on the gene list. For more information, see “Details in the gene list audit record” on page 249.
3. In the **Audit Details** screen:

Option	Description
Review the audit trail details.	Each action that was performed for the gene list, such as gene list creation or edits, is listed.
Export the audit record.	Click  Download to generate a PDF file. Depending on the browser settings, the software automatically downloads the file or prompts you to open or save the file.
Close the Audit Details screen.	Click  Back to return to the Manage Gene Lists screen.

Details in the gene list audit record

The **Audit Details** screen for gene lists shows the actions that are performed on a gene list.

Audit details for gene lists

Item	Description
User Name	The name of the person who was signed in when the action was performed.
Gene List Name	The name of the gene list.
Data Object Name	The data object that is affected by the action. For gene lists, the data object is Gene List .
Action Performed	Actions performed on the gene list include: Create, Edit, Lock, Delete, and Obsolete. Content version updates that change the genes in system installed gene lists are also included.
Time Stamp	The time and date of the action.
ID	An identifier for the gene list.
New Values	A list of new values in the gene list for the listed action. If a value changed as a result of the action, the new value is shown. If no change was made to the genes in the gene list, the value is listed as NA for the action. For example, when the action is to change the default gene list, but there is no change to the genes included in the list, Genes=NA is shown. A change to the default gene list is designated as Primary . For example, Primary = false indicates that the action made another gene list the default gene list.
Old Values	A list of the previous values in the assay. Values are listed for actions that resulted in a change.



System administration and management (manager/administrator)

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Manager-level users use tools in the ⚙️ **(Settings)** menu to:

- Create and link user accounts to get software updates from the Connect App Store, and use Ion Reporter™ Software.
- View system information and settings.

Administrator-level users use tools in the ⚙️ **(Settings)** menu to:

- View and export audit records.
- Configure user email settings, set up laboratory information, and audit these settings.
- Create and link user accounts for to get software updates from the Connect App Store, use Ion Reporter™ Software.
- Create and manage account settings, and audit these accounts.
- Manage software updates.
- Manage data backup, disk cleanup settings, and disk space usage.
- Manage log files.
- Manage services for servers, services, and active jobs.
- Create and manage user accounts.

Linked accounts in Genexus™ Software (manager/administrator)

Manager- and administrator-level users can configure Genexus™ Software to link the following accounts in Genexus™ Software.

Account Type	Description
Thermo Fisher Connect	<p>When a Thermo Fisher Connect account is configured and active, Administrator-level users can perform the following tasks in the Genexus™ Software.</p> <ul style="list-style-type: none"> • Download the latest software updates • Download more plugin software • Download assay definition files • Download software configuration packages <p>The first Thermo Fisher Connect account that is added in the software is designated as the administrator (Admin) account and cannot be unlinked or removed.</p>
Genexus	Upload samples results to another Genexus™ Software server.
Ion Reporter Local	Upload samples results to an Ion Reporter™ Software on Connect account, and then use the Ion Reporter™ Software to further analyze those results.
Ion Reporter Server	Upload samples results to an Ion Reporter™ Server, and then use the Ion Reporter™ Software to further analyze those results.

Create a Connect account

Before you configure a **Thermo Fisher Connect** account in Genexus™ Software, you must have a valid Connect account on the thermofisher.com website.

1. Navigate to thermofisher.com/connect.
2. Click **Sign up now**, then enter the information to create a new account.
3. Click **Create account** at the bottom of the screen to complete the registration.



Configure a Thermo Fisher™ Connect account with a QR code (manager/administrator)



Manager- and administrator-level users can link their Connect account in Genexus™ Software with a QR code generated in the software.

When a Connect account is configured and active, administrator-level users can perform the following tasks in the Genexus™ Software:

- Download the latest software updates
- Download more plugin software
- Download assay definition files
- Download software configuration packages

Before you link a Connect account to create a **Thermo Fisher Connect** account, you must have a valid Connect account. You must also have the InstrumentConnect mobile app to link the account with a QR code that is scanned on a mobile device. For more information, see the *Thermo Fisher™ Connect Help System* (Pub. No. MAN0010954).

1. Click  **(Settings) ▶ Thermo Fisher Account**.
2. Click  **Link Account to Device**.
 The **Link Account to Device** dialog box opens and a QR code image and QR code text are shown.
3. Link the Connect account with the software.


Option	Description
Link a Connect account with the InstrumentConnect mobile app	<ol style="list-style-type: none"> 1. Open the InstrumentConnect app, then sign in. 2. In the app, click . 3. Click QR Code, then scan the QR code shown in the Link Account to Device dialog box in Genexus™ Software.
Link the account through the Connect website	<ol style="list-style-type: none"> 1. Click the link in the QR Code to open the Thermo Fisher Connect website. 2. In the Connect website, click  InstrumentConnect in the left sidebar. 3. Click Link instrument in the upper right corner of the screen. 4. Enter the QR code text shown in the Link Instrument screen. 5. Click Link.


The linked account is listed in the  **(Settings) / Thermo Fisher Account** screen. A successfully authenticated account has **Active** listed in the **Status** column.


When the **Thermo Fisher Account** is active, software updates are available in Genexus™ Software. Administrator-level users can download and install the updates. For more information, see “Software updates” on page 265.

Link Genexus™ Software accounts (manager/administrator)

Manager- and administrator-level users can link Genexus™ Software accounts. After the accounts are linked, you can transfer results from one Genexus™ Software account to another.

1. In Genexus™ Software, click  **(Settings)** ▶ **Thermo Fisher Account**.
2. In the **Thermo Fisher Account Settings** screen, click **+ Create Account**.
3. In the **Create Thermo Fisher Account** dialog box, enter the information that is required to create the account.

Item	Description
Account Type	Select Genexus .
Name	Enter a name to identify the account in the Thermo Fisher Account Settings screen in Genexus™ Software. The name can contain only alphanumeric characters (0-9, Aa-Zz), periods (.), underscores (_), or hyphens (-). For example, enter <i>Lab_Admin</i> .
User Name	Enter the name that is used to sign in to the Genexus™ Software account that you want to link.
Password	Enter the password for the Genexus™ Software account that you want to link.
Server	Enter the name of the Genexus™ Software server that you want to use to upload results files.
Port	Enter the port of the Genexus™ Software server.
Version	Click  Get Ion Reporter Software Versions , then select the software version.
Set as Default Account	Click the toggle switch to make the account the default account that is used to upload results files.

The configured account is listed in the  **(Settings)** / **Thermo Fisher Account** screen. The **Account Type** is **Genexus**. A successfully authenticated account has **Active** listed in the **Status** column.



Configure an Ion Reporter™ Software on Connect account (manager/administrator)


Manager- and administrator-level users can configure a link to an Ion Reporter™ Software on Connect account in Genexus™ Software.

Ion Reporter™ Software on Connect, the cloud-based platform, is available at <https://ionreporter.thermofisher.com>.

When the link is configured, you can upload results files from Genexus™ Software directly to the Ion Reporter™ Software on Connect and use Ion Reporter™ Software for further data analysis, annotation and reporting. Unmapped BAM files for the sample results are uploaded.

Access to Ion Reporter™ Software on Connect is controlled by your [thermofisher.com](https://www.thermofisher.com) user name and password. Before you configure the Ion Reporter™ Software on Connect account, you must have a valid Connect account on the [thermofisher.com](https://www.thermofisher.com) website. Users who use the account to access Ion Reporter™ Software on Connect must also have a valid Thermo Fisher account.

1. Click  **(Settings) ▶ Thermo Fisher Account**.
2. In the **Thermo Fisher Account Settings** screen, click  **Create Account**.
3. In the **Create Thermo Fisher Account** dialog box, complete the information required to configure the account.

Option	Description
Account Type	Select IonReporter Cloud .
Name	Enter a name to identify the account in the list of Thermo Fisher Account Settings in Genexus™ Software.
User Name	This must be the user name (email address) for a valid Ion Reporter™ Software on Connect account.
Password	Enter the password for the Ion Reporter™ Software on Connect account.
Version	Click  Get Ion Reporter Software Versions , then select the software version.
<i>(Optional)</i> Set as Default Account	Click the toggle switch if you want to make the account the default account that is used to upload results files.



4. Click **Submit**.

The configured Ion Reporter™ Software on Connect account is added to the **Thermo Fisher Account Settings** table. (See the row with the **Account Name** that you used listed as a **Thermo Fisher Cloud** account.) The account can be selected in the **Setup** step of a run plan to automatically upload results file to Ion Reporter™ Software when the run is complete.


Results files can also be uploaded to Ion Reporter™ Software from the **Results / Run Results** screen. For more information, see “Upload results files to Ion Reporter™ Software” on page 171.

Configure an Ion Reporter™ Server account (manager/administrator)

Manager- and administrator-level users can configure a link to an Ion Reporter™ Server account in Genexus™ Software. When the server is configured, you can upload results files from Genexus™ Software directly to the Ion Reporter™ Server and use Ion Reporter™ Software for further data analysis, annotation and reporting. The uploads contain unmapped BAM files.


1. Click  **(Settings) ▶ Thermo Fisher Account**.
2. In the **Thermo Fisher Account Settings** screen, click  **Create Account**.

3. In the **Create Thermo Fisher Account** dialog box, complete the information required to configure the account.

Option	Description
Account Type	Select IonReporter Local
Name	A name to identify the account in the list of Thermo Fisher Account Settings
User Name	This must a username for a valid Ion Reporter™ Server account.
Password	Enter the password for the Ion Reporter™ Server account.
Server	Enter the name of the Ion Reporter™ Server that you want to use to upload results files.
Version	Click  Get Ion Reporter Software Versions , then select the software version.
Port	Enter the port of the Ion Reporter™ Server.
<i>(Optional)</i> Set as Default Account	Enable the toggle switch to make this account a default account to upload results files.

4. Click **Submit**.

The configured Ion Reporter™ Server account is added to the **Thermo Fisher Account Settings** screen. The account can be selected in the **Setup** step of a run plan to automatically upload sample results files and launch analyses in Ion Reporter™ Software. Results files can also be uploaded to Ion Reporter™ Software from the **Results / Run Results** screen. For more information, see “Upload results files to Ion Reporter™ Software” on page 171.


The configured account is listed in the  **(Settings) / Thermo Fisher Account** screen. A successfully authenticated account has **Active** in the **Status** column. The software version for the Ion Reporter™ Software is listed in the **Version** column.

Manage connected accounts

Manager- and administrator-level users can edit, unlink, delete, obsolete, and view changes made to the accounts that are connected to Genexus™ Software.

Edit a connected account (manager/administrator)


Manager- and administrator-level users can edit the connected accounts in Genexus™ Software. For example, edit a connected account when the password of the connected account has changed.

1. Click  **(Settings) ▶ Thermo Fisher Account**.
2. In the **Thermo Fisher Account Settings** screen, place the pointer over the row of the account that you want to edit, then click **Edit**.
3. In the **Edit Thermo Fisher Account** dialog box, enter the new information, then click **Submit** to save the changes.

Unlink a Thermo Fisher™ Connect account (manager/administrator)

Manager- and administrator-level users can unlink Thermo Fisher™ Connect accounts in Genexus™ Software.


The first Thermo Fisher™ Connect account that is added is designated as the administrator (Admin) account and cannot be unlinked or removed in the software.

1. Click  **(Settings)** ▶ **Thermo Fisher Account**.
2. Place the pointer over the row of the account that you want to unlink, then click **Unlink**.

The Thermo Fisher™ Connect account is no longer linked in Genexus™ Software.


Obsolete a connected account (manager/administrator)

Manager- and administrator-level users can obsolete accounts that are connected in Genexus™ Software.

1. Click  **(Settings)** ▶ **Thermo Fisher Account**.
2. In the **Thermo Fisher Account Settings** screen, place the pointer over the row of the account that you want to obsolete, then click **Obsolete**.

Delete a connected account (manager/administrator)



Manager- and administrator-level users can delete accounts that are connected in Genexus™ Software.

1. Click  **(Settings)** ▶ **Thermo Fisher Account**.
2. In the **Thermo Fisher Account Settings** screen, place the pointer over the row of the account that you want to delete, then click **Delete**.


The account is removed from the **Thermo Fisher Account Settings** screen.

View the changes made to a connected account (manager/administrator)

Manager- and administrator-level users can view the changes made to connected accounts in Genexus™ Software.

1. Click  **(Settings)** ▶ **Thermo Fisher Account**.
2. In the **Thermo Fisher Account Settings** screen, place the pointer over the row of the account that you want to audit, then click **Audit**.
3. In the **Audit Details** screen, view the most recent change for the selected account.
4. In the **Audit Record Details** pane, click a time stamp of interest to view the details for other changes that have been made to the selected account.
5. (Optional) Click  **Download** to export the record of the single change to a PDF file.

User accounts (administrator)

Administrator-level users can perform user management actions in the  **(Settings) / User Management** screen.



- Add new user accounts
- Assign user privileges (roles)
- Edit user information
- View user account audit trails
- Manage user account security policies

Add a new user account (administrator)

Administrator-level users add new user accounts to the software, and grant some users permissions to electronically sign reports.


This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

The email settings in the software must be configured before new users can receive email notifications. For more information, see “Configure email notifications (manager/administrator)” on page 263.


1. In the menu bar, click  **(Settings) ▶ User Management**.
2. In the **User Management** screen, click  **Add New**.
3. In the **Create User** dialog box, enter the user account information.
User names can contain alphanumeric characters and underscores, periods, and hyphens.
4. From the **Role** list, select the appropriate user-access level (**Administrator**, **Manager**, or **Operator**).
5. To allow a user to sign reports (manager- and administrator-level users only), click the **Electronic Signature** toggle switch.
6. Click **Save**.
The software sends an email notification containing a temporary password to the email address of the new user.

Sort the user list (admin)


Administrator-level users can sort the user list by user name, first name, and last name.

1. In the menu bar, click  **(Settings) ▶ User Management**.
2. In the **User Management** screen, click the column for **User Name**, **First Name**, or **Last Name** to sort the column alphabetically.
3. Click the column name a second time to reverse the order.

Edit user details (administrator)

Administrator-level users can edit user details in the  **(Settings) / User Management** screen. An administrator can also suspend or disable user accounts. A history of user account changes is maintained by the audit feature.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

1. In the menu bar, click  **(Settings) ▶ User Management**.
2. In the **User Management** screen, place the pointer over the row of the user account, then click **Edit**.
3. In the **Edit User** dialog box, edit the user details.
 - Enter changes in the appropriate account information fields.
 - In the **Role** list, select the user-access level (**Administrator**, **Manager**, or **Operator**).
 - To enable the ability to sign reports (manager- and administrator-level users only), click the **Electronic Signature** toggle switch to the enabled position.
 - In the **State** list, select the appropriate state (**Active**, **Suspended**, or **Disable**).
4. Click **Save** to make the changes.

Reset user password

An administrator-level user resets passwords for user accounts. A history of user password changes is maintained by Genexus™ Software.


After a password is reset, the software sends an email notification to a user of a temporary password, which they must personalize when logging in.

Periodically, the software sends a system-generated email notification to a user that their password is about to expire. The operator follows the directions included in the email notification of a pending password expiration. A user can also request that an administrator reset their password.

Passwords can contain only alphanumeric characters and no special characters or spaces.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

The email settings in the software must be configured before new users can receive email notifications. For more information, see “Configure email notifications (manager/administrator)” on page 263.



1. Click  **(Settings)** ▶ **User Management**.
2. Place the pointer over the row of the user of interest, then click **Reset Password**.
3. In the **Change Password** dialog box, you can change the password in one of two ways.
 - Click **Send password in Email** to send a software-generated password in an email message to the user.
 - Enter a new password into the **Password** field to change the password for the user. Reenter the new password into the **Confirm Password** field, then click **Change**.

The confirmation message "Password reset link sent to your email account" or "Password changed successfully" appears to indicate the password reset.

View user account history (administrator)

An administrator-level user can view the user account history in Genexus™ Software. The user account audit trail is a record of when the user account was created and modified.



This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

1. In the menu bar, click  **(Settings)** ▶ **User Management**.
2. In the **User Management** screen, place the pointer over the row of the user of interest, then click **Audit**.
3. In the **Audit Details** screen, view the audit trail details for the selected user. For more information, in the **Audit Record Details** column, click the time stamp of interest.
4. (Optional) In the **User Management** screen, click  **Download** to export the record to a PDF file.


Set user account security policies (administrator)

An administrator-level user sets security policies for Genexus™ Software user accounts. User account security policies include the permissible number of failed sign-in attempts, password lifetime, and the length of inactivity before users are automatically signed out of the software. These settings apply to all users.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.



1. In the menu bar, click  **(Settings)** ▶ **User Management**.
2. In the **User Management** screen, click  **Policies**.
3. In the **Policies** dialog box, select the appropriate values to set the user-account suspension period, password, and session timeout policies.
4. Confirm that the **Enabled** toggle switch is set to enable the session policy settings.
5. Click **Save**.

Configure an SSH key (system administrator)


System administrator-level users can set up an SSH key in their user profile in the  **(Profile)** / **My Profile** screen. SSH, also known as Secure Shell or Secure Socket Shell, is a network protocol that gives administrator-level users a secure way to access a computer over an unsecured network. SSH also refers to the suite of utilities that implement the SSH protocol.

Configuring an SSH key is optional, but best practice is to configure one or more SSH keys. The ionadmin and ionservice users may need to configure SSH keys so that they can access the system at the operating system level to configure some system-level settings, for example mounting an archive directory on the system. A user can have more than one SSH key.

Before configuring an SSH key, you must first generate keys by using the Keygen command on your local system. When the public and private keys are generated, copy the text string from the public key to use in this procedure.

1. In the menu bar, click  **(Profile)** ▶ **My Profile**.
2. In the **SSH Keys** section, click  **Add new SSH Key**.
The **SSH Keys** section is available only to ionadmin and ionservice users.
3. Complete the **Add new SSH Key** dialog box.
 - a. In **Label**, type a unique name for this SSH key.
 - b. In **SSH Key**, paste or type the SSH key string generated by using the Keygen command on your local system.
 - c. Click **Save**.


Audit records (administrator)

Administrator-level users can use the tools in the  **(Settings) / Audit Records** screen to configure and manage audit records. Information that can be audited includes activities by users on samples, libraries, assays, planned runs, system setup of the sequencer, accounts, and software updates.


The **Audit Records** screen lists actions that are performed on any data object, including the user who performed the action, the action performed, the data object name, and the date and time of the action. Additional details concerning the action are available in an accompanying audit record.

The actions can be searched and sorted, and the audit records can be exported and printed.



This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

Note: Library batches and runs that are created in a batch with other objects in a single LIMS transaction do not have a "Create" action that is listed in the  **(Settings) / Audit Records** screen. However, you can view complete library batch audit records from the **Samples / Manage Libraries** and **Runs / Manage Runs** screens. Any subsequent actions that are performed on library batches are listed in the **Audit Records** screen. A library batch or run that is created individually in a LIMS transaction is listed.

Search audit records (administrator)

An administrator-level user can use the search tools in the  **(Settings) / Audit Records** screen to search for an existing audit record.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.


1. In the menu bar, click  **(Settings) ▶ Audit Records**.
2. In the **Audit Records** screen, select the search parameters.
 - a. To select a date range, click  **Calendar** next to **Start Date** and **End Date**.

If an end date is not selected, the search results include all records from the start date through the current date.
 - b. To limit the results list to actions that are performed by a specific user, select the user from the **Select User** list.
 - c. To limit the results to specific actions, select from the **Select Action** list.
 - d. To list more items in the table, select from the data object list.


The list is set to **Sample** by default and all items listed in the **Audit Records** screen have the data object of **Sample** in the **Data Object Name** column. You can change the type of objects that are shown in the table. For example, when you select **Planned Runs** in the data object list, all planned runs are listed.

3. Click **Search**.
4. Click **Clear** to return to the default list of records, with the data object of **Sample** in the **Data Object Name** column.


Sort audit records (administrator)

An administrator-level user can sort the columns in the  **(Settings) / Audit Records** screen to display the list of audit records in a different order. The default order is the newest record on top.



This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

1. In the menu bar, click  **(Settings) ▶ Audit Records**.
2. In the **Audit Records** screen, click the column header of interest.
The list of records is reordered based on the heading selected. **User**, **Action Performed**, and **Data Object Name** sort alphabetically.
3. Click the column header a second time to reverse the order of records displayed.
4. Click **Clear** to return to the sorted column to the default order.
5. Click the **Timestamp** heading to return to the default order, with the most recent record on top.

Export audit records (administrator)

An administrator-level user uses the  **(Settings) / Audit Records** screen to export a print-ready PDF file of selected audit records.



This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

1. In the menu bar, click  **(Settings) ▶ Audit Records**.
2. In the **Audit Records** screen, select the record or records to be exported by clicking the checkbox next to the record of interest. Select all of the records in the screen by selecting the checkbox above the column.
3. Click  **Export**.
A PDF file is generated. Depending on your browser settings, the software automatically downloads the file or prompts you to open or save the file.
4. Open the PDF file in an appropriate viewer to print.

Update the audit configuration (administrator)

An administrator-level user configures the audit details by requiring that a reason for a change be included in audit records when a user changes information. When a reason is required, the user must supply a reason for change when editing the information, and the reason is included in the audit record.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

1. In the menu bar, click  **(Settings)** ▶ **Audit Records**.
2. In the **Audit Records** screen, click  **Audit Configuration**.
3. In the **Audit Configuration** dialog box, select the **Require Reason** checkbox next to the **Data Object Name** for which a reason for change is required.
4. Click **Save**.

To remove a reason for change requirement, open the **Audit Configuration** dialog box, deselect the checkbox, then click **Save**.


Configure email settings (manager/administrator)

Manager- and administrator-level users configure email settings. Administrator-level users can configure support contact information, such as the lab contact and the IT contact for the organization.


Configure email notifications (manager/administrator)

Manager- and administrator-level users configure the Genexus™ Software to send email notifications to a specified administrator.

Administrator-level users receive email notifications when there is a restore system failure. Administrator-level users can also select to receive email notifications when other conditions are met, such as when the system has limited free disk space remaining. For more information, see “Change email notification settings (administrator)” on page 279.

1. Obtain the SMTP server URL and port information from your IT department.
2. Click  **(Settings)** ▶ **Configuration**.
3. In the **Configuration** screen, in the **Email Settings** section, enter the server and port information supplied by IT, and the Genexus™ Software user name, password, and email address of the administrator.
 - Keep the **Use TLS** toggle switch enabled to send email notifications with transport layer security.
 - Click the **Use TLS** toggle switch to send email notifications without transport layer security.
4. Click **Update**.
5. Click **Send Test Email** to send a test email to the email address provided in the settings.

Lab information


The **Lab Information** tab in the  **(Settings) / Configuration** screen provides a reference for support personnel, listing points of contact if a problem with the connected Genexus™ Integrated Sequencer occurs.

- **Lab Contact**—This is the person in your organization who should be notified during a support request of problems related to the sequencer.
- **IT Contact**—This is the person in your organization who should be notified during a support request of problems related to the sequencer hardware or the network environment.

IMPORTANT! Administrator and Manager-level users should review the contact information periodically to ensure that it is current and accurate.



Edit the reference lab information (manager/administrator)

Manager- and administrator-level users can edit the reference lab information in Genexus™ Software. You can view the history of changes to the contact information. For more information, see “View the changes made to the lab information (manager/administrator)” on page 264.

1. Click  **(Settings) ▶ Configuration**.
2. In the **Lab Information** section, below the **Lab Contact** or **IT Contact** pane, click **Edit**.
The fields for the **Lab Contact** or **IT Contact** are now available to edit.
3. Enter the updated contact information.
4. Click **Save** to save the new selections or click **Reset** to revert to the original information.

View the changes made to the lab information (manager/administrator)

Manager- and administrator-level users can view the changes made to the lab information in Genexus™ Software.

1. Click  **(Settings) ▶ Configuration**.
2. In the **Lab Information** section, in the **Lab Contact** or **IT Contact** pane, click **Audit**.
3. In the **Audit Details** screen, view the most recent change for the selected contact.
4. In the **Audit Record Details** pane, click a time stamp of interest to view the details for other changes that have been made for the selected contact.
5. (Optional) Click  **Download** to export the record of the single change to a PDF file.

Software updates

An administrator-level user can select **Software Updates** in the ⚙️ (**Settings**) menu to:

- See the currently installed software versions for each software module, assay definition files, plugin, and configuration package installed on the system under the **Installed Software** link.
- Check for new software updates that are available to be downloaded and installed under the **Software Updates** link.

For users who have set up a Thermo Fisher™ Connect account, 📶 (**Notification**) appears in the Genexus™ Software banner on the top of the screen upon login when updated software packages are available for download from the Connect cloud-based platform.

Updated software packages are available from the App Store at apps.thermofisher.com. Software packages can include assay definition files, for example. Administrator-level users can download and install software directly from the App Store if a Thermo Fisher™ Connect account has been configured. Alternatively, a USB drive can be used to install and update the software if the instrument is not connected to the internet.

Option	For more information, see this section
Download and install software directly	“Download and install software from the App Store (administrator)” on page 266
Download and install software with a USB drive	“Format a USB drive for a software update” on page 267 “Download and install software with a USB drive (administrator)” on page 267

Note: For first-time update of system software from the 5.100.2 version, contact a Field Service Engineer (FSE) to update the system software.

IMPORTANT! Before updating the software, ensure that the instrument is idle, no runs are in progress or are queued to run, and the screen on the instrument is the main menu screen.

Available versions of software update packages

When a software update package is available to install, you can view the available version of the package in the **Software Updates** table in ⚙️ (**Settings**) / **Software Updates**. The available version, for example 1.0.0, is listed for all assay definition files, plugins, configuration packages, and software updates that are available to install.

Assays that are available to install are listed in the **Category of Assay Definition Files** in the **Software Updates** table. The software files that contain the assay settings and parameters are packaged in a ZIP file called the assay definition file (ADF). You can install and use multiple versions of assays in the software. For more information about version numbers for assays, see “Assay details” on page 238.

Download and install software from the App Store (administrator)

In order to download and install software updates from the App Store on thermofisher.com/connect, you must first:

- Configure a Thermo Fisher Connect account. For instructions, see “Configure a Thermo Fisher™ Connect account with a QR code (manager/administrator)” on page 251.
- Ensure that the Genexus™ Integrated Sequencer has the latest software version, Genexus™ Software 6.6.
- Connect the Genexus™ Integrated Sequencer to the internet.

1. In the menu bar, click  **(Settings)** ▶ **Software Updates**, then click **Software Updates** in the upper right corner.

The **Software Updates** screen opens with **App Store** enabled.

2. (Optional) Click the desired package name to view the installation details and version information in the **View Package Details** pane.
3. Click **Download** in the **Actions** column in the row of the software package of interest. After the download is complete, the **Download** link changes to an **Install** link.
4. Click **Install**. When the confirmation window appears, click **Yes** to confirm that you want to install. If you are performing a system software module installation, a command window opens to show the progress of the installation.

Note: There may be a delay before the command window opens.

5. When installation of the software package is complete, click **Installed Software** to verify the installation.

Format a USB drive for a software update



Pre-requisites:


- An Ubuntu Linux desktop machine with disk utility software.
 - A USB drive with a capacity of 60 GB or more.
1. Insert the USB drive into the Ubuntu Linux machine.
 2. With Ubuntu running, open the disk utility by navigating to **System Administration** ▶ **Disk Utility**.
 3. Select the USB Flash Drive (SMI USB DISK) under **Peripheral Devices from Storage Devices** by clicking on it. Highlight the volumes (New Volume FAT >60 GB), click **Unmount Volume**, then click **Delete Partition**. Click **Delete** to confirm.
 4. Select the free USB volume, then click **Create Partition**.
The **Create partition on SMI USB DISK** window opens.
 5. Deselect the **Take ownership of file system** check box.
 6. Select **ext3** from the **Type** dropdown menu, then select the entire size of the partition. Name the partition *usbinstall*.
The partition name must be completely lowercase.
 7. Click **Mount**.
The mount point of the USB drive is `/media/usbinstall`.

Proceed to “Download and install software with a USB drive (administrator)”.

Download and install software with a USB drive (administrator)

In order to download and install software updates, you must first:

- Create or access your Thermo Fisher Connect account. For instructions, see “Configure a Thermo Fisher™ Connect account with a QR code (manager/administrator)” on page 251.
 - Format a USB drive for the software update. For more information, see “Format a USB drive for a software update” on page 267.
1. Sign in to the Thermo Fisher App Store on Connect at apps.thermofisher.com.
 2. On the left side of the App Store screen, click  (**AppConnect**).
 3. On the right side of the screen, under **Resource Libraries**, click **Ion Torrent™ Genexus™**.
 4. Click **Software Update**.
 5. In the Ion Torrent™ Genexus™ Software table, select the row that contains the software package for the version that you want to install.
 6. Click  (**Download**) to download the package.
 7. Copy the downloaded package (.zip) to the `/media/usbinstall` folder in the USB drive.

8. After completion, unmount the USB drive, then insert the drive into one of the USB ports at the back of the Genexus™ Integrated Sequencer.
9. In the menu bar of the Genexus™ Software, click  **(Settings)** ▶ **Software Updates**, then click **Software Updates** in the upper right corner.
10. Select **USB**.
11. Click **Install**. When the confirmation window appears, click **Yes** to confirm that you want to install.


When installation of the software package is complete, click **Installed Software** to verify the installation.

Manage backup settings and restore runs (administrator)

Administrator-level users can view and manage backup settings and restore runs in Genexus™ Software with the following actions:

- Monitor system storage space usage.
- Manage settings to backup and delete files and data.
- View an audit trail of any changes made.
- View and change email notification settings.
- Restore runs.

Disk space usage and backup settings (administrator)

An administrator-level user monitors disk space usage for the Genexus™ Integrated Sequencer and changes data backup settings in Genexus™ Software. System disk space usage information is available in the  **(Settings) / Backup & Restore** menu.

To maintain sufficient disk space, best practice is to implement a systematic plan to archive older run data, results files, variants information, and signed reports to an external file storage system. A system administrator or Field Service Engineer (FSE) is responsible for setting up a data backup strategy and disk cleanup settings. An administrator-level user can then configure the backup schedule. For more information, see “Change the backup settings (administrator)” on page 275.

The software ensures that disk space is available for runs. If the system has less than 1 terabyte (TB) of free disk space, no new runs can be started on the sequencer. In this case, an alert notifies the user that there is insufficient disk space when setting up a run on the sequencer. The run cannot proceed until data are backed up and deleted.

Contact a local system administrator to backup and delete data manually. An administrator-level user can also change the schedule to backup and delete data.

Results and reports that have been backed up can be restored in Genexus™ Software. For more information, see “Restore run results for a single run (administrator)” on page 280.

Specifications and disk space usage

The amount of disk space used for a run on the Genexus™ Integrated Sequencer depends on the nucleic acid sample type and the assay that is used for sequencing. The system can store 40 to 50 whole chip data runs. If the system has less than 1 terabyte (TB) of available storage space, no new runs can be started. For more information, see “Disk space usage and backup settings (administrator)” on page 268.

The following table lists chip specifications and data usage for a run with the Ion Torrent™ GX5™ chip.

Assay	Oncomine™ Precision Assay GX		Oncomine™ Myeloid Assay GX v2	Oncomine™ Comprehensive Assay v3 GX	Oncomine™ TCR Beta-LR Assay GX
	cfTNA	FFPE DNA+RNA	DNA+RNA	DNA+RNA	RNA
Number of reads per chip (4 lanes)	48–60 million	48–60 million	48–60 million	48–60 million	48–60 million
Number of reads per lane	12–15 million	12–15 million	12–15 million	12–15 million	12–15 million
Signal processing input size per chip ^[1]	~2 terabytes	~2 terabytes	~3.5 terabytes	~2 terabytes	~3.5 terabytes
Basecalling input size per chip ^[2]	200–300 gigabytes	200–300 gigabytes	400–600 gigabytes	200–300 gigabytes	400–600 gigabytes
Data size of intermediate and results files and logs ^[3]	120–180 gigabytes	120–180 gigabytes	160–240 gigabytes	120–180 gigabytes	160–240 gigabytes
Data size per lane of intermediate and results files and logs	30–45 gigabytes	30–45 gigabytes	40–60 gigabytes	30–45 gigabytes	40–60 gigabytes
Data size of backed up files and logs	30–45 gigabytes	30–45 gigabytes	40–60 gigabytes	30–45 gigabytes	40–60 gigabytes
Data size of results files per lane ^[4]	~10–20 gigabytes	~8–24 gigabytes	~32–48 gigabytes	~9–15 gigabytes	~16–48 gigabytes

(continued)

Assay	Oncomine™ Precision Assay GX		Oncomine™ Myeloid Assay GX v2	Oncomine™ Comprehensive Assay v3 GX	Oncomine™ TCR Beta-LR Assay GX
Recommended number of samples per lane	1 sample per lane	4 samples per lane	8 samples per lane	1.5 samples per lane	4 samples per lane
Data size of the results per sample	~10–20 gigabytes	~2–6 gigabytes	~4–6 gigabytes	~6–10 gigabytes	~2–6 gigabytes


^[1] The signal processing input on the sequencer is deleted after analysis is complete and successful.

^[2] Administrator-level users can schedule when basecalling input files are deleted. By default, these files are not retained. For more information, see “Set a schedule to permanently delete files (administrator)” on page 277.

^[3] Includes QC, plugins, logs, results, and other assay and variant database files.

^[4] For more information about results files, see “Results files” on page 169.

Configure backup settings (administrator)

An administrator-level user views and configures sequencer backup settings in the  **(Settings) / Backup & Restore** screen.

Update backup settings for various reasons, including:

- The backup location has limited free disk space remaining.
- The backup location is not accessible when the system tries to back up a run.

Consult a local Field Service Engineer to set up a backup system. For more information, see “Set up a backup server (administrator)” on page 274.

The following options are available only after a backup directory has been configured:

- View the backup disk space.
- Change the backup settings.

1. In the menu bar, click  **(Settings) ▶ Backup & Restore**

2. In the **Backup Settings** tab, edit the backup settings.

Option	Description
View the system disk space	View the chart that shows the percentage of system disk space that is used. View the total amount of disk space, the amount of free disk space, and the amount of used disk space, shown in terabytes.
View the backup disk space ^[1]	View the chart that shows the percentage of backup disk space that is used. View the total amount of backup disk space, the amount of free disk space, and the amount of used disk space, shown in terabytes.

(continued)

Option	Description
Change the backup directory	Change the backup directory. For more information, see “Add or change the backup directory (administrator)” on page 271.
Change the email notification settings	Select when administrator-level users receive email notifications. For more information, see “Change email notification settings (administrator)” on page 279.
Change the backup settings ^[1]	Backup .wells files and intermediate files. For more information, see “Change the backup settings (administrator)” on page 275.
Change the disk cleanup settings	Set the schedule to permanently delete analysis results files. Set the conditions to permanently delete other files. For more information, see “Set a schedule to permanently delete files (administrator)” on page 277.
Enable backup and cleanup	Click the Backup & Cleanup Enabled toggle switch to the enabled position to start the backup and cleanup process at the scheduled time.
Disable backup and cleanup	Click the Backup & Cleanup Enabled toggle switch to the disabled or off position for temporary instances, such as when the backup server is down. This setting overrides all other backup settings.
Start backup	Click Start Now to start: <ul style="list-style-type: none"> the backup of system data and analysis results to the selected backup directory. if enabled, the backup of sequencing output and intermediate files to the selected backup directory. data clean up based on cleanup settings. Backup can take a considerable amount of time when the backup is set up for first time on the system.


^[1] This option is only available when **Network** is the selected **Connection type**.

3. Click **Save**.

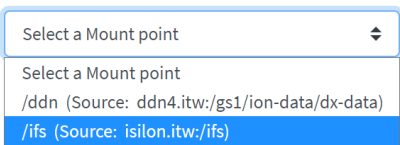
Add or change the backup directory (administrator)

Administrator-level users can use a Network File System (NFS) to set up an external storage device that is a backup server to store system data. When the current backup directory has limited free disk space remaining, change the backup directory used by the system.

IMPORTANT! We recommend NFS version 3 (NFSv3) or NFS version 4 (NFSv4) with Linux Operating System external storage devices for Genexus™ Software.

1. In the menu bar, click  (**Settings**) ▶ **Backup & Restore**.
2. In the **Backup & Restore** screen, select either **Network** or **Cloud** as the connection type.


3. Edit the backup settings.



Option	Description
Add a new mount point	<ol style="list-style-type: none"> In the Backup Disk Space section, click Add Mount. In the Add Mount dialog box: <ul style="list-style-type: none"> Enter the name of the mount directory. For example, ddn. <hr/> <p>Note: Do not include information from a hostname in the mount directory name. For example, if the host name is gnx001, do not use gnx001 in the mount directory name.</p> <hr/> <p>You can type a new name to create a new directory.</p> <ul style="list-style-type: none"> Enter the storage source. For example, ddn.itw:/gs1/ion-data/dx-data (Optional) Select Automount to allow the system to mount backup directories automatically when the system is restarted. <p>When the option is unselected, you must mount a directory after a restart.</p>
Change the existing backup directory	<p>In the Backup Disk Space section, select the directory that you want to use for backups from the Backup Directory dropdown menu.</p> <p>You must add a mount point before it can be selected in the dropdown.</p> <p>Backup directory: ?</p> 
Remove a backup directory that is not in use, for example a directory that is full or is no longer available	<ol style="list-style-type: none"> Click Remove Mount. In the Remove Mount dialog box, select the mounted drive you want to remove. Click Remove. <p>If you remove a directory that is in use for backup, backups cannot not be performed until you select a new backup directory.</p>

4. In the **Backup & Restore** screen, click **Save** to save the new selections.

Add or change cloud backup directory (administrator)

You can change the cloud backup directory that is used by the system. For example, you might change the directory if the current backup directory has limited storage space. You can also add a new directory, or remove a directory.

1. In the menu bar, click  **(Settings)** ▶ **Backup & Restore**.
 The **Backup & Restore** screen opens.
2. Under **Backup Disk Space** select **Cloud** as the connection type.
3. Edit the backup settings.

Option	Description
Add a new mount point	<ol style="list-style-type: none"> 1. In the Backup Disk Space section, click Add Mount. 2. In the Add Mount dialog box: <ul style="list-style-type: none"> • Enter the name of the Mount Point directory. For example, <i>ddn</i>. You can type a new name to create a new directory. • Enter the Cloud Provider. For example, <i>AWS-S3</i>. 3. Enter the Configuration information. <ul style="list-style-type: none"> • Nick Name—A name to uniquely identify the directory and mount point on the directory. Do not use spaces or special characters in the name. Note: The following information must be entered correctly to ensure that the mount works correctly. Information should be provided by an administrator for the network or Amazon™ Web Services (AWS). • Bucket Name—Name of the AWS S3 bucket. • Access Key Id—A key identifier that, similar to a username, provides programmatic and CLI access to AWS resource, such as the S3 bucket • Secret Access Key—An access key that, similar to a password, provides programmatic and CLI access to AWS resource, such as the S3 bucket. • Region—AWS region in which for the S3 bucket resides. For example, <i>us-east-1</i>, <i>us-west-1</i>. 4. (Optional) Select Automount to allow the system to mount backup directories automatically when the system is rebooted. When the option is unselected, you must mount a directory after a reboot. 5. Click Save.
Change the existing backup directory	<p>In the Backup Disk Space section, select the directory that will be used for backups from the Backup directory drop-down list. You must add a mount point before for the backup directory can be selected.</p> <p>Backup directory: </p> <div data-bbox="537 1728 935 1864" style="border: 1px solid #ccc; padding: 5px;"> <p>Select a Mount point </p> <p>Select a Mount point</p> <p>/ddn (Source: ddn4.itw:/gs1/ion-data/dx-data)</p> <p>/ifs (Source: isilon.itw:/ifs)</p> </div>

(continued)

Option	Description
Remove a backup directory that is not in use, for example a directory that is full or is no longer available	<ol style="list-style-type: none"> 1. Click Remove Mount. 2. In the Remove Mount dialog box, select the mounted drive you want to remove. 3. Click Remove. 4. Click Save. <p>If you remove a directory that is in use for backup, backups cannot not be performed until you select a new backup directory.</p>

4. In the **Backup & Restore** screen, click **Save** to save the new selections.

The changes that you made are shown in the **Backup Disk Space** section of the **Backup Settings** screen.

Set up a backup server (administrator)

Administrator-level users can set up an external storage device to be a backup server to store system data with this procedure. Alternatively, administrator-level users can directly set up a backup server in Genexus™ Software. For more information, see “Add or change the backup directory (administrator)” on page 271.

IMPORTANT! We recommend NFS version 3 (NFSv3) or NFS version 4 (NFSv4) with Linux Operating System external storage devices for Genexus™ Software.

1. Mount your NFS to the sequencer.

The following example is for users who have an existing—read and write access—Network File System (NFS). Mount your NFS to the sequencer using the *fstab* file so that the mount point is persistent (NFS mounts automatically after rebooting).

Example:

For NFS share: `cqtsop4:/md1200`

Mount point: `nfs1`

2. Create a mount point to mount the external device on nfs share.

- a. Create the directory `/nfs1` using the following command:

```
sudo mkdir /nfs1
```

- b. Append the mount point entry in `/etc/fstab`.

Command example:

```
sudo vi /etc/fstab
cqtsop4:/md1200 /nfs1 nfs defaults 0 0
```

- c. Save, then exit by pressing **Esc** on your keyboard, then type `:wq`

3. Create a permanent or temporary mount on nfs share:

- Permanent mount command: `sudo mount -a`
- Temporary mount command (example): `sudo mount cqtsop4:/md1200 /nfs1`

4. Verify that the NFS share is mounted using the `df -kh` command.

```
ionadmin@eternity1:~$ df -kh
Filesystem Size Used Avail Use% Mounted on
/dev/sda9 253G 5.9G 235G 3% /
none 4.0K 0 4.0K 0% /sys/fs/cgroup
udev 63G 4.0K 63G 1% /dev
tmpfs 13G 1.3M 13G 1% /run
none 5.0M 0 5.0M 0% /run/lock
none 63G 0 63G 0% /run/shm
none 100M 0 100M 0% /run/user
/dev/sda5 19G 2.9G 15G 17% /var
/dev/sda1 938M 37M 854M 5% /boot
/dev/sda6 9.1G 1.7G 7.0G 20% /home
/dev/mapper/raid-serverdata 8.4T 4.3T 4.1T 52% /serverdata
/dev/mapper/raid-servershare 1.0T 9.0G 1015G 1% /servershare
/dev/mapper/raid-scratch 1.0T 838M 1023G 1% /scratch
/dev/sda7 92G 61M 87G 1% /tmp
pluto:/md1200 37T 1.1T 36T 3% /nfs2
penguin-ts1:/local 11T 9.3T 978G 91% /nfs
ddn4.itw:/gs1/ion-data/dx-data 1.1P 1001T 115T 90% /ddn
cqtsop4:/md1200 37T 37T 77G 100% /nfs1
```

The mounted directory is listed in the **Backup Directory** dropdown list in the **Backup & Restore** screen.

For more information, see help.ubuntu.com/lts/serverguide/network-file-system-configuration.

Change the backup settings (administrator)


A backup directory must be set up before an administrator-level user can change the backup settings. For more information, see “Disk space usage and backup settings (administrator)” on page 268.

The following types of data are backed up daily at midnight and cannot be changed:

- **System Data** – Includes the system database, reference files, assay definition files, annotation content, and custom and system-installed panel files, hotspot files, fusion reference files, and CNV baseline files.
- **Analysis Results** – The final results generated after sequencing and analysis. Analysis results include run details, all BAM files, filtered results, graphs, run reports, metrics, plugin results, annotated variant data, and VCF files.

IMPORTANT! The first time you back up the system, it takes a considerable amount of time. The amount of time required for system backup is dependent on the number of runs that require backup and the amount of disk space used. For more information, see “Specifications and disk space usage” on page 269.

A system restart also takes time. When the system restarts, it resumes the backup where it left off.


1. In the menu bar, click  **(Settings)** ▶ **Backup & Restore**.
2. In the **Backup & Restore** screen, in the **Backup Settings** tab, edit the backup settings.

Option	Procedure
Back up .wells files	Select the Sequencing Output checkbox. Sequencing output backup takes time.
Back up intermediate files, such as intermediate BAM files and log files	Select the Intermediate Files checkbox.
Change the frequency or timing of the .wells and intermediate files backup	Select Daily or Weekly , then select the time and day, if applicable, for the backup. Intermediate file backup takes time. Consider scheduling the backup of intermediate file on a weekend or at a time when the system is not in use.

3. Click **Save**.



View and export the audit trail for backup settings (administrator)

An administrator-level user can view the audit trail for backup settings, disk cleanup settings, and email notification settings in Genexus™ Software.

1. In the menu bar, click  **(Settings)** ▶ **Backup & Restore**
2. In the **Backup & Restore** screen, view the audit trail.

Option	Procedure
View the audit trail for the backup settings	In the Backup Settings section, click audit to review changes made to the backup settings.
View the audit trail for email notification settings	In the Email Notification Settings section, click audit to review changes made to the email notification settings.
View the audit trail for disk cleanup settings	In the Disk Cleanup Settings section, click audit to review changes made to the disk cleanup settings.

3. In the **Audit Trail - Archive Settings** screen:

Option	Procedure
Export the record to a PDF file	Click  Download .
Return to the Backup Settings tab.	Click  Back .

Set a schedule to permanently delete files (administrator)

Results files and `.wells` files, and intermediate files that have been backed up can be scheduled to be deleted by an administrator-level user in Genexus™ Software.

Intermediate and `.wells` files include intermediate BAM files, log files, and Sequencing output files. These files are not backed up by default. When you set a schedule to delete these files, by default, the files are deleted when they are not backed up and the system disk space falls below 4 terabytes (TB). You can change the system disk space threshold to delete these files.


Run results that are retained are never deleted in the software, even when you set a schedule to permanently delete run results in the software. For more information, see “Retain run results” on page 112.

IMPORTANT!

- By default, when the Disk Cleanup Settings are enabled, the software is set to delete results after two years from the time the run was last updated and backed up. You can change this setting.
- Intermediate and `.wells` files are not backed up unless an administrator-level user sets a schedule to back up these files. By default, these files are auto-deleted when the free disk space threshold is met, even when these files are not backed up. If you select to keep the default auto-delete setting for intermediate and `.wells` files and the files are deleted, the impact is minimal.

You cannot perform the following actions when these files are deleted.

- Reanalyze a run from basecalling.
 - Upload to Ion Reporter™ Software.
 - Run the CustomerSupportArchive plugin.
-

1. In the menu bar, click  **(Settings)** ▶ **Backup & Restore**
2. In the **Backup & Restore** screen, in the **Disk Cleanup Settings** section:

Option	Procedure
Do not auto-delete analysis results at any time.	In Delete Analysis Results: , click No .
Change the schedule to permanently delete analysis results. ^[1]	<ol style="list-style-type: none"> 1. In Delete Analysis Results:, click Yes. 2. In months after completion, set the schedule to delete the files: <ul style="list-style-type: none"> • 3 months • 6 months • 9 months • 12 months • 24 months – default setting • 36 months
Do not auto-delete .wells files and intermediate files at any time.	In Delete additional files: , click No .
Change the schedule to permanently delete .wells files and intermediate files. ^[2]	<p>In Delete additional files:, click Yes, then select the threshold of free disk space on the instrument, from 1 to 10 TB, that triggers the system to delete these files.</p> <p>The system auto-deletes .wells and intermediate files when the disk space falls below the selected threshold.</p>

^[1] Analysis results are only deleted if the results have been backed up.

^[2] If you have enabled the backup of intermediate files and .wells files, these additional files must be backed up before they can be deleted.

3. Click **Save**.

Data output directory

The data output directory is the primary location on the hard drive where data generated by the Genexus™ Integrated Sequencer is stored.

The directory location is locked and cannot be changed. Users should configure their laboratory information management systems (LIMS) to access this folder location to receive sequencing output files.

After a sequencing run and data analysis are complete, the software creates a separate results folder in the data output directory for each sample, and uses the following naming conventions:

```
AssayDev_<Sample Name>_<Assay Name>_<RunPlanShortCode>_<RunPlanShortCode>
```


Email notifications

Administrator-level users receive email notifications when there is a restore run failure. Administrator-level users can also receive email notifications when other conditions are met, such as when the system has limited free disk space remaining. To review or update these settings, see “Change email notification settings (administrator)” on page 279.

Before administrator-level users can receive email notifications, an administrator or manager must first set up notifications for valid Genexus™ Software user accounts. For more information, see “Configure email notifications (manager/administrator)” on page 263.

Change email notification settings (administrator)

Administrator-level users can select when to receive email notifications, such as when the system has limited disk space remaining.

1. In the menu bar, click  **(Settings)** ▶ **Backup & Restore**
2. In the **Backup & Restore** screen, in the **Email Notifications Settings** section, select the checkbox next to one, some, or all of the following options so that administrator-level users receive an email message when the following conditions occur. Or, deselect the checkbox next to an option so that administrator-level users do not receive email messages for that option.
 - Backup fails or has insufficient storage space.
 - Backup is successfully completed.
 - Genexus™ Integrated Sequencer has insufficient system disk space. Select the amount of sequencer disk space that must be available in the dropdown list.
 - Backup fails or has insufficient disk space. Select the amount of backup disk space that must be available in the dropdown list.
3. Click **Save**.

Changes to the email notifications take effect immediately.

Restore run results (administrator)

An administrator-level user can configure Genexus™ Software to automatically back up older run data, results files, and signed reports to external data storage or a cloud storage space and set a schedule to automatically delete backed up runs after a specified amount of time. For more information, see “Disk space usage and backup settings (administrator)” on page 268, “Add or change the backup directory (administrator)” on page 271, and “Set a schedule to permanently delete files (administrator)” on page 277. Runs, for which analysis results have been backed up and deleted in the software, are listed in the **Run Results** screen and are denoted with the archived symbol (🗄️) after the run name.

An administrator-level user can later restore the run results, files, and reports in the software.

There are two ways to restore run results in Genexus™ Software:

- For information about how to restore run results for a single run, see “Restore run results for a single run (administrator)” on page 280.
- For information about how to restore or import multiple run results, see “Restore or import multiple archived run results (administrator)” on page 280.

Restore run results for a single run (administrator)

An administrator-level user can restore run results, files, and reports that have been archived in Genexus™ Software.

Archived runs that can be restored are listed in the **Results / Run Results** screen with the **Restore** link active when you place the pointer over the row of run. Runs, for which analysis results have been backed up and deleted in the software, are listed in the **Run Results** screen and are denoted with the archived symbol (☐) after the run name.

1. In the menu bar, click **Results ▶ Run Results**.
2. In the **Run Results** screen, click the run name of interest.
3. In **⋮ (More Options)**, click **Restore**.
4. Click **OK** in the confirmation dialog box.
Click the run name in the **Run Results** screen to view run results.

Restore or import multiple archived run results (administrator)

An administrator-level user can:

- Restore run results, files, and reports that have been archived.
- Import run results, files and reports from external data storage or another Genexus™ Integrated Sequencer.

Results that have been backed up, then deleted in the software, are listed in the **Results / Run Results** screen with ☐ (archived) after the run name.


Before you can import an run from external data storage or another Genexus™ Integrated Sequencer:

- The run data must be archived to external data storage, and the same external data storage must be connected to the Genexus™ Integrated Sequencer to which you will import the run.
- The samples for the run or runs that you want to import must have been created in the software with identical names. For more information about how to import a sample from another system, see “Import samples” on page 34.
- The assay for the run or runs that you want to import must exist in the software. Ensure that the name of the assay, the barcode kit, and the extraction method type for the source system and the target system are identical. Other assay parameters, such as hotspots, the filter chain, gene list, and report template do not need to match and are not imported to the target system. For information about how to import an assay, see “Import an assay (manager/administrator)” on page 240.
- You can search the list of runs that can be restored by the run name or a sample tag name. However, if you restore a run that has an assigned sample tag name that has been deleted in the software, the tag is not shown in the software when you restore the run. In this case, you can reassign the tag if needed. For more information, see “Sample tags” on page 46.

Note: Do not attempt to import verification runs because they are specific to a single system.

Sample to Result details for the Genexus™ Purification Instrument are not displayed in the target system (the system results are imported to) if the target system does not have a connected purification instrument.

After a run is imported, you can access **Run Results** and **Sample Results** in the target system. Genexus™ Integrated Sequencer details are also listed with the instruments in the target system.

1. In the menu bar, click  **(Settings) ▶ Backup & Restore**.
2. In the **Backup & Restore** screen, click the **Restore Runs** tab:

Option	Description
Restore archived run results	<ol style="list-style-type: none"> 1. In the type of runs dropdown menu, keep the selection of Archived runs. 2. In the dropdown menu, select the mounted drive that contains the runs you want to restore, then click Browse. 3. In the Select backup folder to list runs dialog box, navigate to the folder with the name of the system that you use, then select the subfolder that contains the runs you want to restore and click Select. 4. In the Restore Runs tab, select the checkbox in the row for each run you want to restore, then click Restore Runs. Use the Search By dropdown list and the search field to search for samples by a run name or sample tags. The process to restore runs takes time. When the run has been restored, you can view the run results for the run in the software. 5. <i>(Optional)</i> When the run has been restored, click Check Log to view if the process was successful or if there are problems.
Import run results from another system	<ol style="list-style-type: none"> 1. In the type of runs dropdown menu, keep the selection of Archived runs. 2. In the dropdown menu, select the mounted drive that contains the runs you want to restore, then click Browse. 3. In the Select backup folder to list runs dialog box, navigate to the folder for a different system, then click Select. 4. In the Restore Runs tab, select the checkbox in the row for each run you want to import, then click Restore Runs. Use the Search By dropdown list and the search field to search for samples by a run name or sample tags. The process to restore runs takes time. When the run has been restored, you can view the run results for the run in the software. 5. <i>(Optional)</i> When the run has been imported, click Check Log to view if the process was successful or if there are any problems, such as if a sample has not been created in the software.
Restore runs if there is a system crash ^[1]	<ol style="list-style-type: none"> 1. In the type of runs dropdown menu, select All runs. 2. In the dropdown menu, select the mounted drive that contains the runs you want to restore, then click Browse. 3. In the Select backup folder to list runs dialog box, navigate to the folder that contains the runs you want to restore, then click Select. 4. In the Restore Runs tab, select the checkbox in the row for each run you want to restore, then click Restore Runs.

Option	Description
	<p>Use the Search By dropdown list and the search field to search for samples by a run name or sample tags.</p> <p>The process to restore runs takes time. When the run has been restored, you can view the run results for the run in the software.</p> <p>5. <i>(Optional)</i> When the run has been restored, click Check Log to view if the process was successful or if there are problems.</p>

^[1] Only use this procedure if there is a system crash and you have worked with a Field Service Engineer (FSE) to restore the data onto a new system. When you select **All runs** in any other instance, the software overwrites existing data with identical data.


Log files (administrator)

Administrator-level users use the **Logs** screen to view log files generated by the software and connected purification and sequencing instruments. Viewing log files is useful for troubleshooting activities.

Administrator-level users can also use the **Logs** screen to manage the storage of log data by setting the retention period for the audit trail function and enabling auto-deletion rules for logs.

View log files (administrator)

Administrator-level users can view software and instrument log files in Genexus™ Software.

1. Click  **(Settings)** ▶ **Logs**.
2. In the **Logs** screen, select an option in the software or connected instrument dropdown menu.

Option	Description
View software log files	Click Ion Torrent Genexus Software
View log files for a connected purification or sequencing instrument	Click Instrument

3. Click the log file name of interest.



File name	Description
TorrentSuite_<date>.log	A log file with information related to some actions on the server, including creation of , user sign-ins and sign-outs, failed attempts to sign-in or sign-out.
InstrumentGX5_date.log	A log of activity on the sequencer instrument.
InstrumentPurif_date.log	A log of activity on the purification instrument.

The log file opens in a new tab in the browser.

Manage log settings (administrator)




An administrator-level user manages the length of time that log files are kept and configures the software to allow log files to be automatically deleted.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

1. Click  **(Settings)** ▶ **Logs**.
2. In the **Logs** screen, click  **Manage Logs**.
3. In the **Manage Logs** dialog box, in **Retention Period**, select the number of months that logs are to be retained on the sequencer.
4. Select **Enable Auto Deletion** to delete log files automatically after the designated retention period.
5. Click **Save**.

View the log settings history

Administrator-level users can view the history of changes made to the settings for the retention of log files in Genexus™ Software.

1. Click  **(Settings)** ▶ **Logs**.
2. In the **Logs** screen, click  **Audit Trail**.
The **Audit Trail - Log Settings** screen opens.
3. Click  **Back** to return to the list of log files.

Panel files (manager/administrator)

Manager- and administrator-level users use the **Assays / Manage Panels** screen to manage panel files and reference files.

Note: Manager- and administrator-level users can create and obsolete panel and reference files using **+ Add New** and **Obsolete**, except for those defined by system-installed or locked assays.

- Panel files (BED format)
- Hotspots files (BED format)
- Fusion reference sequences (FASTA format)
- Reference genome files (FASTA format)
- DNA barcode sequence files
- Control fragments

Manage panels (manager/administrator)

Manager- and administrator-level users can add new panels, export existing panel files, and make panels that have been added in the software unavailable in Genexus™ Software.

Operators can view the panels that are available in the software.

View panel details

You can view details about system-installed and panels that have been added in Genexus™ Software.

1. In the menu bar, click **Assays ▶ Manage Panels**.
The **Panels** tab in the **Manage Panels** screen opens.
2. View the panel details.

Add new panel files (manager/administrator)

Manager- and administrator-level users can add new panel files to Genexus™ Software.

You can upload BED panel files or you can upload a ZIP file that contains the panel file and related files.

- For DNA panels, you can upload a BED panel file or you can upload a ZIP file that contains the panel file and the hotspot file.
 - For DNA and Fusion panels, you can upload a target region BED file and a Fusion panel file individually and select a fusion reference file, or you can upload a ZIP file that contains a target region file, a hotspot file, and a fusion reference file.
 - For Fusion panels, you can upload a Fusion panel file individually and select a fusion reference file, or you can upload a ZIP file that contains the target region file and the fusion reference file.
 - For RNA panels, BED is the only supported file format.
1. In the menu bar, click **Assays ▶ Manage Panels**.
The **Panels** tab in the **Manage Panels** screen opens.
 2. Click **+ Add New**.
 3. In the **Add Panel** dialog box, enter the required information.

Field	Description
Name	Enter a name for the new panel.
Panel Type	Select the panel type, such as DNA and Fusion Panel or DNA Panel .
Reference	Select a reference genome.
File Type	Select the file format when multiple file types are supported.
Target Regions	Navigate to the target regions file.
Fusion Reference	For Fusions panels and DNA and Fusions panels, select the fusion reference.
Fusion Panel	Navigate to the fusion panel file.

4. Click **Upload**.

Note: Large files take time to upload. You can see the file listed only when the upload is complete.

The new panel is listed in the **Panels** tab and is available to add to a custom assay. For more information, see “Assay creation overview” on page 193. Hotspot uploads are listed in the **Hotspot** tab and are available to add to a custom assay. For ZIP file uploads, the name of the hotspot is the

name of the panel followed by **_Hotspot**. Fusion references are listed in the **Panels** tab with more details available in the **Fusion References** tab.


Import panel files from Ion AmpliSeq™ Designer (manager/administrator)

Manager- and administrator-level users can import custom or fixed panel files into Genexus™ Software from Ion AmpliSeq™ Designer.

Before you can import custom panel files, ensure that the following prerequisites are complete.

- You know the username and access code for an [AmpliSeq.com](https://www.ampliseq.com) account.
- You have ordered a custom panel at [AmpliSeq.com](https://www.ampliseq.com).

For more information, see *Ion AmpliSeq™ Designer Help* (Pub. No. MAN0018937).

1. In the menu bar, click **Assays ▶ Manage Panels**.
 The **Panels** tab in the **Manage Panels** screen opens.
2. Click  **Import from AmpliSeq**.
3. In the **Import from AmpliSeq** dialog box, enter the information for the panel that you want to import.

For Custom Panels

Item	Description
Panel type	Select the panel type, such as DNA and Fusion Panel or DNA Panel .
Reference	Select a reference genome.
Library Chemistry	Select the type of library chemistry for the panel, such as Ion AmpliSeq™ HD.
AmpliSeq Panels	Select Custom Panel .
AmpliSeq Username	Enter the username that you used when you created an account for AmpliSeq.com .
AmpliSeq Access code	Enter the access code the you generated at AmpliSeq.com .
List My AmpliSeq Panels	Select the panel you want to import from the dropdown list. Only panels that have a status of ordered in Ion AmpliSeq™ Designer are listed. Draft and results ready panels are not listed.

For Fixed Panels

Item	Description
Panel type	Select the panel type, such as DNA and Fusion Panel or DNA Panel .
Reference	Select a reference genome.
Library Chemistry	Select the type of library chemistry for the panel, such as Ion AmpliSeq™ HD.
AmpliSeq Panels	Select Fixed Panel .
Fixed Panel	Select the panel you want to import from the dropdown list.

4. Click **Import**.

The imported panel appears in the list of panels in the **Manage Panels** screen and can be added to an assay. For more information, see “Assay creation overview” on page 193.

Any hotspots and fusions references associated with the imported panel are also listed in the respective **Hotspots** and **Fusion References** tabs after panel file upload.

Export panel files (manager/administrator)

Manager- and administrator-level users can export existing panel files from Genexus™ Software.

1. In the menu bar, click **Assays ▶ Manage Panels**.
The **Panels** tab in the **Manage Panels** screen opens.
2. Place the pointer over the row of the panel that you want to export, then click **Export** to download the panel BED file.

Obsolete panels (manager/administrator)

Manager- and administrator-level users can make panels unavailable that have been added or imported in Genexus™ Software. You cannot designate as obsolete system-installed panels.

1. In the menu bar, click **Assays ▶ Manage Panels**.
The **Panels** tab in the **Manage Panels** screen opens.
2. Place the pointer over the row of the panel that you want to remove from the software, then click **Obsolete**.

The panel is listed in the **Panels** table with a **Status** of **Obsolete**. Panels that have been made obsolete cannot be added to a new assay. Manager- and administrator-level users can reactivate panels that have been made obsolete. For more information, see “Reactivate panels (manager/administrator)” on page 287.

Reactivate panels (manager/administrator)

Manager- and administrator-level users can reactivate panels that have been made obsolete in Genexus™ Software.

1. In the menu bar, click **Assays ▶ Manage Panels**.
The **Panels** tab in the **Manage Panels** screen opens.
2. Place the pointer over the row of the panel that you want to reactivate, then click **Reactivate**.

The panel is listed in the **Panels** table with a **Status** of **Locked** and is available to add to a new assay.

Manage hotspot files (manager/administrator)

Manager- and administrator-level users add, view, and manage the hotspot BED files that are available in Genexus™ Software.

Operators can view the hotspot files that are available in the software.

View hotspot file details

You can view details about system-installed hotspot files and hotspot files that have been added in Genexus™ Software.

1. In the menu bar, click **Assays ▶ Manage Panels**.
The **Panels** tab in the **Manage Panels** screen opens.
2. Click the **Hotspots** tab.
3. In the **Hotspots** table, view the details for the hotspot files.

Add new hotspot files (manager/administrator)

Manager- and administrator-level users can add new hotspot files in Genexus™ Software.

Hotspot files can also be added when you add or import a panel file. For more information, see “Add new panel files (manager/administrator)” on page 284 and “Import panel files from Ion AmpliSeq™ Designer (manager/administrator)” on page 285.

1. In the menu bar, click **Assays ▶ Manage Panels**.
The **Panels** tab in the **Manage Panels** screen opens.
2. Click the **Hotspots** tab.
3. Click **+ Add New**.
4. In the **Add Hotspot File** dialog box, enter a name, select a panel, then select the BED file that you want to upload.
5. Click **Upload**.
Large files take time to upload and may not appear immediately.

The hotspot file is listed in the **Hotspot** table and is available to select when you create or copy and edit some custom assays.

Export hotspot files (manager/administrator)

Manager- and administrator-level users can export hotspot files from Genexus™ Software.

1. In the menu bar, click **Assays ▶ Manage Panels**.
The **Panels** tab in the **Manage Panels** screen opens.
2. Click the **Hotspots** tab.
3. Place the pointer over the row of the hotspot file that you want to export, then click **Export** to download the hotspot BED file.

Obsolete hotspot files (manager/administrator)

Manager- and administrator-level users can make hotspot files that have been added to the software unavailable in Genexus™ Software. You cannot designate as obsolete system-installed hotspot files.

1. In the menu bar, click **Assays ▶ Manage Panels**.
The **Panels** tab in the **Manage Panels** screen opens.
2. Click the **Hotspot** tab.
3. Place the pointer over the row of the hotspot file that you want to remove from the software, then click **Obsolete**.

The hotspot file is listed in the **Hotspot** table with a **Status** of **Obsolete**. Hotspot files that have been made obsolete cannot be added to a new assay. Manager- and administrator-level users can reactivate hotspot files that have been made obsolete. For more information, see “Reactivate hotspot files (manager/administrator)” on page 288.

Reactivate hotspot files (manager/administrator)

Manager- and administrator-level users can reactivate hotspot files that have been made obsolete in Genexus™ Software.

1. In the menu bar, click **Assays ▶ Manage Panels**.
The **Panels** tab in the **Manage Panels** screen opens.
2. Click the **Hotspots** tab.
3. Place the pointer over the row of the hotspot file that you want to reactivate, then click **Reactivate**.

The hotspot file is listed in the **Hotspots** table with a **Status** of **Locked** and is available to add to a new assay. Hotspot files are only available to select for some assays.

Manage fusion reference files (manager/administrator)

Manager- and administrator-level users add, view, and manage the fusion reference files that are available in Genexus™ Software.

Operators can view the fusion reference files that are available in the software.

View fusion reference file details

You can view details for system-installed fusion reference files and fusion reference files that have been added in Genexus™ Software.

1. In the menu bar, click **Assays ▶ Manage Panels**.
The **Panels** tab in the **Manage Panels** screen opens.
2. Click the **Fusion References** tab.
3. In the **Fusion References** table, view the details for the fusion reference files.

Add fusion reference files (manager/administrator)

Manager- and administrator-level users can add new fusion reference files in Genexus™ Software.

Fusion reference files can also be added when you add or import a panel file. For more information, see “Add new panel files (manager/administrator)” on page 284 and “Import panel files from Ion AmpliSeq™ Designer (manager/administrator)” on page 285.

1. In the menu bar, click **Assays ▶ Manage Panels**.
2. In the **Manage Panels** screen, select the **Fusion References** tab.
The **Panels** tab in the **Manage Panels** screen opens.
3. Click the **Fusion References** tab.
4. Click **+ Add New**.
5. In the **Add Fusion Reference** dialog box, enter a name, select a reference genome, then select the FASTA file that you want to upload.
Large files take time to upload and may not appear immediately.
6. Click **Upload**.

The fusion reference file is listed in the **Fusion References** table and is available to select when you create or copy and edit some custom assays, or when you add a new panel.

Export fusion reference files (manager/administrator)

Manager- and administrator-level users can export fusion reference files from Genexus™ Software.

1. In the menu bar, click **Assays ▶ Manage Panels**.
The **Panels** tab in the **Manage Panels** screen opens.
2. Click the **Fusion References** tab.
3. Place the pointer over the row of the fusion reference file that you want to export, then click **Export** to download the fusion reference FASTA file.

Obsolete fusion reference files (manager/administrator)

Manager- and administrator-level users can make fusion reference files that have been added to the software unavailable in Genexus™ Software. You cannot designate as obsolete system-installed fusion reference files.

1. In the menu bar, click **Assays ▶ Manage Panels**.
The **Panels** tab in the **Manage Panels** screen opens.
2. Click the **Fusion References** tab.
3. Place the pointer over the row of the fusion reference file that you want to remove from the software, then click **Obsolete**.

The fusion reference file is listed in the **Fusion References** table with a **Status** of **Obsolete**. Fusion reference files that have been made obsolete cannot be added to a new assay. Manager- and administrator-level users can reactivate fusion reference files that have been made obsolete. For more information, see “Reactivate fusion reference files (manager/administrator)” on page 290.

Reactivate fusion reference files (manager/administrator)

Manager- and administrator-level users can reactivate fusion reference files that have been made obsolete in Genexus™ Software.

1. In the menu bar, click **Assays ▶ Manage Panels**.
The **Panels** tab in the **Manage Panels** screen opens.
2. Click the **Fusion References** tab.
3. Place the pointer over the row of the fusion reference file that you want to reactivate, then click **Reactivate**.

The fusion reference file is listed in the **Fusion References** table with a **Status** of **Locked** and is available to add to a new assay. Fusion reference files are only available to select for some assays.

Manage reference genome files (manager/administrator)

Manager- and administrator-level users add new DNA reference genome files so that they are available in Genexus™ Software. New reference genomes are uploaded in a FASTA file.

Operators can view the reference genome files that are available in the software.

For information about the system-installed reference genome files, see “System-installed reference genome files” on page 367.

View reference genome file details

You can view details for system-installed reference genome files and reference genome files that have been added in Genexus™ Software.

1. In the menu bar, click **Assays ▶ Manage Panels**.
The **Panels** tab in the **Manage Panels** screen opens.
2. Click the **References** tab.
3. In the **References** table, view the details for the reference genome files.

Add reference genome files (manager/administrator)

Manager- and administrator-level users can add new reference genome files in Genexus™ Software.

1. In the menu bar, click **Assays ▶ Manage Panels**.
2. In the **Manage Panels** screen, click the **References** tab, then click **+ Add New**.
3. In the **Add Reference Genome** dialog box, enter the required and optional information, then select the FASTA file to upload the reference sequence information.
Large files take time to upload and may not appear immediately.
4. Click **Upload** when you have finished.

The reference genome file is listed in the **References** table and is available to select when you add or import a panel or a fusion reference file. For more information, see “Add new panel files (manager/administrator)” on page 284, “Import panel files from Ion AmpliSeq™ Designer (manager/administrator)” on page 285, and “Add fusion reference files (manager/administrator)” on page 289.

Obsolete reference genomes (manager/administrator)

Manager- and administrator-level users can make reference genomes that have been added to the software unavailable in Genexus™ Software. You cannot designate as obsolete system-installed reference genomes.

1. In the menu bar, click **Assays ▶ Manage Panels**.
The **Panels** tab in the **Manage Panels** screen opens.
2. Click the **References** tab.
3. Place the pointer over the row of the reference genome that you want to remove from the software, then click **Obsolete**.

The reference genome is listed in the **References** table with a **Status** of **Obsolete**. Reference genomes that have been made obsolete cannot be added to a panel or a new fusion reference file. Manager- and administrator-level users can reactivate reference genomes that have been made obsolete. For more information, see “Reactivate reference genomes (manager/administrator)” on page 292.

Reactivate reference genomes (manager/administrator)

Manager- and administrator-level users can reactivate reference genomes that have been made obsolete in Genexus™ Software.

1. In the menu bar, click **Assays ▶ Manage Panels**.
The **Panels** tab in the **Manage Panels** screen opens.
2. Click the **References** tab.
3. Place the pointer over the row of the reference genome that you want to reactivate, then click **Reactivate**.

The reference genome is listed in the **References** table with a **Status** of **Locked** and is available to add to a new panel or fusion reference.

Manage barcode sets (manager/administrator)

Manager- and administrator-level users add new barcode sets to Genexus™ Software, and view the barcode IDs and sequences for each barcode set that is available in the software.

For information about system-installed barcode sets, see “System-installed barcode sets” on page 350.

View barcode set details (manager/administrator)

You can view details for system-installed barcode sets and barcode sets that have been added in Genexus™ Software.

1. In the menu bar, click **Assays ▶ Manage Panels**.
The **Panels** tab in the **Manage Panels** screen opens.
2. Click the **Barcode Sets** tab.

3. In the **Barcode sets** table, view the list of barcode sets.
4. Click the name of a barcode set in the list to view the barcode IDs, sequences, and other details for the barcode set.
5. Click **X (Remove)** to return to the complete list of barcode sets.

Add new barcode sets (manager/administrator)

Manager- and administrator-level users add new barcode sets to Genexus™ Software. You can upload XLS or XLSX barcode set files. An example barcode set file is available in the software.

1. In the menu bar, click **Assays ▶ Manage Panels**.
The **Panels** tab in the **Manage Panels** screen opens.
2. Click the **Barcode Sets** tab.
3. Click **+ Add New**, enter the required information in the **Add Barcode Set** dialog box, and browse to and select the barcode set file.
Click the **Click here** link to download an example barcode file.
4. Click **Upload**.

The barcode set is listed in the **Barcode Sets** table and is available to select in the reagent step when you create an assay. For more information, see “Reagent step assay options” on page 199.

Obsolete barcode sets (manager/administrator)

Manager- and administrator-level users can make barcode sets that have been added to the software unavailable in Genexus™ Software. You cannot designate as obsolete system-installed barcode sets.

1. In the menu bar, click **Assays ▶ Manage Panels**.
The **Panels** tab in the **Manage Panels** screen opens.
2. Click the **Barcode Sets** tab.
3. Place the pointer over the row of the barcode set that you want to remove from the software, then click **Obsolete**.
4. In the confirmation dialog box that appears, click **Yes** to confirm that you want to obsolete the barcode set.
You cannot reactivate barcode sets that have been made obsolete.


Control fragments (manager/administrator)

Control fragment sequences are system-installed in Genexus™ Software and are included in reagents. These internal controls are predefined and cannot be modified by users.

Manager- and administrator-level users can view control fragment information in the **Assays / Manage Panels** screen, in the **Control Fragments** tab.

Column	Description
Name	The control fragment name.
Sequence	Single-letter nucleotide sequence of the control fragment.
Control	The process in which the control fragment is used.
Status	Indicates whether the control fragment information is locked and cannot be modified by the user.

System services (manager/administrator)


Manager- and administrator-level users can view information about system services using the  **(Settings) / Services** screen. The information can be useful when troubleshooting error messages that are received during an active job or why a run failed.


Service	Function
Jobs Server	Shows a list of server information and active (running) software.
Services	Shows the service name, description, and status. The Services list can be used for troubleshooting. If a service has stopped during a run, an option to start the service is visible for manager- and administrator-level users. For more information, see “Start a stopped service (manager/administrator)” on page 295.
Active Jobs	<ul style="list-style-type: none"> Shows a list of runs in progress that are active and queued on the sequencer. The message “No records available” indicates that no jobs are currently active. Allows manager- and administrator-level users to stop a job. For more information, see “Terminate an active job (manager/administrator)” on page 295.
ionCrawler Service Details	Provides information about the data transfer process from the Genexus™ Integrated Sequencer. The information includes whether the service was started or stopped, which user completed the action, and a timestamp for the action.
Smart Monitoring Service Details	Allows remote monitoring of the Genexus™ Integrated Sequencer. Allows manager- and administrator-level users to start or stop the service. For more information, see “Start a stopped service (manager/administrator)” on page 295.

(continued)


Service	Function
Download Instrument Diagnostics	Contains the initialization logs of the Genexus™ Integrated Sequencer, and allows you to download the diagnostic archive file in ZIP format.
System Check Report	Manager and administrator-level users can click Run System Check to view the system check report.


Terminate an active job (manager/administrator)

If an active job is not going to be useful or if a different job is urgent, you can opt to terminate an active job. A manager- or administrator-level user can stop an actively running job in the  **(Settings) / Services** screen.

1. In the menu bar, click  **(Settings) ▶ Services**.
2. In the **Services** screen, in the **Active Jobs** section, find the active job that you want to stop, then click **Terminate**.
A confirmation message opens, **Are you sure you want to terminate <job name> ?**.
3. Click the **Terminate** to terminate the job.

Start a stopped service (manager/administrator)

A manager- or administrator-level user can use the  **(Settings) / Services** screen to restart a stopped service if a service has stopped running for some reason. The two database services, **ntp** and **deemplaser** are the only services that can be restarted.


1. In the menu bar, click  **(Settings) ▶ Services**.
2. In the **Services** screen, in the **Services** section, find the service that you want to reactivate, then click **Start**.
A confirmation message opens, **Are you sure you want to Start <service name>?**.
3. Click **Yes** to restart the service.

Disable or enable the Smart Monitoring service (manager/administrator)

The Smart Monitoring service allows Thermo Fisher Scientific personnel to monitor the status of the Genexus™ Integrated Sequencer remotely through an internet connection. Smart Monitoring employs multiple layers of security, including a Secure Sockets Layer (SSL) and Lightweight Directory Access Protocol (LDAP) authentication, to provide real-time troubleshooting and problem resolution for the Genexus™ Integrated Sequencer.

The Smart Monitoring service is active by default and can be disabled by manager- or administrator-level users, if required.

- Disable the service.
 - a. In the menu bar, click  **(Settings) ▶ Services**.

- b. In the **Services** screen, in the **Smart Monitoring Service Details** section, click **Stop**.
 - c. In the confirmation message, click **Yes** to disable the service.
The **Stop** option changes to **Start**.
 - Enable the service.
 - a. In the menu bar, click  **(Settings)** ▶ **Services**.
 - b. In the **Services** screen, in the **Smart Monitoring Service Details** section, click **Start**.
 - c. In the confirmation message, click **Yes** to enable the service.
The **Start** option changes to **Stop**.

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You can expand the analysis capabilities of Genexus™ Software with plugins that are preinstalled with the software. The plugin results are added to the report summary and can be used for a various purposes.

An administrator-level user can download and install more plugins from the Connect cloud-based platform.

coverageAnalysis plugin in Genexus™ Software

Use the coverageAnalysis plugin to view statistics and graphs that describe the level of sequence coverage produced for targeted genomic regions. The results for a run analyzed with the plugin vary based on the library type that you select when you configure the plugin. You can export some charts as graphics, such as the Amplicon Coverage Chart and the Reference Coverage Chart.



Review coverageAnalysis plugin results



The coverageAnalysis plugin generates a Coverage Analysis Report. This report includes read statistics and several charts. The statistics and charts that are presented depend on the library type for the analysis.

The report summary lists the barcode, the sample, the number of mapped reads, the percentage of on target reads, mean base coverage depth, and base coverage uniformity. Microsoft™ Excel™-compatible reports are also generated, including differential expression tables. Additional details regarding read coverage are also provided on a per-barcode basis, along with a list of gene annotations for each sequenced region.

You can download coverageAnalysis plugin output files from the **Results** screen for a sample. For more information, see “Results files” on page 169 and “Output files generated by the coverageAnalysis plugin” on page 305.

1. In the menu bar, click **Results** ▶ **Sample Results**.
2. In the **Sample Results** screen, in the **Sample Name** column, click the sample of interest.

3. Click the **Plugins** tab.
A summary table of the coverage analysis, by barcode, is included in the **coverageAnalysis** summary pane.
4. (Optional) From the **Executed At** dropdown list, select an alternate timestamp, if available, to view additional reports.
5. (Optional) Click  **View Log** to view the coverageAnalysis log.
6. (Optional) Click  **Delete** to delete the coverageAnalysis plugin output for the selected timestamp.

IMPORTANT! If you click  **Delete**, the report is deleted without the appearance of confirmation dialog window. Ensure that you intend to delete the report before clicking  **Delete**.

7. In the **coverageAnalysis** summary pane, in the **Barcode Name** column, click the link in the row of the barcode of interest.
The detailed **Coverage Analysis Report** for the barcode opens in a separate window.

Reads statistics

The detailed **Coverage Analysis Report** that is available when you select a barcode in the coverageAnalysis plugin summary for a run includes a variety of reads statistics. The statistics that are displayed in your report depend on the type of library that is used in your sequencing experiment. Definitions are in tooltips. Almost every statistic, plot, link, and functional widget in the report provides tooltips with definitions. Hover over a heading or description in the report to view the tooltip.

General statistics

Statistic	Description
Number of mapped reads	The total number of reads mapped to the reference genome.
Percent reads on target	The percentage of filtered reads mapped to any targeted region relative to all reads mapped to the reference. If no target regions file is specified, this value will be the percentage of reads passing uniquely mapped and/or non-duplicate filters, or 100% if no filters were specified. A read is considered on target if at least one aligned base overlaps at least one target region. A read that overlaps a targeted region but where only flanking sequence is aligned, for example, due to poor matching of 5' bases of the read, is not counted.

Amplicon read coverage statistics

The following statistics describe the reads that are assigned to specific amplicons. Each sequence read is assigned to exactly one of the amplicons specified by the targets file. If a read spans multiple amplicon targets, the target region that the reads covers most is assigned. In the event of a tie, the target that is the closest to the 3' end is assigned.

Statistic	Description
Number of amplicons	The number of amplicons that is specified in the target regions file.
Percent assigned amplicon reads	The percentage of reads that were assigned to individual amplicons relative to all reads mapped to the reference. A read is assigned to a particular (inner) amplicon region if any aligned bases overlap that region. If a read might be associated with multiple amplicons, it is assigned to the amplicon region that has the greatest overlap of aligned sequence.
Average reads per amplicon	The average number of reads assigned to amplicons.
Uniformity of amplicon coverage	The percentage of amplicons that had at least 20% of the average number of reads per amplicon. Cumulative coverage is linearly interpolated between nearest integer read depth counts.
Amplicons with at least <i>N</i> reads	The percentage of all amplicons that had at least <i>N</i> reads.
Amplicons with no strand bias	The percentage of all amplicons that did not show a bias towards forward or reverse strand read alignments. An individual amplicon has read bias if it has ≥ 10 reads and the percentage of forward or reverse reads to total reads is greater than 70%. Amplicons with < 10 reads are considered to have no strand bias.
Amplicons reading end-to-end	The percentage of all amplicons that were considered to have a sufficient proportion of assigned reads (70%) that covered the whole amplicon target from 'end-to-end'. To allow for error, the effective ends of the amplicon region for read alignment are within 2 bases of the actual ends of the region.
Amplicon base composition bias	A number that represents the proportion of amplicons showing low representation ($< 0.2 \times$ mean reads) in the lower and/or upper quartiles of amplicons ordered by increasing G/C base pair content of their insert sequences. The value is relative to that in the center 50th percentile of amplicons and weighted by the standard deviation of representation over all amplicons. An RMS (root mean square) value is used so that a bias greater in either upper or lower quartiles produces a larger value than a mean bias seen more equally in both outer quartiles. The value is 0 if the uniformity of amplicon coverage metric is 100%, however, the value is not necessarily high at lower amplicon uniformity.

Target base coverage statistics

The following statistics describe the targeted base reads of the reference. A base covered by multiple target regions is counted only once per sequencing read.

Statistic	Description
Bases in target regions	The total number of bases in all specified target regions of the reference.
Percent base reads on target	The percent of all bases covered by reads aligned to the reference that covered bases in target regions. Clipped bases, deletions, and insertions (relative to the reference) are not included in this percentage. If no specific target regions were specified, the whole genome is the targeted regions.

Target base coverage statistics (continued)

Statistic	Description
Average base coverage depth	The average number of reads of all targeted reference bases. This is the total number of base reads on target divided by the number of targeted bases, and therefore includes any bases that had no coverage.
Uniformity of base coverage	The percentage of bases in all targeted regions (or whole genome) that is covered by at least 20% of the average base coverage depth reads. Cumulative coverage is linearly interpolated between nearest integer base read depths.
Target base coverage at Nx	The percentage of target bases covered by at least N reads.
Target bases with no strand bias	The percentage of all target bases that did not show a bias toward forward or reverse strand read alignments. An individual target base is considered to have read bias if it has ≥10 reads and the percentage of forward or reverse reads to total reads is greater than 70%. Target bases with <10 reads are considered to have no strand bias.
Percent end-to-end reads	The percentage of on-target reads that fully cover their assigned amplicon (insert) from 'end-to-end'. To allow for error, the effective ends of the amplicon region for read alignment are within 2 bases of the actual ends of the region.
Bases in targeted reference ^[1]	The total number of bases in all target regions of the reference.
Bases covered (at least 1x) ^[1]	The total number of target bases that had at least one read aligned over the proximal sequence. Only the aligned parts of each read are considered. For example, unaligned (soft-cut) bases at the 5' ends of mapped reads are not considered. Covered target reference bases can include sample DNA read base mismatches, but does not include read base deletions in the read, nor insertions between reference bases.
Average base read depth ^[1]	The average number of reads of all targeted reference bases that were read at least one time.
Genome Base Coverage ^[1]	Summary statistics for base reads of the reference genome.
Genome base coverage at Nx ^[1]	The percentage of reference genome bases covered by at least N reads.

^[1] Applicable to whole genome library analyses only.

Example charts generated by the coverageAnalysis plugin

Many of the charts in the detailed **Coverage Analysis Report** include a **Plot** menu that allows you to change characteristics of the chart. For example, you can show both strands (see callout 3 in Figure 6).

The **Q** button (in the top right corner of a chart) opens the chart **Viewing Options** panel. The **?** button opens a description of the chart.

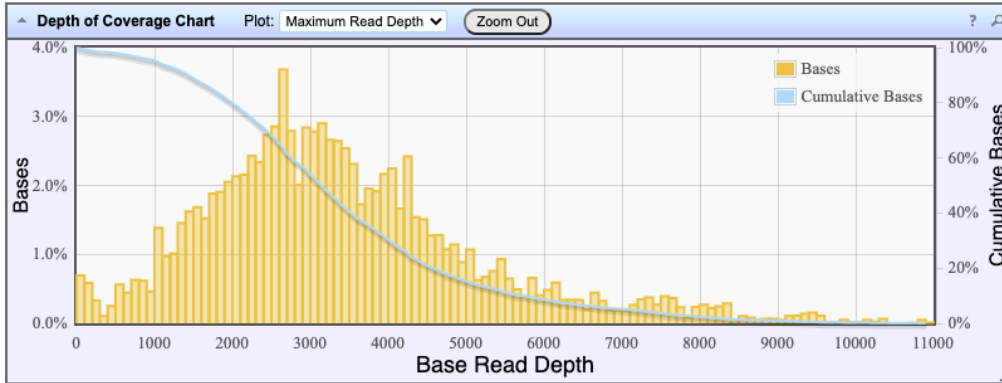


Figure 2 Representative Depth of Coverage Chart

The Depth of Coverage Chart shows the distribution of targeted base coverage. The X-axis represents the base read depth. The left Y-axis represents the number of reads at a given base read depth or a range (bin) of base read depths, as a percentage of the total number of base reads. The right Y-axis represents the cumulative count of the number of reads at a given read depth or greater, as a percentage of the total number of reads. The individual orange bars represent the percentage of reads in the specific range of base read depths. The blue curve measures the cumulative reads at a given base read depth or greater. If your analysis includes a regions of interest file, this chart reflects only target regions (reads that fall within a region of interest).

In most charts, you can click a data point to open a detail pane for that data. For example, in the Depth of Coverage Chart in Figure 2, click an individual orange bar to open the detail pane for bases within a specific range of base read depths. Click a point on the blue curve to open the detail pane for cumulative bases at that base read depth or greater. For example detail panes, see Figure 3. Similarly, you can click any point or bar within the Amplicon Coverage chart to view details for each amplicon coverage bin (see Figure 5).

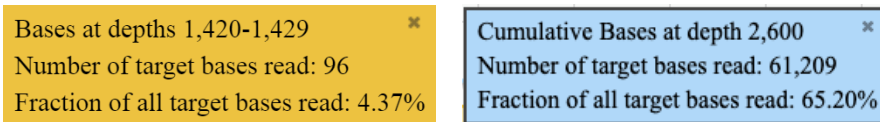


Figure 3 Example detail panes

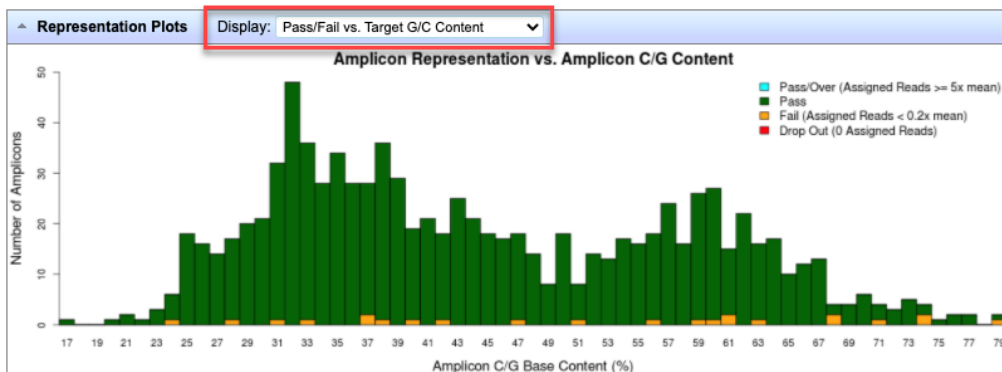


Figure 4 Representation Plots

Use the **Display** list to view different amplicon representation plots. This figure shows an example Pass/Fail vs. Target G/C Content plot.

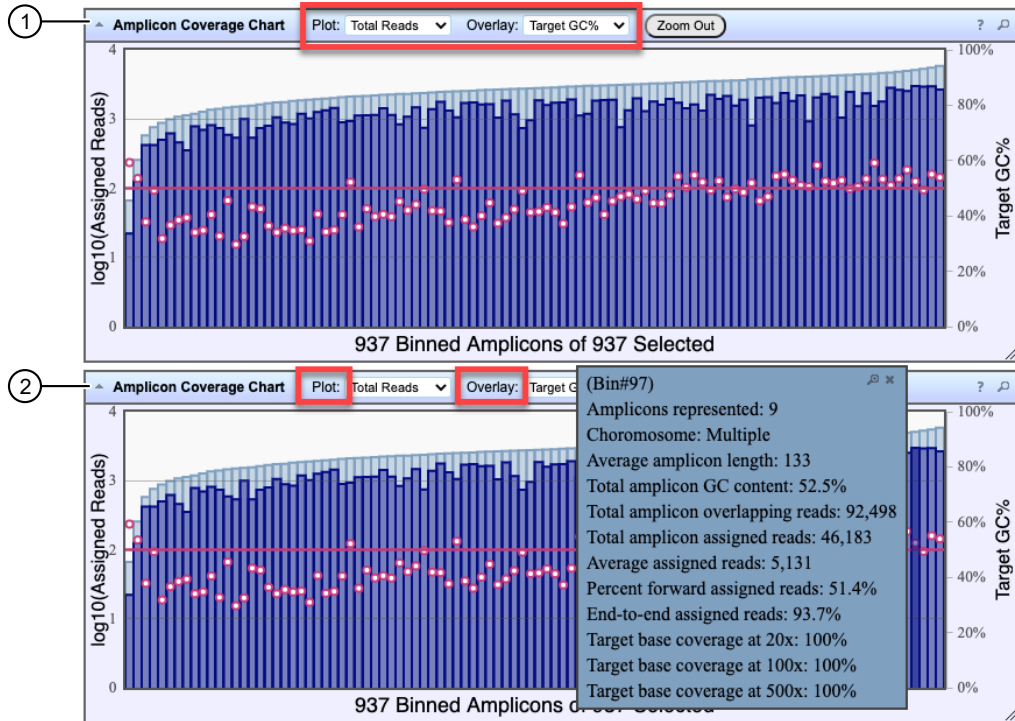


Figure 5 Representative Amplicon Coverage Chart

The Amplicon Coverage charts summarize the amplicon coverage results. Several plots views are available. The X-axis in all plots represents individual amplicons. The left Y-axis in all plots represents assigned reads (on a base 10 log scale). Use the **Plot** and **Overlay** lists to customize the chart view. The right Y-axis varies depending on the plot view that is selected from the **Overlay** dropdown list. Click on any bar or any point on the chart to view the detail pane for that data.

- ① Example Total Reads plot with a Target GC percentage overlay
- ② Click on a bar to open the detail pane for the specific amplicon. Click **View in Reference Coverage Chart** to zoom in on the amplicon in the **Reference Coverage Chart** (see callout 3 in Figure 6).

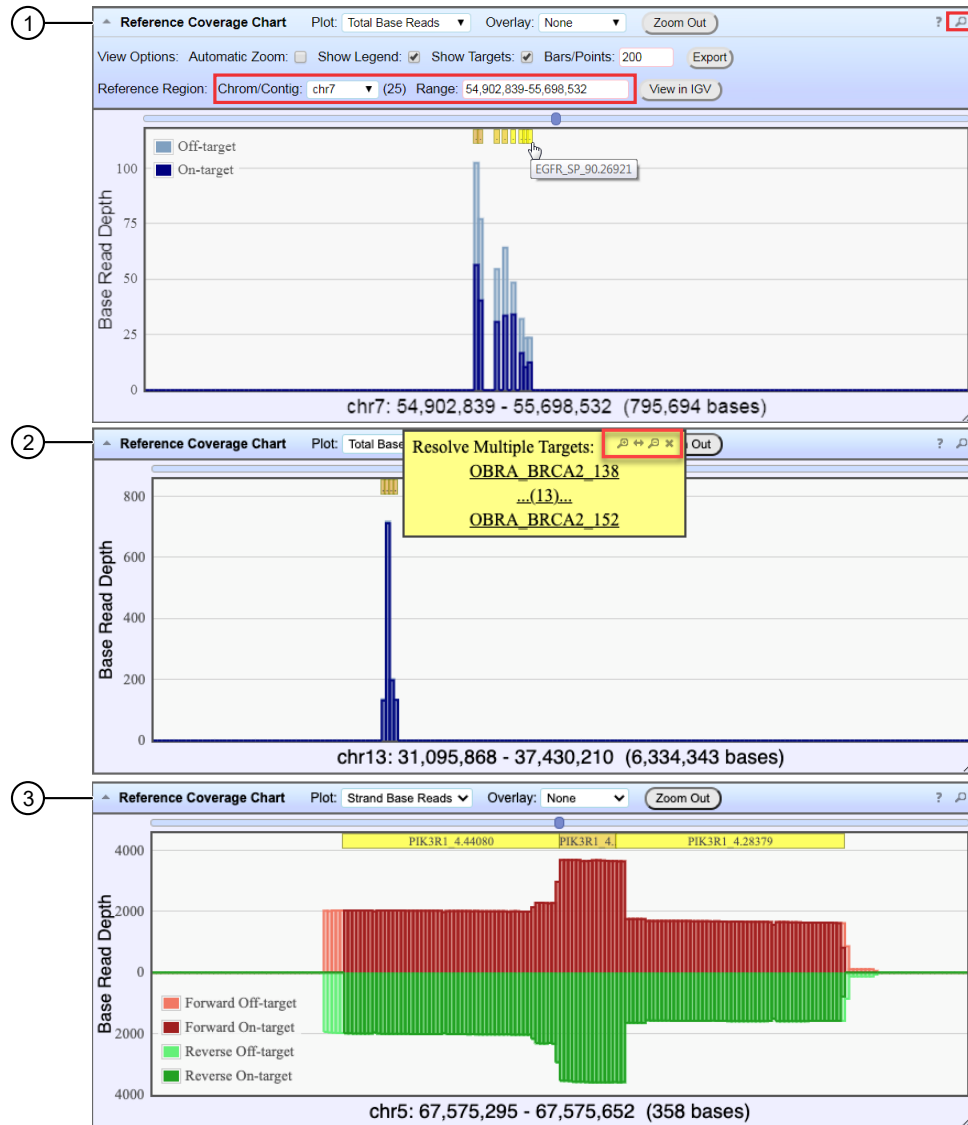



Figure 6 Representative Reference Coverage Chart

The Reference Coverage Chart is an overlay of where target regions are defined and overlap on the reference. The X-axis represents the target region chromosomal position. The Y-axis represents the Base Read Depth.

- ① Example Total Base Reads plot with Viewing Options panel expanded. The plot is zoomed in on a specific chromosomal region. Click a yellow bar (top of the chart) to open the detail pane for a specific amplicon in the Amplicon Coverage Chart (see callout 2 in Figure 5).
- ② Example Total Base Reads plot where a chromosomal region has multiple targets. In the Resolve Multiple Targets pane, click  to zoom in on the region, or click one of the links to open the detail pane for the specific amplicon in the Amplicon Coverage Chart.
- ③ Example Strand Base Reads plot that is zoomed in on one specific amplicon. Click the yellow box to open the detail pane for the specific amplicon in the Amplicon Coverage Chart.

Example Coverage Analysis Report

The detailed Coverage Analysis Report that is available when you select a barcode in the coverageAnalysis plugin summary for a run includes a variety of reads statistics.

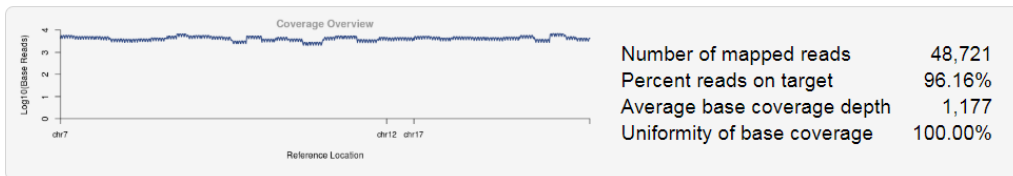
Coverage Analysis Report

Sample Name:

Library type: AmpliSeq HD - DNA and Fusions (Single Library)

Reference: hg19 (DNA)

Target regions:



Amplicon Read Coverage		Target Base Coverage	
Number of amplicons	39	Bases in target regions	2,195
Percent assigned amplicon reads	96.16%	Percent base reads on target	53.78%
Average reads per amplicon	1,201	Average base coverage depth	1,177
Uniformity of amplicon coverage	100.00%	Uniformity of base coverage	100.00%
Amplicons with at least 1 read	100.00%	Target base coverage at 1x	100.00%
Amplicons with at least 20 reads	100.00%	Target base coverage at 20x	100.00%
Amplicons with at least 100 reads	100.00%	Target base coverage at 100x	100.00%
Amplicons with at least 500 reads	100.00%	Target base coverage at 500x	100.00%
Amplicons with no strand bias	100.00%	Target bases with no strand bias	100.00%
Amplicons reading end-to-end	100.00%	Percent end-to-end reads	96.21%
Amplicon base composition bias	0.000		

Output files generated by the coverageAnalysis plugin

You can download coverageAnalysis plugin results files from the **Results** screen for a sample. For more information, see “Results files” on page 169.

Note: Sometimes the file name can be too long to open in applications such as Microsoft™ Excel™. To resolve this problem, right-click the file and click **Save As** to rename the downloaded file.

The following tables describe the files that can be generated by the coverageAnalysis plugin. The list of files depends on the application type that was selected during assay creation.

File download selections in the Genexus™ Software

Selection	File Name
DNA Coverage Statistics	<DNA Barcode><ExpName>.stats.cov.txt
DNA Chromosome base coverage summary	<DNA Barcode><ExpName>.chr.cov.xls
DNA Base depth of coverage	<DNA Barcode><ExpName>.base.cov.xls
DNA Amplicon coverage summary	<DNA Barcode><ExpName>.amplicon.cov.xls
DNA Coverage Analysis Summary (.pdf)	<DNA Barcode><ExpName>.summary.pdf
RNA Coverage Statistics	<RNA Barcode><ExpName>.stats.cov.txt
RNA Amplicon coverage summary	<RNA Barcode><ExpName>.amplicon.cov.xls
RNA Coverage Analysis Summary (.pdf)	<RNA Barcode><ExpName>.summary.pdf
TNA Coverage Statistics	<TNA Barcode><ExpName>.stats.cov.txt
TNA Chromosome base coverage summary	<TNA Barcode><ExpName>.chr.cov.xls
TNA Base depth of coverage	<TNA Barcode><ExpName>.base.cov.xls
TNA Amplicon coverage summary	<TNA Barcode><ExpName>.amplicon.cov.xls
TNA Coverage Analysis Summary (.pdf)	<TNA Barcode><ExpName>.summary.pdf

File contents

File	Description
Coverage Statistics	A summary of the statistics presented in the tables at the top of the plugin report. The first line is the title. Each subsequent line is either blank or a particular statistic title followed by a colon (:) and its value.
Chromosome base coverage summary	<p>Base reads per chromosome summary data that is used to create the default view of the Reference Coverage Chart. This file contains the following fields.</p> <ul style="list-style-type: none"> • <code>chrom</code>: the name of the chromosome or contig of the reference. • <code>start</code>: the coordinate of the first base in this chromosome. This is always 1. • <code>end</code>: the coordinate of the last base of this chromosome. Also its length in bases. • <code>fwd_basereads</code>: the total number of forward strand base reads for the chromosome. • <code>rev_basereads</code>: the total number reverse strand base reads for the chromosome. • <code>fwd_trg_basereads</code> (if present): the total number of forward strand base reads that mapped over at least one target region. • <code>rev_trg_basereads</code> (if present): the total number of reverse strand base reads that mapped over at least one target region. • <code>total_reads</code>: the total number of sequencing reads that are mapped to individual contigs.
Base depth of coverage	<p>Coverage summary data used to create the Depth of Coverage Chart. This file contains the following fields.</p> <ul style="list-style-type: none"> • <code>read_depth</code>: the depth at which a (targeted) reference base has been read. • <code>base_cov</code>: the number of times any base was read (covered) at this depth. <p>Note: Lines (read depths) for which <code>base_cov</code> is 0 are omitted to avoid excessively large files being produced in specific situations.</p> <ul style="list-style-type: none"> • <code>base_cum_cov</code>: the cumulative number of reads (coverage) at this read depth or greater. • <code>norm_read_depth</code>: the normalized read depth (depth divided by average base read depth). • <code>pc_base_cum_cov</code>: same as <code>base_cum_cov</code> but represented as a percentage of the total base reads.

File contents (continued)

File	Description
Amplicon coverage summary	<p>Coverage summary data used to create the Amplicon Coverage Chart. This file contains the following fields:</p> <ul style="list-style-type: none"> • <code>contig_id</code>: the name of the chromosome or contig of the reference for this amplicon. • <code>contig_srt</code>: the start location of the amplicon target region. Note: This coordinate is 1-based, unlike the corresponding 0-based coordinate in the original targets BED file. • <code>contig_end</code>: the last base coordinate of this amplicon target region. Note: The length of the amplicon target is given as $tlen = (contig_end - contig_srt + 1)$. • <code>region_id</code>: the ID for this amplicon as given as the fourth column of the targets BED file. • <code>gene_id</code> or <code>attributes</code>: the gene symbol or attributes field as provided in the targets BED file. • <code>gc_count</code>: the number of G and C bases in the target region. The %GC that is uses this count divided by the amplicon (insert) length. • <code>overlaps</code>: the number of times this target was overlapped by any read by at least one base. Note: Individual reads might overlap multiple amplicons where the amplicon regions themselves overlap. • <code>fwd_e2e</code>: the number of assigned forward strand reads that read from one end of the amplicon region to the other end. • <code>rev_e2e</code>: the number of assigned reverse strand reads that read from one end of the amplicon region to the other end. • <code>total_reads</code>: the total number of reads assigned to this amplicon. This value is the sum of <code>fwd_reads</code> and <code>rev_reads</code> and is the field that rows of this file are ordered by (then by <code>contig id</code>, <code>srt</code>, and <code>end</code>). • <code>fwd_reads</code>: the number of forward strand reads that are assigned to this amplicon. • <code>rev_reads</code>: the number of reverse strand reads that are assigned to this amplicon. • <code>covNx</code>: the number of bases of the amplicon target that had at least N reads. There are 3 such columns for the specified coverage tiers, which by default are <code>cov20x</code>, <code>cov100x</code>, and <code>cov500x</code>.
Coverage Analysis Summary (.pdf)	<p>A PDF file that contains the Coverage Analysis Report, including read statistics and charts that are generated by the coverageAnalysis plugin.</p>

molecularCoverageAnalysis plugin in Genexus™ Software

Use the molecularCoverageAnalysis plugin to view statistics and graphs that describe the level of sequence molecular coverage produced for targeted genomic regions. The plugin generates statistics, downloadable data files, and interactive visualization of molecular coverage over targeted regions of the reference genome.

The plugin is only compatible with Ion AmpliSeq™ HD library chemistry.

Review molecularCoverageAnalysis plugin results



The molecularCoverageAnalysis plugin generates a Molecular Coverage Analysis Report. This report includes molecule statistics and several charts. The report summary lists the barcodes, the samples, the median molecular coverage, the molecular uniformity, and the median reads per functional molecule, and the median percentage of functional reads. Details regarding molecular coverage are also provided on a per-barcode basis, along with a list of gene annotations for each sequenced region.

You can download statistics files from the file links at the bottom of the results screen when the **Plugins** tab is selected. After the sequencing run completes, review the plugin results in the report summary.

1. In the menu bar, click **Results** ▶ **Sample Results**.
2. In the **Sample Results** screen, in the **Sample Name** column, click the sample of interest.
3. Click the **Plugins** tab.

A summary table of the molecular coverage analysis, by barcode, is included in the **molecular CoverageAnalysis** summary pane, located beneath the **coverageAnalysis** summary pane, if present.

4. In the **molecularCoverageAnalysis** summary pane, view detailed plugin information.

Option	Description
View a detailed Molecular Coverage Analysis Report for a barcoded sample.	In the Barcode Name column, click the barcode name of interest.
View the molecularCoverageAnalysis log.	Click  View Log .
View more reports.	In Executed At , select an alternate timestamp, if available.
Delete the molecularCoverageAnalysis plugin output for the selected timestamp.	Click  Delete .
Download a JSON file that shows the parameter settings for the plugin.	Click the Parameters link.

5. Click the links at the bottom of the **Molecular Coverage Analysis Report** to download associated statistics and summary files for each barcoded sample in the run.

Molecule statistics

The detailed **Molecular Coverage Analysis Report** that is available when you select a barcode in the coverageAnalysis plugin summary for a run includes a variety of reads statistics. The statistics that are displayed in your report depend on the type of library that is used in your sequencing experiment. Definitions are in tooltips. Almost every statistic, plot, link, and functional widget in the report provides tooltips with definitions. Hover over a heading or description in the report to view the tooltip.

Statistic	Description
Median Functional Molecular Coverage per Amplicon	A functional molecule is a collection of reads that covers the amplicon and satisfies all criteria associated with the parameters (for example, <code>min_tag_fam_size</code> and <code>min_fam_per_strand_cov</code>) in the parameter file. Molecular coverage is the number of functional molecules. The median is calculated across all amplicons.
Uniformity of Molecular Coverage for all Amplicons	Percentage of amplicons having molecular coverage between 0.5x and 2x of the median molecular coverage.
Percentage of Amplicons larger than 0.8x Median Functional Molecular Coverage	Percentage of amplicons having molecular coverage more than 0.8x of the median functional molecular coverage
Median Total Molecular Coverage per Amplicon	The number of molecules that satisfies size criteria in the parameters file. The median is calculated across all amplicons.
Percentage of Reads with Perfect Molecular Tags	Percentage of reads whose molecular tags are exactly the same with design.
Median Functional Molecular Loss due to Strand Bias per Amplicon	The metric is calculated by $1 - (\text{Functional Molecular Coverage}) / (\text{Molecular Coverage without Strand Constraint})$, which measures the loss of functional molecules due to strand constraint for molecular functionality. The median is calculated across all amplicons. This statistic is available in Ion AmpliSeq™ HD runs only.
Median Percentage of Functional Molecules out of Total Molecules per Amplicon	The percentage of functional molecules out of the number of molecules that satisfies size criteria. The median is calculated across all amplicons.
Median Reads per Functional Molecule	For each amplicon, the number of reads supporting each functional molecule is averaged across all functional molecules for that amplicon to determine the number of reads per functional molecule at the amplicon level. The median is calculated across all amplicons.
Median Reads Contributed to Functional Molecules per Amplicon	For each amplicon, the percentage of reads supporting functional molecules is $(\text{Number of Reads Supporting Functional Molecules}) / (\text{Number of Reads})$. The median is calculated across all amplicons.
Percentage of Amplicons below (around) n% LOD	LOD is calculated based on the number of functional molecules for each amplicon and variant calling parameters in the parameter file. Around x n% means in the range from 0.5x n% to 2x n%.

Example Molecular Coverage Analysis Report

The following is an example of a **Molecular Coverage Analysis Report** is available when you select a barcode in the molecularCoverageAnalysis plugin summary.

Molecular Coverage Analysis Report

Sample Name:

Library type: AmpliSeq HD - DNA
Reference: hg19 (DNA)
Target regions:

Number of amplicons	272
Median functional molecules per amplicon	2.521
Uniformity of amplicon molecular coverage	90.81%
Amplicons with LOD below 5%	99.26%
Amplicons with LOD below 1%	95.59%
Amplicons with LOD below 0.5%	85.66%
Amplicons with LOD around 0.1%	1.47%
Median number of reads per molecule	26.8
Median molecular loss due to strand bias	78.90%
Median reads to molecular conversion rate	80.56%

Example charts generated by the molecularCoverageAnalysis plugin

The charts in the detailed **Molecular Coverage Analysis Report** include **Plot** and **Overlay** menus that allow you to customize the data that is displayed in each chart.

Click **Q (Search)** (in the top right corner of a chart) to open the chart **Viewing Options** panel. Click **? (Help)** to open a description of the chart.

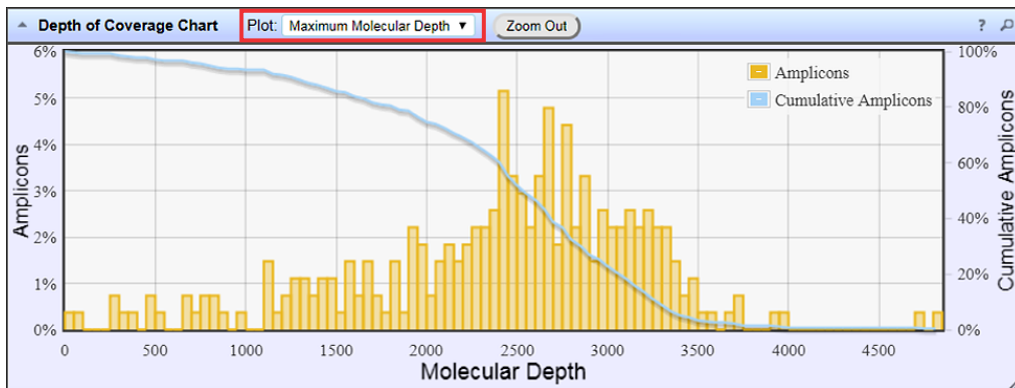


Figure 7 Representative Depth of Coverage Chart

The Depth of Coverage Chart summarizes the amplicon depth of coverage results. The X-axis represents the amplicon molecular depth. The left Y-axis represents the number of amplicons at a given molecular depth or a range (bin) of molecular depths, as a percentage of the total number of amplicons. The right Y-axis represents the cumulative count of the number of amplicons, at a given molecular depth or greater, as a percentage of the total number of amplicons. The individual orange bars represent the percentage of amplicons in the specific range of molecular depths. The blue curve measures the cumulative amplicons at a given molecular depth or greater. Use the **Plot** dropdown list to switch between **Maximum Read Depth**, **99.9% of All Reads**, and **Normalized Coverage** plots.

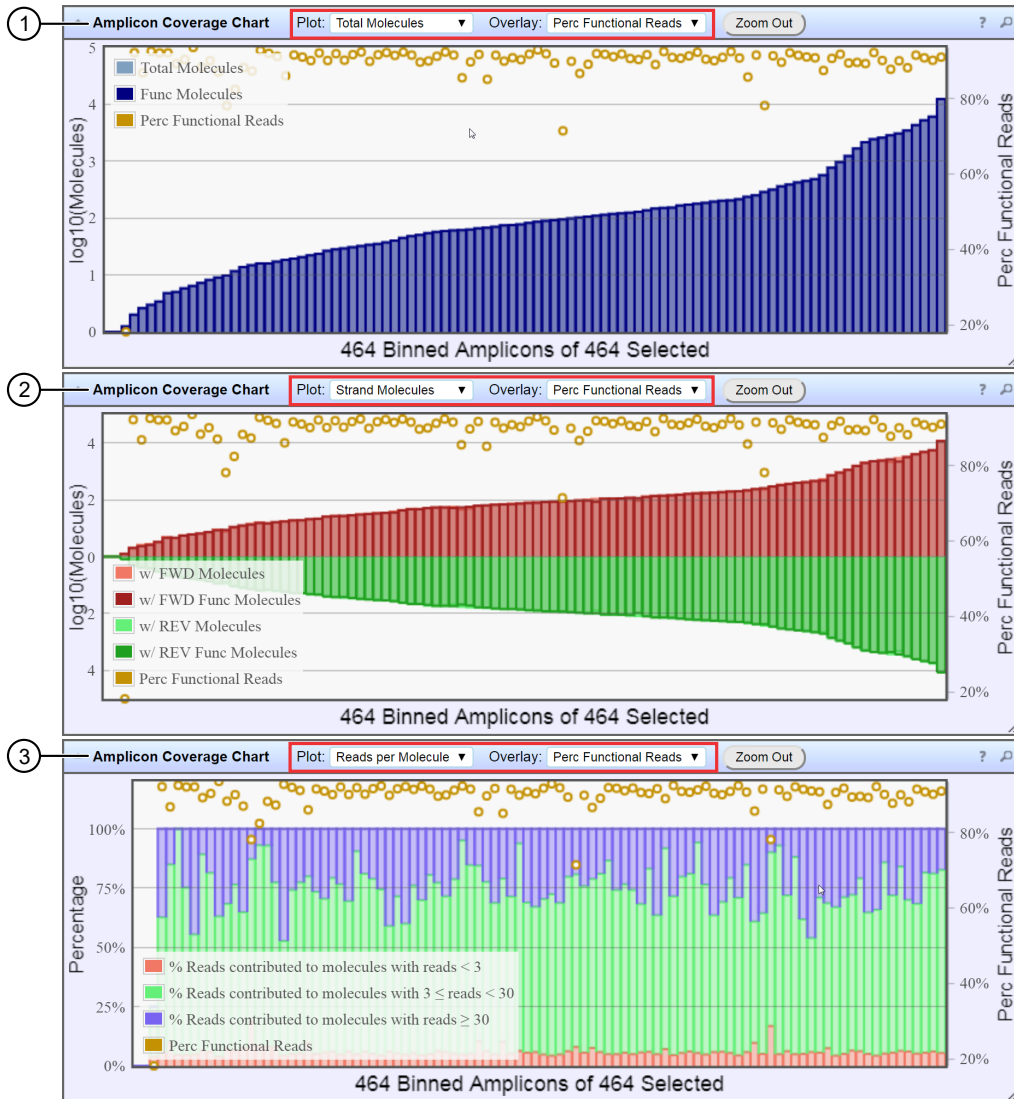


Figure 8 Representative Amplicon Coverage charts

The Amplicon Coverage charts summarize the amplicon molecular coverage results. The X-axis in all plots represents amplicons that are grouped into bins, where each bin contains amplicons that have the same molecular coverage. Depending on the plot selection, the left Y-axis represents either the average number of molecules (on a base 10 log scale) or the percentage of functional molecules in a given bin. The right Y-axis represents the percentage of functional reads in a given bin. Use the **Plot** and **Overlay** dropdown lists to customize the chart view. To zoom in on a subset of amplicons, use the pointer to draw a rectangle that contains the region of interest. Use **Zoom Out** to revert to the initial view.

- ① Representative **Total Molecules** plot with an **Perc Functional Reads** overlay
- ② Representative **Strand Molecules** plot with an **Perc Functional Reads** overlay
- ③ Representative **Reads per Molecule** plot with an **Perc Functional Reads** overlay

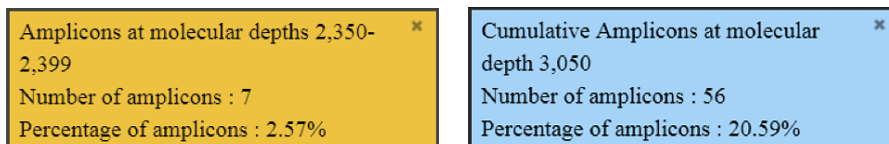


Figure 9 Example detail panes

In most plots, you can click a data point to open a detail pane for that data. For example, in the Depth of Coverage Chart in Figure 7, click an individual orange bar to open the detail pane for amplicons in a specific range of molecular depths or click a point on the blue curve to open the detail pane for cumulative amplicons at that molecular depth or greater. Similarly, you can click any point or bar within each Amplicon Coverage chart to view details for each amplicon coverage bin. For example detail panes, see Figure 9.

Output files generated by the molecularCoverageAnalysis plugin

You can download plugin results files from the **Results** screen for a sample. For more information, see “Results files” on page 169.

Note: Sometimes the file name can be too long to open in applications such as Microsoft™ Excel™. To resolve this problem, right-click the file, then click **Save As** to rename the downloaded file.

The following tables describe the results files that can be generated by the molecularCoverageAnalysis plugin. The list of files depends on the application type that was selected during assay creation.

File download selections in the Genexus™ Software

Selection	File name
TNA Molecular Coverage Statistics	<Barcode><ExpName>.cov.stats.txt
TNA Molecular depth of coverage	<Barcode><ExpName>.mol.cov.xls
TNA Molecular Coverage Analysis Summary	<Barcode><ExpName>.summary.pdf
TNA Molecular Amplicon coverage summary	<Barcode><ExpName>.amplicon.cov.xls

File contents

File	Description
Molecular Coverage Statistics	This file contains a summary of the statistics presented in the tables at the top of the Molecular Coverage Analysis Report plugin report. The first line is the title. Each subsequent line is either blank or a particular statistic title followed by a colon (:) and its value.
Molecular depth of coverage	Molecular coverage summary data used to create the Depth of Coverage Chart. This file contains the following fields: <ul style="list-style-type: none"> • <code>mol_depth</code>: the depth at which a (targeted) reference amplicon has been read. • <code>amp_cov</code>: the number of times any amplicon was read (covered) at this depth. • <code>amp_cum_cov</code>: the cumulative number of reads (coverage) at this read depth or greater. • <code>norm_mol_depth</code>: the normalized read depth (depth divided by average molecular read depth). • <code>pc_amb_cum_cov</code>: same as <code>amp_cum_cov</code> but represented as a percentage of the total reads.
Molecular Coverage Analysis Summary	A PDF file that contains the Molecular Coverage Analysis Report, including molecule statistics and charts that are generated by the <code>molecularCoverageAnalysis</code> plugin.

File contents (continued)

File	Description
Molecular Amplicon coverage summary	<p>Amplicon molecular coverage summary data used to create the Amplicon Coverage Chart. This file contains the following fields:</p> <ul style="list-style-type: none"> • <code>contig_id</code>: the name of the chromosome or contig (from contiguous) of the reference for this amplicon. • <code>contig_srt</code>: the start location of the amplicon target region. Note that this coordinate is 1-based, unlike the corresponding 0-based coordinate in the original targets BED file. • <code>contig_end</code>: the last base coordinate of this amplicon target region. Note that the length of the amplicon target is given as $(contig_end - contig_srt + 1)$. • <code>region_id</code>: the ID for this amplicon as given as the 4th column of the targets BED file. • <code>gene_id</code> or <code>attributes</code>: the gene symbol or attributes field as provided in the targets BED file. • <code>func_mol_cov</code>: functional molecular coverage utilized for variant calling. • <code>lod</code>: LOD (limit of detection) calculated from the number of functional molecules. • <code>strict_func_omt_rate</code>: the percentage of functional molecules used with strict molecular tags. • <code>func_mol_cov_loss_due_to_strand</code>: the percentage of functional molecules not formed. • <code>fwd_only_mol_cov</code>: the number of molecules containing forward strand only. • <code>rev_only_mol_cov</code>: the number of molecules containing reverse strand only. • <code>both_strands_mol_cov</code>: the number of molecules containing both forward strand and reverse strand. • <code>r2m_conv_rate_all</code>: the percentage of reads contributed to functional molecules. • <code>reads_per_func_mol</code>: the average reads per functional molecules. • <code>perc_to_mol_(<3_reads)</code>: the percentage of reads contributed to small size molecules (size <3). • <code>perc_to_mol_(>=3&<30_reads)</code>: the percentage of reads contributed to median size molecules (size ≥ 3 && size <30). • <code>perc_to_mol_(>=30_reads)</code>: the percentage of reads contributed to large size molecules (size ≥ 30).

IonReporterUploader plugin

Sample results files that are generated in Genexus™ Software can be directly transferred to Ion Reporter™ Software with the IonReporterUploader plugin.

Ion Reporter™ Software performs analysis on unmapped BAM files that are output from Genexus™ Software. To transfer the BAM files to Ion Reporter™ Software, you must add one or more Ion Reporter™ Software accounts in Genexus™ Software. To configure an Ion Reporter™ Server account that you can use for data uploads, see “Configure an Ion Reporter™ Server account (manager/administrator)” on page 254 and “Configure an Ion Reporter™ Software on Connect account (manager/administrator)” on page 253.

After an account is configured, there are two ways that you can transfer files to Ion Reporter™ Software.



- When you plan a run, you can select to have Genexus™ Software directly transfer output files to Ion Reporter™ Software. For more information, see “Plan a Sample to Result run” on page 65, “Plan a Nucleic Acid to Result run” on page 71, and “Plan a Library to Result run” on page 75.
- You can select to upload data manually to Ion Reporter™ Software. Use this option if, for example, you want to upload data to multiple Ion Reporter™ Software servers. For more information, see “Upload results files to Ion Reporter™ Software” on page 171.

Note: Ion Reporter™ Software is available under separate license and is not included with Genexus™ Software.

Review IonReporterUploader plugin results

After the analysis is complete, review the plugin results.

1. In the menu bar, click **Results ▶ Run Results**.
2. In the **Run Results** screen, in the **Run Name** column, click the run of interest.
3. In **Select Assay**, select the assay of interest.
4. Click the **Plugins** tab.
A summary table of the IonReporterUploader plugin is shown.
5. From the IonReporterUploader plugin, you can view information that is related to the data transfer, including the name of the Ion Reporter™ Server used, the version of Ion Reporter™ Software that is on the server, the server folder that contains the uploaded files, and the Ion Reporter™ Software organization and user account that was used. You can also review details about barcoded samples that were uploaded with the IonReporterUploader plugin, or failed to upload.

To do this	Click
View error messages associated with the plugin run.	Errors
View warnings that contain details about the barcoded samples that are used in the plugin run.	Warnings
Show or hide a detailed status of the pre- and post-processing of the data transfer.	Show detailed status/Hide detailed status
Open the plugin log files in a separate browser tab.	 View Log
Download a CSV file that contains a list of the uploaded and defined samples.	Download CSV list of samples that are uploaded and defined.
Download a compressed folder of the IonReporterUploader plugin log files and other plugin files	Download IRU logs
Delete the plugin table	 Delete

CustomerSupportArchive plugin

The CustomerSupportArchive plugin generates a downloadable archive that a technical support representative can use to troubleshoot and diagnose issues with sequencing runs or with Genexus™ Software. The archive contains log files and other technical data about the software and other files from sequencing runs on a Genexus™ Integrated Sequencer.

For more information, see “Download a customer support archive” on page 371.

sampleID plugin

Use the sampleID plugin to track samples or possibly identify misassignment or mix up between samples and barcodes in a sequencing run. The sampleID plugin produces a unique identification code (**SampleID**) for each barcode in a sample.

The plugin can be used with the Ion AmpliSeq™ Sample ID Panel, which is a human SNP genotyping panel, to ensure that the accuracy of samples increase confidence in sample data management. The Ion AmpliSeq™ Sample ID Panel is composed of the identified human sample gender and IUPAC base letters for eight high-frequency noncoding SNPs. Ion AmpliSeq™ Sample ID Panel contains nine primer pairs that can be combined with any Ion AmpliSeq™ Ready-to-Use Panel or Custom Panel.

For the samples to work with this plugin, the Ion AmpliSeq™ library must have been prepared with Ion AmpliSeq™ sample tracking amplicons.

The sampleID plugin is preconfigured and does not require input.

Review sampleID plugin results in Genexus™ Software

After the sequencing run completes, review the plugin results.

1. In the menu bar, click **Results** ▶ **Sample Results**.
2. In the **Sample Results** screen, in the **Sample Name** column, click the sample of interest.
3. Click the **Plugins** tab.
A summary table of the **sampleID** results is shown.
4. Click a barcode name to open the **Sample ID Report**.
5. To return to Genexus™ Software, click back in the browser.



Genexus™ Software web services API

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Genexus™ Software has a web services API that allows you to automate the retrieval of key information from the system.

This section provides examples to show you how to leverage the API.

LIMS introduction

Genexus™ Software integrates with laboratory information management systems (LIMS) to help with sample, library, and run plan creation. The interaction is through the LIMS calling a set of RESTful APIs provided by the software (see the following table). The LIMS connects to the software by making an HTTP call to the URL of an API, and sending a user name and a password in the HTTP header. The user name and password must be for a registered active user account of the software. The LIMS passes the input information in JSON format within the HTTP body, while the response from the software is returned in JSON format as well. Users must configure their LIMS to create JSON files containing the required attributes for each LIMS request file.



Outlined in the following table are the API names, functions, and URLs of the five RESTful APIs that are provided.

API	Function	API URL
<p>Get a list of Ion Reporter™ Software accounts</p> <p>For more information, see “Get list of Ion Reporter™ Software accounts” on page 321.</p>	<p>Returns a list and the details of active Ion Reporter™ Software accounts.</p>	<p>https:// <host>/genexus/api/lims/v2/accounts</p>
<p>Get report templates</p> <p>For more information, see “Get report templates” on page 322.</p>	<p>Returns report templates.</p>	<p>https:// <host>/genexus/api/lims/v2/report-templates</p>
<p>Get a list of assays</p> <p>For more information, see “Get assay list” on page 322.</p>	<p>Returns a list of the assays in the system. PQ assays are excluded.</p>	<p>https:// <host>/genexus/api/lims/v2/assays</p>
<p>Get a list of samples</p> <p>For more information, see “Get samples” on page 323.</p>	<p>Returns a list of the samples in the system.</p>	<p>https:// <host>/genexus/api/lims/v2/samples</p>
<p>Get a list of library batches</p> <p>For more information, see “Get library batches” on page 324.</p>	<p>Returns a list of the library batches in the system.</p>	<p>https:// <host>/genexus/api/lims/v2/library-batches</p>
<p>Get a list of run plans</p> <p>For more information, see “Get run plans” on page 326.</p>	<p>Returns a list of available run plans in the system.</p>	<p>https://<host>/genexus/api/lims/v2/plans</p>
<p>Get results</p> <p>For more information, see “Get results” on page 327.</p>	<p>Returns results, including plugins and plugin results. Returns a JSON object for each plugin. All plugins for the run and the sample are returned.</p>	<p>https:// <host>/genexus/api/lims/v2/results</p>
<p>Get a list of signed off samples</p> <p>For more information, see “Get signed off samples” on page 333.</p>	<p>Returns a list of the samples that are designated with (Signed Off).</p>	<p>https:// <host>/genexus/api/lims/v2/signedOffSamples</p>

(continued)

API	Function	API URL
Download files for completed runs For more information, see “Download files for completed runs” on page 334.	Returns a compressed file in ZIP format that contains the requested files for the sample.	https:// <host>/genexus/api/lims/v2/download
Create samples For more information, see “Create samples” on page 334.	Create samples.	https:// <host>/genexus/api/lims/v2/samples
Create a library batch For more information, see “Create a library batch” on page 336.	Create a library batch from existing samples.	https:// <host>/genexus/api/lims/v2/library-batches
Create a Sample to Result (purification) run For more information, see “Create a Sample to Result (purification) run” on page 337.	Create a Sample to Result run from one or more samples and an assay.	https://<host>/genexus/api/lims/v2/plan-by-purification
Create a Nucleic Acid to Result run For more information, see “Create a Nucleic Acid to Result run” on page 338.	Create a Nucleic Acid to Result run from one or more samples and an assay.	https://<host>/genexus/api/lims/v2/plan-by-samples
Create a library run For more information, see “Create a library run” on page 339.	Create a library run from one or more library batches and an assay.	https://<host>/genexus/api/lims/v2/plan-by-library

LIMS system FTP setup or drive mapping

To enable the LIMS system to retrieve data analysis files from the sequencer, a manual FTP setup or drive mapping is required. This configuration is not provided as part of the sequencer installation, and must be set up as part of the LIMS system by the system integration or laboratory IT group. See the sections in this appendix for file paths and other setup information.



API common parameters

If the JSON download process converts @ symbols to %40, do not change the text. Instead, copy the JSON files as is into the browser.

Parameter	Entry
BASE_URL	https://<host>/genexus/api/lims/v2
Request	Authorization:Basic Accept: application/json
Response	Content-Type: application/json

Response fields

Code	Status
200	Successful response
400	Bad request Error response: <pre>{ "errors": [{ "field": "validation error" }] }</pre>
401	Unauthorized
500	Internal Server Error

Get requests

Type of request	Entry
Get list of resources	BASE_URL/resource_url/
Get single object	BASE_URL/resource_url?id=object_id

Common request parameters

Parameter	Entry	Value
Starting page number	pageNumber	The default value is "1".
Number of items to fetch ^[1]	pageSize	The default value is "20".



Common request parameters (continued)

Parameter	Entry	Value
A filed based on which, the result set needs to be ordered	orderBy	Only specific values are allowed. If no value is provided, the result set is ordered by ID.
Order direction of the result set	sortOrder	Valid values are "asc" or "desc". The default value is "asc".

[1] You can limit the number of items that are fetched when you add parameters to the request. For example, add "status" to the get assay list request.

Get list of Ion Reporter™ Software accounts

accounts	
URL	BASE_URL/accounts
Description	Returns a list and the details of active Ion Reporter™ Software accounts.
Method	GET
Example request	https://<host>/genexus/api/lims/v2/accounts
Request query parameters	<ul style="list-style-type: none"> irversion String. <i>Mandatory.</i> Ion Reporter™ Software version.
Example response	<pre>{ "total": 2, "objects": [{ "id": "2", "displayName": "TFAccount_Local_516 (Default)", "serverType": "HTTPS", "port": "443", "state": "Active", "userName": "firstname.lastname@tfc.com", "org": "IR Org", "status": "ENABLED", "server": "think15dx.itw", "irVersion": "IR516", "defaultAccount": true, "accountType": "Ion Reporter Local", "usedAccount": false, "irAccountListError": false, "irAccount": true, "createdBy": "Auto1Admin" }] }</pre>



Get report templates

report-templates	
URL for a list of resources	BASE_URL/report-templates
URL for a single object	BASE_URL/report-templates?id={id}
Description	Returns report templates. API supports multiple filters.
Method	GET
Example URL	https://<host>/genexus/api/lims/v2/report-templates
Request query parameters	<ul style="list-style-type: none"> id String. <i>Optional</i>. Get the report template for the given report template id. assayName String. <i>Optional</i>. Get the report template for the given assay. Use the Assay Name. Do not use the Assay Full Name.
Example response	<pre>{ "mode": "AssayDev", "reportingTemplate": [{ "name": "default", "id": 1, "description": "Default", "locale": "en" }, { "name": "oca3_dr_oncominepositive", "id": 9, "assayName": "Oncomine Comprehensive v3 - GX5 - DNA and Fusions - w5.0.2", "assayId": 50, "description": "OCA3_DR_OncominePositive", "locale": "en" }], "total": 1 }</pre>

Get assay list

assays	
URL	BASE_URL/assays
Description	Returns a list of the assays in the system. PQ assays are excluded.
Method	GET
Example URL	https://<host>/genexus/api/lims/v2/assays

(continued)

assays	
Supported parameters	<ul style="list-style-type: none"> assayName String. <i>Optional</i>. Assay name. status String, <i>Optional</i>. Valid values are "draft", "obsolete", or "locked".
Example response	<pre>{ "total": 1, "objects": [{ "id": 122, "assayName": "copy_of_OCAv3_GX5_DNA_RNA_w5.0.2", "assayShortName": "OCAv3 DNA Fus w5.0.2", "assayDisplayedName": "Oncomine Comprehensive v3 - GX5 - DNA and Fusions - w5.0.2", "application": "DNA+RNA", "createdBy": "System Installed", "createdDate": "2021-06-22T16:50:15.000+0000", "isReanalysisAssay": false, "genomeReference": "hg19", "targetRegions": "OCAv3_w5.0.2_dna_fusions_20200626.GX5.designed.bed", "hotspots": "OCAv3_w5.0.2_dna_fusions_20201117.GX5.hotspots.bed", "fusionReference": "OCAv3_w5.0.2_designs_072720_Reference.fasta", "fusionTargetRegions": "OCAv3_w5.0.2_dna_fusions_designs_072720_Reference.bed", "chipType": "Ion Torrent™ GX5™ Chip", "panelKit": "Oncomine™ Comprehensive Assay v3 DNA and RNA, GX", "controlKit": "Other", "extractionKit": "Ion Torrent™ Genexus™ FFPE DNA/RNA Purification Combo Kit", "libraryKit": "Ion Torrent™ Genexus™ Library Strips 1 and 2-AS", "templatingKit": "Ion Torrent™ Genexus™ Templating Strips 3-GX5 and 4", "barcodeKit": "Ion Torrent™ Genexus™ Barcodes 1-96 AS", "sequencingKit": "Ion Torrent™ Genexus™ Sequencing Kit", "flows": 400, "templatingSize": 200, "status": "Locked", "tumorCellularity": "1", "reanalysisAssay": false }] }</pre>

Get samples

samples	
URL	BASE_URL/samples
Description	Returns a list of the samples in the system.
Method	GET
Example URL	https://<host>/genexus/api/lims/v2/samples

(continued)

samples	
Request query parameters	<ul style="list-style-type: none"> sampleId String. <i>Optional</i>. Sample name.
Example response	<pre>{ "total": 1, "objects": [{ "otherAttributes": {}, "id": 554, "sampleId": "Test_Sample1_127", "collectionDate": "2018-10-02", "gender": "Male", "sampleType": "DNA", "tumorCellularity": "70", "origin": "Non-Small Cell Lung Cancer", "cancerStage": "Stage 0", "diseaseCategory": "Solid Tumor" }] }</pre>

Get library batches

library-batches	
URL	BASE_URL/library-batches
Description	Returns a list of the library batches in the system.
Method	GET
Example URL	https://<host>/genexus/api/lims/v2/library-batches

(continued)

library-batches	
Request query parameters	<ul style="list-style-type: none"> id String. <i>Required.</i> Library batch ID.
Example response	<pre>{ "total": 1, "objects": [{ "libraries": [{ "libraryType": "DNA", "barcodeID": "IonHDDual_0102", "libraryName": "NTC_DNAOPA_DNA_Fusion_Library_Bacth_013", "inputQuantity": 3.0, "sampleId": "NTC", "tumorCellularity": null, "controlKitBarcode": "9110008037510224548317211230", "extractionKitBarcode": null }, { "libraryType": "RNA", "barcodeID": "IonHDDual_0104", "libraryName": "NTC_RNAOPA_DNA_Fusion_Library_Bacth_014", "inputQuantity": 3.0, "sampleId": "NTC", "tumorCellularity": null, "controlKitBarcode": "9110008037510224548317211230", "extractionKitBarcode": null }, { "libraryType": "DNA", "barcodeID": "IonHDDual_0101", "libraryName": "DNA_01", "inputQuantity": 3.0, "sampleId": "Test_Sample11_218", "tumorCellularity": null, "controlKitBarcode": null, "extractionKitBarcode": "9110009055910123456717211019" }, { "libraryType": "RNA", "barcodeID": "IonHDDual_0103", "libraryName": "RNA_01", "inputQuantity": 3.0, "sampleId": "Test_Sample11_218", "tumorCellularity": null, "controlKitBarcode": null, "extractionKitBarcode": "9110009056010123456717211231" }], "id": 233, "assayName": "Oncomine Precision - GX5 - Solid Tumor - DNA and Fusions - w3.0.0", "libraryBatchId": "OPA_DNA_Fusion_Library_Bacth_01", "libraryKitBarcode": "", "panelKitBarcode": "", "libraryPrepType": "DNA+RNA", "ampliSeqHd": false }] }</pre>



Get run plans

plans	
URL	BASE_URL/plans
Description	Returns a list of available run plans in the system.
Method	GET
Example URL	https://<host>/genexus/api/lims/v2/plans
Request query parameters	<ul style="list-style-type: none"> planName String. <i>Optional</i>. Name of the run plan. viewType String. <i>Optional</i>. Valid values are "all" or "toBeProcessed".
Example response	<pre>{ "objects": [{ "id": 184, "planName": "OPA_DNA_Fusion_PlanRun_01", "planShortId": "4RSKR", "type": "sample", "createdBy": "System Installed", "updatedOn": "2021-07-22T06:08:03.000+0000", "noOfSamples": 2, "noOfLibraries": 4, "samples": [{ "id": 973, "sampleId": "NTC", "assayName": "Oncomine Precision - GX5 - Solid Tumor - DNA and Fusions - w3.0.0", "dnaBarcodes": [], "rnaBarcodes": [], "libraryBatchId": "234", "libraryPrepType": "DNA+RNA", "pauseForReview": false }, { "id": 972, "sampleId": "Test_Sample11_278", "assayName": "Oncomine Precision - GX5 - Solid Tumor - DNA and Fusions - w3.0.0", "dnaBarcodes": [], "rnaBarcodes": [], "libraryBatchId": "234", "libraryPrepType": "DNA+RNA", "pauseForReview": false }] }, { "runAssays": ["Oncomine Precision - GX5 - Solid Tumor - DNA and Fusions - w3.0.0"] }], "total": 1 }</pre>



Get results

results	
URL	BASE_URL/results
Description	Returns results, including plugins and plugin results. Returns a JSON object for each plugin. All plugins for the run and the sample are returned.
Method	GET
Example URL	https://<host>/genexus/api/lims/v2/results



(continued)

results	
Request query parameters	<ul style="list-style-type: none">• <code>id</code> String. <i>Optional</i>. Results for the selected result id. Example: 123• <code>assay_id</code> String. <i>Optional</i>. Results for the assay id. Example: 123• <code>plan_id</code> String. <i>Optional</i>. Results for selected run plan id. Example: 123• <code>sampleId</code> String. <i>Optional</i>. Results for the selected sample name. Example: MySample• <code>planName</code> String. <i>Optional</i>. Results for selected run plan name. Example: MyPlan• <code>analysisComplete</code> String. <i>Optional</i>. Results for completed runs. Results included for failed and successfully completed runs. Example: true• <code>updatedBefore</code> String. <i>Optional</i>. Returns results updated before the selected date. Use date format: <code>yyyy-MM-dd HH:MM</code>. Example: 2019-01-15 10:30• <code>updatedAfter</code> String. <i>Optional</i>. Returns results updated after the selected date. Use date format: <code>yyyy-MM-dd</code> Example: 2019-01-15• <code>orderBy</code> String. <i>Optional</i>. Parameter to sort results by selected field. The default sort is by <code>updatedBefore</code> or <code>updatedAfter</code>. Results are listed in descending order, with the most recent results listed first.

Example response for get results

```
{
  "objects": [
    {
      "id": 356,
      "planName": "Gx516-Gx516",
      "sampleId": "pv_vv_gx-gx_ocav3_1",
      "assay": {
        "id": 95,
        "value": "Oncomine Comprehensive v3 - GX5 - DNA"
      },
      "version": "w5.0.0",
      "accountType": null,
      "isGreaterVersion": false,
      "runType": null,
      "greaterVersion": false,
      "planRun": {
        "id": 42,
        "value": "Gx516-Gx516"
      },
      "accountType": null,
      "isGreaterVersion": false,
      "runType": null,
      "greaterVersion": false,
      "sample": {
        "id": 248,
        "value": "pv_vv_gx-gx_ocav3_1",
        "accountType": null,
        "isGreaterVersion": false,
        "runType": null,
        "greaterVersion": false,
        "planShortCode": "L7VEN",
        "resultName": "Auto_R_2019_05_30_15_16_21_ionservice_VB12-15-Int1_R149696_OCAv3_DNA-RNA_DNA15TA10LA30uL_1.1.6_42_356",
        "libraryNames": "pv_vv_gx-gx_ocav3_1_DNA561, pv_vv_gx-gx_ocav3_1_DNA562",
        "runStartDate": "2021-06-28T00:00-0400",
        "runCompleteDate": "2021-07-11T06:20-0400",
        "updatedAt": "2021-07-11T06:27-0400",
        "libraryprepId": 388,
        "analysisComplete": true,
        "analysisVersion": "6.5.25",
        "isThumbnail": false,
        "isArchived": false,
        "isReanalysis": false,
        "runStatus": {
          "id": null,
          "name": "Analysis",
          "value": "Completed"
        },
        "qcStatus": "Failed",
        "instrumentName": "TestRig",
        "outputsLink": "/analysis_output/Auto_R_2019_05_30_15_16_21_ionservice_VB12-15-Int1_R149696_OCAv3_DNA-RNA_DNA15TA10LA30uL_1.1.6_42/Auto_R_2019_05_30_15_16_21_ionservice_VB12-15-Int1_R149696_OCAv3_DNA-RNA_DNA15TA10LA30uL_1.1.6_42_20210628082804990/ChipLane1/pv_vv_gx-gx_ocav3_1_LibPrep388/outputs",
        "outputsDir": "/serverdata/AssayDevdata/IR/data/analysis_output/Auto_R_2019_05_30_15_16_21_ionservice_VB12-15-Int1_R149696_OCAv3_DNA-RNA_DNA15TA10LA30uL_1.1.6_42/Auto_R_2019_05_30_15_16_21_ionservice_VB12-15-Int1_R149696_OCAv3_DNA-RNA_DNA15TA10LA30uL_1.1.6_42_20210628082804990/ChipLane1/pv_vv_gx-gx_ocav3_1_LibPrep388/outputs",
        "loadingMetrics": [
          {
            "name": "Total Addressable Wells",
            "count": 2550093
          },
          {
            "name": "Wells With ISPs",
            "count": 1733428,
            "percentage": 67.98
          },
          {
            "name": "Live ISPs",
            "count": 1705332,
            "percentage": 98.38
          },
          {
            "name": "Control ISPs",
            "count": 444,
            "percentage": 0.03
          },
          {
            "name": "Library ISPs",
            "count": 1704888,
            "percentage": 99.97
          }
        ],
        "filteringMetrics": [
          {
            "name": "Filtered: Polyclonal",
            "count": 717991,
            "percentage": 42.11
          },
          {
            "name": "Filtered: Primer-Dimer",
            "count": 59986,
            "percentage": 3.52
          },
          {
            "name": "Filtered: Low Quality",
            "count": 92384,
            "percentage": 5.42
          },
          {
            "name": "Usable Library Reads",
            "count": 834527,
            "percentage": 48.95
          },
          {
            "name": "Final Reads",
            "count": 834527
          }
        ],
        "runQCMetrics": [
          {
            "data": {
              "metricName": "Key Signal",
              "value": "40",
              "referenceRange": "Not Set",
              "qcStatus": "Not Calculated",
              "percentValue": false,
              "iscosmic": false,
              "isPQControl": false,
              "uniqueName": "RunQC_KeySignal",
              "isDisplayMetric": false,
              "isDisplayQc": false,
              "libraryType": "NA",
              "dataType": "INT",
              "pqcontrol": false,
              "displayMetric": false,
              "displayQc": false
            },
            "metricName": "Percent Loading",
            "value": "68",
            "referenceRange": "Not Set",
            "qcStatus": "Not Calculated",
            "percentValue": true,
            "iscosmic": false,
            "isPQControl": false,
            "uniqueName": "RunQC_PercentLoading",
            "isDisplayMetric": false,
            "isDisplayQc": false,
            "libraryType": "NA",
            "dataType": "FLOAT",
            "pqcontrol": false,
            "displayMetric": false,
            "displayQc": false
          },
          {
            "data": {
              "metricName": "Raw Read Accuracy",
              "value": "98.6",
              "referenceRange": "Not Set",
              "qcStatus": "Not Calculated",
              "percentValue": false,
              "iscosmic": false,
              "isPQControl": false,
              "uniqueName": "RunQC_RawReadAccuracy",
              "isDisplayMetric": false,
              "isDisplayQc": false,
              "libraryType": "NA",
              "dataType": "FLOAT",
              "pqcontrol": false,
              "displayMetric": false,
              "displayQc": false
            },
            "metricName": "Raw Read Accuracy",
            "value": "98.6",
            "referenceRange": "Not Set",
            "qcStatus": "Not Calculated",
            "percentValue": false,
            "iscosmic": false,
            "isPQControl": false,
            "uniqueName": "RunQC_RawReadAccuracy",
            "isDisplayMetric": false,
            "isDisplayQc": false,
            "libraryType": "NA",
            "dataType": "FLOAT",
            "pqcontrol": false,
            "displayMetric": false,
            "displayQc": false
          }
        ],
        "sequence": 0,
        "localizationKey": "RunQC",
        "inlineControlQc": false,
        "customApp": "",
        "images": []
      },
      "purificationQCMetrics": [
        {
          "title": "CF-1",
          "data": [
            {
              "metricName": "Average Reads Per Lane",
              "value": "0",
              "referenceRange": "Not Set",
              "qcStatus": "Not Calculated",
              "percentValue": false,
              "iscosmic": false,
              "isPQControl": false,
              "uniqueName": "InternalControlQC_AvgReads_PerLane_DNA",
              "isDisplayMetric": false,
              "isDis"
            }
          ]
        }
      ]
    }
  ]
}
```



```

playQc":false,"libraryType":"NA","dataType":"FLOAT","pqcontrol":false,"displayMetric":false,"displayQc":false},{"metricName":"Base Call Accuracy","value":"0","referenceRange":"Not Set","qcStatus":"Not Calculated","percentValue":true,"iscosmic":false,"isPQControl":false,"uniqueName":"InternalControlQC_BaseCallAccuracy_DNA","isDisplayMetric":false,"isDisplayQc":false,"libraryType":"NA","dataType":"FLOAT","pqcontrol":false,"displayMetric":false,"displayQc":false},{"metricName":"Mean AQ20 Read Length (bp)","value":"0","referenceRange":"Not Set","qcStatus":"Not Calculated","percentValue":false,"iscosmic":false,"isPQControl":false,"uniqueName":"InternalControlQC_MeanAQ20ReadLength_DNA","isDisplayMetric":false,"isDisplayQc":false,"libraryType":"NA","dataType":"INT","pqcontrol":false,"displayMetric":false,"displayQc":false}],{"sequence":0,"localizationKey":"CF1","inlineControlQc":false,"customApp":"","libraryType":"NA","images":[]},"sampleQCmetrics":[{"title":"DNA","data":[{"metricName":"MAPD","value":"0.91","referenceRange":"<=0.5","qcStatus":"Failed","percentValue":false,"iscosmic":false,"isPQControl":false,"samples":{"pv_vv_gx-gx_ocav3_1":{"value":"0.91","qcStatus":"Failed"}},"uniqueName":"SampleQC_MAPD_DNA","isDisplayMetric":false,"isDisplayQc":false,"libraryType":"DNA","dataType":"FLOAT","pqcontrol":false,"displayMetric":false,"displayQc":false},{"metricName":"Mapped Reads","value":"138678","referenceRange":"Not Set","qcStatus":"Not Calculated","percentValue":false,"iscosmic":false,"isPQControl":false,"samples":{"pv_vv_gx-gx_ocav3_1":{"value":"138678","qcStatus":"Not Calculated"}},"uniqueName":"SampleQC_MappedReads_DNA","isDisplayMetric":false,"isDisplayQc":false,"libraryType":"DNA","dataType":"INT","pqcontrol":false,"displayMetric":false,"displayQc":false},{"metricName":"Mean AQ20 Read Length (bp)","value":"92","referenceRange":"Not Set","qcStatus":"Not Calculated","percentValue":false,"iscosmic":false,"isPQControl":false,"samples":{"pv_vv_gx-gx_ocav3_1":{"value":"92","qcStatus":"Not Calculated"}},"uniqueName":"SampleQC_MeanAQ20ReadLength_DNA","isDisplayMetric":false,"isDisplayQc":false,"libraryType":"DNA","dataType":"INT","pqcontrol":false,"displayMetric":false,"displayQc":false},{"metricName":"Mean Read Length (bp)","value":"110","referenceRange":"Not Set","qcStatus":"Not Calculated","percentValue":false,"iscosmic":false,"isPQControl":false,"samples":{"pv_vv_gx-gx_ocav3_1":{"value":"110","qcStatus":"Not Calculated"}},"uniqueName":"SampleQC_MeanReadLength_DNA","isDisplayMetric":false,"isDisplayQc":false,"libraryType":"DNA","dataType":"INT","pqcontrol":false,"displayMetric":false,"displayQc":false},{"metricName":"Uniformity Of Amplicon Coverage","value":"49.63","referenceRange":"Not Set","qcStatus":"Not Calculated","percentValue":true,"iscosmic":false,"isPQControl":false,"samples":{"pv_vv_gx-gx_ocav3_1":{"value":"49.63","qcStatus":"Not Calculated"}},"uniqueName":"SampleQC_UniformityAmpliconCoverage_DNA","isDisplayMetric":false,"isDisplayQc":false,"libraryType":"DNA","dataType":"FLOAT","pqcontrol":false,"displayMetric":false,"displayQc":false}],{"sequence":0,"localizationKey":"SampleQC","inlineControlQc":false,"customApp":"","libraryType":"DNA","images":[{"title":null,"filePath":"analysis_output/Auto_R_2019_05_30_15_16_21_ionservice_VB12-15-Int1_R149696_OCAv3_DNA-RNA_DNA15TA10LA30uL_1.1.6_42/Auto_R_2019_05_30_15_16_21_ionservice_VB12-15-Int1_R149696_OCAv3_DNA-RNA_DNA15TA10LA30uL_1.1.6_42_20210628082804990/ChipLane1/outputs/BaseCallingActor-00/IonDual_0101-IonDual_0102_rawlib.read_len_histogram.png","status":true,"type":""}, {"title":null,"filePath":"analysis_output/Auto_R_2019_05_30_15_16_21_ionservice_VB12-15-Int1_R149696_OCAv3_DNA-RNA_DNA15TA10LA30uL_1.1.6_42/Auto_R_2019_05_30_15_16_21_ionservice_VB12-15-Int1_R149696_OCAv3_DNA-RNA_DNA15TA10LA30uL_1.1.6_42_20210628082804990/ChipLane1/outputs/BaseCallingActor-00/IonDual_0101-IonDual_0102_rawlib.inline_control.png","status":true,"type":"inlineControl"}]}],"sampleInlineControlQCmetrics":[],"ntcQCmetrics":[],"plugins":[{"id":518,"pluginName":"CustomerSupportArchive","pluginVersion":"0.5.9","upd

```

```
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```



```
RNA_DNA15TA10LA30uL_1.1.6_42/Auto_R_2019_05_30_15_16_21_ionservice_VB12-15-Int1_R149696_OCAv3_DNA-RNA_DNA15TA10LA30uL_1.1.6_42_20210628082804990/ChipLane1/plugins/IonReporterUploader_3.0/1625476344547", "runType": "ASSAY"}, {"id": 1097, "pluginName": "IonReporterUploader", "pluginVersion": "3.0", "updateOn": "2021-07-05T09:23:05.000+0000", "status": "COMPLETED", "path": "/serverdata/AssayDevdata/IR/data/analysis_output/Auto_R_2019_05_30_15_16_21_ionservice_VB12-15-Int1_R149696_OCAv3_DNA-RNA_DNA15TA10LA30uL_1.1.6_42/Auto_R_2019_05_30_15_16_21_ionservice_VB12-15-Int1_R149696_OCAv3_DNA-RNA_DNA15TA10LA30uL_1.1.6_42_20210628082804990/ChipLane1/plugins/IonReporterUploader_3.0/1625476984593", "runType": "ASSAY"}, {"id": 1100, "pluginName": "IonReporterUploader", "pluginVersion": "3.0", "updateOn": "2021-07-05T09:58:00.000+0000", "status": "COMPLETED", "path": "/serverdata/AssayDevdata/IR/data/analysis_output/Auto_R_2019_05_30_15_16_21_ionservice_VB12-15-Int1_R149696_OCAv3_DNA-RNA_DNA15TA10LA30uL_1.1.6_42/Auto_R_2019_05_30_15_16_21_ionservice_VB12-15-Int1_R149696_OCAv3_DNA-RNA_DNA15TA10LA30uL_1.1.6_42_20210628082804990/ChipLane1/plugins/IonReporterUploader_3.0/1625479079891", "runType": "ASSAY"}, {"id": 1111, "pluginName": "IonReporterUploader", "pluginVersion": "3.0", "updateOn": "2021-07-06T09:30:24.000+0000", "status": "COMPLETED", "path": "/serverdata/AssayDevdata/IR/data/analysis_output/Auto_R_2019_05_30_15_16_21_ionservice_VB12-15-Int1_R149696_OCAv3_DNA-RNA_DNA15TA10LA30uL_1.1.6_42/Auto_R_2019_05_30_15_16_21_ionservice_VB12-15-Int1_R149696_OCAv3_DNA-RNA_DNA15TA10LA30uL_1.1.6_42_20210628082804990/ChipLane1/plugins/IonReporterUploader_3.0/1625563823609", "runType": "ASSAY"}, {"id": 1120, "pluginName": "IonReporterUploader", "pluginVersion": "3.0", "updateOn": "2021-07-06T21:33:23.000+0000", "status": "COMPLETED", "path": "/serverdata/AssayDevdata/IR/data/analysis_output/Auto_R_2019_05_30_15_16_21_ionservice_VB12-15-Int1_R149696_OCAv3_DNA-RNA_DNA15TA10LA30uL_1.1.6_42/Auto_R_2019_05_30_15_16_21_ionservice_VB12-15-Int1_R149696_OCAv3_DNA-RNA_DNA15TA10LA30uL_1.1.6_42_20210628082804990/ChipLane1/plugins/IonReporterUploader_3.0/1625607203138", "runType": "ASSAY"}, {"id": 1512, "pluginName": "IonReporterUploader", "pluginVersion": "3.0", "updateOn": "2021-07-11T10:14:26.000+0000", "status": "COMPLETED", "path": "/serverdata/AssayDevdata/IR/data/analysis_output/Auto_R_2019_05_30_15_16_21_ionservice_VB12-15-Int1_R149696_OCAv3_DNA-RNA_DNA15TA10LA30uL_1.1.6_42/Auto_R_2019_05_30_15_16_21_ionservice_VB12-15-Int1_R149696_OCAv3_DNA-RNA_DNA15TA10LA30uL_1.1.6_42_20210628082804990/ChipLane1/plugins/IonReporterUploader_3.0/1625998465941", "runType": "ASSAY"}, {"id": 1515, "pluginName": "IonReporterUploader", "pluginVersion": "3.0", "updateOn": "2021-07-11T10:20:53.000+0000", "status": "COMPLETED", "path": "/serverdata/AssayDevdata/IR/data/analysis_output/Auto_R_2019_05_30_15_16_21_ionservice_VB12-15-Int1_R149696_OCAv3_DNA-RNA_DNA15TA10LA30uL_1.1.6_42/Auto_R_2019_05_30_15_16_21_ionservice_VB12-15-Int1_R149696_OCAv3_DNA-RNA_DNA15TA10LA30uL_1.1.6_42_20210628082804990/ChipLane1/plugins/IonReporterUploader_3.0/1625998853257", "runType": "ASSAY"}, {"id": 1509, "pluginName": "IonReporterUploader", "pluginVersion": "3.0", "updateOn": "2021-07-11T10:06:50.000+0000", "status": "COMPLETED", "path": "/serverdata/AssayDevdata/IR/data/analysis_output/Auto_R_2019_05_30_15_16_21_ionservice_VB12-15-Int1_R149696_OCAv3_DNA-RNA_DNA15TA10LA30uL_1.1.6_42/Auto_R_2019_05_30_15_16_21_ionservice_VB12-15-Int1_R149696_OCAv3_DNA-RNA_DNA15TA10LA30uL_1.1.6_42_20210628082804990/ChipLane1/plugins/IonReporterUploader_3.0/1625998009550", "runType": "ASSAY"}] ] ] ], "total": 1 }
```

Get signed off samples

signedOffSamples	
URL	BASE_URL/signedOffSamples
Description	Returns a list of the samples that are designated with <i>(Signed Off)</i> .
Method	GET
Example URL	https://<host>/genexus/api/lims/v2/signedOffSamples
Request query parameters	<ul style="list-style-type: none"> • start_date String, <i>Optional</i>. Provide the start date as yyyy-mm-dd. • end_date String, <i>Optional</i>. Provide the end date as yyyy-mm-dd. • signedOff String, <i>Required</i> A Boolean data type. Valid values are "true" or a "false". The default value is "true".
Example response	<pre>{ "meta": { "pageSize": 20, "next": null, "pageNumber": 0, "previous": null, "total_count": 76 }, "objects": [{ "sample": { "assay_name": "qualification_ampliseq_genexus_pq_template516_4.0.0", "base_url": "AssayDev_PQSample1_qualification_ampliseq_genexus_pq_template516_4.0.0_A9GHS_A9GHS", "checksum_file": "CHECKSUM", "created_date": "2021-06-23", "sample_file": "Info.csv", "sample_id": "92", "sample_name": "PQSample1", "planName": "IQ_Lib" } }, { "sample": { "assay_name": "qualification_ampliseq_genexus_pq_template516_4.0.0", "base_url": "AssayDev_PQSample2_qualification_ampliseq_genexus_pq_template516_4.0.0_A9GHS_A9GHS", "checksum_file": "CHECKSUM", "created_date": "2021-06-23", "sample_file": "Info.csv", "sample_id": "93", "sample_name": "PQSample2", "planName": "IQ_Lib" } }] }</pre>



Download files for completed runs

download	
URL	BASE_URL/download
Description	Returns a compressed file in ZIP format that contains the requested files for the sample.
Method	GET
Example URL	https://<host>/genexus/api/lims/v2/download
Request query parameters	<ul style="list-style-type: none"> file_list String. <i>Required.</i> Provide the file name in comma separated format. path String, <i>Required.</i> Provide the LIMS folder path of the analyzed sample. You can request the folder path when you get signed off samples. The folder path is listed as the "base_URL". For more information, see "Get signed off samples" on page 333.
Example request	<ol style="list-style-type: none"> 1. Perform a checksum function to query for the list of files for a sample that has been sequenced and analyzed. <pre>https://BASE_URL/download? file_list=CHECKSUM&path=AssayDev_Sample_C1_qualification_ampliseq_Genexus_pq_template516_1.3.2_3MI8Q_3MI8Q</pre> 2. Parse the checksum to get the file name, then request that file. <pre>https://BASE_URL/download? file_list=Info.csv,BaseCaller.json,analysis.log&path=AssayDev_Sample_C1_qualification_ampliseq_Genexus_pq_template516_1.3.2_3MI8Q_3MI8Q</pre> 3. Ensure that the name of the downloaded file matches the checksum requested in step 1.

Create samples

samples	
URL	BASE_URL/samples
Description	Create 1 to <i>n</i> samples. "All" or "None" is followed for all samples in a request. If the creation of any one sample fails, creation of the entire batch fails.
Method	POST
Example URL	https://<host>/genexus/api/lims/v2/samples

(continued)

samples	
Attribute definitions	<ul style="list-style-type: none"> • <code>sampleId</code>. String. Required. Sample name. • <code>sampleType</code>. String. Required. Examples of valid values are "DNA" or "RNA". • <code>collectionDate</code>. String. Required. The date that the sample was collected. • <code>gender</code>. String. Required. The biological sex of the sample: Female, Male, or Unknown. • <code>diseaseCategory</code>. String. Required. The sample application category. • <code>cancerStage</code>. String. Required. Cancer stage. • <code>origin</code>. String. Required. Cancer type. • <code>notes</code>. String. Required. An open-entry field for more sample information. • <code>necrosis</code>. String. Optional. The percentage of cellular necrosis in the sample. • <code>tumorCellularity</code>. String. Optional. The percentage of tumor cells over normal cells in the sample. • <code>condition</code>. String. Optional. The condition of the sample.
Example request	<pre>{ "objects":[{ "sampleId":"Sample_NSCLC_Tag_{{\${randomInt}}}", "diseaseCategory":"Solid Tumor", "sampleType":"DNA & RNA", "origin":"Non-Small Cell Lung Cancer", "cancerStage":"Unknown", "necrosis":100, "tumorCellularity":31, "gender":"Unknown", "collectionDate":"2018-10-02", "condition":"good", "tagDisplayName":["NSCLC"], "notes":"LIMS API created Cancer sample and existing Tag NSCLC" }] }</pre>



Create a library batch

library-batches	
URL	BASE_URL/library-batches
Description	Create a library batch from one or more existing samples. If any requested sample does not exist in the software, the creation of the entire library batch fails.
Method	POST
Example URL	https://<host>/genexus/api/lims/v2/library-batches
Example request	<pre>{ "objects": [{ "libraryBatchId": "LibBatch_DNA_NTC_01", "assayName": "Oncomine Comprehensive v3 - GX5 - DNA - w5.0.2", "libraryPrepType": "DNA", "libraryKitBarcode": "01ABC0012300000017221123102-321AB234240A40252", "panelKitBarcode": "01ABC0012300000017221123102-321AB234240A44351", "libraries": [{ "barcodeID": "IonDual_0101", "inputQuantity": 77.0, "libraryType": "DNA", "libraryName": "DNA_Lib1_99", "sampleId": "Test_Sample1_127", "extractionKitBarcode": "9110009055910123456717211019" }, { "barcodeID": "IonDual_0102", "inputQuantity": 77.0, "libraryType": "DNA", "libraryName": "DNA_Lib2_99", "sampleId": "Test_Sample1_127", "extractionKitBarcode": "9110009055910123456717211019" }, { "barcodeID": "IonDual_0103", "inputQuantity": 77.0, "libraryType": "DNA", "libraryName": "", "sampleId": "NTC", "controlKitBarcode": "9110008037510224548317211230" }, { "barcodeID": "IonDual_0104", "inputQuantity": 77.0, "libraryType": "DNA", "libraryName": "", "sampleId": "NTC", "controlKitBarcode": "9110008037510224548317211230" }] }] }</pre>

Create a Sample to Result (purification) run

plan-by-purification	
URL	BASE_URL/plan-by-purification
Description	Create a Sample to Result run from one or more samples and an assay. The sample or samples and the assay must exist in the system.
Method	POST
Example URL	https://<host>/genexus/api/lims/v2/plan-by-purification
Example request	<pre>{ "irAccountName": "iraccount", "reportTemplateName": "mytemplate", "samples": [{ "assayName": "Purification FFPE DNA and Fusion Assay", "sampleId": "Sample1", "purificationProtocol": "FFPE DNA + RNA", "elutionVolumeDNA": 50, "elutionVolumeRNA": 50, "pauseForReview": false }, { "assayName": "Purification DNA Assay", "sampleId": "Sample2", "purificationProtocol": "Blood 50-100 µl", "elutionVolumeDNA": 40, "pauseForReview": false }, { "assayName": "Purification DNA Assay", "sampleId": "Sample3", "purificationProtocol": "Blood 50-100 µl", "elutionVolumeDNA": 40, "pauseForReview": false }, { "assayName": "Purification RNA Assay", "sampleId": "Sample4", "purificationProtocol": "Biofluid", "elutionVolumeDNA": 90, "pauseForReview": false }, { "assayName": "Purification RNA Assay", "sampleId": "Sample5", "purificationProtocol": "Cells and tissue", "elutionVolumeDNA": 90, "pauseForReview": false }], "notes": "my notes", "planName": "plan_purification_run" }</pre>



Create a Nucleic Acid to Result run

plan-by-samples	
URL	BASE_URL/plan-by-samples
Description	Create a Nucleic Acid to Result run from one or more samples and an assay. The sample or samples and the assay must exist in the system.
Method	POST
Example URL	https://<host>/genexus/api/lims/v2/plan-by-samples
Example request	<pre>{ "irAccountName": "iraccount", "reportTemplateName": "mytemplate", "samples": [{ "assayName": "Generic Sequencing Ampliseq Assay", "sampleId": "Sample1" }, { "assayName": "Custom DNA Assay", "sampleId": "Sample2" }, { "assayName": "Custom DNA Assay", "sampleId": "Sample3" }], "notes": "my notes", "planName": "plan_sample_run" }</pre>

Create a library run

plan-by-library	
URL	BASE_URL/plan-by-library
Description	Create a library run from one or more library batches and an assay. The library batch or batches and the assay must exist in the system.
Method	POST
Example URL	https://<host>/genexus/api/lims/v2/plan-by-library
Example request	<pre>{ "irAccountName": "iraccount", "samples": [{ "assayName": "Generic Sequencing Ampliseq Assay", "libraryBatchId": "LibraryBatch1" }, { "assayName": "Custom DNA Assay", "libraryBatchId": "LibraryBatch2" }, { "assayName": "Custom DNA Assay", "libraryBatchId": "LibraryBatch3" }], "notes": "my notes", "planName": "plan_library_run" }</pre>



Reference information

- Annotation criteria for Onco[™] Variant Annotator 340
- Variant identifiers for Onco[™] panels 348
- System-installed annotation sets 349
- System-installed barcode sets 350
- System-installed filter chains 351
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Annotation criteria for Onco[™] Variant Annotator

In Genexus[™] Software, analysis results are the final results generated by a run analysis. The analysis results include run details, all BAM files, filtered results, graphs, run reports, metrics, plugin results, annotated variant data, and VCF files. When you apply Onco[™] variant annotations to an assay, analysis results data from more than 24,000 exomes across solid tumor and hematological cancer types is used as a reference to annotate variants that are relevant to cancer. For information about applying Onco[™] variant annotations to a custom assay, see “Panel step assay options” on page 197.

This section summarizes the requirements that must be met in order for the Onco[™] Variant Annotator to annotate variants in results for each Onco[™] Research Assay and Ion AmpliSeq[™] HD Panel.

For each variant type in these tables, Onco[™] Variant Annotator annotates a variant only if all conditions in the corresponding Annotation Criteria column are satisfied. The annotations are shown in results in the **Onco[™] Gene Class** and **Onco[™] Variant Class** columns.

Note: You can find all relevant annotation criteria in VCF files.

OncoPrint™ Comprehensive Assay v3 GX

Variant type	OncoPrint™ Gene class	OncoPrint™ Variant class	Annotation criteria
Copy number amplification	Gain-of-Function	Amplification	<ul style="list-style-type: none"> SVTYPE = "CNV" FILTER = "GAIN" Occurs in a designated copy-gain gene
Long deletion	Loss-of-Function	LongDel	<ul style="list-style-type: none"> Positive mutation call SVTYPE = "LongDel"
Gene fusion	Gain-of-Function	Fusion	<ul style="list-style-type: none"> SVTYPE = "Fusion" FILTER = "PASS" Is a targeted fusion isoform
RNA exon variant	Gain-of-Function	RNAExonVariant	<ul style="list-style-type: none"> SVTYPE = "RNAExonVariant" or "Fusion" FILTER = "PASS" Is a targeted RNA exon variant
Loss-of-function truncating de novo mutation	Loss-of-Function	Truncating	<ul style="list-style-type: none"> Positive mutation call Functional impact is frameshift block substitution, frameshift deletion, frameshift insertion, or nonsense Occurs in a loss-of-function gene
Missense hotspot mutation	Gain-of-Function Loss-of-Function	Hotspot	<ul style="list-style-type: none"> Positive mutation call Functional impact is missense Transcript and codon position occur in predefined missense hotspot list
In-frame hotspot mutation	Gain-of-Function Loss-of-Function	Hotspot	<ul style="list-style-type: none"> Positive mutation call Function, transcript, and coding syntax occur in predefined in-frame hotspot list
Splice site hotspot mutation	Gain-of-Function Loss-of-Function	Hotspot	<ul style="list-style-type: none"> Positive mutation call Transcript, location, and exon occur in predefined splice site hotspot list
Intronic hotspot mutation	Gain-of-Function Loss-of-Function	Hotspot	<ul style="list-style-type: none"> Positive mutation call Transcript, location, and coding syntax occur in predefined intronic hotspot list
Promoter hotspot mutation	Gain-of-Function Loss-of-Function	Hotspot	<ul style="list-style-type: none"> Positive mutation call Transcript, location, and coding syntax occur in predefined promoter hotspot list

(continued)

Variant type	OncoPrint™ Gene class	OncoPrint™ Variant class	Annotation criteria
Gain-of-function truncating hotspot mutation	Gain-of-Function	Hotspot	<ul style="list-style-type: none"> Positive mutation call Function, transcript, and coding syntax occur in predefined truncating hotspot list Occurs in a gain-of-function gene
Loss-of-function truncating hotspot mutation	Loss-of-Function	Truncating	<ul style="list-style-type: none"> Positive mutation call Function, transcript, and coding syntax occur in predefined truncating hotspot list Occurs in a loss-of-function gene
MNV hotspot mutation	Gain-of-Function Loss-of-Function	Hotspot	<ul style="list-style-type: none"> Positive mutation call Transcript and coding syntax occur in MNV hotspot list
EGFR exon 19 deletion ^[1]	Gain-of-Function	EGFRExon19Deletion	<ul style="list-style-type: none"> Positive mutation call Functional impact is nonframeshift deletion, nonframeshift block substitution Deletion impacts codons 744-761 of EGFR
EGFR exon 20 insertion ^[1]	Gain-of-Function	EGFRExon20Insertion	<ul style="list-style-type: none"> Positive mutation call Functional impact is nonframeshift insertion, nonframeshift block substitution Insertion impacts codons 762-775 of EGFR or variant is COSM26720
ERBB2 exon 20 insertion ^[1]	Gain-of-Function	ERBB2Exon20Insertion	<ul style="list-style-type: none"> Positive mutation call Functional impact is nonframeshift insertion, nonframeshift block substitution Insertion impacts codons 770-783 of ERBB2
MET exon 14 skipping ^[1]	Gain-of-Function	METExon14Skipping	<ul style="list-style-type: none"> Positive mutation call Location is splice site in MET exon 14, is intronic >= 4bp deletion in 30 nucleotides preceding MET exon 14, or variant is in MET Exon 14 Skipping confirmed list

^[1] For more information, see "Ion Torrent™ Class-Based Variants" on page 346.

OncoPrint™ Myeloid Assay GX v2

Variant type	OncoPrint™ Gene class	OncoPrint™ Variant class	Annotation criteria
Gene fusion	Gain-of-Function	Fusion	<ul style="list-style-type: none"> SVTYPE = "Fusion" FILTER = "PASS" Is a targeted fusion isoform
RNA exon variant	Gain-of-Function	RNAExonVariant	<ul style="list-style-type: none"> SVTYPE = "RNAExonVariant" or "Fusion" FILTER = "PASS" Is a targeted RNA exon variant
Loss-of-function truncating de novo mutation	Loss-of-Function	Truncating	<ul style="list-style-type: none"> Positive mutation call Functional impact is frameshift block substitution, frameshift deletion, frameshift insertion, or nonsense Occurs in a loss-of-function gene
Missense hotspot mutation	Gain-of-Function Loss-of-Function	Hotspot	<ul style="list-style-type: none"> Positive mutation call Functional impact is missense Transcript and codon position occur in predefined missense hotspot list
In-frame hotspot mutation	Gain-of-Function Loss-of-Function	Hotspot	<ul style="list-style-type: none"> Positive mutation call Function, transcript, and coding syntax occur in predefined in-frame hotspot list
Splice site hotspot mutation	Gain-of-Function Loss-of-Function	Hotspot	<ul style="list-style-type: none"> Positive mutation call Transcript, location, and exon occur in predefined splice site hotspot list
Promoter hotspot mutation	Gain-of-Function Loss-of-Function	Hotspot	<ul style="list-style-type: none"> Positive mutation call Transcript, location, and coding syntax occur in predefined promoter hotspot list
Gain-of-function truncating hotspot mutation	Gain-of-Function	Hotspot	<ul style="list-style-type: none"> Positive mutation call Function, transcript, and coding syntax occur in predefined truncating hotspot list Occurs in a gain-of-function gene
Loss-of-function truncating hotspot mutation	Loss-of-Function	Truncating	<ul style="list-style-type: none"> Positive mutation call Function, transcript, and coding syntax occur in predefined truncating hotspot list Occurs in a loss-of-function gene
MNV hotspot mutation	Gain-of-Function Loss-of-Function	Hotspot	<ul style="list-style-type: none"> Positive mutation call Transcript and coding syntax occur in MNV hotspot list

(continued)

Variant type	OncoPrint™ Gene class	OncoPrint™ Variant class	Annotation criteria
FLT3 internal tandem duplication	Gain-of-Function	FLT3ITD	<ul style="list-style-type: none"> Positive mutation call Functional impact is a non-frameshift insertion in exon 14/15 of FLT3 or SVTYPE = "FLT3ITD"

OncoPrint™ Precision Assay GX

Variant type	OncoPrint™ Gene class	OncoPrint™ Variant class	Annotation criteria
Copy number amplification	Gain-of-Function	Amplification	<ul style="list-style-type: none"> SVTYPE = "CNV" FILTER = "GAIN" Occurs in a designated copy-gain gene
Copy number deletion	Loss-of-Function	Deletion	<ul style="list-style-type: none"> SVTYPE = "CNV" FILTER = "LOSS" Occurs in a designated copy-loss gene
Gene fusion	Gain-of-Function	Fusion	<ul style="list-style-type: none"> SVTYPE = "Fusion" FILTER = "PASS" Is a targeted fusion isoform
RNA exon variant	Gain-of-Function	RNAExonVariant	<ul style="list-style-type: none"> SVTYPE = "RNAExonVariant" or "Fusion" FILTER = "PASS" Is a targeted RNA exon variant
Expression imbalance	Gain-of-Function	ExpressionImbalance	<ul style="list-style-type: none"> SVTYPE = "RNAExonTiles" FILTER = "PASS" Record meets Targeted Isoforms Detected Requirement
Missense hotspot mutation	Gain-of-Function Loss-of-Function	Hotspot	<ul style="list-style-type: none"> Positive mutation call Functional impact is missense Transcript and coding syntax occur in predefined missense hotspot list
In-frame hotspot mutation	Gain-of-Function Loss-of-Function	Hotspot	<ul style="list-style-type: none"> Positive mutation call Function, transcript, and coding syntax occur in predefined in-frame hotspot list
Splice site hotspot mutation	Gain-of-Function Loss-of-Function	Hotspot	<ul style="list-style-type: none"> Positive mutation call Transcript, location, and exon occur in predefined splice site hotspot list

(continued)

Variant type	OncoPrint™ Gene class	OncoPrint™ Variant class	Annotation criteria
Intronic hotspot mutation	Gain-of-Function Loss-of-Function	Hotspot	<ul style="list-style-type: none"> Positive mutation call Transcript, location, and coding syntax occur in predefined intronic hotspot list
Gain-of-function truncating hotspot mutation	Gain-of-Function	Hotspot	<ul style="list-style-type: none"> Positive mutation call Function, transcript, and coding syntax occur in predefined truncating hotspot list Occurs in a gain-of-function gene
Loss-of-function truncating hotspot mutation	Loss-of-Function	Truncating	<ul style="list-style-type: none"> Positive mutation call Function, transcript, and coding syntax occur in predefined truncating hotspot list Occurs in a loss-of-function gene
MNV hotspot mutation	Gain-of-Function Loss-of-Function	Hotspot	<ul style="list-style-type: none"> Positive mutation call Transcript and coding syntax occur in MNV hotspot list
EGFR exon 19 deletion ^[1]	Gain-of-Function	EGFRExon19Deletion	<ul style="list-style-type: none"> Positive Hotspot mutation call Functional impact is nonframeshift deletion, nonframeshift block substitution Deletion impacts codons 744–761 of EGFR
EGFR exon 20 insertion ^[1]	Gain-of-Function	EGFRExon20Insertion	<ul style="list-style-type: none"> Positive Hotspot mutation call Functional impact is nonframeshift insertion, nonframeshift block substitution Insertion impacts codons 762–775 of EGFR or variant is COSM26720
ERBB2 exon 20 insertion ^[1]	Gain-of-Function	ERBB2Exon20Insertion	<ul style="list-style-type: none"> Positive Hotspot mutation call Functional impact is nonframeshift insertion, nonframeshift block substitution Insertion impacts codons 770–783 of ERBB2
MET exon 14 skipping ^[1]	Gain-of-Function	METExon14Skipping	<ul style="list-style-type: none"> Positive Hotspot mutation call Location is splice site in MET exon 14, is intronic \geq 4bp deletion in 30 nucleotides preceding MET exon 14, or variant is in MET Exon 14 Skipping confirmed list

^[1] For more information, see “Ion Torrent™ Class-Based Variants” on page 346.

Ion Torrent™ Class-Based Variants

Ion Torrent™ Class-Based Variants are a set of logic-based rules that Genexus™ Software uses to computationally identify and annotate novel mutations in important genomic variant classes including EGFR exon 19 deletions, EGFR exon 20 insertions, ERBB2 exon 20 insertions, and MET exon 14 skipping mutations. The annotation logic is based on scientific literature evidence. Details about the logic follow.

EGFR exon 19 deletion

- **Background:** Epidermal growth factor receptor (EGFR) is a transmembrane receptor protein made up of 1210 amino acids and located on chromosome 7p11.2. EGFR has 31 exons— exons 2-4 and 8-12 encode the ligand binding domains, exons 5-7 and 13-16 encode the cysteine rich domain, exons 18–24 encode the tyrosine kinase domain (TKD), and exons 25-28 encode the autophosphorylation region [PMID: 17318210]. Mutations in EGFR often target the TKD which make up the ATP-binding pocket of the enzyme [PMID: 17318210]. Exon 19 deletions are indels that range between 15-18 bp in length and usually occur within codons 746-756 [PMID: 23768755, PMID: 22190593, PMID: 26933124, PMID: 24163741]. In non-small cell lung cancer (NSCLC), EGFR exon 19 deletions, located within codons 746-750, represent 45-50% of somatic mutations [PMID: 26933124, PMID: 24163741]. Exon 19 deletions result in constitutive activation of the receptor tyrosine kinase and hyperactivation of downstream signaling pathways [PMID: 29455648]. Exon 19 deletions increase sensitivity to EGFR tyrosine kinase inhibitors such as erlotinib [FDA-erlotinib], gefitinib [FDA-gefitinib], afatinib [FDA-afatinib], and osimertinib [FDA-osimertinib]. It has been observed that both common and rare EGFR exon 19 deletions exhibit similar responses to EGFR TKI's [PMID: 23768755].
- **CBV Criteria:** The boundaries defined to capture EGFR exon 19 deletions as a class-based variant start at codon 744 and end at codon 761, which encompass common and rare indels [PMID: 30473385]. Codon 744 captures a unique indel found on the N-terminal region of EGFR (p.I744_E749>LKR) that is associated with erlotinib sensitivity [PMID: 15912202]. Classifying the EGFR exon 19 boundary to codon 761 boundary captures a rare indel (p.S752-I759) located in the C-terminal region of the EGFR exon 19, that is associated with sensitivity to gefitinib and erlotinib [PMID: 17368623, PMID: 19057270].

EGFR exon 20 insertion

- Background:** Epidermal growth factor receptor (EGFR) is a transmembrane receptor protein made up of 1210 amino acids located on chromosome 7p11.2. EGFR has 31 exons— exons 2-4 and 8-12 encode the ligand binding domains, exons 5-7 and 13-16 encode the cysteine rich domain, exons 18-24 encode the tyrosine kinase domain (TKD), and exons 25-28 encode the autophosphorylation region [PMID: 17318210]. Mutations in EGFR often target the TKD which make up the ATP-binding pocket of the enzyme [PMID: 17318210]. In-frame insertions within exon 20 of EGFR are the third most common type of mutation found in NSCLC, representing 4-12% of all EGFR mutations in NSCLC [PMID: 31208370]. EGFR exon 20 encompasses codons 762 to 823. Exon 20 insertions commonly involve codons 762 to 774 wherein codons 762-766 make up the c-helix and codons 767-774 make up the activation loop [PMID: 31208370, PMID: 30854234]. EGFR exon 20 insertions do not alter the receptor binding affinity; however, it is suggested that the location of exon 20 insertions stabilizes its active conformation [PMID: 30854234, PMID: 27843613]. This stabilization of the ATP receptor site leads to resistance to EGFR tyrosine kinase inhibitors [PMID: 27843613]. Tumors harboring EGFR exon 20 insertion mutations involving codons 767, 768, 770, 772 and 773 display decreased sensitivity to gefitinib and erlotinib [PMID: 29551130, PMID: 29387949].
- CBV Criteria:** The boundaries defined to capture EGFR exon 20 insertions as a class-based variant start at codon 762 and end at codon 775. EGFR exon 20 insertions such as p.A763_Y764insFQEA is associated with lowered sensitivity to clinically achievable doses of reversible EGFR TKIs, erlotinib and gefitinib, as well as the irreversible EGFR TKIs, neratinib, afatinib, and dacomitinib [PMID: 26096453, PMID: 30854234, PMID: 24891042]. The boundary involving codon 775 captures the p.V774_C775insPR variant which is associated with insensitivity to first-generation EGFR TKIs [PMID: 24891042].

ERBB2 exon 20 insertion

- Background:** Erb-b2 receptor tyrosine kinase 2 (ERBB2) is a transmembrane glycoprotein located on chromosome 17q12 [PMID: 25276427]. ERBB2 (also known as HER2) has 27 exons— exons 2-4 and 9-12 encode the extracellular receptor L domains, exons 5-8 encode the furin like domain, exons 13-16 encode the growth factor receptor domain IV, exon 17 encodes the transmembrane domain, and exons 18-24 encode the tyrosine kinase domain (TKD) [PMID: 22761469, PMID: 29420467]. In lung cancer, the most recurrent ERBB2 activating mutations include in-frame exon 20 insertions [PMID: 30425522]. ERBB2 exon 20 involves codons 770 to 831 and majority of exon 20 insertions occur between codons 775 and 781 within the kinase domain [PMID: 29686424, PMID: 22761469]. Insertions at the C-terminal end of ERBB2 exon 20 induce a change in conformation of the α -C helix leading to a constitutively active formation which affects the drug-binding pocket [PMID: 29686424].
- CBV Criteria:** The boundaries defined to capture ERBB2 exon 20 insertions as a class-based variant start at codon 770 and end at codon 783. ERBB2 exon 20 insertions, such as Y772_A775dup prevent binding of the noncovalent ERBB2/HER2 inhibitor lapatinib, which binds to ERBB2/HER2 in the inactive conformation [PMID: 29686424].

MET exon 14 skipping

- **Background:** The MET proto-oncogene is a receptor tyrosine kinase made up of 1390 amino acids and is located on chromosome 7q31 [PMID: 15735036]. MET has 21 exons containing three main structural domains—an extracellular Sema domain in exon 2, a juxtamembrane domain in exon 14, and a tyrosine kinase domain in exons 15-21 [PMID: 28376232, PMID: 9380410]. Splice site mutations flanking exon 14 are observed in 4% of non-small cell lung cancer (NSCLC). These mutations include canonical splice site mutations affecting exon 14 and deletions that extend into the splicing motifs within intron 13 [PMID: 25971938, PMID: 27343443]. Such mutations disrupt splicing leading to the formation of an alternative transcript that joins exon 13 directly to exon 15 and skips exon 14 entirely. The MET exon 14 skipping transcript lacks the juxtamembrane domain that contains the recognition motif for ubiquitin-dependent proteolysis and thus leads to a marked increase in steady-state level of the MET protein [PMID: 28164087]. MET exon 14 skipping is a target of several c-MET tyrosine kinase inhibitors including crizotinib, capmatinib, and cabozantinib [PMID: 27223456].
- **CBV Criteria:** The boundaries defined to capture MET exon 14 skipping as a class-based variant can be summarized into four individual rules: (a) has transcript = NM_001127500.3, exon = 14, and location = splicesite_3 (at c.2942) or splicesite_5 (at c.3082), (b) is an intronic deletion ≥ 4 bp impacting 30 nucleotides preceding exon 14 (c) is a missense variant at c.3082 [PMID: 27343443, PMID: 26729443, PMID: 25971938, PMID: 25898962, PMID: 25898965, PMID: 31472177], (d) is one of the following variants with confirmed skipping defined as c.3082delG [PMID: 25971938, PMID: 27343443], c.3075_3082del [PMID: 31472177], or c.3080_3081delAA [PMID: 26729443].

Variant identifiers for OncoPrint™ panels

Hotspots BED files

The BED file specification (<http://genome.ucsc.edu/FAQ/FAQformat.html#format1.7>) indicates that the fourth column is the name of the BED line, and is used to label the variant region in the UCSC genome browser or IGV. This label is also used to populate the ID field in the output VCF files as well as the Variant ID columns in Genexus™ Software.

OncoPrint™ panel hotspots files contain genomic representations that correspond to somatic variants that have been frequently observed in cancer samples, and thus are likely to be relevant to the cancer phenotype. These files also contain less frequently observed variants, implicated in literature reports as functionally relevant, for example, activating/inactivating variants that are associated with sensitivity or resistance to targeted agents, or associated with pathogenicity. When possible, variants within the hotspots files are assigned an identifier consistent with a publicly accessible data source, preferentially COSMIC (<https://cancer.sanger.ac.uk/cosmic>) but also including dbSNP (<https://www.ncbi.nlm.nih.gov/snp/>) and ClinVar (<https://www.ncbi.nlm.nih.gov/clinvar/>). If a variant cannot be found in COSMIC, it may receive an arbitrarily assigned identifier to aid in variant calling interpretation and troubleshooting; these identifiers (for example, BT144, OM3324, OMINDEL700, MAN103) should be consistent across OncoPrint™ panels. When hotspots files are updated, Thermo Fisher Scientific reviews the COSMIC database to determine whether variants with such identifiers have been assigned COSMIC IDs, and replace them with the more meaningful COSMIC IDs.

COSMIC ID changes due to database updates

The COSMIC database updates approximately four times a year, and these updates involve the addition of newly curated variants, removal of a smaller number of variants, and changes in genomic representations to a very small number of variants. There is, therefore, a chance that the variant identifiers in the hotspots files might be out of sync with, or no longer be found on the COSMIC website. Additionally, prior to COSMIC version 90, the same normalized genomic variant (see <https://pubmed.ncbi.nlm.nih.gov/25701572/>) might have had multiple redundant COSMIC variant identifiers (COSM); since COSMIC version 90, these identifiers have been replaced with one consistent COSV identifier, and multiple COSMIC variants may map to these identifiers.

Fusions files

A comprehensive, universally recognized database of oncogenic gene fusion breakpoints does not exist. Therefore, gene fusion isoform identifiers are generated by concatenating the two gene symbols with a hyphen, and then combining the first letter of the 5' partner with the last retained 5' exon along with the first letter of the 3' partner with the first retained 3' exon. For example, EML4-ALK.E6A17 involves a fusion between the sixth exon of EML4 with the 17th exon of ALK. Fusion isoforms involving junctions between incomplete exons, or involving intronic insertions may contain "ins" or "del" modifiers followed by the number of nucleotides removed or added. Additionally, assay names may contain additional identifiers such as COSMIC COSF ids or GenBank accession numbers.

System-installed annotation sets

Genexus™ Software includes system-installed annotation sets as presets to use in assays. System-installed annotation sets are locked and cannot be changed.

System-installed annotation sets	Description
All_hg19_516	A system-installed annotation set for use with hg19 assays.
All_GRCh38_516	A system-installed annotation set for use with GRCh38 assays.
Performance Qualification Annotations -1.9.0 ^[1]	A system-installed annotation set for use with Performance Qualification assays.
Integrated Purification Performance Qualification Annotations -1.9.2 ^[1]	A system-installed annotation set for use with Performance Qualification assays for the integrated Genexus™ Integrated Sequencer.

^[1] This annotation set is available when the assay parameter set for it is enabled in the system.

System-installed barcode sets

The Genexus™ Software includes system-installed barcode sets as presets to use in assays. System-installed barcode sets are locked and cannot be changed.

System-installed barcode set	Description
Ion AmpliSeq™ HD Dual Barcode Kit 1–24	A set of 24 unique dual-barcode primers designed and validated for optimal performance with Ion GeneStudio™ S5 Series sequencers. For use with Ion AmpliSeq™ HD assays.
IonCode™ Barcodes 1-32	A unique set of 32 barcode adapters designed for optimal performance with Ion GeneStudio™ S5, Ion PGM™, and Ion Proton™ sequencers.
IonCode™ Barcodes 1-384	A unique set of 384 barcode adapters designed for optimal performance with Ion GeneStudio™ S5, Ion PGM™, and Ion Proton™ sequencers.
Ion Select BC Set-1	A unique set of 16 barcode adapters supplied with the Ion PGM™ Select Library Kit.
Ion SingleSeq™ Barcodes	A unique set of 96 barcode adapters optimized for use with Ion ReproSeq™ libraries.
Ion Torrent™ Dual Barcode Kit 1–96	A set of 96 unique dual-matched barcode adapters designed to enable optimal performance of amplicon libraries and Ion semiconductor sequencers. For use with Ion AmpliSeq™ HD and OncoPrint™ assays.
Ion Torrent™ Genexus™ Barcodes 1–32 HD	A unique set of 32 barcode adapters designed and validated for optimal performance with the Genexus™ Integrated Sequencer. For use with Ion AmpliSeq™ HD assays.
Ion Torrent™ Genexus™ Barcodes 1–96 AS	A unique set of 96 barcode adapters designed and validated for optimal performance with the Genexus™ Integrated Sequencer.
Ion Xpress™ Barcodes	A unique set of 96 barcode adapters for use with Ion AmpliSeq™ libraries and Ion Xpress™ Plus fragment libraries.

System-installed filter chains

Genexus™ Software includes system-installed filter chains as that are included as presets in assays to filter in or out variants of interest in run results. The filter chains can also be used to dynamically change the view of the variants as you view results. System-installed filter chains are locked and cannot be changed. You cannot edit system-installed filter chains, but you can create custom filter chains to refine the results in the software.

System-installed filter chain	Available versions ^[1]	Description
Oncomine™ Extended	<ul style="list-style-type: none"> 5.14 5.16 	<p>This filter chain includes all Oncomine™-annotated variants and variants that can be relevant to cancer due to their inclusion in one or more of the following classes:</p> <ul style="list-style-type: none"> Non-targeted fusions. CNV variants with FILTER value of GAIN or LOSS. Likely somatic mutations based on dbSNP, 5000Exomes, ExAC, and UCSC Common SNPs annotation source databases. The minor allele frequencies range lies between 0.0 and 1.0E-6. Mutations must also be nonsynonymous and occur in exonic or splice-site regions. Variants with ClinVar annotations of pathogenic or likely pathogenic.
Oncomine™ Variants	<ul style="list-style-type: none"> 5.14 5.16 	This filter chain includes all Oncomine™-annotated variants.
Variant Matrix Summary	<ul style="list-style-type: none"> 5.14 5.16 	Returns variant types in a visualization. Variant types returned are SNV/INDEL, CNV, fusions, and RNAExonVariants.
TMB (Non-germline Mutations)	not applicable	This filter chain returns results for somatic variants based on dbSNP, 5000Exomes, ExAC, and UCSC Common SNPs annotation source databases. The minor allele frequencies range is between 0.0 and 1.0E-6. This filter chain also filters out variants of homopolymer lengths greater than 7.
Oncomine Variants AND Non-Targeted Fusions	not applicable	<p>A logical filter group that contains multiple filters that might include unannotated variants.</p> <p>Filters are applied in the following order.</p> <ul style="list-style-type: none"> Oncomine = In Variant Type in FUSION Filter in PASS
No Filter	not applicable	Select this option to remove a previously applied filter chain and view all called variants.

^[1] The version depends on the assay parameter set installed in the software.

System-installed filters

Genexus™ Software includes system-installed filters that you can add to custom filter chains. A filter chain is a set of filters that the software uses to narrow the list of variants that are included in results. When you create a filter chain, you can dynamically change the view of variants in results, or you can filter in or out variants from results by adding the filter chain to an assay that is used for a sequencing run.

The system-installed filters are based on public and proprietary annotation sources and data types that are included in the software.

Annotation source filters

You can create filter chains that identify variants based on the annotations associated with those variants. The software uses annotation sources from public and proprietary genomic databases to apply these annotations to variants during analysis.

Filters that are made from annotation sources are available in the software and you can use these filters to build filter chains that narrow the list of variants that appear in the analysis results.

5000Exomes filters

The 5000Exomes filters can be added to filter chains to detect minor allele frequencies (MAF) for specific population groups in the software. The filters are derived from the 5000Exomes annotation database. The specific filters are:

- 5000Exomes AfricanAmerican MAF
- 5000Exomes AfricanAmerican MAF(20161108)
- 5000Exomes EuropeanAmerican MAF
- 5000Exomes EuropeanAmerican MAF(20161108)
- 5000Exomes Global MAF
- 5000Exomes Global MAF(20161108)

Filter conditions include the ability to set minor allele frequency ranges from 0.0 to 0.5. The option to include or exclude unannotated variants in filtered analysis results is also available. The **Inclusive** option, to include or exclude boundary values in the range, is selected by default and cannot be changed.

The filter returns minor allele frequency results in samples that align with the 5000Exomes minor allele frequency database.

Clinvar filter

This filter can be added to filter chains in the software to evaluate the impact of variants observed in samples that match those in the National Center for Biotechnology Information (NCBI) ClinVar database. The specific filters are:

- Clinvar(20180725)
- Clinvar(20190909)
- Clinvar(20200329)

The impact values that can be included in the filter chain are: Pathogenic, Likely benign, Benign, other, Likely pathogenic, Uncertain significance, not provided, and drug response.

The filter returns all variants with the selected impact values that match those in the NCBI ClinVar database.

COSMIC filter

This filter can be added to filter chains in the software to compare variants to the catalog of somatic mutations in tumor tissue as compiled by the COSMIC database. The specific filters are:

- COSMIC(85)
- COSMIC(89)
- COSMIC(91)

Filter conditions include the ability to select all COSMIC values, to select specific annotation values, and to include or exclude unannotated variants in filtered analysis results.

The filter returns variants that match those in the COSMIC database.

dbSNP filter

This filter can be added to filter chains in the software to compare single nucleotide polymorphism variants in samples against the dbSNP database. The specific filters are:

- dbSNP(151)
- dbSNP(153)
- dbSNP(154)

Filter conditions include the ability to select all dbSNP values (In), or to exclude all dbSNP values (Not In).

The filter detects SNP variants that match those in the dbSNP database, which can be included in or excluded from filtered analysis results.

DGV filter

This filter can be added to filter chains in the software to detect human genomic structural variants that match those in the Database of Genomic Variants (DGV). The specific filters are:

- DGV filter(20160515)
- DGV filter(20200225)

Filter conditions include the ability to include all DGV variants (In), or to exclude all DGV variants (Not In) in filtered analysis results.

The filter detects human genomic structural variants as defined by DGV, which can be included in or excluded from filtered analysis results.

DrugBank filter

This filter can be added to filter chains in the software to detect variants that are correlated with drugs and drug targets listed DrugBank database. The specific filters are:

- DrugBank (20180731)
- DrugBank (20190723)
- DrugBank (20200619)

Filter conditions include the ability to select specific values, to include all DrugBank values (In), or to exclude all DrugBank values (Not In). In addition, unannotated variants can be included or excluded in filtered analysis results.

This filter detects variants that are correlated with drugs and drug targets in DrugBank, which can be included in or excluded from filtered analysis results.

ExAC filters

These filters can be added to filter chains to detect rare gene variants for specific population groups in the software. The filters are derived from the Exome Aggregation Consortium (ExAC) database. The specific filters are:

- ExAC AAF(1)
- ExAC EAAF(1)
- ExAC EFAF(1)
- ExAC ENFAF(1)
- ExAC GAF(1)
- ExAC LAF(1)
- ExAC OAF(1)
- ExAC SAAF(1)

Filter conditions include the ability to set a range from 0.0 to 1.0. The option to include or exclude unannotated variants in filtered analysis results is also available. The **Inclusive** option, to include or exclude boundary values in the range, is selected by default and cannot be changed.

The filter detects rare gene variant results that align with the ExAC database, which can be included or excluded from filtered analysis results.

FATHMM Scores filter

This filter can be added to filter chains in the software to calculate Functional Analysis through Hidden Markov Models (FATHMM) Scores for coding variants, nonsynonymous single-nucleotide variants (nsSNVs), and noncoding variants. The specific filters are:

- FATHMM Scores(85)
- FATHMM Scores(89)
- FATHMM Scores(91)

Filter conditions include the ability to set a range from 0.0 to 1.0 for the score. The option to include or exclude unannotated variants in filtered analysis results is also available. The **Inclusive** option, to include or exclude boundary values in the range, is selected by default and cannot be changed.

The filter calculates FATHMM scores for coding variants, nsSNVs, and noncoding variants.

Gene Ontology (GO) filter

This filter can be added to filter chains in the software to detect various gene ontologies (functions of specific genes). The specific filters are:

- go(20171101)
- go(20190930)
- go(20200511)

Filter conditions include the ability to select all (In), none (Not In), or to select specific annotations, and to include or exclude unannotated variants in filtered analysis results.

This filter returns gene ontologies of samples based on the selections made.

Gene Symbol filter

This filter can be added to filter chains in the software to report on specific genes.

Filter conditions include the ability to select all (In), none (Not In), or specific gene symbols from the list, and to include or exclude unannotated variants in filtered analysis results.

This filter returns the gene symbols of genes that are found in your samples.

Minor Allele Frequency filter

This filter can be added to filter chains in the software to detect variants with minor allele frequencies that match those in the dbSNP database from the 1000 genomes project.

Filter conditions include the ability to set a range from 0.0 to 0.5 for minor allele frequencies. The option to include or exclude unannotated variants in filtered analysis results is also available. The **Inclusive** option, to include or exclude boundary values in the range, is selected by default and cannot be changed.

The filter returns variants in samples that match those in the dbSNP database from the 1000 genomes project.

OMIM filter

This filter can be added to filter chains in the software to detect variants that match those in the Online Mendelian Inheritance in Man® (OMIM) database. The specific filters are:

- OMIM(20180823)
- OMIM(20191001)
- OMIM(20200508)

Filter conditions include the ability to search for all (In), none (Not In), or specific annotations, and to include or exclude unannotated variants in filtered analysis results.

The filter returns variants in samples that match those in the OMIM database.

Pfam filter

This filter can be added to filter chains in the software to detect protein domain families in the coded proteins as defined by the Pfam database. The specific filters are:

- Pfam(31)
- Pfam(32)
- Pfam(33)

Filter conditions include the ability to include all (In), none (Not In), or specific annotations, and to include or exclude unannotated variants in filtered analysis results.

The filter returns variants in the samples that match the variants in the Pfam database.

PhyloP Scores filter

This filter can be added to filter chains in the software to measure the conservation of protein across a wide range of organisms in 16S metagenomics analyses. The specific filters are:

PhyloP Scores(20160919)

Filter conditions include the ability to set a score range from –14.0 to 3.0. The option to include or exclude unannotated variants in filtered analysis results is also available. The **Inclusive** option, to include or exclude boundary values in the range, is selected by default and cannot be changed.

This filter returns PhyloP scores for 16S metagenomics samples.

UCSC Common SNPs filter

This filter can be added to filter chains in the software to detect variants that match those in the UCSC Genome Browser Common SNPs database.

Filter conditions include the ability to include (In) or exclude (Not In) variants that match the UCSC Common SNPs in filtered results.

The filter returns variants in samples that match the UCSC Common SNPs database.

Data type filters

You can create filter chains that are based on variant data types in the software, such as allele frequency and allele ratio. Many data types that are included in the results tables are available as filters.

Allele fraction filter

The Allele fraction filter can be added to filter chains in the software to report the frequency of alleles observed in raw data.

Filter conditions include the ability to set allele frequency ranges from 0.0 to 1.0. The option to include or exclude unannotated variants in filtered analysis results is also available. The **Inclusive** option, to include or exclude boundary values in the range, is selected by default and cannot be changed.

The filter returns allele frequencies of alleles observed in raw data.

Allele Ratio filter

This filter can be added to filter chains in the software to narrow analysis results to nonreference allele frequencies.

Filter conditions include the ability to set allele ratio ranges from 0.0 to 1.0. The option to include or exclude unannotated variants in filtered analysis results is also available. The **Inclusive** option, to include or exclude boundary values in the range, is selected by default and cannot be changed.

The filter returns all variants that have at least one nonreference allele reported with a frequency in the selected filter range.

Allele Read-Count filter

This filter can be added to filter chains in the software to set the minimum count for genotype alleles.

Filter conditions include the ability to set allele read-count ranges from 0 to 100,000. The option to include or exclude unannotated variants in filtered analysis results is also available. The **Inclusive** option, to include or exclude boundary values in the range, is selected by default and cannot be changed.

The filter removes variant candidates that do not have the required numbers of supporting reads from analysis results.

Alternate Allele Count filter

This filter can be added to filter chains in the software to set the minimum count for alternate alleles.

Filter conditions include the ability to set alternate allele read-count ranges from 0 to 100,000. The option to include or exclude unannotated variants in filtered analysis results is also available. The **Inclusive** option, to include or exclude boundary values in the range, is selected by default and cannot be changed.

The filter removes variant candidates that do not have the required numbers of reads from analysis results.

CNV Confidence Range filter

This filter can be added to filter chains in the software to return copy number variants (CNV) with confidence levels between 10 and 10,000,000 and other variants.

Filter conditions include the ability to set the CNV confidence range from 10 to 1.0E7. The option to include or exclude unannotated variants in filtered analysis results is also available. The **Inclusive** option, to include or exclude boundary values in the range, is selected by default and cannot be changed.

The filter returns copy number variants with high confidence levels and other variants.

CNV Confidence Range—CNVs Only filter

This filter can be added to filter chains in the software to return copy number variants (CNV) with confidence levels between 10 and 10,000,000.

Filter conditions include the ability to set the CNV confidence range from 10 to 1.0E7. The option to include or exclude unannotated variants in filtered analysis results is also available. The **Inclusive** option, to include or exclude boundary values in the range, is selected by default and cannot be changed.

The filter returns only copy number variants with high confidence levels.

CNV Somatic Confidence Range filter

This filter can be added to filter chains in the software to make ploidy estimates with lower and upper confidence values on somatic samples. The filter detects extra copies over the expected normal ploidy value (2 for autosomes, 2 for X chromosomes in females, and 1 for X chromosomes in males). The 5% lower confidence bound value is the ploidy estimate where there is a 5% chance that the true ploidy is below that value. The 95% upper confidence bound is the ploidy estimate where it is 95% certain that the true ploidy is below that value. The lower bound is the most important for gains. The upper bound is most important for losses.

By default, the filter is set to detect gains and losses using the confidence interval values of 5% confidence interval for Minimum Ploidy Gain over the expected value and 95% confidence interval for Minimum Ploidy Loss under the expected value. The default boundary values are set to 0.0.

If you set the **Minimum Ploidy Gain (5% CI) over expected to 2.0** ($2+2=4$) and the **Minimum Ploidy Loss (95% CI) under expected to 0.0**, you can expect the following example CNV call CI data:

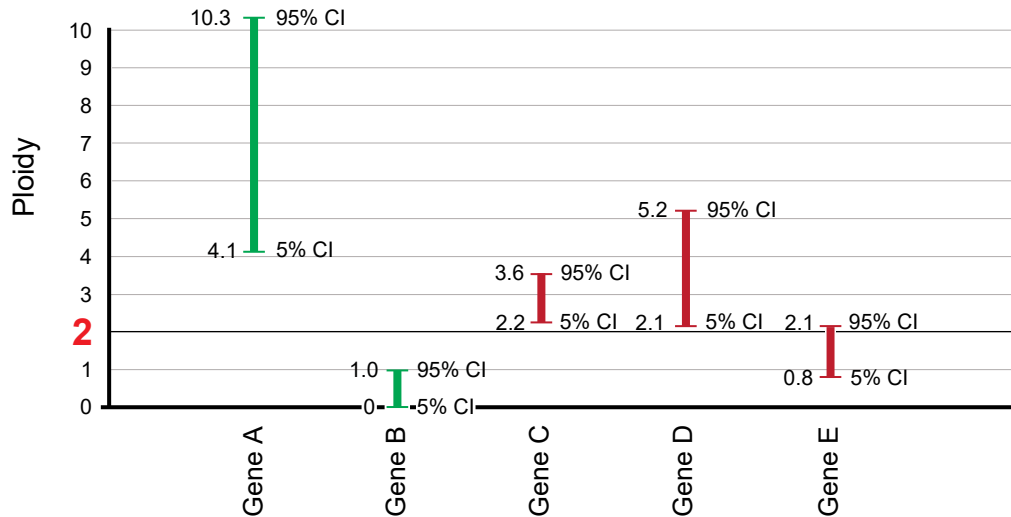
Gene A—A gene with suspected gain of 5% CI = 4.1 and 95% CI = 10.3 filtered in.

Gene B—A gene with suspected loss of 5% CI = 0 and 95% CI = 1.0 filtered in.

Gene C—A gene with suspected gain of 5% CI = 2.2 and 95% CI = 3.6 filtered out (2.2 is less than 4).

Gene D—A gene with suspected gain of 5% CI = 2.1 and 95% CI = 5.2 filtered out (2.1 is less than 4).

Gene E—A gene with 5% CI = 0.8 and 95% CI = 2.1 filtered out (0.8 is less than 4 and 2.1 is greater than 2).



CNV Somatic Confidence—CNVs Only filter

This filter can be added to filter chains in the software to make ploidy estimates with lower and upper confidence levels.

Filter conditions include the ability to enable and set the range for Minimum Ploidy Gain (5% CI) over expected, to enable and set the Minimum Ploidy Loss (95% CI) under expected ploidy values. The option to include or exclude unannotated variants in filtered analysis results is also available. The **Inclusive** option, to include or exclude boundary values in the range, is selected by default and cannot be changed.

The filter returns only somatic copy number variants within the lower and upper limits of the set confidence ranges.

Confident Somatic Variants filter

This filter detects confident somatic variants in tumor-normal analyses. The filter compares tumor samples to matched normal samples. It does not apply to single sample analyses.

Filter conditions include the ability to include the confident somatic variants (In), or to exclude the confident somatic variants (Not In) in filtered analysis results.

Deamination filter

This filter can be added to filter chains in the software to filter C:G>T:A SNVs that are detected at all allele frequencies.

Filter conditions include the ability to exclude (Not In) or include (In) these variants in filtered results.

Default DNA and Fusions View filter

This filter can be added to filter chains in the software to detect all of the fusion variants, expression control markers, assay 5'/3' markers, and all DNA variants that the predefined Default DNA and Fusions View filter chain detects.

Filter conditions include the ability to select specific annotations (Present or Absent), to include all default DNA and Fusions view variants (In), or to exclude all Default DNA and Fusions view variants (Not In) in filtered analysis results. In addition, unannotated variants can be included or excluded in filtered analysis results.

The filter detects the fusion variants, expression control markers, assay 5'/3' markers, and all DNA variants, which can be included or excluded in filtered analysis results.

Default Fusions View filter

This filter can be added to filter chains in the software to detect all of the fusion variants, expression control markers, and assay 5'/3' markers that the predefined Default Fusions View filter chain detects.

Filter conditions include the ability to select specific annotations (Present or Absent), to include all Default Fusions View variants (In), or to exclude all Default Fusions View variants (Not In) in filter analysis results. In addition, unannotated variants can be included or excluded in filtered analysis results.

The filter detects fusion variants, expression control markers, and assays 5'/3' markers, which can be included or excluded in filtered analysis results.

Disease Research Area (DRA) filter

This filter can be added to filter chains in the software to annotate disease research categories as defined by the Disease Research Area (DRA) annotation source database. The specific filters are:

- Disease Research Area (DRA)(20170914)

Filter conditions include the ability to select specific annotations, to include all Disease Research Area values (In), or to exclude all Disease Research Area values (Not In) in filtered analysis results. In addition, unannotated variants can be included or excluded in filtered analysis results.

This filter annotates disease research areas that match the DRA database, and can be included or excluded.

Expected Normal Ploidy Buffer filter

The Expected normal ploidy buffer filter can be added to filter chains in the software to detect mosaic chromosomal aneuploidies in research samples and to set a buffer for normal ploidy values (2 for autosomes, 2 for X chromosomes in females, and 1 for X and Y chromosomes in males) so as to identify the most likely aneuploidy samples. Ploidy calls do not have to be integers because mosaicism detection allows decimal-level Copy Number Gain or Loss calls.

By default, the filter is set to filter out all copy number variant segments of gain or loss within 0.2 ploidy value of expected normal. Gains must be above ploidy 2.2 and below ploidy 1.8 to remain filtered in on autosome and female X, and ploidy 1.2 and ploidy 0.8 on male Y. Filter conditions include the ability to change the amount of buffer by adjusting the From and To values. This Expected Normal Ploidy Buffer overrides any Confidence filtering that may be in effect within the buffer zone.

If default settings are applied, ploidy values of 1.8 to 2.2 are filtered out as normal for autosomes and X chromosomes in females, and 0.8 to 1.2 are filtered out as normal for X and Y chromosomes in males. Values outside this range are detected as chromosomal ploidy samples.

Filter filter

This filter can be added to filter chains in Genexus™ Software to show or exclude specific annotations.

- REF
- PASS
- NOCALL
- LOSS
- GAIN
- FAIL
- ABSENT_HOM_V
- ABSENT_HET_V
- ABSENT_HET_RV
- .

Filter conditions include the ability to include all (In), none (Not In), or specific annotations, and to include or exclude unannotated variants in results when you apply the filter chain.

Filtered Coverage filter

This filter can be added to filter chains in the software to review results that meet a specific coverage threshold. By default, the filter reports all results with a coverage threshold greater than 100. Or, you can use the filter settings to change the threshold to include or exclude boundary values for the threshold, and to include or exclude unannotated variants in filtered analysis results. The default threshold setting is 100.

Filter conditions include the ability to set a minimum and a maximum value for minor allele frequency for the coverage threshold. Ranges from 0.0 to 0.0045 can be selected for the minimum in the **From** field and in the **To** field for the maximum. The option to include or exclude unannotated variants in filtered analysis results is also available. The **Inclusive** option, to include or exclude boundary values in the range, is selected by default and cannot be changed.

Functional Scores filter

This filter can be added to filter chains in the software to provide functional scores based on SIFT, PolyPhen, and Grantham scores. SIFT and PolyPhen scores are predictions of the functional effect of a variant on a protein. A Grantham score attempts to predict the distance between two amino acids, in an evolutionary sense (lower is less distance, higher is greater distance).

Filter conditions include the ability to include or exclude SIFT, PolyPhen, and Grantham scores, and to set ranges for those scores. SIFT and PolyPhen ranges can be set from 0.0 to 1.0, and Grantham can be set from 0.0 to 215.0. The option to include or exclude unannotated variants in filtered analysis results is also available. The **Inclusive** option, to include or exclude boundary values in the range, is selected by default and cannot be changed.

The filter returns a functional score based on the three (or fewer) scores.

Fusion Read Counts filter

This filter can be added to filter chains in the software to produce counts for fusion reads.

Filter conditions include the ability to set a minimum count of 0 or higher, to include the boundary value for the count, and to include or exclude unannotated variants in filtered analysis results.

This filter returns results that meet the fusion read minimum count value.

Genetic Category Type filter

This filter can be included in a filter chain in the software to detect variants of specific genetic category types.

Filter conditions include the ability to select all, none, or specific genetic category type annotations, and to include or exclude unannotated variants in filtered analysis results.

Genetic category types include:

- IsNewlyHomozygousRef
- IsNewlyHomzygousNonRef
- IsNewlyHeterozygousNonRef
- InconsistentWithParents
- InconsistentWithMother
- InconsistentWithFather
- InTransPhaseCompoundHeterozygote
- InCompoundHeterozygote
- HasUnknownX
- HasMaleMaternalX
- HasDeNovoRefAllele
- HasDeNovoNonRefAllele

This filter returns genetic category type annotations for the variants included in the samples.

Genomic Coordinates filter

This filter can be added to filter chains in the software to narrow analysis results to specific genomic regions. The filter can also be used to identify regions that are targeted by an amplicon or amplicons in a user-defined assay.

The filter value is one chromosome region, or multiple regions that are separated by OR operators or commas.

The Genomic Coordinates filter returns variants in samples that are in the range for the genomic coordinates that you set.

IMPORTANT! Do not use AND, OR, or NOT when adding values to the Genomic Coordinates filter.

Homopolymer Length filter

This filter can be used in a filter chain in the software to detect INDELS of specific homopolymer lengths.

Filter conditions include the ability to set a range from 0 to 1,000 for homopolymer length. The option to include or exclude unannotated variants in filtered analysis results is also available. The **Inclusive** option, to include or exclude boundary values in the range, is selected by default and cannot be changed.

This filter returns INDEL variants in samples that meet the specified homopolymer length.

Hotspot filter

The Hotspot filter can be added to filter chains in Genexus™ Software to show or exclude variants that are hotspots.

Filter conditions include the ability to include (In) or exclude (Not In) hotspot position overlaps in results when the filter chain is applied.

The filter returns variants that overlap hotspots that are defined in an uploaded hotspots file.

Location filter

This filter can be added to filter chains in the software to detect variants in specific positions, such as exonic.

Filter conditions include the ability to select all locations or specific locations in filtered analysis results. Locations include:

- unknown
- intergenic
- intronic
- exonic
- utr_5
- utr_3
- splicesite_5
- splicesite_3
- upstream
- downstream
- exonic_nc
- intronic_nc
- ncRNA
- nonCoding

The filter returns variants in samples that match the selected filter locations.

Minimum Limit of Detection at Genomic Location filter

The Minimum Limit of Detection at Genomic Location filter can be added to filter chains to filter for a minimum limit of detection (LOD) at chromosomal locations within a selected range.

This LOD calculation depends on the depth of coverage at a given chromosomal location and the probability that is defined by `Minimum Callable Probability` parameter.

This filter is for use with Ion AmpliSeq analysis results and analysis workflows only. For more information, see “Parameters step assay options” on page 201.

Filter conditions include the ability to set a minimum and a maximum value for minor allele frequency. Ranges from 0.0 to 0.0045 can be selected for the minimum in the **From** field and in the **To** field for the maximum. The option to include or exclude unannotated variants in filtered analysis results is also available. The **Inclusive** option, to include or exclude boundary values in the range, is selected by default and cannot be changed.

Named Variants filter

This filter can be added to filter chains in the software to detect known variants in the Ion AmpliSeq™ Community Panel for the cystic fibrosis transmembrane regulator (CFTR) gene, for example: c.2054delA.

Filter conditions include the ability to select all named variants, to select specific named variants, and to include or exclude unannotated variants in filtered analysis results.

The filter returns variants in samples that match the CFTR variants that are specified in the filter.

OncoPrint™ filter

This filter is the default filter for most OncoPrint™ assays. It presents the user with OncoPrint™ driver variants that are based on their OncoPrint™ Gene Class and OncoPrint™ Variant Class annotations. It can be added to filter chains in the software to detect OncoPrint™-annotated variants in specific OncoPrint™ panels.

Analysis workflows for OncoPrint™ assays and analysis workflow templates for Ion AmpliSeq™ HD panels in Ion Reporter™ Software include the OncoPrint™ Variant Annotator plugin. The plugin integrates into analysis results data from more than 24,000 exomes across solid tumor and hematological cancer types, and annotates variants relevant to cancer with OncoPrint™ Gene class and OncoPrint™ Variant class information. For more information and a full list of annotation rules for each OncoPrint™ assay, see “Annotation criteria for OncoPrint™ Variant Annotator” on page 340, or contact your local support representative, Field Bioinformatics Specialist (FBS), or Clinical Account Consultant (CAC).

The OncoPrint™ filter (OncoPrint = In) returns variants in the samples that have OncoPrint™ Gene class and OncoPrint™ Variant class annotation.

This filter can be used only if an OncoPrint™ analysis workflow was used for the analysis. You can apply filter chains that include the OncoPrint™ filter only if the analysis workflow that was used for the analysis includes the OncoPrint™ Variant Annotator plugin. Examples of filter chains that include this filter are the OncoPrint™ Variants and OncoPrint™ Extended filter chains.

Possible Polyploidy Alleles filter

The Possible Polyploidy Alleles filter can be added to filter chains to detect searches alleles which have passed all variant calling filters used by the Torrent Variant Caller (TVC) module but missed by genotype.

The Possible Polyploidy Alleles filter is useful for identification of variants beyond diploid genomes, that is, cancer cells or polyploidy species.

Filter conditions include the ability to select **YES** to find alleles that have passed all filters, **NO**, to find alleles that have *not* passed all filters in addition to genotype, or both **YES** and **NO**.

PValue filter

This filter can be added to filter chains in the software to determine the p -value of variants. The p -value is a statistical method for the detection of variant calls from next-generation sequencers.

Filter conditions include the ability to set a range from 0.0 to 1.0. The option to include or exclude unannotated variants in filtered analysis results is also available. The **Inclusive** option, to include or exclude boundary values in the range, is selected by default and cannot be changed.

The filter returns p -values for variants calls in the samples.

Variant Classification filter

This filter can be added to filter chains in the software to narrow results to the following user-set variant classifications:

- Unclassified
- Unknown
- Benign
- Suspected Benign
- Likely Benign
- Suspected Deleterious
- Deleterious
- Pathogenic
- Likely Pathogenic
- VUS
- Uncertain Significance
- Technical Artifact

Filter conditions include the ability to include all (Filter Option set to In), none (Filter Option set to Not In), or to select specific classifications in filtered analysis results.

The filter returns variants that are classified by software users.

Variant Effect filter

This filter can be added to filter chains in the software to detect the effect of variants on coding sequences. Specific values include:

- refAllele
- unknown
- synonymous
- missense
- nonframeshiftInsertion
- nonframeshiftDeletion
- nonframeshiftBlockSubstitution
- nonsense
- stoploss

- frameshiftInsertion
- frameshiftDeletion
- frameshiftBlockSubstitution

Filter conditions include the ability to select all (Filter Option set to In), none (Filter Option set to Not In), or to select specific values, and to include or exclude unannotated variants in filtered analysis results.

The filter returns variants in samples that match the selected variant effects.

Variant Subtype filter

This filter can be added to filter chains in the software to detect variant subtypes, including:

- BigDel
- BigDup
- GeneCNV
- REF
- NOCALL

Filter conditions include the ability to select all or specific variant subtypes in filtered analysis results.

The filter returns variants that match the selected specific variant subtypes.

Variant Type filter

This filter can be added to filter chains in the software to detect variants that match the following variant types:

- | | |
|----------------|-------------------|
| • SNV | • ASSAYS_5P_3P |
| • INDEL | • RNA_HOTSPOT |
| • MNV | • GENE_EXPRESSION |
| • REF | • RNAExonVariant |
| • NOCALL | • ProcControl |
| • CNV | • FLT3ITD |
| • LONGDEL | |
| • FUSION | |
| • EXPR_CONTROL | |

Note: The FLT3ITD variant type is detected only in OncoPrint™ Myeloid Assay analysis workflows.

Filter conditions include the ability to select all variant types or specific variant types in filtered analysis results.

The filter returns variants in analyses that match the selected specific variant types.

Zygoty filter

This filter can be added to filter chains in the software to detect variants that are homozygous or heterozygous.

Filter conditions include the ability to search for both homozygous and heterozygous variants, or either of them, in filtered analysis results.

The filter returns variants in analyses that are homozygous or heterozygous.

System-installed plugins

Genexus™ Software includes system-installed plugins that expand the analysis capabilities of the software. System-installed plugins are locked and cannot be changed.

System-installed plugin	Description
coverageAnalysis	This plugin provides statistics and graphs that describe the level of sequence coverage that is produced for targeted genomic regions.
CustomerSupportArchive	This plugin generates a downloadable archive that a technical support representative can use to troubleshoot and diagnose problems with sequencing runs or with Genexus™ Software. The archive contains log files and other technical data about the software and other files from sequencing runs on a Genexus™ Integrated Sequencer. For more information, see “Download a customer support archive” on page 371.
molecularCoverageAnalysis	This plugin provides statics and graphs that describe the level of sequence molecular coverage that is produced for targeted genomic regions.
sampleID	This plugin produces a unique identification code (sampleID) for each barcode in a sample.
IonReporterUploader	This plugin allows you to directly transfer sample results files that are generated in Genexus™ Software to Ion Reporter™ Software.

System-installed reference genome files

The Genexus™ Software includes system-installed reference genome files as presets to use in assays. System-installed reference genome files are locked and cannot be changed.

System-installed reference genome file	Description
hg19	Human reference genome.
GRCh38	Human reference genome.

System-installed report templates


Genexus™ Software includes system-installed report templates that can be used directly to generate variant reports. System-installed report templates are locked and cannot be changed. However, system-installed report templates can be used as a template when you create a custom variant report.

System-installed report template	Description
Default	A system-installed report template for use with all application categories.
Default-Oncology	A system-installed report template for use with samples that are designated with the application category of Cancer.

System-installed sample attributes

The following table lists and describes system-installed sample attributes. System-installed sample attributes cannot be edited.

Note: Custom sample attributes are not listed in this table.

Sample attribute	Description
Sample Name ^[1]	<p>A unique identifier representing the sample.</p> <p>The sample name can contain only alphanumeric characters (0–9, Aa–Zz), periods (.), underscores (_), or hyphens (-), cannot contain spaces, and is limited to a maximum of 20 characters.</p> <p>IMPORTANT! To prevent erroneous sample selection during run planning, ensure that you assign a unique and distinguishable sample name for each sample.</p> <p>Note:</p> <ul style="list-style-type: none"> • Samples that have been used in a run cannot be deleted. • To prevent duplication, the software checks all sample names and returns an error message if a non-unique sample name is detected.
Collection Date ^[1]	<p>The date that the sample was collected.</p> <p>Click  Calendar to select the date in the correct format.</p>
Gender ^[1]	<p>The biological sex of the sample: Female, Male, or Unknown.</p> <p>IMPORTANT! Male or Female must be selected for proper measurement of AR CNV.</p>
Sample Type ^[1]	<p>A term that describes the sample, for example, RNA. You can also select Other, then enter a custom sample type.</p>

(continued)

Sample attribute	Description
Application Category ^[1]	The sample application category, such as Cancer (Germline) or Solid Tumor . Note: If you select an oncology application category in this list, the Cancer Stage , Cancer Type , % Cellularity , and % Necrosis attributes listed below become available in the Add New Sample dialog box.
Cancer Type ^[1]	The type of cancer that is represented by the sample. Select the type of solid or hematologic cancer. If cancer type is unknown, select Unknown Primary Origin .
Cancer Stage ^[1]	The stage of the cancer from which the sample was collected. Select Stage 0–IV , or Primary , Unknown , or Other .
% Cellularity	The percentage of tumor cells over normal cells in the sample. This is a whole number between 1 and 100. The % Cellularity attribute is applicable to FFPE samples only. IMPORTANT! <ul style="list-style-type: none"> • If not set, % Cellularity is assumed to be 100% in calculations that use this attribute. • % Cellularity is a required attribute for CNV analyses. Do not leave the field blank. • (FFPE samples only) If % Cellularity value is set to <100, then the magnitude of copy number gain or loss can be decreased. For more information, see “CNVs table” on page 141.
% Necrosis	The percentage of cellular necrosis in the sample. This is a whole number between 1 and 100.
Notes	An open-entry field for more sample information.

^[1] Required attribute

System-installed verification templates

Field Service Engineers use the system-installed verification templates during Genexus™ Integrated Sequencer installation.

IMPORTANT! The **Assays / Verification Templates** screen lists the verification templates that are used only by service engineers during Genexus™ Integrated Sequencer installation and performance qualification testing. These templates should not be used or modified in any way.

Details about each verification template is available when you click the name of the template in the software.

- Performance Qualification Assay
- System Install Qualification Assay
- Integrated Purification Performance Qualification Assay



Troubleshooting

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Download Instrument Diagnostics

You can download diagnostics files for troubleshooting purposes. Instrument diagnostics files include:

- Diagnostic archive file (ZIP format)
- Init.log file (TXT format)
- Installation Acceptance Report (PDF format)

You must upload the instrument diagnostics files from the Genexus™ Integrated Sequencer before you can download them in Genexus™ Software.

For more information, see the *Genexus™ Integrated Sequencer User Guide* (Pub. No. MAN0017910).

1. In the menu bar, click  **(Settings)** ▶ **Services**.
2. Scroll to the bottom of the screen.
3. In the **Download Instrument Diagnostics** section, in the **Select Instrument** dropdown menu, select the instrument.
4. Click **Download**.



Customer Support Archive (CSA)

You can download an archive that a Technical Support representative can use to diagnose Genexus™ Software issues. The Customer Support Archive (CSA) contains log files and other technical data about your Genexus™ Software and other files from sequencing runs on a Genexus™ Integrated Sequencer.

You can access customer support archive files from the **Results / Sample Results** and the **Results / Run Results** screens.

Download a customer support archive

When you download a customer support archive (CSA) in Genexus™ Software, files are included for all samples for a specific assay. If the run includes only one assay, files for all samples are included. If the run includes more than one assay, you can specify the assay of interest when you download the CSA.

1. In the menu bar, click **Results ▶ Sample Results**.
2. In **Sample Results** screen, place the pointer over the row of the sample result of interest, then click **CSA**.

Alternatively, you can download CSA files three other ways.

- Click **Results ▶ Sample Results**. Click the sample name of interest. Then, click **⋮ (More Options) ▶ CSA**.
- Click **Results ▶ Run Results**. Click the run name of interest. Place the pointer over the run of interest, then click **CSA**. In the **Download CSA** dialog box that appears, select the assay of interest, then click **Download**.
- Click **Results ▶ Run Results**. Click the run name of interest. Then, click **⋮ (More Options) ▶ CSA**. In the **Download CSA** dialog box that appears, select the assay of interest, then click **Download**.

An XZ compressed TAR archive (TXZ) file is downloaded to the folder that you specified to download files from the browser. This location depends on the browser settings. You can attach the archive to an email to send to Customer Support.

Troubleshoot Genexus™ Software

Observation	Possible cause	Recommended action
Cannot sign in to the Genexus™ Software	You have either forgotten your password or are signed out due to several failed login attempts.	Contact the Genexus™ Software system administrator.
Batch sample import fails	One or more entries in the sample-import spreadsheet contains special characters, lines breaks, unexpected spaces, incorrect entry length, incorrect date formatting, or other formatting errors.	Check each entry for correct formatting, correct any errors, and repeat the import.



Observation	Possible cause	Recommended action
Batch sample import fails <i>(continued)</i>	Blank rows were copied into the sample-import template file from a different source.	Rows that appear empty can contain hidden formatting that conflicts with the import function. Start with a clean sample-import template file, and be careful to copy only those rows that contain actual data.
	The sample import spreadsheet contains a nonunique sample name.	Every sample name in the software must be unique. Ensure that the spreadsheet does not contain any duplicate sample names, and repeat the import. Note that the system check is not case-sensitive, so a sample name of ABC1 conflicts with abc1.
	The headers in the sample import spreadsheet do not match the sample attributes in the software.	The headers must match the sample attributes in the software exactly. Check the headings for spelling or other errors. Map a sample attribute if needed.
Library batch import fails	One or more entries in the library batch import spreadsheet contains special characters, lines breaks, unexpected spaces, incorrect entry length, incorrect date formatting, or other formatting errors.	Check each entry for correct formatting, correct any errors, and repeat the import.
	Blank rows were copied into the library batch import template file from a different source.	Rows that appear empty can contain hidden formatting that conflicts with the import function. Start with a clean library batch import template file, and be careful to copy only those rows that contain actual data.
	The library batch import spreadsheet contains a nonunique Library Batch ID.	Each Library Batch ID in the software must be unique. Ensure that the spreadsheet does not contain any duplicate IDs, and repeat the import. The system check is not case-sensitive. For example, a Library Batch ID of ABC1 conflicts with abc1.
	A sample name entered in the library batch import spreadsheet does not match a sample name listed in the Manage Samples screen.	Ensure that the sample names entered into the spreadsheet are correct and match an existing sample name added to the software.
	The Barcode ID name format does not exactly match the format that is used in the Prepare Library Batch dialog box.	Use the name format following the Barcode ID name format found in the Barcode Set reference lists (Settings ▶ References ▶ Barcode Set), for example: lonDual_0101 through lonDual_0196, or lonHDdual_0101 to lonHDdual_0132.



Observation	Possible cause	Recommended action
Library batch import fails <i>(continued)</i>	An invalid library, control, or panel kit barcode has been entered in the spreadsheet.	Ensure that you have correctly entered a valid kit barcode in the appropriate cell of the spreadsheet.
	The spreadsheet template that you used is from a previous software version.	New fields in the template file can be added with new software versions. Ensure that you download the template file from the current software version.
Cannot upload my panel or hotspots	Issues with BED file format or files do not end in <i>.bed</i> .	Ensure your file is in the correct BED format and has a <i>.bed</i> extension.
Variants tab is missing hotspot entries. Details: The remaining entries are present.	Hotspot BED file contains entries that are incorrectly formatted.	Check that BED file entry is correctly formatted. See the following examples: SNP entry: chr1 2337276 2337277 SVA_322 0 + REF=C;OBS=T;ANCHOR=G AMPL Deletion entry: chr1 201341175 201341180 SVA_497 0 + REF=AGAAG;OBS=;ANCHOR=C AMPL Insertion entry: chr1 236978992 236978992 SVA_621 0 + REF=;OBS=TCTG;ANCHOR=T AMPL
		Confirm the REF values match the actual reference coordinate of hg19.
No information for my loci of interest in the results	The wrong hotspot or BED file is associated with the assay.	<ol style="list-style-type: none"> 1. Check the hotspot and BED files associated with the assay. If either is incorrect, create a new assay. 2. Plan a new run for the sample or sample library with the correct assay. 3. Repeat sequencing of the sample or sample library.
Cannot download run result files	The run failed.	Create an assay with the correct configuration for the samples, then reanalyze the samples.

Other

Observation	Possible cause	Recommended action
MSI QC Failure Details: Error message: "Not enough markers with coverage X"	MSI marker(s) had insufficient coverage to reliably calculate MSI. This is common in NTC samples.	If the sample is not an NTC, contact support (see "Customer and technical support" on page 374).
MSI QC Failure Details: Error message: "RMC Calibration Failure"	Insufficient RMC read coverage for calibration. RMC reagent may have been omitted during library preparation.	Remake library including RMC in the target amplification reaction, then sequence the new library.



Documentation and support

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Related documentation

Document	Publication number
<i>Genexus™ Integrated Sequencer User Guide</i>	MAN0017910
<i>Genexus™ Integrated Sequencer Quick Reference</i>	MAN0017912
<i>Genexus™ Integrated Sequencer Site Preparation Guide</i>	MAN0017918
<i>Genexus™ Purification System User Guide</i>	MAN0018475
<i>Genexus™ Purification System Quick Reference Guide</i>	MAN0018476
<i>Genexus™ Purification System Site Preparation Guide</i>	MAN0018477
<i>Ion Reporter™ Software 5.18 User Guide</i>	MAN0024776
<i>Oncomine™ Precision Assay GX User Guide</i>	MAN0018508
<i>Oncomine™ Comprehensive Assay v3 GX User Guide</i>	MAN0018512
<i>Oncomine™ TCR Beta-LR Assay GX User Guide</i>	MAN0018513

Customer and technical support

Visit thermofisher.com/support for the latest service and support information.

- Worldwide contact telephone numbers
- Product support information
 - Product FAQs
 - Software, patches, and updates
 - Training for many applications and instruments
- Order and web support

- Product documentation
 - User guides, manuals, and protocols
 - Certificates of Analysis
 - Safety Data Sheets (SDSs; also known as MSDSs)

Note: For SDSs for reagents and chemicals from other manufacturers, contact the manufacturer.

Limited product warranty

Life Technologies Corporation and/or its affiliate(s) warrant their products as set forth in the Life Technologies' General Terms and Conditions of Sale at www.thermofisher.com/us/en/home/global/terms-and-conditions.html. If you have any questions, please contact Life Technologies at www.thermofisher.com/support.

Glossary

Genexus™ Software definitions

adapter dimer (ISPs)

ISPs that carry DNA insert length of less than 8 bp.

aligned read length

The aligned length of a read at a given accuracy threshold is defined as the greatest position in the read at which the accuracy in the bases up to and including the position meets the accuracy threshold. For example, the AQ17 length of a read is the greatest length at which the read error rate is 2% or less. The "perfect" length is the longest perfectly aligned segment. For all of these calculations, the alignment is constrained to start from position 1 in the read—that is, no 5' clipping is allowed.

aligned reads

The number of bases covered by reads aligned to the reference sequence.

AQ score

An alignment quality (AQ) score defines read accuracy when compared to the reference sequence. The discrepancy can be biological (real variant) or technical (sequencing error). For example, an AQ score of 17 represents 2% aligned read error rate, while an AQ score of 20 represents 1% aligned read error rate when compared to the reference sequence.

archive

The process that occurs when runs on the Ion Torrent™ Genexus™ Integrated Sequencer server reach the maximum time to be on the server and the run is eligible for deletion from the server.

average base coverage depth

The average number of reads of all targeted reference bases.

average base read depth

The average number of reads of all targeted reference bases that were read at least once.

backup

The process of data being copied to an external storage device or system from the Ion Torrent™ Genexus™ Integrated Sequencer. Backups of data are done each night, based on backup settings, and data that is backed up is not deleted from the Genexus™ Integrated Sequencer. When data is backed up, the same data remains on both the server for the sequencer and on the external storage system.

BAM file

A BAM (binary alignment map) file (.bam) is the binary version of a SAM (sequence alignment map) file (.sam). A SAM file is a tab-delimited text file that contains sequence alignment data. A BAM file contains aligned reads sorted by reference location.

barcode

A barcode is a machine-readable code in the form of numbers and a pattern of parallel lines of varying widths, printed on and identifying a product.

There are several applications for barcodes. Libraries can be molecularly barcoded with unique nucleic acid sequence identifiers. Library barcodes are used during data analysis to sort the sequencing results from sequencing reactions that contain combined libraries. Chips and sample tubes also contain unique numeric barcodes that aid in the setup of the experimental analysis workflow.

base calling

Base calling is the process by which raw data from the sequencing instrument is converted to nucleotide sequences. This is performed by base calling software that is usually run from the instrument itself.

basecalling input file

Signal processing input files are converted to a single condensed basecalling input file that represents the processed signal. Basecalling input files are required files for basecalling.

bead loading

The percentage of chip wells that contain live ISPs. The percentage value considers only potentially addressable wells. Bead loading is calculated as number of loaded ISPs divided by the number of potentially addressable wells.

bp

Abbreviation for "base pair(s)".

cellularity (%)

The percentage of tumor cells in a given sample.

BED file

Browser Extensible Data file—BED file—defines chromosome positions or regions.

CNV

Copy number variation (CNV) is the variation in copy number of any given gene between two samples. CNV is a phenomenon in which sections of the genome are repeated and the number of repeats in the genome varies between individuals in the human population.

contig

A contig (from contiguous) is a set of overlapping DNA segments that together represent a consensus region of DNA.

control sequence

Control nucleic acid sequences can be added to DNA or RNA samples to facilitate post-sequencing data analysis. Two types of control sequences can be used during sample preparation. ERCC RNA Spike-In Mix is used with RNA samples to achieve a standard measure for data comparison across gene expression experiments. Ion AmpliSeq™ Sample ID Panel, comprised of nine specially designed primers, can be added prior to template amplification to generate a unique ID for each sample during post-sequencing analysis.

CSV file

A comma-separated values (CSV) file is a delimited text file in which each line represents a data record with information fields separated by a comma. A CSV file stores tabular data (numbers and text) in plain text. Each line of the file is a data record.

CSV files are easily opened using spreadsheet software, such as Microsoft™ Excel™ or Apache® OpenOffice™ Calc, where each comma-separated field is listed in a separate column.

DAT file

A DAT file is a generic data file. It contains data that may be opened or referenced by a specific application. While some DAT files can be opened directly, most contain program data that is referenced the program when it is running. Therefore, most DAT files are not meant to be opened manually.

empty wells

Wells on a chip that do not contain an ISP.

enrichment (%)

Predicted number of live ISPs that have a key signal identical to the library key signal. The percent enrichment value reported is the number of loaded ISPs that are library ISPs, after taking out the test fragment ISPs.

FASTA file

A FASTA file is a text-based format for representing either nucleotide sequences or peptide sequences, in which base pairs or amino acids are represented using single-letter codes. A sequence in FASTA format begins with a single-line description, followed by lines of sequence data.

FASTQ file

A FASTQ file is a text-based format for storing both a biological sequence (usually nucleotide sequence) and its corresponding quality scores. Both the sequence letter and quality score are each encoded with a single ASCII character for brevity.

FD (flow disruptiveness)

A data filtering parameter that is used instead of INDEL, SNP, and MNP.

final library reads

Number (and percentage) of reads, passing all filters that are recorded in the unmapped BAM file. This value may be different from the total number of reads located in the Library Summary Section due to specifications associated with read trimming beyond a minimal requirement resulting in total number of reads being slightly less than Final Library Reads.

flow

A flow is the event of exposing a chip to one particular dNTP (deoxyribonucleotide triphosphate), followed by a washing step.

flow order

The order in which a chip is exposed to each particular dNTP. The default Samba flow order consists of a 32-base sequence, repeated. This flow order resists phase errors by providing opportunities for out-of-phase molecules to catch up and is designed to sample all dimer (nucleotide pair) sequences efficiently. Samba is the default flow order because it improves sequencing accuracy for longer reads by resisting phase errors.

flow transfer

Progress of the sequencing run expressed as number of total flows completed. For example, a sequencing run set to 500 flows shows 250/500 flows in the status column of the **Runs in Progress** table when the sequencing run is half completed.

fusions

A target technique used for detection and annotation of gene fusions (or translocation of genetic material) in samples.

hotspots file

A BED or a VCF file that defines regions in the gene that typically contain variants. Specifying a hotspots file to use in a run enables the Torrent Variant Caller module to identify if a specific variant is present or absent. A hotspot file instructs the Torrent Variant Caller to include these positions in its output files, including evidence for a variant and the filtering thresholds that disqualified a variant candidate. A hotspot file affects only the variantCaller plugin, not other parts of the analysis pipeline.

If you don't specify a hotspots file, the software tells only the difference between your sequence and the reference genome.

IA

Isothermal application (IA) is a template preparation technique that uses nonemulsion isothermal reaction to clonally amplify DNA onto an ISP surface.

INDEL

INDEL is an abbreviation used to designate an insertion or deletion of bases in the genome of an organism.

intermediate files

Intermediate files contain information used for debugging runs.

ISPs

Ion Sphere™ Particles (ISPs) are particles that contain bound copies of a single (ideally) DNA fragment amplified during template preparation.

key signal

Average 1-mer signal in the library key.

library ISPs

Live ISPs that have a key signal identical to the library key signal.

LIMS metadata

Laboratory Information Management System (LIMS) software is used for recording sequencing metadata. Entered text is associated with the Planned Run and can be extracted using APIs for LIMS consumption.

live ISP

An ISP with a signal of sufficient strength and composition to be associated with the library or test fragment key.

low quality ISP

An ISP with a low or unrecognizable signal.

library key

A short known sequence of bases used to distinguish a library fragment from a test fragment (for example, "TCAG").

MAPD

Acronym for median absolute pairwise difference. Assuming that adjacent amplicons in the genome most likely have the same underlying copy number in a sample, the difference between the $\log_2(\text{read count ratio})$ values against the reference baseline for all adjacent amplicons contains information for the noise level of the data. The median of the absolute values of all such difference in $\log_2(\text{read count ratio})$ is the measure for how informative the results fare for copy number estimates.

mapped reads

Sequencing reads that have been mapped to the reference sequence.

mean raw accuracy

Average raw accuracy of 1-mers at a specific position in the read.

microsatellite instability

Microsatellite instability (MSI) is a form of genomic instability in the replication of repetitive DNA that leads to the appearance of multiple alleles at microsatellite loci. MSI markers can be detected with some DNA and DNA and Fusions assays.

missense SNV

A point mutation that changes the amino acid of the respective protein. SNV is an acronym for single nucleotide variation, which means at one base there is a difference.

MNP

Multiple nucleotide polymorphism (MNP) is a genetic mutation in an allele that differs from the reference allele of the same length by >1 nucleotide.

nonsense SNV

A point mutation that changes one of the 20 amino acids into a stop codon, hence a shorter or unfinished protein product. SNV is an acronym for single nucleotide variation, which means at one base there is a difference.

on target reads

Sequencing reads mapped to any target region of the reference. A read is considered to be on target if at least one aligned base overlaps a target region. A read that overlaps a targeted region but where only flanking sequence is aligned, for example, due to poor matching of 5' bases of the read, is not counted.

primer dimer

A primer dimer (PD) is a potential by-product in PCR, a common biotechnological method. As its name implies, a PD consists of primer molecules that have attached (hybridized) to each other because of strings of complementary bases in the primers

primer dimer ISP

An ISP that carries an insert length of less than 8 base pairs.

read length

The length of called reads measured in base pairs.

read

The sequence of a section of a unique fragment obtained after the end of the sequencing process.

reference library

A consensus nucleotide sequence that represents the genome of a particular species. The results from a sequencing run are compared to the reference library to identify sequence variants.

SAM file

A SAM (Sequence Alignment Map) file is a tab-delimited text file that contains sequence alignment data. It stores biological sequences aligned to a reference sequence.

sample

Genetic material from one source (for example, DNA from one individual).

signal processing input files

Signal processing input files consist of the raw voltage measurement data collected during the sequencing run. These files are required to reanalyze the run from signal processing.

SNP

Single nucleotide polymorphism (SNP) is a genetic mutation in an allele that differs from the reference allele of the same length by one nucleotide.

SSH

SSH, also known as Secure Shell or Secure Socket Shell, is a network protocol that gives users, particularly system administrators, a secure way to access a computer over an unsecured network. SSH also refers to the suite of utilities that implement the SSH protocol.

structural variants

Genetic mutations that cause a change in the organism's chromosome structure, such as insertions, deletions, copy number variations, duplications, inversions, and translocations.

target base coverage

Summary statistics for targeted base reads of the reference. A base covered by multiple target regions is counted only once per sequencing read.

target regions file

A BED file that specifies all of the regions that a panel represents such as the amplified regions that are used with target sequencing. The complete software analysis pipeline, including plugins, is restricted to only these specified regions instead of analyzing the entire reference library.

test fragment

A test fragment (TF) is a known nucleotide sequence that is used to monitor system characteristics.

test fragment ISPs

Live ISPs with a key signal that is identical to the test fragment key signal.

test fragment key

A test fragment key (TF key) is the nucleotide sequence that is used to identify test fragment reads.

TF key peak counts

Signal strength of the first three bases of the TF key.

total reads

Total number of filtered and trimmed reads independent of length reported in the output BAM file.

TSV file

A tab-separated values (TSV) file is a tab-delimited file that is used with spreadsheet software. TSV files are essentially text files, and the raw data can be viewed by text editors, though they are often used when moving raw data between spreadsheets. See also *VCF file*

uBAM file

An unmapped BAM (uBAM) file is a variant form of the BAM file format in which the read data does not contain mapping information. This is basically an "off-label" use of the BAM format (which was specifically designed to contain mapping information) that is used for data management reasons. It allows you to attach metadata to the reads from as early on in the analysis process as possible.

unaligned reads

Nucleotide bases covered by reads that are not aligned to the reference.

uniformity of base coverage

The percentage of bases in all targeted regions (or whole genome) covered by at least 0.2X the average base coverage depth.

usable sequence / usable reads

Usable reads consist of library ISPs that pass the polyclonal, low quality, and primer dimer filter.

VCF file

A variant call format (VCF) file specifies a variant of interest and its location. This file stores the differences between the BAM file and the reference file.

wells with ISPs

Number of wells that were determined to be positive for the presence of an ISP within the well. Wells containing ISPs have a delayed pH change due to the presence of an ISP slowing the detection of the pH change from the solution.

XLS file

Files that use the .xls extension. XLS files can be created by Microsoft™ Excel™ and other similar spreadsheet programs.

