

Genexus™ Software 6.2.1

Release Notes

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Revision History

Revision	Date	Description
D.0	31 May 2022	Added issue 25474. Potential pool imbalance when selecting UDG option for AmpliSeq DNA and AmpliSeq DNA&RNA Assays.
C.0	17 January 2022	Updated with issue 17803. Run reports for runs that are started on or completed on 26–31 December display the incorrect year.
B.0	07 January 2021	Updated release notes for Ion Torrent Genexus™ Software 6.2.1 and for Known Issue 15753.
A.0	07 July 2020	New release notes for Ion Torrent Genexus™ Software 6.2.

Release Notes Overview

This document outlines major enhancements as compared with the existing software platforms, new features and known issues to be fixed in subsequent releases.

Genexus 6.2.1 Software Overview

Genexus™ Software 6.2.1 is a recommended upgrade if you are using Genexus™ Software 6.2. The upgrade is mandatory if you are using Genexus™ Software 6.0.

Genexus™ Software 6.2.1 improves the stability of the Ion Torrent Genexus™ Integrated Sequencer. For more information, see Issues fixed in Genexus™ Software 6.2 and 6.2.1 on page 4.

Genexus™ Software Overview

Genexus™ Software is the fully integrated software for the Genexus™ System that includes components to run the instrument and provide access to the Genexus™ System through touch screens. The browser-based component provides users with tools to plan runs, create and manage runs and assays, view results, prepare customized reports, manage users, and complete other administrative tasks. The touch screens on the instrument provide access to instrument operations, and to run plans created in the browser-based component. Together, the software components combine much of the functionality inherited from the existing Torrent Suite™ Software and Ion Reporter™ Software to provide a sample-to-report NGS solution.

In addition to offering an end-to-end user experience, the software presents major features for ease of use, cybersecurity, quality control and interoperability. While Genexus™ Software includes most downstream features of Ion Reporter™ software, it continues to allow uploads of data to, and analysis in, Ion Reporter™ Software on Connect or Ion Reporter™ Server.

Key Enhancements in Genexus™ Software 6.2

Performance improvements

Genexus™ Software 6.2 provides a faster completion time for analysis pipelines and faster rendering of user interface views.

Reanalyze samples

The new reanalysis feature allows you to reanalyze samples that have previously been sequenced and analyzed. Reanalysis can be started from basecalling or alignment with an assay that has been used previously or with a different, compatible assay. Reanalysis using a new or a modified assay allows analysis parameter tuning and assay implementation.

Multi-Lane IQ/PQ Assays

This feature allows you to plan a verification (PQ/IQ) run for up to four chip lanes. Multi-lane IQ/PQ runs allow for comprehensive testing of the fluidic system in a single run. Verification run results are displayed per lane, and the run information includes Run QC, Templating QC, Control QC and Library QC. Results also contain reagent information such as part number, lot number, and expiration date.

Genexus™ Software 6.2 improvements to the Results screens

The user interfaces and views of the Sample Results and Assay Metrics screens are more user friendly and responsive than in the previous software version.

Multi-sample comparison and visualization

This feature allows you to compare and visualize variants across multiple samples and compare and plot sample results over time. You can select quantitative attributes to add to charts. For example, you can visualize the **Allele Fraction** for **single nucleotide variants (SNVs)** across samples. Samples analyzed with the OncoPrint™ TCR Beta-LR Assay GX assay can only be compared against other samples analyzed using the same assay.

Save and sequence unused libraries

The plate well locations of the leftover barcoded libraries from a completed sample run are now shown in the **Run Summary** tab. Access the **Run Summary** in the **Results / Run Results** screen. Click a sample name to open the **Run Summary** tab. You can retain unused libraries and sequence and analyze them when you plan a library run in the future.

New assays enabled

- **OncoPrint™ Comprehensive v3 – DNA and Fusions:** Detects and annotates low frequency somatic variants (SNPs, INDELs, and CNVs) from targeted DNA libraries, as well as gene fusions from targeted RNA libraries, from the OncoPrint Comprehensive Assay v3 using Ion AmpliSeq™ technology.
- **OncoPrint™ Precision for Tissue – DNA and Fusions:** Detects and annotates low frequency (to 0.5% limit of detection) somatic variants (SNPs, INDELs and CNVs) from targeted DNA libraries, as well as gene fusions from matching targeted RNA libraries using Ion AmpliSeq™ HD technology. This assay is compatible with DNA and RNA purified separately from tissue samples.
- **OncoPrint™ TCR Beta-LR GX:** Analyzes human T-cell receptor beta chains amplified from non-FFPE RNA/cDNA using Framework 1 and Constant Gene targeting primers on the GX5™ chip. The long amplicon (330 bp average length) provides coverage of all three CDR domains to enable detection of polymorphisms within the T-cell receptor variable gene. This assay reports the frequency and sequence features of clonotypes and performs secondary analyses of repertoire features. This assay uses the IMGT database of variable, diversity and joining genes as a reference.

No template control (NTC)

Genexus™ Software 6.2 eliminates the need for manual creation of NTC samples in the system. You can plan a run with one NTC sample per assay and the NTC QC metrics are reported separately. When planned and sequenced with an NTC, run results contain relevant QC metrics for the NTC sample. The software assigns input plate location for NTC samples and prevents allocation of chip space to an NTC sample. This improves ease of use and efficiency of run planning.

In-line quality control (QC) metrics

In-line QC metrics are now reported in the QC results. These metrics characterize insufficient sample input and/or poor sample quality. In-line QC is accomplished through the addition of a set of six control amplicons (covering a range of amplicon lengths) and spike-in nucleic acid into sample library preparation. Measuring the ratio of spike-in to endogenous reads from the six amplicons helps identify insufficient sample input and/or poor sample quality.

Issues fixed in Genexus™ Software 6.2 and 6.2.1

Issue number	Description
1031	Cooling capacity was limited because the second radiator fan on the instrument did not function with Genexus™ 6.2 Software. The fan works automatically upon upgrade to Genexus™ Software 6.2.1, thereby increasing increase the cooling capabilities of the instrument.
995	Intermittent message corruption for an internal software command that causes a motor to move caused some runs to be terminated in Genexus™ Software 6.2. This issue is fixed in Genexus™ Software 6.2.1 with the addition of a retry to send the message again in case of message corruptions.
848	A defect in Genexus™ Software 6.2 was leading to an issue called "salt bridging" which caused liquid to leak out of the squid over time and dry on the chip. Genexus™ Software 6.2.1 adds Safestate, a procedure that prevents liquid from leaking out of the squid when a chip is moved away from the sequencing position. This procedure was added is run any time the chip is moved from the sequencing position.
1044	The chip shuttle on the Genexus™ System has a solenoid pin that holds the shuttle in place at the pipette position. On rare occasions this pin fails to retract, which blocks movement of the chip shuttle. This issue is fixed in Genexus™ Software 6.2.1. If this issue occurs, the shuttle is shifted, the pin retracts, and the movement is retried.
11951	The status-progress-bar for a sequencing run in the Monitor > Instrument view of Genexus™ software was stuck at an intermediate step even when the run was successfully completed. This issue is fixed in Genexus™ Software 6.2.
11684	The table that shows Consumables Installed Ready To Use in the Review step of the sample run plan creation sometimes did not list the panel kit as a required consumable. This issue is fixed in Genexus™ Software 6.2.
11371	The Variation Matrix Summary Filter chain did not work as expected for custom AmpliSeq™ HD assays. This issue is fixed in Genexus™ Software 6.2.
11762	The Parameters step for users who create an assay and use the Fusions assay template (in the Create Assay > Fusions menu) incorrectly included Tumor Mutational Burden (TMB) parameters. This issue is fixed in Genexus™ Software 6.2.
11769	Some of the variant filters that Ion Reporter™ Software provided were missing in Genexus™ Software 6.0. This issue is fixed in Genexus™ Software 6.2.
11455	The variants shown in downloaded Summary.tsv and CNV.tsv files did not match results displayed in Genexus™ Software 6.0. This issue is fixed in Genexus™ Software 6.2.
12013	When users created a custom assay in the Assays > Create Assays > DNA Somatic template, the resulting parameters were the Germline parameters. This issue is fixed in Genexus™ Software 6.2.
11936	The value of the MAPD parameter for a given assay appeared as -2 when viewed in the Manage Assays / Assay screen even when the value was not set during assay creation. This issue is fixed in Genexus™ Software 6.2.
11735	Interactive features for the CoverageAnalysis plugin results did not work. This issue is fixed in Genexus™ Software 6.2.
11674	For sample runs, library runs, and verification run planning, incorrect consumable kit barcodes were listed in the Lab Report and info.csv file. This issue is fixed in Genexus™ Software 6.2.
11561	Sorting on the Analysis Version column in the Assays step of the Plan Sample Run screen caused a system error. This issue is fixed in Genexus™ Software 6.2.
11446	The Variation Classification variant filter option sometimes resulted in incorrect results and did not reflect custom classifications that were created. This issue is fixed in Genexus™ Software 6.2.

Issue number	Description
11387	The user account was suspended even when incorrect login attempts occurred past the expiration time that was set under User Management > Policies > Suspension policies . This issue is fixed in Genexus™ Software 6.2.
8128	When a user removed assays from a run plan, the recalculation and allocation of chip lanes to assays and samples did not occur as expected. This issue is fixed in Genexus™ Software 6.2.
12161	The SNV and INDEL type variants were not listed when these variants had multiple Absent calls. This issue is fixed in Genexus™ Software 6.2.
12167	The Software Updates screen showed the instrument name as Valkyrie. This issue is fixed in Genexus™ Software 6.2.
11168	When the system recovered or rebooted due to power or workflow failure, the results of the ongoing analysis at the time of the failure were sometimes corrupted. In addition, subsequent analysis sometimes failed at basecalling. This issue is fixed in Genexus™ Software 6.2.

Known issues in Genexus™ Software 6.2

Issue number	Issue Summary	Impact and Mitigation
25474	<p>The Uracil DNA Glycosylase (UDG) Treat DNA setting is used to clean up deaminated Cytosine bases from degraded samples. If you enable the UDG Treat DNA parameter in an assay, you may observe read imbalance in the Barcodes with Reads Reported table. Read imbalance can occur because the on-instrument dilution of input sample nucleic acid is impacted by this setting.</p>	<p>This issue affects assays for DNA or DNA and Fusions if you enable the UDG Treat DNA parameter in assay creation or when you copy, then edit an assay. The issue can result in over dilution of nucleic acid prior to library prep for AmpliSeq, and over dilution of the second pool in AmpliSeq and AmpliSeq HD two pool assays, leading to pool imbalance in the final reads.</p> <p>Mitigation: If possible, do not enable the UDG Treat DNA parameter for AmpliSeq HD assays with two or more pools and for AmpliSeq assays. Manually dilute the DNA sample if UDG treatment is necessary. Contact an FAS for help.</p>
17803	<p>The dates for runs that are started on or completed on 26–31 December display the incorrect year. The date that is shown is one year later than the actual date. For example, for a run started and completed on 2021-12-30, the started on and completed on date is incorrectly shown as 2022-12-30.</p>	<p>This issue affects all runs that are started on or completed on 26–31 December. The incorrect date is shown in the Lab Report PDF, in the Run Report PDF, in the Run Summary file (the info.csv file), in the Sample Results screen, and in the Run Results screen.</p>
15753	<p>When you copy the Oncomine™ Myeloid Assay GX (using the Manage Assays > Copy workflow) to modify system-installed assay settings the PCR denature temperature parameter from the system-installed assay is not passed to the copied assay.</p>	<p>This issue impacts copied assays created from the factory shipped assay. See “Mitigation procedure for Known Issue 15753: Copy and edit an assay” on page 9 to copy and edit the Oncomine™ Myeloid Assay GX in Genexus™ Software 6.2. Contact your Field Bioinformatics Specialist (FBS) for assistance with modifying any system-installed assay settings if you need further assistance.</p>
13234	<p>For a multi-assay run, when the Customer Support Archive (CSA) plugin finishes execution for one of the assays, the status of the CSA plugin for the other assays also erroneously gets updated to a status of Completed.</p>	<p>This issue impacts only multi-assay runs. Even though CSA plugin status for other assays is marked Completed prematurely, the plugin results eventually become available when the plugin execution finishes. This issue will be fixed in an upcoming release of the software.</p>
11310	<p>Analysis for any assay that uses a non-hg19 reference genome fails.</p>	<p>This issue only impacts runs that use a non-hg19 custom reference. Even though the analysis fails with an error, users can still get mapped BAM files for downstream analysis. In order to avoid the error, users can run custom reference workflows using the Generic Sequencing Application assay type which completes with mapped BAM files. This issue will be fixed in an upcoming release of the software.</p>

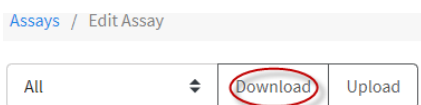
Issue number	Issue Summary	Impact and Mitigation
12692	Missing variant label and legend chart options for charts generated with the Multi-sample comparison and visualization feature described in the Key Enhancements in Genexus™ Software 6.2 section above.	This is a chart customization issue that does not affect sequencing results. This issue will be fixed in an upcoming release of the software.
13222	Deck loading instructions jump between sections before completion of each section.	This issue may result in a suboptimal user experience in setting up the deck. It does not, however, cause any misplacement of deck consumables. This issue will be fixed in an upcoming release of the software.
11298	The session time-out on the Genexus™ Integrated Sequencer touchscreen does not work after a user aborts a run or after the post-run clean stage.	This issue is only applicable when a user has aborted a run or post-run clean. Also, it only affects the instrument interface and not the browser-based Genexus™ Software 6.2. The session time out works if the run is allowed to complete. This issue will be fixed in an upcoming release of the software.
12378	Sporadic failure while importing an assay.	This is an intermittent failure and seems to get resolved when user tries to import the same assay again. This issue will be fixed in an upcoming release of the software.
13213	An incorrect message displayed when new user is prompted to change password.	This is a software user interface issue and does not impact the functionality for logging in as a new user. This issue will be fixed in an upcoming release of the software.
13199	An incorrect start time is displayed for reanalyzed runs and assays.	This is a software user interface issue and does not impact the functionality of the reanalysis feature. The run start time is accurate for the original run but fails to update sporadically when the run is reanalyzed. This issue will be fixed in an upcoming release of the software.
13233	Users may experience a frozen popup window when adding a new user from Settings > User Management .	In Settings > User Management , when the Create User dialog window freezes, an administrator-level user must manually close the window, click + Add New and re-enter the new user details. This issue will be fixed in an upcoming release of the software.
13192	The Download files menu fails to open intermittently.	Clicking on the Download files link from the Sample Results screen fails to open the Download files window but clicking a second time opens the window. This issue will be fixed in an upcoming release of the software.
13190	For some assays, when a user creates a library batch that does not include a no template control (NTC), then edits the library batch to include the NTC, unique barcodes are not assigned.	Although unique barcodes are not assigned in this case, users can change and correct the barcode assignments when editing a library batch. This issue will be fixed in an upcoming release of the software.

Issue number	Issue Summary	Impact and Mitigation
13223	The Forgot password user link in the sign in screen forces the user to set a new password twice.	This issue does not block the main functionality of resetting a password. This issue will be fixed in an upcoming release of the software.
13318	Plugins applied in Genexus™ Software 6.2 fail to run on samples sequenced with Genexus™ Software 6.0.	This issue will be resolved in an upcoming release of the software.
13238	The minimum cellularity threshold for a given assay should prevent only samples with a cellularity lower than the threshold from being added to a run plan. However, samples with passing criteria are also not added to a run plan.	To resolve this issue, the user can remove the cellularity threshold from the assay definition. The sample cellularity should still be specified when relevant (and works as expected) for downstream analysis. This issue will be resolved in an upcoming release of the software.
13593	Spearman's correlation coefficient is calculated incorrectly when performing a pairwise sample comparison in the OncoPrint™ TCR Beta-LR GX assay.	This issue will be resolved in an upcoming release of the software
13478	If a user clicks the link for the ExAC browser from variant annotation in the View Annotation Sources window, the following message is displayed: "The ExAC browser is no longer available. ExAC data is available in the gnomAD browser or can be downloaded from gs://gnomad-public/legacy . View this page in the gnomAD browser. You will be automatically redirected in 24 seconds. "	To resolve this issue, click the updated gnomAD browser link or wait to be redirected. This issue will be resolved in an upcoming release of the software
13362	For an imported assay, the chip type value is NA in the audit record.	This issue has minimal impact. There is only one chip, GX5, in Genexus Software 6.2 so there is no ambiguity as to the chip type used. The imported assay retains the correct value of chip type and performs as expected. This issue will be resolved in an upcoming release of the software.

Mitigation procedure for Known Issue 15753: Copy and edit an assay

The Oncomine™ Myeloid Assay GX parameter sets and the associated plugins are installed from **Thermo Fisher Connect** in Genexus™ Software using the **Settings > Software Updates** menu or from a media storage device (USB) by qualified support personnel (FSE – Field Service Engineer).

1. In the menu bar, click **Assays > Manage Assays**.
2. In the **Manage Assays** screen, in the **Actions** column in the row of an assay that you want to copy, click **Copy**.
3. In the **Copy Assay** screen, proceed through the workflow steps. When finished, enter a new name in the **Assay Name** field and a short name in the **Assay Short Name** field for the copied assay, then click **Save**.
4. In the **Manage Assays** screen, in the **Actions** column in the row of your copied assay, click **Edit**, then in the **Edit Assay** screen proceed to the **Parameters** step.
5. At the top of the screen in the **Parameters** step, select **All** from the dropdown list, then click **Download**.



A ZIP folder that contains the **all.json** file for the assay is downloaded to your local storage.

6. Open the **all.json** file using a text editor, then search for “**amplifyTargetPCRProfile**”.

The **all.json** file contains two matches for the “**amplifyTargetPCRProfile**” search entry. A text string “**value": "[[990],[xxx],x],[[990,xxx],[xx,xxx],xx],[[xxx],[xx],x]]”** should follow each instance of the matched text.

7. Replace each instance of the 990 value with **980** in the text string (a total of four instances), then save the edited **all.json** file to your local storage.
8. Return to the **Edit Assay** screen in the Genexus™ Software, then at the top of the screen in the **Parameters** step, click **Upload**.
9. Under **Advanced Parameter Configuration**, click **Select files**, select the edited **all.json** file, then click **Open**.

When upload is complete, **Parameter file uploaded successfully** message is shown.

10. Proceed through the rest of the workflow steps, then click **Save** to save the assay.
11. In the **Manage Assays** screen, in the **Actions** column in the row of your edited assay, click **Lock** to lock the assay.
12. In the **Actions** column, in the row of your edited assay, click **Download Parameters**.
13. Open the **all.json** file and verify that four **980** replacements in the text string are included in the file.

The modified assay is now ready for use in a run.

System Requirements

Genexus™ Software 6.2 has a browser-based interface that can be viewed using Google™ Chrome™ version 64 and above (version 64 has been tested) and is best viewed with a 1440 x 900 screen resolution. The software is accessed on an independent client computer with a private web server.

Operating system: Ubuntu® 18.04 LTS with Apache web service to provide web pages to client computers. Installation procedure

Genexus™ Software 6.2 is installed on top of the manufacturing software (version 5.100.2). The software is installed from a media storage device (USB) by a qualified Field Service Engineer (FSE).

Package and version information

Release Date	January 2021
Release Version	6.2.1

Software packages versions in 6.2.1

These tables list the versions for each of the latest packages and plugins for Genexus™ Software.

Packages

Package	Version
ts-assaydev-analysis514	6.2.1
ts-assaydev-annotdelta514	6.2.1
ts-assaydev-crawler	6.2.1
ts-assaydev-factory-template	6.2.1
ts-assaydev-genxscriptupdates	6.2.1
ts-assaydev-genxupdates	6.2.1
ts-assaydev-igvservlet	6.2.1
ts-assaydev-iruploader	6.2.1
ts-assaydev-manager	6.2.1
ts-assaydev-ocrp514	6.2.1
ts-assaydev-ovat-plugin514	6.2.1
ts-assaydev-seeddata	6.2.1
ts-assaydev-server514	6.2.1

Package	Version
ts-assaydev-ui	6.2.1
ts-config	6.2.1
ts-deeplaser	6.2.1
ts-fonts	6.2.1
ts-mongo	6.2.1
ts-softwareupdate	6.2.1
ts-tomcat8	6.2.1
ts-reference-delta	6.2.1

Plugins

Plugin	Version
customersupportarchive	6.2.1
coverageanalysis	6.2.1
molecularcovrageanalysis	6.2.1
sampleid	6.2.1

The information in this guide is subject to change without notice.

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