

## **Converge™ Software v2.1 Release Notes** **Case Management, Kinship and NGS Data Analysis Modules for STR, SNP and Mito**

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### PRODUCTS AFFECTED

- Converge Software v2.1
  - HID Genotyper Plugin v2.1
  - Next-Generation Sequencing Module for STR, SNPs and Mito - Converge-NGS-1.1.zip
  - Upgrade Installer Files - ConvergeUpgrade-2.1-0.x86\_64.rpm, upgradecvg
  - Precision\_ID\_Panel\_Definitions.zip
- Precision ID mtDNA Panels
  - Precision ID mtDNA Whole Genome Panel
  - Precision ID mtDNA Control Region Panel
- Precision ID SNP Panels
  - Precision ID Ancestry Panel
  - Precision ID Identity Panel
- Precision ID STR Panel
  - Precision ID GlobalFiler™ NGS STR Panel v2
- Precision ID Manual Library Preparation Kits
- Precision ID DL8 Chef Kit
- NGS Run Template on TSS 5.10
  - Run Template - Precision ID mtDNA Control Region Panel - S5
  - Run Template - Precision ID mtDNA Whole Genome Panel - S5
  - Run Template - Precision ID Ancestry Panel - S5
  - Run Template - Precision ID Identity Panel - S5
  - Run Template - Precision ID GlobalFiler STR Panel - S5

### SOFTWARE OVERVIEW

Converge v2.1 is a third release of a multi-phased product suite for Next Generation Software Platform. This release of Converge Software provides analysis support for all current NGS Panels (STR, mtDNA, ancestry, identity and YSNPs), in addition to Paternity and Kinship testing. The software is built on a modular platform offering data storage and workflow capabilities, genotype calling, in addition to downstream tertiary sample analysis tools after genotyping.

### KEY FEATURES IN CONVERGE v2.1:

- Analysis Modules - NGS analysis modules integrates with the Torrent Suite™ Software v5.10 for automatic import of sample data. The following new modules are available.
  - NGS mtDNA module for mitochondrial DNA- Perform Secondary Analysis of Mito sample data.
    - EMPOP classification and haplogroup prediction using April 2017 EMPOP release..
    - IGV Developed version – 1.01b
    - Mito Variant Caller version - 1.01b

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- NGS SNP module for single nucleotide polymorphisms - Perform Secondary and Tertiary analysis of SNP sample data.
  - Torrent Variant Caller version – 5.2.25.
  - Ancestry predictions based on ALFRED and Identity likelihood based on 1000 Genomes Phase 3.
  - Y Haplogroup prediction based on Phylotree ISOGG 2014
  - Exclusion of SNPs and reanalysis of Tertiary analysis workflow in Converge.
- Profile Dashboard—Manage NGS and/or GeneMapper™ ID-X Software profiles.
- Sample Dashboard—Manage NGS sample information.
- Batch Dashboard—Manage NGS Batches, Create a New Batch for a run.
- Manage Applications—Install and uninstall applications, such as the NGS modules – STR, SNPs, Mito. (Manage Applications is located in the Admin Dashboard under Global Settings.)

## SYSTEM REQUIREMENTS CONVERGE v2.1:

- TSS v5.10 / S5 / S5XL™ or Ion GeneStudio™ Series Sequencer
- Converge Software Server & its components
  - Dell™ PowerEdge™ T130 Tower Server, motherboard v2 or later
  - Red Hat™ Enterprise Linux™ operating system
  - Apache™ Tomcat™ application server that runs on Converge software
  - PostgreSQL database server that stores the data for the server and software
  - Google™ Chrome™ browser
  - Automatic configuration of IP, domain name service (DNS), and Windows internet name service (WINS) settings via dynamic host configuration protocol (DHCP)
- Converge Software Server Specifications
  - Processor - Intel™ Xeon™ Processor E3-1270 v6, 3.8 GHz, 8M cache, 4C/8T,turbo (72 W)
  - Memory - 16 GB of memory (2 × 8 GB), UDIMM, 2400 MT/s, Single Rank, x8 Data Width, DVD ROM, SATA, Internal
  - Hard Drive (2) - 2 TB 7.2 K RPM NLSAS, 12 GB/s, 3.5-in cabled hard drive (RAID1)
  - Data Storage - RAID 1; PERC H330 Integrated Controller for 3.5-inch cabled hard drive
  - Operating System - Red Hat™ Enterprise Linux™ operating system
  - Browser - Google Chrome™ 66 or later
- Recommended Software (not provided)
  - Adobe™ Acrobat Reader
  - Microsoft Excel
- Verified Converge v2.1™ software workflow on Google Chrome™ and MAC Safari browsers.

## INSTALLATION / UPGRADE:

Refer to Converge Software v2.1 SETUP AND REFERENCE GUIDE - Publication Number 100039539, Rev D for following instructions

- Initial setup and configuration of Converge Software Server and Converge Software.
- Managing the Converge™ Software Server and licenses.
- In addition, following sections covers Upgrade and Fresh Install workflow plus enhanced troubleshooting section.
  - Appendix A - Troubleshooting Server networking, Password Issues, Access to log files, Restart Services, Reset IP address, Account Configuration and Dell T110 USB recognition.
  - Appendix B - Upgrade to Converge v2.1 on Dell T110 and T130 Appliance Servers.
  - Appendix C - Fresh Installation of Converge v2.1 on Dell T130 Appliance Sever.

Steps below provide additional reference links for TSS upgrade to 5.10 and supplemental files that need to be downloaded prior to starting an end to end run from TSS and generating a batch file on Converge™ Software.

NOTE: In general, all the S3 Chrome Browser links provided below, downloads files to “Downloads” folder in your user account. When the download finishes, you'll see the linked file at the bottom of your Chrome window. To find a

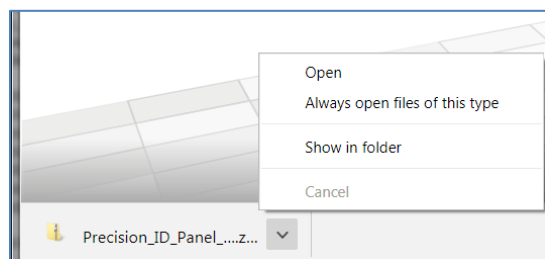
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file on your computer, next to the filename, click the arrow next to the file name > Show in folder link. Copy the downloaded files on to an external USB drive and insert into a readable port of the Converge appliance server for use.

- TSS - Upgrade your Torrent Server, Ion Chef, and Ion S5/S5 XL to TSS v5.10. For TSS v5.10 documentation, refer to the [TSS 5.10 User Guide](#) and [Release Notes](#).
- Precision\_ID\_Panel\_Definitions.zip - Includes relevant Reference/BED and JSON Files and must be installed onto TSS v5.10 before analyzing data generated with Precision ID Chemistry. The zip file can be downloaded from [S3](#) or [TF.com](#) link.

Snapshot of the Downloaded Precision\_ID\_Panel\_Definitions.zip file at bottom of your Chrome window. Click “Show in folder” to locate the file and copy it to an external USB drive and insert into a readable port of the Converge appliance server for use.

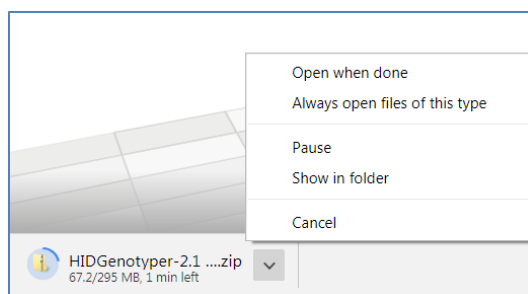


The Precision ID Panel definition.zip includes the following list of files:

- a. Mito Panel –
    - Mito Reference - Precision\_ID\_mtDNA\_rCRS.fasta
    - Mito CR Target File - Precision\_ID\_mtDNA\_Control\_Region\_Panel\_Targets\_v1.0.bed
    - Mito WG Target File - Precision\_ID\_mtDNA\_Whole\_Genome\_Panel\_Targets\_v1.0.bed
    - Mito Analysis Parameter File - Precision\_ID\_mtDNA\_Panel\_AnalysisParams\_v1.0.json
  - b. SNP Panel -
    - Ancestry Target File - Precision\_ID\_Ancestry\_Panel\_Targets\_v1.0.bed
    - Ancestry Hotspot File - Precision\_ID\_Ancestry\_Panel\_Hotspot\_v1.0.bed
    - Ancestry Analysis Parameter File - Precision\_ID\_Ancestry\_Panel\_AnalysisParams\_v1.0.json
    - Identity Target File - Precision\_ID\_Identity\_Panel\_Targets\_v1.0.bed
    - Identify Hotspot File - Precision\_ID\_Identity\_Panel\_Hotspot\_v1.0.bed
    - Identity Analysis Parameter File - Precision\_ID\_Identity\_Panel\_AnalysisParams\_v1.0.json
  - c. STR Panel
    - Precision\_ID\_GlobalFiler\_NGS\_STR\_Panel\_Target\_v1.1.bed
    - Precision\_ID\_GlobalFiler\_NGS\_STR\_Panel\_Hotspot\_v1.1.bed
    - Precision\_ID\_GlobalFiler\_NGS\_STR\_Panel\_AnalysisParams\_v1.1.json
    - Precision\_ID\_GlobalFiler\_NGS\_STR\_Control\_Sample\_male007\_v1.1.json
    - Precision\_ID\_GlobalFiler\_NGS\_STR\_Control\_Sample\_9947A\_v1.1.json
    - Precision\_ID\_GlobalFiler\_NGS\_STR\_Control\_Sample\_NegCtrl\_v1.1.json
    - Precision\_ID\_GlobalFiler\_NGS\_STR\_Control\_Sample\_9947A\_and\_male007\_and\_NegCtrl\_v1.1.json
- HID Genotyper v2.1 Plugin - Download the plugin HIDGenotyper-2.1.zip from [S3 link](#) or [TF.com](#) for installation. Snapshot of the Downloaded HID Genotyper v2.1.zip file visible at bottom of your Chrome window is shown below. Click “Show in folder” to locate the file and copy it to an external USB drive.

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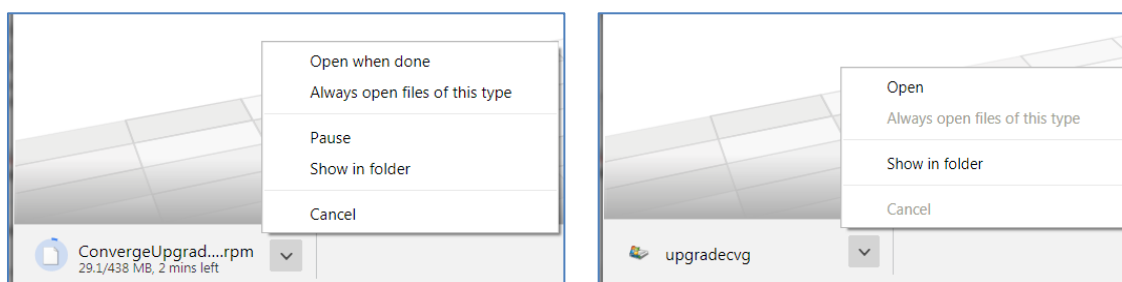
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Upload / configure plugin on supported Ion S5/S5XL™ or Ion GeneStudio Series Torrent Suite server. An example of successful configuration of HID Genotyper v2.1 Plugin with TSS and Converge v2.1 software is shown below.

- Upgrade Installer Package – Download following two files (Serial Number - df70f6b-201808231621) from S3 link or [TF.com](#) for upgrading Converge v2.0.1 to Converge v2.1 software installed on Dell T110 or T130 Server.
  - ConvergeUpgrade-2.1-0.x86\_64.rpm - [S3 link](#)
  - upgradecvg - [S3 link](#)

Snapshot of the Downloaded ConvergeUpgrade-2.1-0.x86\_64.rpm and upgradecvg files at bottom of your Chrome window. Click “Show in folder” to locate the file and copy it to your local directory or an external USB drive and insert into a readable port of the Converge appliance server for use.



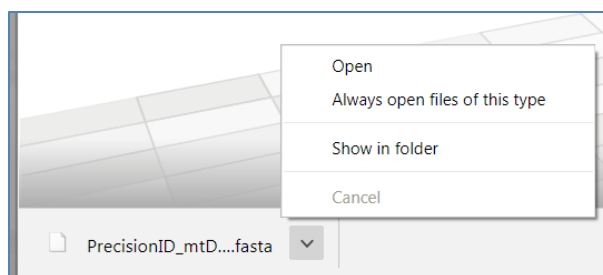
Refer to Converge™ Software v2.1 SETUP AND REFERENCE GUIDE - Publication Number 100039539, Rev D (Appendix B) for instructions on steps to upgrade to Converge v2.1 software

- Download Mito reference file “Precision\_ID\_mtDNA\_rCRS.fasta” (NCBI reference NC\_012920) file from [S3 link](#) or [TF.com](#). Snapshot of the Downloaded Precision\_ID\_mtDNA\_rCRS.fasta file at bottom of your Chrome window. Click “Show in folder” to locate the file and copy it to an external USB drive. Upload the file onto TSS > Reference Page as shown below.

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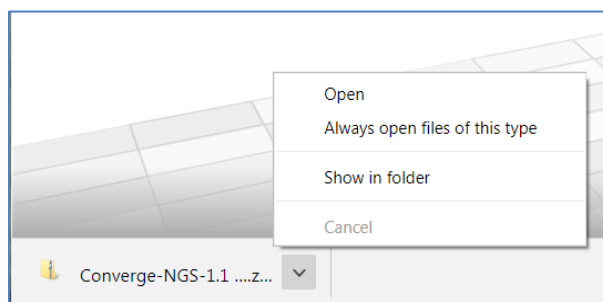
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Reference Sequences		Reference Sequences					Import Preloaded Ion References	Import Custom Reference
Reference Sequences								
Obsolete Reference Sequences								
Target Regions								
Hotspots								
Test Fragments								
Barcodes								
Upload History								
Short Name	Description	Notes	Enabled	Date	Status			
HPV	GeneTree		true	Feb 19 2018	Successfully Completed			
PrecisionID_mtDNA_rCRS	Mito		true	Aug 21 2017	Successfully Completed			
hg19	Homo sapiens		true	Nov 5 2016	Successfully Completed			
e_coli_dh10b	E. coli DH10B		true	Feb 14 2013	Successfully Completed			

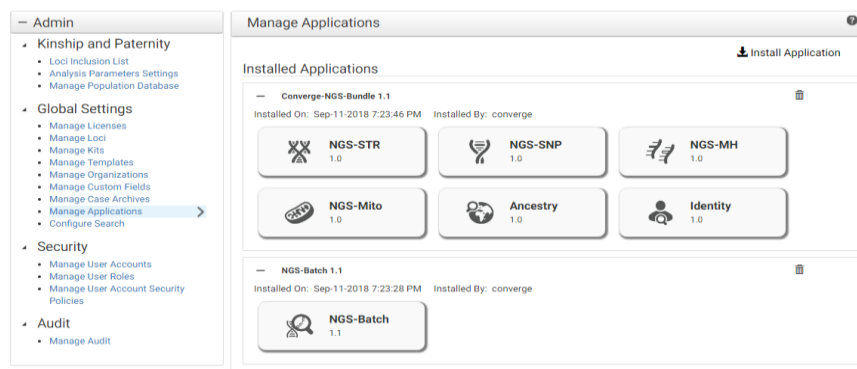
- Readme.txt instructions can be downloaded from [S3 link](#) or [TF.com](#) to a USB and inserted into a readable port of the Converge appliance server for use. For any technical support on upgrade path, contact local FAS team member.
- Converge-NGS-1.1.zip - Download Converge-NGS-1.1.zip file from [S3](#) or [TF.com](#) links. Snapshot of the Downloaded Converge-NGS-1.1.zip file at bottom of your Chrome window. Click “Show in folder” to locate the file and copy it to an external USB drive.



- Post upgrade from Converge v2.0 / v2.0.1 > Converge v2.1 version, install Case management license at minimum, followed by uploading NGS Application Bundle package (Converge-NGS-1.1.zip) onto Converge > Admin > Manage Application Software page (snapshot shown below).
- Once installed proceed to install and activate NGS license downloaded from <https://licensing.appliedbiosystems.com/web/login> web portal.

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## NOTE:

- During upgrade from Converge v2.0 and v2.1, install NGS Application Bundle and NGS License prior to navigation through the Converge Software workflow. If user has not installed the NGS Application bundle and corresponding licenses, certain actions for example deletion of batches may lead to loss of user data.
- For Fresh Installation on Dell T130, Converge-NGS-1.1 application modules comes pre-installed by default.

## UPDATES TO CONVERGE™ SOFTWARE v2.1 HELP TOPICS

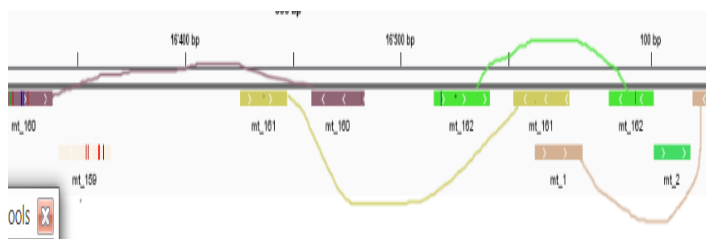
Refer to Converge™ Software v2.1 Help contained in the software for following updates::

- New workflow and Analysis procedures.
  - NGS mtDNA & SNP workflow
  - Sample Dashboard
  - Profile Dashboard
  - Manage Applications
- Troubleshooting procedures.
  - NGS Module workflow, Display Issues, Profile Management, and Kinship & Paternity features.

## KNOWN ISSUES AND LIMITATIONS:

- Data Related:
  - Median Coverage of Amplicon calculation excludes overlapping amplicon read coverage, as such, when reads for overlapping amplicons are removed amplicon coverage is zeroed out (as seen in variant\_colored.xls output file and screenshot below).

Amplicon	Median Coverage
Sample median	18777
15927-16092(165)	47432
16034-16159(125)	27825
16083-16251(168)	8061
16196-16365(169)	6189
16312-16483(171)	38828
16426-16578(152)	22198
81-16541(16460)	0
1-143(142)	43935
102-268(166)	41953
219-354(135)	3012
271-435(164)	5070
360-503(143)	9796
436-566(130)	5806
500-635(135)	2778



- For overlapping amplicons, the software selects only one amplicon coverage value for display even when there are significant coverage differences between the amplicons. Example: Software displays the coverage for position 263, spanned by 2 amplicons with coverages of 4839x and 542x. as of now it is considering the lesser of the two values which is 542x. In addition, comments in the variant colored xls sheet displays incorrect information on the coverage stats

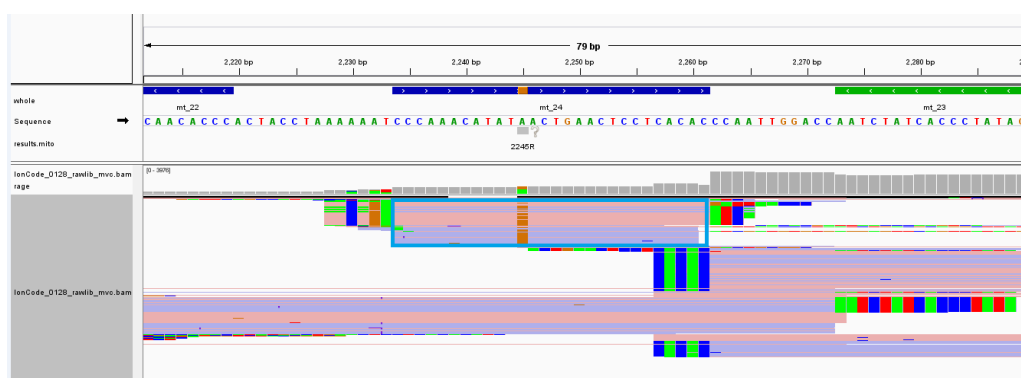
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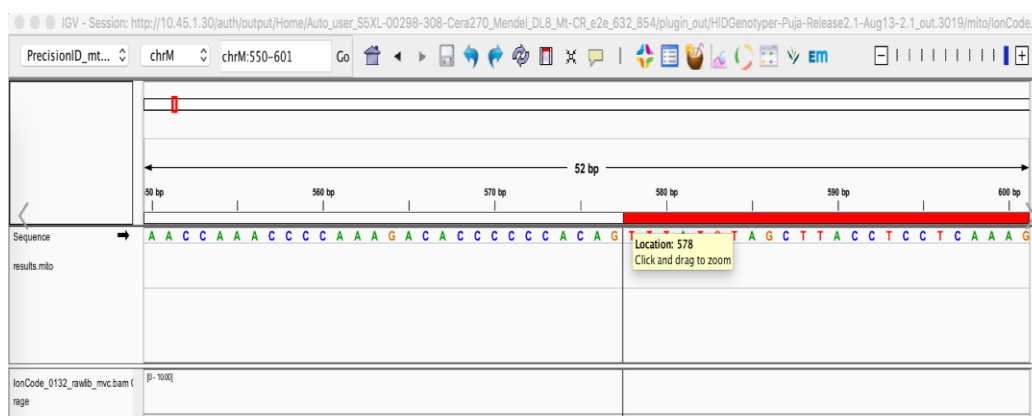
	A	B	C
1	Amplicon	Median Coverage	
2	Sample median	4017	
3	1-143(142)	6882	
4	102-268(166)	4839	
5	219-354(135)	542	
6	271-435(164)	417	
7	360-503(143)	541	
8	436-566(130)	490	
9	500-635(135)	196	
10	571-732(161)	5428	
11	663-830(167)	4798	
12	771-930(159)	5347	
13	868-1029(161)	2450	
14	964-1138(174)	4717	
15	1092-1266(174)	3687	
16	1212-1377(165)	7844	
17	1317-1491(174)	7918	

Date	2_out_5850/mito/ionCode_0149/results/ionCode_0149_rawlib_mvc.bam								
Sam file									
Sample									
Close hapl									
Regions									
Position	Ref	State	Frequency	Artifact	Var Strand	Read Strand	EMPOP	Score	Comment
54	uncl	54.5	Point	True	5.5	5.6	unchecked	0.133	no strand bias in variant reads; not listed in X261 or its subgroups; found 3.06% in EMPOP;
73	confirmed	99.8	True	variant	5.5	5.6	unchecked	1	no strand bias in variant reads; 730 is expected in X261 or its subgroups; found 76.26% in EMPOP;
155	confirmed	99.7	True	variant	5.5	5.5	unchecked	1	no strand bias in variant reads; 195C is expected in X261 or its subgroups; found 20.78% in EMPOP;
204	confirmed	99.7	True	variant	5.5	5.5	unchecked	1	no strand bias in variant reads; 204C is expected in X261 or its subgroups; found 6.13% in EMPOP;
207A	confirmed	99.5	True	variant	5.5	5.5	unchecked	1	no strand bias in variant reads; 207A is expected in X261 or its subgroups; found 4.52% in EMPOP;
263	confirmed	100	True	variant	0.5	0.6	unchecked	0.903	no strand bias in variant reads; The coverage is only 544 compared to the median 3686 for this amplicon (might be sequencing artefact); 263G is expected in X261 or its subgroups; found 96.88% in EMPOP;
509	likely	51.7	Length	True	5.8	5.7	confirmed	0.683	The reference has HP 7 (might be indel artefact); (the sample sequence might have HP 8); no strand bias in v
515	likely	56.2	Length	True	5.8	5.7	confirmed	0.693	The reference has HP 5 (might be indel artefact); (the sample sequence might have HP 6); no strand bias in v
709	confirmed	99.1	True	variant	5.5	5.5	unchecked	1	no strand bias in variant reads; 709A is expected in X261 or its subgroups; found 14.33% in EMPOP;
730	confirmed	97.5	True	variant	5.5	5.5	unchecked	1	no strand bias in variant reads; 730G is expected in X261 or its subgroups; found 98.96% in EMPOP;
748	confirmed	99.2	True	variant	5.5	5.6	unchecked	1	no strand bias in variant reads; 1438G is expected in X261 or its subgroups; found 95.74% in EMPOP;
719	confirmed	98.1	True	variant	5.5	5.6	unchecked	1	no strand bias in variant reads; 719A is expected in X261 or its subgroups; found 5.73% in EMPOP;
726	confirmed	99.7	True	variant	5.5	5.7	unchecked	1	no strand bias in variant reads; 726G is expected in X261 or its subgroups; found 79.33% in EMPOP;
769	confirmed	99.8	True	variant	5.5	5.5	unchecked	1	no strand bias in variant reads; 769G is expected in X261 or its subgroups; found 98.39% in EMPOP;
839	possible	51.9	True	variant	5.5	5.8	unknown	0.512	no strand bias in variant reads; not listed in X261 or its subgroups;
886	confirmed	99.4	True	variant	5.5	5.7	unchecked	1	no strand bias in variant reads; 886G is expected in X261 or its subgroups; found 0.16% in EMPOP;
921	confirmed	99.9	True	variant	5.5	5.5	unchecked	1	no strand bias in variant reads; 921C is expected in X261 or its subgroups; found 3.13% in EMPOP;
971	confirmed	98.4	True	variant	5.5	5.6	unchecked	1	no strand bias in variant reads; 971T is expected in X261 or its subgroups; found 1.16% in EMPOP;
991	confirmed	98.4	True	variant	5.5	5.5	unchecked	1	no strand bias in variant reads; 991G is expected in X261 or its subgroups; found 0.17% in EMPOP;
928	confirmed	99	True	variant	5.5	5.6	unchecked	1	no strand bias in variant reads; 928T is expected in X261 or its subgroups; found 80.48% in EMPOP;
953	confirmed	100	True	variant	5.5	5.9	unchecked	1	no strand bias in variant reads; 953C is expected in X261 or its subgroups; found 0.28% in EMPOP;
980	confirmed	98.4	True	variant	5.5	5.8	unchecked	1	no strand bias in variant reads; 980G is expected in X261 or its subgroups; found 99.48% in EMPOP;
1110	confirmed	99.6	True	variant	5.5	5.8	unchecked	1	no strand bias in variant reads; 1110A is expected in X261 or its subgroups; found 74.81% in EMPOP;

- In certain samples with low amount of DNA (< 0.1ng) or degraded DNA and / or high amount of primer dimer, not all primer sequence get removed. In these cases, both the target sequence that contributes to the variant call and primer sequences are used to determine the variant classification.



- Excluded region in IGV is off by 1 base, independent of what "virtual kit"/ regions analysed is selected. Note - The beginning of the marker (highlighted in red in IGV, where start position 578 instead of 577, is off by 1. All the end positions are correctly displayed though.



- Confirmed variants are used for mtDNA haplotype profile comparisons at the batch and case level. In some instances, match results may differ between batch and case levels. In the example below, the reference is missing a variant at position 150 that is present in the unknown profile; the variant is considered for match% calculation, and the software displays this as a mismatch for the case level comparison.

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For the same comparison at the batch level, position 150 is ignored

Batch Level Comparison Match % (4/10 = 40%)

Case Level Comparison Match % (4/15 = 26.7%)

Manual_Mt_CR_Chr_Rep1 (N)	Manual_Mt_CR_HL6_Rep2 (22b1a)
64Y	Meta Data
73G	Percentage Match: 48
195C	
204C	
207A	
263G	263G
309c	309c
309.1C	
315.1C	315.1C
16183C	
16189C	
16193.1C	
16223Y	
16224Y	
16278T	16278T

Position	Profile 1326 (Precision_ID_mDNA_Control_Region_Panel_v1.0)	Profile 1327 (Precision_ID_mDNA_Control_Region_Panel_v1.0)
73	73G	73G
195	195C	-
204	204C	-
207	207A	-
263	263G	263G
16189	16189C	-
16278	16278T	16278T
16519	16519C	-
16642	16642G	16642G
150	-	150T
152	-	152C
295	-	295T
489	-	489C
Variant Match %		26.7%

- Converge software calls variants in accordance with ISFG and IUPAC recommendations. As such, lower case variants should be displayed as "del". The following examples highlights inconsistencies in nomenclature for certain software displays.

- 16640g is incorrectly displayed in the variant grid and should be "16640del" with a "-" deletion icon.

- Deletion variants are being saved with upper case "DEL" and lower case "del", although they reflect the same deletion w.r.t to comparison but is different w.r.t to frequency/display..

- Mito Results Page > Variants classified as Degraded in "Grid>Unexpected" tab are not captured in "variant\_colored.xls" excel output file..

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[Variants](#)
[Uncalled Variants](#)
[Unexpected Variants](#)
[Excluded Variants](#)

Variant	Frequency	Status	Var Strand Bias	Classification
16319R	11.29	False	0.8	Degraded
16320Y	12.86	False	0.77	Degraded
16327Y	20.29	False	0.6	Degraded
309del	67.29	Unclear	0.87	Length Heteroplasmy
16325Y	14.96	Unclear	0.73	Point Heteroplasmy

Unexpected variants of J2b1a1a that were listed

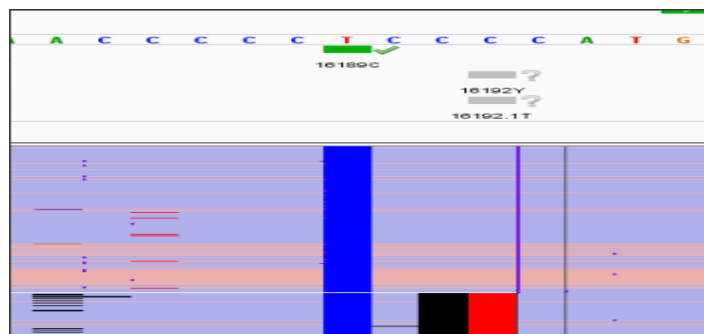
Variant	Frequency	State	Var Strand	EMPOP state
309del	67.3	unclear	0.9	UNEXPECTED
16325Y	15	unclear	0.7	UNEXPECTED

- Mito Quality Scores are rounded off up to 2 decimal places but original values are used for state determination. For e.g. Calls with score 0.82 has state as “Likely” when score  $\geq 0.82$  maps to “Confirmed” &  $< 0.82$  maps to “Likely” state. The reason is that the actual score is 0.815 (as seen in variant\_colored.xls output file) but it is rounded up to 2 decimal places & displayed as 0.82.

16311C	93.11	Likely	Unchecked	0.53	True Variant	2758	0.82
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- Both LHP & PHP variants get reported at same location for single source. Example two calls observed at position 16192 - 16192Y & 16192.1T. For a single source it is not expected to have variants classified as both LHP & PHP. As seen in the variant\_colored.xls export file (snapshot below), 23.5% of T inserts ideally should be classified as a sequence artefact and not a true positive call for a single source.

Position	Ref	Sampl	Var	Var Freq	Type	Read Cov	Read Cov	Cov	Allele Cov	Allele Cov	Allele G%	A%	T%	C%	N%	ins%	del%	Polymorp	Control R	State	Frequency	Artefact	Var Stranc	Read Stra
16192	C	T	Y	60.5	SNP	227	747	589	134	455	0	0.1	60.5	35.4	0	0.1	4	16192Y	16192Y	possible	60.5	Point Het	0.5	0.8
16192	C	+	T	21.3	INS	227	748	208	56	152	0	0	0	0	23.5	0		16192.1T	16192.1T	possible	21.3	Length He	0.5	0.8



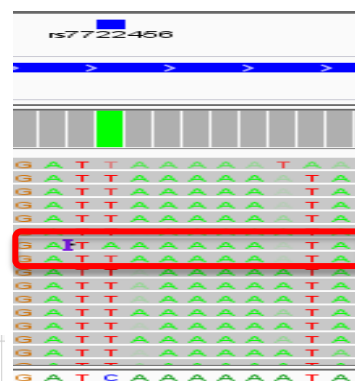
- Consensus.fasta downloaded from Mito Results grid contains the entire reference genome as opposed to three different contigs covering HV1 , HV2 , HV123 or intervening regions should have been masked with an “N” calls.
- Genotype errors are around homopolymer regions with too many wrong A reads. For example rs7722456 (aiSNP) which lies near homopolymer region (6As) because of which some A bases incorrectly align with rs7722456 position & hence, “A” Genotype call is added. ~15% of reads show an “A” call as seen in the contig assembly snapshot in IGV Browser and could be due to a sequence artefact around homopolymer regions.

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# SOFTWARE RELEASE NOTES

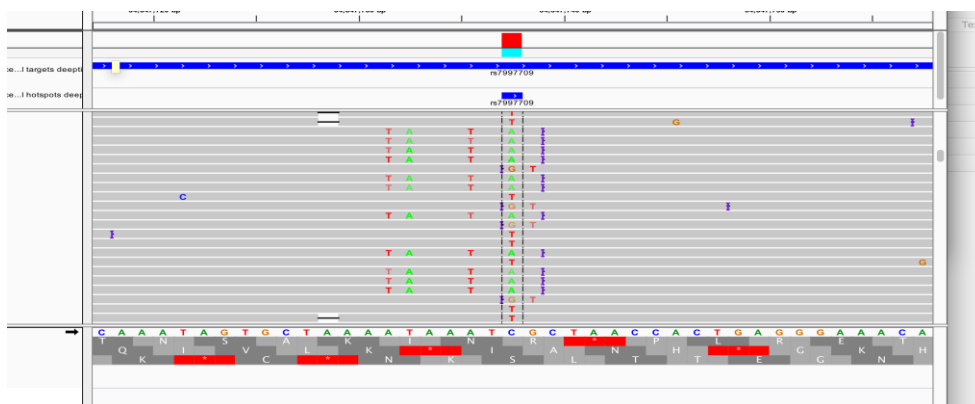
Snapshot of Converge SNP Results > Grid view and IGV Contig assembly view

rs7722456	chr5:170202984	AT	6973	
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Additional SNP id's with similar noise issues –

- rs772262 – C noise is added to this SNP position because of poly Cs (5Cs)
- rs13400937- T noise is added to this SNP position because of poly Ts (4 Ts)
- rs7997709 - A & G noise added due to polyA region close by (screenshot below)



- The following aiSNPs do not have frequency values in the seeded population table and thus excluded from tertiary calculations: rs6541030, rs2814778, rs13400937, rs1369093, rs3811801, rs6556352, rs7722456, rs6422347, rs731257, rs6464211, rs3943253, rs2306040, rs2073821, rs4918842, rs214678, rs9319336, rs17642714, rs7238445, rs3907047, rs310644.
- SNPs are assumed to be bi-allelic; we rarely observe a third allele for some SNPs (e.g., rs1503767). Since we don't have frequencies for these alleles, they are ignored from the calculation.

rs1503767 [Homo sapiens]  
1. GCTACCTCCCTTCCACCAAGGCTACC [A/C/T] GATTTTAAATGATCTCCTCTATA

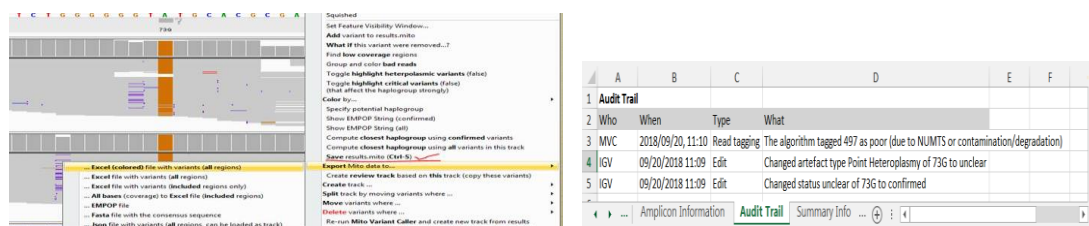
- SNP ID rs938283 from the Identity Panel does not have frequency values in 1000 Genomes population table and hence ignored in Random Match Probabilities (RMP) calculations.

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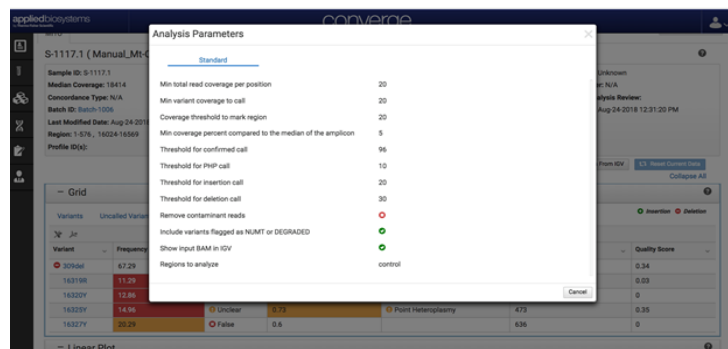
# SOFTWARE RELEASE NOTES

## ➤ General Functionality, IGV and TSS Workflow:

- Converge v2.1 does not have audit capability features enabled for the supported mtDNA analysis workflow. Further, Mito IGV contains advanced user analysis settings in an unsupported workflow. Variant edits made in Mito IGV are auditable (steps & screenshot below) to a limited extent; changes to advanced analysis settings are not auditable. Steps to perform the Mito IGV analysis workflow are as follows:
  - Launch IGV > Make edits > Right click in the variants track
  - Select “Save results.mito” > Right click again > Select “Export Mito Data to”
  - Followed by “Excel colored file with all variants”



- Grid “Preference Settings” for Batch Details > Mito Results Card does not have options to be configured and saved.
- “Export Results” from Batch Details > Mito Results Card works only for a smaller set of sample results (up to 10).
- A user may reanalyse samples in IGV with modified parameters using “Load from IGV” workflow in Converge. Post Reanalysis in IGV with modified parameters, the analysis parameters on Converge > Mito Results > Summary Pane does not get updated although genotype changes get recorded.



- Profile comparison grid does not export the entire grid (pdf format) and screen captures are required to capture the entire grid.
- User Defined Custom Templates created in Converge 2.0 version needs to be reimported using “Admin > Manage Template” Workflow, post upgrade from Converge v2.0 > v2.1.
- Batch (.bef) file generation fails if TSS is offline mode, since Converge Software fails to fetch valid host IP. Meaning if TSS server runs as a stand-alone (non-networked) server where the plugins are hosted, then the HIDGenotyper plugin would fail while trying to fetch the host id of the server. And though the analysis will complete for all the samples involved, but the previous exceptions would prevent it from generating the .bef

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(batch export file).

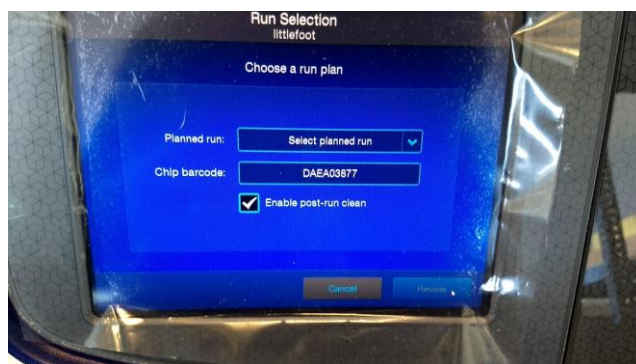
This known issue will be addressed in subsequent release of HID Genotyper Plugin.

- In certain network topology, S5 Data Collection system does not automatically sync the run plan on the chip. There is hence a delay in Data Collection server updating run plan on TSS v5.10 server and hence expected run plans do not get updated. The workaround is to restart Data Collection server and restart S5 sequence analysis workflow.

This known issue will be addressed in next patch release of Data Collection and TSS server v5.10.1.

Steps to be taken and workaround –

- a. In the run selection page, notice that S5 does not automatically sync the run plan in the “Planned run field” when the chip barcode gets displayed (snapshot below)



- b. Workaround, is to keep chip in S5 > Restart Data Collection > Run > selecting "Next" in "Load Chip" screen > S5 automatically selects correct run plan



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# SOFTWARE RELEASE NOTES

## RELEASE & COMPATIBILITY SUMMARY:

SYSTEM	TYPE	DESCRIPTION	VERSION / DATE STAMP
TSS	Software	TSS Compatibility	v5.10
	Run Templates	Precision ID mtDNA Control Region Panel - S5	June 23 <sup>rd</sup> 2018
		Precision ID mtDNA Whole Genome Panel - S5	June 23 <sup>rd</sup> 2018
		Precision ID Ancestry Panel - S5	June 23 <sup>rd</sup> 2018
		Precision ID Identity Panel - S5	June 23 <sup>rd</sup> 2018
		Precision ID GlobalFiler STR Panel - S5	June 23 <sup>rd</sup> 2018
TSS Control Publisher	Publisher	Control_Samples	v1.01
	STR Control Files	Precision_ID_GlobalFiler_NGS_STR_Control_Sample_male007	v1.1
		Precision_ID_GlobalFiler_NGS_STR_Control_Sample_9947A	v1.1
		Precision_ID_GlobalFiler_NGS_STR_Control_Sample_NegCtrl	v1.1
		Precision_ID_GlobalFiler_NGS_STR_Control_Sample_9947A_and_male 007_and_NegCtrl	v1.1
		Precision_ID_mtDNA_Whole_Genome_Panel_Targets_v1.0.bed	v1.0
	NGS mtDNA Module	Precision_ID_mtDNA_Control_Region_Panel_Targets_v1.0.bed	v1.0
		Precision_ID_mtDNA_Panel_AnalysisParams_v1.0.json	v1.0
		Precision_ID_Ancestry_Panel_Targets_v1.0.bed	v1.0
	BED/ JSON File	NGS SNP Module	Precision_ID_Ancestry_Panel_Hotspot_v1.0.bed
Precision_ID_Identity_Panel_Target_v1.0.bed			v1.0
HID Genotyper	NGS STR Module	Precision_ID_Identity_Panel_Hotspot_v1.0.bed	v1.0
		Precision_ID_GlobalFiler_NGS_STR_Panel_Target	v1.1
		Precision_ID_GlobalFiler_NGS_STR_Panel_Hotspot	v1.1
	Precision_ID_GlobalFiler_NGS_STR_Panel_AnalysisParams	v1.1	
	Plugin	HIDGenotyper-2.1 (Serial No.)	2.1_df70f6b
	Software	About (Serial No.)	df70f6b- 201808231621
	Module	Platform	v2.1
Converge Software	License	Case Management	v1.0
	Module	Kinship and Paternity Analysis	v1.0
	License	Converge Kinship	v1.0
	Module	NGS Application	v1.1
	License	Converge NGS	v1.0
	Upgrade Installer	ConvergeUpgrade-2.1-0.x86_64.rpm (Serial No.)	df70f6b- 201808231621
	Components	upgradecvg (Serial No.)	
Supplemental	Converge-NGS-1.1.zip	v1.1	

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**appliedbiosystems**  
by Thermo Fisher Scientific

Files	Precision_ID_Panel_Definitions.zip	Oct 15 <sup>th</sup> 2018
	Precision_ID_mtDNA_rCRS.fasta File (NCBI reference NC_012920)	July 19 <sup>th</sup> 2017

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October 2018

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