

# Genexus™ Software 6.8.1.1

## Release Notes

Publication No. MAN0029968 Rev. A.0

### Contents

Revision History .....	2
Release Notes Overview .....	2
Languages supported in Genexus™ Software 6.8 .....	2
Antivirus software .....	2
Genexus™ Software 6.8 Key Features .....	3
Ion Torrent Genexus™ Integrated Sequencer security .....	3
Assay Application Support .....	3
Oncomine™ Precision Assay has the following new features and enhancements .....	3
Oncomine™ BRCA w4.1.0 assays have the following new features and enhancements .....	3
Increased sample capacity .....	3
Support for 2-pool designs for AmpliSeq™ HD custom panels .....	3
Ability to update annotation sources .....	3
Rules for variant names .....	3
Download variant results in TSV format .....	3
New sample attribute: Nucleic Acid Type .....	3
Download sample results for a selected assay .....	4
Electronic signature enhancements .....	4
Data and annotation source updates .....	4
New user-access level for users to view and electronically sign off reports .....	4
New summary report design .....	4
View and download Final Reads QC metric .....	4
Issues fixed in Genexus™ Software 6.8.1.1 .....	5
Issues fixed in Genexus™ Software 6.8.0 .....	5
Known issues in Genexus™ Purification Instrument Control Software 6.8.1.1 .....	7
Known issues in Genexus™ Integrated Sequencer Control Software 6.8.1.1 .....	8
Known issues in Genexus™ Software 6.8.1.1 .....	8
System Requirements .....	25
Version information .....	25
Software plugin versions .....	25

## Revision History

Revision	Date	Description
A.0	05 September 2023	Release notes for Ion Torrent Genexus™ Software 6.8.1.1. The following issues are fixed: 39633, 36388, 40320, 35277, 42846, and 36955, and Known Issues 42847, 42946, 43231, 43896 and 42946. Known Issue 1796 was added for Genexus™ Integrated Sequencer Control Software 6.8.1.1.

## Release Notes Overview

This document outlines major features in Ion Torrent™ Genexus™ Software 6.8, issues fixed in Genexus™ Software 6.8.0 and 6.8.1.1, and known issues to be fixed in subsequent releases.

## Languages supported in Genexus™ Software 6.8

Genexus™ Software 6.8 supports the following languages.

- English
- Chinese
- French
- German
- Italian
- Japanese
- Spanish

## Languages supported by the help system and documentation

The Genexus™ Software 6.8 help system, user guide and release notes are available only in English.

## Antivirus software

We have tested Genexus™ Software 6.8 with the following antivirus software products and found that the products are compatible as antivirus solutions.

- Bitdefender GravityZone™ Business Security 7.0.3.2115 <https://www.bitdefender.com/business/>
- AVAST Premium Business Security 4.2.0 <https://www.avast.com/>

**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.

# Genexus™ Software 6.8 Key Features

## Ion Torrent Genexus™ Integrated Sequencer security

The system configuration is enhanced for security on the Ion Torrent Genexus™ Integrated Sequencer. Genexus™ Software includes all updates contained in the Genexus™ Security Package v1.0.0.

## Assay Application Support

Genexus™ Software now expands support of assays and applications. Default analysis parameters are now optimized to better support implementation of human custom assays.

## OncoPrint™ Precision Assay has the following new features and enhancements

- Novel variant calling is enabled for the entire panel for FFPE samples.
- Re-sequencing for cTNA is enabled, which increases the maximum number of samples to 6 samples with controls in a single sequencing run on a GX5 chip.
- CNV calling module is integrated for both cTNA and FFPE, and parameters are adjusted to increase the specificity in the FFPE pipeline.

## OncoPrint™ BRCA w4.1.0 assays have the following new features and enhancements

- The liquid handling workflows are updated to improve assay robustness.
- The sequence variant baseline, copy number baseline and hotspot files are further optimized for variant calling with an updated version of the Torrent Variant Caller module to increase the sensitivity and specificity for both germline and somatic workflows.
- The CNV analysis pipeline is updated to increase accuracy of CNV reporting.

## Increased sample capacity

Compatible with up to 48 barcode reactions for increased sample capacity per run.

## Support for 2-pool designs for AmpliSeq™ HD custom panels

Genexus™ Software 6.8 supports 2-pool designs for AmpliSeq™ HD custom panels, allowing sequencing runs that target full genes on the Genexus™ Integrated Sequencer with these panels.

## Ability to update annotation sources

An administrator-level user can get the latest additions, deletions, or changes to the ClinVar database in Genexus™ Software.

## Rules for variant names

Genexus™ Software provides standard conventions for the names of genomic variants. Standard variant names are derived from sample and results data and properties that are available in the software for each variant, such as variant, gene name, and amino acid change. The conventions are used for both de novo variants and variants that are included in a hotspots file.

## Download variant results in TSV format

When you view variant results in Genexus™ Software, you can now download the information for the variants and the annotations associated with the variants in TSV format.

## New sample attribute: Nucleic Acid Type

Nucleic Acid Type is now available as a sample attribute.

### Download sample results for a selected assay

You can now download sample result files of your choice from an assay that you select in Genexus™ Software. Select the assay in the software, and the samples of interest to download a compressed folder in ZIP format.

### Electronic signature enhancements

Notification and confirmation messages appear when edits are made to an electronic signature. Signed reports will show first and last name of the signature user.

### Data and annotation source updates

The following annotation sources are updated.

- ClinVar
- COSMIC
- dbSNP
- DrugBank
- GO
- RefSeq Gene Model
- Ensemble Gene Model
- OMIM
- Pfam
- PhyloP Scores

### New user-access level for users to view and electronically sign off reports

Administrator-level users can now create users whose primary role is to view and sign off on reports. Users who are assigned the Report role can do the following actions in the software.

- Access the Sample Results and Run Results screens to view results
- Generate, view, and sign variant reports
- Send notifications
- Download results files
- View and edit notes for the sample result
- View the audit trail for sample results
- View notifications


### New summary report design

PDF reports generated in Genexus™ Software have a new design layout.

### View and download Final Reads QC metric

The number of library reads that pass all filters for sample results, and sample results from runs that are resequenced, are now included as a QC metric in the software. You can view Final Reads and Final Reads - Resequencing in the QC tab of the Results screen, download and generate the metric in summaries of run results, and find the metric recorded in output BAM file outputs.

## Issues fixed in Genexus™ Software 6.8.1.1

Issue number	
42846	An issue was discovered which prevented restore functionality in Genexus™ Software 6.8.0 for instruments that have a hyphen (-) in the server name. This includes the default factory-shipped instrument names, which contain a hyphen (-) in the server name. No data was lost and data remained in the stored location(s). Data could still be backed up or archived, but could not be restored from the backup or archived location.
39633	A warning symbol  with a message that stated “Sample concentration out of range” was shown in error for automated sample dilution in the <b>Run Plan</b> screen when you planned a run, or viewed the Run Summary screen, in Genexus™ Software.
36388	The expiration date for verification runs that were performed during installation of Genexus™ Integrated Sequencer or for performance qualification (PQ) did not match the expiration date shown on the Genexus™ GX5 or Genexus™ GX7 chip product labels. The incorrect date was shown in the Reagents section of the Verification Results screen, and in the Verification PDF report for verification runs.
40320	Oracle™ Java™ documentation states that the Java™ security feature has been updated, which caused issues with the JNLP not loading in updated versions of Java™, due to JARs that are signed with weak algorithms. This applied to Java version 8 Update 351 and later, and affected the JNLP file that is required for use of the Broad Institute Integrative Genomics Viewer (IGV).
35277	Some of the context-sensitive help that was shown in the Assay Metrics tab of run results was not displayed correctly. When you hovered over either the blue or gray sections of the bar graph for "Loading" wells the help pop-up showed a count for a number of "Wells with ISPs". The gray sections of the graph now show information for the number "Empty Wells."
36955	The following issue was listed incorrectly in Rev. A of these release notes as a Known Issue, however, it was not an issue for Genexus™ Assay Development Software: Some QC values of "N/A" were shown intermittently in the Run Results screen.

## Issues fixed in Genexus™ Software 6.8.0

30178	If you clicked Run Plugin for a sample result while data backup is in progress, the 1.wells files, the basecaller output, and the coverageAnalysis plugin source files for the sample result are deleted. If this issue occurred, the Loading Density image and histograms were not shown in the Assay Metrics and Run Report screens and values are absent for some QC metrics. Note: The affected run report data are available in the Run Report PDF.
28945	Sequencing run results that were imported to another Genexus™ Integrated Sequencer, showed a signed report that was locked for electronic signatures as a draft report on the system with the imported results.
29228	If there were three report templates marked as Default for an assay or application category, you were unable to edit the report template.
29089	Users assigned to accounts with the Manager Role were unable to access Audit records for the Services screen and Sample Attribute mapping.
28490	Custom baselines (CNV baselines, sequence variant baselines, exon tile assay baseline) that were created on one Genexus™ Integrated Sequencer could not be imported to another Genexus™ Integrated Sequencer in ZIP format.
25307	The manual signature title of a report did not show correctly when the default report template was used.
20701	Downloads of sequencing results from the Oncomine™ TCR Beta-LR Assay GX assay that included unmapped BAM and FASTQ files had no dialog box to indicate that the download started and was successful.

25474	The Uracil DNA Glycosylase (UDG) Treat DNA setting is used to clean up deaminated Cytosine bases from degraded samples. If you enabled the UDG Treat DNA parameter in an assay, you might have observed read imbalances in the Barcodes with Reads Reported table. Read imbalances can occur because the on-instrument dilution of input sample nucleic acid is impacted by this setting.
29239	If manual and automated backups occurred at the same time, the intermediate files might have been deleted without a successful backup.
29905	If you enabled <b>Generate Report</b> when you planned a run, you could not download lab reports when you clicked <b>(More Options) ▶ Download Files</b> and selected the <b>Reports</b> checkbox in the <b>Download Files</b> dialog box.
30233	If you enabled Generate Report when you planned a run, the report that was generated was always in English regardless of the language that is associated with the report template.
30234	If you enabled Generate Report when you planned a run, the language was not shown in the pane for the report in the Reports tab.
28908	For some results, the ClinVar track was not shown in the Integrative Genomics Viewer (IGV) view that is generated with the JNLP files that are downloaded from Genexus™ Software.
21585	When you edited multiple samples at once and changed the cellularity or necrosis value for one sample, the new value was applied to all samples in the Edit Sample screen.
21293	If you created a custom sample attribute with an attribute name that contained a full stop or period (.), the entries for that sample attribute were not shown in the Manage Samples table.
33765	The Run Summary tab in the Sample Results screen 6.6 sorted 2-library positions that contained multiple barcodes alphabetically, instead of showing the actual order of pools as used in the run.
13478	When you clicked any of the links for the Exome Aggregation Consortium (ExAC) browser in the Annotations tab shown below the Variants table, a message redirected you to the gnomAD browser was shown.
20720	If you added all available columns to the list of samples in the Samples / Manage Samples screen and then tried to filter the list of samples with the Tags column, an error message was shown.
21985	If you performed a run with a sample, then performed a second run with the same sample, you were unable to edit the sample information after the second run.
20742	If you enabled the option to upload immune repertoire BAM files to another Genexus™ Integrated Sequencer when you planned a run, the option to perform an analysis in the target software automatically after run completion was not available.
23545	NTC QC results for Sample to Result runs were visible only for samples that were included in the purification batch with the NTC.
21485	If you tried to compare immune repertoire results from BAM samples that had been uploaded from another Genexus™ Integrated Sequencer, no comparison results were shown.
22549	In the Key Findings tab, in the Coverage Graph for genes that are transcribed from the reverse strand, the order of the exons was shown incorrectly when you placed the pointer over the transcript track. In previous releases of Genexus™ Assay Development Software, the orientation of transcripts that are transcribed from the reverse strand could only be viewed in the Broad Institute Integrative Genomics Viewer (IGV) or with other genome browsers, such as the UCSC Human Genome Browser.
22883	If you selected a gene that was not shown by default in the Coverage Graphs, in some cases the coverage graph was not shown for the gene that you selected.
21778	For some assays, when you viewed all of the variants in the Variants table, you could add the P-Value column to the table twice.
23399	The option to create a custom filter chain to filter variant results by allele frequency did not appear to be available when you selected Assays▶Manage Presets▶Filter Chains▶Add New▶Choose Filter.
21930	If you created a custom filter chain for a variant classification, then tried to apply the custom filter chain to a result, a “Data not available” message appeared.
21794	In the Gene Browser, in some instances, the target region was represented with a black color instead of green.

23057	When you viewed the exon tile fusion imbalance charts for a sample, then selected a different sample, the charts were not updated for the new sample that is selected.
23586	For the Oncomine™ Myeloid v2 - GX5 - DNA and Fusions - w4.2.2 assay, Purification QC was not included by default.
22005	When you tried to download a BAM result file larger than 2 GB, the file that was downloaded was 0 KB and did not contain any data.
21486	If you uploaded a DNA & RNA sample result from one Genexus™ Integrated Sequencer to another sequencer and the name of the sample was more than 50 characters long, two samples were created on the target sequencer. One sample was for DNA (with a sample type of DNA & RNA) and another was for RNA (with a sample type of RNA).
21357	In the Manage Assays screen, when you selected DNA and Fusions, DNA, Fusions to refine the assay list with the filter tool in the Research Application header, no records were shown even if records were available.
21712	If you sorted the list of copy number baselines, sequence variant baselines, or exon tile assay baselines by name, an error message appeared that indicated the action could not be completed.
1316	If the error message “Lane clog check failed” was shown after a clean instrument procedure, the message appeared only briefly.
31901	Spearman's correlation coefficient was calculated incorrectly using all clones when performing a pairwise sample comparison in the Oncomine™ TCR Beta-LR GX assay.

## Known issues in Genexus™ Purification Instrument Control Software 6.8.1.1

Issue number	Issue Summary	Impact and Mitigation <sup>1</sup>
933	When you sign in on the Genexus™ Purification Instrument the error message “Error in response from TS” might be shown on the instrument screen, or the countdown timer runs continuously.	To resolve the issue, close the error message then try to sign in again. If the countdown timer runs continuously, you must reboot the instrument, then wait one to two minutes, and try to sign in again.
1796	The <b>About This Instrument</b> screen shows Software Version: 6.8.1 on the Genexus™ Integrated Sequencer RUO instrument. The <b>About This Instrument</b> screen shows Software Version: 6.8.1 when you are signed into Assay Development mode on the Genexus™ Dx Integrated Sequencer.	<b>Workaround:</b> Sign into Genexus™ Software on a computer to see the full 4-digit software version number.
794	If a USB update for the purification instrument failed, there is no notification in the software.	When you attempt to update the purification instrument software from a USB, the message “Install Completed; Please Reboot Now” appears regardless of whether the software update is successful. Tap Instrument Settings ► About to view the software version and confirm that the software update is complete.

## Known issues in Genexus™ Integrated Sequencer Control Software 6.8.1.1

Issue number	Issue Summary	Impact and Mitigation <sup>1</sup>
933	When you sign in on the Genexus™ Integrated Sequencer the error message "Error in response from TS" might be shown on the instrument screen, or the countdown timer runs continuously.	To resolve the issue, close the error message then try to sign in again. If the countdown timer runs continuously, you must reboot the instrument, then wait one to two minutes, and try to sign in again.

## Known issues in Genexus™ Software 6.8.1.1

Issue number	Issue Summary	Impact and Mitigation
42946	If you copy and edit two different versions of the OncoPrint-Precision-GX5 assay, the CNV calling parameter file from the version of the assay that was initially edited is incorrectly used for both copies of the assay.	Contact your Field Bioinformatics Specialist (FBS) or Clinical Account Consultant (CAC) for instructions to enable the correct CNV parameters.
39861	When the Bam Uploader is used to transfer BAM files between more than one Genexus™ Integrated Sequencer, the software appends the sample names to add version and nucleic acid type to the names for use on the destination sequencer. If a sample name is approximately 50 characters or more, the software might delete part of the original sample name. As a result, multiple BAM files might be listed under a single sample in the BAM files that are transferred.	Do not use any samples that unexpectedly or incorrectly lists multiple BAM files under a single sample, since analysis results will not be correct. Review the sample names in each BAM file on the destination server. If multiple BAM files are listed under a single sample, create a new sample BAM file for each BAM file that is needed on the target sequencer.
39367	If you have a new installation of the Genexus™ Integrated Sequencer and do not yet have any fusion references installed, an upload of a fusions panel ZIP file (that contains a reference.bed and reference.fasta) directly from AmpliSeq.com the upload can fail.	Ensure that you upload a fusions reference <i>before</i> you upload the fusions panel ZIP file. Alternatively, import (from the software updates screen) or upload (from the <b>Manage Assays</b> screen) an assay definition file (ADF) from Thermo Fisher Connect that contains a fusion reference.



Issue number	Issue Summary	Impact and Mitigation
40236	<p>The name of two gnomAD Annotation genomic references are shown incorrectly in the Annotation tab of the Results screen.</p> <ul style="list-style-type: none"> <li>• "Alternate allele frequency in samples of Non-Finnish European ancestry" is shown as "GnomAD FNFAF". The correct name is "GnomAD ENFAF".</li> <li>• "Alternate allele frequency in samples of male ancestry" is shown as "GnomAD MAF". The correct name is "GnomAD MAL".</li> </ul>	N/A
42847	<p>When multiple sample PDF reports are downloaded from the Sample Results or Run Results screens, folders for the downloads are sometimes empty.</p>	Review and download individual reports for each sample in the <b>Sample Reports</b> .
42946	<p>PQ runs with names that exceed 25 characters are shown incorrectly in PDF reports. The run plan name overlaps with other fields such as PQ status in the reports.</p>	Field service representatives should use PQ run names with fewer than 25 characters.
43231	<p>The <b>Required</b> column in the sample attributes table for the <b>Sample Type</b> and <b>Collection Date</b> attributes might not display values correctly in the <b>Manage Attributes</b> screen. The value might be No, when it should be Yes. This has no impact on the creation of a sample in the software.</p>	N/A
43896	<p>In the <b>Annotations</b> tab of the <b>Manage Presets</b> screen, the value in the <b>Last Modified On</b> column in a system-installed annotation set that has been associated with an assay changes from N/A to a date. The content of the system-installed annotation set is not changed when the annotation set is associated with an assay.</p>	N/A

Issue number	Issue Summary	Impact and Mitigation
36669	<p>If you use have run results with one variant that has more than one allele and more than one of those alleles is annotated as a UCSC Common SNPs annotation, and a filter chain that includes the UCSC Common SNPs filter was applied, the results for returned variants are not as expected. This includes the TMB score filter chain and the <b>Oncomine Extended</b> filter chain.</p>	<p>When you create custom filter chains in Genexus™ Software 6.8, we recommend that the UCSC Common SNPs filter not be used. Use other population database filters such as dbSNP, ExAC, and 5000Exomes population filters.</p> <p>Alternatively, for Genexus™ Software runs that use a system-installed assay, you can use the Download All Variants function in the software to get the VCF file from Genexus™ Software and upload the VCF file to Ion Reporter™ Software. Then in Ion Reporter™ Software, launch the appropriate annotate analysis workflow that uses the same assay name and apply the predefined or custom-created filter chain that includes the UCSC Common SNPs annotation filter to get the expected results.</p> <p>For Genexus™ Software runs that use a custom assay, you can use the Download All Variants function in the software to get the VCF file from Genexus™ Software and upload the VCF file to Ion Reporter™ Software. Then in Ion Reporter™ Software, launch the Annotate variants single sample analysis workflow and apply the predefined or custom-created filter chain that includes the UCSC Common SNPs annotation filter to get the expected results.</p>
35975	<p>While planning a run, if you select an Ion Reporter™ Software account is used that has a EULA that is not accepted, an error is shown.</p>	<p>You must sign into the Ion Reporter™ Software account and accept the EULA. Alternatively, select another Ion Reporter™ Software account to use in the run plan.</p>
35963	<p>Some content that appears in the software is not correctly translated into supported languages. The content is shown in English in the software screens.</p>	N/A
35269	<p>If a run is edited within the same day that automatic or manual backups or cleanups are completed, run data might be deleted during the cleanup process.</p>	<p>Ensure that you wait 24 hours after run results are restored or cleaned up in the software before runs are edited.</p>
36910	<p>When you create a sample from a BAM file and select <b>Breast Cancer</b> as cancer type sample attribute, the system-installed <b>Breast Cancer</b> gene list is applied to the run results by default. The Breast Cancer gene list does not include BRCA1 or BRCA2 genes.</p>	<p>Select “No Gene List” in the Key Findings tab to enable the display of BRCA1 or BRCA2 in the software.</p> <p>Alternatively, create a custom gene list with cancer type of <b>Breast Cancer</b> that includes BRCA1 and BRCA2 for use with the BRCA Assay.</p>

Issue number	Issue Summary	Impact and Mitigation
35191	When you export an assay that is created from a custom panel ZIP file that is downloaded from ampliseq.com, and then import the assay to another Genexus™ Integrated Sequencer, on which the same custom panel ZIP file has been uploaded, a validation error for a panel mismatch will occur.	Create a custom assay on the Genexus™ Integrated Sequencer on which you want to import the assay (that is, the target sequencer), that uses the same presets as that of source Genexus™ Integrated Sequencer. Assay parameters can then be exported from the source sequencer and imported to the target sequencer.
36378	When you create a report template that includes a section for Images, the option "Browse Image", which is used to browse for an image and add it to the template, does not work if the template is used to automatically generate a report during a run	Use the report template to generate a variant report after the run is completed. In the Generate Report dialog box, click Upload Image, then select Browse Image and click Select File, then select the image or images to include in the report. Enter a title for the image, and if needed enter a description and footnote for the image. Continue with the steps to generate the variant report, as described in the Genexus™ Software help system.
34986	Responses for an API request for a report-templates that include a specific report template ID, include all report templates instead of only the report template that is associated with the report template ID.	N/A
36942	When you review run results for PQ runs in Genexus™ Software. from the Dashboard, you will see links named Download Files, Report and Reanalysis. If you click any of the links, you might receive an error or see incorrect results.	The errors do not affect the original PQ run or the state of the Genexus™ Integrated Sequencer. <b>Mitigation:</b> Do not review PQ sample results from Dashboard. Instead, go to Results > Verification Results to view PQ run results.
36195	The Non-Human Reference assay type is not currently supported for Bam to Result runs that are planned with Genexus™ Software.	N/A
21701	Software assays that are created in Genexus™ Software, or assays that are copied and edited, and contain a Sequence Variant Baseline file that was exported from Genexus™ Software 6.2 cannot be imported into a Genexus™ Integrated Sequencer that uses Genexus™ Software 6.6 or later.	Upon upgrade from Genexus™ Software 6.6 to Genexus™ Software 6.8, you can copy the assay that will be exported, or enter at least one edit to the assay. When one of these actions is done, the assay can then be exported and imported to Genexus™ Integrated Sequencer that uses Genexus™ Software 6.6 or later.
21491	For SARS assay results, when a sample is uploaded to another Genexus™ Integrated Sequencer, the sample is defined as DNA BAM Sample instead of RNA (the SARS assay is an RNA assay).	When the import to the target sequencer is complete, you can edit the sample, map the BAM to the RNA BAM file attribute and relaunch the BAM to Result run.
33039	When you create a new report template and select the option to add an image, some formats which are not supported are shown in the software.	Select only JPEG, PNG, GIF, or TIFF formats in the software. Other image formats are not supported.

Issue number	Issue Summary	Impact and Mitigation
34392	When you create a new report template with the "Other" application category, the new report template shows the application category as "Default" instead of "Other".	N/A
31888	If you copy and edit an Oncomine™ TCR Beta-LR - GX5 - w1.3.0 assay and set the chip type to Ion Torrent™ GX7™ Chip, the Templating Kits field in the software workflow step contains the incorrect selection. And, if you select the GX7 templating kit, the software might show an error.	The Oncomine™ TCR Beta-LR - GX5 - w1.3.0 assay is not compatible with the Ion Torrent™ GX7™ Chip. We recommend that you start over on the edit of the assay from the Manage Assays screen, and use only selections for the Ion Torrent™ GX5™ Chip.
34543	Software counts in the Manage Gene Lists screen show a count that incorrectly includes obsoleted/inactivated gene list records and should not be counted.	Inactivated gene list records will get added to the counts of the Gene Lists total items that are available in table, but not show up in the Gene Lists table.
34226	When a new report template is created for an Oncomine™ Comprehensive Plus - DNA Assay or an Oncomine™ Comprehensive Plus - DNA and Fusions Assay, if the Complex Biomarkers section is removed from the Configuration pane, the section cannot be added again from the Add Section dropdown menu, or with the Bulk Add option.	This affects reports for Oncomine™ Comprehensive Plus - DNA Assay and Oncomine™ Comprehensive Plus – DNA Assay and Fusions Assay. To restore the Complex Biomarkers section to the Configuration pane in the Report Template, refresh the page and select the Oncomine™ Comprehensive Plus assay and select starting template as Oncomine™ Comprehensive Plus from Start From Existing Report Template dropdown.
34359	Previous versions of the CoverageAnalysis and MolecularCoverageAnalysis plugins are shown in Genexus™ Software 6.8. However, the previous versions are not compatible with the latest version of the software.	If you use Genexus™ Software 6.8, ensure that you use CoverageAnalysis 5.18.02 plugin and MolecularCoverageAnalysis 5.18.02 plugin. If you use Genexus™ Software 6.6, ensure that you use CoverageAnalysis 5.16.0.4 and MolecularCoverageAnalysis 5.15.0.5 plugin.
34494	If you create a custom Germline assay for use with the GX5 and GX7 chips, and change the templating size to 400 bp, the assay shows an incorrect number of flows. (550 flows instead of 852 flows).	After you create the assay, edit the sequencing flow to 852, which is the default settings for all other templates.
35417	The option to select an assay of interest to upload the sample results (BAM files) and automatically start an analysis with the SARS assay on another Genexus™ Integrated Sequencer is not available. The option to Upload Only is available.	If you transfer a SARS assay to another Genexus™ Integrated Sequencer, create a BAM to result planned run to analyze the run on the second Genexus™ Integrated Sequencer.

Issue number	Issue Summary	Impact and Mitigation
29273	A single BAM file that includes all barcodes for a single sample is generated in addition to PNG files for sequencing runs that use multiple pools. Genexus™ Software does not generate individual BAM and PNG files for each pool-wise barcode of single sample. As a result, these BAM and PNG files are not available to download.	For sequencing runs that use multiple pools, download a single BAM file or a PNG file that includes all barcodes for a single sample.
34360	When you use the DNA and Fusions application category to create or copy and edit an assay for the OncoPrint™ Plus panel, the option is available. However, the MSI is not supported for DNA and Fusions samples.	N/A
35268	If a run is edited within the same day that automatic or manual backups or cleanups are completed, edits that affect the run data might be deleted during the cleanup process.	Ensure that you wait 24 hours after run results are archived or restored in the software before runs are edited.
30298	When you create an exon tile assay baseline and select samples, you must select samples for the appropriate panel and for the correct library chemistry.	Ensure that you use only samples from the panel for which the baseline will be used. The samples should not be from multiple panels or multiple library chemistry types. For example, do not mix AmpliSeq™ and AmpliSeqHD™ samples in the same baseline.
33953	The template file used to import library batches has a column named "Extraction Method Type" for the Nucleic Acid Type template item.	Use the column named "Extraction Method Type" to enter information about nucleic acid types in the library batch template file
34418	A gene symbol that does not have a valid HGNC ID is not displayed under the Annotations tab in sample results shown in the Variants tab in the software.	Export the samples file (TSV or XLS format) to view all gene symbols, and make corrections to the sample entries if desired.
11380	The sorting function of the Locus column on the Run Results screen does not work as expected. Although you can click on the column heading to sort the data in the column, the list is not sorted numerically by chromosome number.	To sort records numerically, sort the list of chromosomes.
35478	If a target regions file includes sample ID (SID) regions, and variants are detected in those regions, then those variants will be listed as a non-hotspot variants in the results in the Variants tab of the Key Findings screen.	You can manually review these variants and not include them in reports, if the variants belong to SID regions.
36096	When you upload duplicate files that have the same file names for DNA and Fusion panel designs, you will see the same error message for both uploads.	Change the filename to resolve the issue and no longer see the error message.

Issue number	Issue Summary	Impact and Mitigation
34784	For aneuploidy run results, no filter chain is associated when no value for a filter chain is selected in the Run Results screen.	Select "No filter chain" or any other filter chain that is shown in the results page, then regenerate the report. After a filter chain is selected from the dropdown menu, the report can be generated successfully.
35940	When you prepare a library batch and use the Include NTC option, the NTC sample is not included in the library batch that is created if you select the maximum number of samples that can be added to an assay. For example, if you use the assay Oncomine™ Precision - GX5 - Solid Tumor - DNA and Fusions - w3.2.0, the maximum allowed samples is 16, and the maximum allowed libraries is 32.	N/A
22891	If you create and save a run plan, then click Edit in the Manage Runs screen, then click through the workflow steps, but do not make any changes, an Edit entry is shown in the Audit screen. The entry has blank values for Old Values and New Values. The entry shows that an Edit of the run plan occurred, although no edits were made to the run plan.	N/A
35258	Although 3 characters are required to search the list of filter dropdown values in the Sample Results table, the entry of 2 characters will generate a list of available filter dropdown values that pertain to the current screen only.	Enter at least 3 characters when using your search for filters in the Sample Results screens to search across all the records.
35450	Reports that are signed off for runs that are performed and backed up on Genexus™ Software version 6.6 cannot be exported from, or imported to, another Genexus™ Integrated Sequencer.	N/A
35291	There are some system-installed assays that are preconfigured to generate RNA Exon Tile Fusion Imbalance results during analysis. If you use a custom assay that is created by copy-editing such assay, you can view RNA Exon Tile Fusion Imbalance results and generate a visualization from the software. If you create an assay without these predefined settings, you cannot view RNA Exon Tile Fusion Imbalance results in the software.	Use a copy of the system-installed assay to analyze or reanalyze the run results, then view the RNA Exon Tile Fusion Imbalance visualization for the sample results for the reanalyzed run.

Issue number	Issue Summary	Impact and Mitigation
36574	BRCA germline analysis workflows are not shown in the list of analysis workflows to select for transfers of samples (BAM files) from Genexus™ Software to Ion Reporter™ Software.	Sign into Ion Reporter™ Software and manually launch the analyses for samples that are transferred from Genexus™ Software.
36047	The software does not enforce the use of password protected SSH keys, but the use of a password for the SSH keys is strongly recommended. Do not share the SSH private key or the credentials for the SSH private key. The SSH key should be used only by the user who creates the SSH key.	Do not share the SSH private key or the credentials for the SSH private key. The SSH key should be used only by the user who creates the SSH key. <b>Note:</b> This information is included in the Genexus™ Software User Guide, but is not in the software help system.
36678	When you create a new assay, or copy-and-edit an assay, the Annotation parameter value “Functional Annotations For All Alleles” does not work correctly.	To create a new assay, or copy-and-edit an assay in Genexus™ Software that includes the Annotation parameter value “Functional Annotations For All Alleles”, you must use an annotation.json file that is manually edited. Contact your Field Bioinformatics Scientist (FBS) or Field Support Engineer (FSE) for assistance with the change.
35679	Manager and administrator-level users can augment an existing copy number baseline. The software allows you to select an existing baseline for augmentation from the dropdown menu, even if the baseline is obsolete.	Do not use select an obsoleted baseline when you augment a copy number baseline.
35976	When Genexus™ Software runs that contain samples which include the Collection Date attribute are uploaded to Ion Reporter™ Software, the Sample Collection Date attribute is not shown for the samples in Ion Reporter™ Software.	Edit the sample in Ion Reporter™ Software and define the Collection Date attribute value.
33289	Under rare circumstances, the Allele Frequency (AF) of a long insertion or deletion (INDEL) is calculated incorrectly such that the frequency may be greater than 100% when the long INDEL is discovered by the long indel assembler of the software.	N/A
34020	If data for annotation tracks in Variant Pileup view have the same start and end base positions, detailed information is not shown in the Gene Browser in the Variant tab, when you click on the green tiles.	To view details for the data in the annotation tracks, you can open the results in the Broad Institute’s Integrative Genomics Viewer (IGV).
35729	When you upgrade to Genexus™ Software 6.8, email notification settings that are set in the Backup and Restore screen might be reset to the default settings.	In the Backup & Restore screen, in the Email Notifications Settings section, set the email notifications to the settings that were used prior to the upgrade.

Issue number	Issue Summary	Impact and Mitigation
35705	When you upload samples to another Genexus™ Integrated Sequencer and select the option to automatically start a run with the selected assay, it might fail to launch the assay although the samples are successfully uploaded, if the assay that you select in the <b>Genexus Workflow</b> column has a trademark in the name.	This affects the system-installed aneuploidy assays in the software, which have trademarks in the names. Plan and use a BAM to Results run on the target Genexus™ Integrated Sequencer for large number of samples that use the assay
37196	In a sequencing run that uses multiple assays, reanalysis of multiple run results with the same assay might result in an error or incorrect results.	Reanalyze only run results that use a single assay at a time.
37524	Manager- and administrator-level users can add new custom genome reference files in Genexus™ Software, which are listed in the References table. Custom genome references can be obsoleted with the actions menu in the table. When the obsolete action is used, the software shows the message "Reference <Reference Genome Name> successfully obsoleted" but the software is not updated to reflect the obsolete state.	An obsoleted custom genome reference file will be available to add or import to a panel. However, we recommend that you do not use reference genomes files that are obsoleted in the software.
37516	The VCF entry for fusions imbalance does not produce READ_COUNT entry in a VCF results file. In such cases, the Genexus™ Software 6.8 Variant tab shows a Read Count of 0.	The issue also affects comparison of results in the software and variant PDF reports.
36284	If a sequencing run is stalled for more than 120 hours, go to Settings -> Services and review the list of runs in the Active Jobs section. The run will not be listed under Active Jobs.	If the run is still active but the run is not listed under the active jobs, reboot the Genexus™ Integrated Sequencer. After the reboot, the run status will be marked as Stalled, and the Reanalyze action will show in the list of runs. Reanalyze the run so that its analysis can be completed.
36417	The CoverageAnalysis results for the DNA Barcode under the Barcode column cannot be opened in the Integrated Genomic Viewer (IGV) if you click the option to Show or Hide the Panel, then select the Total Reads or Strand Reads options from the 'Reference Coverage Chart'. An error is shown when the IGV.jnlp file is downloaded and opened.	The .jnlp file that is downloaded from CoverageAnalysis results does not launch IGV. However, you can view and download the .jnlp file from Variants tab for use with IGV.



Issue number	Issue Summary	Impact and Mitigation
35198	If you restore a run that has no backup of one or both of these file types: sequencing output files and intermediate files, or one of these file types, an incorrect audit record is displayed. When the run restoration is completed under these circumstances, the audit record includes "Sequencing Output & Intermediate Files Deleted = false" under New Values. However, this audit record is incorrect because the backups are not available, and therefore the sequencing output files and intermediate files for the given run cannot be restored.	N/A
35155	When an administrator user tries to edit the profile for an already existing user who has lock-level sign off permissions, the lock symbol is not shown next to the selected Electronic signature type in the User management screen. The lock symbol is shown correctly when an administrator user creates a new user and wants to assign the locked Electronic signature from the new user configuration page.	N/A
34115	If you reanalyze a sequencing run that has been imported to another Genexus™ Integrated Sequencer, information about reagents is missing from the Reagent section of the Run Summary screen.	View the information in the Run Report PDF that you can download from the Reports tab.
32559	Some sample results screens list additional incorrect statuses in the Sample Status column when using the "Terminated" filter, including Library Preparation Aborted, and Resequencing Aborted. The sample results screen should instead show statuses only of Terminated for such results.	N/A
34074	A plugin cannot be rerun on a sample result if a plugin run is currently in progress for the sample result.	To avoid the potential loss of plugin results data, do not rerun a plugin on a sample result when a run for the same plugin is in progress. Wait until the plugin run is complete before you rerun the plugin.
35785	In some cases the software will show an error on the Sample Results page if you apply a filter in any column of the table and click on the "Started On" column.	The error does not affect the results.

Issue number	Issue Summary	Impact and Mitigation
37197	When uploading sample files in BAM format from one Genexus™ Integrated Sequencer to another, if any sample name is a partial name of another sample name, sample attributes, for the transferred sample on the sequencer to which the samples are transferred, are likely be incorrect.	After transferring samples in BAM format from one sequencer to another, check any sample attribute names in which a partial name of another sample name is included in any sample name. Some sample attributes, such as cancer type and tumor cellularity directly impact sample results. If sample attributes are incorrect after sample files are transferred in BAM format, edit the sample attributes on the sequencer to which you transferred the samples.
37336	Gene coverage graphs display correctly in run results but do not display correctly after the BAM result file is transferred to another Genexus™ Integrated Sequencer.	N/A
37629	If a new assay is created in Genexus™ Software 6.6 or earlier, then copied and edited in Genexus™ Software 6.8 or later and chip type is changed to GX7, the advanced Command Line Arguments parameters are not correct in the newly created assay.	Only the GX5 chip can be used with Genexus™ Software 6.6 or earlier. The GX7 chip can only be used with Genexus™ Software 6.8 or later.
37625	When you view results in the Variants tab that include the variant type RNAExonTiles, if the "Reads" column for the genes, NTRK1, NTRK2, or FGFR3 has a value of 0, and you click on the variant ID, the Pileup will not load any reads. The Pileup view will be blank.	Close and reopen the results screen to view other variants subsequently in the Pileup view.
37455	Information is missing from the info.csv file that is downloaded for runs completed on the Genexus™ Purification System.	Review the purification run name and instrument serial number are available in the Run Summary in the software. Alternatively, work with your Field Service Engineer (FSE) to get the information from a Customer Support Archive file.
37452	To complete sequencing runs with the Ion Torrent™ Genexus™ Total RNA Purification Kit for a cell/tissue sample type, the Sample to Report run is not supported.	Plan a Genexus™ Purification Instrument only run in standalone mode, then transfer samples to a Genexus™ Integrated Sequencer to perform a sequencing run.
36863	In rare cases, if the variant is not properly right-align in the input VCF file from a given analysis, the cdot annotation might be reported incorrectly.	N/A

Issue number	Issue Summary	Impact and Mitigation
36333	In rare cases, a reported fusion with good reads count, might show zero or a very few number of reads with the Broad Institute's Integrated Viewer (IGV), which are much less than the number of reads reported in the Ion Reporter™ Software Analyses Results screen. This could be caused by an additional exon that is amplified and sequenced by existing primers, which becomes unmapped due to the presence of a large insertion. There could also be other technical or biological reasons.	Contact your Field Bioinformatics Scientist (FBS) or Field Support Engineer (FSE) for assistance.
36318	When the Genexus Security Package v1.0.0 is installed, at the end of the Software Update installation process, on the Server Update screen, the "Go To Application" button does not get enabled even if the upgrade has completed.	The workaround is to ensure that the logs show the message: "Software Update completed successfully". When you see this message, refresh the screen, and return to the Update screen, where the "Go To Application" button has become active.
35505	The option to run a plugin is not shown immediately after a signed-off report is deleted from the Reports screen.	Refresh the Reports screen to see the option to run a plugin.
34802	The Ploidy Plot is missing from the PDF report for runs that use the ReproSeq™ PGS - GX5 Assay kit. run when the run results are Aneuploidy Positive or Aneuploidy Negative.	N/A
34414	Sometimes when the run planning page is printed, some information is truncated in the printout.	Adjust the page set up and layout prior to printing the run planning page.
34373	If a variant is an INDEL located on UTR regions, you will see the cdot annotation includes position and sequence change information but the information is not in an HGVS-compliant format.	This issue affects results shown in both Genexus™ Software and Ion Reporter™ Software.
33902	For some Genexus™ Software assays, a PDF report that uses a custom template and includes additional columns that were not in the original assay, might show no values the value in the additional columns in the report.	Contact your Field Bioinformatics Scientist (FBS) or Field Support Engineer (FSE) for assistance.

Issue number	Issue Summary	Impact and Mitigation
25297	If you create a custom sample attribute that has no space between the words that is named the same as a mandatory sample attribute that does include a space between the words on the same on one Genexus™ Integrated Sequencer, then transfer the samples through the IonReporterUploader plugin, the Plugins tab will show sample transfer is completed and valid. However, the uploaded samples are not listed in the target Genexus™ Integrated Sequencer.	Create sample attribute with underscore to separate the words or use different letter character case or words.
36387	A known issue in Genexus™ Software that might yield false positives in OncoPrint™ Precision Assay. When a de novo deletion c.700_712delTACAACACTACATGT (i.e., CACATGTAGTTGTA to C at chr17:7577568) in TP53 gene is present in the sample, it potentially causes false positives for the hotspot SNPs COSM44321 and COSM45329.	Examine the evidence of the hotspot SNP in a Gene Browser to confirm whether the SNP calling is correct. The call is correct if both the de novo deletion and the hotspot SNP are reported as present by the software.
36793	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. Each of these software processes must be done individually, that is, you cannot create, copy or edit multiple assays in Genexus™ Software simultaneously. Opening more than one tab and trying to create, copy or edit assays in the multiple open tabs will result in errors and loss of entered data.	Do not open multiple tabs in Genexus™ Software for procedures that involve data entry.
35786	A Sample Status of "BaseCallingActor: Running" is sometimes shown for in sample results for runs that use the OncoPrint™ Comprehensive v3 Assay and are terminated.	N/A
28966	If you create an assay in the software with the Non-Human Reference assay type, the Annotation tab is shown in the Parameters step of the Create Assay workflow bar. This tab should not be visible.	The Annotation tab has no effect on the creation of an assay for the Non-Human Reference assay type.

Issue number	Issue Summary	Impact and Mitigation
35391	When you compare sample results in Genexus™ Software, you are able to select samples with a sample result Status of Obsolete to be included in the comparison and include the comparison in a report for multiple samples.	The use of obsoleted samples for sample comparisons is not recommended.
37535	The software allows you to create a report template that has the same name as a report template that is obsolete.	The use of report templates names that are used for an obsolete report template is not recommended.
21541	If you import a custom exon tile assay baseline file in an unsupported file format, you are not notified that the import failed. The unsupported file appears in the exon tile assay baselines list after import. However, analysis fails if the custom assay baseline is used in a run plan.	This issue occurs only if you attempt to import a custom exon tile assay baseline file in an unsupported file format. <b>Mitigation:</b> Ensure that custom exon tile assay baselines that you import are in a supported file format.
20696	In any column header, when you click <b>Filter</b> , enter any information in the search field, then click <b>Enter</b> , all options that are listed in the filter dialog box are selected.	This issue applies to all searches in the filter dialog boxes in all screens. <b>Mitigation:</b> In the filter dialog boxes, do not click <b>Enter</b> except to select all options.
12692	Missing variant label and legend chart customization options for charts generated when you compare samples.	This is a chart customization issue that does not affect sequencing results.
22895	<p>In the <b>Key Findings</b> tab, in the <b>Key Variants</b> matrix, variants with more than one gene annotation can be listed as <b>Not Assayed</b> even if one of the annotated genes is present in the panel. Affected variants are shown in the <b>Key Variants</b> matrix only when the <b>Not Assayed</b> checkbox is selected and appear at the end of the matrix in a white tile instead of a blue tile.</p> <p><b>Example:</b></p> <div data-bbox="558 1493 919 1640" style="border: 1px dashed gray; padding: 5px;"> <p>U2AF1L5 SNV/Indel  COSM166866  AA Change: p.?  Allele Frequency: 0.066</p> </div>	<p>This issue can affect variants with more than one gene annotation: for example, U2AF1L5/U2AF1.</p> <p><b>Identify if a variant is affected by this issue.</b></p> <ol style="list-style-type: none"> <li>For any gene tile that is shown only when the <b>Not Assayed</b> checkbox is selected, click the Variant ID to open the <b>Variants</b> tab.</li> <li>Scroll up to view the variant in the Variants table.</li> <li>In the <b>Variants</b> table, in the <b>Gene</b> column, if the variant is listed with ** to indicate that more than one gene is detected, click the <b>Annotations</b> tab.</li> <li>Scroll to the <b>Gene</b> row to view the multiple annotations for the gene.</li> </ol> <p><b>Mitigation:</b> When you view the <b>Key Variants</b> matrix, ensure that you select the <b>Not Assayed</b> checkbox, if available, to review variants that are not assayed.</p>

Issue number	Issue Summary	Impact and Mitigation
17425	In rare instances, the order of the reads shown in the BAM track is not consistent each time you view the BAM track in the <b>Gene Browser</b> .	<p>This issue occurs only when the total read count exceeds the maximum read count threshold that is set for optimal browser performance. When the threshold is exceeded, the <b>Gene Browser</b> shows a specified number of randomly sampled alignments configured by down sampling parameters. The coverage track is not affected by this issue.</p> <p><b>Mitigation:</b> You can adjust the down sample reads parameter and view the full BAM track with the Broad Institute Integrative Genomics Viewer (IGV). For more information, see “View variants with IGV” in the software help system.</p>
23068	If you create a custom panel for the OncoPrint™ BRCA Assay GX, exon numbering remains set by the system-installed panel for the assay. As a result, when you use a custom panel that is based on a reference sequence version that is different from the system-installed panel, exon numbering for BRCA1 is not correct when you visualize exon and whole gene CNVs.	This rare issue affects the visualization of BRCA1 for the OncoPrint™ BRCA Assay GX if you use a custom panel with a version of the reference sequence that is different from the system-installed panel for the assay. The system-installed version of the reference sequence is NM_007294.4.
21320	If the name of a variant report template includes a special character of @, *, #, or \$, you cannot view or download variant reports generated with the template.	<p>This issue affects any variant report with @, *, #, or \$ in the name of the variant report template.</p> <p><b>Mitigation:</b> Create a new variant report template without @, *, #, or \$ in the name, then generate a new report.</p>
23081	When you try to prepare a library batch with more than 24 samples, an error message is shown for some DNA + RNA assays.	<p>This issue affects some system installed and custom DNA + RNA assays when you select more than 24 samples during library batch preparation.</p> <p><b>Mitigation:</b> Create an XLS or XLSX file that contains the library batch information from a template file provided in the <b>Import Library Batch</b> screen, then import the library batch file.</p>
20579	For runs that use an assay that has been enabled for resequencing, the log files for the resequencing portion of the run are located in a separate folder called “Reseq”.	This issue affects all runs that include resequencing.
20738	Downloading multiple Customer Support Archive (CSA) files simultaneously fails.	<p>This issue occurs when you try to download a CSA file while another CSA file is downloading. This issue occurs in the <b>Run Results</b>, <b>Sample Results</b>, and <b>Verification Results</b> screens.</p> <p><b>Mitigation:</b> You must wait for a CSA file to download successfully before you can download another CSA file.</p>

Issue number	Issue Summary	Impact and Mitigation
16765	Results generated by the coverageAnalysis plugin for RNA samples with an application category of oncology are not informative.	This issue occurs only when you view the coverageAnalysis plugin results in the <b>Plugins</b> tab for an oncology RNA sample. <b>Mitigation:</b> For oncology RNA samples, you can ignore the coverageAnalysis plugin results shown in the <b>Plugins</b> tab.
16763	If you import run results from another Genexus™ Integrated Sequencer that has a different version of a plugin used in the run, you cannot see the plugin results in the <b>Plugins</b> tab when you view the results on the target server.	This issue affects results that are imported in a Genexus™ Software server that has a different version than the source Genexus™ Integrated Sequencer. <b>Mitigation:</b> Run the plugins again in the target server (from a sample result, click the <b>More Options ► Run Plugin</b> ).
21807	When you create some custom assays or copy and edit some assays, you can enable, edit, and view Microsatellite Instability (MSI) parameters in the <b>Parameters</b> step of assay creation even though the MSI parameters are not applied in the analysis.	This issue affects some custom assays. <b>Mitigation:</b> Ignore MSI parameter settings in custom assays when results do not show MSI metrics.
20696	In any column header, when you click <b>Filter</b> , enter any information in the search field, then click <b>Enter</b> , all options that are listed in the filter dialog box are selected.	This issue applies to all searches in the filter dialog boxes in all screens. <b>Mitigation:</b> In the filter dialog boxes, do not click <b>Enter</b> except to select all options.
23074	The <b>Oncomine Extended (5.14)</b> filter chain launched in Genexus™ Software 6.2.1 was updated in Genexus™ Software 6.6. The improvements, which are part of <b>Oncomine Extended (5.16)</b> filter chain, are also included in the <b>Oncomine Extended (5.14)</b> filter chain in Genexus™ Software 6.6. This update can result in rare differences to Filtered Variants in Genexus™ Software 6.6 when compared to results viewed with the <b>Oncomine Extended (5.14)</b> filter chain in Genexus™ Software 6.2.1.	This rare issue affects results that are viewed when the <b>Oncomine Extended (5.14)</b> filter chain is applied and compared to results in Genexus™ Software 6.2.1. This rare issue could affect the following Filtered Variants. Likely somatic de novo splice variants that are not hotspot splice variants or ClinVar pathogenic variants are filtered in. (Hotspot splice site variants and ClinVar pathogenic variants are not affected by this issue.)
20875	For samples with a disease category of cancer that were created in Genexus™ Software 6.2, you cannot plan a run or generate a report unless you first edit the application category of the samples.	This issue affects samples created in Genexus™ Software. Disease category was a sample attribute in version 6.2, but in version 6.6, the correlated attribute is application category. (Cancer is not an available option in version 6.6.) <b>Mitigation:</b> For samples with a disease category of cancer created in Genexus™ Software 6.2, edit the application category before you plan a run or generate a report with the samples in Genexus™ Software 6.6. Alternatively, you can copy samples that you created in version 6.2, edit the application category for version 6.6, then plan a run with the new samples.

<b>Issue number</b>	<b>Issue Summary</b>	<b>Impact and Mitigation</b>
21487	BAM to Result runs are not compatible with assays that are created or installed in previous versions of the software.	This issue affects all system-installed or custom assays from Genexus™ Software 6.2.1 or earlier.



## System Requirements

Genexus™ Software 6.8 has a browser-based interface that can be viewed using Google™ Chrome™ version 90 and above (version 90 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.

### Version information

Release Date	September, 2023
Release Version	6.8

## Software plugin versions

Plugin	Version
customersupportarchive	0.6.5
coverageanalysis	5.18.0.2
molecularcoverageanalysis	5.18.0.2
sampleid	5.18.0.2

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

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05 September 2023