Bionano VIA™ Software Version 7.2.2 Release Notes

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Revision History

REVISION	NOTES
A	Initial release.
B	Updates for VIA 7.2 release
C	Updates for VIA 7.2.1 update
D	Updates for VIA 7.2.2 update

Bionano VIA™ Software

This document describes the v7.2.1 release of VIA software. This document provides an overview of what is changing with this release so that users may better understand the impact of moving to this version. Should users have any questions please contact software-support@bionano.com.

Introduction

Variant Intelligence Applications™ (VIA) software is a complete and integrated solution for the visualization, interpretation and reporting of genomic variants from multiple technology types. By supporting multiple genome-wide data modalities, VIA provides the most comprehensive view of genomic variants of any interpretation, annotation, and reporting software tool available. As a platform-agnostic tertiary analysis solution, VIA stores and manages distinct types of genomic data from various platforms enabling the extraction of meaningful insights from standalone or combined analysis.

The software includes algorithms to detect copy number variants (CNV) from major microarray vendors, optical genome mapping (OGM), and next generation sequencing (NGS) methodologies as well as Absence of Heterozygosity (AOH), from data types that assess B-allele frequency. VIA also provides interpretation assistance to analyze CNVs, Loss of Heterozygosity (LOH) and Structural Variants (SV) from OGM data. As a centralized analysis solution spanning technologies and application areas, VIA provides an efficient environment to keep pace with advancements in technology while retaining access to historical platform data. By being adaptive to whichever technology is used to generate CNV, LOH, or SV genomic variants, VIA provides rich annotations for the co-analysis of sequence variants from NGS to provide a complete picture of genomic variation and reveal more answers for disease association.

Supported Implementation Options

VIA is a scalable enterprise solution comprised of three components: the VIA Server, VIA Processing, and VIA Client applications. The server is the primary component of the system that manages the data in the repository. Installed adjacent to the Server is the Processing application that executes processing jobs for segmentation and annotation. The Client software is the User Interface (UI) utilized by all users to access sample data hosted in the VIA Server. There are three implementation options to deploy the VIA system; installation of the VIA Server and VIA Processing on the Bionano Access Server (BAS), local network server, or hosted in a cloud environment as illustrated below in **Figure 1**. In each implementation option, the VIA Client software is installed locally on each analyst's workstation with network access to the VIA Server. Further information on hardware requirements is available in the *Bionano VIA System Requirements* guides (CG-30577 and CG-30580).

- Option 1: VIA Server and VIA Processing are hosted on the BAS and is a common installation strategy for OGM users primarily interested in analyzing OGM data in VIA software.
- Option 2: VIA Server and VIA Processing are hosted on a local network and is a common installation strategy for NxClinical users primarily interested in array or NGS technology.
- Option 3: VIA Server and VIA Processing are hosted on a site-administered cloud environment and is a common installation strategy for users seeking production-scale analysis with VIA.

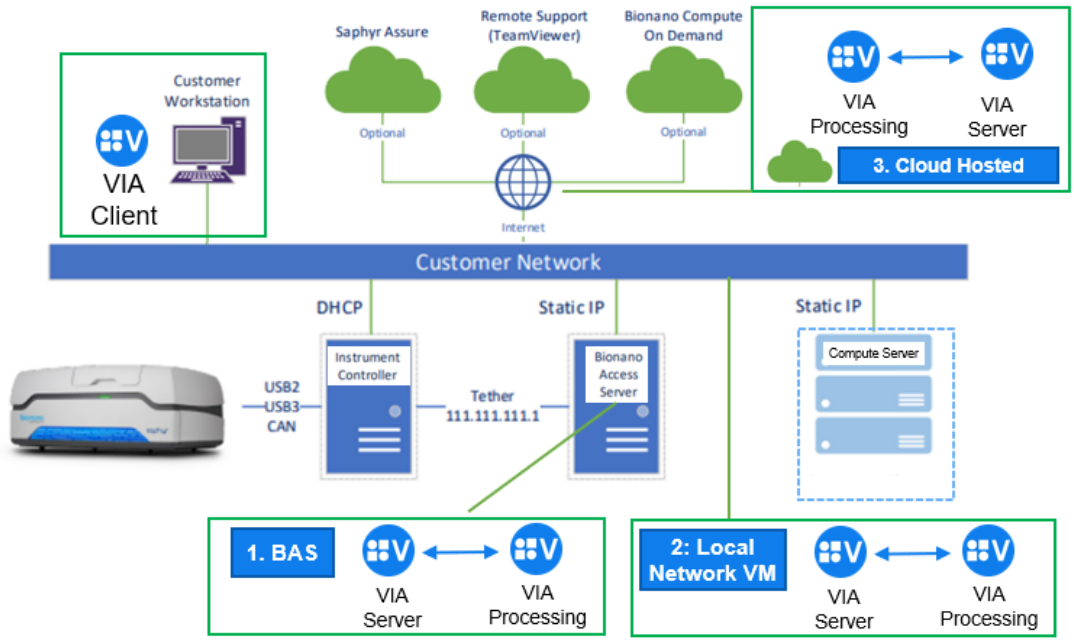


Figure 1. VIA implementation options illustration

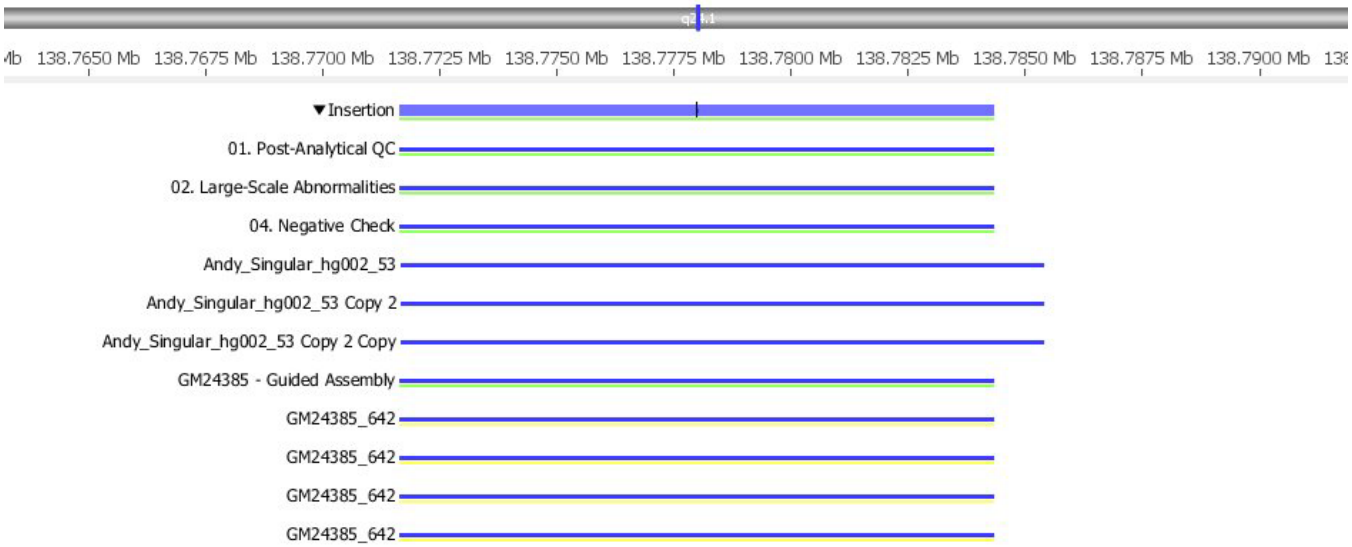
Compatibility

VIA 7.2.1 is compatible with Bionano AccessTM 1.8.3 and the Bionano Solve® v3.8.3 pipeline running on either Saphyr® Compute, Bionano Compute servers or Bionano Compute On Demand v1.4.3. Please refer to the Bionano VIA System Requirements guide (CG-30577) for hardware requirements to install the software on local network and client workstations.

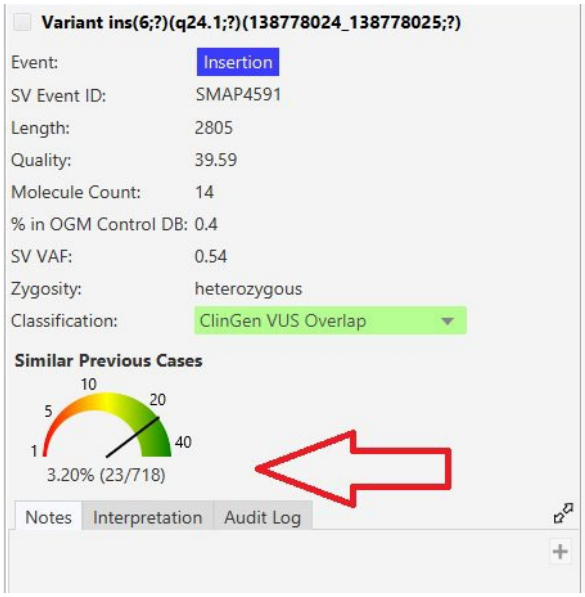
New Features in Version 7.2

Similar Previous Cases for Structural Variants

VIA now supports similar previous cases for structural variants. In the track view if similar previous cases are available a carrot (triangle) will be displayed just the left variant type. If you click on the carrot, the system will display similar cases as shown below. Click the carrot again to collapse the similar previous cases. You can click on a similar previous case name in the track view to open that sample.



Similar previous cases for structural variants also appear in the Variant Details (image below), Event Table, and SV Filtering. Similar previous cases for decision tree classification of structural variants will be supported in future versions.



Specific Region Sample Search

On the home page users can search for samples containing structural variants at specific regions or that overlap specific genes. Here are some examples:

Insertion:EGFR
Insertion:chr7:100000-150000

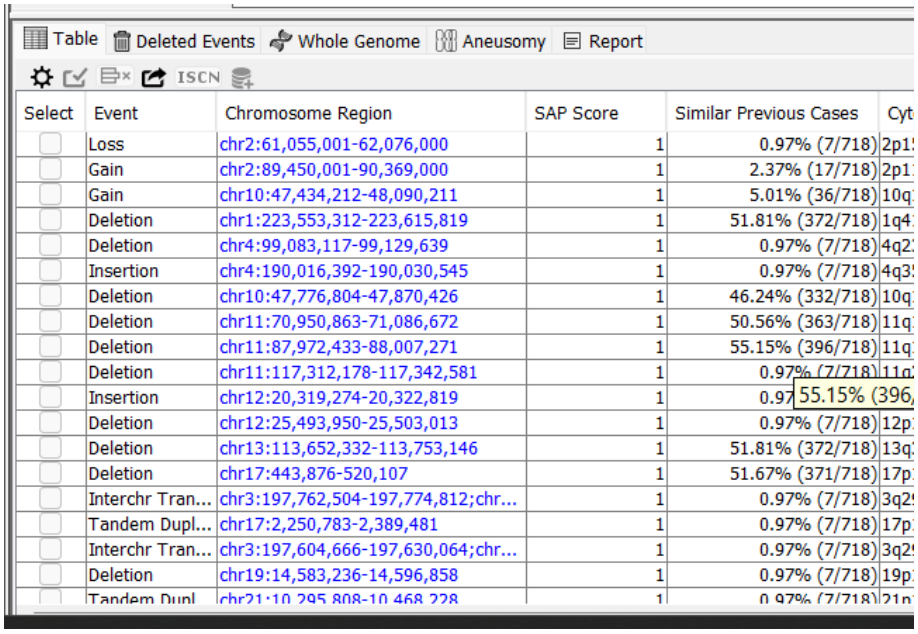
The structural variant types that are supported include the following:

- unpaired_inversion
- paired_inversion
- interchr_translocation
- intrachr_fusion
- inverted_duplication
- sv-deletion
- sv-insertion
- sv-tandem_duplication
- sv-inversion
- inversion_breakpoint

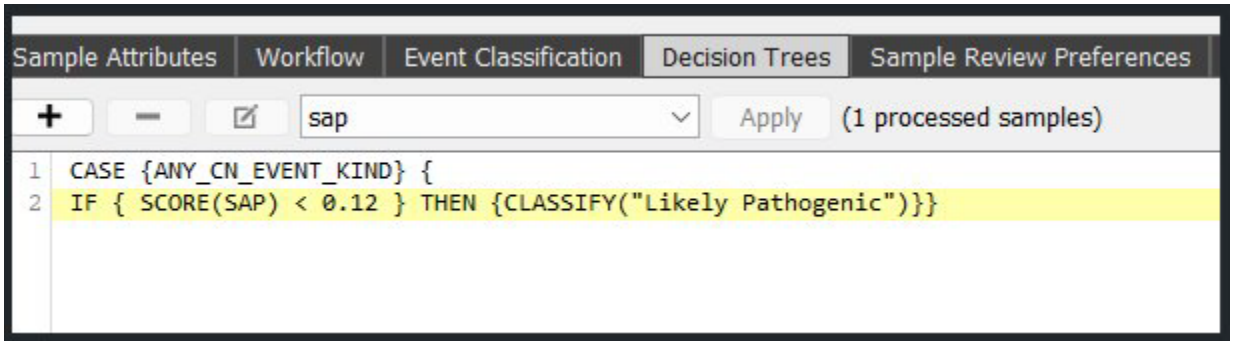
SAP Score for SV Events

The Significance Associated with Phenotype (SAP) score is now calculated for structural variant event types. The SAP Score column is available in the Event Table and can be used in Decision Trees to classify variants. For more information about SAP scores you can refer to this article:

https://www.sciencedirect.com/science/article/abs/pii/S1525157824000291



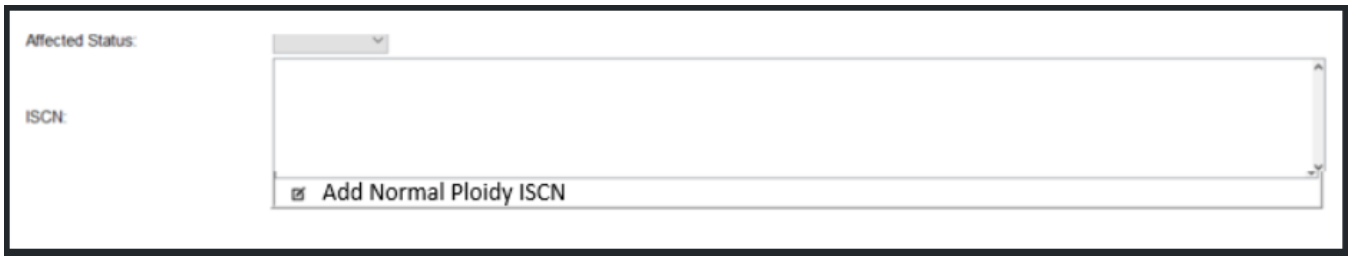
Select	Event	Chromosome Region	SAP Score	Similar Previous Cases	Cyt
	Loss	chr2:61,055,001-62,076,000	1	0.97% (7/718)	2p1
	Gain	chr2:89,450,001-90,369,000	1	2.37% (17/718)	2p1
	Gain	chr10:47,434,212-48,090,211	1	5.01% (36/718)	10q
	Deletion	chr1:223,553,312-223,615,819	1	51.81% (372/718)	1q4
	Deletion	chr4:99,083,117-99,129,639	1	0.97% (7/718)	4q2
	Insertion	chr4:190,016,392-190,030,545	1	0.97% (7/718)	4q3
	Deletion	chr10:47,776,804-47,870,426	1	46.24% (332/718)	10q
	Deletion	chr11:70,950,863-71,086,672	1	50.56% (363/718)	11q
	Deletion	chr11:87,972,433-88,007,271	1	55.15% (396/718)	11q
	Deletion	chr11:117,312,178-117,342,581	1	0.97% (7/718)	11q
	Insertion	chr12:20,319,274-20,322,819	1	0.97 55.15% (396/	
	Deletion	chr12:25,493,950-25,503,013	1	0.97% (7/718)	12p
	Deletion	chr13:113,652,332-113,753,146	1	51.81% (372/718)	13q
	Deletion	chr17:443,876-520,107	1	51.67% (371/718)	17p
	Interchr Tran...	chr3:197,762,504-197,774,812;chr...	1	0.97% (7/718)	3q2
	Tandem Dupl...	chr17:2,250,783-2,389,481	1	0.97% (7/718)	17p
	Interchr Tran...	chr3:197,604,666-197,630,064;chr...	1	0.97% (7/718)	3q2
	Deletion	chr19:14,583,236-14,596,858	1	0.97% (7/718)	19p
	Tandem Dupl	chr21:10,295,808-10,468,228	1	0.97% (7/718)	21p



Add ISCN for Normal Samples

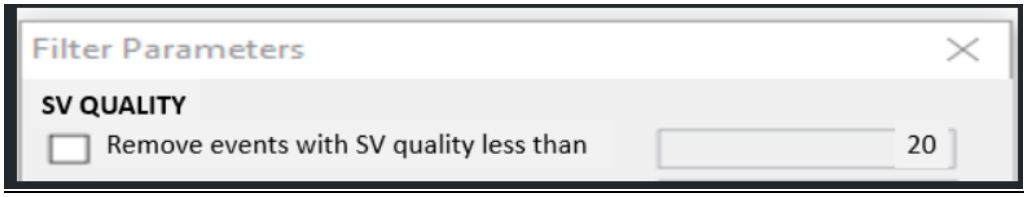
In this release it is possible to add ISCN values to normal samples that have no events to report. The ISCN values generated would match the table below.

	array	ogm
female	arr(X,1-22)x2	ogm (X,1-22)x2
male	arr(X,Y)x1,(1-22)x2	ogm (X,Y)x1,(1-22)x2



New SV Quality Filtering

In this release it is possible to filter structural variants based on their quality value as show below.

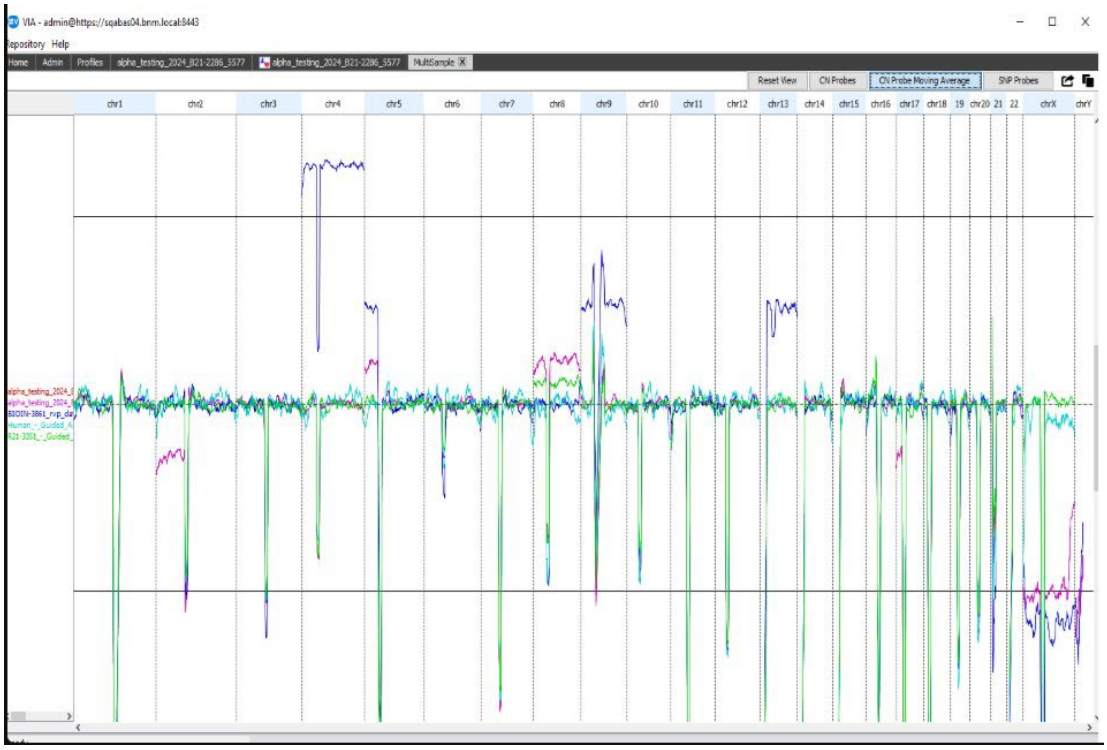


Support for Illumina Connected Annotation Files

Version 7.2 supports the import of data in Illumina Connected Annotations JSON format. For help creating a sample type to support this data format please contact our support team.

Mutli-Sample View Improvement

The mutli-sample view now sports an extra tab to show the 'CN Probe Moving Average' (image below). This new tab displays the average CN value for each sample along the genome. The left column includes a legend so you can determine which color matches to which sample.



Tickets

The features released with version 7.2 are listed in **Table 1**.

Table 1. Released features in v7.2

Summary	Ticket
Show the matched sample count of previous similar cases on the SV track	NC-5804
Show similar previous cases in the table	NC-5946
API to retrieve sample types from Access	NC-6216
Enable tags for QC metrics so that these can be added and populated in the report template	NC-6311
Support reporting for genes from databases used to annotate events	NC-6312
Add the OGM chip serial number as standard attribute for OGM Sample Type	NC-6334

Display Similar previous cases dial for SV events	NC-6343
Modify SV index to store/use different overlap intervals based on event type	NC-6355
Connect the REST API to the SV index via QueryManager to retrieve number of matched SV samples	NC-6358
Add new SV previous cases parameters to the sample query menu	NC-6365
Allow export of HGVS nomenclature for SeqVars in the «T:Whole Genome Results» table	NC-6370
Add similar to previous cases filter for SV events	NC-6396
Connect the REST API to the SV index to retrieve samples with similar case data	NC-6397
Add new sort-order option "Sample Name" for SV previous similar cases	NC-6400
Add query breakend direction to inputs for gene disruption queries parameterization	NC-6401
Display a CN probe moving average line for a set of samples in one chart	NC-6405
Support export and copy to clipboard for the multisample view image	NC-6417
Allow VIA to fall back to a default CRAM reference name when the UR field is missing.	NC-6426
Implement new DT Function SV_MOLECULE_COUNT	NC-6472
Implement new DT Function SV_VAF	NC-6473
Chromosome Region Serialize/Deserialize refactor for 7.2	NC-6481
Calculate a SAP score for SV event types	NC-6487
export SV data in JSON	NC-6493
Create methods to access the SV index	NC-6517
Vertical zoom for the multi sample CN moving average	NC-6520
Ignore corrupted samples from the homepage	NC-6534
Allow users to search for samples containing SVs at specific regions or gene from the homepage	NC-6538
Render expanded similar cases events in SV track	NC-6542
Allow users to include custom region values for an event in the word report	NC-6545
Display the sample name in the CN prove moving average tab only if there are probes available	NC-6548

Allow sv track query sample reference to display various attributes	NC-6560
Create new criteria using new header and decouple Gson parser version from Illumina Schema version.	NC-6564
Disambiguate DbSnp parsing in ICA	NC-6565
Parse transcript format flat map with source field in ICA	NC-6566
Only show SV prev sim cases query tab for samples with SVs	NC-6569
Perform aneusomy calculation for Affy CyChp data type	NC-6582
SV Previous Similar Cases Tool Tip	NC-6589
Display classification colored bar on SV track and Similar Previous Cases	NC-6590
Double right click on the probe moving average line plot should reset the plot	NC-6594
Add a Y-axis to the probe moving average line plot of the multi-sample view	NC-6595
Sv Previous Similar Cases options in admin preferences menu	NC-6599
SV previous similar case query should use confidence interval	NC-6605
Add ability to specify normal ISCN for samples regardless of test type	NC-6608
Allow user to click on the label of a similar previous case to open the sample in a new tab	NC-6617
Change Clinvar.Significance into a string array instead of single string	NC-6624
Remove ICA data version check (but keep the step name and the filter options)	NC-6625
Clinvar VCV links should link out to variant page	NC-6628
Create a set of similar previous cases setting for Deletion, Duplication and another set for Insertion and Inverted duplication	NC-6629
Add percent overlap parameter to similar previous case setting for Deletion and Duplication	NC-6630
Avoid unnecessary listener notifications for the SV similar cases preferences menu	NC-6636
SV track mouse for closed track plus text label font size	NC-6637
Graceful start on Ignite connection failure	NC-6696

The defects that have been addressed in Bionano Access version 1.8 are listed in **Table 2**.

Table 2. Defects Addressed in v7.2

Summary	Ticket
Compile DT Scripts in UTF-8 encoding	NC-6149
Server reloads license without checking request key	NC-6392
When only using a VCF or only concerned with SeqVar, the list of the genes in panel will not appear on export	NC-6467
Prevent admin from adding attribute that contains only whitespace	NC-6512
SCORE(EVIDENCE) for SV events should be prevented with useful message	NC-6514
OGM BAF sample clustering exception when no probes at all	NC-6524
Gene annotations should be cleared when a sample is duplicated or reset	NC-6531
SeqVar processing types should not be selectable if the seqvar modality is not selected for the sample type	NC-6541
Attempting to navigate to the sample type associated with a sample from the homepage generates an error	NC-6550
Parent of origin probe coloring should show up in whole genome view tab initially at sample opening	NC-6552
Completion Exception - undefined manufacturer generated when adding a sample type with sample classes Array only, Methylation, NGS & Array and GxA-Cyto	NC-6555
Address secondary matching edge cases	NC-6557
Table columns such as parent of origin, inheritance comment fail to update if an event is manually edited or created	NC-6558
Memory leak in dpiScalingPreferences listeners	NC-6559
Unable to login due to error in sample type preference migration	NC-6561
Sample order in the multi-sample view window should follow the sample order on home page	NC-6572
Synchronize column width in main table and preferences table layout	NC-6584
Failure to query samples from the homepage based on sequence variants at a particular location	NC-6596
Error using «T:Whole Genome Results» tag with single-position Seqvar events	NC-6604
OGM label density systematic correction apply to all probes	NC-6611
KB Relevant Genes Dialog exception with blank notes	NC-6612
Admin decision tree columns re-calculation SeqVar index bounds check	NC-6615

Exception manually adding CNV events to SeqVar samples	NC-6616
Seqvar samples not processing with VCF proc type	NC-6631
Decision tree SeqVar event fix for ANY_SEQVAR_EVENT_KIND predicate	NC-6638
BAF Segment Value isn't being calculated for allelic events	NC-6639
cnScaledProbes and cnScaledAvg temp files should be deleted along with normal probe and snp files.	NC-6645
Nirvana JSON loading accept "Illumina Annotation Engine" annotator	NC-6653
Column preference UI ArrayIndexOutOfBoundsException	NC-6654
Add Logging for Ignite	NC-6661
exception when loading older duo samples	NC-6667
mouse wheel scrolls in the wrong direction	NC-6669
SeqVar JSON omitted genotype field interpreted as dot (.)	NC-6680
ISCN correction from alpha testers	NC-6681
Similar cases score for multiple matches from same sample	NC-6686
Decision tree throwing error 22	NC-6691
Change Ignite Configuration	NC-6695

Table 2. Defects Addressed in v7.2.1

Summary	Ticket
Incorrect ISCN generated for Normal Ploidy	NC-6768
Thread performance issue	NC-6756
S3 batch loading .crai suffix fix	NC-6792

Table 3. Defects Addressed in v7.2.2

Summary	Ticket

Latest ICS JSON file has formatting changes that impact import into VIA	NC-6844

Known Issues

We have observed in some cases the VIA client is not allocated sufficient memory (NC-6871). We plan to update the memory settings in the VIA Client installer in the next release. For help adjusting the memory configuration for your existing VIA Client please contact support.

There is a known issue with IDAT Methylation probes on ChrX and ChrY (NC-6839). These issues will be resolved in the next release.

Technical Assistance

For technical assistance, contact Bionano Technical Support.

You can retrieve documentation on Bionano products, SDS's, certificates of analysis, frequently asked questions, and other related documents from the Support website or by request through e-mail and telephone.

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