



Bionano Access™ Software User Guide

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Revision History

REVISION	NOTES
N	<p>Update for Bionano Access 1.7</p> <ol style="list-style-type: none"> 1) Removed references to Irys line of products. 2) Updated various legacy terms and associated procedures to conform with Saphyr terms. 3) Updated Jobs Menu 4) Included EnFocus™ Fragile X Analysis Options to menu. 5) Added NxClinical Bundle Download feature. 6) Added Chips Metrics Dashboard feature. 7) Added Perform Duo and Trio Analysis execution steps. 8) Enhanced explanation of the following features <ol style="list-style-type: none"> a. Circos Plot visualization b. Enhanced Whole Genome visualization. c. Curated Variant List 9) Added Visualization Reporting functionality. 10) Updated View option settings <p>Updated Settings section to reflect Access 1.7 updates</p>
O	<p>Updated for Bionano Access 1.8</p> <ol style="list-style-type: none"> 1) Revised section formatting for conciseness and clarity without changing meaning. 2) Changed listing of Terms and Definitions to table format. 3) Reworded redundant information or deleted altogether. 4) Created table for reference documents table. 5) Put “Job Options” and “Job Operations” text into tabular form.
P	<p>Updated for Bionano Access 1.8.1</p> <ol style="list-style-type: none"> 1) Added Guided Assembly section.
Q	<p>Updated to include information on Stratys™ and Stratys™ Compute</p>

Introduction

The Bionano Access™ Software enables users to manage experiments, view Saphyr® or Stratys™ run results in real time, perform a variety of bioinformatics analyses (**Table 1**), and visualize the resulting structural variants. This document guides users through capabilities available in the Access software installed on the Bionano Access Server. See **Table 2** and **Table 3** for more information.

Table 1. Bioinformatics operations including, but not limited to, the following:

Operation	Description
<i>In silico</i> digestion of data sequences	Perform <i>in silico</i> digestion by transforming a FASTA file into a Bionano CMAP file format. Calculates the estimated label density of FASTA file.
Merge molecule data sets	Merge two or more molecule jobs into a single molecule job.
Filter molecule jobs	Filter molecules jobs by length and total amount with random selection or enrich for longest molecules.
Generate <i>de novo</i> assemblies	Assemble single molecules into consensus maps for structural variant (SV) detection and hybrid scaffold applications.
Perform rare variant analysis	Detect structural variants at low allelic fraction.
Generate EnFocus™ FSHD analysis	Analyze regions relevant to facioscapulohumeral muscular dystrophy (FSHD).
Generate EnFocus™ Fragile X syndrome analysis	Analyze regions relevant to Fragile X syndrome
Align maps	Compare two different maps or molecules to maps by aligning them to each other.
Build hybrid scaffolds	Merge maps with sequence assemblies to produce contiguous hybrid scaffolds that represent the chromosome structure.
Annotate structural variants	Provide variant annotation for downstream filtering and prioritization of SVs based on Bionano internal human database or custom control database. Perform dual or trio comparisons.
Convert SMAP to VCF file	Convert insertion, deletion, duplication, inversion, and translocation breakpoint calls in an SMAP file to a dbVar-compliant VCF v4.2 format.

Compatibility

Bionano web Access™ 1.8 is compatible with the Google Chrome web browser. Bionano Access malfunction may be caused by using other, unvalidated browsers.

Table 2. Bionano Access Terms

Term	Definition
Cluster	Multiple compute servers can be organized into units, or clusters, which can be combined and scaled to user computing requirements.
Compute On Demand (COD)	A cloud-based computing platform, optimized for Bionano pipelines to perform bioinformatics analyses. The pipelines can work on custom clusters with custom configurations.
Job	Also known as Objects in version 1.6.1 or earlier - a distinctive set of output files that depend on a chosen operation. Examples include molecule data, map alignments, assemblies, hybrid scaffold results, and variant annotations.
Prep	Also known as a Molecule Set, this is the experiment setup consisting of the sample, reference, and enzyme in one flow cell.
Cohort	The number of subgroups that each scan is divided into for real-time analysis. One scan contains multiple cohorts, and run metrics are generated for each cohort.
Consensus Character MAP (CMAP)	The Bionano CMAP file is a raw data file which provides location information for label sites within a genome map or an <i>in silico</i> digestion of reference or sequence data. CMAPs may refer to a reference as well as a query (sample consensus map). The CMAP is a tab-delimited text-based file. See <i>OGM File Format Specification Sheet</i> (CG-00045) for more details.
Board Neutral Exchange (BNX)	The Bionano BNX file is a raw data view of molecule and label information, and quality scores per channel identified during a run. BNX v1.3 supports one or two label channels, or colors. See <i>OGM File Format Specification Sheet</i> (CG-00045) for more details.

Table 3. Bionano Access User Roles

Roles	Description
Administrator	Authorized to set security standards, assign user roles, create, and manage user accounts and access to projects, manage references, chips, and projects, permanently delete, or restore projects.
Project Lead	Administer user access to view, create, and manage projects, chips, and references. Can view and edit all projects in the Projects list.
User	Access, view and edit chips and projects, based on permissions.
Read Only	View only access. Cannot run chips.

Login

With the Bionano Access software installed on the server, a URL is needed for login, although a standalone user may use `localhost:3005` to open Access on the web. A user account is also required. Click **Forgot Password** to receive an email to reset a user password.

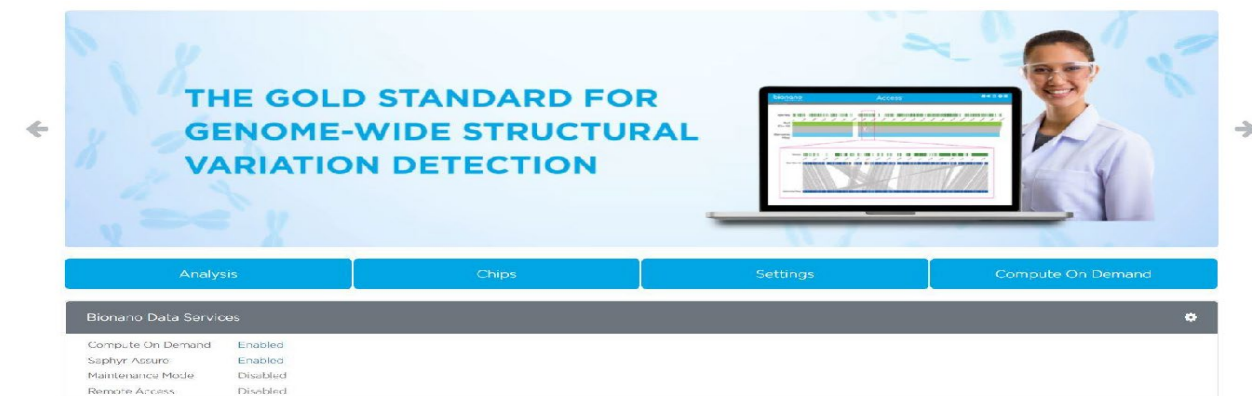


Figure 1. Access software Home Screen

The **Bionano Access Home** page presents certain modules for users, shown in **Table 4**. All modules are dependent on the types of user roles, system service settings, and installation instances. See details in *Bionano Access Installation Guide* (CG-30170).

Table 4. Bionano Access modules

Module	Description	Availability	Note
Analysis	Users create, view, edit, and manage projects. Users can also import and export data.	All user roles.	Name changed from "Projects" to "Analysis" in Access 1.7
Chips	Users can set up and manage Saphyr or Stratys experiments, view templates, and track chip run progress.	Only for systems integrated with an Instrument. (Not available for Read-Only)	Name changed from "Experiments" to "Chip" in Access 1.7
<i>in silico</i> Digestion	Users can perform <i>in silico</i> digestion, update settings, and view results.	All user roles.	Moved to Settings in Access 1.7
Settings	Users can create and manage user accounts, add references, configure software, upload BED files and Control Databases, and enable maintenance mode, enable Compute On Demand, and test the connection.	Only available to "Administrator" and "Project Lead" user roles.	
Compute On Demand	Users can redeem and transfer tokens and check token balances and transactions.	Only available for systems enabled with Compute On Demand.	

Help

Users can click the **Help** icon to access the **Help** page in Access. Software version information for Access and Solve, Compute On Demand version information for Bionano Tools and Solve, if enabled, video tutorials, security patch information and support contact information are all available here.

User Profile

Click the **User Profile** icon to change email address or password and to enable or disable email notifications after the initial log in.

1. In the **User Name** field, the username displays.
2. In the **Full Name** field, the full name displays.
3. In the **User Role** field, user role displays.
4. In the **Email Address** field, type the email address.

NOTE: By default, the **Send email notifications** checkbox is selected. This box can be unchecked to disable email notifications.

5. In the **Current Password** field, type the current password.
6. In the **New Password** field, type the new password.
7. In the **Confirm Password** field, type the new password again.
8. In the **Expiration date** field, the expiration date of the password is displayed.

Message Center

Click the **Message Center** icon to read and delete individual messages. Users can mark all messages as read and delete all read messages. When new messages about job status in Access are ready to be read, a red circle with a number will show at the **Message Center** icon. By default, the system will attempt to notify users via email from the message center. Users can disable email notifications on the **User Profile** page.

Alerts

Bionano Access sends alerts to notify system administrators of errors or system conditions that require attention. This is the foundation of proactive system diagnostics where the system can monitor itself and alert those responsible when action is needed. This feature works similarly to the messaging and message center, where notifications are sent to specific users regarding job status and other system events. Alerts are broadcast to all users in the Administrator role. The **Alerts** icon will appear in the header of Bionano Access next to the existing **Message Center** icon. The system will also post alerts that pertain to a chip run on the dashboard sent by the instrument.

Menu

Click the **Menu** icon to access the different modules, including **Home**, **Analysis**, **Chips**, **Settings**, **Compute On Demand**, **Profile**, **Help** and **Logout**. Use the **Job ID Search** function to access the specific job directly. The system will then point to the project and highlight the specific job.

Analysis

In the **Analysis** module, users can create new projects, manage existing projects, and edit, import, and download jobs. Users can also filter the projects by project name and export all the jobs in the project by clicking the **Select-All** option. Additionally, users can perform bioinformatics analysis such as alignment, molecules merge, hybrid scaffold, and other commands for individual jobs in the project. Jobs can be shared across all projects.

Create Project

Users in the Project Lead or the Administrator role can create Projects.

1. From the Bionano Access main menu, select **Analysis**. The **Projects** window appears.
2. Click **Create Project**. The **Project** dialog box appears.
3. At the **Project Name** field, type the name of the project.
4. [Optional] At the **Key** field, type a key (e.g., a project code) to associate with the project.
5. [Optional] At the **Description** field, type a brief description.
6. Click **Submit**. The project appears in the **Projects** list.

Sample Search

Users can find which projects contain certain samples.

1. From the **Bionano Access** main menu, select **Analysis**. The **Projects** window appears.
2. At the **Sample Name** field, type the name of the sample.
NOTE: The search for sample name is case sensitive.
3. Click **Search**. The project list will show the projects that contain the sample.

Export Project

Users in the Project Lead or the Administrator role can export a project containing multiple jobs, such as molecules, assembly, SV merge and variant annotation pipeline.

1. From the **Bionano Access** main menu, select **Analysis**. The **Projects** window appears.

2. Click the **Export** icon of the project. The **Export Project** window appears.
3. At the **Export File Name** field, type in the name.
4. [Optional] At the **Description** field, type the description.
5. Select the required jobs (i.e., assembly, molecules, map, SV merge and variant annotation pipeline).
6. Click **Submit**.
7. The exported file (.bng) will be automatically saved to the **Export** folder, which is in the **webServerShare** directory, as configured in the Access configuration file. For more details, see *Bionano Access Installation Guide* (CG-30170).

Import Project

Users can import an exported project containing multiple jobs, such as molecules, assembly, and variant annotation pipeline. Only users with the Project Lead or the Administrator role can import projects.

1. Copy the exported file (.bng) to the “import” folder, which is in the **webServerShare** directory, as configured in the **Access Configuration** file. See *Bionano Access Installation Guide* (CG- 30170).
2. Go to **Bionano Access** main menu, select **Analysis**. The **Projects** window appears.
3. Click the **Import** icon of the project. The **Import Project** window appears.
4. At the **Import File Name** field, select the file desired to import.
5. Click **Submit**.

Project Settings

Users in the Project Lead or the Administrator role can edit project settings to automatically generate various operations for each example.

1. Go to **Bionano Access** main menu, select **Analysis**. The **Projects** window appears.
2. Click the **Settings** icon of the project. The **Project Settings** window appears.
3. Under the **Auto Analysis (recurring)** panel, switch on auto-analysis for the desired operations.
4. Click **Submit**.

NOTE: The system will not generate a new job for a given sample if the molecule data does not meet the minimum coverage or quantity for the operation or if the operation has already been run on the sample.

Delete Project

Users with the Project Lead or the Administrator role can delete projects. Deleting a project is a two-step process: a Project Lead can delete the project from the list. However, only the administrator can delete the project permanently from Access, as the second step. For more details, see the “Deleted Projects” section in this guide.

1. From the **Bionano Access** main menu, select **Analysis**. The **Projects** window appears.
2. Click the **Delete** icon of the project to delete. The **Confirm** dialog box appears.
3. Click **Yes**. The project is deleted from the list.

NOTE: There are several rules that would prevent a user from deleting a project:

- If the project contains experiments, a user cannot delete it because experiments cannot be deleted.
- If the jobs in the project are not removed, a user cannot delete the project.

Thus, before deleting a project, make sure it does not contain experiments, and that the jobs are deleted first.

Edit Project

Users in the Project Lead or the Administrator role can edit project information.

1. From the **Access** main menu, select **Analysis**. The **Projects** window appears.
2. Select one project from the project list.
3. Click **Edit**. The **Project** dialog box appears.
4. At the **Project Name** field, type the name of the project.
5. [Optional] At the **Key** field, type a key (e.g., a project code) to associate with the project.
6. [Optional] At the **Description** field, type a brief description.
7. Click **Submit**.

Give Users Access to View Projects

Project Lead or Administrator roles can give User and Read-only roles to access and view projects.

1. From the **Bionano Access** main menu, select **Analysis**. The **Projects** window appears.
2. Click the **Access** icon of the project. The **Project Access Control** screen appears.
3. In the **All Users** pane, select the users who will be given access, and then click **Grant Access**. Users in the Project Lead or Administrator role are not listed. The user's names appear in the Users with **Access** pane.

Deleted Projects

Users must have administrator privileges to perform this task. Users can restore a project that was deleted by a Project Lead.

1. From the **Bionano Access** main menu, select **Analysis**. The **Projects** window appears.
2. Click the **Deleted Projects** icon at the top right-corner of the screen. The **Deleted Projects** screen appears.

3. With the project to restore highlighted, click the **Restore** icon. The software restores the project and adds it back to the project list.
4. With the project to delete highlighted, click the **Delete** icon. The project would be permanently deleted.

Jobs

When users select a project, the **Project Browser** page opens (**Table 5**). The page contains menu options, the list of job(s), the selected-job details, and the options and operations for the selected job, seen in **Table 6** through **Table 9**.

Table 5. Project Browser Page menu

Menu	Description
Import	To import output files from clusters or Bionano files from another system.
Remove	To remove a job from the project.
Copy	To copy data from the job and paste it in another project.
Edit	To edit job name, sample, reference, tags, and description.
Jobs	To view a list of jobs and their state (Active, Working, Complete, Cancelled, Failed) for a project.
Samples	To view a list of samples associated with the jobs in this project.
Reset	To remove all filters currently applied in the job list.

Table 6. All the jobs listed belong to the selected project.


Info	Description
Sample	The sample name.
Type	The job type (e.g., <i>de novo</i> assembly, molecules, map, scaffold, FASTA file).
Name	The user-defined name for the job.
Curation	The curation status for the job
Tag	The Tag feature allows users to create and edit keywords that are tagged to this job. Users can filter jobs with the same keywords to easily manage jobs in a project. To create or edit tags for the job, select the job, and then click Edit . At the Tags field, type the keyword, and then press Tab . Repeat to add more keywords. Click the Delete icon to delete the keyword.
Created	The date when the job was created.
	Click this icon to sort and filter the information in the columns.

Table 7. Sample Details

Info	Description
Name	The sample name that is assigned to the job.
Sample UID	System generated global unique identifier.
Created	The date when the job was created.

Table 8. Job Details

Info	Description
Name	The job name should be unique to a project. The same job can have different names in different projects.
Reference	[Optional] The reference map.
Description	[Optional] The description of the job.
Created	The date the job was created.
Operation	The type of job performed.
Status	The status of the job.
User	The user who created the job.
Job ID	The identifier of the job. Job Error Reporting icon shows there is returned error message.
Command	Detailed information about the job and the Bionano Tools command issued to compute servers.
VIA Status	The status of the VIA upload.
VIA Uploaded	The date the job was uploaded to VIA.

Job Options

The **Options** pane contains a list of options that can be visualized or downloaded with the selected job. The options are dependent on the type and status of the job. The links in the **Options** pane may be disabled and greyed out if a previous operation is still in progress.

Table 9. Options Pane

Type of Options	Capability
Molecules	<ul style="list-style-type: none"> • View Molecule Quality Report • View Molecules Size Distribution Chart • Download Molecules • View Dashboard

Type of Options	Capability
Alignment	<ul style="list-style-type: none"> • View Results • Download Alignment
<i>De novo</i> Assembly Annotated <i>de novo</i> Assembly	<ul style="list-style-type: none"> • View Results • View Molecule Alignment • View Informatics Summary • Download Assembly • Download ogm.bam. • Download ogm.bai. • Download ogm.vcf. • Upload to VIA
Rare Variant Analysis Annotated Rare Variant Analysis	<ul style="list-style-type: none"> • View Results • View Informatics Summary • Download Rare Variant Analysis • Download ogm.bam. • Download ogm.bai. • Download ogm.vcf. • Upload to VIA
Guided Assembly Annotated Guided Assembly	<ul style="list-style-type: none"> • View Results • View Molecule Alignment • View Informatics Summary • Download Assembly • Download ogm.bam. • Download ogm.bai. • Download ogm.vcf.
Variant Annotation Pipeline	<ul style="list-style-type: none"> • View Results • Download Variant Annotation
EnFocus™ FSHD Analysis Options	<ul style="list-style-type: none"> • View Results • View Maps • View Informatics Summary • Go to Molecules Job • Download EnFocus™ FSHD. • Download EnFocus™ FSHD JSON • Download EnFocus™ FSHD Report
EnFocus™ Fragile X Analysis	<ul style="list-style-type: none"> • View Results • View Maps • View Informatics Summary • Go to Molecules Job • Download EnFocus™ Fragile X • Download EnFocus™ Fragile X JSON • Download EnFocus™ Fragile X Report
BED	<ul style="list-style-type: none"> • Download Feature

Type of Options	Capability
FASTA	<ul style="list-style-type: none"> Download FASTA
Map	<ul style="list-style-type: none"> Download Map
Scaffold	<ul style="list-style-type: none"> View Results View Results with Conflicts View Informatics Summary Export NCBI Package Download Hybrid Scaffold

Job Operations

The **Operations** pane (**Table 10**) contains a list of operations that can be performed within the selected job. The **Compute On Demand** pane contains the same list of operations but uses tokens. The operations are dependent on the type and status of the job. The links in the **Operations** pane may be disabled and greyed out if a previous operation is still in progress.

NOTE: The only operations available in the **Operations** pane for Access servers connected to a Stratys Compute are **Generate Molecule Quality Report** and **Generate Guided Assembly**.

Table 10. Job Operations

Type of Operation	Capability
<i>De novo</i> Assembly Annotated <i>de novo</i> Assembly	<ul style="list-style-type: none"> Generate Variant Annotation – Single Generate Variant Annotation – Dual Generate Variant Annotation – Trio Align Maps Generate Hybrid Scaffold Generate 2-Enzyme Hybrid Scaffold
Rare Variant Analysis Annotated Rare Variant Analysis	<ul style="list-style-type: none"> Generate Variant Annotation – Single Generate Variant Annotation – Dual Generate Variant Annotation - Trio
Guided Assembly Annotated Guided Assembly	<ul style="list-style-type: none"> Generate Variant Annotation – Single Generate Variant Annotation – Dual Generate Variant Annotation - Trio
Map	<ul style="list-style-type: none"> Align Maps Generate Hybrid Scaffold Generate 2-Enzyme Hybrid Scaffold
Molecules	<ul style="list-style-type: none"> Generate Molecule Quality Report Generate <i>de novo</i> Assembly Generate Rare Variant Analysis Generate Guided Assembly Generate EnFocus™ FSHD Analysis Generate EnFocus™ Fragile X Analysis

- Filter Molecule Job
- Merge Molecule Jobs
- Align Maps

Import Job

Users can import data from output files that are based on user-defined commands and generated from the cluster, or from another system that is in an acceptable Bionano file format.

IMPORT MOLECULES

Users must have Bionano molecules data in a *.bnx or *.bnx.gz file to import to Bionano Access.

1. From the **Bionano Access** main menu, select **Analysis**. The **Projects** window appears.
2. Select the project to view from the list.
3. Click **Import**. The **Import** dialog box appears.
4. At the **Job Type** field, select **Molecules**.
5. At the **Molecules Name** field, type a name for this job.
6. At the **Sample** field, click the drop-down list to select the sample associated with a file or **New** to add a new sample, when the **Sample** dialog box appears. At the **Sample Name** field, type a name for this sample.
7. Click **Import**.
8. The **File Upload Status** is displayed. The progress bar indicates the status of the data uploading to the server.

IMPORTANT: Do not close the page until the upload is finished as this may interrupt the process.

9. The **Upload Completed** shows up when the upload is finished.
10. Click **Ok**. The screen switches back to the **Project Browser** page. Access sends an email notification when the data is imported.

IMPORT CONSENSUS MAP

Users must have consensus map data in a *.cmap file to import to Bionano Access.

1. From the **Bionano Access** main menu, select **Analysis**. The **Projects** window appears.
2. Select the project to view from the list.
3. Click **Import**. The **Import** dialog box appears.
4. At the **Job Type** field, select **Consensus Map**.
5. At the **Consensus Map Name** field, type a name for this job.
6. At the **Sample** field, click the drop-down list to select the sample associated with file.

7. [Optional] At the **Reference** field, click the drop-down list to select the reference that was used.
8. [Optional] At the **Tags** field, type the keywords to associate with the file.
9. [Optional] At the **Description** field, type a brief description.
10. At the **Consensus Map File** field, click **Browse** to choose the cmap file (.cmap).
11. Click **Import**.
12. The **File Upload Status** appears. The progress bar indicates the status of the data uploading to the server.

IMPORTANT: Do not close the page until the upload is finished. If users close the page while the data is uploading, the uploading process may be interrupted.

13. **Upload Completed** appears when the upload is finished.
14. Click **Ok**. The screen switches back to the **Project Browser** page. Bionano Access sends an email notification when the data is imported.

IMPORT AN ALIGNMENT JOB

Users must have all three of these files to import an alignment to Bionano Access: **.xmap, _r.cmap, and _q.cmap**

1. From the **Bionano Access** main menu, select **Analysis**. The **Projects** window appears.
2. Select the project to view from the list.
3. Click **Import**. The **Import** dialog box appears.
4. At the **Job Type** field, **Alignment** is auto selected.
5. At the **Alignment Name** field, type the name of the alignment.
6. At the **Sample** field, select the sample from the drop-down list.
7. [Optional] At the **Reference** field, select a reference from the drop-down list.
8. [Optional] At the **Tags** field, type the keywords to associate with the alignment job.
9. [Optional] At the **Description** field, type a brief description.
10. At the **Alignment Type** field, choose one of the following options for Bionano to display the files in:
 - **Anchor to Genome Maps**— Display a consensus map as contigs. Select this option if the alignment is between two consensus maps.
 - **Anchor to Molecules**—Display a consensus map as molecules. Select this option if the alignment is between molecule file to a reference or assembly consensus map.
11. At the **Reference Map** field, click **Browse** to choose the reference cmap file (r.cmap).
12. At the **Alignment Map** field, click **Browse** to choose the alignment xmap file (.xmap).
13. At the **Query Map** field, click **Browse** to choose the query cmap file (q.cmap)

14. Click **Import**.

15. The **File Upload Status** shows up.

The progress bar indicates the status of the data uploading to the server.

IMPORTANT: Do not close the page until the upload is finished. If users close the page while the data is uploading, the uploading process may be interrupted.

16. The **Upload Completed** shows up when the upload is finished.

17. Click **Ok**.

The screen switches back to the **Project Browser** page. Bionano Access sends an email notification when the data is imported.

IMPORT FASTA

Users must have fasta data in a ***.fasta**, ***.fa** or ***.fna** file to import to Bionano Access.

1. From the **Bionano Access** main menu, select **Analysis**. The **Projects** window appears.

2. Select the project to view from the list.

3. Click **Import**. The **Import** dialog box appears.

4. At the **Job Type** field, select **FASTA**.

5. At the **FASTA Name** field, type a name for this job.

6. [Optional] At the **Tags** field, type the keywords to associate with the file.

7. [Optional] At the **Description** field, type a brief description.

8. At the **FASTA** field, click **Browse** to choose the fasta file (fasta,.fa or .fna).

9. Click **Import**.

10. The **File Upload Status** shows up.

The progress bar indicates the status of the data uploading to the server.

IMPORTANT: Do not close the page until the upload is finished. If users close the page while the data is uploading, the uploading process may be interrupted.

11. The **Upload Completed** shows up when the upload is finished.

12. Click **Ok**.

The screen switches back to the **Project Browser** page. Access sends an email notification when the data are imported.

IMPORT SCAFFOLD

Users must have the hybrid scaffold data in a ***.zip** file to import to Bionano Access.

IMPORTANT: Hybrid Scaffold result generated by Bionano Solve through the command line **MUST** have output directory set in ***/output**.

1. From the **Bionano Access** main menu, select **Analysis**. The **Projects** window appears.
2. Select the project to view from the list.
3. Click **Import**. The **Import** dialog box appears.
4. At the **Job Type** field, select **Scaffold**.
5. At the **Hybrid Scaffold Name** field, type a name for the scaffold.
6. At the **Sample** field, select the sample from the drop-down list.
7. [Optional] At the **Tags** field, type the keywords to associate with the alignment job.
8. [Optional] At the **Description** field, type a brief description.
9. At the **Hybrid Scaffold File** field, click **Browse** to choose the scaffold file (.zip).
10. Click **Import**.
11. The **File Upload Status** shows up.

The progress bar indicates the status of the data uploading to the server.

IMPORTANT: Do not close the page until the upload is finished. If users close the page while the data is uploading, the uploading process may be interrupted.

12. The **Upload Completed** shows up when the upload is finished.
13. Click **Ok**.

The screen switches back to the **Project Browser** page. Access sends an email notification when the data are imported.

IMPORT DE NOVO ASSEMBLY

Users must have *de novo* Assembly or Annotated *de novo* Assembly data generated by Bionano Solve in ***.gz** or ***.zip** file to import to Bionano Access.

IMPORTANT: *de novo* Assembly results generated by Bionano Solve through the command line **MUST** have output directory set in ***/output**.

1. From the **Bionano Access** main menu, select **Analysis**. The **Projects** window appears.

2. Select the project to view from the list.
3. Click **Import**. The **Import** dialog box appears.
4. At the **Job Type** field, select **de novo Assembly**.
5. At the **Assembly Name** field, type a name for this job.
6. At the **Sample** field, click the drop-down list to select the sample associated with file.
7. [Optional] At the **Reference** field, click the drop-down list to select the reference used.
8. [Optional] At the **Tags** field, type the keywords to associate with the file.
9. [Optional] At the **Description** field, type a brief description.
10. At the **de novo assembly File** field, click **Browse** to choose the assembly file (.zip).
11. Click **Import**.
12. The File Upload Status shows up.

The progress bar indicates the status of the data uploading to the server.

IMPORTANT: Do not close the page until the upload is finished. If users close the page while the data is uploading, the uploading process may be interrupted.

13. The **Upload Completed** shows up when the upload is finished.
14. Click **Ok**.

The screen switches back to the **Project Browser** page. Bionano Access sends an email notification when the data are imported.

IMPORT BED

Users must have BED data in *.bed file to import to Bionano Access.

1. From the **Bionano Access** main menu, select **Analysis**. The **Projects** window appears.
2. Select the project to view from the list.
3. Click **Import**. The **Import** dialog box appears.
4. At the **Job Type** field, select **BED**.
5. At the **Name** field, type a name for this job.
6. [Optional] At the **Tags** field, type the keywords to associate with the file.
7. [Optional] At the **Description** field, type a brief description.

8. At the **BED File** field, click **Browse** to choose the bed file (.bed).
9. Click **Import**.
10. The **File Upload Status** appears.

The progress bar indicates the status of the data uploading to the server.

IMPORTANT: Do not close the page until the upload is finished. If users close the page while the data is uploading, the uploading process may be interrupted.

11. **Upload Completed** appears when the upload is finished.
12. Click **Ok**.

The screen switches back to the **Project Browser** page. Access sends an email notification when the data are imported.

IMPORT CYTOBAND

Users must have cytoband data in *.bed file to import to Bionano Access.

1. From the **Bionano Access** main menu, select **Analysis**. The **Projects** window appears.
2. Select the project to view from the list.
3. Click **Import**. The **Import** dialog box appears.
4. At the **Job Type** field, select **Cytoband**.
5. At the **Cytoband Name** field, type a name for this job.
6. [Optional] At the **Tags** field, type the keywords to associate with the file.
7. [Optional] At the **Description** field, type a brief description.
8. At the **Cytoband File** field, click **Browse** to choose the cytoband file (.bed).
9. Click **Import**.
10. The **File Upload Status** shows up.

The progress bar indicates the status of the data uploading to the server.

IMPORTANT: Do not close the page until the upload is finished. If users close the page while the data is uploading, the uploading process may be interrupted.

11. **Upload Completed** appears when the upload is finished.
12. Click **Ok**. The screen switches back to the **Project Browser** page and an email notification is sent when the data are imported.

IMPORT RARE VARIANT ANALYSIS

Rare Variant Analysis or Annotated Rare Variant Analysis data must be generated by Bionano Solve in a *.zip file to import to Bionano Access.

IMPORTANT: Rare Variant Analysis results generated by Bionano Solve through command line must be within the output directory set in */output.

1. From the **Bionano Access** main menu, select **Analysis**. The **Projects** window appears.
2. Select the project to view from the list.
3. Click **Import**. The **Import** dialog box appears.
4. At the **Job Type** field, select **Rare Variant Analysis**.
5. At the **Rare Variant Analysis Name** field, type a name for this job.
6. At the **Sample** field, click the drop-down list to select the sample associated with file.
7. [Optional] At the **Reference** field, click the drop-down list to select the reference that was used.
8. [Optional] At the **Tags** field, type the keywords to associate with the file.
9. [Optional] At the **Description** field, type a brief description.
10. At the **Rare Variant Analysis File** field, click **Browse** to choose the analysis file(.zip).
11. Click **Import**.
12. **File Upload Status** appears.

The progress bar indicates the status of the data uploading to the server.

IMPORTANT: Do not close the page until the upload is finished. If users close the page while the data is uploading, the uploading process may be interrupted.

13. The **Upload Completed** shows up when the upload is finished.
14. Click **Ok**.

The screen switches back to the **Project Browser** page. Bionano Access sends an email notification when the data are imported.

IMPORT VARIANT ANNOTATION

Users must have variant annotation analysis data generated by Bionano Solve in *.zip file to import to Access.

IMPORTANT: Variant Annotation Pipeline Analysis result generated by Bionano Solve through the command line **MUST** have output directory set in */output.

1. From the **Bionano Access** main menu, select **Analysis**. The **Projects** window appears.
2. Select the project to view from the list.
3. Click **Import**. The **Import** dialog box appears.
4. At the **Job Type** field, select **Variant Annotation**.
5. At the **VAP Analysis Name** field, type a name for this job.
6. At the **Sample** field, click the drop-down list to select the sample associated with file.
7. [Optional] At the **Reference** field, click the drop-down list to select the reference that was used.
8. [Optional] At the **Tags** field, type the keywords to associate with the file.
9. [Optional] At the **Description** field, type a brief description.
10. At the **Variant Annotation Pipeline** field, click **Browse** to choose the VAP analysis file (.zip).
11. Click **Import**.
12. The **File Upload Status** shows up.

The progress bar indicates the status of the data uploading to the server.

IMPORTANT: Do not close the page until the upload is finished. If users close the page while the data is uploading, the uploading process may be interrupted.

13. The **Upload Completed** shows up when the upload is finished.
14. Click **Ok**.

The screen switches back to the **Project Browser** page. Bionano Access sends an email notification when the data are imported.

IMPORT FSHD ANALYSIS

FSHD analysis data must be generated by Bionano Solve in *.zip file to import to Bionano Access.

IMPORTANT: FSHD Analysis Users must have FSHD result generated by Bionano Solve through the command line MUST have output directory set in */output.

From the **Bionano Access** main menu, select **Analysis**. The **Projects** window appears.

1. Select the project to view from the list.
2. Click **Import**. The **Import** dialog box appears.
3. At the **Job Type** field, select **FSHD Analysis**.

4. At the **FSHD Analysis Name** field, type a name for this job.
5. At the **Sample** field, click the drop-down list to select the sample associated with file.
6. [Optional] At the **Reference** field, click the drop-down list to select the reference that was used.
7. [Optional] At the **Tags** field, type the keywords to associate with the file.
8. [Optional] At the **Description** field, type a brief description.
9. At the **FSHD Analysis File** field, click **Browse** to choose the FSHD analysis result file (.zip).
10. Click **Import**.
11. **File Upload Status** appears.

The progress bar indicates the status of the data uploading to the server.

IMPORTANT: Do not close the page until the upload is finished. If users close the page while the data is uploading, the uploading process may be interrupted.

12. **Upload Completed** appears when the upload is finished.
13. Click **Ok**.

The screen switches back to the **Project Browser** page. Bionano Access sends an email notification when the data are imported.

IMPORT FRAGILE X ANALYSIS

Users must have Fragile X analysis data generated by Bionano Solve in a ***.zip** file to import to Bionano Access.

IMPORTANT: Fragile X Analysis result generated by Bionano Solve through the command line **MUST** have output directory set in ***/output**.

1. From the **Bionano Access** main menu, select **Analysis**. The **Projects** window appears.
2. Select the project to view from the list.
3. Click **Import**. The **Import** dialog box appears.
4. At the **Job Type** field, select **Fragile X Analysis**.
5. At the **Fragile X Analysis Name** field, type a name for this job.
6. At the **Sample** field, click the drop-down list to select the sample associated with file.
7. [Optional] At the **Reference** field, click the drop-down list to select the reference that was used.
8. [Optional] At the **Tags** field, type the keywords to associate with the file.
9. [Optional] At the **Description** field, type a brief description.

10. At the **Fragile X Analysis File** field, click **Browse** to choose the Fragile X analysis result file (.zip).
11. Click **Import**.
12. **File Upload Status** appears.

The progress bar indicates the status of the data uploading to the server.

IMPORTANT: Do not close the page until the upload is finished. If users close the page while the data is uploading, the uploading process may be interrupted.

13. **Upload Completed** appears when the upload is finished.
14. Click **Ok**.

The screen switches back to the **Project Browser** page. Bionano Access sends an email notification when the data are imported.

Remove Job

Users can remove jobs from a project, but it will not affect the same data that are shared/copied with other projects. To remove a job that exists in several projects, users would need to remove the job from each project individually.

1. From the **Bionano Access** main menu, select **Analysis**. The **Projects** window appears.
2. Select the project to view from the list.
3. In the Jobs list, select the job to remove.
4. Click **Remove**. The **Remove Job from Project** dialog box appears.
5. Click **Yes**.
6. When jobs are deleted in the **Project Browser** they go into the trash. To permanently remove a job or restore a previously deleted job in a project, select the project and click on the **Deleted Jobs** icon at the top-right corner of the **Access** webpage to manage deleted jobs. There is a button to allow Administrators only to delete all the jobs in the trash, so they are not deleted one at a time.

Copy Job

Users can copy jobs from one project and add them to another project.

1. From the **Bionano Access** main menu, select **Analysis**. The **Projects** window appears.
2. Select the project to view from the list.
3. In the **Jobs** list, select a job (i.e., assembly, molecules, map) to share.
4. Click **Copy**. The **Copy Job to Project** dialog box appears.
5. At the **Select the target project** field, click the drop-down list to another project.

6. Click **Submit**.

Users can view the data from the project that was selected to share.

Edit Job

Users can edit a job's name, sample reference, tags, and description information.

1. From the **Bionano Access** main menu, select **Analysis**. The **Projects** window appears.
2. Select the project to view from the list.
3. Click **Edit**. The **Edit Job** dialog box appears.
4. At the **Name** field, click the drop-down list to select the sample associated with file.
5. [Optional] At the **Reference** field, click the drop-down list to select the reference that was used. (Can only be changed for Molecule Jobs)
6. [Optional] At the **Tags** field, type the keywords to associate with the file.
7. [Optional] At the **Description** field, type a brief description.
8. Click **Submit**.

Users can view all recently (default: last 30 days) submitted jobs' status, cancel running jobs, and sort and filter the columns of a job listing (**Table 11**). A menu for Samples is seen in **Table 12**.

Table 11. Jobs

Info	Description
Project	The project name of the job.
Job ID	The unique job number auto generated by the Access server when a job is created.
Operation	The operation of the job, such as Alignment, Import Molecule, DLE-1 <i>de novo</i> Assembly, or Variant annotation.
Name	The created job name.
User	The name of the user who submitted the job.
Created	The date when the job was created/submitted.

State	The current state of the job. Active: the job is running. Working: copying input files; job has not started Complete: job completed successfully Cancelled: job cancelled Failed: job failed
Status	The status of the job. System reads status.xml file in the compute server for each job.
Server	The IP address or hostname of the assembly server.
Actions	To view job details or cancel the job: Bionano Access automatically cancels jobs that are inactive for more than 10 days. Jobs that are inactive for longer than 10 days are assumed to have been interrupted or orphaned.

Samples

Table 12. Menu

Menu	Description
Add	To add a new sample.
Remove	To remove a sample. NOTE: to remove a sample, one must remove all the associated jobs.
Copy	To copy sample from the job and paste it in another project.
Edit	To edit sample name, description, and comment.

Users can view all information, sort, and filter the columns of the sample list (**Table 13**).

Table 13. Sort and filter

Info	Description
Sample	The sample name that is assigned to jobs.
ID	System generated global unique identifier.
Description	The description of the sample.

Type	The type of sample that was created.
Indication	The indication of the sample created
Comments	The comment of the sample created.
Created	The date the sample was created.

Reset

After applying filters to the job list, users can clean all the filters applied.

Deleted Job

Users must have administrator privileges to perform this task.

1. From the **Bionano Access** main menu, select **Projects**. The **Projects** window appears.
2. Select the project that contains the job to delete.
3. Click the **Deleted Jobs** icon at the top right-corner of the screen. The **Deleted Jobs** screen appears.
4. Click **Delete all jobs** if it is desired to delete all the jobs permanently in all projects.
5. Select the job from the list to restore, click the **Restore** icon.
6. The software restores the project and adds it back to the job list.
7. Select the job from the list, and then click the **Delete** icon.

The software permanently deletes the job from the project. If the job is shared with other projects, the job still exists in those projects. To permanently delete the same job that is in other projects, repeat Steps 1 through 4 for the other projects.

Download Job

1. From the **Bionano Access** main menu, select **Projects**. The **Projects** window appears.
2. Select the project to view from the list.
3. In the **Jobs** list, select the job to download.
4. In the **Options** pane, click the download [job type] option. The software downloads the data onto the workstation running the browser.

Chips

In the **Chips** module, users can add new projects, manage projects, track run performance, and view real-time metrics. Additionally, users can view and edit templates. In Access 1.8, each chip can now be associated to more than one project. Before inserting the Saphyr Chip or Stratys Chip into the instrument, create a project or projects to associate with the chip. The run results are posted for the project or projects that are associated with the chip.

A project created on Bionano Access is in *pending* status until it is associated with a Saphyr Chip or Stratys Chip. Users can modify or delete pending projects. Once a project is associated with a scan, users cannot modify or delete the project.

Add Chip

Only users in the Administrator or Project Lead role can create chip runs.

From the Bionano Access main menu, select **Chips** and a list appears (**Table 14**).

Table 14. Chips list

Info	Description
Start	Time that the chip run was started.
Finish	Time that the chip run was completed.
Name	The chip name.
Instrument	The name of the instrument.
Chip Run	The chip run ID generated by the system.
Serial Number	The serial number of the chip.
Operator	The user's name who set up the chip run.
Expiration	The chip expiration date
Action	To view the dashboard associated with the chip, click View Dashboard . Click Delete to delete a chip.

Click the **Expand** icon for more information about each experiment as suggested below (**Table 15**):

Table 15. Expanded information.

Info	Description
Location	The location of the flowcell on each chip. FC – 1, FC – 2 or FC – 3. (Applicable only to Saphyr Chips)
Project	The name of the project.
Sample	The sample origin. Human samples or others.
Label	The label colors. Green 01 or Red 01.
Enzyme	The enzyme name (e.g., DLE-1)
Reference	The reference name.
Auto Assemble	False or true.
Auto RVA (Rare Variant Analysis)	False or true.
Auto FSHD	False or true.
Auto Fragile-X	False or true.
Isolation kit	Name of the DNA Isolation kit used.
Labeling kit	Name of the DNA Labeling kit used.

1. Select **Add Chip**. A dialog box appears.
2. At the **Chip Name** field, type the name of the chip that is associated with the experiment.
3. At **Chip Description** (optional) field, type a description.
4. At the **Chip Part Number** field, select the correct Chip Part Number.
5. At the **Chip Serial Number** (optional) field, type the serial number of the chip.
6. At the **Chip Template** (optional) field, select a chip template (e.g., Fragile X Analysis, FSHD Analysis, Mosaic Analysis).

7. Under **Sample Slot**, at the **Project** field, select a Project or create a new Project.
8. A window will open if **New Project** is selected.
9. Under **Project Name**, type the name of the project. Type a **Key** (optional) and **Description** (optional). Click **Submit**.
10. Under **Sample**, select a Sample or create a New Sample.
11. A window will open if **New Sample** is selected.
12. Under **Sample Name**, type the name of the Sample. Type **Sample Type** (optional), **Indication** (optional), **Description** (optional), or **Comment** (optional). Click **Submit**.
13. Click **Next**.
14. At the **Throughput Target (Gbp)** field under the **Molecule Set**, type the target throughput in Gbp. This field only applies to instruments running Stratys™ Chips or Saphyr® ICS 4.8 or greater. Please refer to *Data Collection Guidelines* (CG-30173) for more detail.
15. At the **Molecule Job Name** field (optional), type the name of molecule job. This field can also be left blank for default naming.
16. At the **Project** field, click the drop-down list to select a project or click **New** to create a new project.
 - When creating a new project, type a name under **Project Name**. **Key** and **Description** are optional. Click **Submit**.
17. At the **Sample** field, click the drop-down list to select a sample or click **New** to create a new sample.
 - When creating a new sample, type the **Sample Name**, **Sample Type** (optional), **Indication** (optional), **Description** (optional), and **Comment** (optional). Click **Submit**.
18. At the **Recognition Enzyme** field, click the drop-down list to select an enzyme or click **New** to add a new enzyme. When creating a new Enzyme, type the **Enzyme Name** and **Sequence** of the recognition site.
19. At the **Reference** field, click the drop-down list to select a genome reference for the experiment.
 - If hg19 (DLE1, BSSSI or BSPQI) or hg38 (BSSSI or BSPQI) is chosen as reference, **Auto de novo Assembly**, and **Auto Rare Variant Analysis** options will be available. Check the box to enable automatic pipeline submission.
 - If hg38 (DLE1) or T2T (DLE-1) is chosen as reference, **Auto de novo Assembly**, **Auto Rare Variant Analysis**, **Auto Guided Assembly**, **Auto EnFocus™ FSHD** and **Auto EnFocus™ Fragile-X** options will be available. Check the boxes to enable automatic pipeline submission.
20. [Optional] At the **Isolation Kit Lot Number** field, type the lot number of the isolation kit used.
21. [Optional] At the **Labeling Kit Lot Number** field, type the lot number of the labeling kit used.
22. Click **Add to Flowcell 1**.

To edit the information for Flowcell 1, click **Remove** under **Flowcell 1** in the **Chip** pane (on the right), and then make changes.
23. Repeat steps 2 through 22 for Flowcells 2 and 3, if applicable.
24. Click **Add to Flowcell 2 (or 3)**.

To edit information for either Flowcell 2 or 3, click **Remove** under the corresponding Flowcell in the **Chip** pane, and then make changes.

25. Click **Save Chip**.

Create an Experiment Template

Users can create an experiment template if the same enzyme and reference are used every time. Only users in the Administrator or Project Lead role can create experiment templates.

1. From the **Bionano Access** main menu, select **Chips**. The **Chips** list appears.
2. Select **Templates**. By default, there are three templates each for Fragile X Analysis and FSHD Analysis using Chip Part Numbers 20319, 20366 and 20367. There is one template for Mosaic Analysis using Chip P/N 20366.
3. Repeat Steps 2-24 in the “Add a Chip” section.
4. [Optional] Click **Save as Template**. The **Experiment Template** window appears.
5. At the **Template Name** field, type in the name of template.
6. At the **Label for sample slot** field, type in the name of the sample slot for each of the different samples. The user can select the saved experiment template.
7. Click **Ok**.

Monitor Run Progress

After the instrument scans the chip for 15 to 20 minutes and raw images are detected, users can monitor the progress of the run and view real-time metrics from the Bionano Access web site.

1. From the **Bionano Access** main menu, click **Chips**.
2. The latest chip run (**Table 16**) appears at the top of the list.

Table 16. Chip run information

Info	Description
Start	Date and time of the chip run start.
Finish	Date and time of the chip run finish.
Name	The name of the chip.
Instrument	The instrument name.

Info	Description
Chip Run	The chip run ID generated by the system.
Serial Number	The serial number of the chip.
Operator	The user's name who set up the chip run.
Expiration	The chip expiration date.
Action	To view the dashboard associated with the chip, click 'View Dashboard.' Click 'Delete' to delete a chip.

3. [Optional] Click the **Filter** icon to filter data in the columns.
4. Select the experiment or chip run to monitor, and then click **View Dashboard**. See *Bionano Access: Dashboard Guidelines* (CG-30304) for more details. The **Dashboard** screen appears showing:
 - Run information (see **Table 17**)
 - Analysis graphs (see **Table 18**)
 - Run metrics table (see **Table 19**)

Table 17. Run Information

Info	Description
Chip	The Saphyr Chip or Stratys Chip bar code.
Run ID	The system-generated run identifier.
Experiment	The experiment name.
Instrument	The instrument serial number.
Min Length	The minimum length of molecules that are used for analysis. The setting for this parameter is system generated.
Min Labels	The minimum labels per molecule that are used for analysis. The setting for this parameter is system generated.
Start Time	The run start time.

Info	Description
End Time	The run end time.

5. To view the raw data, click **Show Data**. Users can export the raw data by clicking **Export to Excel**.

Table 18. Analysis Graphs

Map	Description
DNA per Scan (Gbp)	This graph shows the amount of DNA per flowcell that is detected per scan.
Map Rate (%)	This graph shows the percentage of molecules that map to the reference genome. The map rate cannot be calculated unless there is a minimum number (1000) of molecules and labels. If the minimum threshold is not acquired, the map rate is defaulted to zero. If no reference genome is provided, the map rate is defaulted to zero.

Table 19. Run Metrics

Metric	Description
Plot	The color used in the plot and checkbox for each flowcell.
Flowcell	The scanned flowcell.
Prep	The names of the sample, channel, enzyme, type, and reference
Avg N50 (>=150kbp) (Mbp)	The molecule length N50 for all molecules that are ≥ 150 kbp in length.
Avg N50 (>=20kbp) (Mbp)	The molecule length N50 for all molecules that are ≥ 20 kbp in length. Molecules that are less than 20 kbp are considered noise by the image detection algorithm.
Avg Label Density (per 100 kbp)	The number of labels that are detected by the image detection algorithm per 100 kbp of DNA length for molecules ≥ 150 kbp.
Avg Map Rate	The percentage of molecules that map to the reference for molecules ≥ 150 kbp. If no reference genome is provided, the metric is blank.
Estimated Effective Coverage	The coverage number is calculated as follows: <ul style="list-style-type: none"> Average Map Rate * Total DNA / length of the reference For human structural variation detection, at least 80X effective coverage is recommended.

Metric	Description
Avg PLV	Average Positive Label Variance: Percentage of molecule labels absent in reference labels.
Avg NLV	Average Negative Label Variance: Percentage of reference labels absent in molecule labels.
DNA Collected (Gbp)	The total amount of DNA that is detected per flowcell during the run.
Scan Count	The Scans Read number reflects the number of scans submitted by the Instrument Control Software to Bionano Access. The Scans Mapped number reflects the number of scans that have had metrics data generated.
Cumulative DNA (Gbp)	Collected: The total amount of DNA that is detected in this flowcell across all runs of this chip. Target: Target throughput of this flowcell as set in the Experiment. Percent: Percent of target throughput collected.

Chip Metrics Dashboard

Users can see chip metrics for the last 30, 60 or 90 days. The dashboard contains chip run information including **Total DNA (>=150kbp)**, **N50 (>=150kbp)**, **Average label density (>=150kbp)**, **Map rate (%)**, **DNA per scan (Gbp)** and **Longest molecule (kbp)**. Each data point represents one flowcell run on a chip.

1. From the **Bionano Access** main menu, select **Chips**. The **Chips** list appears.
2. Select **Metrics**.
 - Users can click on data points to open the corresponding dashboard.

Bioinformatics Analysis

Analysis Types

Users can perform the following analyses in Bionano Access (**Table 20**):

Table 20. Analysis types

Analysis	Description
Molecules Merge	Merge two or more molecules' jobs into a single molecules job.
Filter molecule jobs	Filter molecules jobs by length and total DNA.
<i>de novo</i> Assembly	Assemble single molecules into consensus maps for SV detection and hybrid scaffold applications.
Guided Assembly	Assembles the whole genome using a reference genome as a seed offering application versatility to detect SVs for both cancer and constitutional research needs
Rare variant analysis	Identify rare variants at low frequencies.
EnFocus™ FSHD analysis	Analyze FSHD relevant regions.
1-Enzyme Hybrid Scaffold	Use a set of Bionano maps and a sequence assembly to build hybrid scaffolds.
2-Enzyme Hybrid Scaffold	Use two sets of Bionano maps and a sequence assembly to build two-enzyme hybrid scaffolds
Alignments	Compare two different maps or molecules to maps by aligning them to each other.
Variant Annotation Pipeline	Annotate SV calls for applications such as identification of rare and potential <i>de novo</i> SVs for trio (mother, father, and proband) or for cancer research.
Convert SMAP to VCF file	Convert insertion, deletion, duplication, and inversion and translocation breakpoint calls in an SMAP file to dbVar-compliant VCF v4.2 format.
EnFocus™ Fragile X analysis	Analyze Fragile X relevant regions

Filter Molecule Jobs

Users can filter BNX files based on molecules length and total DNA. The Bionano BNX file is a raw data view of molecule and label information and quality scores per channel identified during a run or runs if data from multiple runs are merged. BNX v1.3 and above supports one or two label channels (colors). If a user has a chip run that generates more data than recommended for the application, it is possible to down sample the data before running an assembly. Users can also filter the data on label count or length. On filtering a BNX file, a new BNX file is generated so that the raw molecules file is always saved as a copy.

To filter a molecule job:

1. From the **Bionano Access** main menu, select **Analysis**. The **Projects** window appears.
2. Select the project to view from the list.
3. In the **Jobs** list, select a molecules job to filter.
4. In the **Operations** pane, select **Filter Molecule Job**. The **BNX Filter** screen appears.
5. At the **Filtered Molecule Name** field, type the new name for the molecules job.
6. At the **Channel Information** field, **Channel ID**, **Enzyme Motif**, **Name**, and **Reference Name** are displayed.
7. At the **Reference** field, an automatically selected reference is shown up.
8. [Optional] At the **Tags** field, type the keywords to associate with the assembly.
9. [Optional] At the **Description** field, type a brief description.
10. At the **Filter Settings** field, the following options are available for user to select.
 - [Optional] At the **Channel to Keep** field, select one of **Keep Only Channel 1**, **Keep Only Channel 2**, or **Keep Both Channels** (Only available on dual-label molecule job).
 - [Optional] Select the **Label Count** checkbox to type value of Minimum label to keep. By default, Bionano Access automatically uses ten as Minimum.
 - [Optional] Select the **Length (kbp)** checkbox to type values of Minimum and Maximum. By default, Bionano Access automatically uses 100 kbp as Minimum and 5000 kbp as Maximum.
 - [Optional] Select the **Total DNA (Gbp)** checkbox and select **Random Seed** to randomly selected the molecules or select **Keep Longest** to keep the longest molecules.

If a dataset is larger than necessary for a desired application, it is recommended to down sample it in Bionano Access. Type the desired volume of DNA here if this is the case.
 - [Optional] Select the **Scan number** checkbox to reduce the range of scans.

This operation offers base filtering options which will be expanded in the future. Molecules are sampled based on a pseudorandom process, which requires a random seed. A random integer is expected. If the same seed is used, the same molecules would be output. **NOTE:** By default, Bionano Access automatically uses 300 Gbp as Total DNA and 17 as Random Seed.

11. Click **Submit**. Users will receive an email when the filtering is complete.

Merge Molecule Jobs

Users may want to combine data from multiple runs to create a super set of their best data or it may require multiple runs to acquire sufficient coverage to perform an assembly. In such cases, users can merge these molecule files generated from different flow cells for assembly.

The program will automatically select molecule jobs with the same sample name and recognition enzymes for users to merge. The user can still choose other molecule jobs that do not have the same sample name, but they must have the same recognition enzymes.

- Merge molecule jobs that have the best quality data.
- Use BNX files that are with the same sample and reference.
- Do not merge BNX files that have different levels of quality data.
- Do not merge BNX files from different generation Systems. Please contact Bionano Technical Support for assistance if needed.
- Do not merge BNX files generated from Bionano Access v1.2 and greater with those generated from Bionano Access v1.0 or v1.1. Please contact Bionano Technical Support for assistance.
- Dual labeled BNX files can only be merged with dual labeled BNX files with matching recognition enzymes.

BEST PRACTICES

1. From the **Bionano Access** main menu, select **Analysis**. The **Projects** window appears.
2. Select the project to view from the list.
3. In the **Jobs** list, select a **Molecules** job to merge.
4. In the **Operations** pane, select **Merge Molecule Jobs**. A dialog box appears.
5. At the **Merge Molecule Name** field, type the new name for the molecules job.
6. At the **Sample** field, select the sample from the drop-down list.
7. [Optional] At the **Reference** field, select the genome reference from the drop-down list.
8. [Optional] At the **Tags** field, type the keywords to associate with the assembly.
9. [Optional] At the **Description** field, type a brief description.
10. Select the checkboxes of the molecule jobs to merge with the molecules job that was selected in **Step 3** from the list of molecules jobs. the description of the information is shown in the list below (**Table 21**):

Table 21. Molecule jobs information

Info	Description
Name	The molecules job name
Sample	The sample name.
Reference	The reference.
Map Rate (%)	The percentage of molecules that map to the reference for molecules ≥ 150 kbp. If no reference genome is provided, the metric is blank.
N Mol	The total number of molecules
Coverage	The coverage number is calculated as follows: Total DNA Throughput / length of the reference
PLV %	Average Positive Label Variance: Percentage of molecule labels absent in reference labels.
NLV %	Average Negative Label Variance: Percentage of reference labels absent in molecule labels.
SR	The quadratic term in sizing error relative to reference.
SF	The minimum expected sizing error relative to reference.

By default, Bionano Access automatically selects the molecule jobs in the project that are a match with the molecule job selected to merge with. For example, it selects the molecule jobs with the same reference and sample information. There is also a **select-all** option.

11. Click **Submit**. Users will receive an email when the merge is complete.

Align Maps

Users can use map or assembly jobs to generate alignment data. If a user has two independent sets of maps, say each from a different nicking enzyme, it is possible to align the maps to each other to understand the similarities or differences between the two maps.

1. From the **Bionano Access** main menu, select **Analysis**. The **Projects** window appears.
2. Select the project to view from the list.
3. In the **Jobs** list, select the map or assembly to use as a reference.
4. In the **Operations** pane, select **Align Maps**. The **Alignment** screen appears.
5. At the **Alignment Name** field, type the name of the alignment.
6. [Optional] At the **Tags** field, type the keywords to associate with the alignment job.
7. [Optional] At the **Description** field, type a brief description.

At the Alignment p-value field, type the p-value for the alignment. Users can select checkboxes for **Output Best Alignment** and **Swap Anchor - Query**.

8. At the **Sample** field, the name of the sample is displayed.
9. At the **Source Anchor Map** field, the name of the selected consensus map or *de novo* assembly results is displayed.
10. At **Target Query Map**, select the map, assembly, or reference to generate an alignment.
11. Click **Submit**. Users will receive an email when the alignment is complete.

Users can also use molecules jobs to generate alignment results.

- a. From the **Bionano Access** main menu, select **Analysis**. The **Projects** window appears.
- b. Select the project to view from the list.
- c. In the **Jobs** list, select the molecules job to use for alignment.
- d. In the **Operations** pane, select **Align Molecules**. The **BNX Alignment** screen appears.
- e. At the **Alignment Name** field, type the name of the alignment.
- f. [Optional] At the **Tags** field, type the keywords to associate with the alignment job.
- g. [Optional] At the **Description** field, type a brief description.
- h. At the **Sample** field, displays the name of the sample.
- i. At the **Source Molecules** field, displays the name of the selected molecules job.

-
- j. At **Target Map** field, select the map, assembly, or reference to generate an alignment.
 - k. At **Configuration** field, select or edit the configuration file.
 - l. Click **Submit**. Users will receive an email when the alignment is complete.

Generate a Molecule Quality Report

Users can generate Molecule Quality Reports (MQR) with molecules data. Bionano Access will automatically generate an MQR after importing molecule files. Refer to *Molecule Quality Report Guidelines* (CG-30223) for a detailed MQR description.

1. From the **Bionano Access** main menu, select **Analysis**. The **Projects** window appears.
2. Select the project to view from the list.
3. In the Jobs list, select the molecules job to generate molecule quality report.
4. In the **Operations** pane, select **Generate Molecule Quality Report (MQR)**. The **Generate Molecule Quality Report** screen appears.
5. At the **Name** field, displays the name of the molecules job.
6. At the **Sample** field, displays the name of the sample.
7. [Optional] At the **Reference** field, displays the reference of the selected molecules job.
8. At **Enzyme of primary molecule channel** field, select the enzyme.
9. Click **Submit**. Users will receive an email when the molecule quality report is complete.
10. Once complete, in the **Options** pane, select **View Molecule Quality Report**. Values that fall below the threshold will be highlighted in red.

Generate *de novo* Assembly

The *de novo* assembly algorithm is built on the overlap-layout-consensus (OLC) strategy with a maximum likelihood model for scoring alignments. Refer to *Bionano Solve Theory of Operation: Structural Variant Calling* (CG-30110) for more details.

Users can generate *de novo* assembly with molecules data.

- a. From the **Bionano Access** main menu, select **Analysis**. The **Projects** window appears.
- b. Select the project to view from the list.
- c. In the **Jobs** list, select the molecules job to perform *de novo* assembly.
- d. In the **Operations** pane, select **Generate *de novo* Assembly**.
- e. At the **Primary Channel to Assemble** field, select the channel to use for assembly. (**Only available on dual-label molecule job**).
- f. At the **Assembly Name** field, type the name of the assembly.
- g. At the **Estimated Genome Size (Gbp)** field, type the estimated genome size.
- h. [Optional] At the **Tags** field, type the keywords to associate with the assembly.
- i. [Optional] At the **Description** field, type a brief description.
- j. The **Selected BNX** field shows the name of the selected BNX job.
- k. The **Reference** field shows the name of the selected reference.
- l. The **Organism** field shows the name of selected organism (Human or non-human).
- m. [VIA Required] At the **VIA Setting** field, select a sync setting to automatically upload the results if the job is successful, or **None** to disable automatic uploading for this job. (With VIA Settings created beforehand)
- n. The following options in the **Advanced Assembly Options** are **optional** to change the default setting. Refer to *Bionano Solve Theory of Operation: Structural Variant Calling* (CG-30110) for more details.
 - a. The **Expected CN Baseline File** is used as the basis for calling genomic regions with abnormal CN states. It is recommended to select **Default Expected CN Baseline** for human samples. Users can upload and select their own file for human or non-human samples.
 - b. The **Control CNV File** is used to reduce variation unrelated to true CNV events in raw coverage data. It is recommended to select **Default CNV control database** for human samples. Users can also upload and select their own file for human or non-human samples.
 - c. The **CNV Mask Bed File** filters CNVs from regions of the genome that are included in the mask. It is recommended to select the corresponding CNV mask bed file for human samples. Users can also upload and select their own CNV mask bed files for their human or non-human samples.
 - d. The **SV Mask Bed File** filters SVs from regions of the genome that are included in the mask. It is recommended to select the corresponding SV mask bed file for human samples. Users can also upload and select their own mask bed files for their human or non-human samples.

- e. At the **Use Custom Config** field,
 - If a user selects **No**, use the dialog to automatically determine the correct configuration.
 - f. At the **Add Pre-Assembly** field, select **Yes** to add this step to *de novo* assembly process to help with samples that have no reference or if the reference is poor. This option will be turned on automatically if the sample has no reference. If a reference is available, users will have the option to enable pre-assembly.
 - g. At the **Assembly Type** field, select one from **haplotype, non-haplotype with extend and split, non-haplotype without extend and split**. Haplotype is recommended for human samples and non-haplotype without extend and split for non-human samples.
 - h. At the **Cut CMPR (Complex Multi-Path Regions)** field, select **Yes** or **No**. Recommend this to be set to **Yes** for most applications of human samples. Refer to *Bionano Solve Theory of Operation: Structural Variant Calling* (CG-30110) for more details.
 - If a user selects **Yes**, then the list of configuration files will be displayed. Select the configuration file from the default list or select a customized configuration.
1. At the **Add Pre-Assembly** field, select **Yes** to add this step to *de novo* assembly process to help with samples that have no reference or if the reference is poor. This option will be turned on automatically if the sample has no reference. If a reference is available, users will have the option to enable pre-assembly.
 2. To customize a configuration, click the **Edit** icon in one of the default configurations. The **Configuration** dialog box appears.
 3. Define the settings for *de novo* assembly, and then click **Save As**.
 4. Type the name of the configuration, and then click **OK**.
 5. The customized configuration appears in the list of configurations.
 - a. At the **VCF Experiment Id** field, type the Id number for the VCF result.
 - b. Select **Disable VCF Breakpoint Uncertainty** checkbox will remove uncertain breakpoint in VCF result.
 - The following default setting for annotation pipeline in the **Advanced Variant Annotation Pipeline Settings** are optional to change when **Run Variant Annotation with control SV database** is enabled.
 6. Select the SV control database for the annotation pipeline. Users can create their own control database. For more detail information, please refer to *Bionano Solve Theory of Operation: Variant Annotation Pipeline* (CG-30190).
 7. At the **Known genes** field, select a bed file from the drop-down list.
 8. Use the default values or enter the values to use for the parameter fields of variant annotation.
 - a. Click **Next**.
 - b. Users can run the *de novo* Assembly with **Original BNX**, or **Recommended**, which is down sampled to 80X effective coverage.

Depending on the volume of data and coverage, the *de novo* assembly run time may vary. Refer to *Bionano Solve Theory of Operation: Structural Variant Calling* (CG-30110) for more details.

If **Variant Annotation** is selected, only one annotated *de novo* assembly job will be generated.

- c. Click **Submit**. Users will receive an email when the assembly is complete.

Generate Guided Assembly

Rather than a complete *de novo* assembly, guided assembly uses the reference genome as seed for the initial assembly, extension, and refinement phases of the pipeline. Refer to *Bionano Solve Theory of Operation: Structural Variant Calling* (CG-30110) for more details.

Users can generate guided assembly with molecules data.

- a. From the **Bionano Access** main menu, select **Analysis**. The **Projects** window appears.
- b. Select the project to view from the list.
- c. In the **Jobs** list, select the molecules job to perform guided assembly.
- d. In the **Operations** pane, select **Generate Guided Assembly**.
- e. At the **Primary Channel to Assemble** field, select the channel to use for assembly. (**Only available on dual-label molecule job**).
- f. At the **Guided Assembly Name** field, type the name of the assembly.
- g. At the **Operation Type** field, select either **Low Allelic Fraction** (default) for somatic mutation analysis or **Constitutional** for constitutional applications.
- h. At the **Estimated Genome Size (Gbp)** field, type the estimated genome size.
- i. [Optional] At the **Tags** field, type the keywords to associate with the assembly.
- j. [Optional] At the **Description** field, type a brief description.
- k. The **Selected BNX** field shows the name of the selected BNX job.
- l. The **Reference** field shows the name of the selected reference.
- m. The **Organism** field shows the name of selected organism (Human or non-human).
- n. [VIA Required] At the **VIA Setting** field, select a sync setting to automatically upload the results if the job is successful, or **None** to disable automatic uploading for this job.
- o. The following options in the **Advanced Assembly Options** are **optional** to change the default setting. Refer to *Bionano Solve Theory of Operation: Structural Variant Calling* (CG-30110) for more details.
 1. The **Expected CN Baseline File** is used as the basis for calling genomic regions with abnormal CN states. It is recommended to select **Default Expected CN Baseline** for human samples. Users can upload and select their own file for human or non-human samples.
 2. The **Control CNV File** is used to reduce variation unrelated to true CNV events in raw coverage data. It is recommended to select **Default CNV control database** for human samples. Users can also upload and select their own file for human or non-human samples.

3. The **CNV Mask Bed File** filters CNVs from regions of the genome that are included in the mask. It is recommended to select the corresponding CNV mask bed file for human samples. Users can also upload and select their own CNV mask bed files for their human or non-human samples.
4. The **SV Mask Bed File** filters SVs from regions of the genome that are included in the mask. It is recommended to select the corresponding SV mask bed file for human samples. Users can also upload and select their own mask bed files for their human or non-human samples.
5. At the **Use Custom Config** field,
 - If a user selects **No**, then the **Assembly Type** field will be displayed. Select one from **haplotype**, **non-haplotype with extend and split**, **non-haplotype without extend and split**. Haplotype is recommended for human samples and non-haplotype without extend and split for non-human samples.
 - If a user selects **Yes**, then the list of configuration files will be displayed. Select the configuration file from the default list or select a customized configuration.
 - a. To customize a configuration, click the **Edit** icon in one of the default configurations. The **Configuration** dialog box appears.
 - b. Define the settings for guided assembly, and then click **Save As**.
 - c. Type the name of the configuration, and then click **OK**.
 - d. The customized configuration appears in the list of configurations.
6. At the **VCF Experiment Id** field, type the Id number for the VCF result.
7. Select **Disable VCF Breakpoint Uncertainty** checkbox will remove uncertain breakpoint in VCF result.
 - a. The following default setting for annotation pipeline in the **Advanced Variant Annotation Pipeline Settings** are optional to change when **Run Variant Annotation with control SV database** is enabled.
8. Select the SV control database for the annotation pipeline. Users can create their own control database. For more detail information, please refer to *Bionano Solve Theory of Operation: Variant Annotation Pipeline* (CG-30190).
9. At the **Known genes** field, select a bed file from the drop-down list.
10. Use the default values or enter the values to use for the parameter fields of variant annotation.
 - b. Click **Next**.
 - c. Users can run the Guided Assembly with **Original BNX**, or **Recommended**, which is down sampled to 80X effective coverage.

Depending on the volume of data and coverage, the guided assembly run time may vary. Refer to *Bionano Solve Theory of Operation: Structural Variant Calling* (CG-30110) for more details.

If **Variant Annotation** is selected, only one annotated guided assembly job will be generated.

- d. Click **Submit**. Users will receive an email when the assembly is complete.

Generate Rare Variant Analysis

The Rare Variant Analysis (RVA) is designed specifically to identify variants at low variant allele frequencies in heterogeneous samples such as cancers or samples with allele mosaicism. For more information on how the algorithm runs, refer to *Bionano Solve Theory of Operations: Structural Variant Calling* (CG-30110). Users can generate rare variant analysis with molecules data.

1. From the **Bionano Access** main menu, select **Analysis**. The **Projects** window appears.
2. Select the project to view from the list.
3. In the **Jobs** list, select a molecule job to perform rare variant analysis.
4. In the **Operations** pane, select **Generate Rare Variant Analysis**.
5. At the **Rare Variant Analysis Name** field, type the name of the analysis.
6. [Optional] At the **Tags** field, type the keywords to associate with the analysis.
7. [Optional] At the **Description** field, type a brief description.
8. The **Selected BNX** field shows the name of the selected BNX job.
9. The **Reference** field shows the name of the selected reference.
10. [VIA Required] At the **VIA Setting** field, select a sync setting to automatically upload the results if the job is successful, or **None** to disable automatic uploading for this job.
11. The following options in the **Advanced Rare Variant Analysis Options** are optional to change the default setting.
 - a. At the **Expected CN Baseline File** field, select **Default Expected CN Baseline** or customer's own baseline file from the drop-down list.
 - b. At the **Control CNV File** field, select **Default CNV control database** for human sample, **mm10 control CNV** for mouse sample, or customer's own CNV control database from the drop-down list.
 - c. At the **CNV Mask Bed File** field, select a bed file from the drop-down list.
 - d. At the **SV Mask Bed File** field, select a bed file from the drop-down list.
 - e. At the **Configuration** field, select the arguments.
12. The following default setting for annotation pipeline in the **Advanced Variant Annotation Pipeline Settings** are optional to change when **Run Variant Annotation with control SV database** is enabled.
 - a. Select the SV control database for the annotation pipeline. Users can create their own control database. For more detail information, please refer to *Bionano Solve Theory of Operation: Variant Annotation Pipeline* (CG-30190).
 - b. At the **Known genes** field, select a bed file from the drop-down list.
 - c. Use the default values or enter the values to use for the parameter fields of variant annotation.

13. Click **Submit**. Users will receive an email when the assembly is complete.

Generate EnFocus™ FSHD Analysis

EnFocus™ FSHD Analysis targets the regions of the genome related to FSHD. Please refer to *Bionano Solve Theory of Operation: Bionano EnFocus™ FSHD Analysis (CG-30321)* for more details. The EnFocus™ FSHD Analysis pipeline cannot run from merged BNX and can only be run on BNX files that come from one flowcell or one chip run. It is only applicable for human samples using hg38 as the reference.

1. From the **Bionano Access** main menu, select **Analysis**. The **Projects** window appears.
2. Select the project to view from the list.
3. In the **Jobs** list, select a molecule job to generate FSHD analysis.
4. At the field of **FSHD Analysis Background Information**, check more information about this analysis.
5. At the **FSHD Job Name field**, type the name of the analysis.
6. [Optional] At the **Tags** field, type the keywords to associate with the analysis.
7. [Optional] At the **Description** field, type a brief description.
8. The **Sample Name** field shows the name of the selected sample.
9. The **Selected BNX** field shows the name of the selected BNX job.
10. The **Reference** field shows the name of the selected reference.
11. The **Show Additional Information** field is enabled by default.
12. Click **Submit**. Users will receive an email when the assembly is complete.

Generate EnFocus™ Fragile X Analysis

EnFocus™ Fragile X Analysis targets the region of the genome related to Fragile X. The EnFocus™ Fragile X Analysis pipeline cannot run from merged BNX and can only be run on BNX files that come from one flowcell or one chip run. It is only applicable for human samples using hg38 as the reference. Refer to the *Bionano Solve Theory of Operation: Bionano EnFocus™ Fragile-X Analysis (CG-30457)* for more details.

- a. From the **Bionano Access** main menu, select **Analysis**. The **Projects** window appears.
- b. Select the project to view from the list.
- c. In the **Jobs** list, select a molecules job to generate Fragile X analysis.
- d. At the field of **Fragile X Analysis Background Information**, check more information about this analysis.
- e. At the **Fragile X Job Name** field, type the name of the analysis.
- f. [Optional] At the **Tags** field, type the keywords to associate with the analysis.

- g. [Optional] At the **Description** field, type a brief description.
- h. The **Sample Name** field shows the name of the selected sample.
- i. The **Selected BNX** field shows the name of the selected BNX job.
- j. The **Reference** field shows the name of the selected reference.
- k. Click **Submit**. Users will receive an email when the assembly is complete.

Generate Hybrid Scaffold

Users can merge Bionano map or assembly jobs with sequence assemblies to produce long hybrid scaffolds that represent the chromosome structure for analysis. Recommendation: users do not use haplotype assemblies to generate a hybrid scaffold.

1. From the **Bionano Access** main menu, select **Analysis**. The **Projects** window appears.
2. Select the project to view from the list.
3. In the **Jobs** list, select the map or assembly to use as a reference.
4. In the **Operations** pane, select **Generate Hybrid Scaffold**. The **Scaffold** screen appears.
5. At the **Scaffold Name** field, type a name for the scaffold.
6. [Optional] At the **Tags** field, type the keywords to associate with the hybrid scaffold job.
7. [Optional] At the **Description** field, type a brief description.
8. At the **Selected Map** field, by default the software shows the map to use for scaffolding.
9. At the **Enzyme Selection** field, select the enzyme from the drop-down list.
10. At the **FASTA** field, select the file from the drop-down list.
11. At the **Conflict Resolution** field, for best practices, select **ResolveConflicts** for both Bionano Assembly and Sequence Assembly. For more information, see more in *Bionano Solve Theory of Operation: Hybrid Scaffold* (CG-30073).
12. At the **Trim Overlapping Sequence Contigs** field, turn it on or off. For more information, see more in *Bionano Solve Theory of Operation: Hybrid Scaffold* (CG-30073).
13. At the **Configuration** field, select the configuration file from the default list or select a customized configuration.
 - a. To customize a configuration, click the **Edit** icon in one of the default configurations. The **Configuration** dialog box appears.
 - b. Define the settings for hybrid scaffold, and then click **Save As**.
 - c. Type the name of the configuration, and then click **OK**. The customized configuration appears in the list of configurations.

14. Click **Submit**. Users will receive an email when the hybrid scaffold is complete.

Generate 2-Enzyme Hybrid Scaffold

Users can also generate a hybrid scaffold using two enzymes. If DLE-1 is one of the enzymes, the DLE-1 assembly or CMAP must be selected first.

1. From the **Bionano Access** main menu, select **Analysis**. The **Projects** window appears.
2. Select the project to view from the list.
3. In the **Jobs** list, select the map or assembly to use as a reference.
4. In the **Operations** pane, select **Generate 2-Enzyme Hybrid Scaffold**. The **2 Enzyme Scaffold** screen appears.
5. At the **Scaffold Name** field, type a name for the scaffold.
6. [Optional] At the **Tags** field, type the keywords to associate with the hybrid scaffold job.
7. [Optional] At the **Description** field, type a brief description.
8. At the **First Map** and **Enzyme** field, by default the software shows the map to use for scaffolding.
9. At the **Second Map** field, select the second map to use for scaffolding. The **Enzyme** field will automatically show up after that.
10. At the **Fasta** field, select the file from the drop-down list.
11. At the **Trim Overlapping Sequence Contigs** field, turn it on or off. For more information, see more in *Bionano Solve Theory of Operation: Hybrid Scaffold (CG-30073)*.
12. At the **Configuration** field, select the configuration file from the default list or select a customized configuration.
 - a. To customize a configuration, click the **Edit** icon in one of the default configurations. The **Configuration** dialog box appears.
 - b. Define the settings for hybrid scaffold, and then click **Save As**.
 - c. Type the name of the configuration, and then click **OK**.

The customized configuration appears in the list of configurations.

13. Click **Submit**. Users will receive an email when the two-enzyme hybrid scaffold is complete.

Perform Variant Annotation - Single

The purpose of the variant annotation pipeline (VAP) is to enable users to determine if a Bionano structural variant (SV) call is relevant to certain physical or disease traits in humans. It can help to identify if a variant overlaps with annotated genes or is a potential false positive call.

1. From the **Bionano Access** main menu, select **Projects**. The **Projects** window appears.
2. Select the project to view from the list.
3. In the **Jobs** list, select the map or assembly to use as a reference.
4. In the **Operations** pane, select **Generate Variant Annotation - Single**.
5. At the **Name** field, type the name for the variant annotation pipeline.
6. [Optional] At the **Tags** field, type the keywords to associate with the job.
7. [Optional] At the **Description** field, type a brief description.
8. At the **Control SV database** field, select the control SV database from the drop-down list.

The control SV database is used to estimate the percentage of similar SVs found in Bionano control samples. It is recommended to select the corresponding control SV database file for human or mouse samples. Users can also upload and select their own control SV bed files for their human or non-human samples. Users can also run variant annotation pipeline with no control SV database file selected. Refer to *Bionano Solve Theory of Operations: Structural Variant Calling* (CG-30110) for more details.

9. At the **Known genes** field, select a bed file from the drop-down list.

The gene annotation bed file is used to annotate SVs. It is recommended to select the corresponding gene bed file for human or mouse samples. Users can upload and select their own file for their human or non-human samples. Users can also run variant annotation pipeline with no gene bed file selected. Refer to *Bionano Solve Theory of Operations: Structural Variant Calling* (CG-30110) for more details.

10. The following default settings for annotation pipeline in the **Advanced Variant Annotation Pipeline Settings** are optional to change. Use the default values or enter the values to use for the following parameter fields.

For more details, see the *Bionano Solve Theory of Operations: Variant Annotation Pipeline* (CG-30190) for guidance on setting these parameters.

11. Click **Submit**. The spinning arrows indicate that the pipeline is uploading data to the server.
12. When the spinning arrows disappear, click **Close**. Bionano Access sends an email to notify the user when the variant annotation pipeline is complete.

Perform Dual Analysis

The purpose of the dual variant annotation pipeline is to identify variant differences in two samples, such as somatic in tumor-normal pair studies.

1. From the **Bionano Access** main menu, select **Projects**. The **Projects** window appears.
2. Select the project to view from the list.
3. In the **Jobs** list, select the map or assembly to use as a reference.
4. In the **Operations** pane, select **Generate Dual Analysis**.
5. At the **Name** field, type the name for the variant annotation pipeline.
6. [Optional] At the **Tags** field, type the keywords to associate with the job.
7. [Optional] At the **Description** field, type a brief description.
8. At the **Control Assembly** field, select the control sample assembled result from the drop-down list.

At the **Control SV database** field, select the control SV database from the drop-down list. The control SV database is used to estimate the percentage of similar SVs found in Bionano control samples. It is recommended to select the corresponding control SV database file for human or mouse samples. Users can also upload and select their own control SV bed files for their human or non-human samples. Users can also run variant annotation pipeline with no control SV database file selected. Refer to *Bionano Solve Theory of Operations: Structural Variant Calling* (CG-30110) for more details.

9. At the **Known genes** field, select a bed file from the drop-down list.

The gene annotation bed file is used to annotate SVs. It is recommended to select the corresponding gene bed file for human or mouse samples. Users can upload and select their own file for their human or non-human samples. Users can also run variant annotation pipeline with no gene bed file selected. Refer to *Bionano Solve Theory of Operations: Structural Variant Calling* (CG-30110) for more details.

10. The following default setting for annotation pipeline in the **Advanced Variant Annotation Pipeline** Settings are optional to change. Use the default values or enter the values to use for the following parameter fields.

For more details, see the *Bionano Solve Theory of Operations: Variant Annotation Pipeline* (CG-30190) for guidance on setting these parameters.

11. Click **Submit**. The spinning arrows indicate that the pipeline is uploading data to the server.
12. When the spinning arrows disappear, click **Close**. Bionano Access sends an email to notify the user when the variant annotation pipeline is complete.

Perform Trio Analysis

Users can identify rare and potential *de novo* SVs for the trio Variant Analysis Pipeline (mother, father, and proband).

1. From the **Bionano Access** main menu, select **Projects**. The **Projects** window appears.
2. Select the project to view from the list.
3. In the **Jobs** list, select the map or assembly to use as a reference.
4. In the **Operations** pane, select **Generate Trio Analysis**.
5. At the **Name** field, type the name for the variant annotation pipeline.
6. [Optional] At the **Tags** field, type the keywords to associate with the job.
7. [Optional] At the **Description** field, type a brief description.
8. At the **Father assembly** field, select the assembly from the drop-down list.
9. At the **Mother assembly** field, select the assembly from the drop-down list.
10. At the **Control SV database** field, select the control SV database from the drop-down list.

The control SV database is used to estimate the percentage of similar SVs found in Bionano control samples. It is recommended to select the corresponding control SV database file for human or mouse samples. Users can also upload and select their own control SV bed files for their human or non-human samples. Users can also run variant annotation pipeline with no control SV database file selected. Refer to *Bionano Solve Theory of Operations: Structural Variant Calling* (CG-30110) for more details.

11. At the **Known genes** field, select a bed file from the drop-down list.

The gene annotation bed file is used to annotate SVs. It is recommended to select the corresponding gene bed file for human or mouse samples. Users can upload and select their own file for their human or non-human samples. Users can also run variant annotation pipeline with no gene bed file selected. Refer to *Bionano Solve Theory of Operations: Structural Variant Calling* (CG-30110) for more details.

12. The following default setting for annotation pipeline in the **Advanced Variant Annotation Pipeline** Settings are optional to change. Use the default values or enter the values to use for the following parameter fields.

For more details, see the *Bionano Solve Theory of Operations: Variant Annotation Pipeline* (CG-30190) for guidance on setting these parameters.

13. Click **Submit**. The spinning arrows indicate that the pipeline is uploading data to the server.
14. When the spinning arrows disappear, click **Close**. Bionano Access sends an email to notify the user when the variant annotation pipeline is complete.

Visualization Features

Navigate to the Viewer

Users can navigate to the **Viewer** screen from the **Project** page.

1. From the **Bionano Access** main menu, select **Analysis**. The **Projects** window appears.
2. Select the project to view from the list.
3. In the **Jobs** list, select an assembly, alignment, rare variant analysis, variant annotation pipeline, or scaffold job. Users can view these jobs on the **Viewer** screen.
4. In the **Options** pane, depending on the job selected, these are possible links to get to *Viewer* screen:
 - View Results
 - View Results with Conflicts
 - View Molecule Alignment
 - View Maps

Tour

Each of the four visualizations have a built-in guided tour. To access the tour for the currently selected view, click the **Info** icon in the **Menu** bar or, to pick a different view, click the **dropdown arrow** next to the icon and select one of the available tours. To return from a tour, click the **Return** icon.

Circos Plot visualization

The Circos Plot is an interactive visualization tool to facilitate the identification and analysis of similarities and structural variants in genomics studies. The Circos plot is displayed by default for human assemblies, rare variant analysis, and variant annotations.

NOTE: For more mouse and keypad shortcuts to maneuver the Circos plot, click on the **?** icon, which is the second to last icon on the top panel. For more detail, please see the “Key and Mouse” shortcuts section.

1. In Bionano Access, navigate to the **Viewer** screen of the job to analyze.
2. The **Circos plot** is the default viewer for human samples. Users can select **Circos Plot** from the drop-down list at upper-left corner of the **Viewer** screen from the viewer.

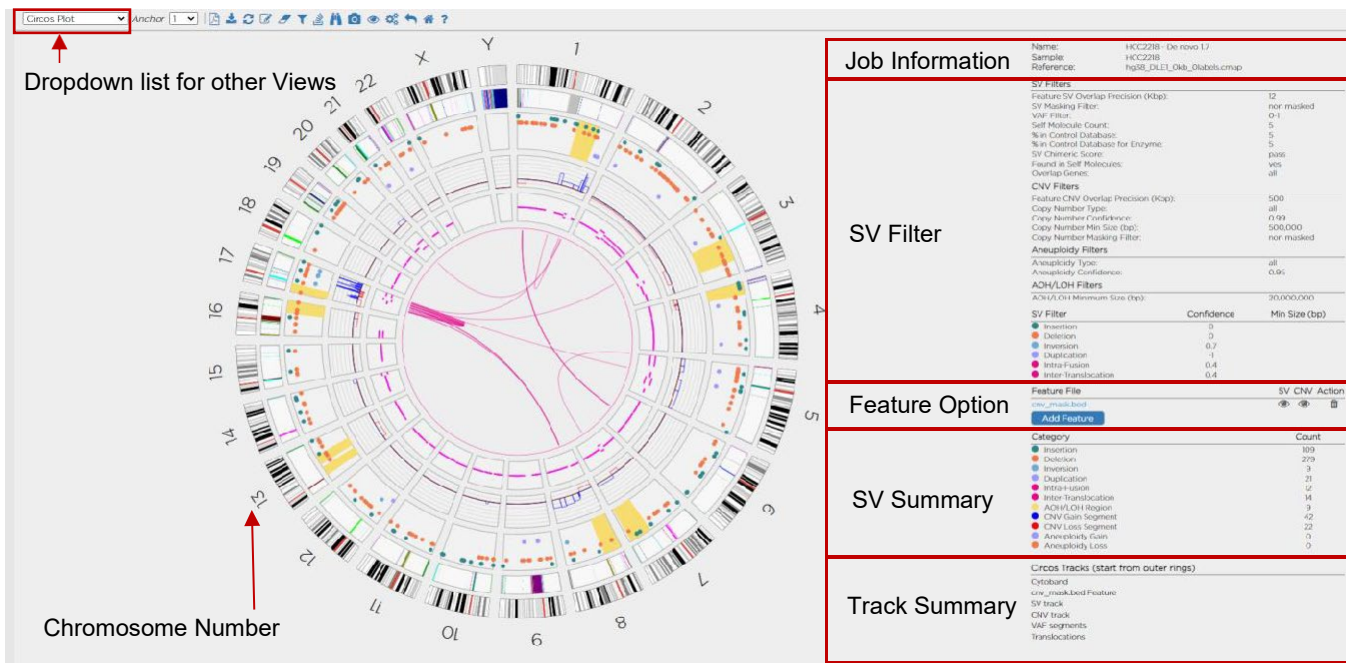


Figure 2. Circos Plot visualization example

- The Circos Plot is displayed as the circular plot on the left, while on the right, the user will find detailed information of the job, SV, CNV and aneuploidy call Filter Criteria, the ability to add feature files, and a color legend containing counts for each call category on the right.
- To zoom in on the Circos Plot, rotate the mouse-wheel forward (scroll it down). The user can also click and drag the plot around using the mouse. In the Circos Plot each chromosome is positioned end-to-end in a large circle. Each track represents a different type of annotation or result.
- Users can change the **SV filter** settings to filter the SV calling. Please follow the “SV filter” section for more detail.
- Users can add current displayed SV calling to the Curated List. For more detail, please follow Adding SV to Curated Variant List/SV report for Circos Plot.
- The outermost numerical track corresponds to the chromosome number, with cytoband information shown in the black-and-white banding pattern. New in Bionano Access 1.7, Chromosomes 1 through 22 (the autosomes) are designated by their chromosome number. The sex chromosomes are designated by X or Y for human samples.
- Below the cytoband, users may find or may add relevant genome annotations (feature files) which can be used for either data filtering or visualization. These can be loaded in the Circos Plot as separate tracks. Each feature in the file is a separate vertical line. Users can zoom in and hover over the lines to get more information about the feature.
- The next track includes the detected SVs as represented by color-coded dots. A user can hover over the dot to view summary info about a given SV call. AOH/LOH regions also show in this track indicated with a yellow- colored block.
- The next track shows copy number calls. For the copy number track, the baseline state of autosomes in a diploid genome is a copy number of 2 for human sample. Three color coded lines are used to visualize the copy number calls. Black is the baseline, blue is CNV gain, and red is CNV loss. If a region shows a copy number gain, the line will move outward from 2 (colored blue), reflecting an increase in the local copy number state. If the region has a loss, it will move inward (colored red).
- The next track shows Variant Allele Fraction (VAF) segments. The segmentation smooth line helps distinguish changes in the VAF pattern across the genome. By default, the VAF smooth lines will only display in *de novo* results.

For RVA results, the VAF track is not displayed by default. Users can change this setting in **View Options** for the Circos Plot.

- Clicking on any **SV dots**, **CNV**, **AOH/LOH region** or **cytoband** in the Circos Plot will navigate to the **Genome Browser Viewer**. Clicking on a **VAF smooth line** segment will navigate to the **Whole Genome Viewer**.
- The user can Shift + Left Click on a **Feature** in the feature track to annotate the corresponding name. Clear feature name(s) by clicking the **Clear Feature Annotations** icon.

NOTE: Please refer to the **SV Workflow** video on <https://bionanogenomics.com/support-page/bionano-access/> to see how the following features can be used in visualizing, filtering for and reporting SVs of interest.

ADD FEATURE

1. Click the **Add Features** icon. The **Feature Filter Settings** dialog box appears.
2. At the **Feature Selection** field, click the drop-down list and select one bed file.
 - To add a new feature file to Access, please follow the **System Features** settings.
3. At the **Structural Variant Overlap Precision (Kbp)** field, type the value. 12 is the system default.
4. At the **Structural Variant Filter Setting** field, select one from **Show all structural variants**, **Show structural variants that overlap BED Regions** and **Hide structural variants that overlap BED Regions**. **Show all structural variants** is the system default. This default setting will load the feature file without filtering the data.
5. At the **CNV Segment Overlap Precision (Kbp)** field, type the value. 500 is the system default.
6. At the **CNV Segment Filter Setting** field, select one from **Show all CNV segments**, **Show CNV segments that overlap BED Regions** and **Hide CNV segments that overlap BED Regions**. **Show all CNV segments** is the system default.
7. Click **OK**.
8. The **Feature** track will be added into Circos Plot.

Genome Browser visualization

The genome browser view is an interactive visualization tool for analyzing variants on a chromosome. The central viewing region includes tracks that display information for a given chromosome.

1. In Bionano Access, navigate to the **Viewer** screen of the job to analyze.
2. Select **Genome Browser** from the drop-down list.
3. **Genome Browser** viewer shows up.

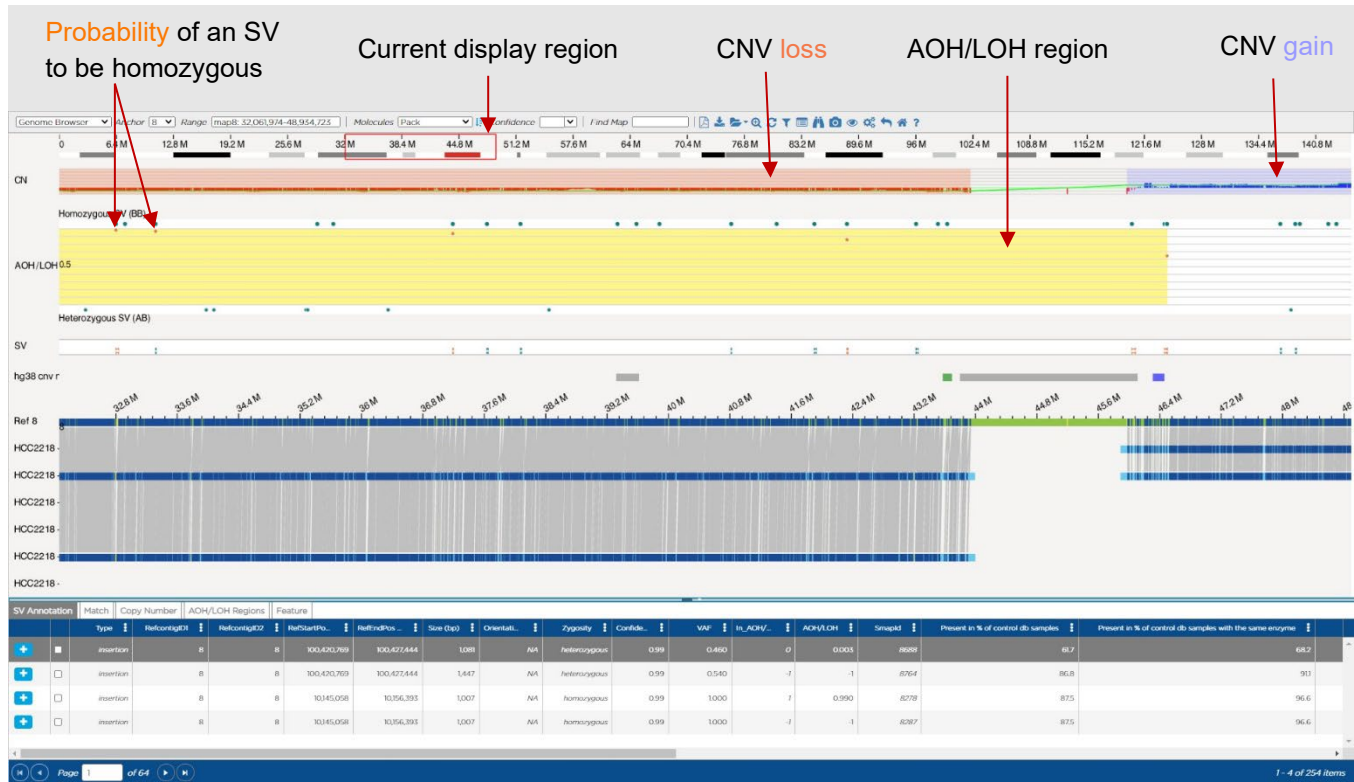


Figure 3. Genome Browser visualization example

- At the **Anchor** field, change the view of other contigs and chromosomes in the same sample.
- At the **Range** field, users can set the range of the coordinates on the same chromosome.
- At the **Molecules** field, user can sort the molecules in the order of pack, start, end, confidence, length, and label density.
- At the **Confidence** field, user can view molecules in the genome maps depending on their levels of confidence. Users can also enter a specific value manually.
- At the **Find Map** field, user can highlight the genome map by typing in the Genome Map ID.
- The **Cytoband** information is shown in the black-and-white banding pattern with centromere in red. The displayed range is marked in a red box. Users can draw a new red box by clicking and dragging the mouse over the cytoband region of interest.
- By default, the **CN (Copy Number)** track is shown as a smooth light green line with highlighted regions for deletions and duplications. After zooming in, it shows the individual copy number values for each label with high copy number in blue and low copy number is red by default.
- The **SV** track marks the location of each SV using the same color code used in the Circos Plot legend.
- The **AOH/LOH** track shows AOH/LOH calls and highlights these regions yellow. Users can show all AOH/LOH probability data points as orange dots (disabled by default). To enable this option, please follow the **View Options** for the **Genome** browser.
- The **Ref** track displays the reference information of the selected chromosome or contig. By default, it is shown as a green map with dark blue labels.
- The **Map** track displays the genome maps that are aligned to the selected chromosome or contig. By default, it is shown as a light blue background with aligned labels in dark blue and unaligned labels in yellow.

- Users can change the **SV filter** settings to filter the SVs called. Please follow the “SV filter” section for more detail.
- Users can add current displayed SV calling to the Curated List. For more detail, please follow *Adding SV to Curated Variant List/SV report* for **Genome** browser.
- If match groups or maps are hidden, click the **Show All** icon to view them again.
- Right click the **Ref** or **Map** track to select the options shown in **Table 22**.

Table 22. Map Track options

Option	Description
Align Maps to Current Label (Ref track only)	Adjust position of all maps by aligning to the selected label in the reference.
Hide	Hide the selected reference or genome map.
Hide the Others	Hide the other reference or genome map(s).
Collapse	Collapse multiple Map tracks into one.
Hide (Show) Ruler	Hide or Show ruler of the reference or genome map.
Hide (Show) Match groups	Hide or Show Match groups of the reference or genome map.
Show Feature (Ref track only)	Select and load a feature file.
Remove All Features	Remove all added features.
Invert	Invert the orientation of the selected reference or genome map.

Whole Genome Visualization

The whole genome view in Bionano Access helps visualize genomic locations across all chromosomes with **Copy number**, **AOH/LOH** and **Variant Allele Fraction (VAF)** in three separate plots. AOH/LOH is plotted only in *the de novo* assembly workflow. For each graph, Bionano displays the data identifier, one or more attributes, and the data.

1. In Bionano Access, navigate to the **Viewer** screen of the job to analyze.
2. Select **Whole Genome** from the drop-down list.
3. **Whole Genome** view shows up.

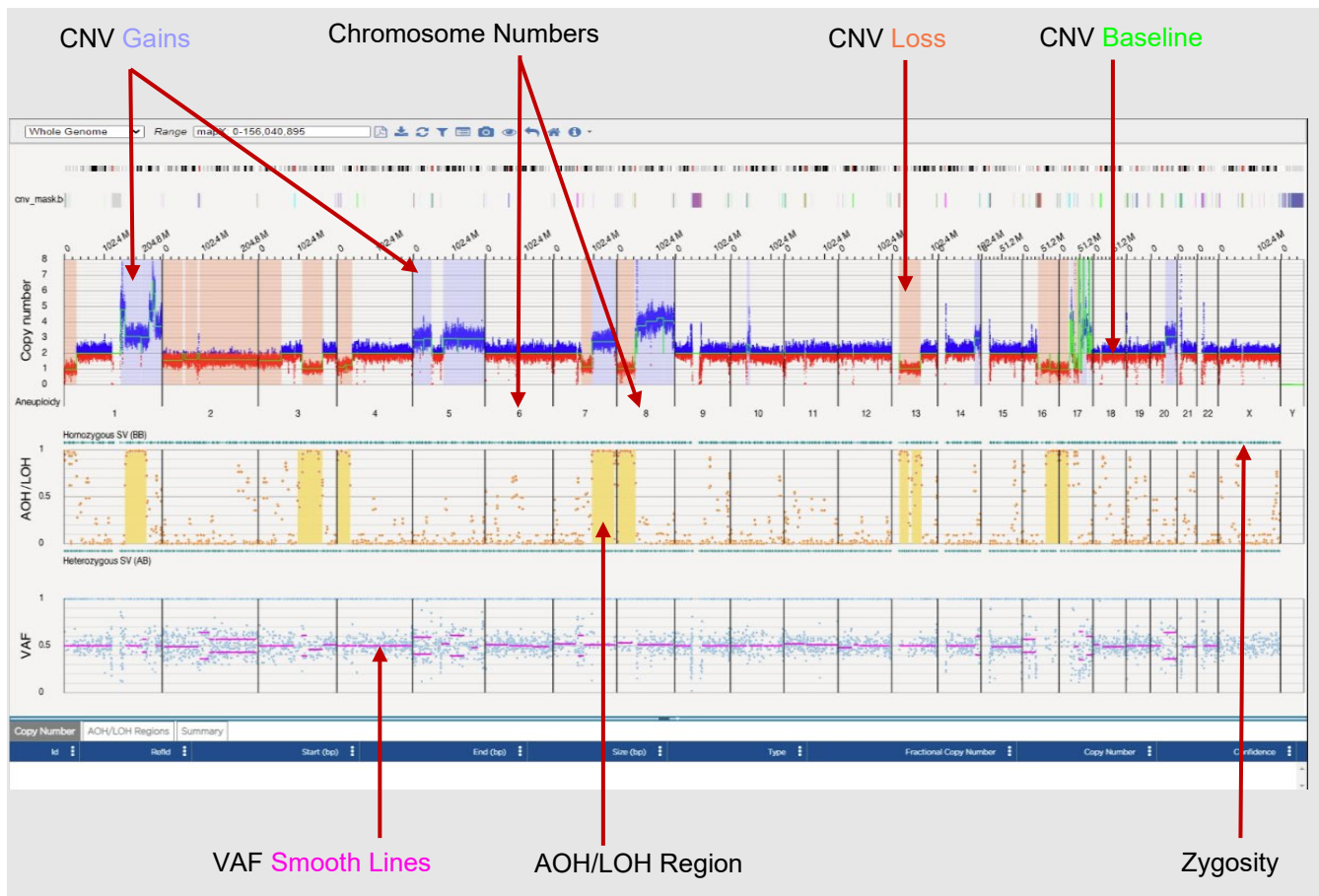


Figure 4. Whole Genome visualization example.

- At the **Range** field, user can set the chromosome and range of the coordinates to view.
- The **Copy Number** plot represents the CNV profiles across the chromosomes. The Y axis shows copy number for each of the chromosomes plotted on the X axis. Molecules showing regions with increased copy number

from the calculated baseline (green) are shown in blue and regions with decreased copy number are shown in red.

- The **AOH/LOH** graph represents regions of AOH/LOH calculated for structural variants identified in the *de novo* workflow. The X axis represents chromosomes, and the Y axis represents the range of zygosity for these genomic locations. The teal dots represent zygosity of the variant. Users can also plot the probability of the SV to be present in the LOH/AOH region (orange dot). The regions of the genome that have a consistently high AOH/LOH calls will be indicated with a yellow-colored block. The AOH/LOH graph is only available on the *de novo* Assembly result.
- The **Variant Allele Fraction (VAF)** plot represents the fraction of copies that are of a particular allele in a defined population. The VAF track provides a visual illustration of this difference with a scatter plot of allele frequencies for a given variant. The Y axis represents the range of the allelic frequency across the different chromosomes plotted on the X axis. The mean of the frequency rests around .5 for diploid genome and is color-coded magenta. If the data show fluctuations in the ploidy, the mean value changes and is visualized by more than one magenta line across the region of the genome that shows polyploidy. The Variant Allele frequency is calculated for all SVs detected in the SV detection pipelines. Users can choose to filter variants based on their allelic frequency.

Ideogram visualization

Ideograms provide a schematic representation of chromosomes. They are used to show the relative size of the chromosomes and their characteristic banding patterns. Bionano Access 1.7 now has an interactive viewer to show the location of the structural variants, copy number variants and loss of heterozygosity on human chromosomes. SVs, CNVs, AOH/LOH can be chosen to display on chromosomes. All chromosomes and cytobands can be viewed via Ideogram.

1. In Bionano Access, navigate to the **Viewer** screen of the job to analyze.
2. Select **Ideogram** from the drop-down list.
3. **Ideogram** view is displayed.

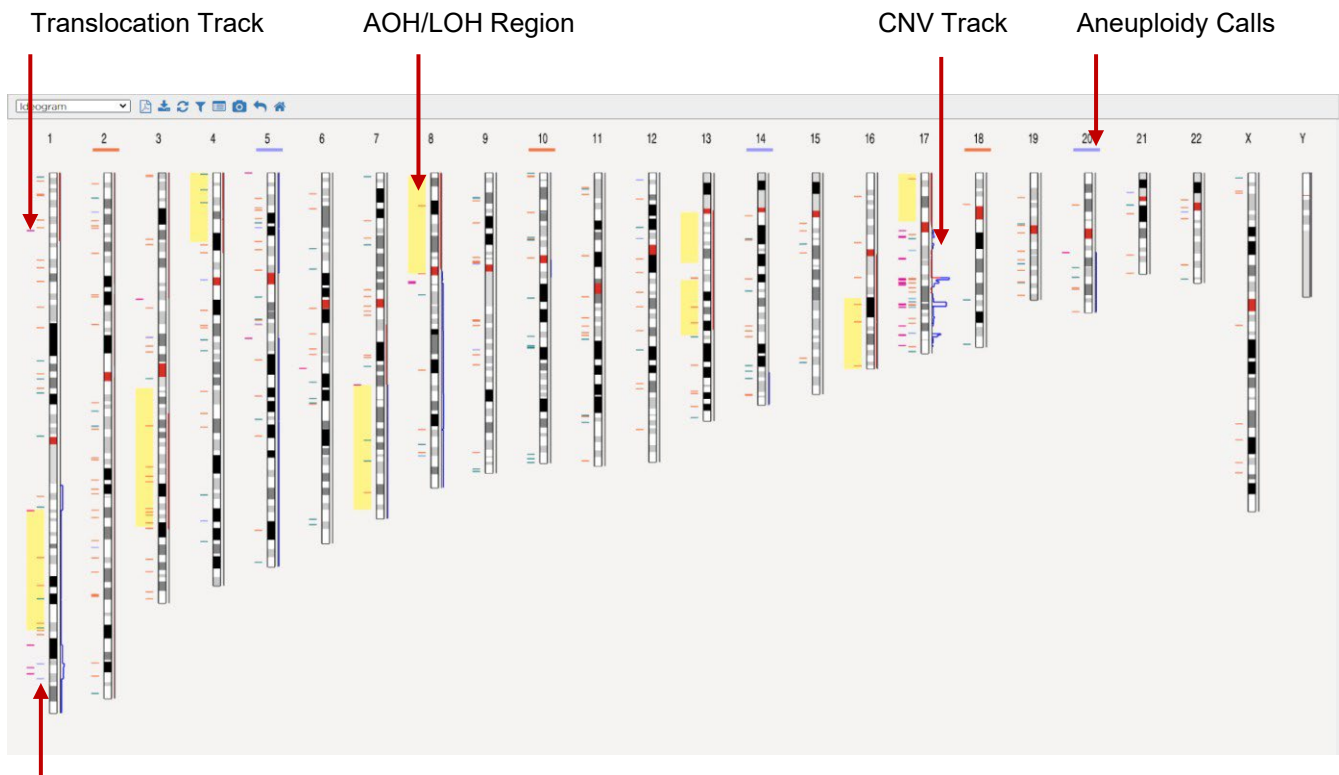


Figure 5. Ideogram view example.

- The Cytoband information is shown in the black-and-white banding pattern with centromere in red.
- The SV track is on the left and the CNV track is on the right of each chromosome.
- Aneuploidy information is also marked on the top under the chromosome number as a blue band for a gained copy of a chromosome and a red band for a lost copy of a chromosome.
- The yellow blocks in the SV track represent the AOH/LOH regions.

Curated Variant List

The curated variant list view contains the SV calls that the user added to the curated list. It contains the SV details and classifications made in the variant classifier view. Administrators and project leaders can generate an SV report with the classification information after the review is completed. Bionano Access provides features for the manual classification of variants per the ACMG guidelines. These features are solely intended for Research Use Only.

1. In Bionano Access, navigate to the **Viewer** screen of the job to analyze.
2. Select **Curated Variant List** from the drop-down list.
3. **Curated Variant List** view shows up.

Classification	Notes	Type	SVID	RefStartPos (bp)	RefEndPos (bp)	Size (bp)	Present in % of controls	Overlap Genes	Putative Gene Fusion	GCN	Chr	Details
Pathogenic		insertion	272	86,581,028	86,582,337	5,939	0	CLCA4-AS1	-	cgmlnGRCh38	1	[Details]
Likely pathogenic		deletion	289	98,577,072	98,579,380	538	1	-	-	cgmlnGRCh38	1	[Details]
Unclassified		insertion	356	120,078,181	120,581,429	740	4	ACM19S21	-	cgmlnGRCh38	1	[Details]
Likely benign		deletion	380	146,700,523	146,729,465	6,077	3	HYDM2	-	cgmlnGRCh38	1	[Details]
Benign		deletion	388	148,729,369	148,764,933	3,969	2	RNAG-IT1/PALOT48	-	cgmlnGRCh38	1	[Details]
Unclassified		deletion	487	207,535,784	207,553,039	1,528	4	CR1	-	cgmlnGRCh38	1	[Details]
Unclassified		duplication	646	38341033	38364098	23,065	0	-	-	cgmlnGRCh38	2	[Details]
Unclassified		deletion	689	22,083,623	22,398,358	3,654	4	AC096501	-	cgmlnGRCh38	2	[Details]
Unclassified		deletion	784	24,462,250	24,469,376	556	2	-	-	cgmlnGRCh38	2	[Details]
Unclassified		deletion	804	88,860,736	88,899,440	12,339	2	ACM4205:DKK4:8KJ:8KJ:8KJ:8KJ:4-DGAVL	-	cgmlnGRCh38	2	[Details]
Unclassified		insertion	848	105,353,684	105,354,776	530	0	-	-	cgmlnGRCh38	2	[Details]
Unclassified		insertion	867	105,353,684	105,354,776	530	0	-	-	cgmlnGRCh38	2	[Details]
Unclassified		deletion	868	105,880,875	105,882,852	3,499	0	NKX2	-	cgmlnGRCh38	2	[Details]
Unclassified		insertion	927	186,709,000	186,729,413	1,333	0	AC092081	-	cgmlnGRCh38	2	[Details]
Unclassified		deletion	935	188,074,348	188,075,630	741	4	-	-	cgmlnGRCh38	2	[Details]
Unclassified		deletion	938	132,243,087	132,249,680	13,270	1	POTEE-FINDM	POTEE-FINDM	cgmlnGRCh38	2	[Details]
Unclassified		deletion	954	132,272,641	132,272,671	6,152	0	-	-	cgmlnGRCh38	2	[Details]
Unclassified		deletion	952	158,802,229	158,804,984	655	2	DAPI1	-	cgmlnGRCh38	2	[Details]
Unclassified		deletion	960	368,869,488	368,936,566	53,880	0	SPC25-ABC8B	SPC25-ABC8B	cgmlnGRCh38	2	[Details]
Unclassified		duplication	961	368,872,263	368,940,833	70,570	0	SPC25-ABC8B	SPC25-ABC8B	cgmlnGRCh38	2	[Details]
Unclassified		deletion	962	132,272,641	132,272,671	6,174	0	-	-	cgmlnGRCh38	2	[Details]
Unclassified		deletion	973	155,409,529	155,405,739	543	0	-	-	cgmlnGRCh38	2	[Details]
Unclassified		deletion	976	158,802,229	158,804,984	655	2	DAPI1	-	cgmlnGRCh38	2	[Details]

Figure 6. Curated Variant list example.

- Users can remove variants from the curation list by selecting the row or rows to remove and then clicking the **Remove Records** button.
- Users can clear the entire curation list by clicking the **Remove all records** button.
- Users can filter and sort the table using multiple columns.
- Users with access to the project will be assigned as Analyst and submit the classification result to the supervisor for review by clicking the corresponding **Classification** icon.
- Administrators and project leaders will be assigned as supervisors. Supervisor accounts can reconcile the records and change the job curation status to **In Review** after clicking the **Reconcile Records** button.
- After the supervisor makes final classifications, clicking **Review Complete** button will allow for SV report with classification information to be generated. The job curation status will change to **Review Complete**.
- After the review is complete, a supervisor can download the SV report with classification information by clicking the **Generate Report** icon. In the **Summary** field, type the summary for the SV report and select the variant classification categories to be included using the checkboxes. The job curation status will change to **Published**.

Variant Classifier

The variant classifier view is an interactive view that allows users to curate and visualize one variant in the curated variant list at a time. Bionano Access provides features for the manual classification of variants per the ACMG guidelines (see **Figure 8** through **Figure 12**). These features are solely intended for Research Use Only.

1. In Bionano Access, navigate to the **Viewer** screen of the job to analyze.

2. Select **Variant Classifier** from the drop-down list.
3. **Variant Classifier** view shows up.
4. Secondly, users can double-click on a variant in the **Curated Variant** list which will also lead to the **Variant Classifier** beginning with the selected variant.



Figure 7. Variant Classifier visualization example.

- The table on the left includes information about a single variant. The rows in this table have information from the Variant calling algorithm that is also found in the **Genome** browser view and SMAP file.
- The genome map in the middle includes the cytoband, SV track, feature track, reference map and the sample specific assembled maps in the viewing region information.
- The lower panel includes navigation tools that allow a user to view a different variant, remove a variant, create images, or add to an existing list.
- Users can re-capture the SV image for the SV report by clicking the **SV Image** icon. The image will be automatically added to the SV report but can also be viewed by clicking the **Image** icon. Users can remove the SV from the curated variant list by clicking **Trash** icon.
- Users can go back to **Curated Variant List** view by clicking the **List** icon or go back to **Genome** Browser view by clicking **Link** icon.
- Users can add notes to the SV by clicking the **Edit** icon.
- In this view, the users can review the ACMG categories assigned to a variant during curation and change them if necessary. The options are **Pathogenic**, **Likely pathogenic**, **Uncertain significance**, **Likely benign**, **Benign** and **Unclassified**. The **Variant Classifier** view is specifically designed for variant curation.
- Users can reset the display of the variant by clicking the **Reset Display** icon.

- Administrators and Project leaders (Supervisor role) can see the classification made by the Analyst (user role) after they click **Classification Complete** and make a final classification.

Structural Variant Examples

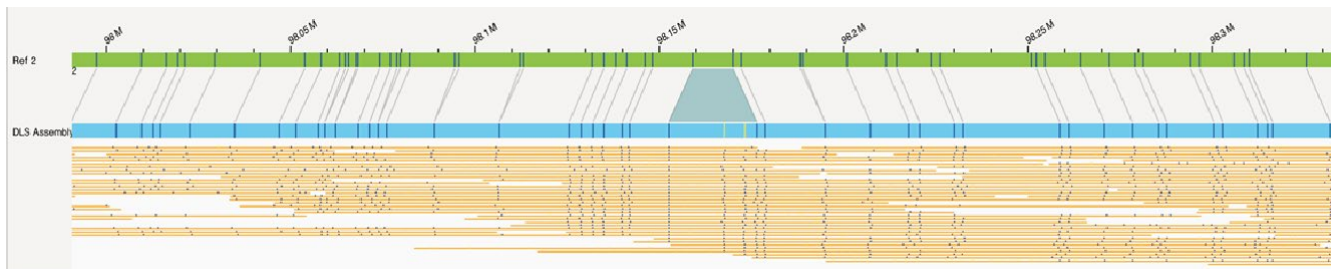


Figure 8. Insertion

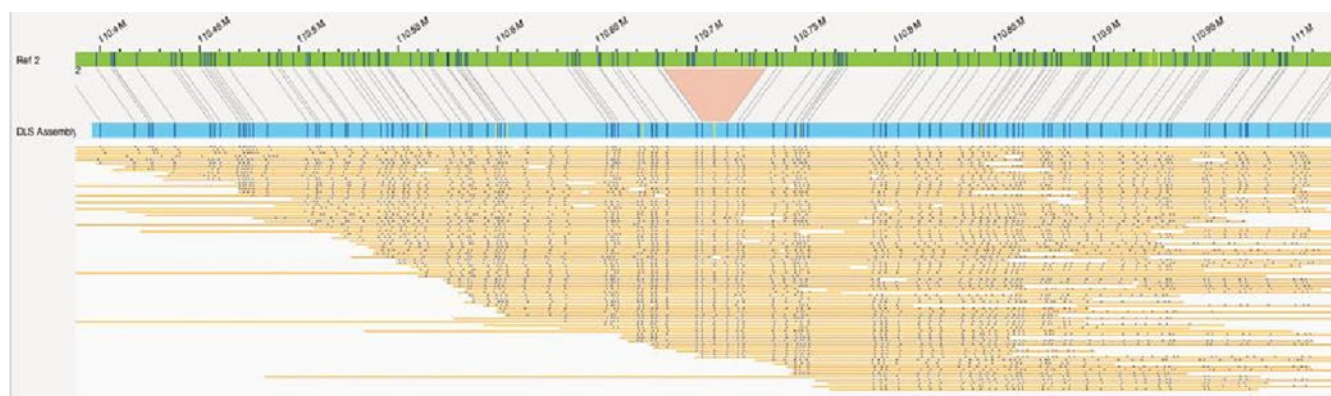


Figure 9. Deletion

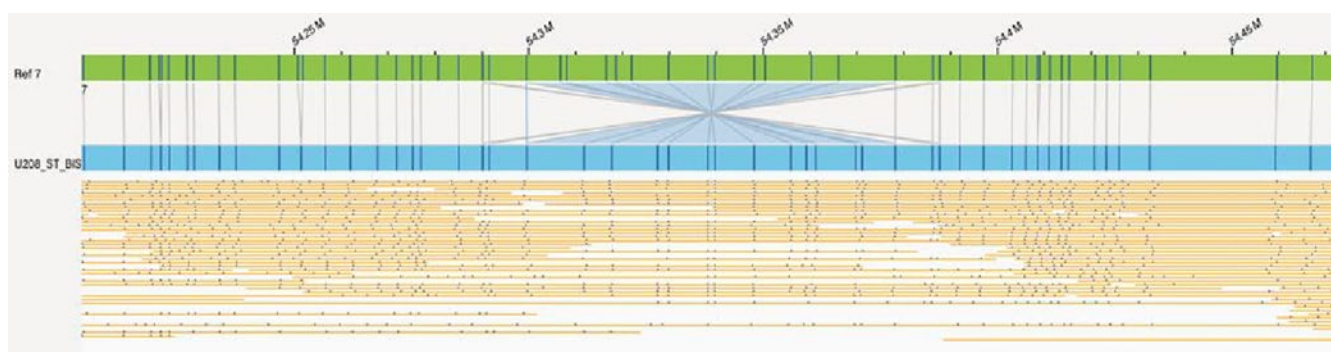


Figure 10. Inversion

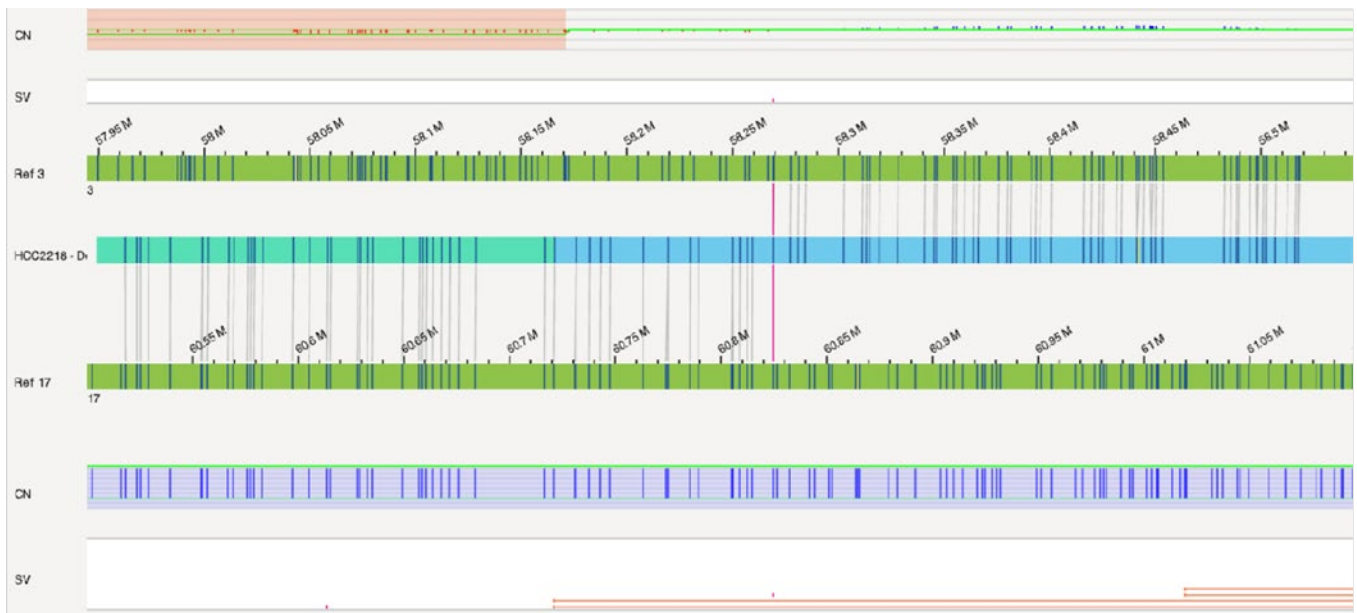


Figure 11. Translocation

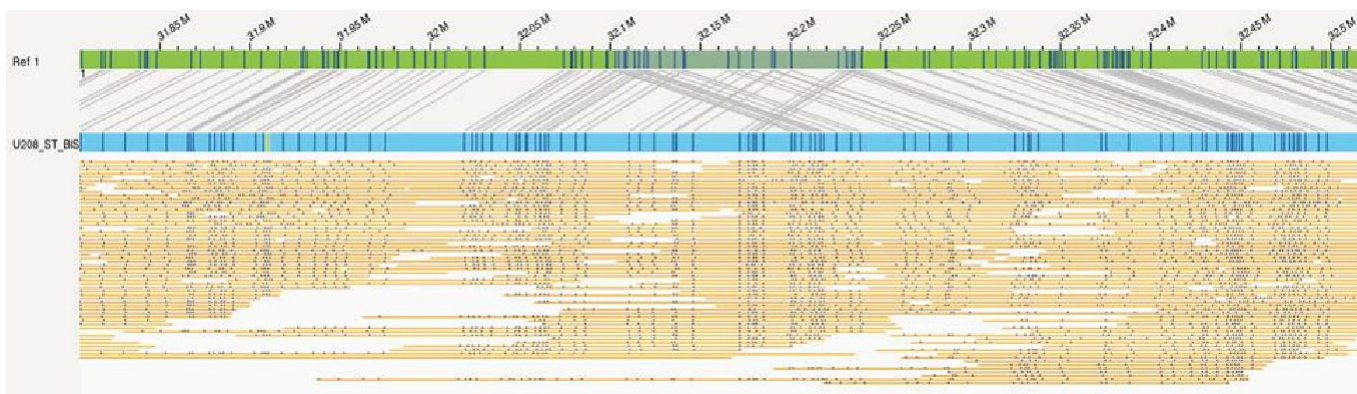


Figure 12. Duplication

FSHD VISUALIZATION

The **Genome** browser has been customized for FSHD results. It will automatically focus on the areas of interest on either chromosome 4 or 10. The consensus maps are shown in blue. The repeat region of interest, labeled as D4Z4, is in purple, and the haplotype-specific regions, labeled as either 4qA or 4qB, are highlighted. Below is an example:

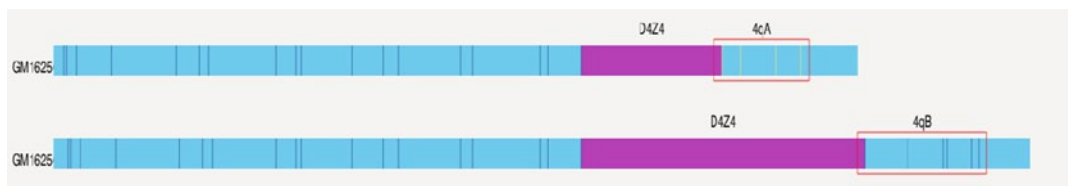


Figure 13. FSHD Visualization example

COMPLEX MULTI-PATH REGION VISUALIZATION

Bionano Access highlights Complex Multi-Path Regions (CMPR). For CMPR detail information, refer to *Bionano Solve Theory of Operation: Structural Variant Calling* (CG-30110) for more details. By default, it is shown in mint green on the assembled map, as below (**Figure 14**):

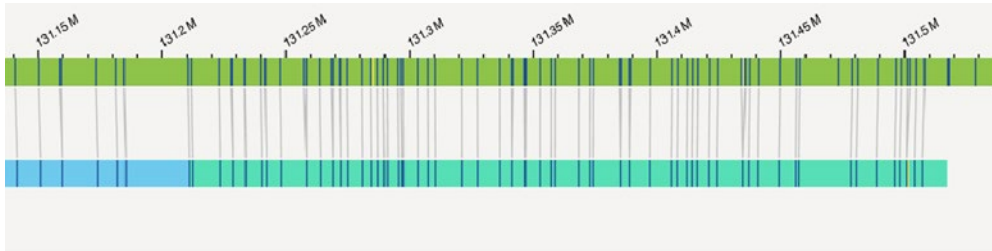


Figure 14. Complex Multi-Path Visualization example

Generate SV Report

Reporting features have been added to the **Visualization** page to allow users to share their findings with others. For a simplified workflow with default options selected:

1. In Bionano Access, navigate to the **Viewer** screen of the job to analyze.
2. Select **Circos Plot**, **Genome Browser**, **Whole Genome**, or **Ideogram** from the drop-down list.
3. Click the **Generate SV Report** icon.
4. At the **Generate PDF report** field, select one from **Filtered Variants** and **Curation Variant List**. **Filtered Variants** are structural variants based on filter criteria. **Curation Variant List** are the SVs that were added to the Curated Variant List.

For a more customizable report, see the “Curated Variant List” section.

The PDF generated will include a rendering of the **Circos plot**, **Ideogram**, and **Whole Genome** views, a screen shot of each structural variant, details on each structural variant, overlapping features, and general information about the dataset.

Download Files

Users can download filtered SV files.

1. At the **Download Files** field, select one from **Filtered Variants** and **Curation Variant List**. **Filtered Variants** are structural variants based on filter criteria. **Curation Variant List** are the SVs that were added to the **Curated Variant List**.

2. Check one or more boxes of **Copy Number Variants (CSV Format)**, **Structural Variants (SMAP Format)**, **Annotated Structural Variants (SMAP Format)**, **Aneuploidy File (.txt)**, **Informatics Report (.txt)**, and **Structural Variants (VCF Format)**.
3. Click **OK** to generate and download reports.

The files will be generated and downloaded to the system.

Refresh

Users can refresh the current view.

1. In Bionano Access, navigate to the **Viewer** screen of the job to analyze.
2. Click the **Refresh** icon.
3. Refresh the current view.

Annotate Genomic Features

The user can annotate specific genes or cytobands on the Circos Plot.

1. In Bionano Access, navigate to the **Circos Plot** screen viewer of the job to analyze.
2. Click the **Annotate Genomic Features** icon.
3. At the **Feature Name** field, type the gene name (i.e., FMR1) on the user-added feature, or the chromosome region (i.e., 4q11) for the cytoband.
4. Click **Annotate**. And the feature will be annotated on the Circos Plot.

Clear Annotations

Users can clear all annotated features on the Circos Plot.

1. In Bionano Access, navigate to the **Circos Plot** screen viewer of the job to analyze.
2. Click the **Clear Annotations** icon.
3. All annotated features will be removed.

SV Filter

The **SV Filter** icon is used to set up customized filter criteria. After setting, the applied filter criteria are displayed on the top right corner of **Circos plot** view. Filter settings will be saved on the user's profile, and each individual user can save different **SV filter** settings.

1. In Bionano Access, navigate to the **Viewer** screen of the job to analyze.
2. Click the **SV Filter** icon on the **Circos Plot** view, **Genome Browser** view or **Whole Genome CNV** view. The **Filter Settings** Window appears.
3. [Optional] From the **Filter Profile** dropdown menu select an existing profile, **Reset Filter** to reset the filter settings to the default, or **Show All** to remove all filter settings.
4. Set the desired filter settings (see below for detailed instructions).
5. [Optional] Click **Save As Filter Profile** to save the current filter settings as a new filter profile. Enter a name for the filter profile. To make the profile visible to other users select the **Shared Profile** checkbox. Click **Submit** to save the Filter Profile. To delete a filter profile, select it from the **Filter Profile** dropdown, click **Delete**, then **Ok** to confirm deletion.
6. Click **Apply Filters** and the viewer will update to show the filtered SVs.

FILTER BY SV TYPE

Users can filter SVs by SV Type, confidence score and minimum size. For more details, please refer to *Bionano Solve Theory of Operation: Structural Variant Calling (CG-30110)*.

- At the **Insertion** field, check the box to display, select one from **All, Recommended, Recommended Prior to 1.6, Recommended for 1.6 (in Access 1.7)** or customized filter from the drop-down list and type SV minimum size (bp).
- At the **Deletion** field, check the box to display, select one from **All, Recommended, Recommended Prior to 1.6, Recommended for 1.6 (in Access 1.7)** or customized filter from the drop-down list and type SV minimum size (bp).
- At the **Inversion** field, check the box to display, select one from **All, Recommended, Recommended Prior to 1.6, Recommended for 1.6 (in Access 1.7)** or customized filter from the drop-down list.
- At the **Duplication** field, check the box to display, select one from **All, Recommended, Recommended Prior to 1.6, Recommended for 1.6 (in Access 1.7)** or customized filter from the drop-down list and type SV minimum size (bp).
- At the **Intra-Translocation** field, check the box to display, select one from **All, Recommended, Recommended Prior to 1.6, Recommended Prior for 1.6 (in Access 1.7)** or customized filter from the drop-down list.
- At the **Inter-Translocation** field, check the box to display, select one from **All, Recommended, Recommended Prior to 1.6, Recommended Prior for 1.6 (in Access 1.7)** or customized filter from the drop-down list.

GENERAL SV FILTERS

Users can filter SVs by chromosome and SV Masking.

1. At the **Chromosomes to Display on Circos Plot** field, select one from **All chromosomes, Only chromosomes that have structural variants**, and **Only chromosomes from this range**. All chromosomes is the system default.
2. At the **SV Masking Filter** field, select one from **All Structural Variants, Masked Structural Variant Only** and **Non-Masked Structural Variant Only**. **Non-Masked Structural Variant Only** is recommended for human analysis if the mask bed file is selected during SV detection.
3. At the VAF filter min and VAF filter max field, type the minimum and maximum value (between 0 to 1) for the variant allele frequency to filter SVs by their variant allele frequency.

VARIANT ANNOTATION FILTERS

Users can filter SVs by percentage in Bionano control samples, chimeric score filter, self/control/parent molecule/assembly check, overlapping genes and self-molecule count. These are only available for the variant annotation pipeline. For more details, please refer to *Bionano Solve Theory of Operation: Variant Annotation Pipeline* (CG-30190).

1. At the SV in less than this % of the Bionano control samples field, type the value. 100 is the system default.
2. At the SV in less than this % of the Bionano control samples with the same enzyme field, type the value. 100 is the system default.
3. At the SV chimeric score filter field, select one from All SVs, Show Failed Chimeric Score, and Show Not Failing Chimeric Score; this is only available for variant annotation of *de novo* assembly.
4. At the SV control assembly check field, select one from All SVs, SV found in control assembly, and SV not found in control assembly; This is only available for dual variant annotation pipeline.
5. At the SV control molecule check field, select one from All SVs, SV found in control molecules, and SV not found in control molecules. It is only available for dual variant annotation pipeline.
6. At the SV parent assembly check field, select one from All SVs, SV found in both parent assemblies, SV found in father assembly, SV found in mother assembly and SV not found in parent assemblies. It is only available for trio variant annotation pipeline.
7. At the SV parent molecule check field, select one from All SVs, SV found in both parent molecules, SV found in father molecules, SV found in mother molecules and SV not found in parent molecules. It is only available for trio variant annotation pipeline.
8. At the SV self-molecule check field, select one from All SVs, SV found in self molecules, and SV not found in self molecules.
9. At the SV overlapping genes filters field, select one from All SVs, SV with overlapping genes, and SV with no overlapping genes.
10. At the Self Molecule Count field, type the value. 5 is the system default.

COPY NUMBER VARIANT FILTERS

Users can filter copy number calls by type, confidence score and minimum size. For more details, please refer to the “Introduction to Copy Number Variation” section in *Bionano Solve Theory of Operation: Structural Variant Calling* (CG-30110).

1. At the Copy Number Variant Type field, select one from All, Deletion, and Duplication.
2. At the Copy Number Variant Confidence field, select one from All, Recommended, Recommended Prior to 1.6, or Recommended for 1.6 (in Access 1.7) from the drop-down list.
3. At the Copy Number Variant Minimum Size (bp) field, type the value. 500000 is the system default.
4. At the Copy Number Variant masking filter field, select All Copy number variants, Masked Copy Number Variants Only, or Non-Masked Copy Number Variants Only to filter CNV.

ANEUPLOIDY FILTERS

1. At the Aneuploidy Type field, select All, Gain or Loss to show the aneuploidy calls.
2. At the Aneuploidy Confidence field, select one from All, Recommended, Recommended Prior to 1.6, or Recommended for 1.6 (in Access 1.7) from the drop-down list.

AOH/LOH FILTERS

1. At the AOH/LOH Minimum Size (bp) field, type the minimum size in bp to filter AOH/LOH calling region. 25,000,000 is the system default.
2. Click **Apply** to apply the filter criteria.
3. Click **Reset Filter** to reset to the default settings.
4. Click **Show All SVs** to show all the SVs.

SV Summary

Users can check SV summary (loaded by default in the **Circos Plot View**), in the **Genome Browser** viewer, **Whole Genome** viewer and **Ideogram** Viewer by clicking the **SV summary** icon. In **Genome Browser** viewer, the SV summary will display the SV summary for the current chromosome. In the **Whole Genome** viewer and in the **Ideogram** Viewer, the SV summary will display the full SV summary.

Search Genomic Features

Users can search for different features on the **Genome Browser** viewer.

1. In Bionano Access, navigate to the **Genome Browser** screen viewer of the job to analyze.
2. Click the **Search Genomic Feature** Icon.
3. Use one of the following options to search for features.

- [Optional] At the **Feature Name** field, type the feature name in the corresponding feature in the drop- down list.
 - [Optional] At the **Smap Id** field, type the Smap Id.
 - [Optional] At the **CNV Id** field, type the CNV Id.
4. Click **Search**. Access will direct to the corresponding feature.

Export to JPEG

Users can download the current view in JPEG file format.

1. In Bionano Access, navigate to the **Viewer** screen of the job to analyze.
2. Click the **Export to JPEG** icon. The dialog box appears.
3. At the **Filename** field, type the name. By default, it is **Access Viewer**.
4. Click **OK**.

View Options

The **View Options** contain different settings on **Circos Plot**, **Genome Browser** and **Whole Genome View**.

CIRCOS PLOT

1. Click the View Options icon. The dialog box appears.
2. At the Show Variants from Curation List Only option, switch on or off to display SV in curated variant list only.
3. At the Show AOH/LOH Regions option, switch on or off to display AOH/LOH calling regions.
4. At the Show CNV Mask Regions option, switch on or off to display CNV mask track.
5. At the Show SV Mask Regions option, switch on or off to display SV mask track.
6. At the Show CytoBand on Circos Chromosome option, switch on or off to display cytoband detail on the cytoband track.
7. At the Show Tooltips option, switch on or off the display of summary information textboxes when highlighting over features in the Circos Plot.
8. At the Show Alternative Transcripts option, switch on or off to display alternative transcript.
9. At the Show VAF Smooth Lines option, select Hidden, Assembly Operations Only, Rare Variant Analysis Only or Both to show VAF smooth lines on the corresponded analysis results. This setting affects all analysis results for that user account. Default is on Assembly Operations Only.
10. Click Close to apply the setting.

GENOME BROWSER

1. Click the View Options icon. The dialog box appears.
2. At the Show Variants from Curation List Only option, switch on or off to display SV in curated variant list only.
3. At the Show Unmatched Labels option, switch on or off to show unmatched labels.
4. At the Show Matched Labels option, switch on or off to show matched labels.
5. At the Grouping Highlighted Molecules option, switch on or off to enable or disable.
6. At the Auto Align Highlighted Molecules option, switch on or off to enable or disable.
7. At the Show Molecule Matchlines option, switch on or off to enable or disable.
8. At the Show Molecule Coverage option, switch on or off to enable or disable.
9. At the Show Feature Track In Overlapping Mode option, switch on or off to enable or disable.
10. At the Show Feature Name In Non-Overlapping Mode option, switch on or off to enable or disable.
11. At the Show AOH/LOH Track option, switch on or off to show the track.
12. At the Show AOH/LOH Data Points option, switch on or off to show the AOH/LOH probability data points.
13. At the Show SV Track option, switch on or off to show the track.
14. At the Show SV Query Individually option, switch on or off to enable or disable.
15. At the Show Copy Number Track option, switch on or off to enable or disable.
16. At the Show CNV Mask Regions option, switch on or off to display CNV mask track.
17. At the Show SV Mask Regions option, switch on or off to display SV mask track.
18. At the Show CytoBand Track option, switch on or off to show the cytoband.
19. At the Show Tooltips option, switch on or off to enable or disable.
20. At the Show Alternative Transcripts option, switch on or off to display alternative transcript.
21. At the Show Molecule Labels option, click the drop-down list, and select one from UnStretch, Stretch, and Stretch Matchgroup. UnStretch is the system default.
22. At the Show Match Lines in option, click the drop-down list, and select Enzyme Color.
23. Click Close to apply the setting.

WHOLE GENOME

1. Click the View Options icon. The dialog box appears.
2. At the Show Variants from Curation List Only option, switch on or off to display curated variant list only.
3. At the Show AOH/LOH Data Points option, switch on or off to show the AOH/LOH probability data points.
4. At the Show CNV Mask Regions option, switch on or off to display CNV mask track.
5. At the Show SV Mask Regions option, switch on or off to display SV mask track.
6. At the Show Tooltips option, switch on or off the display of summary information textboxes when highlighting over features in the Circos Plot.
7. At the Show Alternative Transcripts option, switch on or off to display alternative transcript.
8. At the Hide VAF Outliers option, switch on or off to hide the VAF outliers' data points.
9. At the Show VAF Smooth Lines option, select Hidden, de novo Assembly Only, Rare Variant Analysis Only or Both to show VAF smooth lines on the corresponded analysis results. This setting affects all analysis results for that user account. Default is on de novo Assembly Only.
10. Click Close to apply the setting.

View Settings

Users can customize the options settings, such as setting enzyme colors, SV colors, map (reference, genome or NGS map) colors, minimum and maximum map height value, molecule and copy number height value, and others. In Bionano Access, navigate to the Viewer screen of the project to analyze.

1. In Bionano Access, navigate to the **Viewer** screen of the project to analyze.
2. Click the **View Settings** icon. The dialog box appears.
3. At the **Minimum map height (6-20 px)** field, type the value. By default, the value is **10**.
4. At the **Maximum map height (20-100 px)** field, type the value. By default, the value is **40**.
5. At the **Molecule height (1-40 px)** field, type the value. By default, the value is **2**.
6. At the **Copy number height (30-150 px)** field, type the value. By default, the value is **40**.
7. At the **SV track height (30-250 px)** field, type the value. By default, the value is **60**.
8. At the **AOH/LOH row height (50-400 px)** field, type the value. By default, the value is **150**.
9. At the **Non-overlapping feature row height (30-400px)** field, type the value. By default, the value is **150**.

10. At the **Copy number zoom threshold (0-400)** field, type the value. By default, the value is **5**.
11. At the **Max copy number to display (0 - 100)** field, type the value. By default, the value is **8**.
12. At the **Track name width (80 px)** field, type the value. By default, the value is **80**.
13. At the **Gap between molecules (0-40 px)** field, type the value. By default, the value is **2**.
14. At the **SV height (2-10 px)** field, type the value. By default, the value is **2**.
15. At the **Minimum gap between rows (10-40 px)** field, type the value. By default, the value is **10**.
16. At the **Maximum gap between rows (40-200 px)** field, type the value. By default, the value is **70**.
17. At the **Font size (%)** field, type the value. By default, the value is **100**.
18. At the **BED Font size (%)** field, type the value. By default, the value is **80**.
19. At the **BED color opacity (%)** field, type the value. By default, the value is **60**.
20. At the **SV color opacity (%)** field, type the value. By default, the value is **40**.
21. At the **Raw copy number opacity (%)** field, type the value. By default, the value is **100**.
22. At the **Ruler text rotation (degree)** field, type the value. By default, the value is **30**.
23. At the **Hybrid cut flashing duration (msec)** field, type the value. By default, the value is **5000**.
24. At the **Circos plot SV size** field, type the value. By default, the value is **0.6**.
25. At the **Circos plot translocation width** field, type the value. By default, the value is **0.5**.
26. At the **Hybrid cut color** field, select the color from the palette.
27. At the **Background color** field, select the color from the palette.
28. At the **Highlight row color** field, select the color from the palette.
29. At the **Selected map border color** field, select the color from the palette.
30. At the **Selected label color** field, select the color from the palette.
31. At the **Molecule panel color** field, select the color from the palette.
32. At the **Molecule color** field, select the color from the palette.
33. At the **Reference map color** field, select the color from the palette.
34. At the **Genome map color** field, select the color from the palette.
35. At the **NGS map color** field, select the color from the palette.
36. At the **Longest transcript color** field, select the color from the palette.

37. At the **Alternative transcript color** field, select the color from the palette.
38. At the **Matchline color** field, select the color from the palette.
39. At the **Label coverage color** field, select the color from the palette.
40. At the **Lasso color** field, select the color from the palette.
41. At the **Highlight molecule color** field, select the color from the palette.
42. At the **Highlight label/matchline color** field, select the color from the palette.
43. At the **Highlight matchgroup color** field, select the color from the palette.
44. At the **Highlight molecule matchline color** field, select the color from the palette.
45. At the **Track background color** field, select the color from the palette.
46. At the **SV insertion color** field, select the color from the palette.
47. At the **SV deletion/CNV loss color** field, select the color from the palette.
48. At the **SV inversion color** field, select the color from the palette.
49. At the **SV translocation color** field, select the color from the palette.
50. At the **SV duplication/CNV gain color** field, select the color from the palette.
51. At the **SV end color** field, select the color from the palette.
52. At the **AOH/LOH color** field, select the color from the palette.
53. At the **AOH/LOH region color** field, select the color from the palette.
54. At the **VAF color** field, select the color from the palette.
55. At the **VAF Segment color** field, select the color from the palette.
56. At the **CMPR color** field, select the color from the palette.
57. At the **Copy number color** field, select the color from the palette.
58. At the **High copy number color** field, select the color from the palette.
59. At the **Low copy number color** field, select the color from the palette.
60. At the **Nt.BspQI enzyme** field, select the color of matched label from the first palette, and unmatched from the second one.
61. At the **Nb.BbvCI enzyme** field, select the color of matched label from the first palette, and unmatched from the second one.

62. At the **Nb.BsmI enzyme** field, select the color of matched label from the first palette, and unmatched from the second one.
63. At the **Nb.BsrDI enzyme** field, select the color of matched label from the first palette, and unmatched from the second one.
64. At the **Nb.BssSI enzyme** field, select the color of matched label from the first palette, and unmatched from the second one.
65. At the **DLE-1 enzyme** field, select the color of matched label from the first palette, and unmatched from the second one. There may be more enzymes if other enzymes are added into the system.
66. Click **Save**. The new settings are saved.
67. [Optional] To revert to the original settings, click **Reset Options**.

Return to Project Browser

1. In Bionano Access, navigate to the **Viewer** screen of the job to analyze.
2. Click the **Return to Project Browser** icon.

Home Page

1. In Bionano Access, navigate to the **Viewer** screen of the job to analyze.
2. Click the **Home Page** icon.

Key and Mouse Shortcuts

1. In Bionano Access, navigate to the **Viewer** screen of the project to analyze when viewing results.
2. Click the **Key & Mouse Shortcuts** icon. The dialog box appears.
3. Here is the list of highlighted features and descriptions. More information is available in Access (**Table 23**).

Table 23. Key and Mouse Shortcuts

Feature	Description
Customize the view of the samples	In the first ruler, press down on the mouse, and then drag left or right to highlight the samples to view. A red box indicates the selected samples that are displayed on the viewer.
Move maps horizontally	Left click on mouse; move left or right.
Move maps vertically	Press Shift; move up or down.

Map options

Right-click on **map**.

- **Hide** - Hide the selected map from the viewer.
- **Hide the others** - Hide the other genome maps from the viewer except for the selected map and reference map.
- **Collapse** - Collapse the genome maps to a single row next to the reference map.
- **Show Ruler** - Attach the ruler to the selected genome map.
- **Show Molecule** - Show all the aligned molecules that are related to the selected genome map. If there are no molecule alignments associated with a map in the viewer the 'Show Molecules' option in the right click menu will no longer be available.
- **Show Molecule for** - Show all the aligned molecules that are related to Proband, Mother and Father for variant annotation results.
- **Hide Matchgroups** - Remove the gray line connecting to the selected genome map from the reference map.
- **Invert** - Invert the orientation of the selected map.

Zoom in or out

Use the mouse to scroll zoom in or zoom out.

SV Tab

1. In Bionano Access, navigate to the **Viewer** screen of the project to analyze.
2. Select **Genome Browser** from the drop-down list at upper left corner of the Viewer.
3. Click the **SV** Tab. This tab is only available for non-annotated jobs: *de novo* assembly and rare variant analysis. This tab includes **Sample, Global_ID, Smap_ID, Type, Zygotity, Confidence, Ref1_ID, Ref2_ID, Ref_Start, Ref_End, Size, Qry_ID, Qry_Start, and Qry_End**. The columns can be sorted. Please refer to the *OGM File Format Specification Sheet* (CG-00045) for more details.

SV Annotation Tab

1. In Bionano Access, navigate to the **Viewer** screen of the project to analyze.
2. Select **Genome Browser** from the drop-down list at upper left corner of the Viewer.
3. Click the **SV Annotation** Tab. This tab is only available for annotated jobs: annotated *de novo* assembly, annotated rare variant analysis, and variant annotation. This tab includes **Type, RefcontigID1, RefcontigID2, RefStartPos, RefEndPos, Size, Zygotity, Confidence Score, SmapId, In_AOH/LOH, AOH/LOH, Present in % of control db samples, Present in % of control db samples with the same enzyme, Fail assembly chimeric score, Overlap Genes, Nearest Non-overlap Gene, Nearest Non-overlap Gene Distance, Putative Gene Fusion, Self Molecule Count, Number Overlap DGV Calls, additional sorting options against the Bionano control database, ISCN, and Found in self molecules**. The columns can be sorted. Please refer to *OGM File Format Specification Sheet* for more details.

Adding SV to Curated Variant List/SV Report

CIRCOS PLOT

Users can add SVs to the **Curated Variant List** for variant classification purposes.

1. In Bionano Access, navigate to the **Circos Plot** screen viewer of the job to analyze.
2. Click the **Add to Curated List** icon.
3. All current remaining SVs will be added to the **Curated Variant** list.

GENOME BROWSER

Users can select an SV of interest to the **Curated Variant** list for Variant Classifier, Generate SV report, or Download Files.

1. In Bionano Access, navigate to the **Viewer** screen of the job.
2. Select **Genome Browser** from the drop-down list at the upper left corner of the **Viewer** screen.
3. To add an SV, click **Add to Report** in the **SV** or **SV Annotation** tab.
4. Select **Curated Variant List** or **Variant Classifier** from the drop-down list at the upper left corner of the Viewer screen. All added SVs will be shown here.

Match Tab

1. In Bionano Access, navigate to the **Viewer** screen of the project to analyze.
2. Select **Genome Browser** from the drop-down list of **Circos Plot**.
3. Click the **Match** Tab. This tab includes **XmapId**, **RefId**, **QryId**, **RefStartPos (bp)**, **RefEndPos (bp)**, **QryStartPos (bp)**, **QryEndPos (bp)**, **Orientation**, **Confidence**. The columns can be sorted and filtered. Please refer to *OGM File Format Specification Sheet* for more details.

Copy Number Tab

1. In Bionano Access, navigate to the **Viewer** screen of the project to analyze.
2. Select **Genome Browser** or **Whole Genome CNV** from the drop-down list.
3. Click the **Copy Number** tab. It contains columns of **Id**, **RefId**, **Start (bp)**, **End (bp)**, **Size (bp)**, **Type**, **Fractional Copy Number**, **Copy Number**, **Confidence**, **VAF**, **Algorith**, **Overlap Genes**, **Nearest NonOverlap Genes**, **Nearest NonOverlap Gene Distance**, **Num Overlap DGV Calls**, and **ISCN**.

The columns can be sorted. Refer to the *Bionano Solve Theory of Operation: Structural Variant Calling* (CG-30110) for more details.

Aneuploid Tab (For Whole Genome View)

1. In Bionano Access, navigate to the **Viewer** screen of the project to analyze.
2. Select **Genome Browser** or **Whole Genome CNV** from the drop-down list of **Circos Plot**.
3. Click the **Aneuploid** tab. It contains per-chromosome aneuploidy call and includes columns of **RefId**, **Type**, **% Chr Coverage**, **Score**, and **Fraction CN State**. The columns can be sorted. It is only available for Access 1.4 or higher. Please refer to *Bionano Solve Theory of Operation: Structural Variant Calling* (CG-30110) for more details.

Summary Tab (For Whole Genome View)

1. In Bionano Access, navigate to the **Viewer** screen of the project to analyze.
2. Select **Whole Genome CNV** from the drop-down list.
3. Click the **Copy Number** Tab. It contains columns of **RefId**, **% Deletion** and **% Duplication**. The columns can be sorted. Refer to *Bionano Solve Theory of Operation: Structural Variant Calling* (CG-30110) for more details.

Repeat Tab (For EnFocus™ FSHD analysis and EnFocus™ Fragile X analysis)

1. In Bionano Access, navigate to **View EnFocus™ FSHD Analysis** of the project to analyze.
2. Click the **Repeat** Tab, which is generated only for human FSHD samples and includes columns of **Qry Id**, **Repeat Count (units)**, **Haplotype**, **Repeat spanning coverage (X)**. The columns can be sorted and filtered. Please refer to *Theory of Operation EnFocus™ FSHD Analysis* for more details.

Conflict Resolutions Tab (For Hybrid Scaffold analysis)

1. In Bionano Access, navigate to **Maps to NGS with Conflicts** of the hybrid scaffold job in any project to analyze.
2. Click the **Conflict Resolutions** Tab. It is generated from hybrid scaffold jobs and includes columns of **RefId**, **Enzyme**, **RefLeftBkpt**, **RefRightBkpt**, **RefLeftBkptToCut**, **RefRightBkptToCut**, **RefToDiscard**, **QryId**, **QryLeftBkpt**, **QryRightBkpt**, **QryLeftBkptToCut**, **QryRightBkptToCut**, **QryToDiscard**. The columns can be sorted. Please refer to *OGM File Format Specification Sheet* for more details.

Settings



Figure 15. Bionano Access settings

In silico Digestion

NOTE: Users must have Administrator or Project Lead privileges to see this setting.

In silico digestion is used to create a genome reference consensus map for any sequence file. This tool uses a FASTA file and the recognition sequence of a labeling enzyme to create a CMAP file that can be used as a reference. The *in silico* digestion results are accessible to all users. Bionano Access saves the FASTA files that are uploaded; users can reuse the files to generate different CMAP files with different enzymes or enzyme-to-channel combinations.

The ***in silico* Digestion** page contains an accordion menu that contains three panes: **Create New Run, Runs in Progress, Completed Runs**. Click the arrow on the pane to expand or collapse the view.

CONFIGURATION SETTINGS

Before performing *in silico* digestion, define the following settings:

1. From the **Bionano Access** main menu, select **Settings**.
2. Select ***in silico* Digestion**.
3. Click the **Configuration Settings** icon in the top right corner. The **Configuration** dialog box appears.
4. At the **Minimum Labels** field, set a value between 0–20 (default as **0**).
5. At the **Minimum Length** field, set a value between 0–250 kbp (default as **0**).
6. At the **Number of Channels** field, set the number of channels to digest. The channel refers to the laser color used.
7. At the **Number of Enzymes per Channel** field, set the number of enzymes per channel to digest.
8. Click **Save**. The software displays the settings in the **Create New Run** pane.

ENZYME MANAGEMENT

Users with administrator privileges can add and edit labeling enzymes and recognition sequences; however, the default enzymes listed in Bionano Access are not editable (see **Table 24**).

1. From the **Bionano Access** main menu, select **Settings**.
2. Select ***in silico* Digestion**.
3. Click the **Enzyme Management** icon in the top right corner. The **Manage Enzymes** dialog box appears:

Table 24. Manage Enzymes dialog box

Info	Description
Name	The enzyme name.
Recognition Sequence	The recognition sequence.
Type	Bionano or user identified.
Matched Label Color	The color.
Unmatched Label Color	The color.

Experiment count	The count of experiments using this enzyme.
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4. Click **Add**. The **Add New Enzyme** dialog box appears.
5. At the **Enzyme Name** field, type the enzyme name.
6. At the **Enzyme Sequence** field, type the recognition sequence of the labeling enzyme (ACGT). The maximum length for the enzyme sequence is twenty nucleotides.
7. At the **Matched Label Color** field, select the color from the palette.
8. At the **Unmatched Label Color** field, select the color from the palette.
9. Click **Save**. The software adds the enzyme to the list.
10. Select one enzyme with the **Type as User** from the list. Then click **Edit**. The **Edit Existing Enzyme** dialog box appears.
11. At the **Enzyme Name** field, type the enzyme name.
12. At the **Enzyme Sequence** field, type the recognition sequence of the labeling enzyme (ACGT). The maximum length for the enzyme sequence is twenty nucleotides.
13. At the **Matched Label Color** field, select the color from the palette.
14. At the **Unmatched Label Color** field, select the color from the palette.
15. Click **Save Changes**.
16. Select one enzyme with the **Type as User** from the list. Then click **Delete**. The **Delete Existing Enzyme** dialog box appears.
17. Click **Yes** to delete this enzyme.

FASTA MANAGEMENT

Users with administrator privileges can use *in silico* Fasta Files Management to manage FASTA files that have been uploaded into the *in silico* Digestion tool (see **Table 25**).

1. From **Bionano Access** main menu, select **Settings**.
2. Select ***in silico* Digestion**.
3. Click the **Fasta Management** icon in the top right corner. The **Fasta Files** list and ***in silico* Digestion Runs** for every fasta file appear.

Table 25. Fasta Files list and *in silico* Digestion Runs

Information	Description
Run Details	System-generated number for the run.
Date	The date the file is uploaded.
Minimum Labels	The user-defined setting for the minimum number of labels per map.
Minimum Length	The user-defined setting for the minimum map length.
Enzyme(s) Channel 1	The enzyme(s) that are used in Channel 1.
Enzyme(s) Channel 2	The enzyme(s) that are used in Channel 2.
Enzyme(s) Channel 3	The enzyme(s) that are used in Channel 3.
Action	To Delete the run.

4. Click **Delete Fasta** to delete the selected Fasta. The **Confirm** dialog box appears. Click **Yes** or **Cancel**.
5. Click **Delete All Runs** to delete all runs of the selected Fasta. The **Confirm** dialog box appears. Click **Yes** or **Cancel**.

CREATE NEW RUN

Depending on the digestion tool settings, users may have one or more label channel parameters to select. Access saves the FASTA files that have been digested and only checks for duplicate file names. If a user uploads a FASTA file that Access has already saved, the software will point to the existing file. For best practice, give FASTA files unique names to easily track them.

The following task is an example of an *in silico* digestion for a one label channel.

1. From the **Bionano Access** main menu, select **Settings**.
2. Select ***in silico* Digestion**.
3. In the **Create New Run** pane, at the **Select file** field, browse to the FASTA file to digest, and then click **Open**.
4. At Label Channel 1, select the enzyme from the drop-down list.
5. [Optional] At the Label Channel 2, select the enzyme from the drop-down list.
6. Click **Launch**.

A message appears to indicate that the file is uploading if it is new, and then the software initializes the run. The digestion progress appears in the **Runs in Progress** pane. Once the run is complete, the digested data appears in the **Completed Runs** pane.

If this FASTA file was previously uploaded to the server, this message appears: *“This FASTA has been uploaded to the server. To use it, click **OK**. To use a new version of this FASTA, click **Cancel**, rename the FASTA, upload, and try again.”*

RUNS IN PROGRESS

Expand the **Runs in Progress** pane to view the following statistics (**Table 26**):

Table 26. Runs in Progress pane.

Statistic	Description
Run	System-generated number for the run.
Date	The date the file is uploaded.
FASTA	The FASTA file name.
Min Labels	The user-defined setting for the minimum number of labels per map.
Min Size	The user-defined setting for the minimum map length.
Enzyme(s)	The enzyme(s) that are used.
Progress	The progress of the run.
Cancel	Click x to cancel the file upload.

COMPLETED RUNS

Expand the **Completed Runs** pane to view the following statistics (**Table 27**):

Table 27. Completed Runs pane.

Statistic	Description
Run ID	System-generated number for the run.
Date	The date the CMAP is generated.
Files FASTA	The FASTA file name.
Files Cmap	The CMAP file that can be downloaded.
Files Key	A file to track CMAP IDs that are associated with the FASTA sequence map names.
Files Gap	The GAP file can be downloaded. The GAP file track N-base gaps in sequence.
Files Summary	A complete summary of the results that can be downloaded.
Enzyme(s)	Enzyme(s) that are used to create the CMAP.
#Maps	Number of maps in the digested sample.
N %	Percent of genome that has N bases.
Sites / 100 kbp Ch1 Sites	The number of labels per 100 kbp in Channel 1.
Action "Add to References"	Clicking on this will automatically copy the newly digested CMAP file to the reference pool. Users do not need to manually download the cmap file and import to Reference pool.

Banner

Users must have Administrator privileges to see this setting.

Users can add login banner to the **login** page of Bionano web Access or add download message that users must acknowledge before files can be downloaded. Users must have administrator privileges to perform this task.

1. From the **Bionano Access** main menu, select **Settings**.
2. Select **Banner**.

3. At the **Login Banner or Download Banner** field, type and edit the information.
4. Click **Submit**.

System Account Settings

At the **Account Login** field, user can edit the following options:

- **Idle User Session Expiration [minutes]** – Bionano Access will automatic logout when no action was performed in a period time. The default is 30 minutes.
- **Maximum User Concurrent Sessions** – The maximum number of sessions a user can open. The default is five.
- **Maximum Login Attempts** – The Maximum login attempts a user can enter before Access locks the account. Default is three times.

At the **Password Retention** field, user can edit the following options:

- **Password Retention Checks** – Enable/Disable password retention check. Default is **On**.
- **Password Expiration [days]** -- Bionano Access will ask user to change password after setting time. The default is **90** days.
- **Previous Password Retention [month]** – How long does Access save old password to prevent the password can be re-used. The default is three months.

At the **Password Settings** field, user can edit the following options:

- **Minimum Characters** – minimum characters required in password. Default is one.
- **Minimum Numerals** – minimum number required in password. The default is zero.
- **Minimum Special Characters** – minimum special characters required in password. The default is zero.
- **Minimum Case Changes** – minimum case changes required in password. The default is zero.
- **Maximum number of identical characters in a row** - maximum number of identical characters in a row allowed in password. The default is two.

At the **Second Factor Authentication** field, user can edit the following options:

- **Check authentication code** - Enable/Disable second factor check authentication code which received by user account email.

User Accounts

- Users must have Administrator privileges to see this setting.
- All user accounts are listed in the **User Accounts** screen, and the user list can be exported by clicking **Export to Excel**.
- Users can **Edit**, **Delete** or **Lock/Unlock** account in the list.

NEW USER

All users should have their own user account with a valid email address. Bionano Access notifies users via email when their job is complete. Do not use shared accounts.

1. From the **Bionano Access** main menu, select **Settings**.
2. Select **User Accounts**.
3. Click **New User**. **Create User** window appears.
4. At the **Username** field, type the account log in username.
5. At the **Full Name** field, type the full name to display in Access.
6. At the **Email Address** field, type the email address. This email address is used to receive Access emails.
7. At the **Password** field, type the password.
8. At the **Confirm Password** field, type the password again to confirm.
9. At the **Role** field, choose one of the following:
 - **Administrator**
 - **Project Lead**
 - **User**
 - **Read-only**
10. At the **User Status** field, chose one of the following:
 - **Active**: The user account is active; the user can log on to Bionano Access.
 - **Disable**: The user account is disabled; the user cannot log on to Bionano Access.
11. At the **Second factor authentication** field, enable/disable second factor authentication when login.
12. Click **Submit**. The new user account appears on the **User Accounts** screen.

Control Database

Users must have Administrator or Project Lead privileges to perform this task. For Control Database format, please go to *Bionano Solve Theory of operation: Variant Annotation Pipeline* (CG-30190) for more detail.

1. From the **Bionano Access** main menu, select **Settings**.
2. Select **Control Database**.
3. Click **Add Control Database**. **Add Control Database** window appears.
4. At the **Control Database Name** field, type the name.

5. At the **Genome Build** field, select the genome build from the list.
6. At the **Control Type** field, select one from **SV**, **CNV** or **ECNB**.
 - **SV** Control database is used for SV calling.
 - **CNV** Control Database is used for CNV calling.
 - **ECNB (Expected Copy Number Baseline)** is used for CNV calling to set the baseline of copy number.
7. At the **Operation Type** field, select from **Assembly** or **RareVariant**, which is only applicable if **SV** is selected for **Control Type**.
8. At the **This is a human control database** field, check the box if it is a human control database.
9. [Optional] At the **Description** field, type a brief description.
10. At the **Control Database File (*.txt)** field, click **Choose File**, and then browse to select the file (*.bed) to use.
11. Click **Open**.
12. Click **OK**.

System Status

Administrators can check the Bionano Access Server (BAS) or customer's own system status by viewing the System Status. It includes **Uptime**, **System Version**, **Disk Usage**, **Memory statistics**, **SSH status**, **I/O Utilization**, **CPU information** and **Processes Running**.

* This feature will not be available when `"BAS":false` is indicated in Access configuration file.

References

Users must have Administrator or Project Lead privileges to perform this task.

Users can **Download** system reference, and **Download**, **Delete** or **Edit** non-system reference.

Users can add references (CMAP files) to use for map alignments.

1. From the **Bionano Access** main menu, select **Settings**.
2. Select **References**. The **Reference List** page appears.
3. Click **Add Reference**. A dialog box appears.
4. At the **Reference Name** field, type the name for the reference.

5. At the **Enzyme** field, select the enzyme from a list of enzymes already known in the system. Users can also click the **Add Enzyme** button to add a new enzyme if needed. More than one enzyme in the list can be selected if needed.
6. At the **Genome Build** field, type the name of the genome.
7. If the reference is human, select the **This is a human reference** checkbox. If this is a human reference, **Genome Build** can only be hg19, hg38 or t2t.
8. At the **Reference File (.cmap)** field, click **Choose File**, and then browse to select the CMAP file to use.
9. Click **Open**.
10. Click **OK**.
11. Select one reference to **Download File, Delete** or **Edit**.

Configurations

Users must have Administrator or Project Lead privileges to perform this task.

When generating certain jobs, users are required to select a configuration that contains a set of pre-defined parameters. Users can select a configuration from:

- A list of default configurations in Bionano Access
- A list of customized configurations based on the existing configurations in Bionano Access
- Configuration files (*.xml) that users upload to Bionano Access

Bionano Access does not validate the formatting or parameter settings of the upload XML file. If the uploaded file does not contain proper parameters or formatting, the file may cause the system to crash. Users can also download the configuration files in the system.

For details on customizing configurations based on existing and validated configurations in Bionano Access, see the following tasks:

- Generate *de novo* Assembly
 - Generate rare variant analysis.
 - Generate Hybrid Scaffold
1. From the **Bionano Access** main menu, select **Settings**.
 2. Select **Configurations**.
 3. Click **Add Configuration**. A dialog box appears.
 4. At the **Configuration Name** field, type the name for the configuration.

5. At the **Configuration Type** field, select the operation type to use this configuration file from the drop-down list.
6. [Optional] At the **Description** field, type a brief description.
7. At the **Configuration File** field, click **Choose File**, and then browse to the configuration file (*.xml) to use.
8. Click **Open**.
9. Click **OK**.
10. Select one configuration file to **Download File, Delete** or **Edit**.

System Warning

Users must have Administrator privileges to post a warning message to all other users.

1. From the **Bionano Access** main menu, select **Settings**.
2. Select **System Warning**.
3. At the **System Warning** field, type the information.
4. Click **Submit**.

System Features

Administrator, Project lead or User privileges to perform this task. Users can add or select features to automatically display on jobs if the Genome Build matches the sample being viewed. Access can accept BED and GTF files.

For BED file format, please go to *OGM File Format Specification Sheet* (CG-00045) for more details.

ADD NEW FEATURES

1. From the **Bionano Access** main menu, select **Settings**.
2. Select **System Features**.
3. Click **Add**. A dialog box appears.
4. At the **Name** field, type the name for the bed.
5. [Optional] At the **Description** field, type a brief description.
6. At the **Feature Type** field, select one from **Generic, Mask, CNVMask** or **Genes**.
 - **Generic** bed files are used for SV and CNV Filter in *de novo* assembly, Rare Variant Analysis and Variant Annotation jobs.

- **Mask** bed files are used for masking putative false positive translocation breakpoint calls, reference nbase gaps (for insertions and deletions) or complicated regions, such as segmental duplications during *de novo* assembly or Rare Variant Analysis.
 - **CNV Mask** bed files are only used for masking coverage variable regions during copy number analysis.
 - **Genes** bed files are used to annotate SVs during Variant Annotation Pipeline job.
7. At the **Genome Build** field, select the genome build from the list.
 8. At the **File (*.bed, *.gtf)** field, click **Choose File**, and then browse to select the file to use.
 9. Click **Open**.
 10. Click **OK**.

AUTO DISPLAY

1. From the **Bionano Access** main menu, select **Settings**.
2. Select **System Features**.
3. Check the **Auto Display** option, system will Auto Display the feature file if the Genome Build matches the sample being viewed. Locks are user specific not system wide.

Queue Status

Users must have Administrator privileges to perform this task.

The **Queue Status** page will list all jobs currently being managed by SGE. If there is more than one SGE cluster in the Access configuration, the **Queue Status** page will list the status of each SGE cluster. This feature offers a quick way to better understand the compute load on systems.

Named Filters

Users must have Administrator or Project Lead privileges to perform this task.

1. From the **Bionano Access** main menu, select **Settings**.
2. Select **Named Filters**.
3. Select **Add Filter**. The **Create Filter** window appears.
4. At the **Filter Name** field, type the name.
5. At the **Insertion Confidence**, type the confidence score cutoff for insertion calls.
6. At the **Deletion Confidence**, type the confidence score cutoff for deletion calls.
7. At the **Inversion Confidence**, type the confidence score cutoff for inversion breakpoint calls.

8. At the **Duplication Confidence**, type the confidence score cutoff for duplication calls.
9. At the **Intra-Translocation Value**, type the confidence score cutoff for intra-translocation breakpoint calls.
10. At the **Inter-Translocation Value**, type the confidence score cutoff for inter-translocation breakpoint calls.
11. At the **Copy Number Value**, type the confidence score cutoff for copy number calls.

Archive Jobs

Users must have Administrator privileges to perform this task.

This operation allows you offload results to enterprise storage so that you can reclaim disk space for additional analysis jobs and or to declutter your projects if needed. You can later restore these jobs if needed. The system will ensure that the restored files match the original files. It is also possible to extend the storage capacity of your Bionano Access server with enterprise storage. Contact support for details on how to mount enterprise storage for job archival or additional storage.

IMPORTANT NOTE:

To reclaim disk space on the Bionano Access Server when jobs are archived it is expected that users are archiving files to a folder location that is in external enterprise storage. Ideally this folder location maps to storage that has been mounted on the Bionano Access Server. Mounting external storage requires some in-depth technical knowledge of networking and Linux. We recommend that you contact Bionano Support prior to mounting any storage. Be aware that support and monitoring of external storage systems is outside the scope of Bionano.

1. From the **Bionano Access** main menu, select **Settings**.
2. Select **Archive Jobs**.
3. From the **Set Archive Folder** dropdown, select either:
 - a. An existing archive folder or,
 - b. **New Archive Folder** to set up a new archive path.
4. Select the checkbox next to the jobs you want to archive.
5. When you are ready, select **Submit Archive**.
6. A dialog will pop up, confirm the number of jobs to archive and select **Yes**.

7. Wait until the archive completes without refreshing the page.

Restore Archived Jobs

Users must have Administrator privileges to perform this task.

1. From the **Bionano Access** main menu, select **Settings**.
2. Select **Restore Archived Jobs**.
3. From the **Set Archive Folder** dropdown, select the path containing the archives.
4. Select the checkbox next to the job archives you want to restore.
5. When you are ready, select **Restore Archive**.
6. A dialog will pop up, confirm the number of job archives to restore and select **Yes**.
7. Wait until the restoration completes without refreshing the page.

System Services Settings

Users must have Administrator privileges to perform this task.

1. From the **Bionano Access** main menu, select **Settings**.
2. Select **System Services Settings**.
3. At the **System Level Services** field, please find the following options:
4. At the **Compute On Demand & Bionano Assure** field, opt in or out **Compute On Demand** and **Bionano Assure**.

The Compute On Demand service provides pay per use elastic computing resources to supplement and accelerate analysis needs. Together with the Bionano Assure service it is possible to monitor the health of the entire workflow. This is the recommended option. Internet access is required to connect to the Bionano Compute on Demand solution. Test the connection by clicking the **Test Connection** button after enabling the service.

5. At the **Bionano Assure Only** field, opt in or out **Bionano Assure**. Use this option to ensure optimal performance of the instrument but are not interested in the Compute On Demand service.
6. At the **VIA Server** field, **Edit** or **Test** the VIA connection, see the “VIA” section for more details.
7. At the **Maintenance Mode** field, toggle maintenance mode.

Toggle **Maintenance** mode to prevent non-administrator level users from accessing the system during maintenance windows. In **Maintenance** mode, only users with the administrator role can log

into the system. This allows administrators to check system operations after upgrades before opening the system to general use.

8. At the **Remote Access** field, opt in or out **Remote Access**.

This option is only available in Bionano Access software, which is installed in a Bionano Access Server with HTTPS (Hypertext Transfer Protocol Secure, the encrypted version of HTTP) enabled. Please refer to *How to enable HTTPS in Bionano Access (CG-30377)* for more details.

By default, this option is off (opt-out). Once opt-in, it allows Bionano support personnel to directly access Bionano Access Server for remote support and troubleshooting.

VIA Software Settings

Users with the Administrator role can create custom VIA sync settings. Bionano Access is preconfigured with default settings for common operations, which users can select from when launching a job or uploading an existing job. For instructions on how to create or edit VIA sync settings, see the “VIA Software” section.

Compute On Demand

Enable Compute On Demand

To enable **Bionano Compute on Demand** within Bionano Access, opt in to **Compute On Demand** in **System** settings (please see above for more details). The **Bionano Compute On Demand Activation** window will pop up (**Figure 16**).

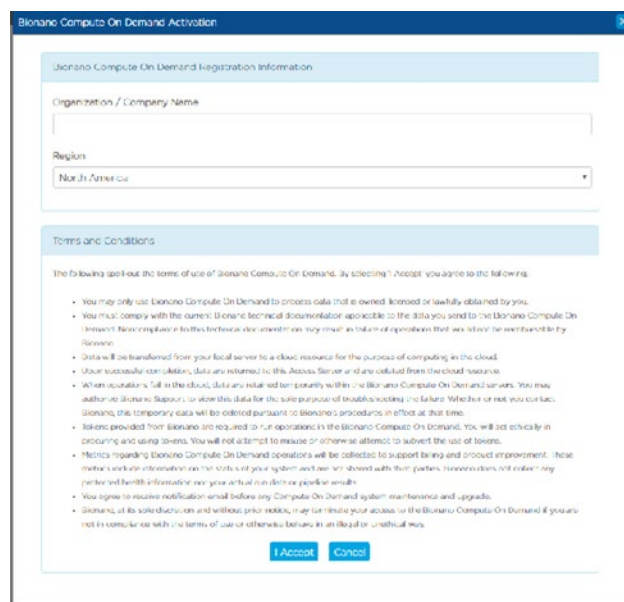


Figure 16. Bionano Compute On Demand Activation window

If not ready to complete the opt-in dialog, click the **Cancel** button, but Administrators will continue to be prompted each time they login until they complete the opt-in dialog or **Compute On Demand** is disabled. Once the opt-in has been completed, the dialog will no longer appear. Please carefully follow the steps below to fill in the two fields in the **Compute On Demand** setting page. Please note that once the information in these fields is submitted by clicking on the **I Accept** button, it is not possible to change them easily.

The first prompt is for **Organization / Company Name**. We recommend setting this value to the domain in the user’s email. For example, if the email were jsmith@bionano.com, we would recommend setting the organization to *bionano*.

The second prompt is for the computing region. We currently offer four computing end points: North America, Canada, Europe, and Germany. Germany server is intended only for customers who are in Germany and Europe server is for customers in Europe but not in Germany. Additional regions will be added in the future based on demand. The region is where the servers will reside that will execute the compute job. Users are not restricted to selected regions but selecting a region closest to where the system resides is best for security and performance reasons. Tokens must be purchased with the same server as registration.

Compute On Demand Operations

Once the opt-in process described above has been completed, Bionano Compute On Demand operations will be accessible through the **Project** browser. In the **Project** browser the **Operations** panel will display different operations users can perform based on the job type selected. When multiple options are enabled (**Saphyr Compute**, **Stratys Compute**, and/or **Compute On Demand**), there will be a drop-down menu in the **Operations** panel to allow the user to switch between the available options. If a local **Saphyr Compute** or **Stratys Compute** is not configured, then the **Compute On Demand** options will be the only ones available. Users can check the status of Compute On Demand by clicking **Compute On Demand Test** in **Settings** to see whether the service is online or not (**Figure 17**).

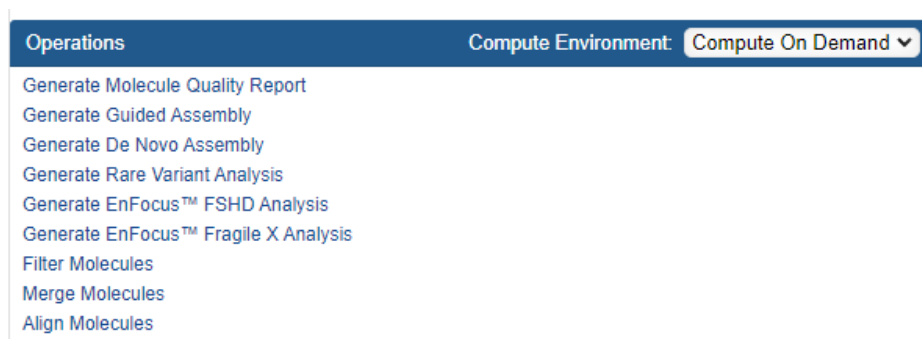


Figure 17. Compute On Demand drop-down toggle

Tokens

When performing a Bionano Solve operation locally, users first will select the required inputs. When performing a Bionano Compute On Demand operation, the inputs are the same as the inputs with one addition, the token cost (**Figure 18**). To proceed users must have enough tokens and must approve the token cost estimates. The token cost estimates will be a range. In the example below, the cost estimate is between 9 and 14 tokens. When users approve the operation the maximum token cost (14 in this example) will be deducted from the token balance. When the job is completed, if the cost was less than the maximum token estimate, the token difference will be refunded. For example, if the job below were to cost twelve tokens upon completion, users would receive a message informing those two tokens have been returned to token balance. Users are guaranteed that the cost will not exceed the maximum token cost quoted.



Figure 18. Token Cost message example

Tokens to execute jobs on our Bionano Compute On Demand service can be purchased through the designated salesperson or by contacting orders@bionanogenomics.com.

Compute On Demand Options

From the **Bionano Access** main menu, select **Compute On Demand**.

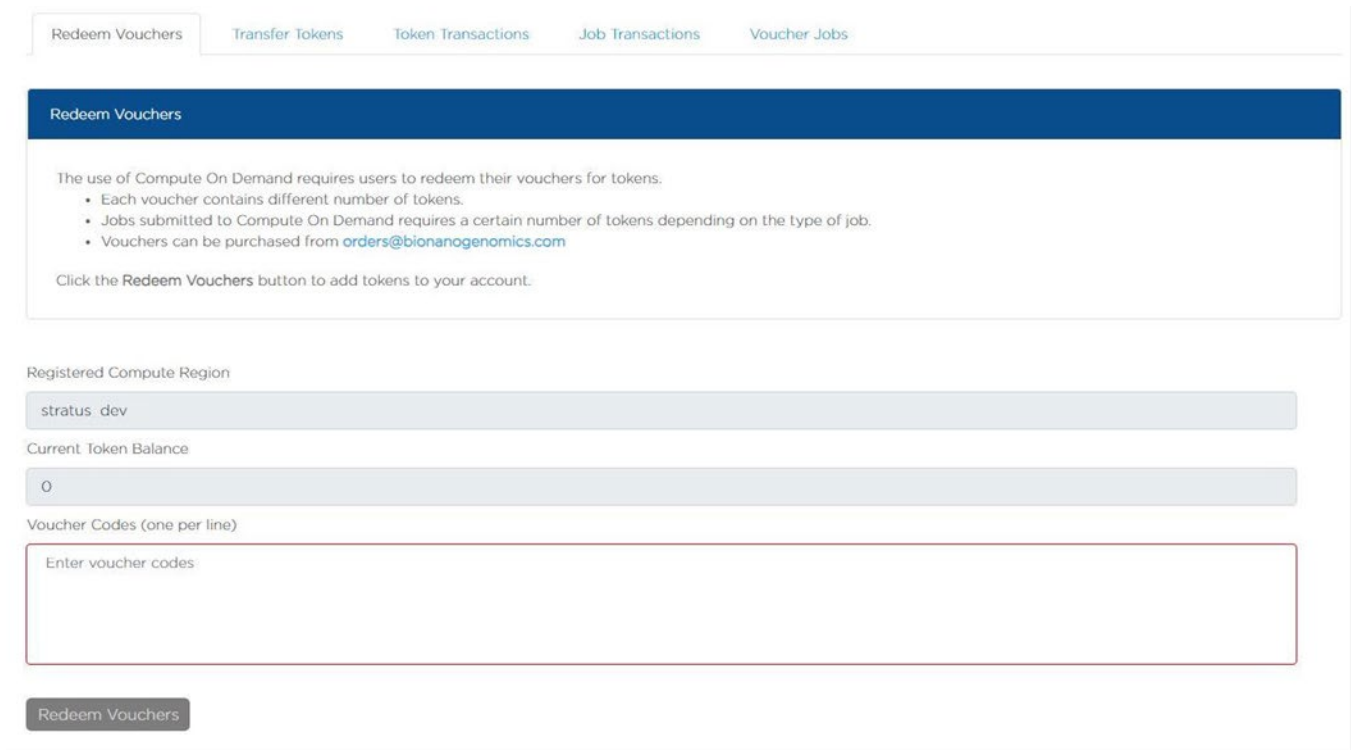


Figure 19. Redeem Vouchers

REDEEM VOUCHERS

1. **Redeem Vouchers** are selected by default.
2. At the **Registered Compute Region** field, display the region.
3. At the **Current Token Balance** field, display the current token balance.
4. At the **Voucher Codes (one per line)** field, input one voucher coder per line.
5. When users purchase tokens, users will receive one or more voucher codes. When a voucher is redeemed, all the tokens for that voucher are granted to the account for the user who is logged in.
6. Click **Redeem Vouchers** to redeem.

TRANSFER TOKENS

1. At the **Your Current Token Balance** field, display the current token balance.
2. At the **Organization Token Balance** field, display all the users and their token balance, email address.
3. At the **Select Token Recipient** field, select the user desired to transfer the token to. Any user who owns tokens can transfer them to other user accounts (except those with Read-Only access) on the same server. Read-Only accounts cannot have tokens because they cannot perform operations. Tokens currently cannot be transferred between Bionano Access servers.
4. At the **Token to Transfer** field, type the token amount to transfer.

5. Click **Transfer** or **Reset**.

TOKEN TRANSACTIONS

Users can view the transferred tokens from each user to a recipient.

JOB TRANSACTIONS

Users can view the reserved and actual cost of the tokens for each job.

VOUCHER JOBS

User can expand the **Voucher** row to see the corresponding jobs for every voucher. Use this view to understand how tokens for a given voucher were spent.

Token Use Recommendation

- Only use the required amount of data needed for the application. Please refer to *Data Collection Guidelines* (CG-30173) for more details
- Use the data down sampling tools available within Bionano Access to help achieve targeted coverage.
- Provide high quality references; this reduces costs by eliminating the need for the pipeline to first generate a rough assembly.

VIA Software

NOTE: The following features are only enabled once VIA software has been installed on the Access server. Contact Bionano Technical Support (support@bionano.com) for assistance setting up the VIA Sample Importer. The Bionano Access configuration file (/home/bionano/access/web/Server/Config/access.txt) file should include the key for viaExe to indicate where the VIA Sample Importer is installed. For example, the viaExe entry line in the access.txt file could look like this - "viaExe":"/home/bionano/VIASampleImporter/VIASampleImporter". VIASampleImporter appears twice in this example because the first is the directory and the second is the executable.

Setup VIA Server Connection

1. From the **Bionano Access** main menu, select **Settings**.
2. Select **System Services Settings**.
3. Under the **VIA Server** panel, click **Edit**. **Edit VIA Connection** window appears.
4. At the **VIA Server** field, type the domain (e.g., www.example.com or the IP address)
5. At the **Protocol** field, select **http** for a standard connection or **https** if the server requires a secure connection.
6. At the **Port** field, type the port used to access the server, typically 8443.
7. At the **VIA User** field, type the username for an active VIA user with the ability to upload samples.
8. At the **Password** field, type the password associated with the user above.
9. Click **Submit** to update the connection details.
10. Click **Test Connection** to check the validity of the connection.

Customize Sync Settings

1. From the **Bionano Access** main menu, select **Settings**.
2. Select **VIA Settings**.
3. To edit an existing sync setting, click the **Edit project sync** icon under the **Action** column, or to create a new sync setting, click **Add New VIA Setting**. The **Edit VIA Settings** window appears.
4. At the **Settings Name** field, type the desired display name for the setting.
5. At the **Bionano Access Job Type** field, select the job type.
6. To make the setting the default, select the **Default** checkbox.

7. At the **Sample Type** field, select the VIA sample type.
8. At the **Reference** field, select the VIA reference genome.
9. At the **Processing** field, select the processing to be launched by VIA when the job is imported.
10. At the **Panel** field, select the VIA panel to check for specific biomarkers.
11. Click **Submit** to save the VIA settings.

Upload to VIA

MANUAL

1. From the **Bionano Access** main menu, select **Analysis**. The **Projects** window appears.
2. Select the project containing the job to upload.
3. Select a completed **de novo Assembly** or **Rare Variant Analysis** job from the list.
4. In the **Options** pane, click **Upload to VIA**. The **VIA Upload** window appears.
5. At the **VIA Sample Name Field**, type the name of the VIA sample.
6. If uploading a job that already exists in VIA, switch **Overwrite** to “on.”
7. At the **VIA Settings** field, select the desired setting. See the “VIA Settings” section for details on how to customize these settings.
8. Click **OK** to start the upload.
9. Monitor the **VIA Status** in the **Job Details** panel.

AUTOMATED

When launching a job, if there are matching VIA sync settings for the reference and operation the user can select which sync setting to use. The results will automatically be transferred to VIA upon successful completion of the job. The default sync setting will be selected initially but can be changed later through manual upload. See the following sections for detailed instructions:

- Generate *de novo* Assembly
- Generate Rare Variant Analysis

Appendix

Red-Labeled Sample Experiment

Bionano Access supports experiments that use the red laser (if applicable) in the Saphyr instrument. These workflows are disabled by default. To enable this operation, users need to contact *Bionano Technical Support* (support@bionanogenomics.com).

RED-ONLY WORKFLOW

If the capability of running red labeled sample has been enabled, when users open the Single Sample workflow, they will have the choice of selecting green or red. If red-labeled sample setup has not been enabled, the system will only generate experiments using green laser.

DUAL-LABELED WORKFLOW

If red is enabled, the **Dual-Labeled workflow** in the experiment design module will be available. The **Dual-Labeled workflow** is designed to process a single sample with motifs in the DNA labeled using either green or red colors. The system will generate label-related metrics on the dashboard for each color. Users can provide separate references for each color, but only one reference will be associated with the final merged BNX file. The user is prompted to select which label (the primary) would retain its reference. The **Dual-Labeled workflow** will generate a single BNX file that contains both green and red labels information.

When users import a dual-labeled BNX file, the reference will automatically be associated with channel 1, as specified in the header or the BNX file. Users can select the **Edit** option to designate either channel for the reference. When users run an assembly using a dual labeled BNX file, they will be prompted to select labels from which channel they want to use for assembly. After the assembly is done, when users choose to show molecules in the viewer, the viewer system will show labels for both labeled motifs.

MULTIPLEX WORKFLOW

If red is enabled, the **Sample Multiplex** workflow will be enabled in the **Experiment Design** module. This workflow is designed to process one sample using green and a second sample using red in the same flowcell. This workflow will generate separate dashboard metrics for each color. This workflow will also generate a separate BNX file for each color. It is possible to use the same sample for both red and green, but the molecules loaded should be labeled with only red or green, but not both.

ISCN Symbols and Abbreviated Terms

Symbols and abbreviated terms used in the description of chromosomes and chromosomal abnormalities are listed below (**Table 28**). For a detailed discussion of ISCN notation produced by the Solve pipeline, please refer to *Bionano Solve Theory of Operation Variant Annotation Pipeline* (CG-30190). Consider the following when searching for chromosomal abnormalities:

Table 28. ISCN Symbols and Abbreviated Terms

Symbol/abbreviation	Description
amp	amplification of interval where copy number is greater than 4.
approximate sign (~)	denotes intervals and boundaries of a chromosome segment or number of chromosomes, fragments, or markers; denotes a range of number of copies of a chromosomal region when the exact number cannot be determined.
brackets, square ([])	surround number of cells or genome build.
dup	Duplication.
fus	fusion between regions of the same chromosome.
ins	Insertion.
inv	inversion.
ogm	Optical genome mapping data.
p	Short arm of chromosome.
parentheses ()	Surround structurally altered chromosomes and breakpoints; surround chromosome numbers, X, and Y in normal and abnormal results; surround coordinates (or nucleotide positions) in abnormal result.
q	Long arm of chromosome.
question mark (?)	Insertion of unknown sequence.
t	Translocation.
x	Copy number.

References

Table 29. References

Document Number	Reference Title
CG-00045	<i>OGM File Format Specification Sheet</i>
CG-30073	<i>Bionano Solve Theory of Operation: Hybrid Scaffolding</i>
CG-30110	<i>Bionano Solve Theory of Operation: Structural Variant Calling</i>
CG-30170	<i>Bionano Access Installation Guide</i>
CG-30173	<i>Data Collection Guidelines</i>
CG-30190	<i>Bionano Solve Theory of Operation: Variant Annotation Pipeline</i>
CG-30223	<i>Molecule Quality Report Guidelines</i>
CG-30304	<i>Bionano Access Dashboard Guidelines</i>
CG-30321	<i>Bionano Solve Theory of Operation: Bionano EnFocus™ FSHD Analysis</i>
CG-30377	<i>How to enable HTTPS in Bionano Access</i>
CG-30457	<i>Solve Theory of Operation: Bionano EnFocus™ Fragile-X Analysis</i>

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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