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## Revision History

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<tr>
<td>L</td>
<td>Update for Bioano Access 1.6.</td>
</tr>
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# Introduction

The Bionano Access® Software enables users to view Saphyr® run results in real time and perform a variety of bioinformatics analyses.

Saphyr and Irys® users can perform bioinformatics operations including, but not limited to, the following:

<table>
<thead>
<tr>
<th>Operation</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>In silico digestion of data sequences</strong></td>
<td>Perform <em>in silico</em> digestion by transforming a FASTA file into a Bionano CMAP file format.</td>
</tr>
<tr>
<td>Merge molecule data sets</td>
<td>Merge two or more molecules objects into a single molecules object.</td>
</tr>
<tr>
<td>Filter molecule objects</td>
<td>Filter molecules objects by length and total amount.</td>
</tr>
<tr>
<td>Generate <em>de novo</em> assemblies</td>
<td>Assemble single molecules into consensus maps for structural variant (SV) detection and hybrid scaffold applications.</td>
</tr>
<tr>
<td>Perform rare variant analysis</td>
<td>Detect structural variants at low allelic fraction.</td>
</tr>
<tr>
<td>Generate EnFocus™ FSHD analysis</td>
<td>Analyze regions relevant to facioscapulohumeral muscular dystrophy (FSHD).</td>
</tr>
<tr>
<td>Align maps</td>
<td>Compare two different maps or molecules to maps by aligning them to each other.</td>
</tr>
<tr>
<td>Build hybrid scaffolds</td>
<td>Merge Bionano maps with sequence assemblies to produce contiguous hybrid scaffolds that represent the chromosome structure.</td>
</tr>
<tr>
<td>Annotate structural variants</td>
<td>Provide variant annotation for downstream filtering and prioritization. Perform dual or trio comparisons.</td>
</tr>
<tr>
<td>Convert SMAP to VCF file</td>
<td>Convert insertion, deletion, duplication, inversion and translocation breakpoint calls in an SMAP file to dbVar-compliant VCF v4.2 format.</td>
</tr>
</tbody>
</table>

Irys users can import, analyze, and visualize their data in compatible format into Bionano Access. Saphyr users can additionally create experiments to run on their Saphyr Instrument and monitor chip runs in real time.
Bionano Access Terms

**Cluster** — Compute servers are required to perform bioinformatic analysis operations. Multiple compute servers can be organized into a cluster to work as unit and complete large amounts of work efficiently. Bionano sells Saphyr and Bionano Compute systems that can be combined into a cluster scaled to meet the customers’ computing requirements. Customers can also utilize Bionano Compute On Demand, a cloud-based computing platform, which is optimized for Bionano pipelines to perform bioinformatics analyses. The bioinformatics pipelines may also work on custom clusters; however, custom configurations may be needed.

**Objects** — Whenever Bionano Access performs a job, it may generate a variety of output data depending on the operation, such as molecule data, map alignments, assemblies, hybrid scaffold results, variant annotations, and others. Each of these distinctive sets of output files is an object (a set of output files). The types of objects include, but are not limited to:

- Alignment
- *De Novo* Assembly
- Rare Variant Analysis
- EnFocus™ FSHD Analysis
- Variant Annotation Results
- Hybrid Scaffold
- Consensus CMAP
- Molecule BNX
- FASTA file
- BED file

Please go to “Bioinformatics Analysis” section for more details.

**Prep** — The information of the sample, reference, and enzyme in one flow cell and related to experiment setup. This is also known as a Molecule Set.

**Cohort** — The number of subgroups that each scan is divided into for real-time analysis. One scan contains multiple cohorts, and we generate run metrics for each cohort.

**Consensus CMAP** — The Bionano Genomics CMAP file is a raw data file which provides location information for label sites within a genome map or an *in silico* digestion of a reference or sequence data. The CMAP is a tab-delimited text based file. Please go to P/N 30039, CMAP File Format Specification Sheet for more details.

**BNX** — The Bionano Genomics 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 (colors). Please go to P/N 30038, BNX File Format Specification Sheet for more details.
Login to Bionano Access

User Roles

There are four distinct user roles in Bionano Access. Users with administrator privileges can assign roles to other users.

<table>
<thead>
<tr>
<th>Roles</th>
<th>Description</th>
</tr>
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<tbody>
<tr>
<td>Administrator</td>
<td>The Administrator can set the security standards, assign user roles, manage user accounts and the access to projects, manage references and projects, delete projects permanently, or restore deleted projects.</td>
</tr>
<tr>
<td>Project Lead</td>
<td>The Project Lead can give users access to view projects, create and manage projects, and manage references. All project leads can view and edit all projects in the Projects list.</td>
</tr>
<tr>
<td>User</td>
<td>The User can view the projects that they have access to and edit experiments.</td>
</tr>
<tr>
<td>Read Only</td>
<td>The Read Only user can view the projects that they have access to.</td>
</tr>
</tbody>
</table>

Login

After installation of the Bionano Access software on the server, a URL is needed for login. To log in, you must first have a user account. Users with the Administrator role can create new accounts. If you do not have an account, please contact your administrator to create a new account. If you forgot your password, click Forgot Password, and you will receive an email to reset your password.

1. Navigate to the Bionano Access web page.
2. Enter user name and password.
3. Click Login.

Bionano Access Modules

The Bionano Access Home page lists the following modules:
Note: Depending on the types of User Roles (see below) and the types of Access install instances (see details in 30170 Bionano Access Installation Guide), not all the modules are available on the Home page.

<table>
<thead>
<tr>
<th>Module</th>
<th>Description</th>
<th>Availability</th>
</tr>
</thead>
<tbody>
<tr>
<td>Projects</td>
<td>This module lets users create, view, edit, and manage projects. Additionally, users can import and export data.</td>
<td>All user roles.</td>
</tr>
<tr>
<td>Experiments</td>
<td>This module lets users set up and manage Saphyr experiments, view templates, and track chip run progress.</td>
<td>Only for systems integrated with a Saphyr Instrument.</td>
</tr>
<tr>
<td>In Silico Digestion</td>
<td>This module lets users perform in silico digestion, update its settings, and view results.</td>
<td>All user roles.</td>
</tr>
<tr>
<td>Settings</td>
<td>This module lets users 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.</td>
<td>Only available to “Administrator” and “Project Lead” user roles.</td>
</tr>
<tr>
<td>Compute On Demand</td>
<td>This module lets users redeem and transfer tokens, check balance and transonation of tokens.</td>
<td>Only available for systems enabled with Compute On Demand.</td>
</tr>
</tbody>
</table>

Help

To get a better understanding of Bionano Access, users can click the Help icon.

The “Help” page has the following information:

a. Software version information of Bionano Access, Tools and Solve.

b. Compute On Demand version information of Bionano Tools and Solve (if enabled).

c. Tutorial Videos.

d. Security patch information.

e. Support contact information.

User Profile

Users can change the email address or password, enable or disable the email notifications after the first time logging in.

In Bionano Access, click the User Profile icon.

1. At the User Name field, user name displays.

2. At the Full Name field, full name displays.
3. At the **User Role** field, user role displays.

4. At the **Email Address** field, type the email address.

   By default, the **Send email notifications** check box is selected. You can uncheck this box to disable email notifications.

5. At the **Current Password** field, type the current password.

6. At the **New Password** field, type the new password.

7. At the **Confirm Password** field, type the new password again.

8. At the **Expiration date** field, the expiration date of the password is displayed.

9. Click **Submit**.

**Message Center**

In Bionano Access, click the **Message Center** icon to read and delete individual messages. Users can also mark all messages as read and delete all read messages.

When there are new messages about job status in Access 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 and 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. Messages are notifications sent to specific users regarding job status and other system events. Alerts are broadcast to all users with 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 sent by the instrument that pertain to a chip run on the dashboard.

**Menu**

User can directly access to different bionano access modules, include **Home, Projects, Experiments, In Silico Digestion, Settings, Profile, Help** and **Logout**.
Projects

In the Projects module, users can add new projects, manage projects, edit, import, and download objects. Users can also filter the projects by project name and export all the objects 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 objects in the project. Objects 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 **Projects**.
   
   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.

Export Project

Users can export a project containing multiple objects, such as molecules, assembly, SV merge and variant annotation pipeline.

Only users in the Project Lead or the Administrator role can export projects.

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

2. Click the **Export** icon of the project.
   
   The *Export Data* window appears.

3. At the **Export File Name** field, type in the name.

4. At the **Description** field, type the description (optional).

5. Select the objects (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, of “webServerShare” directory,
as configured in the access web server configuration file. For more details, please go to Bionano Access Installation Guide (PN 30170).

**Import Project**

Users can import an exported project containing multiple objects, such as molecules, assembly, and variant annotation pipeline.

Only in the Project Lead or the Administrator role can import project.

1. Copy the exported file (.bng) to the “import” folder of “webServerShare” directory, as configured in the targeted access web server.

2. Go to Bionano Access main menu, select **Projects**.

   The **Projects** window appears.

3. Click the **Import** icon of the project.

   The **Import Data** window appears.

4. At the **Import File Name** field, select the one you would like to import.

5. Click **Submit**.

**Delete Project**

Users in the Project Lead or the Administrator role can delete projects. Deleting a project is a two-step process. As the first step, a project lead can delete the project from the project list. However, only the administrator can delete the project permanently from Bionano Access, as the second step. For more details, see the **Deleted Projects** section.

1. From the Bionano Access main menu, select **Projects**.

   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.

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

1. If the project contains experiments, you cannot delete it because experiments cannot be deleted.

2. If the samples and objects in the project are not removed, you cannot delete the project.

Thus, before deleting the project as a whole, make sure this project does not contain experiments and the samples and objects are already deleted. Please see **Samples** section for more details.
Edit Project

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

1. From the Bionano Access main menu, select Projects.
   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

Users in the Project Lead or the Administrator role can give others access to view projects.

1. From the Bionano Access main menu, select Projects.
   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 to give access, and then click Grant Access. Users in the Project Lead or the Administrator role are not listed in the pane.
   The users 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 Projects.
   The Projects window appears.
2. Click the Deleted Projects icon at the top right-corner of the screen.
   The Deleted Projects screen appears.
3. At the project to restore, click the Restore icon.
The software restores the project and adds it back to the project list.

4. At the project to permanently delete, click the Delete icon.

The project would be permanently deleted.
Objects

When users select a project, the Project Browser page opens. The page contains menu options, the list of object(s), the selected-object details, and the options and operations for the selected object.

Menu

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

Objects List

All the objects that are listed belong to the selected project.

<table>
<thead>
<tr>
<th>Info</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>Name</td>
<td>The user-defined name for the object.</td>
</tr>
<tr>
<td>Sample</td>
<td>The sample name.</td>
</tr>
</tbody>
</table>
| Tag | The Tag feature allows users to create and edit keywords that are tagged to this object. Users can filter objects with the same keywords to easily manage objects in a project.  
1. To create or edit tags for the object, select the object, and then click Edit.  
2. At the Tags field, type the keyword, and then press Tab. Repeat to add more keywords.  
3. Click the delete “x” icon to delete the keyword. |
| Type | The object type (e.g. de novo assembly, molecules, map, scaffold, FASTA file). |
| Created | The date when the object was created. |
| | Click this icon to sort and filter the information in the columns. |
Object Details

<table>
<thead>
<tr>
<th>Info</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>Name</td>
<td>The object name should be unique in a project. The same object can have different names in different projects.</td>
</tr>
<tr>
<td>Sample</td>
<td>The sample name that is assigned to the object.</td>
</tr>
<tr>
<td>Description</td>
<td>[Optional] The description of the object.</td>
</tr>
<tr>
<td>Created</td>
<td>The date the object was created.</td>
</tr>
<tr>
<td>Status</td>
<td>The current status of the job.</td>
</tr>
<tr>
<td>User</td>
<td>The user who created the job.</td>
</tr>
<tr>
<td>Job ID</td>
<td>The job number of the object.</td>
</tr>
<tr>
<td>Command</td>
<td>Detailed information about the job and the Bionano Tools command issued to compute servers.</td>
</tr>
</tbody>
</table>

Icon of Job Error Reporting shows there is returned error message.

Object Options

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

Alignment Options

- Download Alignment Object
- View Maps Alignment
- View Molecules Alignment

**De Novo Assembly or Annotated De Novo Assembly Options**

- *De Novo* Assembly Report
- Molecules to Maps
- Maps to Reference with SV
- Download De Novo Assembly
- Download VCF file
Rare Variant Analysis or Annotated Rare Variant Analysis Options

- Rare Variant Analysis Report
- Maps to Reference with SV
- Download Rare Variant Analysis
- Download VCF file

Variant Annotation Pipeline Options

- View variant annotation results
- Download SV Annotation file
- Download Variant annotation Pipeline

EnFocus™ FSHD Analysis Options

- View EnFocus™ FSHD Analysis
- Maps to Reference with SV
- Go to Molecules Object
- Download EnFocus™ FSHD File
- Download EnFocus™ FSHD Report JSON File
- Download VCF file

Bed Option

  Download BED file

FASTA Option

  Download FASTA file

Map Options

  Download Map

Molecules Options

  Download Molecules File
  Show Molecule Quality Report (MQR)
**Scaffold Options**

- Hybrid Scaffold Report
- Maps to Next Generation Sequencing (NGS) with Conflicts
- Maps and Next Generation Sequencing (NGS) to Hybrid Scaffold
- Download Hybrid Scaffold
- Export NCBI Package

**Object Operations**

The *Operations* pane contains a list of operations that can be performed with the selected object. The operations are dependent on the type and current status of the object. The links in the *Operations* pane may be disabled and greyed out if a previous operation is still in progress.

**De Novo Assembly or Annotated De Novo Assembly Operations**

- Generate Hybrid Scaffold
- Generate 2-Enzyme Hybrid Scaffold
- Variant Annotation Pipeline
- Align Maps

**Rare Variant Analysis or Annotated Rare Variant Analysis Operations**

- Variant Annotation Pipeline

**EnFocus™ FSHD Analysis Operations**

- Generate EnFocus™ FSHD Analysis Report

**Map Operations**

- Generate Hybrid Scaffold
- Generate 2-Enzyme Hybrid Scaffold
- Align Maps

**Molecules Operations**

- Filter Molecule Object
- Merge Molecule Objects
- Align Maps
• Generate Molecule Quality Report (MQR)
• Generate De Novo Assembly
• Generate Rare Variant Analysis
• Generate EnFocus™ FSHD Analysis

**Import Object**

Users can import data from the following:

• Data output files that are generated from the Saphyr cluster, which are based on user-defined commands.
• Data from another system that is in the acceptable Bionano file format (e.g. Irys data generated by AutoDetect 2.1.4 or IrysSolve 2.1.0)

**Import Molecules**

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

1. From the Bionano Access main menu, select Projects.

   The Projects window appears.

2. Select the project to view from the list.

3. Click Import.

   The Import dialog box appears.

4. At the Object Type field, select Molecules.

5. At the Molecules Name field, type a name for this object.

6. At the Sample field, click the drop-down list to select the sample associated with file or New to add a new Sample. Once click New, the Sample dialog box appear.

6.1 At the Sample Name field, type a name for this sample.

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

6.3 [Optional] At the Comment field, type a brief comment.

7. [Optional] At the Reference for Channel 1 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 Molecules File field, and click Browse to choose the molecules file (.bnx or .bnx.gz).

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 is imported.

**Import Consensus Map**

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

1. From the Bionano Access main menu, select **Projects**.

   The **Projects** window appears.

2. Select the project to view from the list.

3. Click **Import**.

   The **Import** dialog box appears.

4. At the **Object Type** field, select **Consensus Map**.

5. At the **Consensus Map Name** field, type a name for this object.

6. At the **Sample** field, click the drop-down list to select the sample associated with the 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, and click **Browse** to choose the cmap file (.cmap).

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 is imported.

**Import an Alignment Object**

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

1. From the Bionano Access main menu, select Projects.

   The Projects window appears.

2. Select the project to view from the list.

3. Click Import.

   The Import dialog box appears.

4. At the Object 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 object.

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 *.*fasta, *.fa or *.fna* file to import to Bionano Access.

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

2. Select the project to view from the list.

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

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

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

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. Bionano 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.

1. From the Bionano Access main menu, select **Projects**.
   
   The *Projects* window appears.
2. Select the project to view from the list.

3. Click Import.

   The Import dialog box appears.

4. At the Object 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 object.

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. Bionano Access sends an email notification when the data are imported.

   Hybrid Scaffold result generated by Bionano Solve through command line must with output directory set in */output.

**Import De Novo Assembly**

Users must have De Novo Assembly or Annotated De Novo Assembly data generate by Bionano Solve in *.gz or *.zip file to import to Bionano Access.

1. From the Bionano Access main menu, select Projects.

   The Projects window appears.

2. Select the project to view from the list.

3. Click Import.

   The Import dialog box appears.

4. At the Object Type field, select De Novo Assembly.
5. At the **Assembly Name** field, type a name for this object.

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 **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.

*De Novo* Assembly result generated by Bionano Solve through command line must with output directory set in */output*.

**Import BED**

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

1. From the Bionano Access main menu, select **Projects**.

   The **Projects** window appears.

2. Select the project to view from the list.

3. Click **Import**.

   The **Import** dialog box appears.

4. At the **Object Type** field, select **BED**.

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

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 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. Bionano 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 Projects.

The Projects window appears.

2. Select the project to view from the list.

3. Click Import.

The Import dialog box appears.

4. At the Object Type field, select Cytoband.

5. At the Cytoband Name field, type a name for this object.

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

12. Click Ok.

The screen switches back to the Project Browser page. Bionano Access sends an email notification when the data
For Research Use Only. 
Not for use in diagnostic procedures.

are imported.

**Import Rare Variant Analysis**

Users must have rare variant analysis or annotated rare variant analysis data generate by Bionano Solve in *.zip file to import to Bionano Access.

1. From the Bionano Access main menu, select **Projects**.

   The **Projects** window appears.

2. Select the project to view from the list.

3. Click **Import**.

   The **Import** dialog box appears.

4. At the **Object Type** field, select **Rare Variant Analysis**.

5. At the **Rare Variant Analysis Name** field, type a name for this object.

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. 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.

   Rare Variant Analysis result generated by Bionano Solve through command line must with output directory set in */output*.

**Import FSHD Analysis**

Users must have FSHD analysis data generate by Bionano Solve in *.zip file to import to Bionano Access.
1. From the Bionano Access main menu, select **Projects**.

   The *Projects* window appears.

2. Select the project to view from the list.

3. Click **Import**.

   The *Import* dialog box appears.

4. At the **Object Type** field, select **FSHD Analysis**.

5. At the **FSHD Analysis Name** field, type a name for this object.

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 **FSHD Analysis File** field, click **Browse** to choose the FSHD 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.

**Important:** FSHD Analysis result generated by Bionano Solve must with output directory set in */output*.

**Import Variant Annotation**

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

1. From the Bionano Access main menu, select **Projects**.

   The *Projects* window appears.

2. Select the project to view from the list.

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

4. At the **Object Type** field, select **Variant Annotation**.

5. At the **VAP Analysis Name** field, type a name for this object.

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.

**Important:** Variant Annotation Pipeline Analysis result generated by Bionano Solve must with output directory set in `*/output`.

### Remove Object

Users can remove objects from a project. Removing an object from a project does not affect that same data that are shared/copied with other projects. To remove an object that exists in several projects, users would need to remove it from each project.

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 Objects list, select the object to remove.

4. Click **Remove**.

   The **Remove Object from Project** dialog box appears.

5. Click **Yes**.
6. When objects are deleted in the Project Browser they go into the trash. To permanently remove an object or restore a previously deleted object in a project, select the project and click on Deleted Objects icon at the top-right corner of Access webpage to manage deleted objects. Administrators can permanently delete objects in the trash. There is a button to allow Administrators to delete all the objects in the trash, so the objects do not have to be deleted one at a time.

**Copy Object**

Users can copy objects from one project and add it to another project.

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 Objects list, select an object (i.e. assembly, molecules, map) to share.

4. Click Copy.
   
   The Copy Object 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 Object**

Users can edit object’s name, sample reference, tags and description informations.

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

2. Select the project to view from the list.

3. Click Edit.
   
   The Edit Object 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.

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.
Jobs

Users can view all submitted jobs’ status, cancel running jobs, sort and filter the columns of job listing.

<table>
<thead>
<tr>
<th>Info</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>Project</td>
<td>The project name of the job.</td>
</tr>
<tr>
<td>Job ID</td>
<td>The unique job number auto-generated by Bionano Access server when a job is created.</td>
</tr>
<tr>
<td>Operation</td>
<td>The operation of the job, such as Alignment, Import Molecule, DLE-1 De Novo Assembly, Variant annotation.</td>
</tr>
<tr>
<td>Object Created</td>
<td>The created object name.</td>
</tr>
<tr>
<td>User</td>
<td>The user name who submitted the job.</td>
</tr>
<tr>
<td>Created</td>
<td>The date when the job was created/submitted.</td>
</tr>
<tr>
<td>State</td>
<td>The current state of the job.</td>
</tr>
<tr>
<td></td>
<td>- <strong>Active:</strong> the job is running.</td>
</tr>
<tr>
<td></td>
<td>- <strong>Working:</strong> copying input files; <strong>job has not started</strong></td>
</tr>
<tr>
<td></td>
<td>- <strong>Complete:</strong> job completed successfully</td>
</tr>
<tr>
<td></td>
<td>- <strong>Cancelled:</strong> job cancelled</td>
</tr>
<tr>
<td></td>
<td>- <strong>Failed:</strong> job failed</td>
</tr>
<tr>
<td>Status</td>
<td>The current status of the job. System reads <code>status.xml</code> file in the compute server for each job.</td>
</tr>
<tr>
<td>Server</td>
<td>The IP address or hostname of your assembly server.</td>
</tr>
<tr>
<td>Actions</td>
<td>To view job details or cancel the job.</td>
</tr>
<tr>
<td></td>
<td>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.</td>
</tr>
</tbody>
</table>

Samples

Menu

<table>
<thead>
<tr>
<th>Menu</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>Add</td>
<td>To add a new sample.</td>
</tr>
<tr>
<td>Remove</td>
<td>To remove a sample. Note: to remove a sample, one has to remove all the associated objects.</td>
</tr>
<tr>
<td>Copy</td>
<td>To copy sample from the object and paste it in another project.</td>
</tr>
<tr>
<td>Edit</td>
<td>To edit sample name, description and comment.</td>
</tr>
</tbody>
</table>

Users can view all samples’ information, sort and filter the columns of sample list.
### Info

<table>
<thead>
<tr>
<th>Sample</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>Created</td>
<td>The date the sample was created.</td>
</tr>
<tr>
<td>Description</td>
<td>[Optional] The description of the sample.</td>
</tr>
<tr>
<td>Comment</td>
<td>The date the object was created.</td>
</tr>
</tbody>
</table>

Users can view all the associated objects while selecting one sample.

<table>
<thead>
<tr>
<th>Type</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>Name</td>
<td>The object name for each sample.</td>
</tr>
<tr>
<td>Tag</td>
<td>The Tag feature allows users to create and edit keywords that are tagged to this object. Users can filter objects with the same keywords to easily manage objects in a project.</td>
</tr>
<tr>
<td>Created</td>
<td>The date the object was created.</td>
</tr>
</tbody>
</table>

### Reset

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

### Deleted Object

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 object to delete.

3. Click the **Deleted Objects** 🗑 icon at the top right-corner of the screen.
   
   The **Deleted Objects screen** appears.

4. Click **Delete all objects** if it is desired to delete all the objects permanently in all projects.

5. Select the object from the list to restore, click the **Restore** icon.
   
   The software restores the project and adds it back to the object list.

6. Select the object from the list, and then click the **Delete** icon.
   
   The software permanently deletes the object from the project. If the object is shared with other projects, the object still exists in those projects.
To permanently delete the same object that is in other projects, repeat steps 1 through 4 for the other projects.

**Download Object**

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 Objects list, select the object to download.

4. In the *Options* pane, click the download [object type] option.
   
The software downloads the data onto the workstation running the browser.
Experiments

In the Experiments module, users can add new experiments, manage experiments, track run performance, and view real-time metrics. Users can create multiple experiments for one project. Additionally, users can view and edit templates.

Add Experiment

Each chip can only be associated to one experiment. Before inserting the Saphyr Chip into the instrument, create an experiment to associate it with the chip. The run results are posted for the experiment that is associated with the chip.

An experiment created on Bionano Access is in pending status until it is associated with a Saphyr chip. Users can modify or delete pending experiments. Once an experiment is associated with a scan on the Saphyr, users cannot modify or delete the experiment.

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

1. From the Bionano Access main menu, select Experiments.

The Experiment list appears. Here is a description of Chip Runs list.

<table>
<thead>
<tr>
<th>Info</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>Project</td>
<td>The project name.</td>
</tr>
<tr>
<td>Experiment</td>
<td>The experiment name.</td>
</tr>
<tr>
<td>Chip Name</td>
<td>The chip name.</td>
</tr>
<tr>
<td>Created</td>
<td>The date and time when the experiment was created.</td>
</tr>
<tr>
<td>Action</td>
<td>To view dashboard or delete the experiment.</td>
</tr>
</tbody>
</table>

Click Expand Icon to get more information about each experiment as below:

<table>
<thead>
<tr>
<th>Info</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>Location</td>
<td>The location of Flow Cell on each chip. FC – 1, FC – 2 or FC – 3.</td>
</tr>
<tr>
<td>Sample</td>
<td>The sample origin. Human samples or others.</td>
</tr>
<tr>
<td>Label</td>
<td>The label color. Green 01 or Red 01.</td>
</tr>
<tr>
<td>Enzyme</td>
<td>The enzyme name.</td>
</tr>
<tr>
<td>Reference</td>
<td>The reference name.</td>
</tr>
<tr>
<td>Auto Assemble</td>
<td>False or true.</td>
</tr>
</tbody>
</table>
2. Select Add Experiment.

   A dialog box appears.

3. At the Project field, click the drop-down list to select the project that is associated with the experiment.

   [Optional] If users do not have a project for this experiment, click New to create a project.

4. At the Experiment Name field, type in the name of the experiment.

5. [Optional] At the Experiment Description field, type in a brief description.

6. [Optional] At the Experiment Template field, click the drop-down list to select a template.

7. Click Next.

8. At the Workflow field, click the drop-down list to select “Single Sample”, “Sample Multiplex” or “Dual Labeled Sample”.

   Please see section “Red Labeled Experiment” below for details to setup “Sample Multiplex” and “Dual Labeled Sample” experiments. The instruction below describes setup for Single Sample.

9. At the Throughput Target (Gbp) field, type the target throughput. This field only applies to Saphyr instruments running ICS 4.8 or greater.

10. At the Molecule Object Name field, type the name of molecule object. You can also leave it blank for default naming.

11. At the Sample field, click the drop-down list to select a sample or click New.

12. At the Label field, click the drop-down list to select “Green 01” (green labeled) or “Red 01” (red labeled) for Single Sample.

13. At the Recognition Enzyme field, click the drop-down list to select an enzyme or click New to add a new enzyme.

14. [Optional] At the Reference field, click the drop-down list to select a genome reference for the experiment.

15. [Optional] At the Comment field, type in a brief comment.

16. At the Chip Part Number field, select the correct Chip Part Number, then click Add Chip.

17. Click Add to Flowcell 1.

   To better distinguish between dual labeled samples and multiplexed flow cells, Prep Type is added to each flow cell listing in the experiment design module.

   To edit information to Flowcell 1, click Remove in the Chip pane, and then make changes.
18. Repeat steps 9 through step 17 for Flowcells 2 and 3, if applicable.

19. Click **Add to Flowcell 2 (or 3)**.

   To edit information to Flowcell 2 (or 3), click **Remove** in the Chip pane, and then make changes.

20. Click **Add Chip**.

   To add more chips for samples in one experiment.

21. Click **Next**.

   A summary of the experiment appears.

22. By default, the **Auto Assemble Human** check box shows up if the reference is a human genome. Users can select the check box to activate auto assembly for a human samples.

   When this option is selected, if there is at least 80X effective coverage, the *de novo* assembly will start using the human haplotype-aware assembly parameters immediately after the molecules file is generated. Data collection will continue till it reaches the throughput target or run time limit for the flowcell. A BED file for masking will be automatically selected based on reference. If Auto Assemble Human is chosen, Complex Multiple Path Regions (CMPRs) will be cut by default during assembly. Please refer to P/N 30110 Theory of Operation SV calling for more details.

23. By default, the **Auto Enfocus™ FSHD** check box shows up if it is a DLE-1 human sample using hg38 as reference.

   When this option is selected, EnFocus™ FSHD analysis will run once 400 Gbp DNA is collected, with a 70% or better average map rate, and a molecule N50 (>=150 kbp) over 200 kbp. Please refer to P/N 30321 Theory of Operation EnFocus FSHD Analysis for more details.

24. Only one of the **Auto Assemble Human** or **Auto Enfocus™ FSHD** boxes can be checked at one time.
25. Click **Done**.

The *Experiment List* screen appears.

### 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 **Experiments**.

   The *Experiment* list appears.

2. Select **Templates**.

   By default, there are 3 templates for FSHD Analysis using Chip Part Number 20319, 20366 and 20367.

3. Repeat steps 2-17 in the Create an Experiment section.

4. [Optional] Click **Save as Template**.

   A window of Experiment Template 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 slot for different samples.

   The experiment template will be saved and you can select it.

7. Click **Ok**.

### Monitor Run Progress

After the instrument scans the chip for 15 to 20 minutes and raw images are being 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 **Experiments**.

2. Use **Experiments** or click **Chip Runs**.

   The latest chip run appears at the top of the list. Here is a description of Chip Runs list.

<table>
<thead>
<tr>
<th>Info</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>Start</td>
<td>The date and time when the chip run was started.</td>
</tr>
<tr>
<td>Finish</td>
<td>The date and time when the chip run was finished.</td>
</tr>
<tr>
<td>Project</td>
<td>The project name.</td>
</tr>
<tr>
<td>Experiment</td>
<td>The experiment name.</td>
</tr>
<tr>
<td>Chip Name</td>
<td>The chip name.</td>
</tr>
</tbody>
</table>
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.

The Dashboard screen appears showing:

- Run information
- Analysis graphs
- Run metrics table

### Run Information

<table>
<thead>
<tr>
<th>Info</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>Chip</td>
<td>The Saphyr Chip bar code.</td>
</tr>
<tr>
<td>Run ID</td>
<td>The system-generated run identifier.</td>
</tr>
<tr>
<td>Project</td>
<td>The project name.</td>
</tr>
<tr>
<td>Experiment</td>
<td>The experiment name.</td>
</tr>
<tr>
<td>Instrument</td>
<td>The instrument serial number.</td>
</tr>
<tr>
<td>Cohorts Per Scan</td>
<td>The number of subgroups that each scan is divided into for real time analysis.</td>
</tr>
<tr>
<td>Min Length</td>
<td>The minimum length of molecules that are used for analysis. The setting for this parameter is system generated.</td>
</tr>
<tr>
<td>Min Labels</td>
<td>The minimum labels per molecule that are used for analysis. The setting for this parameter is system generated.</td>
</tr>
<tr>
<td>Start Time</td>
<td>The run start time.</td>
</tr>
<tr>
<td>End Time</td>
<td>The run end time.</td>
</tr>
</tbody>
</table>

### Analysis Graphs

<table>
<thead>
<tr>
<th>Map</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>DNA per Scan</td>
<td>This graph shows the amount of DNA per flowcell that is detected per scan.</td>
</tr>
</tbody>
</table>
### 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.

### Run Metrics Table

<table>
<thead>
<tr>
<th>Metric</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>Plot</td>
<td>The color used in the plot and check box for each flow cell.</td>
</tr>
<tr>
<td>Flowcell</td>
<td>The scanned flowcell.</td>
</tr>
<tr>
<td>Prep</td>
<td>The names of the sample, channel, enzyme, type and reference</td>
</tr>
<tr>
<td>Avg N50 (&gt;=150kbp) (Mbp)</td>
<td>The molecule length N50 for all molecules that are ≥ 150 kbp in length.</td>
</tr>
<tr>
<td>Avg N50 (&gt;=20kbp) (Mbp)</td>
<td>The molecule length N50 for all molecules that are ≥ 20 kbp in length.</td>
</tr>
<tr>
<td></td>
<td>Molecules that are less than 20 kbp are considered noise by the image detection algorithm.</td>
</tr>
<tr>
<td>Avg Label Density (per 100 kbp)</td>
<td>The number of labels that are detected by the image detection algorithm per 100 kbp of DNA length for molecules ≥ 150 kbp.</td>
</tr>
<tr>
<td>Avg Map Rate</td>
<td>The percentage of molecules that map to the reference for molecules ≥ 150 kbp.</td>
</tr>
<tr>
<td></td>
<td>If no reference genome is provided, the metric is blank.</td>
</tr>
<tr>
<td>Estimated Effective Coverage</td>
<td>The coverage number is calculated as follows:</td>
</tr>
<tr>
<td></td>
<td>Average Map Rate * Total DNA / length of the reference</td>
</tr>
<tr>
<td></td>
<td>For human structural variation detection, we recommend at least 80X effective coverage.</td>
</tr>
<tr>
<td>Avg PLV</td>
<td>Average Positive Label Variance: Percentage of molecule labels absent in reference labels.</td>
</tr>
<tr>
<td>Avg NLV</td>
<td>Average Negative Label Variance: Percentage of reference labels absent in molecule labels.</td>
</tr>
<tr>
<td>DNA Collected (Gbp)</td>
<td>The total amount of DNA that is detected per flowcell during the run.</td>
</tr>
<tr>
<td>Scan Count</td>
<td>The scans “Recd” number reflects the volume of scans submitted by Saphyr Control Software to Bionano Access.</td>
</tr>
<tr>
<td></td>
<td>The Scans “Mapped” number reflects the number of scans that have had metrics data generated.</td>
</tr>
<tr>
<td>Cumulative DNA (Gbp)</td>
<td>The total amount of DNA that is detected in this flowcell across all runs of this chip.</td>
</tr>
</tbody>
</table>

Please refer to Bionano Access Dashboard Guidelines 30304 for more details.
## Bioinformatics Analysis

### Analysis Types

Users can perform the following analyses in Bionano Access:

<table>
<thead>
<tr>
<th>Analysis</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>Molecules Merge</td>
<td>Merge two or more molecules objects into a single molecules object.</td>
</tr>
<tr>
<td>Filter molecule objects</td>
<td>Filter molecules objects by length and total DNA.</td>
</tr>
<tr>
<td>De novo Assembly</td>
<td>Assemble single molecules into consensus maps for SV detection and hybrid scaffold applications.</td>
</tr>
<tr>
<td>Rare variant analysis</td>
<td>Identify rare variants at low frequencies.</td>
</tr>
<tr>
<td>EnFocus™ FSHD analysis</td>
<td>Analyze FSHD relevant regions.</td>
</tr>
<tr>
<td>1-Enzyme Hybrid Scaffold</td>
<td>Use a set of Bionano maps and a sequence assembly to build hybrid scaffolds.</td>
</tr>
<tr>
<td>2-Enzyme Hybrid Scaffold</td>
<td>Use two sets of Bionano maps and a sequence assembly to build two-enzyme hybrid scaffolds</td>
</tr>
<tr>
<td>Alignments</td>
<td>Compare two different maps or molecules to maps by aligning them to each other.</td>
</tr>
<tr>
<td>Variant Annotation Pipeline</td>
<td>Annotate SV calls for applications such as identification of rare and potential de novo SVs for trio (mother, father, and proband) or for cancer research.</td>
</tr>
<tr>
<td>Convert SMAP to VCF file</td>
<td>Convert insertion, deletion, duplication, and inversion and translocation breakpoint calls in an SMAP file to dbVar-compliant VCF v4.2 format.</td>
</tr>
</tbody>
</table>

### Filter Molecule Objects

User can filter BNX files based on molecules length and total DNA.

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 Objects list, select a molecules object to filter.
4. In the **Operations** pane, select **Filter Molecule Objects**.  
   The BNX Filter screen appears.
5. At the **Filtered Molecule Name** field, type the new name for the molecules object.
6. At the **Channel Information** field, displays the Channel ID, the enzyme motif, name and reference name.

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. [Optional] At the **Channel to Keep** field, select one of **Keep Only Channel 1**, **Keep Only Channel 2** or **Keep Both Channels**.

11. [Optional] Select the check box of **Channel 1 Label Count** to type value of **Minimum**.

    By default, Bionano Access automatically uses 10 as Minimum.

12. [Optional] Select the check box of **Length (kbp)** to type values of **Minimum** and **Maximum**.

    By default, Bionano Access automatically uses 100 kbp as Minimum and 5000 kbp as Maximum.

13. [Optional] Select the check box of **Total DNA (Gbp)** and **Random Seed** to randomly selected a number of DNA.

    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. By default, Bionano Access automatically uses 300 Gbp as Total DNA and 17 as Random Seed.

14. Click **Submit**.

    A new molecules object will appear in the project browser.

    Users will receive an email when the filtering is complete.

### Merge Molecule Objects

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

### Best Practices:

- Merge molecule objects 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 1st generation Saphyr System and 2nd generation. Please contact Bionano Genomics 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 Genomics Technical Support for assistance.

- Dual labeled BNX files can only be merged with dual labeled BNX files with matching recognition enzymes.

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 Objects list, select a molecules object to merge.

4. In the **Operations** pane, select **Merge Molecule Objects**.
   
   A dialog box appears.

5. At the **Merge Molecule Name** field, type the new name for the molecules object.

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 check boxes of the molecule objects to merge with the molecules object that was selected in step 3 from the list of molecules objects. Here is the description of the information in the list:

<table>
<thead>
<tr>
<th>Info</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>Name</td>
<td>The molecules object name</td>
</tr>
<tr>
<td>Sample</td>
<td>The sample name.</td>
</tr>
<tr>
<td>Reference</td>
<td>The reference.</td>
</tr>
<tr>
<td>Map Rate (%)</td>
<td>The percentage of molecules that map to the reference for molecules ≥ 150 kbp.</td>
</tr>
<tr>
<td></td>
<td>If no reference genome is provided, the metric is blank.</td>
</tr>
<tr>
<td>N Mol</td>
<td>The total number of molecules</td>
</tr>
<tr>
<td>Coverage</td>
<td>The coverage number is calculated as follows:</td>
</tr>
<tr>
<td></td>
<td>Total DNA Throughput / length of the reference</td>
</tr>
<tr>
<td>PLV %</td>
<td>Average Positive Label Variance: Percentage of molecule labels absent in reference labels.</td>
</tr>
<tr>
<td>NLV %</td>
<td>Average Negative Label Variance: Percentage of reference labels absent in molecule labels.</td>
</tr>
</tbody>
</table>
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 objects in the project that are a match with the molecule object selected to merge with. For example, it selects the molecule objects 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 objects to generate alignment data.

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 Objects 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 object.

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

8. At the Alignment p-value field, type the p-value for the alignment. You can also check the boxes of Output Best Alignment and Swap Anchor - Query.

9. At the Sample field, displays the name of the sample.

10. At the Source Anchor Map field, displays the name of the selected consensus map or de novo assembly results.

11. At Target Query Map, select the map, assembly, or reference to generate an alignment.

12. Click Submit.

   Users will receive an email when the alignment is complete.

Users can also use molecules objects to generate alignment data.

1. From the Bionano Access main menu, select Projects.
For Research Use Only.
Not for use in diagnostic procedures.

The *Projects* window appears.

2. Select the project to view from the list.

3. In the Objects list, select the molecules object to use for alignment.

4. In the *Operations* pane, select **Align Maps**.

   The *BNX 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 object.

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

8. At the **Sample** field, displays the name of the sample.

9. At the **Source Molecules** field, displays the name of the selected molecules object.

10. At **Target Map** field, select the map, assembly, or reference to generate an alignment.

11. Click **Submit**.

   Users will receive an email when the alignment is complete.

---

**Generate Molecule Quality Report**

Users can generate molecule quality report with molecules data. Bionano Access will automatic generate a molecule quality report after importing molecule files.

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 Objects list, select the molecules object 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 object.

6. At the **Sample** field, displays the name of the sample.

7. [Optional] At the **Reference** field, displays the reference of the selected molecules object.

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.
Generate *de novo* Assembly

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

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 Objects list, select the molecules object to perform *de novo* assembly.

4. In the **Operations** pane, select **Generate de novo Assembly**.

5. At the **Assembly Name** field, type the name of the assembly.

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

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

8. The **Selected BNX** field shows the name of selected BNX object.

9. The **Reference** field shows the name of selected reference

10. The **Organism** field shows the name of selected organism (Human or non-human).

11. [Optional] At the **Expected CN Baseline File** field, select “Default Expected CN Baseline” or customer’s own baseline file from the drop-down list.

   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. Please refer to P/N 30210 Introduction to Copy Number Analysis for more details.

12. [Optional] 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..

   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. Please refer to P/N 30210 Introduction to Copy Number Analysis for more details.

13. [Optional] At the **CNV Mask Bed File** field, select a bed file from the drop-down list.

   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. Please refer to P/N 30210 Introduction to Copy Number Analysis for more details.

14. [Optional] At the **SV Mask Bed File** field, select a bed file from the drop-down list.

   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. Refer to P/N 30110 Structural Variant Calling Theory of Operation for more details.

15. At the Use Custom Config field,

1) If you select “No”, you can use the dialog to automatically determine the correct configuration to use.
   a) At the Est. Genome Size (Gbp) field, type the estimated genome size.
   b) 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 you have a reference, you will have the option to enable pre-assembly.
   c) 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.
   d) At the Cut Complex Multi-Path Regions field, select “yes” or “no”. Recommend this to be set to ‘Yes’ for most applications of human samples. Refer to P/N 30110 Structural Variant Calling Theory of Operation for more details.
   e) At the Instrument field, select “Saphyr” or “Irys” for the instrument that was used to collect the data. It is only available for non-DLE1 data.

2) If you select “yes”, then the list of configuration files will be displayed. You can select the configuration file from the default list or select a customized configuration.
   a) At the Est. Genome Size (Gbp) field, type the estimated genome size.
   b) 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 you have a reference you will have the option to enable pre-assembly.
   c) To customize a configuration, click the Edit icon in one of the default configurations. The configuration dialog box appears.
   d) Define the settings for de novo assembly, and then click Save As.
   e) Type the name of the configuration, and then click OK.

The customized configuration appears in the list of configurations.

16. Click Next.

17. At the field of MQR Details, you can check MQR information of the BNX file by clicking on the up-arrow.

For the following options, please go to “Generate Variant Annotation Pipeline” for more details.

18. The Run Variant Annotation with control SV database check box shows up. Check this box to enable variant annotation pipeline.
19. At the **Known genes** field, select a bed file from the drop-down list.

20. Use the default values or enter the values to use for the parameter fields of variant annotation.

21. Click **Submit**.

Users will receive an email when the assembly is complete.

Depending on the volume of data and coverage, the de novo assembly process may take a while to complete. If Variant Annotation is selected, only one annotated de novo assembly object will be generated.

**Generate Rare Variant Analysis**

Users can generate rare variant analysis with molecules data.

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 Objects list, select a molecules object 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 selected BNX object.

9. The **Reference** field shows the name of selected reference.

For the following four options, please go to “Generate De Novo Assembly” for more details.

10. [Optional] At the **Expected CN Baseline File** field, select “Default Expected CN Baseline” or customer’s own baseline file from the drop-down list.

11. [Optional] 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.

12. [Optional] At the **CNV Mask Bed File** field, select a bed file from the drop-down list.

13. [Optional] At the **SV Mask Bed File** field, select a bed file from the drop-down list.

14. At the **Configuration** field, select the Arguments.

15. Click **Next**.

16. At the field of **MQR Details**, you can check MQR information of the BNX file by clicking on the up-arrow.

For the following options, please go to “Generate Variant Annotation Pipeline” for more details.

17. The **Run Variant Annotation with control SV database** check box shows up. Check this box to enable
variant annotation pipeline.

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

19. Use the default values or enter the values to use for the parameter fields of variant annotation.

20. Click Submit.

**Generate EnFocus™ FSHD Analysis**

Users can generate FSHD analysis with molecules data. It is only applicable for human samples using hg38 as the reference.

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 Objects list, select a molecules object to generate FSHD analysis.

4. At the field of FSHD Analysis Background information, you can 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 selected sample.

9. The Selected BNX field shows the name of selected BNX object.

10. The Reference field shows the name of selected reference.

11. The Show Additional information field is enabled by default.

12. Click Submit.

**Generate Hybrid Scaffold**

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

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 Objects 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 alignment object.

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 Resolve Conflicts for both Bionano Assembly and Sequence Assembly. For more information, see more in P/N 30073 Bionano Solve Theory of Operation Hybrid Scaffold.

12. At the Trim Overlapping Sequence Contigs field, turn it on or off. For more information, see more in P/N 30073 Bionano Solve Theory of Operation Hybrid Scaffold.

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 Projects.
   The Projects window appears.
2. Select the project to view from the list.
3. In the Objects 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 alignment object.

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 P/N 30073 Bionano Solve Theory of Operation Hybrid Scaffold.

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 Pipeline**

This analysis lets users identify rare and potential *de novo* SVs for trio (mother, father, and proband) or dual (parent and proband) research. This analysis is also used for cancer research.

The following task is an example of the trio variant annotation pipeline (proband, father, mother).

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 Objects list, select the map or assembly to use as a reference.

4. In the **Operations** pane, select **Variant Annotation Pipeline**.
5. At the **Name** field, type the name for the variant annotation.

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

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

8. At the **Variant Annotation Result Type** field, select **Trio** from the drop-down list.

9. Click **Next**.

10. At the **Proband de novo assembly** field, select the assembly from the drop-down list.

11. 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. User can also run variant annotation pipeline with no control SV database file selected. Refer to P/N 30110 Structural Variant Calling Theory of Operation for more details.

12. 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 P/N 30110 Structural Variant Calling Theory of Operation for more details.

13. At the **Father de novo assembly** field, select the assembly from the drop-down list.

14. At the **Mother de novo assembly** field, select the assembly from the drop-down list.

15. Use the default values or enter the values to use for the following parameter fields.

   For more details, see the *Bionano Solve Theory of Operation, Variant Annotation Pipeline (document #30190)* for guidance on setting these parameters.

16. Click **Submit**. The spinning arrows indicates that the software is uploading data to the server.

17. When the spinning arrows disappear, click **Close**.

   Bionano Access sends an email to notify the user when the variant annotation 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 Projects. The Projects window appears.
2. Select the project to view from the list.
3. In the Objects list, select an assembly, alignment, rare variant analysis, variant annotation pipeline, or scaffold object. Users can view these objects on the Viewer screen.
4. In the Options pane, depending on the object selected, these are possible options to get to Viewer screen:
   - Molecules to Maps
   - Maps to Reference with SV
   - View variant annotation results
   - View EnFocusTM FSHD analysis
   - Maps to NGS with Conflicts
   - Maps to NGS with Hybrid Scaffold

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.

1. In Bionano Access, navigate to the Viewer screen of the object to analyze.
2. Select Circos Plot from the drop-down list.

   The Circos plot is displayed by default for human assemblies, rare variant analysis and variant annotations. On the left, the Circos plot displays the cytoband, SV, CNV and Aneuploidy. Information about Filter Criteria, BED Files, Samples, and Color legend for each SV type is on the right. Here is an example of human variant annotation pipeline visualization page:
3. Click any SV, CNV or cytoband navigate to **Genome Browser Viewer**.

4. Click the **Add BED** icon,  
**BED Filter Settings** dialog box appears.

5. At the **BED Selection** field, click the drop-down list and select one bed file.

6. At the **Structural Variant Overlap Precision (Kbp)** field, type the value. **12** is the system default.

7. 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 variant** is the system default.

8. At the **CNV Segment Overlap Precision (Kbp)** field, type the value. **500** is the system default.

9. 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.

10. Click **OK**.

11. The **BED** track is added into Circor Plot.

12. Shift + Right Click of the **BED** track to display the corresponding name.

13. Clear BED name(s) by clicking **Clear BED 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.
Whole Genome CNV visualization

The Whole Genome CNV is an interactive CNV visualization tool.

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

2. Select **Whole Genome CNV** from the drop-down list.

3. Whole Genome CNV shows up.

   The individual label copy number data points are plotted in blue or red. The segmented copy number line is in green and the copy number variant segments identified are shaded according to the variant type. Aneuploidy information is also marked on the bottom. Zoom in/out to see the scale of chromosome.

4. Click CNV or chromosome to navigate to **Genome Browser Viewer**.

5. Right click CNV plot to Show BED from the list.

6. Right click BED track to Configure BED Filters.

   **BED Filter Settings** dialog box appears. Please see “Circos Plot visualization” for more details.

7. Right click BED track to Remove BED.

Genome Browser visualization

The Genome Browser Viewer is an interactive SV and CNV visualization tool for each chromosome.

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

2. Select **Genome Browser** from the drop-down list.

3. Genome Browser viewer shows up.

4. At the **Anchor** field, hange the view of other contigs and chromosomes in the same sample.

5. At the **Range** field, set the range of the coordinates on the same chromosome.

6. At the **Molecules** field, sort the molecules in the order of pack, start, end, confidence, length, label density.

7. At the **Confidence** field, view molecules in the genome maps depending on their levels of confidence. Users can also enter a specific value manually.

8. At the **Find Map** field, highlight the genome map by typing Genome Map ID here.
9. The **Cytoband** information is shown in the black-and-white banding pattern with centromere in red. The displayed range is marked in red box.

10. By default, **CN** track is shown as a smooth light blue 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.

11. **SV** track marks the location of each SV.

12. **Ref** track displays the reference information of the selected chromosome or contig. By default, it is shown

13. **Map** track displays the genome maps that are aligned to the selected chromosome or contig. By default, it is shown as blue background with aligned labels in dark blue and unaligned labels in yellow.

14. Right click **Ref** or **Map** track to select the following options:

<table>
<thead>
<tr>
<th>Option</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>Hide</td>
<td>Hide the selected reference or genome map.</td>
</tr>
<tr>
<td>Hide the Others</td>
<td>Hide the other reference or genome map(s).</td>
</tr>
<tr>
<td>Hide (Show) Ruler</td>
<td>Hide or Show ruler of the reference or genome map.</td>
</tr>
<tr>
<td>Hide (Show) Matchgroups</td>
<td>Hide or Show matchgroups of the reference or genome map.</td>
</tr>
<tr>
<td>Show BED</td>
<td>Select BED file (Only available for <strong>Ref</strong> track).</td>
</tr>
<tr>
<td>Collapse</td>
<td>Collapse multiple Map tracks into one.</td>
</tr>
<tr>
<td>Invert</td>
<td>Invert the orientation of the selected reference or genome map.</td>
</tr>
</tbody>
</table>

15. If matchgroups or maps are hidden, click the **Show All** icon to view them again.

**Genome Maps Viewing Features**

**Insertion**
Deletion

Inversion

Translocation

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) and the haplotype-specific region (labeled as either 4qA or 4qB) are highlighted. Below is an example:

Complex Multi-Path Region Visualization

Bionano Access highlights Complex Multi-Path Regions. By default it is shown in mint green as below:

SV Filter

SV Filter icon is used to set up your own customized filter criteria. The filter criteria only displays on the top right corner of circos plot.

1. In Bionano Access, navigate to the Viewer screen of the object to analyze.
2. Click the **SV Filter** icon.
   The **Filter Settings** Window appears.
3. Click the **Filter by SV Type** Tab, and you can filter SVs by SV Type, confidence score and minimum size. For more details, please refer to Theory of Operation SV calling.
4. At the **Insertion** field, check the box to display, select one from **All, Recommended** or your customized filter from the drop-down list and type SV minimum size (bp).
5. At the **Deletion** field, check the box to display, select one from **All, Recommended** or your customized filter from the drop-down list and type SV minimum size (bp).
6. At the **Inversion** field, check the box to display, select one from **All, Recommended** or your customized filter from the drop-down list.
7. At the **Duplication** field, check the box to display, select one from **All, Recommended** or your customized filter from the drop-down list and type SV minimum size (bp).
8. At the **Intra-Translocation** field, check the box to display, select one from **All**, **Recommended** or your customized filter from the drop-down list.

9. At the **Inter-Translocation** field, check the box to display, select one from **All**, **Recommended** or your customized filter from the drop-down list.

10. Click the **General SV Filters** Tab, and you can filter SVs by chromosome and SV Masking.

11. 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.

12. 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 the recommended for human analysis if mask bed file is selected during de novo assembly and rare variant analysis.

13. Click the **Variant Annotation Filters** Tab, and you can filter SVs by percentage in Bionano control samples, chimeric score filter, self/control/parent molecule/assembly check, overlapping genes and self molecule count. It is only available for variant annotation pipeline. For more details, please refer to Theory of Operation Variant Annotation Pipeline 30190.

14. At the **SV in less than this % of the Bionano control samples** field, type the value. **100** is the system default.

15. 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.

16. At the **SV chimeric score filter** field, select one from **All SVs**, **Show Failed Chimeric Score**, and **Show Not Failing Chimeric Score**. It is only available for variant annotation of de novo assembly.

17. At the **SV control assembly check** field, select one from **All SVs**, **SV found in control assembly**, and **SV not found in control assembly**. It is only available for dual variant annotation pipeline.

18. 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.

19. 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.

20. 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.

21. At the **SV self molecule check** field, select one from **All SVs**, **SV found in self molecules**, and **SV not found in self molecules**.

22. At the **SV overlapping genes filters** field, select one from **All SVs**, **SV with overlapping genes**, and **SV
with no overlapping genes.

23. At the **Self Molecule Count** field, type the value. 5 is the system default.

24. Click the **Copy Number Variant Filters** Tab, and you can filter copy number calls by type, confidence score and minimum size. For more details, please refer to Introduction to Copy Number Variation.

25. At the **Copy Number Variant Type** field, select one from **All**, **Deletion**, and **Duplication**.

26. At the **Copy Number Variant Confidence** field, select one from **All**, **Recommended** and your customized filter.

27. At the **Copy Number Variant Minimum Size (bp)** field, type the value. **500000** is the system default.

28. Click **Apply** to apply the filter criteria.

29. Click **Reset Filter** to reset to the default settings.

30. Click **Show All SVs** to show all the SVs.

**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 of **Circos Plot**.

3. Click the **SV Tab**. This tab is only available in de novo assembly and rare variant analysis. This table includes Sample, Global_ID, Smap_ID, Type, Zygosity, 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 SMAP File Format Specification Sheet 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 of **Circos Plot**.

3. Click the **SV Annotation** Tab. This tab is only available in annotated de novo assembly and rare variant analysis or variant annotation pipeline. This table includes Sample, Type, Algorithm, RefcontigID1, RefcontigID2, RefStartPos, RefEndPos, Size, Zygosity, Score, SmapId, link SmapId, Present in % of BNG control samples, Present in % of BNG control 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, Found in self molecules. The columns can be sorted. Please refer to SV Annotation Pipeline File Format Specification Sheet for more details.

**Selected SV Tab**

Selected SV Tab is used to save all the manually selected SVs. During SV Report, selected SVs can be outputted in SMAP format.
1. In Bionano Access, navigate to the Viewer screen of the object to analyze.

2. Select Genome Browser from the drop-down list of Circos Plot.

3. Click Add to Report icon on the SV or SV Annotation Tab to add this SV to SV Selected Tab.

4. Click Selected SV Tab to visualize all selected SVs. The table on the Selected SV Tab includes columns of Smap_ID, Sample, Description, Type, Ref1_ID, Ref2_ID, Ref_Start, Ref_End and Size. All the columns are sortable and Type is filterable.

5. Select one SV in Selected SV Tab for further analysis.

6. Click Preview icon to visualize this SV. Users can modify the display and then click Camera icon to save the changes. It will be updated in SV Report too.

7. Click Edit icon to add or modify description. Then click Update icon to update or Cancel icon to cancel.

8. Click Delete icon to remove SV from the Selected SV Tab.

Generate SV Report

Reporting features have been added to the visualization page to allow users to share their findings with others during reviewing data.

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

2. Select Circos Plot from the drop-down list.

3. Click the Generate SV Report icon.

4. At the Generate and download the following files as field, select one from Filtered Variants and Selected Variants. Filtered Variants are structural variants based on filter criteria. Selected Variants are in Selected SV list.

5. The additional files offered will include all variants that meet the filter criteria active in your viewer. Check one or more boxes of Copy Number Variants (CSV Format), Structural Variants (SMAP Format), Annotated Structural Variants (SMAP Format), Aneuploidy File, Informatics Report and Structural Variants (VCF Format).

6. At the VCF Input Values field, Reference Accession and Experiment Id display. They are greyed out. Reference Access is the reference that used for the analysis. Experiment Id is the job Id in Bionano Access.

7. Click OK to generate PDF report.
The files will be generated and downloaded to your system. The PDF generated will include a screen shot of the circos plot complete with the legend, a screen shot of each structural variant, details on each structural variant, and general information about the dataset.

**Export to JPEG**

Users can download the current view in JPEG file format.

1. In Bionano Access, navigate to the Viewer screen of the object 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 AccessViewer.
4. Click OK.

**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 Non-overlapping BED row height (30-400px) field, type the value. By default the value is 150.
8. At the Copy number zoom threshold (30-400px) field, type the value. By default the value is 5.
9. At the Max copy number to display (0 - 100) field, type the value. By default the value is 8.
10. At the Track name width (80 px) field, type the value. By default the value is 80.
11. At the Gap between molecules (0-40 px) field, type the value. By default the value is 2.
12. At the SV row height (2-10 px) field, type the value. By default the value is 2.
13. At the Minimum gap between rows (10-40 px) field, type the value. By default the value is 10.
18. At the Maximum gap between rows (40-200 px) field, type the value. By default the value is 70.

19. At the Font size (%) field, type the value. By default the value is 100.

20. At the BED color opacity (%) field, type the value. By default the value is 60.

21. At the SV color opacity (%) field, type the value. By default the value is 40.

22. At the Raw copy number opacity (%) field, type the value. By default the value is 100.

23. At the Ruler text rotation (degree) field, type the value. By default the value is 30.

24. At the Hybrid cut flashing duration (msec) field, type the value. By default the value is 5000.

25. At the Circos plot SV size field, type the value. By default the value is 0.6.

26. At the Circos plot translocation width field, type the value. By default the value is 0.5.

27. At the Hybrid cut color field, select the color from the palette.

28. At the Background color field, select the color from the palette.

29. At the Highlight row color field, select the color from the palette.

30. At the Selected map border color field, select the color from the palette.

31. At the Selected label color field, select the color from the palette.

32. At the Molecule panel color field, select the color from the palette.

33. At the Molecule color field, select the color from the palette.

34. At the Reference map color field, select the color from the palette.

35. At the Genome map color field, select the color from the palette.

36. At the NGS map color field, select the color from the palette.

37. At the Matchline color field, select the color from the palette.

38. At the Label coverage color field, select the color from the palette.

39. At the Lasso color field, select the color from the palette.

40. At the Highlight molecule color field, select the color from the palette.

41. At the Highlight label/matchline color field, select the color from the palette.

42. At the Highlight matchgroup color field, select the color from the palette.

43. At the Highlight molecule matchline color field, select the color from the palette.

44. At the SV track color field, select the color from the palette.

45. At the SV insertion color field, select the color from the palette.
46. At the **SV deletion color** field, select the color from the palette.

47. At the **SV inversion color** field, select the color from the palette.

48. At the **SV translocation color** field, select the color from the palette.

49. At the **SV duplication color** field, select the color from the palette.

50. At the **CMPR color** field, select the color from the palette.

51. At the **Copy number color** field, select the color from the palette.

52. At the **High copy number color** field, select the color from the palette.

53. At the **Low copy number color** field, select the color from the palette.

54. At the **Nt.BspQI enzyme** field, select the color of matched label from the first palette, and unmatched from the second one.

55. At the **Nb.BssSI enzyme** field, select the color of matched label from the first palette, and unmatched from the second one.

56. At the **DLE-1 enzyme** field, select the color of matched label from the first palette, and unmatched from the second one.

   You may see more enzymes if other enzymes are added into your system.

57. Click **Save**.

   The new settings are saved.

58. [Optional] To revert back to the original settings, click **Reset Options**.

### View Options

Users can show or hide unmatched labels, copy numbers, molecule coverage, molecule match lines, and choose stretched alignment for molecule labels when viewing assemblies.

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

2. Click the **View Options** icon.

   The dialog box appears.

3. At the **Show unmatched labels** option, switch on and off to disable and enable.

4. At the **Show matched labels** option, switch on and off to disable and enable.

5. At the **Grouping highlighted molecules** option, switch on and off to disable and enable.

6. At the **Auto align highlighted molecules** option, switch on and off to disable and enable.

7. At the **Show molecule matchlines** option, switch on and off to disable and enable.
8. At the **Show molecule coverage** option, switch on and off to disable and enable.

9. At the **Show BED track in overlapping mode** option, switch on and off to disable and enable.

10. At the **Show BED names in non-overlapping mode** option, switch on and off to disable and enable.

11. At the **Show SV track** option, switch on and off to disable and enable.

12. At the **Show SV query individually** option, switch on and off to disable and enable.

13. At the **Show copy numbers** option, switch on and off to disable and enable.

14. At the **Show copy numbers masked BED** option, switch on and off to disable and enable.

15. At the **Show CytoBand** option, switch on and off to disable and enable.

16. At the **Show CytoBand on circos chromosome** option, switch on and off to disable and enable.

17. At the **Show tooltips** option, switch on and off to disable and enable.

18. 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.

19. At the **Show match lines in** option, click the drop-down list and select **Enzyme Color**.

20. Click **Close**.

**Refresh**

Users can refresh the current view.

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

2. Click the **Refresh** icon.

3. Refresh the current view.

**Return to Project Browser**

Users can return to Project Browser.

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

2. Click the **Return to Project Browser** icon.

3. Return to Project Browser.

**Home Page**

Users can return to Home Page.

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

2. Click the **Home Page** icon.
3. Return to Project Browser.

## Key and mouse shortcuts

Users can use key and mouse shortcuts when viewing results.

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

2. Click the **Key & Mouse Shortcuts**  icon.

   The dialog box appears.

3. Here is the list of highlighted feature and description. You can find more information in Access.

<table>
<thead>
<tr>
<th>Feature</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Customize the view of the samples</strong></td>
<td>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.</td>
</tr>
<tr>
<td><strong>Move maps horizontally</strong></td>
<td>Left-click on mouse; move left or right.</td>
</tr>
<tr>
<td><strong>Move maps vertically</strong></td>
<td>Press Shift; move up or down.</td>
</tr>
</tbody>
</table>

### 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.

### Zoom in or out

Use the mouse to scroll zoom in or zoom out.

## Create a Container for Two or More Assemblies

Users can view more than one assembly at the same time in the **Viewer** screen. Additionally, users can create a container for two or more assemblies so that they can be viewed at a later time.

The following task is an example of creating a container for two assemblies.

1. In Bionano Access, navigate to the **Viewer** screen of the project to analyze.
2. Click the File menu icon, and then click Select Alignments.
3. In Alignments, select the check box of the second alignment assembly to view.
4. Click OK.
5. Click the File menu, and then click Save.
6. At the Container Name field, type the container name.
7. [Optional] At the Tags field, type the keywords to associate with this container.
8. [Optional] At the Description field, type a brief description.
9. Click Save.
10. Return to the Project page.
    The container that contains the two assemblies appears in the Objects list.
11. Select the container name, and then in the Options pane, select the view container option.
    The Viewer screen shows the two assemblies together.

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 table includes XmapId, RefId, QryId, Ref_Start (Kbp), Ref_End (Kbp), Qry_Start (Kbp), Qry_End (Kbp), Orientation, Confidence. The columns can be sorted and filtered. Please refer to XMAP File Format Specification Sheet for more details.

Aneuploid 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 of Circos Plot.
3. Click the Aneuploid Tab. It contains per-chromosome aneuploidy call and includes columns of Type, Length, Score and FractCN. The columns can be sorted. It is only available for Access 1.4 or higher. Please refer to Introduction to Copy Number Analysis 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 of Circos Plot.
3. Click the Copy Number Tab. It contains columns of Id, Start (Kbp), End (Kbp), Size, Type, Fractional
Copy Number, Copy Number and Confidence. The columns can be sorted. Please refer to Introduction to Copy Number Analysis for more details.

Summary Tab

1. In Bionano Access, navigate to the Viewer screen of the project to analyze.
2. Select Whole Genome CNV from the drop-down list of Circos Plot.
3. Click the Copy Number Tab. It contains columns of RefId, % Deletion and % Duplication. The columns can be sorted. Please refer to Introduction to Copy Number Analysis for more details.

Repeat Tab

1. In Bionano Access, navigate to View EnFocus™ FSHD Analysis of the project to analyze.
2. Click the Repeat Tab. It is only generated 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

1. In Bionano Access, navigate to Maps to NGS with Conflicts of the 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 Hybrid Scaffold Conflict Cut Status File Format for more details.
**In Silico Digestion**

*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 by 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 **In Silico Digestion**.

2. Click the **Configuration Settings** icon.
   - The icon is at the top right-corner of the screen.
   - The *Configuration* dialog box appears.

3. At the **Minimum Labels** field, set a value between 0–20 (default as 0).

4. At the **Minimum Length** field, set a value between 0–250 kbp (default as 0).

5. At the **Number of Channels** field, set the number of channels to digest. The channel refers to the laser color used.

6. At the **Number of Enzymes per Channel** field, set the number of enzymes per channel to digest.

7. Click **Save**.
   - The software displays the settings in the *Create New Run* pane.

**Enzymes Management**

Users can add and edit labeling enzymes and recognition sequences; however, the default enzymes listed in Bionano Access are not editable.

Users must have administrator privileges to perform this task.

1. From the Bionano Access main menu, select **In Silico Digestion**.

2. Click the **Enzyme Management** icon.
   - The icon is at the top right-corner of the screen.
The `Manage Enzymes` dialog box appears. Here is a description of Enzyme list.

<table>
<thead>
<tr>
<th>Info</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>Name</td>
<td>The enzyme name.</td>
</tr>
<tr>
<td>Recognition Sequence</td>
<td>The recognition sequence.</td>
</tr>
<tr>
<td>Type</td>
<td>Bionano or user identified.</td>
</tr>
<tr>
<td>Matched Label Color</td>
<td>The color.</td>
</tr>
<tr>
<td>Unmatched Label Color</td>
<td>The color.</td>
</tr>
<tr>
<td>Experiment count</td>
<td>The count of experiments using this enzyme.</td>
</tr>
</tbody>
</table>

3. Click **Add**.

   The `Add New Enzyme` dialog box appears

4. At the **Enzyme Name** field, type the enzyme name.

5. At the **Enzyme Sequence** field, type the recognition sequence of the labeling enzyme (ACGT). The maximum length for the enzyme sequence is 20 nucleotides.

6. At the **Matched Label Color** field, select the color from the palette.

7. At the **Unmatched Label Color** field, select the color from the palette.

8. Click **Save**.

   The software adds the enzyme to the list.

9. Select one enzyme with the Type as User from the list. Then click **Edit**.

   The `Edit Existing Enzyme` dialog box appears.

10. At the **Enzyme Name** field, type the enzyme name.

11. At the **Enzyme Sequence** field, type the recognition sequence of the labeling enzyme (ACGT). The maximum length for the enzyme sequence is 20 nucleotides.

12. At the **Matched Label Color** field, select the color from the palette.

13. At the **Unmatched Label Color** field, select the color from the palette.

14. Click **Save Changes**.

15. Select one enzyme with the Type as User from the list. Then click **Delete**.

   The `Delete Existing Enzyme` dialog box appears.

16. Click **Yes** to delete this enzyme.

**FASTA Management**
Users can use *in silico* Fasta Files Management to manage FASTA files that have been uploaded into the *in silico* digestion tool.

Users must have administrator privileges to perform this task.

1. From Bionano Access main menu, select **In Silico Digestion**

2. Click the **Fasta Management** icon.

   The icon is at the top right-corner of the screen.

   The *Fasta Files* list and *In Siloco Digestion Runs* for every fasta file appears. Here is a description of Run list for every fasta file.

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

3. Click **Delete Fasta** to delete the selected Fasta.

   The *Confirm* dialog box appears.

   Click **Yes** or **Cancel**.

4. 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 channels parameters to select.

Bionano Access saves the FASTA files that have been digested. Bionano Access only checks if there are duplicate file names. If a user uploads a FASTA file that Bionano Access already has saved, the software will point to the existing file. For best practices, give FASTA files unique names to easily track them.
The following task is an example of an *in silico* digestion for one label channel.

1. From the Bionano Access main menu, select *In Silico Digestion*.

2. In the *Create New Run* pane, at the *Select file* field, browse to the FASTA file to digest, and then click *Open*.

3. At the *Label Channel 1*, select the enzyme from the drop-down list.

4. At the *Label Channel 2*, select the enzyme from the drop-down list (optional).

5. 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 your FASTA, upload, and try again.*

### Runs in Progress

Expand the *Runs in Progress* pane to view the following statistics:

<table>
<thead>
<tr>
<th>Statistic</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>Run</td>
<td>System-generated number for the run.</td>
</tr>
<tr>
<td>Date</td>
<td>The date the file is uploaded.</td>
</tr>
<tr>
<td>FASTA</td>
<td>The FASTA file name.</td>
</tr>
<tr>
<td>Min Labels</td>
<td>The user-defined setting for the minimum number of labels per map.</td>
</tr>
<tr>
<td>Min Size</td>
<td>The user-defined setting for the minimum map length.</td>
</tr>
<tr>
<td>Enzyme(s)</td>
<td>The enzyme(s) that are used.</td>
</tr>
<tr>
<td>Progress</td>
<td>The progress of the run.</td>
</tr>
<tr>
<td>Cancel</td>
<td>Click x to cancel the file upload.</td>
</tr>
</tbody>
</table>
**Completed Runs**

Expand the *Completed Runs* pane to view the following statistics:

<table>
<thead>
<tr>
<th>Statistic</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>Run ID</td>
<td>System-generated number for the run.</td>
</tr>
<tr>
<td>Date</td>
<td>The date the CMAP is generated.</td>
</tr>
<tr>
<td>Files FASTA</td>
<td>The FASTA file name.</td>
</tr>
<tr>
<td>Files Cmap</td>
<td>The CMAP file that can be downloaded.</td>
</tr>
<tr>
<td>Files Key</td>
<td>A file to track CMAP IDs that are associated with the FASTA sequence map names.</td>
</tr>
<tr>
<td>Files Gap</td>
<td>The GAP file can be downloaded. The GAP file track N-base gaps in sequence.</td>
</tr>
<tr>
<td>Files Summary</td>
<td>A complete summary of the results that can be downloaded.</td>
</tr>
<tr>
<td>Enzyme(s)</td>
<td>Enzyme(s) that are used to create the CMAP.</td>
</tr>
<tr>
<td>#Maps</td>
<td>Number of maps in the digested sample.</td>
</tr>
<tr>
<td>N %</td>
<td>Percent of genome that has N bases.</td>
</tr>
<tr>
<td>Sites / 100 kbp Ch1 Sites</td>
<td>The number of labels per 100 kbp in Channel 1.</td>
</tr>
</tbody>
</table>

**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.
Settings

In the Settings module, users can do the following:

- Add login banner
- Add System Beds and Genes files
- Change system settings:
  - Enable maintenance mode
  - Enable Compute On Demand
  - Add security settings
- Perform administrator tasks to manage User Accounts
- Add Control Databases
- Create system warning
- Add references
- Add and edit configurations
- Check queue status
- Add named filters

Login Banner

Users must have administrator privileges to perform this task.

1. From the Bionano Access main menu, select Settings.
2. Select Login Banner.
3. At the Content for login page field, type and edit the information.
4. Click Submit.

System BEDs & Genes

Users must have administrator or project lead privileges to perform this task. For BED file format, please go to P/N 30110 Theory of operation - Structural Variants Calling, P/N 30190 Theory of operation - Variant Annotation Pipeline, P/N 30210 Theory of operation - Copy Number Variation and P/N 30164 BED File Format Specification Sheet for more details.

1. From the Bionano Access main menu, select Settings.
2. Select System BEDs & Genes.
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 Bed 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 Pipeline object.

   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.

   CNVMask 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 object.

7. At the Genome Build field, select the genome build from the list.

8. At the File (*.bed) field, click Choose File, and then browse to select the bed file (*.bed) to use.

9. Click Open.

10. Click OK.

System Settings

Users must have administrator privileges to perform this task.

1. From the Bionano Access main menu, select Settings.

2. Select System Settings.

3. At the System Level Services field, please find the following options:

4. At the Compute On Demand & Saphyr Assure field, opt in or out Compute On Demand and Saphyr Assure.

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

5. At the Saphyr Assure Only field, opt in or out Saphyr Assure. Use this option if you want to ensure optimal performance of your instrument, but are not interested in the Compute On Demand service.
6. 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.

7. At the **Remote Access** field, opt in or out Remote Access. Enable this option to allow remote support personnel to access your environment for support and troubleshooting. With this option disabled there is no remote access to the Bionano systems.

8. At the **User Account Settings** field, please find the following options:

9. At the **Idle User Session Expiration [minutes]** field, it is 120 by default.

10. At the **Maximum User Concurrent Sessions** field, it is 2 by default.

11. At the **Maximum Login Attempts** field, it is 3 by default.

12. At the **Password Expiration [days]** field, it is 150 by default.

13. At the **Previous Password Retention [month]** field, it is 3 by default.

14. At the **Password Settings** field, please find the following options:

15. At the **Password Validity Check** field, enable or disable validity check.

16. At the **Validity Period [days]** field, type the number of validity days if validity check is on.

17. At the **Minimum Characters** field, it is 1 by default.

18. At the **Minimum Numerals** field, it is 0 by default.

19. At the **Minimum Special Characters** field, it is 0 by default.

20. At the **Minimum Case Changes** field, it is 0 by default.

### User Accounts

#### New User

Users must have administrator privileges to perform this task.

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 **User Name** field, type the user name.

5. At the **Full Name** field, type the full name.

6. At the **Email Address** field, type the email address.

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:
   - User
   - Project Lead
   - Administrator
   - 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. Click **Submit**.

    The new user account appears on the **User Accounts** screen.

12. Select one account to **Edit**, **Delete** or **Unlock**.

**Control Database**

Users must have administrator or project lead privileges to perform this task. For Control Database format, please go to P/N 30110 Theory of operation - Structural Variants Calling, P/N 30190 Theory of operation - Variant Annotation Pipeline, P/N 30210 Theory of operation - Copy Number Variation.

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 during *de novo* assembly or Rare Variant Analysis object.

CNV Control Database is used for CNV calling during *de novo* assembly or Rare Variant Analysis object.

ECNB (Expected Copy Number Baseline) is used for CNV calling to set the baseline of copy number during *de novo* assembly or rare variant analysis object.

7. At the **Operation Type** field, select one from **Assembly** or **RareVariant**. It is only applicable if **SV** is selected for **Control Type**.

8. At the **This is a human control database** field, check the box if applicable.

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 Warning**

Users must have administrator privileges to perform this task.

When administrator want to post a warning message to all user, system warning can show the message to all user.

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**.

**References**

Users must have administrator or project lead privileges to perform this task.

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. You can also click the **Add Enzyme** button to add a new enzyme if needed. You can select more than one enzyme in the list 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** check box.

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 a *de novo* assembly or hybrid scaffold, 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
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.

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 you have more than one SGE cluster in your Access configuration, Queue Status page will list the status of each SGE cluster. This feature offers a quick way to better understand the compute load on your 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.
Compute On Demand

How to enable Compute On Demand

To enable the Bionano Compute On Demand within Bionano Access, you can opt in in Compute On Demand in System settings (see above for more details). After that, it will pop up Bionano Compute On Demand Activation window:

![Compute On Demand Activation Window](image)

If you are not ready to complete the opt-in dialog, you can 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 the 2 fields in the Compute On Demand setting page. Please note that once the information of these fields are submitted by clicking on "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 your email. For example if your email was jsmith@bionanogenomics.com, we would recommend setting your organization to 'bionanogenomics'.

The second prompt is for the computing region. We currently offer three computing end points: North America, 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 your compute job. Users are not restricted to selected region, but selecting a region closest where your 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 appear in the Project Browser. In the Project Browser the operations panel will display different operations users can perform based on the object type selected. When Bionano Compute On Demand is enabled, there will be an additional panel visible below the Options panel (image below). The Bionano Compute On Demand options are listed separately since they require tokens. Like the existing Operations panel the hyperlinks on the Compute On Demand panel will change depending on the type of object selected. If a local Saphyr Compute is not configured, then the Compute On Demand operations will be the only ones displayed. 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.

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 (image below). To proceed users must have sufficient number of 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 your token balance. When the job completes, 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 12 tokens upon completion, users would receive a message informing that 2 tokens have been returned to token balance. Users are guaranteed that the cost will not exceed the maximum token cost quoted.
Tokens to execute jobs on our Bionano Compute On Demand service can be purchased through your designated sales person or by contacting orders@bionanogenomics.com.

### Compute On Demand Options

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

#### Redeem Vouchers

The use of Compute On Demand requires users to redeem their vouchers for tokens. The following rules apply:

- Each voucher contains different number of tokens.
- Jobs submitted to Compute On Demand requires a certain number of tokens depending on the type of job.
- Vouchers can be purchased from orders@bionanogenomics.com

Click the Redeem Vouchers button to add tokens to your account.

1. **Redeem Vouchers** is 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.

   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.

5. Click **Redeem Vouchers** to redeem.
6. Select **Transfer Tokens**.
7. At the **Your Current Token Balance** field, display your current token balance.

8. At the **Organization Token Balance** field, display all the users and their token balance, email address.

9. At the **Select Token Recipient** field, select the user you want 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.

10. At the **Token To Transfer** field, type the token amount that you want to transfer.

11. Click **Transfer** or **Reset**.

12. Select **Voucher Jobs**.

13. You can also expand the voucher row to see the corresponding jobs for every voucher.

14. Select **Voucher Balance**.
   
   This view displays the balance for each voucher. You can expand the voucher row to see which individuals currently possess tokens from the selected voucher.

**Token Use Recommendation**

- Only use as much amount of data as needed for the application, which is defined in the various theory of operations Bionano

- Use the data down sampling tools available within Bionano Access to help achieve a targeted coverage

- Provide good quality references; this reduces costs by eliminating the need for the pipeline to first generate a rough assembly.
Appendix

Red Labeled Sample Experiment

The Access system 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 Genomics 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.
Technical Assistance

For technical assistance, contact Bionano Genomics 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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<tr>
<td>Email</td>
<td><a href="mailto:support@bionanogenomics.com">support@bionanogenomics.com</a></td>
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<td></td>
<td>Monday through Friday, 9:00 a.m. to 5:00 p.m., PST</td>
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