



A Sysmex Group Company

OGT Handbook

Interpret Cloud User Guide

Interpret Cloud User Guide v1-20241029095209

For Research Use Only; Not for Use in Diagnostic Procedures

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Accessing Interpret Cloud

Following deployment of an installation of the Interpret software, OGT support will provide users with:

- 1. The URL of the Interpret deployment (<u>https://web-app.*.interpret-ogt.com</u>, where "*" is a name specific to the deployment. E.g. <u>https://web-app.mylab.interpret-ogt.com</u>).
- 2. A user name (e.g. "admin")
- 3. A password for the user name.

To access Interpret:

1. Open a web browser and navigate to the URL provided. A screen like the following may appear, indicating that the software is loading. This may take a few minutes.



Figure 1: The Interpret start page, indicating that the software is loading

2. Once the software has loaded, a login screen like the following will be displayed. Enter the user name and password provided, and click **Log In**.



Figure 2: The Interpret login page, displayed when the software has loaded



Access Restrictions

Please note that it is possible to configure Interpret such that it is only accessible from specific IP addresses. If the message "You do not have permission to access this resource" is displayed instead of the loading or login screens, please contact OGT for support.



Automatic shutdown

In order to optimise use of computing resources, the Interpret web interface will shut down automatically after a period of user inactivity (between 10 and 20 minutes). When this occurs, a message like the following will appear at the top-right corner of the screen:

Server connection lost, trying to reconnect...

To access Interpret, simply refresh the page and login again when prompted. Please note that processing of samples by the analysis pipeline is unaffected by this shutdown.

Uploading FASTQ Files

There are currently 2 methods for providing FASTQ files to the system:

- 1. Via the AWS web "console".
- 2. Via the "Upload FASTQs" page in the Interpret software.



UMIs

If the intention is to run UMI processing on the FASTQ files, they must have beeen generated with UMIs included. If UMIs are not included then the analysis will not complete.

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Uploading via the AWS web console

To access the AWS web console, which provides the most reliable upload mechanism, the following information will be provided:

- 1. A URL to the upload page (similar to <u>https://s3.console.aws.amazon.com/s3/</u> <u>upload/ogt-data-mylab?region=eu-west-2&prefix=incoming/</u>)
- 2. An AWS account ID (a 12-digit number)
- 3. A user name
- 4. A password

To upload FASTQ files to the system:

- 1. Navigate to the upload page URL in a web browser.
- 2. Select IAM user.
- 3. Enter the Account ID provided and click Next.
- 4. Enter the user name and password provided and click Sign in.
- 5. Click either Add Files (to select FASTQ files) or Add Folder (to select a folder containing FASTQ files), and select the appropriate file/folder from the file system.
- 6. Scroll down to the bottom of the page and click Upload.
- 7. Upload progress will be displayed in the next page. Do not navigate away from this page until the upload is complete, otherwise it may fail.
- 8. When the upload is complete, confirmation will be displayed on the page.
- 9. To verify that all FASTQ files have been added to the system, in Interpret, select **B atches** -> **Run Batch**, and check that they have appeared in the samples table.



Automatic Sample ID

In order to monitor changes in Variant Allele Frequency between samples from the same source in different batches, all samples must be assigned the same Sample ID when uploaded into the system. When FASTQ files are uploaded via the AWS console, the system will automatically extract a Sample ID from the name of the FASTQ file using the **standard naming convention**. If necessary, rename the FASTQ files before upload to ensure they are assigned the correct Sample ID.

Uploading via the Interpret software

Once logged in to Interpret, to upload FASTQ files to be processed by the system:

- 1. Click on the **Batches** button in the toolbar and select **Upload FASTQs**.
- 2. Click **Select FASTQ Files**, select the FASTQ files from your file system and click **Open**. The FASTQ files should be automatically paired and listed in the **Paired FASTQs** table.
- 3. If necessary, modify the Sample ID assigned to each pair of FASTQs by clicking on the button in the **Sample ID** column, typing the correct name of the sample in the input field, pressing Enter and clicking **Done**.



Note that, in order to monitor changes in Variant Allele Frequency between samples from the same source in different batches, all samples must be assigned the same Sample ID when uploaded into the system

- 4. Click Upload Paired FASTQ Files.
- 5. Click **Ok**.
- 6. Click No.

Progress of the upload can be monitored in the **Upload FASTQs** page, and an **Estimated Time Remaining** for all selected files to be uploaded is provided, which will be dependent on the total size of the FASTQ files and the upload speed.



While the upload is in progress, please note the following:

- 1. It is essential that the user does not navigate away from the page before upload is complete.
- 2. If using the Chrome web browser, ensure that Upload FASTQs tab is the one selected in the browser window, as selecting another tab will result in the upload being paused.

User Interface

The Dashboard View

The dashboard view displayed below comprises 3 sections:

- Menu Bar
- · Dashboard buttons to provide function shortcuts
- User account options

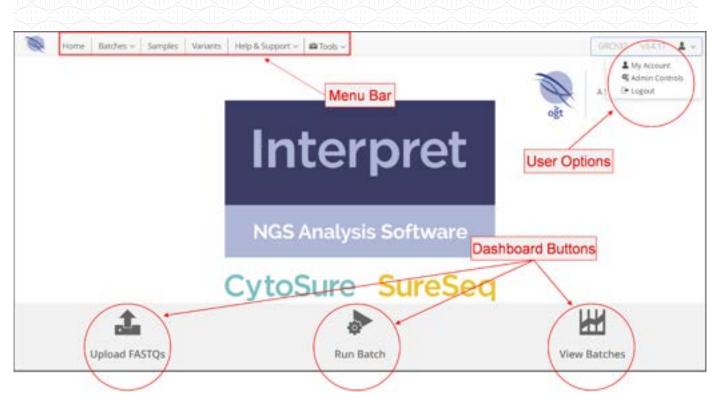


Figure 3: Annotated view of the dashboard

Menu Bar

The menu bar provides access to the functionality:

- Home Link back to the Dashboard View
- · Batches Setting up and reviewing analysis batches
- Samples Sample related functions
- Variants Provides a means to view all data from a variant centric view
- Help & Support A means to provide feedback as well request support
- Tools Access to any additional tools

Home Batches - Samples Variants Help & Support - ATON	~
---	---

Figure 4: The menu bar from the dashboard

Dashboard Buttons

These provide shortcuts to the common actions required by users.

- Upload FASTQs Select and upload FASTQ files.
- Run Batch Run an analysis of a batch of loaded sample files
- View Batches View the results of the batch analyses

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1	>	1
Upload FASTQs	Run Batch	View Batches

Figure 5: Shortcut icons on the dashboard view

User Options

The User Options drop down menu gives the user access to a range of administration tools. Additionally this section of the dashboard displays the build of the genome being used as well as the version of the software. In this case it is GRCh37 and v3.3.61.

The drop down options are as follows:

- My Account Your account details
- Admin Controls Additional options described in detail in the Admin Options section of the guide
- Logout Return to the Login page

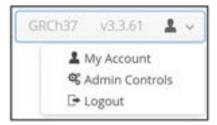


Figure 6: User account options

Viewing Samples

Once the FASTQ files have been uploaded the samples will be available to view in the Samples page

Accessing this is via the Samples button on the dashboard menu bar shown in the figure below.



Figure 7: Selection of Samples from the Dashboard menu bar

Samples and status are displayed on the left hand side of the window and when a sample is selected further information is displayed on the right hand side.

When a sample is first loaded there is no additional information present.

H 4 Page 1 of	1 (1 - 10 of 10) + HH Page Size	25			
Sample	Status		10384		
10004		Status	To Do	~	
10384 8210	To Do To Do	Mother		~	
7408	To Do	Father		~	
6937	To Do	Runs			¥
5881	To Do				
4315	To Do				
14130	To Do				
12878	To Do				
11516	To Do				
10847	To Do				

Figure 8: The initial view of samples in the Sample page

Samples can be searched by entering a part of the sample name in the search box. In the example below all samples containing "10" are displayed.

II Samples						
HI I Page 1 of	1 (1 - 3 of 3) ► ► Page Size: 25 ~					
Sample	Status					
10						
10384	To Do					
8210	To Do					
10847	To Do					

Figure 9: Searching for samples containing 10

The status of a sample can be updated. When first loaded, the status will be set to "To Do" and will be updated to "Running Pipeline" once processing has begun. Once sample processing is complete, the status will be further updated to "In Review". Users can assess the results if the analysis and manually update the sample status to "Completed" as required.

Sample	10384	
Status	To Do 🗸	
Mother	To Do	
	Running Pipeline	
Father	In Review Completed	
Runs		~

Figure 10: Modifying the status of a sample in the Sample view

It is also possible to specify the mother and father of a sample if they are also loaded in Interpret.

Sample	10384			
Status	To Do	~		
Mother	1	~		
Father	10047			
Runs	10847 11516	-	~	
	12878			
	14130			
	4315			
	5881			
	6937			
	7408			
	8210			

Figure 11: Specifying the mother of a sample

Initially, before any analysis, the run drop-down list will be empty.

Sample	10384		
Status	To Do	~	
Mother		~	
Father		~	
Runs			~

Figure 12: A sample that has not been processed yet having no run data listed

When a sample has been analysed, each run can be accessed from the drop-down menu. Each run will have a set of data which is displayed by 3 tabs. These are for general run information, QC metrics and results of the analysis.

The General tab displays basic information about the analysis and provides a link to batch view.

)384					
Status	in Review	~				
Mother		~				
Father		*				
Runs	14 Jan 2021 12	:14:27	~			
General	QC Metrics 8	lesuits				
Date	14 Jan 2021	12:14:27				
Batch	CytoSure N	GS Batch 00001				
Status	Completed					
Panel	CytoSure N	GS Panel				
Protocol	Default Pro	tocol				
ISCN	seq[GRCh3]	7] (1-22,X)x2				
FASTQ read	10384_7_L0	01_R1_001.fastq.gz				
FASTO read	10384 7 10	01_R2_001.fastq.gz				

Figure 13: Viewing the General tab for a sample run

The QC Metrics tab gives an overview of the metrics of the sample. The data will be colour-coded according to the metric set that was defined for the analysis protocol.

There is further information on metric sets in the section of the guide that covers the admin options.

Sample	10384				
Status	In Review	~			
Mother		~			
Father		~			
Runs 14 jan 2021 12:14:27		2021 12:14:27			
General	QC Metrics	Results			
% Reads	Aligned		% Duplication	Mean Target Coverage	1
99.63			8.03	314.04	
Targets N	lot Covered		% Usable On Target Reads	% Usable On And Near Target Reads	
136.0			73.08	82.55	
Off Targe	t Reads		% Reads Mapping Quality 0	Average Quality	
17.45			2.71	33.1	
Average I	Insert Size		Insert size std	Eveness	
206.2			66.3	87.5	
Uniformi	ty		Sample Sex	# Exon Targets Not Covered	
1.348			0.0	15.0	
# SegDup	Exon Targets Not	Covered			- 1
15.0					

Figure 14: Viewing the QC Metrics tab for a sample run

Finally, the Results tab provides links (in green) to download files from the analysis as well to view (in blue) the different variants that have been detected.

	10384					
Status	In Review	~				
Mother		~				
Father		~				
Runs	14 Jan 2021 12:	14:27		~		
General	QC Metrics R	esults				
View S	iNVs		View CN	Vs/LOH Calls	View Translo	cations
BAM	Downlo	ad	BAI	Download	QC	Download
/CF	Downlo	ad	CGH	Download	Translocations	Download
.og	Downlo	ad				
	nerate Report					

Figure 15: Viewing the Results tab for a sample run

Adding User-defined Variables

In order to enable the user to capture and report custom information related to samples processed in Interpret, the admin controls section provides a means to create variables of different data types via Admin Controls > Analysis > Manage Samples > Variables.

🔢 Manage Samples	
Oven/kw VirUbles	
Manage Sample Variables	
+ Add New Variable Category	
Save Changes R Discard Changes	← return

Figure 16: The manage samples page in the Admin Controls

Selecting **Add New Variable Category** provides a text box to name the new variable and clicking **Add** adds a new sub-tab to the **Variables** tab.

Create	new Variable	Category	×
Name	Additional		
	🖺 Add	× Cancel	

Figure 17: Creating a new variable category named Additional

With the new category called "Additional" generated, users can create variables associated with the category by selecting **Add New Variable**.

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Manage Samples
Verlables
Manage Sample Variables
Actitional
Add New Variable Remove Empty Category
+ Add New Variable Category
E Save Changes X Discard Changes + return

Figure 18: An empty custom category named "Additional"

To delete an empty category, click the **Remove Empty Category** button. To create new variables in the category, click the **Add New Variable** button, assign a **Name** to the variable, confirm the **Category** with which it should be associated, and select the appropriate data type from the **DataType** drop-down box, then click the **Add** button.

Name	Phenotype		
Category	Additional	~	
DataType		~	
	+ Add	× Cancel	

Figure 19: Creating a new variable named "Phenotype" in a category named "Additional".

Add New V	ariable		×
Name	Phenotype		
Category	Additional	\sim	
DataType	Text - Multiple I	Line 🗸	
	Integer		
	Floating Point	Number	
	Percentage		
	Text - Single Lir	ne	
	Text - Multiple	Lines	
	True or False		
	Date		
	Date and Time	8	
	List - Select On	e 🚽	

Figure 20: Selecting the appropriate data type for the new variable - in this case, "Text - Multiple Lines"

Once a variable has been created, it will be listed, along with its data type, in the appropriate category in the **Manage Sample Variables** section. To delete a variable, click on the cross next to the variable name.

W Ma	anage Samples	
Overview	v <u>Variables</u>	
Ш м	lanage Sample Variables	
Additio	init .	
[Add New Variable Phenotype (Text - Multiple Lines) ×	
+ 4	Idd New Variable Category	

Figure 21: A custom field named "Phenotype" listed under the "Additional" category

Having been created in the system, custom fields may be populated for each sample in the **Samples** view, and will also be displayed in the sample run page whenever the sample has been processed in a batch (accessible by clicking on the sample row in the **C ompleted Samples** table in the **Batch Overview** page).

II Samples		
14 Page 1 of	10-20-020 P. H. Age Not 25 -	
Garges	344	Sample Sample-1
		Status Hi Review V
CR007-012	It Revew	Mother Sample-1
CR007-008	to Review	
CR007-001	It Review	Refer Sample-1 v
C8007-008	ti Review	Num 20 Oct 2021 13:58:16
CR007-011	IT Review	General QCMency Results Additional
C8007-009	ti Reveni	
CR017-017	In Review	Wenotype The phenotype has been defined for this patient:
C8007-010	At Review	HP10000488 Retinopathy Noninflammatory retina disease HP10000156 Retinal dystrophy
CR017-002	II Review	
CH007-005	II Revex	
Sample-4	In Review	Instant C Yes 🕡 No
Sample 3	at Revex	
Sample-2	It Review	Et law
Carryne F	1.1	

Figure 22: Editing the content of the "Phenotype" field in the Samples page

Interpret also provides a framework enabling the development of plug-ins to import sample data in bulk from other sources, such as spreadsheets, text files or a LIMS. If you are interested in importing data in bulk, contact OGT – a suitable plug-in may be available, or it may be possible to develop a plug-in to satisfy your requirements.

Running an Analysis

On the dashboard either select "Run Batch" in the drop down from the 'Batches' menu item.

A	Home	Batches ~	Samples	Variants	Help & Support 🗸	Tools 🗸
		L Upload	i FASTQs e Copy FASTQ	ks		
		🕭 Run Bi	itch			
		W View 8	atches			

Figure 23: Selection of Run Batch from the Dashboard menu bar Batches drop down menu

Or, click on the 'Run Batch' icon on the dashboard page

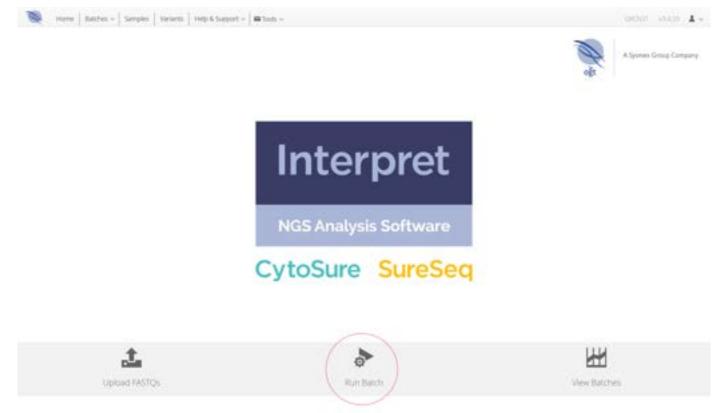


Figure 24: Selection of Run Batch from the dashboard short-cut buttons

Either choice leads to the initial Run Batch page is as follows:

Running an Analysis

Run Ba	ch			
atch Name *	3530-01-0717144-08	Panel *	ur ∈ Protocol *	
munt for farm	nin te large D			
10.1	Symple ID	AND TO Read 1	NUTU Next 2	
a 1.	5881	5881,7,1001,81,001,feotuga	5881,7,0001,82,001.fexsu.gz	
0.2	6827	8807,7,1001,81,001.feeta.gp	6837_7_0801_82_001.5aves_ga	
H 8.	7408	7408,7,1001,81,001 Aning gr	7408,7,0001,82,001 Aexts gp	
1. 4.	8210	8210,7,0001,81,001 Avenue	8210,7,5005,82,001.6xmg.gt	
= S	10384	10384,7,1001,91,801.famoj.gz	18384_7,1001_92_001.fwtmp.gz	
1. 6.	10647	10847_7_L001_R1_801.fax89_81	10847,7,1001,82,001.fwite.gr	
11. 7	11576	11516_1_U001_81_001.faxes, gx	11516,7,1001,82,001.fwin,gt	
- +			10000 0 - 101 00 001 0	
nacted Samples	Sample II	Habits Read 1	Author Read 2	

Figure 25: Initial view of the Run Batch window

Besides showing the list of available samples there are additional text fields and drop-down menus.

In order to run an analysis the user needs to

- 1. Select samples for the analysis
- 2. Select the correct panel for the samples
- 3. Select the analysis protocol

Pun Batch	e Sele	ct Panel	Select Proto	col
Batch Name * _ appoint on Fight of	Fanal *	7	- Protocol +	
Search for Samplin by Sample D	Search for Sample	s		

Figure 26: Input fields for the Run Batch window

Optionally users can specify a name for the batch analysis. A default batch name is provided with the date followed by the time in the format YYYY-MM-DD HH:MM:SS.

In the example below the user has created the batch name CytoSure NGS Batch 00001

🏶 Run Batch			i i i i i i i i i i i i i i i i i i i
Batch Name 1 Cycolum NG3 Bech 00001	Panel *	w Protocul *	19
Search for langular to langue (b)			

Figure 27: Entering a batch name

The samples have been processed with OGTs CytoSure NGS panel so that is the selection to make from the Panel dropdown menu.

	Panel *	Protocol +	
🏕 Run Batch			

Figure 28: Selecting the appropriate Panel

The user now specifies the protocol that will be used, in this case the Default Protocol.

Aun Batch				
Batch Name * CytoSure NGI Batch (0001	Partel * Cytology MIS Partel	- Protocal *	BECKER WEITER	
South for Samples by Sample (S			Default Womani Venant Protocol	

Figure 29: Selecting the protocol to use for processing the batch



Panel-Protocol Compatibility

Only protocols whose **Pipeline Type** are included in the list of pipeline types supported by the selected **Panel** will be listed in the **Protocol** drop-down list. Additionally, if any **Pipeline Capabilities** supported by the protocol are not supported by the selected panel, a warning will be displayed indicating which processes will not be run.

Lastly, the user specifies the samples to be analysed.

There may be a large number of samples loaded into the system, so to enable easier sample selection it is possible to add a search term. In this case the user is looking for all samples containing the number 5. Additionally, search terms are independent of the case used.

ð Ru	n Batch			
Batch Name * Cytochure NGS Batch (6001		Panel * Constant ALL Revel	w Protocol * Default Protocol	
1				
4.1	R.F. Cheyell	HOD Real (NOT PARTY.	
10	4 1881.	1001,7,201,01,201Aerage	MMIT, T JOHY, N2, OHY Triving (
	8 1998a	+1510_3_2200_00_001_b00_b00_pp	111110,7,1201,82,501 fwhigt	
	10 4015	4115,7 (401,81,001.5em.pt	4315,7 Little A2, bit tamage	

Figure 30: Filtering loaded samples with a search term

Selecting the checkbox next to a sample moves a loaded sample into the Selected Samples table.

Running an Analysis

9.1	tun Ba	lun					
latch	atch Name * Cytofune INGE Barch 00001			Fanal * Cytobure MGS Panal		w Protocol * Extent Protocol	0
-	h ta jan	Giving Sumply D					
		Sept.1.		INCOMENTATION AND A DESCRIPTION OF A DES	INCIDENCE		
		1 Ages		- Sent 2 add (p) just here ar		sant 7,1007,02,000 Annuger	
	5.	1004		10384,7,200,25,305,5anugr		102864,7,1001,32,001 family gr	
11	λ.	1060		10407, F.; 200, Jr.; 200, hum, gr		1047,7,001,82,001 family	
	+	11916		11114_7,200.01_00.001/weige		11816_F_001_RE_001.tem.gt	
	1			Construction of the second s		Contraction in the second	
41.18	d Samphes						
-		Sargin C.	ALC: No.		(A)(2) (a)		
=		2441	180 (7,502,50,502 here		1481,7,1287,92,287,944,87		
	1	407	3627,7,201 97,301 Mag		4447,7306,70,905 Aug.g		
	1	1428	1418, 1, 101, 31, 301, here		1406_7_000_81_001-benug		
	4	1414	1011_1/101_01347344	P	4210, 1,200,30 (51 Autor)		

Figure 31: Adding a sample to an analysis batch

Clicking on the minus icon 🗧 will remove the sample from the Selected Samples tables.

				and a second			_
atch Name * Cyschure (403) Bakm (0000)				Panel * Conduce Mills Panel		Prosocal * Delaus Protocol	
Silan	to Ser	give by Sample D					
*	10.4	Tampie III		Fed http://www.ti		EASTIN local 2	
	1	5481		Stat, J, Jort, 41, Millionaga		sent (, cent, Ad, etc. terrage	
		6607		NRT 7 JOH (R) MITHING &		AND TANK READ WANTED	
				100.7.1.001.01.001/ming.gt		Tells, Collector, McCollector, and Annual and	
				8210,73.001,81,001.0wnegt		ATTER A LINE AD ANT Annual	
				1984 (Color Jonger		10164,7,1001,92,001 Seringge	
		1040		UNKEY CARE AN ADD AND AND A		19847, 7, 1091, 93, (41, 164), ggi	
				11116_Compromising		1935a, 7 July (42,001,004) gr	
darla.	Lamater	ina -					
		Servela (0	Exciting thread 1		(ASTQ Read 3	£	
1	1	2467	INT.CONTROLMAND		teetr, F, Letter, Md, Jerr, Naves, gp.		
U	inter D	ngter frankrisk i	WITCOM UNDER SHEEP	é.	4107,7,300,90,000 Average		
-	1	7408	7406,7,3007,81,001.5empg	1.5 C	1408_1(001_R0_001.hemgs		
8	4	8/14	829,7,399,91,391,645.pt	çi.	4119,7,1001,92,001 Renge		
	3	10384	10384,7,5301,81,801,8416	*	10004, 7 Julion, NO, Juni, Bernarge		
	4	10847	TOWN, T, LANS, M. (1997) Average	F	ead-of, P., Mart, AQ, Jan, Sarraga		
	1	11114	11014_7_1001_01_001.0010	r .	11116,7,5804,	1,52,301 herey gr	
8	8	10878	12076,7,1001,81,001.5e4q		10878,7,0001	(Action Reside	

Figure 32: Removing a sample from an analysis batch

When all selections have been made the run can be started by selecting

Run Batch Batch Name * Oyusture 1603 Batch 00001 Protocal * Detaut Protocal Partel * Opsidure MSI Partel and the second 1001 and install 00000 instrument and an and INCOMPANY. No. 1, Str. A. W. Song P. \$70,7387.00.00 Aug pt APR, SARE AS, MY MILLION inert (1,000,00,00) terrup 000 1047,7,10131,31,301041pt DERIGTION ACTIVATES

Figure 33: Starting an analysis

If the selected protocol has "Enable CNV and LOH Calling" set to "Yes", CNVs will be detected by comparison with a set of reference samples which need to be defined in the protocol as either "All Batch Samples" or a specific set of reference samples whose FASTQ files have already been uploaded and designated to the system by the user.

In the latter case, OGT may provide a set of data files that can be used as a reference set for CNV analysis. As more samples are processed users may extend the reference pool by adding any samples they believe are suitable as controls for CNV calling. A user can modify samples designated as reference pool in the protocol in Admin Controls-Manage Samples-Protocols.

If CNV calling is enabled without a reference data set being defined, then, on

	The selected Protocol includes CNV calling but no references
	have been added.
Invalid Protoco	
	Please update the protocol with the required references or turn
	off CNV calling.

Figure 34: Error message for using an invalid protocol

Click on the message to remove the warning and select Admin Controls > Analysis > Protocols to set reference samples. More details are in the Protocols section of this User Guide.

Otherwise, a popup presents the chosen files and selected parameters. Following this there is a request for confirmation and upon confirmation the analysis run will be initiated.

Larve .	CytoSure NGS Br	Rip 00001						
ureSeq Panel	 DytoSure NGS Pa 	ne -						
votocel	Default Protocol	Default Protocol						
amples (10)								
0	Sargini O	MCQ Real 1	1401Q Baat 3					
1	5887	5881_T_L001_R1_001.helte.gz	5881_7_L001_82_001 faste gr					
2	6337	6937_7_L005_R1_001 Autogst	4937_7_1001_R2_001 failing gr					
3	7406	7408_7_L001_R1_001.factorgr	7400_7_L001_R2_001.fastra.gz					
4	8210	#210,7,1001,81,301.tetta.gt	#218,7,0001_#2,001 twing gt					
5	10364	10384,7,1001,R1,001 tastagr	10384_7_L001_92_001 failing gt					
+	10647	10847,7,1001,R1,001.tarts.gt	10647,7,1001,82,001 faits.gr					
7	(1316	11516,7,0001,81,001.tastuggt	11516,7,1001,#2,001 fasts.gz					
1	12878	12878_7_L001_R1_001.featugg	12676,7,1001,92,001.temisgr					
9	14130	14130_7_L001_R1_001.textq.gt	14130_7_L001_R2_001_MMq.gs					
10	4315	4315_7_001_R1_001 htmggt	4315,7,000,83,001 fairs gr					

Figure 35: Window requesting confirmation to run an analysis

Selecting will start the analysis and the display will change to show information about batch being analysed.

Within this there is an overview window providing an overview of the analysis and a sample window giving information about the status of each sample.

Running an Analysis

Batch - CytoSune NGS Batch 0001		Overview window	
* Depines			
	en 2020 12:17:38	2	
D 🔒 File Statur 🖌 Copy Santh 🔳 Stop Santh			
tariyin Matr	Unge Waiting to start		Program
4927	Waiting to start		
7406	Weiting to start	R	
K210	Weiting to start.		1.15
10384	Warting to start		
	Waiting to start.	1	1
10647	Waiting to start		
11516	Waiting to start		
10847 11556 12878 14130	Waiting to start Waiting to start		

Figure 36: The batch processing view with the overview window and sample window highlighted

Initially the status of the samples will be listed in the overview window as "Waiting to start"

Controller			
Openatur address Balan 2020	12.17.00		
Parent Cytochaire NGS Parent Instant Mailting to	e start		
Pressed Default Protocol Multiple Resident Casherently	un examples		
teres and the second second second			
🗅 🐞 File Status 🖡 Copy Batch 🗰 Stop Batch 🗟	Developed and Property		
argin .	hap	Ingen	
588T	Mailing to start		
NRET .	Maining to start		
1408	Waiting to start		
	Waiting to start		
1210	Waiting to start		
1210 Ciller			
1270 12984 12647	Waiting to start		
1210 1084 10847 1556	Waiting to start Waiting to start		
10000 10000 10000 10515 11516 12078	Waiting to start Waiting to start Waiting to start		

Figure 37: Initial batch status before analysis starts

Waiting for a reference to be generated

If a reference pool needs to be generated the status shown in the batch overview will report this and provide a means to track progress of the reference pool creation.

¥ Oversee		Villevi	
Operator	admin	Dete	8 Jan 2039 14:51:53
tent :	CytoScire NGS Panel	toms	Pre-processing Reference Samples - 1/9, Alignment:
Permit	Dafault Protocol	MalifyC Report	Currently usualable
¥ Outroom			
tpears	admin	0400	8 jan 2828 5451:53
field	CytoSure NGS Panel	Itatos	Pre-processing Reference Samples - 1/8, Counts
Protocol	Default Protocol	MultiQC Report	Currently unavailable

Figure 38: The analysis status in the overview window, highlighted, showing reporting the status of pre-processing of the reference samples

The status of reference building can also be tracked in the View Batches window which is discussed in the View Batches section of this User Guide.

ны в	atches							
Actors	for selected. (at comm							
- 26 .	Same	Panel .	Present	Number of Langes	Status.	Det .		
							() () () () () () () () () () () () () (
- 10	Cytofium NGS Batch 0001	CytoSure NGS Panel	Default Protocol	10	Pre-processing Reference Samples - 5% Algorment	8 jan 2020 143	9153	

Figure 39: Reference building status being shown in the View Batches window

If the protocol performs CNV analysis and samples in the analysis are to be used to generate the reference pool against which to make CNV calls then the overview will report the combining of the reference samples.

Once the reference samples have been aligned and counted, they are combined into a pool for the CNV analysis

₩0mmen		
Penal	admin Cytobure NGS Panat Default Francial	8 Jan 2001 14:51:53 Pre-processing Reference Tamptes - Containing Currently unavailable

Figure 40: The analysis in the over window, highlighted, reporting the combining of the reference samples into a pool

Samples will be queued until there is capacity available in the pipeline. Once this is available the software will start processing the samples sequentially. The stage of the process is updated and the overall progress can be monitored in the progress bar.

♥ Overvees			
Operator adverse	bes.	Rjan 2020 14:51:53	Overview of sample analysis
Pariet Cytoflare NGS Pariet	Dates	Running - 6/10 samples completed	Je
Pressore Darlandt, Prostocol	MultiQC Separat	Currently unavailable	
			Progress of current sample
O 🔒 File Statum 🌢 Copy Bant	n 🖬 Stop Batt	R Germanner	
tanyo		1 Mage	Property
5881		Running - FASTQ File QC	*
6817		Waiting to start	

Figure 41: The batch view showing progress of analysis

Once analysis started the stage of each sample is displayed and can be followed

tangin	Trap.	Properti
5481	Running - FASTQ File QC	
Sample	Toge	Propessi
5881	Running - Courts	
Sample	Rep	Property
5881	Russning - Target Coveráge	
tanges	Stept	Popos
5881	Running - Variant Calling	
lungit	Tesp	Propess
5681	Running - Variant Annotation	
faingle	Tage	Propess
5881	Bunning - LOH calling	
langin	Bage	Progenia
5881	Running - CNV Calling	
tample	tage	Program
5881	Running - Importing Results	
6937	Running - Initialization	

Figure 42: Tracking progress of a sample processing

Once a sample has been analysed the overview updates the count and a summary of the analysis is displayed in a Completed Samples table.

Running an Analysis

Batch - CytoSune NGS Batch 0001						
* Derve					sis	
Operanai admin Dara Penal Cytoflaire MIS Parlet Status (Process) Default Process) Mudroc Hysert	II Jan 2020 1451:51 Burning - 1/10 samples completed	Overview	w of sample	f sample analysis		
O & Ne Status & Copy Banth & Stop Ban Semple	alt 🖉 German Halant -		Program.			
SAUT	Compreses					_
8937	Runging - Courts		-			-
2117	Warding to start					-
7408						
8270	Watting to start					_
10084 Completed san	nple Watting to start					
Completed san	wating to start		-			
Completed san	wating to start Wating to start Weing to start		:			
10004 Completed san	wating to start		:			
E270 TODA4 TODA7 TTST4 TURTU TURTU TURTU	wating to start Wating to start Weing to start		:			
E270 TODA4 TODA7 TTST4 TURTU TURTU TURTU	waiting to start Waiting to start Waiting to start Waiting to start Waiting to start		-			
8270 TOBA TOBA TOBA TOBA TOBA TOBA TOBA TOBA	Notice to start Waiting to start Waiting to start Waiting to start Waiting to start Waiting to start					
8250 10084 10847 11516 12878 14738 8275	Nple Waiting to start Waiting to start Waiting to start Waiting to start Waiting to start Waiting to start Waiting to start	s # CMIS		Ingeri	84	

Figure 43: The first completed sample is displayed below the samples to be processed

When all samples are completed

herries .										
Carrier adm	-	Date	# Jan 2020 14 51:53	Overvi	iew of sam	ple analysi	is			
funal Cyne	(liere Nil)	3.Panel Sunsi	Completed							
Protocol Data	nuk.Frank	and shoo	Chapter 3 Matters			0	ompleted s	amples	1	
		S Copy Batch II	Ing Turch 🔋 Generate Report				1		-	
man had when	Complet	ted Gamples					+			
Housed, view	-	famples fample	Vere		4 (99)	# CNIN	rates	August	QK.	3
	_		ter tour Let	•	# 1900 2,754	# CNIR	e almo 16	Aspert	6K 1980	3
independente .	1	Sample								3
independente .		Lampie 1681		• vel	2,754		16		1.000	1
independente .	1.1	lampin 5681 6937	The Scene Ave a		2,754 2,695	# 13	16 15	*	-	
independente .		See1 6837 7428	The Douse Art 1		2,754 3.685 2,740	8 13 7	16 15 12	:	11	
independente .		Sear Sear 6837 7428 K250	The Doub Ave a transmission of the transmissio		2,754 1,695 2,740 2,666	8 13 7 10	16 15 12 16	÷	1111	2
independente .		Sample S681 4937 7438 8210 10384	The Desire Art 1 The Desire Art 1 The Desire Art 1 The Desire Art 1 The Desire Art 1		2,754 3,685 2,740 2,666 2,690	8 13 7 10 4	16 15 12 16 17	:	11	
independente .		Sergin Selat 6837 7438 8210 10384 10384 10847	The Desire Art 1 The Desire Art 1		2,754 2,885 2,740 2,666 2,650 2,669	8 13 7 10 4 5	16 15 12 16 17 13		10 10 10 10 10 10 10 10 10 10	
independente .		Sergin Selat 4827 7408 8210 10384 10384 10847 71516	The Doub Ave a The Doub Ave a		2,754 2,895 2,349 2,666 2,660 2,669 2,571	8 13 7 10 4 5 7	16 15 12 16 17 13 18		10 10 10 10	

Figure 44: An analysis with all samples analysed

There is no need to wait until all samples have been processed to view the results for a completed sample.

This will be discussed in the Viewing Analysis Results section of the manual.

Viewing Analysis Batches

On the dashboard either select "View Batches" in the drop down from the 'Batches' menu item.



Figure 45: Selecting View Batches from the menu bar drop down menu

Or, click on the 'View Batches' icon on the dashboard page

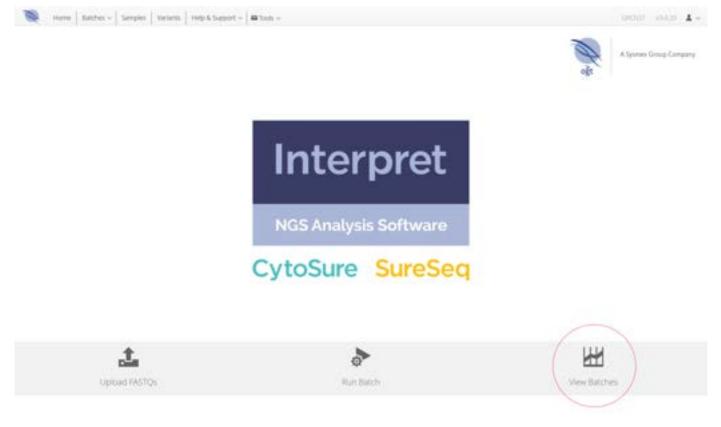


Figure 46: Selecting View Batches from the dashboard shortcut buttons

The Batches are presented in a table as below.

	atches An another at some								
	Name .	Fanal	Frank	Number of Tangets	Table .			100	
					1		2.0		,
-	CytoSure NG5 Batch 0002	CytoSure NISS Panet	Default Protocal	3	Mating to start	10 jan 2020 11	34:27	admin	
10	Cytoflame NGS Batch 0001	Cytofiare NGS Panel	Default Protocal	10	Completed	8 Jan 3530 145	153	admin	

Figure 47: Initial view of the Batches window

As with other tables in Interpret where there is a column selector icon 🗉 a user can add or remove columns from the display

Batches						Colum	n Selector	-	
Accord for selected:	-								
St. Aste	Farmel	Percei		Number of Samples	threat	0.000		THAT	-)*
			14				2.0		~

Figure 48: Column selection options for the Batches window

Column names annotated with a tick are in the current display and changes can easily be made to add or remove columns

~ 1	Name
✓ F	Panel
✓ F	Protocol
~ 1	Number of Samples
¥ 5	Status
- 0	Date
~ 1	User
5	Samples

Figure 49: Selection of columns to display in the Batches window

By default, all batches are presented in the first instance but these can easily be filtered.

Where the column header has a text field, users can type in a search term and all batches with that text contained somewhere in the name, will be retained. The text search is independent of lower- or upper-case letters, "Demo" will return the same samples as "demo".

Alternatively, where there is a drop-down menu selecting one of the values in the menu will lead only to the batches matching the selection being displayed, for example, below only batches that have completed will be displayed.

ctions	for selected X (most								
а.	have	Parel	Protocol.	Number of Sampley	Solut.	date:		iner.	
		1941	7.4		-		2.0		
8	Cytorilarie NGS Barton 2002	CytoSore NSS Panel	Default Protocol	1		10 per 2020 11	38.27	admin	
н.	Cytocharme NGS Barts In 2021	Cytoliure AIGS Panel	Default Protocol	-18	maning	II Jan 2020 143	1.53	admin	
					Completed				
					Falled				
					tilled Transforringfortur				
					Prepricessing				

Figure 50: Filtering batches on status

Lastly, there are date fields, allowing selection of batches run within a set time frame.

-	Ar selected a second											
й.	Rate	Aunal		Parent.	Summer of Samples	Natio					Uter	
			-	(÷			- w (2	0		
11	CytoSure NGS Batch 0002	CytoSure NGS Panel		Default Protociol	3	Completed		0	49 2020	1.91	admin	
11	Cytatione NGS Batch 0001	Cytoflure NGS Panel		Default Protocol	10	Completed		the large	24. 44 5	1.100	admin	
								N 10 3 8 7 8 10 14 10 10 27 20 10 38 30 1 4 7				

Figure 51: Filtering batches on date of processing

Deleting Batches

In the batch view it is possible to delete batches. When first opened there is a greyed out Delete button in the display.

If a batch is selected it is highlighted in blue and Delete button is now active, Clicking the delete button will delete the batch from the software.

	i Valli Valli V	ado Vado Vad	nValnVa	In Value	Zalla	Vala	adhValhVi	ada Vada Va	
Batches									
iners for salected (x uners)	-	Protocol	Number of Longins	Tarra .		Tate -		1041	
					1.4		×		
Batches			Delete						
tans for selected K inters	*	L							
i tana	Agent	Protocol	Assesses of Samples.	1044		Date		-	
						8	2 = .		
E Cynelwre MGS Result Mitz	Canadiane 1955 Percer	Defeat Presson	1.4	Component	1000	10 (44 2000 111	1944)	(1000)	

Figure 52: Selection of a batch to delete highlights the delete button

If the delete button is selected there will be a popup box requesting confirmation of the deletion.

Selecting ewill lead to the batch being deleted.

Once a batch is deleted it **CANNOT** be recovered.

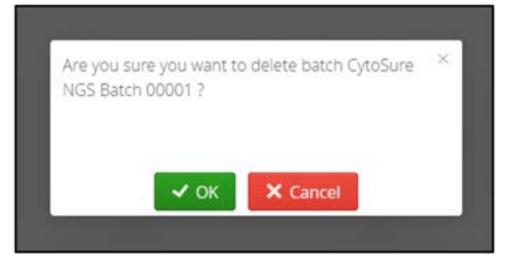


Figure 53: Popup box requesting confirmation of batch deletion

Individual Batches

Clicking on a row in the View Batches page will open a new page showing the selected batch in more detail.

There are 3 parts to the information provided,

1. Overview

The overview provides information about the analysis

2. Batch Functions

The batch functions allow users to download files form the analysis, repeat the analysis or generate a report

3. Sample Details

In this part there is the headline information form the run about each sample such as the number of SNVs and CNVs called.

Batch - Cytos	Sure N	GS Batch 00	0001		0	verview	1	Batch Fu	nctions	Samp	le results		
• Overview					/	/		- /					
	in Sure NGI wit Proto	iPanel 1	Sate Matus MuttiQC Report	Complete									
C File So	allocate and	Copy Eatch	R Sob P	en B	Generate Report	5		1					
in selected, view:		tample •	Vari				# 59/14	# CN/A	#104	# Travelocations	Report	QC.	
Shork-Indets	0-			E CHANNEL	-	1.1p	2,774	2	31	0		(ten)	
CW/s5DH Cafe	11-	10647	2.00	a contrai	B furnament.	• Logi	2,786	2	30	a		in the second	
Constant Case	04	11516	a sin	I chince	-	B logs	2,685	5	38	0		these .	
	- CL_	12878	8 1mm	a central	-	• top	2,731	6	34	0		. Steel	
	0-	14130	-	-	II fermature	₿ logi	2,734	4	31	0	а.	(Hew)	
	11_	4315	8 mil	Course .	-	 Imp. 	3,514	4	16	0		. Alexand	
	0-	5881	10 mm	-	-	∎ ug	2,883	\$	33	0		(these)	
	3_	6937	E 1995	E Central	-	B logs	2,815	8	34	0		See	
	0-	7408	II 1995		B 1	1 top	2,846	6	25	0		(1000)	

Figure 54: The sections of the batch analysis window

Batch QC

Included in the Batch page are two QC reports.

In the batch overview there is a link to a MultiQC report which gives an overview of all the samples that were in the batch.

Additionally, each sample in the completed table has a FastQC report for each read file.

Examples of both of these QC reports are shown below.

Batch - Cytos	Sure N	GS Batch 00	0001									
• Overview												
	in Gure NGS wit Posto	Panel 1	Sofe Katus WARQC Repor	Complete	-	_	Batch	QC		Sampl	le QC	
C File Sta		Copy Batch	II Sep I	en B	Generate Report						/	
Stonaste		tample .	Vari				# 555%	# CN/A	#108	#Thansfocations	Report	¢¢.
	0	10384		E CHINGH		 Logs 	2,774	2	31	0	. *	Net.
Variabacamore	0.	10847	8.00	E Couldi	-	a logi	2,786	2	30	0		- Face
Participation	0-	10847 11516	8 mm	E cheatrai	B formation	● Logs ● Logs	2,786 2,685	2 5	и я	0 0	*	-
Nartsbacamore	0-	10847 11516 12878	2 min 2 min 2 min	E Constant E Constant E Constant	E formant. E formante	LogsLogsLogs	2,786 2,685 2,731	2 5 6	30 39 34	0 0 0	•	111
Variabacamore	0-0-0-	10847 11516 12878 14130	8 m		E formation E formation E formation E formation	 Logs Logs Logs Logs 	2,796 2,685 2,731 2,734	2 5 6 4	33 38 34 31	0 0 0 0	•	1997 1997 1997
arteles arteres	0-00-0-0	10847 11516 12878 14130 4315	2 m		E formation E formation E formation E formation	 Logs Logs Logs Logs 	2,786 2,685 2,731 2,734 3,514	2 5 4 4	33 38 34 31 36	0 0 0 0	*	New New New New New
Variabacamore		10847 11516 12878 14130 4315 5881	2 m	E Creation E Creation E Creation E Creation E Creation E Creation	E formation E formation E formation E formation E formation	 Lop Lop Lop Lop Lop Lop 	2,796 2,685 2,731 2,734	2 5 6 4	33 38 34 31	0 0 0 0 0	*	1997 1997 1997
Sumburger		10847 11516 12878 14130 4315 5881 6837			E formation E formation E formation E formation	 Logs Logs Logs Logs 	2,786 2,685 2,731 2,734 3,514 2,883	2 5 4 4 5	33 34 31 36 33	0 0 0 0	*	Ner Ner Ner Ner

Figure 55: Links to QC reports for a batch and a sample

MultiQC

MultiQC is a reporting tool for the whole batch of samples. It parses summary statistics from results and log files generated by other bioinformatics tools.

When you launch MultiQC, it recursively searches through any provided file paths for specific files. These files are parsed for relevant information and used to generates a single stand-alone HTML report file. It also saves a directory of data files with all parsed data for further use downstream. To save MultiQC report to user's computer, right click on the page, and choose "Save as...".

Additional information about MultiQC can be found in the next section of this guide.

1.1	Quality	Control	Report										>		
a tan ng (tana) Tan (anan ng	Project some Pariet Generate bolt Monter of a Report pros	e lemant by: mytes in a balant.		Control on Section 1 Control on Section 2 Section Address 10 2000-01-08, 10-	land								og	t	
	Change sample in	ares Design	and State										A Sysmex Comp	any	5
them.	OGT's s	ummary t	able												
in the	& Day title	E Contanto Concesso	Are many	lane of Same											
a lanky	Surryte Servel	Mapped water	Explored rests	Ang matty	Arg inset size	Inset som ME.	MPGHE	Occupit		Othergel	Mean son.	Bet nevered	Evenness source	1.00.00	les.
10	10,540	10.025	Barn.	11.11	101.00	11.96	317%	21.07%	81.295	1115	26.0	141	8195	1.014	Farmer
	1,000	MARKS.	3.00	31.0	2000	1.31	1.79%	11404	31.875	1115	346.21	18	41276-	1.539	Ayesta
	1,140	9117h	88.79%	38.4	040.00 C	4630	1.00%	21.995	HIMS.	9405	1000	40	\$179m	1.108	Funds
	1,100	the larty	Arrs.	82.91	210.00	34.96	2.98%	dates.	BLUPS .	0.118	201.00	101	81 mu	1.100	Twee
	4,544	MARK .	Barry .	11.11	20430	44.00	2.97%	75.445	31375	191	106.13	00	81205	1.146	Tennis
	8,8am	01105	100	10.49	200.00	HO.	2.16%	15.00%	all times	10.00	14114	14	stars.	1:344	Farmer
	8,540	10.075	8.1m	32.91	20100	44.00	1,0%	20.62%	31.0Ph	0.05	211.09	141	#15m	1.349	Twiste
	T_ban	min.:	\$255	31.00	219.00		1105	70.07%	81.01%	1115	107.01	185	813/6	1.348	Family
		101.111.	8475	33.00	211.00	86.00	1345	ILM'S	HIMS	4 1/5	348.99	101	41304	1.201	Twiste
	8,5em	the state of the second s													

Figure 56: Example of a MultiQC report

Sample QC

FastP is used for sample QC data generation. Clicking on the	View	button on the
sample view will open up a new tab in the web browser with t		

1	
Summary	
General	
fastp version:	0.20.0 (https://github.com/CrenGene/fasts)
sequencing:	paired end (151 cycles + 151 cycles)
mean length before filtering:	147bp, 147bp
mean length after filtering:	1475p, 1475p
duplication rate:	8.1035194
Insert size peak:	173
Before filtering	
total reads:	32.384588 M
total bases:	4.776467 G
Q20 bases:	4.399816 G (92.114459%)
Q30 bases:	4.130451 G (86.475024%)
GC content:	47.035643%
After filtering	
total reads:	30.184820 M
total bases:	4.440014 G
Q20 bases:	4.185280 G (94.262757%)
Q30 bases:	3.945594 G (88.864444%)
GC content:	46.795308%
Filtering result	
reads passed filters:	30.184820 M (93.201606%)
reads with low quality:	2.094360 M (6.466751%)
reads with too many N:	52.744000 K (0.162858%)
reads too short:	54.664000 K (0.168786%)

Figure 57: Example of a FastP report

Batch Functions

Below the overview section there are a set of buttons providing a set of option – when the batch has finished processing the Stop Batch button is disabled.

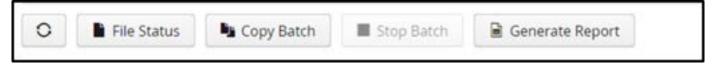


Figure 58: Batch options

File Status

File status provides shows the files that have been generated for each sample during the analysis.

Files provided are:

- 1. Alignment files
- 2. QC files
- 3. VCF files
- 4. CGH files for loading into CytoSure Interpret
- 5. Log files

Where a green tick is displayed, that file is available for download and this can be

achieved by clicking on the button.

+ feseni	a Batch Ball Down	interest in the second s						(
	Sample	840	84	er.	#1	- 104	ling .	Actions
21	5861	V Derivat	and the second second	and the second second	¥	and the second	V Dented	
22	6807	~	- Caretar	4 Easter	V Contract	×	af Descent	
23	7408	and the second	- Constant	and the second second	V Instant	and the second second	and the second second	
24	6210	V Trainer	af Dented	V Desited	V Contact	V Sector	V Inner	
25	10084	V Descent	and the second	and the state	of Contract	at Destail	and the second	
26	10647	~	and the second	and the second second	¥	* Inc.	~ E	
27	11516	V Decent	and the second	and the second	×	V Second	and the second	
28	12676	and a second	and the start	4 Testal	V Contain	and the second	V Dented	
29	14130	and the second	of Constant.	and the second	4 Constant	and the second	and the second second	
30	4315	V Daniel	and the second		V Territori	and the second	V 2000	

Figure 59: Status of files generated by the pipeline for each sample

It is possible to download all files, or selected files, simultaneously via the bulk download button

4		Sample	BAM	BAL C	qc 🗌	VCF 🛄	CGH	Log 🗌
21		5881	0		0	0		
22		6937						
23		7408				0		
24		8210						
25		10384			0			
26		10847						
27		11516			Ð			
28		12878		10				
29	0	14130					0	0
30		4315						

Figure 60: Bulk download file selector

Specific files can be selected as below

d		Sample	BAM	BAI 🗌	QC	VCF	сан	Log
21	4	5881	×		*	2		
22		6937						
23		7408		0				
24		8210						
25		10384						
26		10847						
27		11516						
28		12878						
29	:0	14130						0
30		4315						

Figure 61: Bulk download with single sample selected

Alternatively, all files can be selected for download

A Downlo	10							
id.	0	Sample	BAM 💌	BAI 💌	QC 💌	VCF 💌	сан 💌	Lúg 🕑
21	1	5881	2	8	1	2	2	2
22		6937					×.	
23	۲	7408			۲	۲	۲	
24	*	8210			-		2	~
25	۲	10384	2		۲	8		
26	۲	10847		×				
27		11516				2	*	2
28	۲	12878	1					
29	×	14130	8	8		×	8	
30	*	4315					2	

Figure 62: Bulk download with all files selected

Files downloaded in bulk can be grouped by sample or file type

ownload Selected (60) Grouped By	Sample 🔹
	File Type
	Sample

Figure 63: Bulk download selecting grouping by Sample of downloaded files

Copy Batch

This function allows the user to repeat the batch analysis with the same settings. When selected a Run Batch window opens and if the user selects to Run Analysis the processing will be repeated.

The software will automatically update the Batch Name but otherwise nothing is changed including the time stamp.

Viewing Analysis Batches

₽R	un Ba	tch		Updated Batch Name
atch	Name	Cytoflare MG3 Batch 00	TIGI Sandeq Panel * Costure MS Panel	w Pratocal * Default Protocal
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*		Dartyle IX	HITS fixed 1	PADQ Base 2
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			0007 / 1/001 / K1 /001 Reine get	entry Later, Manufactures ga
		100	1400,7,1001,01,001 Avenues	Table 7 parts (42,001 Array gal
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		106.01	1940, Spins Jr. Williams gr	Head / June 10, Set head go
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la la	Lampter	1997		
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=	4	8216	APROXIMITER AND A REAL	4214,7,000,42,000 hers,gt
	1	10000	10384,7,1381,91,001Annugr	46044,7,,684,30,891 hverugt
=	4	10647	10847_2_1407_01_001.0x44ajgr	Maket 2 (2009) (30) Anni (pr
=		11118	11216,7,1001,01,001,000,gr	11348, F. (001, A0, 011, Farry ga
-	8	1001	10070,23400,01,001Aerouge	1000(2).000.000.000.000.000

Figure 64: A batch analysis being repeated using the Copy Batch option showing the updated batch name

Report Generation

Report Generation shows a drop down in which the user can select the report to be generated

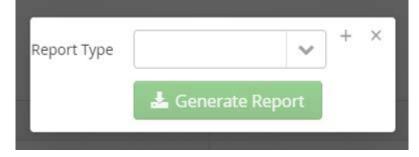


Figure 65: Initial view of the report options

Currently, the only template loaded is the Batch Report

Report Type + × Batch Report with template

Figure 66: Selecting a report type for the QC of the run

Report Type	Batch Report with	\sim	+	×				
Template	QC Report	~						
	🛓 Generate Repo	ort						

Figure 67: Selection of a template for the report

When the report is generated the output is a table with a set of metrics for each sample in the batch.

Sam ple	Percent Reads Aligned	Percent Duplicat ion	Mean Target Coverage	Targets Not Covered	Aligned Reads GC	Aligned Reads Per Base Quality	Usable On Target Reads	Usable On Target Bases
5881	99.4	43.3	536	0	40	37.4	53.9219	35.5563
693 7	99.4	43.1	627	0	40	37.4	55.4457	36.6627
740 8	99.1	50.2	440	0	40	37.3	39.6108	25.9758
821 O	99.1	50.7	418	0	41	37.3	40.9108	26.9532

Table 1: Example output of the QC report for a batch Selecting a completed sample or samples allows viewing of the variant information and this is described in Viewing Analysis Results section.

Viewing Analysis QC

• Overview Operator addr									
Operator addr									
	in .	Date 14 jan	2021 12:14:27						
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		TO A SAME T							
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		 Variation 		# Shirk	# CNM	#108	# Translocations	Report	QC.
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Surridozamon Gwintor Cata	18384 19847 11516 12878 14130 4315 5881		31 31 Termination 4 Logs 32 31 Termination 4 Logs 33 31 Termination 4 Logs 34 31 Termination 4 Logs	2,774 2,786 2,685 2,731 2,734 3,514 2,883	2 2 5 6 4 4 5	31 33 38 34 31 56 33	0 0 0 0 0 0	* * * *	100 100 100 100 100 100

Figure 68: Selecting a batch to view

Viewing Analysis QC

When a batch of samples is processed, besides individual sample metrics that were discussed in the previous section, there is a batch QC report generated. This uses MultiQC and fastp to collate a set of metrics for each sample and merge into a set of graphs and tables.

The report can be accessed from in the batch overview displayed once a batch has completed analysis.

Batch - C	ytoSure NGS Batch	00001	
• Contribut			
Operator	admin	Date	54 jun 2021 12:54:27
Panel	CytoSure NOS Pariet	Status	MultiQC Report
Protocol	Detail Protocol	MultiQC Report	· IMARCE REPORT
0	The Status 🕒 Copy Batt	a Baip	Luith 🔒 Generate Report

Figure 69: Accessing the Batch QC report

When the user clicks on the MultiQC Report link a new tab opens up in the browser displaying the QC report. The view is divided into 3 parts – the quality control report for the batch, which comprises the bulk of the display, and 2 tabs that come into the

view from the left and the right of the page. These tabs can be viewed and hidden by clicking on their respective buttons. The second tab provides the MultiQC toolbox for:

- 1. The Quality Control report for the batch
- 2. The report short cut tab
- 3. The tool box tab

At the head is the quality control report; this provides general information about the analysis such as the date of the analysis and which user performed it.

Quality Control Report		~
Napel Joseph Name Marchine Ladd Analyse participation Analyse participation Report parameters:	Sandwar HOT Hand HUTH Spatial and Hung Sandwar HUTH HUTH HANNE HUTH Sandwar HUTH Sandwar HUTH	oğt
		A Systems Group

Figure 70: Example batch overview details

OGT's Summary Table

Each sample has a row in the table with some key metrics.

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Figure 71: Example sample QC summary table

The column names from the summary table are listed in the table below with some additional detail as to their meaning.

Column Name	Description
Mapped reads	The percentage of reads that mapped to the reference genome
Duplicate reads	The percentage of reads that were duplicated
Avg. quality	The average read quality reported by Samtools stats
Avg. insert size	The average insert size reported by Samtools stats
Insert size std	The standard deviation of the insert size reported by Samtools stats
MPQ = 0	The percentage of reads that were mapped that have a mapping quality of O
On-target	The percentage of reads that map on target that are not duplicate reads
<u>+</u> 250bp	The percentage of reads that overlap target regions extended by 250bp

Interpret Cloud User Guide v1-20241029095209

Column Name	Description
Off-target	The percentage of reads that are neither on target nor within the specified flanking region
Mean cov.	The mean target coverage
Not covered	The number of targets with a coverage of less than 1
Evenness score	The fraction of the whole sequencing output that is correctly distributed
Fold-80	The fold of additional sequencing that would be required to ensure that 80% of targeted bases achieve the mean target coverage.
Sex	The chromosomal sex of the sample predicted from the distribution of reads that map to the sex chromosomes

Table 2: Column names and their description from the QC summary table

Targets Not Covered

Any targets not covered are detailed, providing that they are not within a segmental duplication.

Targets Not Covered Sector Uptote constraints and the term at any sector tables.

Figure 72: Example targets not covered summary

Coverage Efficiency

The efficiency of coverage as a measure of depth are displayed.

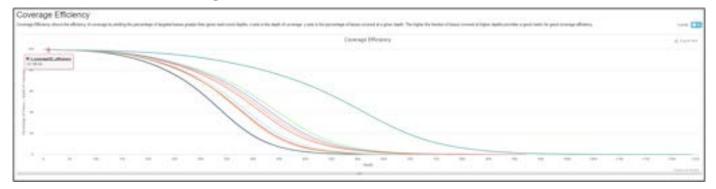


Figure 73: Example QC report summary

Insert Sizes Samtools

The distribution of insert sizes for each sample is displayed.

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Figure 74: Example QC report summary

Percent Mapped

The percentage of base calls at each position for which an N was called.

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Figure 75: Example QC report summary

Alignment Metrics

The alignment metrics for all the samples in the batch are plotted.

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Charlest period																					1.0					

Figure 76: Example of the alignment metrics

Filtered Reads

The filtered reads graph shows the number or percentage of reads that have been removed by the filter.

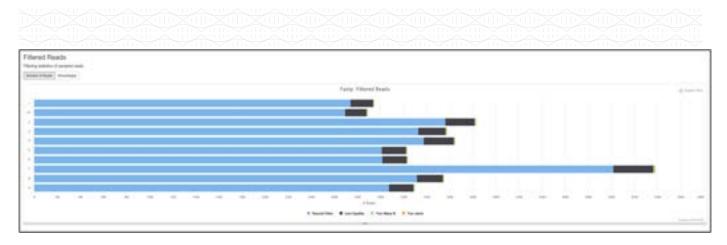


Figure 77: Example QC report summary

Duplication Rates

The relative level of duplication found for each sample as a percentage.

-		
	- \	

Figure 78: Example QC report summary

Sequence Quality

The mean sequence quality or Phred score of each base in the insert for each sample.

Sequence Quality	
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Figure 79: Example QC report summary

GC Content

Viewing Analysis QC

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Figure 80: Example QC report summary

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Figure 81: Example QC report summary

Sample QC

Sample QC information can also be access via the Sample page. For a particular run selecting the QC metrics tab will provide the relevant information. Colours are defined by the metric set used in the analysis protocol.

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		16.0		

Figure 82: Viewing the metrics for an individual sample

Interplay Interplay

Figure 83: Accessing the Sample QC report from the sample results tab

Summary		
General		
fastp version:	0.20.0 (https://github.com/OpenGene/fastp)	
sequencing:	paired end (151 cycles + 151 cycles)	
mean length before filtering:	146bp, 146bp	
mean length after filtering:	145bp, 145bp	
duplication rate:	7.220084%	
Insert size peak:	171	
Before filtering		
total reads:	28.905316 M	
total bases:	4.227335 G	
Q20 bases:	3.894346 G (92.122952%)	
Q30 bases:	3.658105 G (86.534532%)	
GC content:	47.4124128	
After filtering		
total reads:	26.944780 M	
total bases:	3.928181 G	
Q20 bases:	3.703276 G (94.274565%)	
Q30 bases:	3.492954 G (88.920399%)	
GC content:	47.151145%	
Filtering result		
reads passed filters:	26.944780 M (93.217386%)	
reads with low quality:	1.862392 M (6.443078%)	
reads with too many N:	46.288000 K (0.160137%)	
reads too short:	51.856000 K (0.179400%)	

Figure 84: Start of a FastP report for an individual sample

Viewing Analysis Results by Sample

Viewing a Sample

Access to the results from running the pipeline are described in the previous section "View Analysis Batches".

Within each batch are the samples processed in that batch comprising analysed variants and QC metrics.

SNW/Undets	10	View			#5N05	# CNVs	#1,0H	# Transfocations	Report	90	% Reads Aligned 18
fransiscations	10	E SAN E CANADA	II Termentary	 Logi 	7	2	0	1		Vater	99.52
CVM/LDH Calls	Ш.		E Vanishing	B top	5	2	0	1	*	Stee	99.47
	0.	E 144 E 1944124	-	8 Hep-	5	2	0	1		Stee	99.51
	. 8		E Internet	a tage	7	1	0	1		Van	99.49
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	Ш.,	E true E Constale	-	8 top	4	2	0	1		ver.	99.31
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Figure 85: View of a set of processed samples in the batch view

As with other tables in Interpret, where there is a column selection icon users can use it to configure which columns are being displayed.

Comple	ted Samples				Column S	elector		0
	Sergia	Vale	# \$503	# CNH	# LOH	Report	QC	>(=)
18	5881	E two E Charles & Kt B Lagr	2.754		16		Tex	4

Figure 86: Column selector button for configuring columns to view in display

The column options for this view are shown in the Figure.



Figure 87: Columns available for selection to display

There is one completed sample per row and for each sample there is a range of information available to view.

SNVs		CNVs	/LOH	Tran	sloc	ations	Lo	gs	Report		QC	
1			1		/		7		/			
For selected, view:	Contra	View	1	1		+5800	# 01/h	#10H	# Turoiscations	report	i)c	% Reals Algred II
Dahlacations	а.	-	Contracted	a Janita attant	1 top	7	2	0	1		Vese	98.52
ENVIAGE Catte	18.	# 1000	B constant	-	110	5	2	0	1		View	99.47
	11	III inter	E CONSULTS	II Summer	B Logo	5	2	0	1		Vee	99.51

Figure 88: Information available for each sample

Variants for a sample can be viewed by selecting the SNVs or CNVs/LOH links present in each row.

Multiple samples can be viewed simultaneously by selecting the check boxes of the required samples which will then activate the SNVs and CNVs buttons on the left hand of the view.

SNVs		Translocation		NVs/L	On				
		/	/						
		/ /							
For selected, serve		Samples		#3905	# 09/6	RICH.	# Transistations	% Raads Aligned	Aspert B
SNVs/Indets			a ser	7	2		10. 10.	-	
CNVs/LOH Calls			. inp						
	°в.	E In E Opploy E tomas		5	2	0	t.	99.51	
	11			7	1	0	1	99.49	
		# the # Constan # Terms		7	1	0		99.6	
	U.			4	2	0	1	39.31	
		B MALE DOMAGN	1 1 1 1 1 1 1 1 1 1 1 1 1 1 1 1 1 1 1	4	3	0	1	99.51	

Figure 89: Selecting multiple samples to view simultaneously

Once selected, the variants will be displayed on a Variants page.

γa	riants													- Neuro Serie	OWVOHO	n.,
	T Protocol Filt	er, Defaut Sky File													-	
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																10

Figure 90: The initial SNV/Indel display page

At the top of the page are buttons that allow the user to toggle between the CNV and SNV views.

	Aberration Type	Toggle Between V	iews
Variants 4	\square	₽ t100 ~	Result2Bach ON6/LOH Gels Tomocodorn
CNVs + LOH C	alls		- Return Elligith SNA Tonskoutions
Translocations	<i>[</i>		+ Return to Batch SNNs OWWADH Calls

Figure 91: Toggling between SNV, CNV and Translocation reports

The Variants viewer is divided into three parts:

- 1. The Protocol Filter, initially this will be showing the Protocol Filter used in the analysis. The filter is modifiable in the Admin Controls (Admin Controls > Analysis > Protocols)
- 2. The Variant Table, showing the variants, one on each row. In the header there is a drop down "Actions" menu options; these are discussed below.
- 3. The Integrated Genome Viewer (IGV) that has been embedded in the software. Further details on using IGV are below.

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12	1111	10.12.12	1.1	6.25	1.44		22 - 1 - 1 - 1 - 1 - 1 - 1 - 1 - 1 - 1 -	1.02.24	1.1.1.1.1.1.1		12.0012-		1.	100201000	-11 C C C C
1.0		. 10,44	676	-			aya nya	41,14			10,00	ma . ma		10.44	41.4 ··· ··

Figure 92: The sections of a sample results view page

Viewing SNV and Indel Events

The variant table has a column selector icon 🔳 allowing user to configure which columns are displayed.

There are different columns available depending on whether you are viewing the SNV variants page or the CNV/LOH variant page. The options for SNVs are displayed below.

Select Displayed Columns	# Ref Reads (-)
 HGVSc (Gene Symbol) 	# Alt Reads (+)
✓ Chromosome	# Alt Reads (-)
✔ Start	Ref Strand Bias
✔ End	Alt Strand Blas
✔ Ref	Reads Placed Left
🖌 Alt	Reads Placed Right
 Allele Frequency 	Most Severe Consequence
 Туре 	Impact
 Ref Depth 	Consequence Terms
 Alt Depth 	PolyPhen Prediction
 Total Depth 	PolyPhen Score
 Quality Score 	SIFT Prediction
 Ref Quality 	SIFT Score
 Alt Quality 	HGVSc
 Sample 	Canonical?
Length	rsiD
Genome Build	Minor Allele Frequency
Genomic Context	Minor Allele
Context Length	ClinVar Significance
HGVSc	Gene ID
HGVSp	Gene Symbol
Classification	Transcript ID
Genotype	Transcript Resolution Method
Zygosity	Protein ID
Inheritance	Exon ID
Log Ratio	Exon Number
# Ref Reads (+)	

Figure 93: Columns available to display in the SNVs variant page

Selection of a variant will load the alignment file in IGV allowing review of the alignment.

A range of variants can be displayed and examples of each of these are:

1. SNV

Viewing Analysis Results by Sample



Figure 94: Example of a SNV being displayed in the IGV browser

2. Deletion

	Lange V.	A HOUSE SHOW SHOW	Construction of the	Cart W	106 5	ours.		Alleis Programs W.	1000	Bard Dapath W.	All Depth #	Table Depitt #	for their P	and Southly T	Donate Science
	1441	elwiperstal alter-6	1.1	TTERALIZED	TRALING	1		11.076	ANV	- 180	180	246	14445-000	4287.00	AREL AT
		ABOUNDED11-1109-1100		1110010040	THE PART	m m T		2 87 9 89 C	- Desire	1.00	1.1		and 12, 500	11100.000	STATE OF
	1881	ARHIGAPOLIA, HEARDA		115254711	118284711			bi date	5AV		246	-	Neet.m	4345.00	80799.011
	5680	ARHIGAPICIE*11367-6		TTRECHART	119413465			45.78%	589	198	168	168	67463.000	3608.00	3879,54
	1887	ABHGAP213.*988,*988	1	110413249	119412249	- A -	AL.	48.55%	everteet.	244	198		8,027,89	11716-00	00/10.04
	3887	AUDIC CONTRACTOR	1.	VIDENALDS.	VIDENEZ38			91.716	1994	1	942	945	12.88	118AE.00	10000.00
	1441	HENDAFIDA TORFO-6	1	115416545	1941034	- 1		15.47%	5NW	148	104	204	- 4891.00	6162.00	80140
	5681	ARMAPSTY. MCH.		115754867	1157548067	- E		44,00%	SNV	967	- 687	254	40653.00	1004.00	3683.25
	1001	ARHIGAPITU,*1234T-1		115417502	115417503	1		31.79%	58%	192	183	10	5200.00	5546.00	205.10
٩.,	3997	ARNIGAPOLL*1198218		119413438	111413430	1		47,016	SNV	100	19	100.	1074.00	6002.00	423.84
10	W 167 (11)		аталтис 0 ,еза											0 6	
	-					100	100								
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								-				-	-		

Figure 95: Example of a deletion being displayed in the IGV browser

3. Insertion

Viewing Analysis Results by Sample

Figure 96: Example of an insertion being displayed in the IGV browser

4. Complex

4	Service.	· a legate plana types	11	Sec.	1007	1010	10.0	And the Property P	Page #	And Departs #	All Depty #	hand Depits #	Ind Quality W.	ALCOHOL *	Quarty Name 41
	9881	AND IN PROPERTY.		0121405	8171401			#1.12%	584		- 294	284	2008.000	0015-00	1748-40
	1941	ANOVIA "BEBRUA		0123425	REPART		.1	89.70%	5544		108	181	86.00	12545-00	11000 90
8	801	Manager Co. Printed. Philadeline		PERSONAL PROPERTY AND	_ MILLINE	ALC: N	XOL	No beau	Terraret,		144	200	10.01	and on	No. 10
	3881	ANOT11.10084-0	×	an intern	ancientes.	1.0	.e.:	98779	1MV	1.1.1	100	150	31.00	3123.00	4211.69
64 T	1841	ABBRELL, PILLIPIA	8	1021004	BUTTHEA		÷.	100%	344		288	200	9.49	1012-00	6299.28
	1887	A4001+10400-4		61246754	0034754	- 1		(6.27%)	550	4	208	100	70.46	111100	8155.07
8. L	1941	AM01111-146(P-6		81,0113.55	0121333	- 4		01,79%	584		281	100	212.00	9095-05	7419.00
	3661	AM0511.125707-6		(1521423	#529423		. 6	19.47%	544	*		104	36.26	11205-00	10000.54
64	1881	AT121/5496-0		1003175	1908175	1	34	10.40%	Row-tops	19	19	100	546.00	4196.00	1075.00
	1941	APROL778P-C		10812342	10812342		. 8.	16.4%	8WV		208	101	100.00	10036-00	8464,78
1	-		interesting in the second	-			-				-	100,000			-
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	and printly deprived														0
	· North Contraction	- 10 C													
	A														and the second second
													_		
													-		

Figure 97: Example of a complex event being displayed in the IGV browser

5. Multi Nucleotide Polymorphism (MNP)



Figure 98: Example of a MNP variant being displayed in the IGV browser

6. Partial Tandem Duplication (PTD)

8 136486482 136486485 136486485 136486764 136486764 136486764 136486764 136486764 136486764 136486764 136486764 136486764 136486764 136486764 136486764 136486764 136486764 136486764 136 <th>8 136486482 136485486 10216 1 2.37% Deletion 1707 4.1 1795 6 42.138 9 136487184 136487184 6 A 99.85% 5NV 3 1964 1567 85.555.2021 113 17 360859 366962 AGST 6666 4.09% Complex 774 83 815 6 26.465 17 366969 366962 AGST 6666 1.65% Skv 774 83 815 6 26.465 17 366969 3469662 AGST 6666 1.65% Skv 774 13 836 6 28.465 17 366969 3469662 AGST 1.66% Skv 774 13 836 6 28.465 17 3669678 6 T 1.66% Skv 855 1872 25.56.4064 34.552 17 3668678 6 C Sev</th> <th>136496492 1364927184</th> <th>136496495 TO 136487184</th> <th>1 010</th> <th>2,37%</th> <th></th> <th></th> <th></th> <th>1295</th> <th></th> <th></th> <th>Al</th>	8 136486482 136485486 10216 1 2.37% Deletion 1707 4.1 1795 6 42.138 9 136487184 136487184 6 A 99.85% 5NV 3 1964 1567 85.555.2021 113 17 360859 366962 AGST 6666 4.09% Complex 774 83 815 6 26.465 17 366969 366962 AGST 6666 1.65% Skv 774 83 815 6 26.465 17 366969 3469662 AGST 6666 1.65% Skv 774 13 836 6 28.465 17 366969 3469662 AGST 1.66% Skv 774 13 836 6 28.465 17 3669678 6 T 1.66% Skv 855 1872 25.56.4064 34.552 17 3668678 6 C Sev	136496492 1364927184	136496495 TO 136487184	1 010	2,37%				1295			Al
5 136487184 136487184 6 A 98,39% 5W 3 1964 1967 45,365,2011 113 17 3669659 3669652 AGGT 6666 A.00% Complex 774 39 836 6 28,467 17 3669659 3669652 AGGT AGGS 1.65% SW 774 13 836 6 28,467 17 3669679 3669678 6 1 56% SW 774 13 836 6 28,467 17 3669678 6 1 56% SW 750 855 117 23,536,604 24,529 17 3699674 596574 6 6 56% SW 805 165 1672 25,556,604 3559 11 1566667 156,405 44,60% 44,60% 191 565 1672 25,556,406 3559 11 156,407,107,407,407,407,407,407,407,407,407,407,4	5 13448/184 13448/184 6 A 94.85% 54V 3 1944 1947 85.85.2011 113 17 7500859 7560852 A057 6055 4.09% Complex 774 83 815 6 24.45% 17 7500859 7560852 A057 A055 1.65% SNV 774 13 835 6 24.45% 17 7500859 7560857 3508154 6 7 1.65% SNV 774 13 835 6 24.45% 17 7500859 3668578 6 7 1.65% SNV 755 11 855 6 24.45% 13 355614 315514 6 C 56% SNV 855 1672 25.56.4694 34.562 11 15566714 1546664 Q.21 xx 44.00% 492 106 106 106 106 106 13 411 15566714 114.460.660 Q.21 xx 44.00% 404 405 404 405 406 406 406 406 406 406 406 406 406 406 406 406 406 406 406		136407184									
17 3689639 3669632 AGGT	17 3689639 3699631 A00T	7 7969659			99.05%	SNV	3	1948		45.965.2001		
17 3669678 3669678 6 1 1.64% 500 11 679 0 24,829 17 3639154 3639154 5 6 5 50% 50% 505 1272 25,355,4004 54,359	17 3469678 369678 6 1 1.64% SNV 655 11 675 6 24,529 17 369678 3636154 6 C 56% 50% 505 107 25,556,6004 34,529 11 15644174 156441745 6 C 56% 50% 50% 107 107 25,556,6004 34,529 11 156441745 15644166 C 56% 34,50% 400 50% 106 34,50%		7609062 .M	06T 6666	4.09%	Complex	774	89	836		28.405	
17 NONISH 20015H 6 C 50% 540 805 805 1072 23.565.400H 34.352 11 Holdwarrik Holdwarrik Generation Holdwarrik	17 3036154 3006154 6 C 50% 50% 505 105 1072 25.56.604 54.50 11 1566661714 1566666714 1566666714 1666666714 166666714 166666714 166666714 16666714 16666714 16666714 16666714 16666714 16666714 1666714 1666714 1666714 1666714 1666714 166714	7 2560659	7609662 Ad	DOT ADDS	1.65%	SNV	774	13	825		28,401	
II Holdering Company Holdering Holderi		2 25009178	2669678	6 T	1.64%	SNV	659	11	870		24,829	
		7 2626154	3636154	6 6	50%	SNV	805	805	1672	25,165.4004	34.392	
	and and and and an	115458714	118451495	< +00.Ph	44.40%	P10	1211	905	198	Contraction of the second	100000000	
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	and a second		1.40000	111	1 1			100				14
	and a second	2 5	A Constant of Constant			1.1			A A			
and a second sec	and the second s											
						-						
69 -			7636154 145561701 2611 • defit 11 64 64 64 64 6	909654 909654 0000000 00000000 anti • avrome,antaco, measurasi Q anti • avrome,antaco, measurasi Q	3536154 3636154 6 C 1555621114 1556621114 1556621114 155662114 an11 • avministration instation of Q, 21 kg 1556 44 1547 1545 1541 111	3536154 3636154 6 C 50% 1056021714 105602000 C 400.00 400.00 an11 - avminite.antizon instaticute Q, 21 to 400.00 400.00 an11 - avminite.antizon instaticute Q, 21 to 400.00 400.00 400.00	ADAITSE SUDATSE 6 C SON SAW Holdwarmal Holdwares 6 applies autom Pro- ant • aventration means and Q, pro- ant • avent means and applies area.	3036154 3036154 6 C 50% 54V 805 155640704 155640605 C 404/04 405 1211 arti	M04154 M04154 G C SNN SNN NDS NDS NSMATRIA NSMATRIA	3036154 3006154 6 C 50% 54V 805 825 1872 1155442774 115442675 C 40.000 40.000 470 1211 MAX 3366 arti - artin resultance instances (Q.31cs) - 40.000 470 1211 MAX 3366 arti - artin resultance instances (Q.31cs) - 40.000 470 1211 MAX 3366 artin - artin resultance instances (Q.31cs) - - 40.000 470 1211 MAX 3366 artin - artin resultance instance (Q.31cs) - - 40.000 470 1211 MAX 3366 40.000 470 40.000 470 470.000 470.0	MORTSH MORTSH G C SNN SNN ND ND	NOVESH SEAL SAVE SEAL <

Figure 99: Example of a PTD being displayed in the IGV browser; the duplication event is highlighted by the transparent red box

SNV Options

Right clicking on a row will generate a popup menu with a range of options.

	Surroyte +	 REAC DAVE SPREAD 	Constructed 7	30817	1.000	1414	10.7	Allegic Prospanny T	Person P.	And Sweet's #	At Depth 7	Total (Rept)s #	Prices 7	an party # 1	Streetly Score 4.8
	1881	ARHIGAPOTIC * STREET &		119416945	115418345			10.47%	5MV	148	- 118	1914 · · · ·	4891.88	0/56.00	404.4
- (-)	1881	ABREATON OFFICE		115294021)	115254821			44,000	SNV	140	480	104	0403.00	1008.00	000125
1.1	100	And Address of Contract		1000010000	- Tribuctory of	1		A to the second			1 August		the second second	1000	-
	1847	ARHEAPTEL/108CH		110417420	11010428	1.4	- 40	ARHOLY DAY TO A PROPERTY		198	176	17	5076.00	0001.00	478.84
	1881	ARHEAPER-LIERCOT	1	119470344	TTN/TD48	- 6	- T	Add to Shortist		184	200	1000	667420	7024-00	8088.82
	1941	ABHGAPST-L-TEMACHT	1.1	TIMUM	THAIRIN		T	Antra.			388	100	8.00	12771-08	11444.40
	1801	AM64711.00014		+14381299	114385291		4	and the second se		187	180	141	5441.00	4274-00	4403.01
	1941	48064701271391,713	1	119417948	THENE	4047		Set Preferent Transco	et 3 👘		190		1072.00	15450.00	#022.90
	1687	ABHOAFOTIC-TEND-A	8.	T182H/11	TROUT		. 6.	Cierofe		2019	240	607	746.00	8045.00	0004.01
	1881	MINIAPELL*UNIT-E	1	VIDEL NEW	THEM	1		Catad		100	148	100	47464.00	9404.00	1075.04
					_			Walate Links	⇒ :=						
10	W 1420 000	+	a Q.m.					Terres							0
100	A CONTRACTOR												and the second second		

Figure 100: Options available for each SNV or Indel variant

Add to a Shortlist

10.1	Alter Langie B. C.	A HOUSE Dance Special	- Description of C	Sillet?	CONT.	oute:	ALL Alley Program		Same P.	Ind Depth W.	All Depth 4	Table Courts W	Introducting W.	and Quantity T	Charley buies # 8
10.	1481	ABCODIA 7128,7109AM	14	11412041	114123-001	GAR.	E 100%		Datation		- 218	207	6.00	10140.00	11022-NO 1
	1997	Design and the second second	10	(section)	A REAL PROPERTY.		A COLUMN TWO IS NOT	_		100	1000	10	-	NUTLINE.	1 844198
18	1881	AMON*19861-C	12	21104213	311034313	· · · · · ·	ABCCHL/1585-4			- 167	1.00	204	4101.00	4008.00	2027.00
	5687	4003121814-0	12	27525625	27528526		Add to thereight	De.	. 1997		349	105	8.00	10012-00	1218.43
10	1001	HBUENL71430-0	10	21020111	27429271		ANIN.	5	5555		. 278	274	0.00	9474-00	8400.02
10.	1881	ABCD14.16380-A		TRETAKING	TRETAKING			~	NW .	144	1.000	248	466.00	0140.00	2254.41
10.1	0441	ABCOT+ ISBND-A	4	TRETANTON	TREASURE		Set Preferred Transor	P. P.	5507	409	- 201	104	6003.1.00	1981.00	1064.43
14	5687	ABCDUS BBOY		102724348	102724308	100	Circuity		SMV	24	165	170	7176.68	\$715.00	3471.16
10.1	1961	440311-1970-0		122744012	122144713	÷			589		140	145	4.04	10404-00	10000.000
201	19901	ARC014:180-C	1	112742542	102742542		Warriere Loting.		100	1	942	343	36.00	11668.00	10121-00
10.1	1881	ABCD1+:71840-4		112725764	112725744		Notes .		INV	29	278	104	4075.00	9795.00	796.N
10.1	tier:	X8051419840-6		VIITAAL	TETANUT		4	-	INV	184	179	140	410.40	0105.00	400.04
21	NAME.	Aduna *1768017		130851424	180887429	4	7 44.55%		5AW	178	540	621	6.04.00	8084-00	1504-45

Figure 101: Adding a variant to shortlist

Once a variant has been added to the shortlist it will be annotated with a tick.

			S	elec	ted		_								
	Lampie T	TTODA plane by the	D-British Barrier	I BALL I	110	-	44.4	Andre Programmy W.	Sec. V.	And Employ M.	AN Depth P	Intel Courts W	der bertigten	and Desires T	County Story 4 1
	14 and 10	MOTIO PERMIT	10	21623084	21623000	644	4	140%	Database		218	289-	8.00	10140-00	11022-00
84	APPS	ANY COLUMNER.	No. 1	distant and	AMAMA	100	35	Assess	ANN .	12.000		11.	ALCOLUL.	MARKING .	and the second
- 18.1	1881	ABCC912*14087v2	18.	(18)4(54	110404	4		50.00%	164	tent .	1977	0.99	4101.00	4034.00	unities 1

Figure 102: Annotation of a selected variant

A variant can be removed from the shortlist using the Remove from Shortlist command.

10.0	Dampin T	+ Hills man burns	Christianistic T.	Tiert W. L	1017	107	100 1	Alate Polyanes T	1000.7	10/26/0.7	Alt Saujer, W	Trand Degree W.	the Daniel T	All County #	County Name &
	8881	ABOONL TOB, TIPSIN	10	210,0001	110,000	644		1076	Deletani		.118	387	0.00	10146.00	1-000 20
51	CHIEF C	ABIT NO. March		ALC: UNK	Contraction of		1.1	ALC: NO.	100 March 100	YAP	144	199	white an	8679.50	424.46
44.111	1801	2-1902/14/1388	Q.	210483	210420		- 6	PERSONAL PROPERTY AND INCOME.	Contraction Inc.	107	128	208	102.00	\$308,00	3089.09
	3861	ABUTH FIELD	- 12	21000608	2100300	¥		Bertunie from She	and the second se		548	100	8.00	10212.00	7676.40
4	4887	AB(0101*1430-6	10	17625211	27609011			Add to.			176	176	0.00	8474.00	4+4100 ftp:
	0001	ARC0101084014		1021440270	1021446275		A			184	118	340	4020-01	0100.00	4000.00
44	1987	ABCOTA 18880-9		TRATAGONE.	TRATEGIES.			Dati Preferrent Tran	merint 3	144	-108	- 249	4006.00	2546.00	2008.41
N	1001	ARCHILTHRO-6	×	TRADep for	192340-01			Gauty		177	207	184	\$590.00	7501.00	506.42
88	8881	MACONAL REPORT		1947/06/04	1527/04/04	- E .	. T	comory.		,008	100	970	2176.48	8718.000	3875.18
	1001	46(3-1)-1979(10)		TRATEMONT.	manageria	10		Variant Lines.	3		100	185	+.00	13439.00	10004
10	1007	A803114-190-0		152540542	15240340		1.6	Apping	2		340	140	10.01	11068,00	10101.00
10	3467	ABC2112*1346-4		102120704	1010394		- A	0.000	-	256	.178	354	1075.00	9736.00	7108.52
	1980	ABUTO TO HAVE		1882-122	TRANSPORTS!		.82	100%	mag-frame		182	784	1.00	11406.00	1006.84

Figure 103: Removing a variant from a shortlist

Subsequently the tick annotation will be removed.

		_	Uns	ele	cted	1									
10.0	large T	A HOVE COME Spread	Distant T	Shell #	- INCT	. Set T	16.8	Allow Press, march 1	700 8	Arthold B	All South T	And Depth #	And Shares T	NUMBER T	Quality Store B.
11	Tees	ABOCHL/TOR, TUNKE	10	21020004	PROPERTY.	GAA .		107%	Deteriore		278	227	0.00	12140.00	1102.00 (2)
	-	Additional Species	1.1.1	Contrast.	1000		10.0	there a		147	144	116	-	1000	1000
1.00	0881	3-1100/*_40380	u.	110401	1104010			10.4/%	anv ;	41.7	129	328	4151-34	4208.00	3404.54

Figure 104: Annotation denoting shortlisting has been removed

Add to New List

Variants can be added to lists that can be used in software; for example a list of variants can be used in a filter as a means to specifically search for a data set.

	Internation of	· · High clara ipelia	Citemanian T	thet T	1017	4414	42.7	Adult Integrating T.	1944	Init Depit: #	in born #	Inne Depth +	Ind Damp 1	ALC: NOT THE OWNER, MICH.	Streetly Same N
14	1041	ABO(3117106,*72564)	16	(*******	(recent)	SAA		1076	Datasian		.318	387	0.00	12740.000	season in the
	1000	AND DESCRIPTION OF	1	2 and the local division of the	The second	1.1	1.1	Contract of the	-	100	199		-	and the second second	-
- 44 1	1881	ARCINA THREE .		17834031	270401	A.:		ANCONL21145-4	1.0		141	- 228	400.46	4208.00	1000.08
	1001	ABCON. 1983A-E	10	2162828	2162828	1		Add to Chordist			309		8.88	WELLOW	9076-80
- 10	8881	480791.*1400-6	14	216/0011	2162011		1.8	AMIN		- 8	476	216	4.46	9474.00	8489.52
	1001	ABCOVA NEMIOR		100744624	1007044021					· Remittet	118	100	4425.45	0100.00	400.04
10	1001	ABCIPTUL MODELINE		15234040	15234640			Sel Proferred Tak	serue a	1- 53	148	140	uteres lat	1943.05	1059.40
10	5681	ARX211.15480-4	х.	103348101	1033/481011			The second s	646 M 20	107	.007	388	4010.00	1901.00	5064.02
	1881	ARC014, 090+1	*	100703046	10750.09	6	. 1	Cassify	P.	279	188	100	1170.46	1715.00	3875.78
	1881	MRD1x/MITC-F		19214412	Internation 1.1	1		Variant Strett.		- 4	180	345	0.00	19434.00	10000-001
- 14	1881	ABCD1+1790-1C		-STATIST	rational		- 6	tecter		1	148	140	an an	11048.00	Hampin pa
1971	5881	28/01/271940-0		ISTONA.	mail (minut					254	276	554	4479.00	whene	Pras.A2
	1881	ABUTE-PE-Manuel		10000400	CONTRACT.			1075	minister		180	104	6.00	11466.00	10000.04

Figure 105: Adding a variant to New List

Initially users will be prompted to create a new variant list by setting the name of the list.

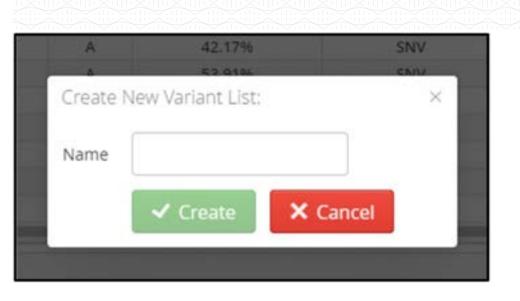


Figure 106: Creating a new variant list

In the example below a list called New List has been created.

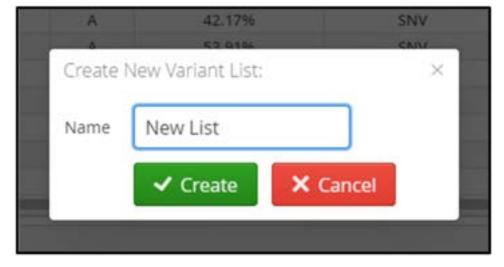


Figure 107: Setting the name of a new variant list

The list New List is now available and variants can be added to it.

8.1	Cartals 7	CA HELDS DAVE MARKED	Distances 1	C DIANCE	1017 047 423		Presidents T	Task T	Hel Dupli, T.	AR Depth #	Table ChipMit T	And Streetly T.	All Loans T.	Darry hore th
0.1	1001	4801701708,*12564	18	01120003	STREEME GAA IS		1275	Designation.		110	187	0.00	12140-00	1100.00
12.1	5661	ARCENLISSAG-A	10.1	greet, level	anational at the		10.479	5AV	647	148	2.8	410.00	1812.00	404136
8	1942	ARCHUTUNITY.	Mr.	20441000	and the second s	-	IN PARTY	399	10.00	- MI	100	440.00	401.08	AND NO.
14.1	1881	38035471818-C	- 42	21403636	ARCONT ADDATE	1.1	100%	350		399	104	1.00	1012-00	8298.80
10	1001	AB(01+171430-0	18.2	21803041	Add to Shortfel		100%	350		-278	278	6.48	9473-00	6491.52
	1881	48/01+174890-4		102144848	Allin			580	144	100	244	454.55	3942.00	1000.41
81	1941	ABCOTIL 19480-14		100140191			 New Ltt. 	. WV	407	397	184	40452.000	7981.00	104442
	1481	ARDIG-DIC-T	×.	1227-0228	Det Preferiet Transition		Number of Street, Stre	- 1997	208	160	112	11/16.00	\$715.00	3871.18
8.1	1881	HECOLUMPTIC-F	8	100144010	Clavely	3	1076	344		289	280	0.01	13436-00	12039-00
14	1841	ARCE14,780-C	A.	HENRY		-148	08.77%	XW	1.	340	945	34.00	11688.05	10101.00
61	1001	ABCD1+LT184D14		VERTORNA	Variant Silves.	×	52.00%	SNV .	278	278	Dist.	8475-88	9755.00	7108.52
	544.0	ABCD11178940-6		HITAKIN	notes		45.00%	5AV	. 184	176	240	605.00	6186.00	#400.M
	5885	AMATE PERMIT		Tains In Pr	TRANSLASS I T		44.55%	10/1	- 178	140	841	1005.00	1014.00	101.45

Figure 108: Adding a variant to the newly created list

Select Transcript

By default the a gene will have the largest canonical transcript set as the preferred transcript.

To change this, users can use the Set Preferred Transcript option to select an alternative transcript from the list available in the database.

10	The second to the second	· HEATS Dans Synthe	Destruction T	list T.	INCT.	1417	48.4	where Programs #	Tare P.	And Deputy #	ALCORDON T	Travel Despire #	inf inverse 1	Latitude T	Gamily Speed #3
10	1881	ABRIGAFO1L*1104C+8	8	1154114,00	1194774,00		4	47.37%	-	198	178	171	1079.00	404,2 000	4279.84
8 :	100	And Address of the owner of the		CONTRACTORS.	TTRATTON	4							and the second s	The second second	and the later of t
ŵ. 1	1987	MINIARD LTIMEOT		111420238	119403230			 MHEAVITE JARDO 			888	396	-9.90	14771-00	111044-00
4	1891	ARHONYS14,4800-0	3.	111082249	118382299	τ.		Add to Shortfelt		187	188	340	Avenue and	9287.00	A403.MT
6	1881	ABMEAPOLS, 11201, 1120	3	11belTball	TTM/THEA	GEALT.		A(24) 10	3	111	185	408	8012.00	11490-00	#600.MB
	1881	ARMAPPLE-INEE-A		115254711	115254711	4		COLUMN TWO IS NOT	-	110	1 mi	461	Twen an	\$163.0h	1010.10
a 1	1881	administration, Printer-III	0.1	115413840	115417840		-6	Set Preferred Trans	- O.	INFORMATION AND		348	#160.56	1004.04	3875.54
	5681	ARHIGAPOTA 1985, 1986		115417548	TEMPT TAR	4	M.	Carloty	3	MA 020754-4 (mill	HGAPTEL -	-	8257-86	11116-06	8016.64
1	1001	Amelantina. Jachtera		115414238	111414038				-	888.006711714.5.0	ADVICAPITY	240	10.00	11840.00	10550.10
1	1881	ARHIAPPIA 10010-E	1	1154185-85	111418548	ε.		WWWWFLINE.		and or makelets.)		214	4411.44	4014.00	0001-00
4	1841	ARHIGAPOTA SEC-4	8	115294881	III THE HERE	8		162545		read by some starts a	Manufacture 201	388	1011.00	1016.00	1481.10
14	SAM1	INTERPOSE PREMIT		119417965	119417965		6	11.785	100	192	163	318	\$2440.000	1014.00	2005.18
61	1441	- address Manuel	4	INTERNAL CONTRACT	INTIMAL			48.73%	1000	154	234	488	40173-00	10071-000	155.70

Figure 109: Setting a preferred transcript for a gene

Variant Classification

Users are able to classify variants in two ways; firstly, a variant can be directly assigned one of the defined classifications.

Additional classifications can be added via the Admin Controls (Admin Controls > Analysis > Classifications).

10.0	Langie V.	Histo Steve Spelled.	Concerns 9	Sini T	247	Bef T	10.0	Allow Departments W	100 1	And Daught W	an Layer, W	Intel Depter #	Bel During W.	And Country W.	Quality 1.00
-	1441	Dista Partic	4	140(76)	140176		- 8	100%	HW C		148	110	0.00	4771.00	er00.18
		THE R PROPERTY AND INCOME.		Case No.	1000			and the second s	-		111	100		10000	-
10	1881	#THE464.47%8-#	- 4	1549-149	1548154		A	DWLY 2000-C		140	117	942	019430	0004-04	4079-1
10.0	5681	ABADDALL PIDADOT	1	15345.00	153453	8.		Add to Shortlist		10	16		718.00	915.00	175.5
10.1	1997	#940(AL11000-C	4	1034402	10440		- T	A2214a	>	189	191	274	1008.00	4943.09	4405.4
12.	1881	361/16274	1.1	- steeler	seens?		3				475	475	8.00	10142.00	1402
10	1007	1814.737820har		12000027	200907	16 C	- 17	Gat Professet Tage	100 pt 1	- 49	117	100	110.00	8121.01	4278.7
10	5681	District, and the		KINGTOOT	100707	6	1	Oanth	100			100	4420.00	-0144100	140.1
14	1881	Examples in Particle		10001-44	1000144		. 6	Sector Sector Sector	100	Liting Guidelines		. >	1148.30	4575.04	405.2
10.1	1881	CARTINE TO TOTAL		100.001	10412017			Variant Links.	0	Deer Claimforder			40.00.00	1988.00	5167.8
14 1.	1881	DARGEOL/19829-A		1747081	19(18)		- A	Things .					\$500 mil	9411.01	475.1
4.1	3847	CHURAN-L*18H,*198H	1.1	750541	7107848	τ.	DAA .	pi are	manital	Derigh			- 201-20	9434.00	TIMA
14	NAME OF COLUMN	CAMPAGE PERMIT		77681/13	7769114	18	1	20%	Debeter	Uncertain significant	rea: Divery hering		456.00	3424.90	- 2419.2
10.1	1841	CAMPRIL, PT798944		1768216	Tradition	64	1.6	10.40%	Selected	Uncertain significant	108		307.64	13239-06	mail a
10.1	1991	CAMINE .* 25 (2mil		Thereise	Thinks	48		155.50m.	Deletter	Divertal agentical		-	2140.00	11148-00	8409.2
44	1441	EMMINTE."25586-A	. 1	Children of	these .		. 4	50.58%	1997		or many leaves		474339	Treast	Millia .
44	5881	DM/9411/101094-0	1	116020	119828		. 0	45.77%	3891	Fallingerin			4040.00	4967.00	Dept.4
1	SAMS .	REFERENCES OF		105/975	105/071	1.		\$1.20m	100	181	144	195	4255.00	6471.05	units I

Figure 110: Variant classification options

A variant classification is selected form the list that is included by default. These are:

- Benign
- Uncertain significance, likely benign
- Uncertain significance
- Uncertain significance, likely pathogenic
- Pathogenic

100	Langie T.	HON SAY SHOE	Character 1.	SAULT.	1017	ALC: Y		Addate Providence T	1047	Participant T	012000.7	Total Depth #	And Country #	AR COUNTY T	Starting GM
12	1991	800147387516	1	1200700	1200790			100%	100		100	108	0.05	4771.00	#100.72
	1015	- POLICE BARRIER	1	THEFT.	DOTO	10	100	100 March 100 Ma	-	2	300	100		The second second	1796
10.1	1881	KORDAX4710-A	1	7820758	7820758	4		210.11.3664-5			333	342	4196.00	HERE	6779-1
44	MART.	diagoni, Health	1.1	1134279	1344378	÷.		Add to Shurtled		21	10	10	198.00	815.05	279.31
41.1	1881	4740341179906-0	4	1524403	1514403		1.6	ANTIN		181		414	4104-00	10103-000	mitta
10.1	1881	140x *********		2000047	2008047		1.6				470	419	0.00	15148.00	1400
41	3867	1411,121803-0		2809847	1000017	,¢.,	-07	Set Preferred Tran	works 3	. 10	187	186	752.00	8033.00	6276.7
44	3681	CR#1011288C-1	1.1	6687707	6881707	¢.	1 -	Classify		100	171	410	4723.00	4144.00	21423
10.1	18.81	DRIFTER ALTER C	1.1	10030144	10087144	T	- E	and the second se		Using Guidelines.		2	7168-00	8175.00	6409-J
10	1001	Chieffelo.compos	1.1	TOADET	THEORY		- A	Variani Linki.		Deer Dauffugtur			1110-00	106105	para l
10	1891	CONTRACK "SHOTLA	1	TheToet	TheTweet			Texted.		CHAR CHERNELER		10.00	1010-00	10795.00	409.5
	1481	0465014*088.*15858	11	1107040	1111543		10.0		-	Bernget		(here)	291,00	003400	Trates
10.1	1881	Configure Provided	. 1	1798111	1108114	14.	. *	1976	Deletteri	Uncertain signific	arcs: Thety benty	- C3 -	614.00	2426.00	1010.2
4.1	1887	CANFING PUTMINE	1.1	7798218	1198218	čk.	1.6	81.87%	Owner	Montain Sentis		~	101.06	0.02394.000	ment
10	1941	CM/9414.703024H		Thereica	Transie	48		11.079	Ordenses				8180-30	11148.00	8446.1
10	SART .	DAMER For POSISIO-A	1.1	Thinkel	Themist			\$1.38%	1000	Uncertain signific	week mind bases		4783-00	7084.06	D-DL
44	1661	CM/8112/20158-6	4	Thesast	THREE.		- 4	45.71%	1000 C	Pathopeni			8945-00	4461.00	Judi-A
100	1444	alar deriver		100000	#151/6/15			11.078	-	144	104		4/16/10	0471.00	1000

Figure 111: Annotating a variant as benign

Once this classification has been made the variant will be annotated with the corresponding colour classification. This colour can be changed in the Admin Controls section of the software (Admin Controls > Analysis > Classifications)

This update will be applied to the variant annotation. As a result where the same variant appears in other samples it will have the same colour coding in the table.

		PERSONAL PROPERTY.												
		01.71273471-0												
		INCLUMANT.	 THATTH	TRAFTS	1	E	11176	100		THE	100	20.00	1919.00	Artist.et
100.0	1847	49/03A18710-4	 1147114	10401144		+	- 45.7%	low.	100	1177	100	01744.00	4011-00	428.04

Figure 112: Update of the annotation to show a variant as benign

A variant classification can be removed using the clear classification method

8	Sample W.	High sizes Spelled	Courses 4	Det W.	1.007	left.	117	All a Constants	1011	Ball Dages #	All Depict #	Intel Digits #	Int Desity T.	Ad Quality #	Quality 5.8
	1441	BHLIG PATTICK	4	1508798	1408/98			100%	and a		108	144	8.00	4771.08	#102.8
	1001	ENGINE MODIFIE		1040110	INCOME	- F	- F	-	_		109			1010.00	8798.
64 L	1841	40404414710-4	1.1	1500150	1520152	4.1	- A	DATE MARKED			+179	141	8754,89	4030-09	- 42581
	5497	#0400Kjt/1540-P		1504028	1504028	- E	1.1	Add to Dwinting		21	18	92	738.48	915.00	195.5
4	1841	KINDLAL T1000-C	1	150milli	110402	4	τ.	A60 to.		103	181	074	4205.00	0001.00	inst. s
a	1841	1861,118021-0		1768147	1248147		- 8				474	479	1.16	12192-08	14000.
66 - L	1941	181+.13183/hpt		LIN660T	1000007	- E C	0	Set Preferred To	secult >	10	107	100	762.00	0011-05	4076.5
	5680	CONTRACTOR OF		8887767	4607107	- E		Cassify			-	100	49623-MI	4684100	140.0
63	1881	CHURKES NOTICE		00001-00	******	+				Orig Contennes.			7148.00	4875-09	+06.0
	1941	CRAFERIA 1000014	1	TRADEST	140827			Tertart Links		Char Carofnato		-	40.00	1082.00	Analy a
	1881	Distancement-e	1	179/381	119(138)			Notes	2			305	DOUGH AND	1891.03	6276.2
	biet.	DMBN/s/1004/1008		THERE.	1111145	£ .	044	56.37%	PORTOR	Berngel		0	291.86	8424/05	71684
10	1881	CAMENTS:*1640eW		27945110	1794714	- DA	. 8	70%	Seletion.	Uncartain agnifu	alox: likely ben	1	450-16	1425-00	2010.2
	1881	CAMPAGE PT THEM		TTRACTO	1746278	-0.		11.17%	Deteriori	Uncertain signific	ania .		207.84	12230-00	10473
10	1887	CARPENS, *252284		1760088	1100004			11.17%	(Veriation)	Uncertain signific	and there are	Contraction (1)	11100.00	11108.00	8499.1
	1881	186/0/14/103980-A	1	Tradeut.	1799647			11.175			and the second second		00.0	7584.00	87533
10	NOR!	LAMBELL-107058-4		1749026	1149026		- 4	45.77%	UNIT .	Fathogenic			5045.88	0017899	3481.4
	SAM!	WHEN A POLYCELAN	. 4	and other	and services.	1	1	41.076	10400		189	100	6719.00	0475.06	40.00.0

Figure 113: Removing a variant classification annotation

The classification will be removed for the variant in the table and all other samples with the same variant will be similarly updated.

1.0	Langle P.	Index plane typetant	Destination #	- Batt	int in	1.641	10.0	adala Pressance V	See 7	And Deputy #	and Department	Children Departs W.	Address T.	All Generally R.	Georg Same 7 10
44	Land L	106.5 a 76875-C		1107166	1222794			100%	100.00		1.00	100	0.00	475.00	#10.3%
100	10001	2010.000-0	1	1142710	TRACTOR		181	0.076	100	- 14 C	315	100	11.00	7904.09	179.47
1.00	1949	4500000000		YEARING	1800188	4.1	1.0	10.01	ing .	165	111	362	4154.04	with the	grander

Figure 114: Update of the annotation to show variant without any classification

Using the American College of Medical Genetics and Genomics (ACMG) Guidelines

An alternative means to derive a classification for a variant is via guidelines described by the ACMG. These guidelines are included with Interpret.

To follow the ACMG guidelines the user provides answers to a specific set of questions. Each answer will navigate the user through the conditions of the guidelines until a classification of the variant can be made.

Selecting to use the guidelines option leads to a new window opening.

	Longin T	Horse place Special	Disconception T	ALC: NOT T	that \$	del T	10.0	Adding the space of the	Jugar T.	And Deputy &	all Dayin 9	Intel Depits # .	that During #	Add Dunling W	Specific L
	1001	1HL14.18871-C		1222780	1222798			100%	1000		100	14	8.00	4771.08	4122.5
8	1011	and a second of the local division of the lo		1000154	1244154	1.7		and the second second	-		-			- 841.00	-
a 1	1997	ADABAN, ATTON	1.14	1638152	1628154	4.		DVI.11.255A+C		100	179	342	2194.00	8054.04	4096-0
	5481	AD40411154017		1504200	1534238	- 6	1.1	Add to Shertfind		21	18	10	798.44	915-00	205.5
1.1	1001	AD40041.18900-C		1534403	15440	- 4	1	Add to.		100	i gen	3/4	6105.00	1045.00	405.1
	5601	1011.710027-0		109441	1008087		1				479	49	8.00	11744.00	14005
0.1	3661	1014/17112/hg		1009917	100947	£ .		Set Proferred Tr	encip. 3	.=.	5427	198	752.04	8125.00	8076.1
	5681	Chiefdat A 2000/1		4447767	4441797	÷.	1	Cantery		444			4105.00	4544.05	19A0.0
1.1	1041	CHARGE AND A MARKED	1.1	0052144	0948144	F.	- 4			Lising Gelowine	100 C	•	ACKED GARBON	A 171.00	1006.0
	1001	CAMPINI 1955514	1.4	Indject	Insided.			Vaciant Links	3	Dawi Classificati			Print and	C 101.00	4447.4
	1001	Lauffalt, "good-a		TTUTION	TTUTION		- A	Nideri	2				55550-000	Sec.m	4(19)
	5861	24445411;11064,118888		TTEPERS.	1757548	- E	444	31.470	Buartoon	Bengh			201.00	8624.08	1108.4
	1001	CAMPACI, THEORY		1796110	1768714	16		79%	Ovieton	Unsertain laged	carrier. Help ber	- m	876.01	3425.00	1813.1
	1941	CANTRILL*17284ar		1798278	1796219	0.6	- E	81.87%	Detellant	Uncertain-sprofi	anes .		807.48	ULINE.OF	19401
	8881	CAAMELA 1313364		1746688	1794604	HE		91.176	Deletion	Uncertain signifi			17.002.005	11149-00	puttin 1
	8861	CARPNIN TOTAL	1.4	THINKT	1168647	4		35.38%	144		care and has	- Alexandre	4763-46	2084.00	81164
	5881	CAMPORTS, *2705A-10		1144226	17985226		- 6	45.775	14M	Partupent -			1045.00	4987.00	5481.6
	- teler	April o Concession			4141414			11.100	1000	100	180	100	174.00	AAPS.im	4016.0

Figure 115: Selection of the inbuilt ACMG classification guidelines

The initial ACMG window, shown below, consists of a progress bar that will report how close to a classification

- Progress Bar Showing the progress of the classification.
 Questions These are the questions to be answered.
- Toggle Allowing the display to be a graph view or a table view.

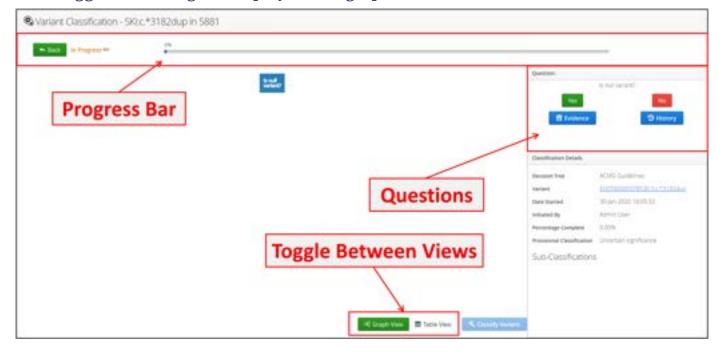


Figure 116: The initial ACMG classification window

As the user answers questions the progress bar will update.

Viewing Analysis Results by Sample

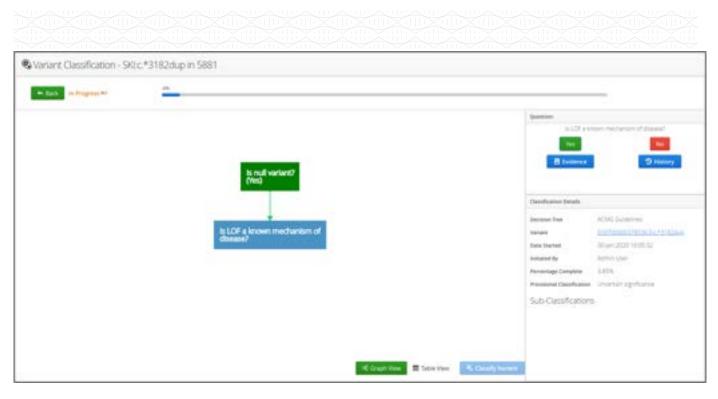


Figure 117: A classification at the start of the guidelines, with 4% progress

· last integration	<u>15</u>		_	2
	Is null variant? (Yes) Is LDF a known mechanism of choose? (Yes)		Darmeter Dar men ih R forder Darohanne barat	
	Post De novo in a partient with the desease and no family followy		Desistant Trap trainert Data transp tentared By Penetroge (penplet	Actid Jammins Exclamation (Contraction) Weak 2020 (Add State Address (Contraction) edd/7% United and Address (Contraction)
		🗟 Graph View 🔳 Techs View	A Carsely secare	

Figure 118: A classification with 67% progress

When sufficient questions have been answered to allow a classification the progress bar will update to show 100% and say Ready to be classified.



Figure 119: A completed ACMG classification

A window will appear showing the classification and give the user the option of making the classification or cancelling.

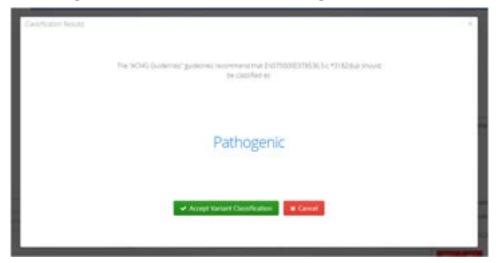


Figure 120: A completed classification showing a Pathogenic all has been made

Selecting the classification will update the variant's annotation accordingly.



Figure 121: Updated annotation for the variant to show its status as pathogenic

It is possible to review the choices made in the guidelines; using the table view, users can see which questions were asked and how they were answered by whom and when.

Variant Classification - SKIc.*3182dup in	5881			
• Date Completed ©				Carofiel As Parkipent
Annual Dation .			100	Surger.
Normal Control of Cont	10.4	Distances in Concession of		Citite pri a Duanterro
In the 1 apparent with the stance within fairing interval	100	Autory User (Briger 2020 Talo 128 C		
a USF a known mechanism of disasa?	200.1	Administration (20) per 2006 16/07 17		
tense serent	164	Remine (Joan 100 (and 2020) 16:07:00		

Figure 122: Table view of a completed classification

Selecting a row from the table view allows a result to be modified if that is required. Alternatively, evidence can be added to support the answer to the question

Variant Classification - SKto	.*3182dup in 588	1			
- Each (semplement @	1004				Cassefied Az: Pathogeni
Anner Dalling				-	Overlage In COV a screen machinest of Issaed
Notest Construct		Take 1	Invested by Chart Investor		ar ma
1. De mois in a justice anti- the disease and m	family many?	1998	Administrati (35)pr-2000 (4(37)38		B henry
2. It LCP a level technicit of dataset. 3. It mational		764	Admit Over 1 35 per 2020 HLISTOR		

Figure 123: Reviewing an answer in the table view

Interpret Cloud User Guide v1-20241029095209

For Research Use Only; Not for Use in Diagnostic Procedures

Variant Links

The software allows users to link out to external sources of documentation. Currently included are:

- EnsEMBL
- ClinView
- ExAC

Additional resources can be added in the Admin Controls (Admin Controls > Analysis > Manage Links).

2.1	Sergie 4	MUCH Elever Spectrum.	Conception I	THEFT	104.8	inter .	10.0	Allele Trappeng P	Law W.	Red Dagets #	ALCOHOL T	Take Depth #	Barrissing W.	ALC: NO. OF CONTRACT,	Darry Sales 12
- 1	No. 1	3011-1-1471-C	8.1	108/98	I MATER			1009	100		100	100	4.00	4771.00	41123.84
	199	THE R P WHEN PARTY OF	11					-	-		100	311	1111	No.	and a set of the set
44.0	1881	AND SHOT STOLE	1	- Materia	1620100		-	SWITE BRANK			1.94	(44)	012428		4000
64 L	3481	ADADDALL*ISADJE	8	159429	159428			And its Shortist		181	14	27	738,60	515.00	2518
64. T	1891	enclass resided	4.5	100460	100eHut		- E.	Add to:		101	100	214	10.000.000	F999.00	4695.87
	4881	inter the second	* / ·	100147	100MA	1	4				410	416	0.00	10140-00	1405.10
81.	10.01	Serv. *Fedding	- C.	100000	100007	20 K	-81	Set Preferred Nam	10.00	- 44	187	146	791.00	8081.00	- 8079.81
41	9661	Gautane, (68) 19	+	4867767	1847107	1.1		Cassify		185	101	102	49523-09	4546.00	1143.87
64	1881	perfects/statute	(1)	10021-04	10001144		£	Contrary of	- 25	318	104	387	7146.00	4975-09	0402.04
64	1887	CRAPRILL10200-4	0.00	The rate of	1940347			Weiser Cires.	Bar 2	Enternia Positioni	38	- 440	910.00	1083.05	0107.63
6	1001	Deletal u PRIStrie	1	797364	1797384	- 8		Notes	()>			101	1000104	1011-22	407578
	16661	CARDINETTING FORM	1.1	1747544	110-044	1.4	Dis.	10.000	Marriel .	Ensembl (Genet	14	- 104	281.00	MIA-00	71844
66 L	3881	(AMERICA*1640MM		1080110	1758114	- 194		70%	14MP	treents (Transister		104	8761.00	\$425.00	215.05
61	2001	Califian L.****8898	1. C	7198278	TYNET	- 24	÷.	10.074	Desire	DWW HRV50	127	229	241.00	122280-005	2047.03
44.	1881	CONTROL * PLUDIE		7764000	77940004			10,129	Destroit	ExAC (Clane)	81	346	12-10120	11148-02	M01.75
44	1001	1047914701880-A	C	research	renter			10.1Ph	201	EAC (Transmith)	B1	100	0703.00	7084-00	ACCURATE VIOLENCE
68.1	1041	Cold Dirack (1994) - G	÷	7798028	7764528	- 8	4	45.71%	- 0.01	ExAC	14	310	8045-00	4887-00	240130
	3981	4444,707-044	1	40109-15	400,815	. 6		91.329			10	311	6275.00	14/5/00	400.04
6 T	1001	48461,*21107-0	1.1	References	444,040			1000	10.00		891	101	1.00	11472:00	

Figure 124: Variant links available in the software

If a source is selected Interpret will show the information in a separate tab in the web browser.

	lamph T	HERE they fairbuild	Character #	Citer T	0.07	Out w.	41.7	Other Dispatry 7.1	Inter #	And Depth. W.	ALCOHOL T.	NALDING T	that South 1	al lotty V	diam'r bres 18
41	++++	346.1 or #3477-0	+	100708	Loughten			100	1100			146	2.00	4771-00	4123.68
	100	and the second s		-	1000			and the second se	-		1.00		10.00	No. of Concession, Name	and the set
10	1881	ADADBAL BTIGAA	1	1008198	10081198	- 6		And an indexed		100	121		012430	1000-00.	4039-04
0	1881	APROLAU, TIGAC-T	1. C	159429	150429	1.6	7	Add to Shorthet		31.	16	31	788.00	111.00	275.16
6	1881	HERCENI, VIEND-C	1 C	104440	1804602	- 4		Ald to.		184	100	314	(12.23.10)	4945.00	4000.17
	1001	0010710027-0	+ .	10894	100007	1.1	- C				471	476	4.00	19192-00	140570
10.1	1611	Jang-Prisidea	1	(Distant)	phieker	4	0	Sid Preferred Transc	197 X	- H	1.65	100	79,5,00	8102-00	80%34
a 1	3481	Destriction	1. C	1007107	4407107	- 4	× .	Clearly		190	194	301	1003-01	4044.00	1940.07
a .	1991	DRAMALL NUMBER	1.5	10007-01	10000-00		Π.		12	218	194	2857	1145.00	8079.00	0000,00
44	1881	CM/9141892-4	+	Tex Call	184.0917	1.1		Kartard Little		Drawnia Proditional		- 416	9581.00	101.07	010740
SI -	68.81	Contail, Netherica	- C	75/161	775/161	1		Holes.	()	Drambt (Gene)		848	10103-04	1010.00	405.78
	5881	100000-1-1008-1-0000	+	Thereas	TROUGH	1.	684	10.30%	- magnine		10	204	281.00	8614-00	1103.48
61	1001	CAMPACIC+TRADRE	÷	798710	1100114	- 14		10%	Deeport	Ensemble (Transcript	· · ·	1.94	828,07	3408.00	2819.75
6	1621	GAATR/ 1.*179894	- C.	79803	1168279	1.0	-E	10.034	Detabation	CREAK (HOV54)	1	.201	0.01.00	12286-00	204110
10	1881	CHAPRILL TOUGHT	1.1	79400	1794004			15.179	Destroy	ExAC (Germi)	0	248	100020	11148-00	Helvis 78
10	5881	DMD1+72880-4	4	THEFT	Chemistry.		4.	01.071	84.	ExAC (Transprint)	20		4701.00	FIG. 00	81248
68	5681	OMINIA NITHIAN	+	Analiga .	1786428			46.71%	0.01	EnAC	0.	919	1045-00	4987.00	3481-89
6111	1001	NUMBER POST OFFICE	- A.	100213	100775	1.1		21.02%	54			201	8,754.00	34(5.0)	with he
10.1	1881	ANNU TIMET-C		anonia.	Antonio I			-128	10.00		1881		1.01	+ iareas	10020-000

Figure 125: Selection of ExAC as an external resource for the gene selected



Figure 126: Example of the software linking out to an external data source, in this case the GnomAD for the gene containing the variant in Interpret

Add Notes

Interpret allows users to add notes for a variant and to also edit notes on the system. This is accessed through the Notes menu item.

18.1	Sample T	HINTO Dava Lynnes,	Designments Y	INCO	int t	ist t	10.4	where fring army #	tare to:	ALCOUNT.	at paper 1.	treast (require #	Including #	and the second of	Quality Scores 7 10
10	188*	311/10/10/10/1	1	1802735	1800755			1405	244		188	148	0.00	471100	-
	194	and a second literation of the lateral second se		1000	Constant State	1			-		100	201		The set	1000
- 10	1447	4003AL8710-9	A	10.01114	10.0110.0			Contraction of the local distance of the loc		100	117	362	#7 THE 201	4/101-00	4270.04
	Mari	ADADAG PERCON	4	1834228	1834236	6	1	Auto to Monthist		31.	16	10	798.00	. 916.00	278.18
- 10	1997	discons 48800-0		1834452	1834452	. 6	- E -	ARE SA.		160	101	376	1008.00	4943.04	469(1.67
	1881	any named	+	indul	104411	+	1.6	1.2.1.1.1.			474	411	436	10143-00	1-4030-14
- 44	1887	later+01404a		100441	Ymeen	ε	- 67	Set Preferred Travits	10.5	38	197	144	192.00	8121.00	4079.01
	5651	CHRYATILINEO-T.	3.5	MATTER	MATTER	1.2		Casally		190	102	812	6523-00	4044.00	Inel.M
14	1001	CANTROL/1010F-C				F	1	and an enter address		218	184	207	1146.00	0075-00	4416.64
1.00	1447	Debte 14,1000014	1	1443017	1443417	1.1		Varianci, Jriac.	2	194		400	Market .	1982.00	anatas.
- 10	1847	Caladratina Perinthia		FTR/SAL	TTR/DAT			Roles .		A DECISION OF	188	100	1000.00	4471.00	4275.79
	5581	Contain view, *1999		1167842	1167542	1.4	644	16.40	POPPOR.	0	188	154	299-30	Minth	Print and
1.0	1887	(whithin to Hawmini		TIMPTOP	TIMOTA .	14		108	Designed	11 V	4.8	134	100.00	Same in	1010.00

Figure 127: Adding a note to a variant

Selecting the Add Note will generate a popup window

 Add Nexis To Vanand: 		
Notes for DVL1:c.366A>G		
Owiedens remaining 250	Existing Notes (0)	
Canval + Add N		

Figure 128: The note template window for the selected variant

Users can add the required text, up to 250 characters, in the text box

+ Anti Note To Xanaro		5
Notes for DVL1:c.366A>G		
Characters remaining 216	E testing Notes 20	
This is a newly discovered variant	Part of the second s	
	A STATE OF THE OWNER	
Cancel + Add Note		

Figure 129: Addition of a note to a variant

Selecting Add Now will append the note to the variant.

Acts Note To Variant			14.14
Notes for DVL1:c.366A>G			
Daraders remaining 250	Exiting Notes (1)		
	Admin User 2020-01-22 17:18:22.0	1	
	This is a ravely discovered outlant		
Cancel Add No	te		

Figure 130: An example of a note on the system

The additional text will now be displayed

Notes for DVL1:c.366A>G			
Characters remaining 250	Exiting Notes (1)		
	Admin User 2020-01-22 17:18:22.0	+ Anat	EL Apply
	Characters remaining 218		
	This is a newly discovered variant		
Cancel + Add N	the		

Figure 131: Appending text to an existing notation

Notes can be modified by clicking on the pen icon. This makes the text box editable

Hot Note To Variant	
lotes for DVL1:c.366A>G	
haracters remaining 250	Existing hotes (1)
	Admin User 2028-01-22 1718/22.0 - Seven Zi Apply Ownaders retaining 191
	This is a newly discovered variant It is likely pathogenic
Carcol + Abb1	

Figure 132: Adding an update to a note

Once any update has been made, selecting Apply will incorporate the changes.

Ourachers remaining 250	Example to ter (1)		
	Admin User 3035-01-33 17-18-22-0	1 1	
	this is a newly discovered variant		
	it is likely participanic.		

Figure 133: A note showing the updated annotation

Similarly, a note can be deleted through the red bin icon.

Users are asked to confirm the delete request after which the note will be removed.

Actil Neter To Variant:		- X.
Votes for DVL1:c.366A>G		
Daracters remaining 250	Costing Notes (1)	
	Admini Char 2020-01-22 17/18/22-0	
	- Cancel A Confirm Delute	
Cancel + Add	And a local and a	

Figure 134: Deleting a variant note

Where there is a note for a variant the note can be viewed through the Notes options seen when right clicking on the variant.

10.0	Tangin T	High Stee Spellet	Distances of the	line T	1044	inter.	10.1	Man Copping V.	Sec. P.	and Depth #	All Depty #	Initial Corplin IP	And Streetly W.	all loads T	Chanty Incire Vill
	1481	BUILING-		1440100	1000100	4.		1209	Sec.	T	. 138	108	8.05	4111.00	0122.89
	100	The second se						1000	-		100	100		1000	1000
H. 1	1001	40634-10710-1	÷	16,00194	10,02124			Delta Transaction		188	+10.1	140	110420	#1751-HZ	4789.04
100	5687	40403444444646	1.1	1884238	103403	1		Add to Shortled			1.146	10	298.40	915-00	.26.4
10.1	1941	#0-0141-14800-C		10.04400	10.04493		÷.	Astro.		180	101	104	COLDN-101	4144.00	4005.67
	1981	10127180742	+	100001	14MM	1.1	۰.				477	470	4.00	12144.00	1446110
44	1011	1011-11120-0	P	1009001	1009901	- 1	-17-	Sat Preferred Transm	的复数	10	127	100	19238	8183.02	1076.01
15	3881	CARDIN 2000-1	1	6827107	6827107	4		Carlos .	1.1	106	180	103	10210	4044.00	814137
4	1001	CANTELL/MINT-C	1.5	10001144	10001-44	1	12	Conserved in the second	1.5.5	200	184	107	7148.00	0075-00	405.N
a 11	4441	(MATE1+18890-4		Translated 1	mantel			Valantinks.	>	104	1.208	445	40103-005	7981-00	instal.
10	1841	Georgers, reports a	1	#Included	+ hutani		4	- Barrent		Contract of the local division of the local	140	1075	1010.00	1833-00	405.00
	5687	CAMPALY, *1304, *13000		7107642	7103648	1.	0.44	104-0472	-	Ven form Dr.	110	104	20140	0524.00	110.40
44	1001	CASPINES,*16a006		2260713	1708714	14		779	painters.	+ ABCTERY		104	610.00	3488.02	proje
61	1821	2567101a-F173808	1.0	1100218	198078	14	-12-	81.899	Defetation (10	201	100	Norse -	102310-007	104742
10	1881	CARTINIA * (1223)ar	1.1	716403	7100004	- 10		11.05	Services	188	187	100	and an	11148.00	8451.79

Figure 135: Selecting a Note to view

The note is displayed on the screen.

10 - II	Langia T	mittaffer planet (gerland)	Coloman P	BALT	DOT -	ber 1		state insurance 7	Type T	Not Depite W	ALCOUNT 1	hist Day is P	Inf During T	ALCOHOL T	Samples The
44.	100	SWLIETHITIC -		1000	1222700			100			1.00	188	1.00	4771.00	4112.00
	100	the second se	1.1.1	1000	Thereit			-		1	100	10		1000	a france in
68 L	2007	APROPRIATE A		10.07.54	1540124	- 8-	1.4	Revenue 11		A	117	86.7	-81704.000	1010-00	4238.94
18	100	READING TENCH	1.1	IDECR.	1124238	5	1.1	Advantation 2020-01-0		87.	10	17	706.00	95.0	275.18
8	100	eticals, 19800-C	1	100462	1204400	- E.	- 5	water the Miller's		100	101	878	0008.00	4945.00	4845.07
10	100	101.73027-0	1.1	ZHENT	2010147	1	1.2	The is a basely shorts	trained terms	18.7	478	475	1.01	1104.05	1400.0
10.	1987	30x79334e	- 1 I - 1	Janet -	100411	£	17	10000000000		20.	107	148	752.00	8105.00	6276.01
10	0401	1-0880-0880-1		468792	6887707	6		A & Skely Jathogeni		100	105	111	6528.26	5844.00	2142.87
68	1000	CMDH 4 NOR-C		And in case	0000144	1	- 2			94	194	187	71.48.28	4079-30	4475.04
10	1001	1.4a23/10.10000-4	1.4	No.	7840241	.4	1.4			194	1.249	80		740.40	010739
10.	1981	Ciefforia Next I a	C.4	79/061	7167641	1				187	1986	104	10000-00	5488.00	4275,78
12	1987	CAMPAN, PORQ PORTS		7554	1167644		144				198	210	201.00	8608-00	7125-44
6	10001	damfalu Presbeau	1	7160114	1188114	94	1.1			24		1,84-	809-00	1407-00	4818.08
	100	Lighting *1708cm		maple -	1798274	0.	1.4			14	1.001	100	10100	10209-00	1047.01
10	town-	0.000/00.00*0520W	1.1	(Notice)	FTREDA.	41.	1.4			dan .	187	800	11-101-00	11140-00	0493.75
	5967	LANKAT L PODMO-A		retear	(100)47	- 6				14	201	100	1/1510	7664.00	2112.02
62	1942	LANDAL + *17(0)A-15-		Producter.	1104534					100.	144	818	1045-10	4001-00	4481.000
	1001	40401-121122-4		0000	4253474						144		4175.70	6475-00	4036.64
	1001	40461270739716		67010	4/11/17		4				100	891	1.04	114/5/00	1008100
	NMAT 1	mmin *1687*4		distant.	42552757		1.1	40.545	- 1000	100	202	404	000100	4404.40	1048167

Viewing CNV and LOH Events

The variant table has a column selector icon allowing user to configure which columns are displayed. The figure below shows the columns available for display.

- ~	Select Displayed Columns =					
-	Chromosome					
-	• Start					
~	End					
~	Туре					
-	• Length					
-	Copy Number					
-	# Markers					
-	• Mean					
-	Confidence					
~	• Overlap					
~	Sample					
*	Genome Build					
	Classification					
	Depth					
	Frequency					
	Inheritance					
	Quality					
	Estimated Tumour Content					
	ISCN					
	Score					
	Mean Standard Error					
	Mosaicism					
	Mosaicism Range					
	Mosaicism Lower Bound					
	Mosaicism Upper Bound					

Figure 136: Columns available to select for display in the CNV/LOH variants page

Selecting a variant will show it in IGV, a track for both CNV and LOH will displayed. Sometimes there will only be a CNV call as in the example below.

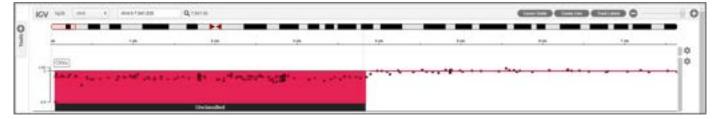


Figure 137: Example of a CNV call only

Sometimes there will only be a LOH call

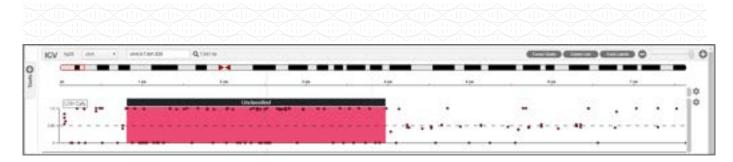


Figure 138: Example of a LOH call only

Sometimes there will be CNV and LOH calls

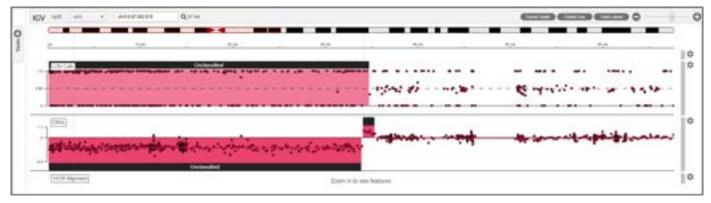


Figure 139: Example of a sample with a CNV call and a LOH call in the same genomic location

CNV and LOH Options

As with the page displaying SNV and Indel calls there are options available for each variant called by the CNV/LOH pipeline,

Right clicking on a variant will provide a menu of the possible options.

18	-12	Times T	1 T.	- Type T.	Longo T	Digg Symbol 7	# Instants T	Allow W	Lindstein T	Directop T.	Servera Balle T	Davellague +	- 240 T	T present	Sample B
	1.1.1	-	Second Second	and strength of the local division of the lo	100			1.4.2	-	oper margine	and the second se	The local division of			800
44	1.48	144.10108	ITTLEMET	Detellary	24280	Add to Shortha	r .	1.810308		(131) (other)	URCHOR	Uniteenfied	111		9218
-	1.1	M/TOPRI	BR/ONAR	Deletion	pola	Carety	3	1.1900		- (00 (arget)	GRONDE	(Inclosed and	. 40		8218
14		2102058	2140258	Deletion	John	Vieta in Catalian	A DESCRIPTION OF	4,7702		OH-hepeti	GNINE	United	216		4218
4		Marine:	techniar:	Dalation	1000	and the second second		-1.200524	1.000	(D) (sept)	UNCTOR .	Universities			4210
4		1001	(BOOK)	Sublish	3.04%	Karlert Links.	>	1.0100	-	(20 Depen	04(1)(8)	increasing and	18.0	0.42	4018
44	. 17	49175805	#425488	Defetions	backenin .		41	4.71410	- mph	COL: (Lergel)	UPCNOE	unitered and	379	6.47	1010
4		287946	205276	Detetion	76.1269			4.90%7		(DC)(Harl)	680108	Unlastfed	-129		8210
48	1.1	INCOME: IN	150369485	Dephasters -	1048		11	6.01116	mp .	(Dispage)	GROOM	Undersified	1386	0.40	8210
44		Services.	ANTIJITION .	Puplication	1008		47	8-401133	No.	(D) Dept	04040	incise/ad	818		4218

Figure 140: Available options for a selected CNV or LOH call

Adding to a shortlist

Variants added to a shortlist are annotated with a tick

1.00	Oursers 1.	Chief P	the second	Company.	Longh N	Charles and the	A MARKING P	interest Pro-	Contraction (P.)	Interna V.	Income Statis	Children P.	Cherris F.	This sector 2	Classifier (B)
100	1	10.1000.000	11,22794.00	Destruction	1008			-4.20405	- magin	COB (tangent)	GACHER	unders the			100
10.0	10	NUTRE OF	CTRAME?	Language 1	3,4,000			1.0000		CER INSTANCE	ORCHER	mounded	100		1070
1.00	T	bertgoes.	THE R. L.	Determs	3009			4.566	-	Childrengelt.	6803486	(maintaine)	100		4016

Figure 141: Variants added to the shortlist displayed

Shortlisted variants can be viewed.

Interpret Cloud User Guide v1-20241029095209

÷					a Angel C	OWNERS @ LONG	ALIN	frage 1 of	n network	M. Applica	The Martine P		and the second	
	Champion 4	Dest T	DOT N	4.7ge T .	Longst T	Day familie T	a intriners T	Aller T	Induced T.	Chertep #	Inter Gamelan Report	Dept. T.	Transmip T	Sample, B
24	- 4	112303338	152303488	Detailor	2016			-4.00940	mgt.	(211 Joseph)	a second s			8216
114	21	14110138	1111.0417	Deterior	1.45/8	1.8		1.01214	inge .	(21)((0)())	a second s	100		6278
11		8875240	Berghad.	Deletion	3006			15.188607	mpt.	COL Hangel)	4 Time U	98.		4210
14	- 16	2100/06	2102258	Existent	2008			4,759	ingh.	(Distinget)	4 If Although 3	205		8210
		14001001	supressed.	beietine.	1040		+	-1,000344	1.000	(distance)	Second Constants	19		82%
		Adda P	Included.	National	1.000		100	1.000	1000	1994 Inscript	carinest contactions	100	0.07	4000

Figure 142: Accessing the shortlist of selected variants

The shortlist opens in a separate view. A variant can be removed from the shortlist be clicking on the red bin icon in the shortlist view.

Value in 19	within the Jampie ADTL (with									
1944 - 1944	-									
Dir .	1041	04	7,00	Longit.	Copy Number	Markets	Aleas 1	Confidence	Overlage	
1	152305335	152305635	DEL	300		. 4	-4.20985	High	costarget	
21	AUG010	17533897	DEL	3421758	1	-0	-1.01204	High	COSOther	

Figure 143: Viewing the shortlist of CNV or LOH variants

Alternatively, a variant can be removed from the shortlist using the CNV options menu.

Case I	Courses T	Start W	1107	- algorito	Longer #	Containing	inter T	Induced V	manual .	Descent State 7	Distance #	They is a	Property P.	Longer B
	A	1000	100175	Surger States	10.000	100 March 100 Ma	diam'r	100	the second	TRONG	and the second s	200		Mint .
24		152385238	15UDEBADE	Datation	2009	Delete from Storright In-	1995	righ	. 201 yargeti-	GROUP	crokesfeet .	. 20		8210
10	31	harmented.	175318447	Deletion	3.40mb	Carsty	5 1014	High	ON other	difficient.	understand	1498		8016
14.7		Serties.	51015040	Deletion	1008	Marrie In Column Street and	BWL-	High	-C211 (1arget)	6403188	inclusified	M		679
14.1	-10	propriet	21912266	Detection	2009	the strange out the	111	nigh	CET (sepera	040108	unidentified	208		8210
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	11	40776895	-	Datasian	14,000	1 4	0.714636	High	(Distance)	GALLOS	indexided	478	647	67-6

Figure 144: Deleting a variant from the shortlist

The shortlist will be updated to reflect the removal of a variant.

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the fact		764	Length	Copy Number	Maters	Man	Conditioners	(noninger	
i 152345335	152305635	045.	300		4	4.20985	mph	CDSTarget	

Figure 145: The shortlist showing that the variant has been removed

Variant Classification

A variant can be classified from the list that is included by default. These are:

- Benign
- Uncertain significance, likely benign
- Uncertain significance
- Uncertain significance, likely pathogenic
- Pathogenic

Additional classifications can be added in the Admin Controls section of the software (Admin Controls > Analysis > Classifications)

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	1	BADDINES.	battelat.	Deletion	1906	Warw in CytoSure to	Alter prof.	1. K. C. S.			68048	Indepted	48		6219
6	4	44967	Distair.	Detectors	3.000			- Denge			GROUP .	Uncharached	144	4.42	10710
	-11	44770888	44225452	· beletim	543885	Earland Lines.		thurt	ain significance:	ikely benign	GPC108	incleasibili	178	1.0	6211
1.1		207108	388276	Deletioni	78.1248	.1		thurt	als significance		OPCN8	Independent	228		8276
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Figure 146: Default classifications

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**	44770888	-46225402	Detection	043845	Veter Links	>	i lonar	Last significance	Marky Isonight	14CH04	Undersfeel	179	6.47	8210
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Figure 147: Classifying a CNV deletion as pathogenic

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0.8411	.7	. Instance	31073546	Deletion	200		R	4,10807	mgh	101 (sept)	880528	United	99		1016

Figure 148: Updating of the variant to show the new classification

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	44	definition .	Automak.	Buginging .	100		10	Autop	-		sachas.	invignment	-		10.0

Figure 149: Removing a variant classification

View Classification History

User can review the classification of a variant by selecting that option in the menu.

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Figure 150: Selecting view classification history option

When chosen a table appears displaying how a variant has been classified, who made the classification and when any changes were made

Classification History		+ ×
lassification history f	or seq[GRCh37] Xq22.1	(100611029_100611266)x3
Classification	User	Date
Unclassified	admin	Oct 21, 2021 4:13:44 PM
Pathogenic	admin	Oct 21, 2021 3:46:36 PM
Unclassified	admin	Oct 15, 2021 9:18:38 AM
Pathogenic	admin	Sep 21, 2021 2:27:30 PM

Figure 151: An example of a variant's classification history

View in CytoSure Interpret

CytoSure Interpret is OGT's class-leading microarray software analysis platform. For existing microarray customers, CNV and LOH events can be loaded into CytoSure Interpret.

38.1	Charmon T.	Bart V.	ing w	.4 1pt 1	in paragric \$1.5	Diese Number W.	0 Hariters W.	Alexan W.	Confidence #	iverag 4	Concerned Bastle W	Conditioner F.	Display W	- Fernance W.	Dangto P
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Figure 152: Selecting to view a CNV deletion in CytoSure Interpret microarray software

It is necessary to have CytoSure Interpret open prior to selecting this option. If it is not yet running Interpret will issue a prompt to the user.



Figure 153: Prompt from Interpret if trying to load data in CytoSure Interpret when it is not running

Variant Links

The software allows users to link out to external sources of documentation. Currently included are:

• EnsEMBL

Interpret Cloud User Guide v1-20241029095209

Additional resources can be added in the Admin Controls (Admin Controls > Analysis > Manage Links).

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Figure 154: Accessing variant links

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25	terristed.	00720008	the privation	4105		1.00	0.407703	ings.	OD-Depet	sitcher	(malassified			6/10

Figure 155: Accessing EnsEMBL as an external source for data annotation

Adding Notes to CNVs

Users can add notes to CNVs

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Figure 156: Selecting the option to add notes to a CNV

When chosen a text editor is displayed as well any pre-existing notes. At the top is an option to choose a file and to then upload it. Below is the text box where details can be entered.

+Add Note To CNV: Notes for 13:50566440-517	82998	
Choose File No file chosen B I U x ₈ x ² R & III - III III S % S Beckground v Foreground v Font v State v	Upload	Existing Notes (0)
Cancel	+ Add Note	

Figure 157: A blank template for creating a note

In the example below a file has been uploaded and text entered.

Add Note To CNA:	
Notes for 13:50566440-51782998	
Additional CNV information.docx	Existing Notes (0)
Background v Foreground v	
Font v Size v	
Additional information that has been gathered for this CNV is included in the attached file.	
Cancel + Add Note	

Figure 158: Addition of text and a file to a note

Selecting + Add Note completes creation of the note and it is added to the existing notes section. Any file that has been uploaded with the note is shown and can be downloaded if required.

+Add Note To CNV. Notes for 13:50566440-517	82998	
Choose File No file chosen	Upload	Existing Notes (1)
	# 5 0	
Eacliground v Fornt v Size v]	▲ 161802 Additional Christianation data B I U x ₁ x ² R A S II II II N N N Beckground Foreground Foreground
Cancel	+ Add No	e

Figure 159: A new note is shown

Now when the user accesses the menu for the variant there is an additional option providing the display of any notes.

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			them in Cy	toliars interp thoat farming											

Figure 160: Existing notes are available to view

If selected, the note(s) are shown in a separate box.



Figure 161: Viewing an existing note

Additional notes can be added as shown below.

•Add Note To CNV:			*:
Choose File No file chosen Upload	Existing Notes (2)		
B I U x ₁ x ³ B ± # 5 0	Admin User 2021-10-22 14:58:01.0	/ 0	- lî
	▲ 145715 Chromosome 13 CNV sets docx		
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	There is an extra file with details of previous chromosome 13	CNV calls.	
	Admin User 2021-10-21 16:19:34.0	1 0	
	B I U x ₁ x ¹ E ± ± S D D		
		Ser -	
	Background V Foreground V Fort V	Size ~	

Figure 162: A blank note template with two existing notes

Manual Creation of CNVs

It may be that the user believes, based on the visual representation of the CNV data, that the software has missed a CNV call and would like to manually generate it. For example, a user may believe that the region highlighted in the screenshot below represents a CNV, but it has not been automatically detected by the software.

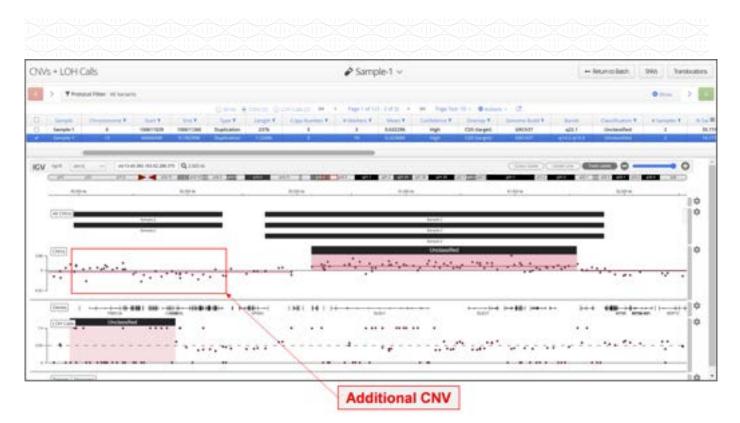


Figure 163: A region, not called by the software as a CNV, that the user wants to manually define as a CNV

In order to manually create the CNV call, the user defines the CNV region by using the mouse to select the region in the chromosome ruler track. Alternatively, the coordinates can be provided in the text box in the menu bar of IGV.

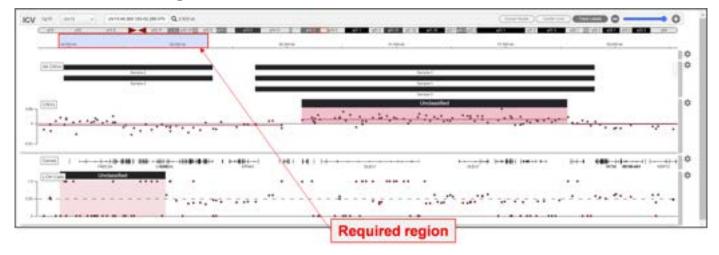


Figure 164: Using the ruler region in IGV to select the region to display

The IGV window resets to the size of the required region

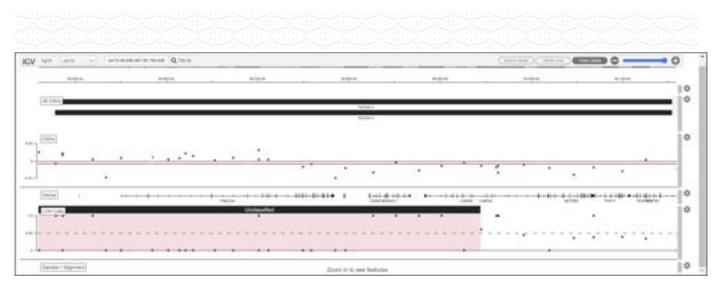


Figure 165: IGV set to the boundaries of the region to be defined as CNV

Select the Add CNV option in the Actions menu.

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e leve	-	annessa.	***	Ballater	1.000			4.50.0000	No.	-	T Har Opener	\$763 gins	inclusion in the		

Figure 166: Selection of the Add CNV option from the Actions menu in the variant table header

Users have the option to define the entire region in IGV as the CNV or the software will snap to the nearest probe at each end.

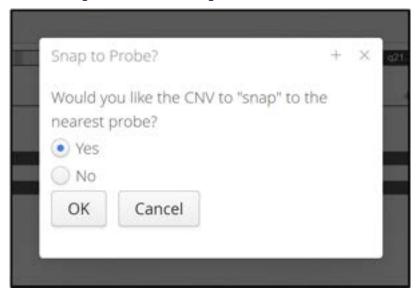


Figure 167: The option to snap to the nearest probe or to keep the region as that displayed in IGV

Once a CNV has been created manually it is **NOT** automatically displayed in the software. The creation of the CNV can be confirmed by the number of CNVs detected that feed into the protocol filter; in the figure below the number has incremented by one to five.

Interpret Cloud User Guide v1-20241029095209

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	April 1		1000114.08	1000011000	(highership)	2216			0.050700	ings.	COLONIDAL	SHOULD .	att+	Unclusional		98.7*

However, in order to display it in the variant table additional steps need to be taken. In the original analysis the protocol and filters did not select the manually defined region as being a CNV and so in order to include it the default protocol CNV filter need to be updated. This can be done in the Admin Controls > Analysis > Protocols section.

Protocols			
• Protocolo		* Protocol DW Rese: Tortaut DW Rese	Q 25.55
Default Protocol		Alt Stravil Baak + 40% 🖕 Reads Point Latt + 4 🔒 Reads Point Right + 4 🖕 Outlig boxe	10.18
Mosaic Protocol		DH .	
CNV Test		PTDN	
	Protacol CNV Filter	Default OW and (OH Filter [32] w	
		Y Protected ONF Filter: Default City and Unit Filter	0.000
		Taple In LOW OR Not + Managerer RA + Make Standard Error on A.S. + # Managerer ro 1. OR OR CB CB CB CB CB CB	Î
	Protocol Translocation Filter	Alt Variants 🖉	
		T Protect Transformation Filter: 40 Instanto	•
		All Test Sector 1	
New Protocol		This Server X Cancel	

Figure 169: The default CNV filter in the default analysis protocol

Users need to create a filter that allows manually created CNVs to be included and this can be added to the default CNV filter. This is shown in the figure below.

Figure 168: The number of CNVs has been incremented by one

Protocols			
₩ Protocols		Y Webson SNV Filmer: Default SNV Film	O Heats
Default Protocol		All Doubles (10%) + (Nami Paratiak () + (Nami Paratiak () + (Out	y binen - 1.0
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	Protocol ONV Filter	(All Variants) or (Source is Manual) (40) 🗸 🗸	
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		Traver (a) ACORE COR Traver (a) ACORE Traver	143
		Tanan vo 2 2 Care handbarr 1 + Meanver 2	
		(Concernence of Mathematics)	
	Protocal Translocation Fill	er Altvalains	

Figure 170: The CNV filter in the Default Protocol has been edited to include OR Source is Manual

Repeating the analysis with the updated filter will result in any manual CNVs being added to the sample variant list.

Merging CNV calls

There are occasions when CNVs are called with small regions in between that the user would like to combine into a single larger CNV.

In order to do this, adjust the scaling in the IGV window such that both CNVs are visible, then right clicking between them in the track will generate a popup menu with the option to Merge Displayed provided.

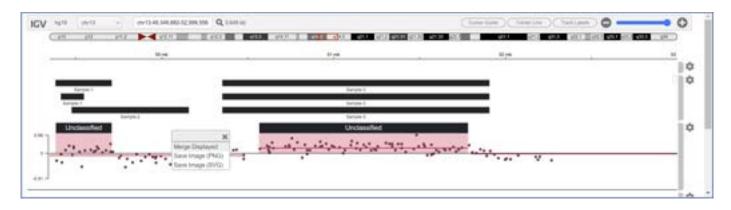


Figure 171: Selecting the option to merge displayed CNVs

If selected the software will request confirmation of the merge option.

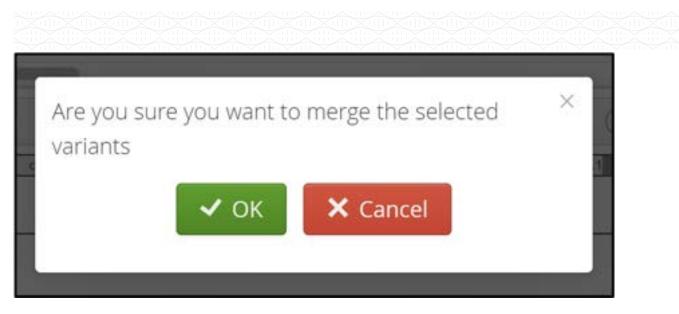


Figure 172: Confirmation of merge option

Following confirmation of the merge option the variant table will be updated. There will be a single row containing the new merged CNV that spans the two previously separate calls. Additionally, the variant counts above the table will be updated.

			(es)	(* (NOVER)	UDW Calls 121	10 .	Page 1 of 1 (1 - 3 of 3).	+ 341 Zug	1 Size. 20 = 1	Actions - C			
	Serge	Chromosome T.	Stort T	DOM: Y	Type W	Langer T	Croy Number T	# Markers W	Mean T.	Carddena T	Overlap T	General Subil V	148
0	Sample-1		1006110079	100611286	Duplication.	2376	3 -	3	0.032296	High	COS (target)	68(3)(7	422.1
	Sample-1	13	50566440	51782998	Dupfication	1.22Mb	9	70	0.203889	High	COS (target)	GRCh/87	q14.3-q1
	Sample 1	13	45354108	49710010	Duplication	126.945	2	21	0.0431794			080437	414.2
	Sector 1		-										
_				* (000)	LOH CARLOS		Page Lat 1 (2 - 2 st 2)		e Size 20 +		Contra V		
0	Sirgin	Outermannin 4	C Card	* (100 C) Del V	UCH+ Calls (2) Type T	M + Lings T	Page 1 of 1 (2 - 2 of 2) Once Names 7	H Pag A Martans T	n San 20 – 1 Mean Y	Confidence V	Overlag 7	Genune Sulti Y	A
0				* (000)	LOH CARLOS				e Size 20 +		Courses 1 COL (sarget) COL (sarget)		

Figure 173: An updated variant table showing the merging of 2 CNVs to a single row in the table as well as the decrease in the number of All variants and CNV variants

Likewise, in the IGV window the two calls are now combined.

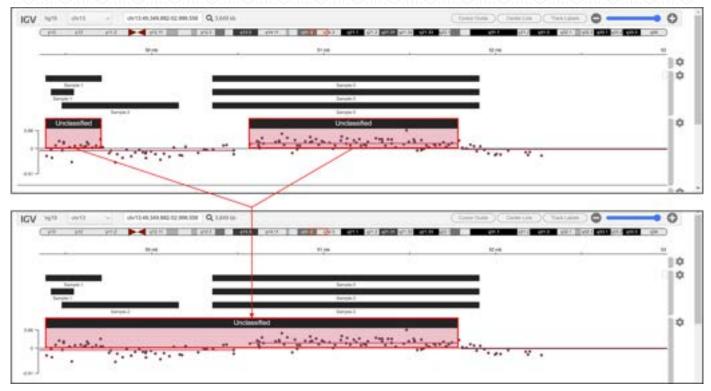


Figure 174: Following the merger of 2 CNVs a single CNV is now displayed

Separating Merged CNV calls

Having been created, users are able to dissolve a merged CNV. Right clicking on the CNV row in the variant table will display the standard popup menu but now with an additional option of Dissolve which will split the merged CNV back into the original separate calls.

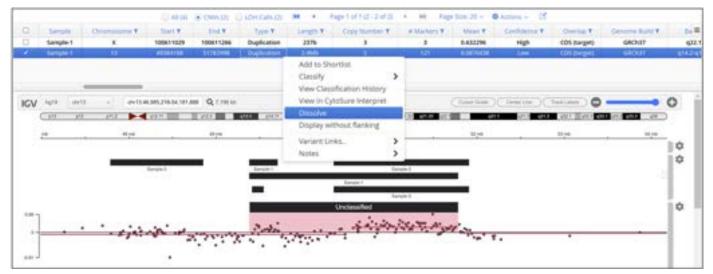


Figure 175: Selecting the dissolve option in CNV variant table

Aneuploidy Plots

Interpret is able to provide an uploidy plots in order for the user to assess whether there is a difference in chromosome number in a set of patients.

This functionality is accessed through the Tools sub-menu in the software dashboard as shown below.

🔆 Home Balthes - Semples Variants Help & Support - 📾 Yook -	(903) (134) 🛔
Charles Assessmentally Paul	A
	A Sysmes Group Company
	oğt

Figure 176: The create aneuploidy plot option

The aneuploidy plot option can only be used when the user is viewing CNV data. If this is not the case then the following error message will be displayed. As with all error messages in the software it can be removed by clicking on it.

Please view the CNV data for which you like to generate an Aneuploidy plot first

Figure 177: Error message from aneuploidy plot option

The figure below illustrates the correct view from which to launch an aneuploidy plot.

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NVs +	LOH Ca	lls				Constant Accounts	Name of State	others ~			- 10	unto Basti	5Nn	Testouton
-	T Posta	of Filter: Definit (NV a	d uphi Filter										0 Tree	2
			CLARINE!	+ OWHIT	O LOW CAR	10,023-00 000	· Page Fordat	40 (r 4) + H	Page Size: 42	0.5mm -	15			
	Sarger	Depressione T	Start #	ins T	Tex V	Langth T	Copy Number 1	a Marterit T	Mean T	Conditions 7	Overlag, T.	Dept. T	(Hequency 1	TTHE LOCAL
	10847		194732218	154702348	Ewletion .	1500		3	4.96529	ange .	CD5-hargeti	1		Not Texted
	12678	1	154722218	154723348	Deletion	1500		87	1,34947	migh .	(D1-harpet)			Not Tented
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	17/018	26	8100230011	##RHOUGH?	Determon	12.9600	.1	8	4.830918	and a	CDI (without)	178		And Tested
	12878	18	MAX21WEE	54521843	Deletter	15/6	1	8	-6.1100004	ange.	CD5 (Larget)	310		Not feeled
	12878	22	223956609	330111626	Deheston	816.6289		12	-6.8140778	math	(24 (other)	152		Not Tested
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	14130	5	78908	33605965	Eviletion	35.8'mm	. t	0.00	-6.990625	High	(04-barget)	175	8.64	Not Texted
	14135	54	106521047	105484801	Deletion	142,9440			4.87598	mpt	(D5 jother)	1		Not Tenned
	34641	14	2152991	(152)47	Deletitor	21496		3	4.957365	might	CH4-Darget)	167		Not Tented
	Aug 1	18	412532946	43123031	Deletter	175.148	1.1		4.417712	mgn .	CDE (withor)	123		Aise Tanied

Figure 178: Selection of aneuploidy plot from menu bar Tools

The user will then be asked to provide the region list that needs to evaluated for plotting. Information on creating a region list is documented in this manual in the Administration Controls > Analysis > Region lists.

Select Regio	n List		+	×
Region List			~	
	View	Cancel	1	

Figure 179: Select a region list pop-up

The available regions lists will be displayed in a drop-down menu; in this example there is a single region list created in the software.

Select Regio	n List	+ >
Region List		~
	Chromosome_1	

Figure 180: Selection of a region from the drop-down list

Following selection of the required region list the user clicks on the View button.

Select Regio			
Region List	Chromos	some_1	~
	View	Cancel	1

Figure 181: Selection of the region Chromosome_1

An example aneuploidy plot is displayed below.

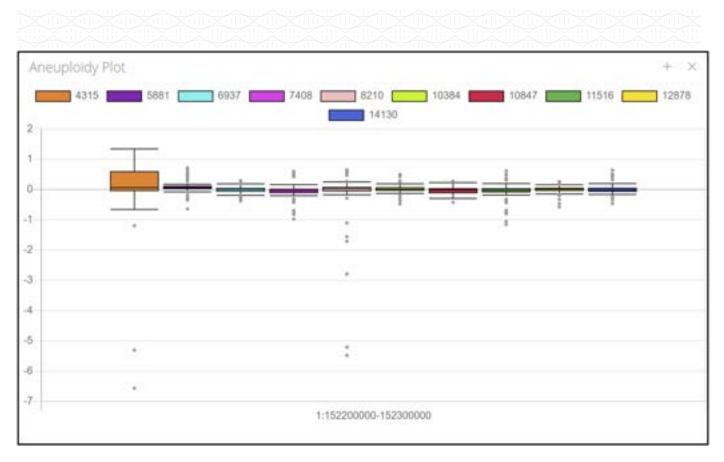


Figure 182: An example of an aneuploidy plot for the chosen region across the samples in the batch

Viewing Translocation Events

The variant table has a column selector icon allowing user to configure which columns are displayed. The figure below shows the columns available for display.

Select Displayed C Sample	xumns =
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cipient Gene	
nor Chromosome	
nor Breakpoint	
cipient Chromosome	
ecipient Breakpoint	
SCN	
onor Locus Reads	
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Value	
Description	
Senome Build	
Гуре	
Classification	
Donor Reads Position	
Donor Orientation	
Recipient Reads Position	
Recipient Orientation	
nheritance	
Proportion	×

Figure 183: Columns available to select for display in the translocations variant page

58427.33.295.805.01.290.805		
	Donor	
	Receiver	
	i. Ar an	

Figure 184: Example of a translocation

Translocation Options

As with the page displaying SNV and Indel calls there are options available for each translocation variant called by the software,

Right clicking on a variant will provide a menu of the possible options.

8	Sample	Donor Gene ¥	Recipient Gene #	Deter Deterosone T	Depor Breakports V	Recover Corporation T	Recipiere Disusport 4	Donor Lucus Reads #	Respond Local
8	111741	-	ARLY	9	Add to Shortlist Class#y	,	1001100		1.41
IGV	1976 (101) (101) (102)	-	C	an ann ann Cant	View Classification H Display with flanking				•
	ene 10	ayana jiyay	19-22-20.310.309-25 Mil-10.296,266.10	antini Adaptine ata	Variant Links	> senctes	0140 100, 101,004 100, 101,014 100,011,00100 100,00	ante acceptione	O CELTRI AND IN

Figure 185: Translocation options

Adding to Shortlist

	Lampie	Durar De		Recipient Carle T	Dunis Desensume T	Dunu Breakpoint T	No.quest	Chromotalite T	Recipseed Breakpoord, T	Dumer Lineus Reads 1	Relignent Locus
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100		0				Add to Shortlist	- One	0.0	and the second	122001	
						Classify	0	5			
IGV	spin -	ant +2	Long Sec.	. 9	21210	View Classification H	atory		Constant of		0
	C #4	11 111	-	11.01 To 12.00 To 10.00	1 101 101 101 101 101 101 101 101 101 1	Doplay with Ranking	81 a	M 11 H	VV 47		C C C C C C C C C C C C C C C C C C C
		marine .	at page 1		200.700. 2020,000 to 2020	Variant Links.	>.		(Av9-130,731,804,130,731,916 100,731,701-00 100,73	one , anyrea	0

Figure 186: Adding a translocation to the shortlist

Once a variant has been added to a shortlist the available option is updated to now allow that variant to be deleted from the shortlist.

*	Lampi	* D	onor Gen	e T Reco	ant fame T	Dance Chromasame T	Danat Breakgairs T	Real Speed	T manual T	Andport Breakpart T	Danie Level Reads T	Recipient Local III
8.	- Mark		808	nski oligi	400.5		2104038		12. No. 1	100171765	CONTRACTOR OF STREET, S	CONTRACTOR NUMBER
182.02	1				- 1999 - 1999 - 1999 - 1999 - 1999 - 1999 - 1999 - 1999 - 1999 - 1999 - 1999 - 1999 - 1999 - 1999 - 1999 - 199		Delete from Shortlist	lbo	(CHERNER AND A		
_			_		10		Classify	0				100
IGV	1428	and .		Louis Search	Q	212.10	Vew Cattlification Hit	story		Contract C		0
	CHI	96	1113	-	1000	#52 PAR 451	Display with flanking		SE 11 H	415 17		
	-	1.00,0	-	1000 Mar 10	0227.000.000-25.0 31.000.000-05	00.715. At 200 per tas 21.20	Variant Links.	>		00-8100.701.00+100.701.010 100.701.70100 100.701.010	ante intigenese	O INCOMPANY
	-		-		-			-	Contraction of the local division of the loc			ALC: NOT THE OWNER WATER

Figure 187: Selecting to delete a variant from the shortlist

Variant Classification

A variant can be classified from the list that is included by default. These are:

- Benign
- Uncertain significance, likely benign
- Uncertain significance
- Uncertain significance, likely pathogenic
- Pathogenic

Additional classifications can be added in the Admin Controls section of the software (Admin Controls > Analysis > Classifications)



Figure 188: Classify the translocation

A variant classification may change over time and it is possible to track the changes and view the classification history.

Interpret Cloud User Guide v1-20241029095209

🗶 Kample Daniel Gene 🕇 Recipient Gene T Daniel Chromosome T	During Drivinguest T	Instance Depression T	And part literat part 7	Dareir Lavest Reads #	Revisient Local II
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ene mayore masjare masjare masjare ma	Variant Links.	>	44481302913041302391318 180292/9596 18029		0 10.711,Mill 16

Figure 189: Viewing a variant's classification history

Initially, the classification will be blank.

		+ ×
290555 > chr9:130731760		
User	Date	
2		

Figure 190: A variant with no classification history

When a classification is made the history table will show the classification type, who made it and when it was made.

Classification History Classification history for	22:23290555 > chr9:130	731760	+ ×
Classification	User	Date	
Benign	admin	21-Apr-2020 16:08:30	

Figure 191: Example of a benign classification

Any updates to the classification will be recorded with previous designations retained.

lassification History ssification history for 22:2329	0555 > chr9:13073176	50 ± ×
Classification	User	Date
Unclassified	admin	21-Apr-2020 16:10:41
Uncertain significance	admin	21-Apr-2020 16:10:08
Benign	admin	21-Apr-2020 16:08:30

Figure 192: Example of a tracking a translocation classification change

Display with Flanking

Users can select to view translocations with flanking sequence

×.	Rangele 1	Darnet Liene Y	Reigneri Gerne	T. Danie Destanante T.	Dural Drakpovit F	Antiper	Distance T	Andport Breakport T	Durin Locus Reads V	Revisient Lintes
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					Classify	>				
IGV	40.00		tion Seatch	Quum	View Classification Histo	ry .			0	0
	C 44 . 15	212	- and	100 CO. 100 CO.	Display with flanking	3-1		11 41		
	and the second	and an an an	4422.23.240.3	86.23.286.711 Fee	Variant Links.	0		44/8130.791.804.130.791.818 181.781/98146 180.791		0

Figure 193: Selecting to show a translocation with flanking sequence

Variant Links

Links to external data sources are available; these are managed in Admin Controls > Analysis > Manage Links

*	Sample	Donor Ge	ur T Rectain	et Gene 🕈 🛛 Dave	r Orienname T	Donce Breakport #	Recipiers	Chrismanne T	Recipient Development 1	Dowor Lance Reads #	Reciptent Locus
				and shares and shares		2528255			530731256	4111	
11.		1.			2.11	Add to Shortlist		1.1	1011/555-01		11111
						Classify	>				1 200
IGV	14.00	1. 1.	Long Steering	Q.212 m		New Classification H	latory		Concession of the local division of the loca	G	0
-	C #10	14 114	-		494 1400	Display with flanking		SE 11 H			
		-	ideal	225,580,886-21,386,711		Variant Links.	62	forward the	saciota		0
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1.0								Google (Gen			14
- 51	T100 Night	train a statement	-					Google Schol	Geriel)	and the second sec	0

Figure 194: Linking out to external data sources

Variant Table Options

Column Sorting

Rows in the variant table can be sorted using the column header. In the example below the results have been sorted by decreasing and increasing allele frequency. Currently, data can only be sorted by one column.

Alt ¥	Abele Frequency T	Type T	ART	* Allele Prequency ¥	Type 🔻	ART	▲ Allele Frequency ▼	Type T
6	500% (M	TRO.	A	100%	SNV	A	21,98%	SNV
c	99.57%	Allele Frequency	T	100%	SNV	G	22.95%	SNV
Α	48.9%		6	100%	SNV	6	24%	Deletio
1	43.24%	SNV	A	100%	SNV	c	24.27%	Deletion
ε	51.07%	SNV	. A	100%	SNV	6	24.69%	Deletio
C	100%	SNV	T	100%	Deletion	A	26.3%	SNV
CT	86.10%	Insertion	6	100%	SNV	c	26.8%	Deletio
t -	40.99%	SNV	6	100%	SNV	AAAACA	27.27%	Comple
¢	47.55%	SNV	T	100%	SNV	6	27.86%	Deletio
A	51.5%	SNV	6	¥ 500%	SNV	G	28.4%	Deletio

Figure 195: Sorting by Allele Frequency

Dynamic Filtering

As shown previously the variants page displays the Protocol Filter, the number of variants detected by the pipeline and presented to the filter is depicted in a red box and the number remaining in a green box.

In the image below you can see that there are 2946 variants (in the red box) detected by the pipeline that are to be filtered based on the settings in the protocol. Subsequently there are 2754 remaining (as shown in the green box).

	SNVs IN	SNVs OUT	
Training plan series We Film			0 142
Said 1	All Marel Sec 1975	· Instructingers · Case, Spectra	2 IN

Figure 196: The filter used by the protocol in the analysis of the sample displayed in the Variants page

However, the user is able to implement additional filtering dynamically. Any column header with the funnel icon **I** can be used as a filter. For example, a user may want to filter on Gene Symbol

- This last play because the transformer to the second tot

Figure 197: Selection of the funnel icon for the Gene Symbol column

In this case they want to see only variants found when the total depth is greater than 200.

Filter on Total I	Depth			×	
Total Depth =					
Total Depth >	200				
Total Depth <					
Total Depth ≥					
Total Depth ≤					

Figure 198: Dynamic filtering of the variants using the Total Depth column

After updating the Variants view now shows a Dynamic Filter window and within it is the "Total Depth > 200" filter.

From the 2754 variants generated by the protocol using the default filter, it can be seen that a further 265 have been removed filtered with 2489 remaining.

Protocol F	Dynamic Filter			
Transations: Select Write		9-18- 1	2	in
Typener Harringt-202	Turket Theyath + 200.0	One Diserter Diserted	2	24

Figure 199: Example of Variants filtered by the protocol filter and a dynamic filter

Dynamic filters can be chained together so additional filters can be added for instance an Allele Frequency greater than 80%

Filter on Allele Fred	uency	×	
Allele Frequency =			
Allele Frequency >	80%		
Allele Frequency <		ci tail	
Allele Frequency ≥			
Allele Frequency ≤			

Figure 200: Selection of another dynamic filter to start creating combinations

Now the Dynamic Filter shows "Total Depth > 200 and Allele Frequency > 0.8" and there are now 1457 variants remaining from the input of 2754.

Taysenia Marte: (Nari of) Mart		Once Streeter Election
3764 3	Total Depth + 2018 . Allete Prequency + 5.4	3. 502

Figure 201: Variants being filtered by a compound dynamic filter

There is no requirement for the user to have to repeat the setting of dynamic filters every time they use the software, there is the option to name the filter and pressing

to retain for re-use.

Alternatively, all dynamic filters can be removed from the display by selecting to

clear the filter

Viewing a Sample in IGV

Selection of a variant in the Variants Table causes it to be displayed in the embedded IGV.

		Thursdown #1	Contract N	and T	ind w	11.4	dama transmis T	Tani T	And Desire #	All Depth #	Table Depity #	Inclusion T	Add (particular)	Quality Scient W.	Lange
	BALLA THEFT.C		1008798	1228/98			Table -	and a	-	108		0.00	4771.00	4122.00	1441
	District Manual		1942152	1942742	1.1	1	41.179	100		102	100	11.01	Nam	6791.47	5461
	displaced to be		+424142	+12010			40.00	0.00	188	127	142	0104.00	4034.00	4775.04	5881
	Adaptala https://www.	1	1114(25	1014020	1		45.04%	544	10	18		1105.000	\$15.00	275.18	5881
	and a hadden	Ý.	Tillevill.	Concession of the			1007	200	141	Apr.	124	National Contraction	4444-05	middar.	3441
1	Min "MUT-C	4.1	13Meter	1204247	1.8.1	1	1979	stev		170	177	6.00	18742-00	14003.78	1001
	Min Welling	1	10HHIT	1000007	5	13	85.19%	insertion	20	147	148	154.00	8111.00	4279.01	144
	CANTRAL 2000-T	+	4847797	6867702	6		40.07%	599	190	180	369	1523.00	4644.99	3142.20	500
	CAMBADS Arthol	+	0002144	00007-04	+	£	47.50%	544	100	164	58/7	7145.07	4079-00	405.04	5481
	CHARGES TOTOL 4	Ť.	TRADERT	164,0007			51.3%	100	194	100	44	1000.00	1181.00	0101.00	548
1	CV 400 ml		-	Q.0	teaper			100,000					100,000		-
	chapma	1.84.0716													
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		the second s		****			*****								
	the second	the second s		****	••••										
		the second s		****	••••										

Figure 202: A variant selected and the aligned displayed in IGV

Within the IGV window there are several options for modifying the data being displayed.

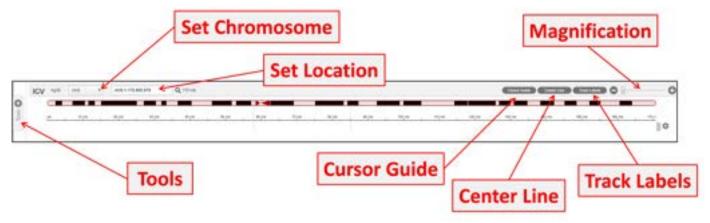


Figure 203: Display options for IGV

By default, the sequence viewer is centered upon the selected variant but users can drag the display upstream and downstream of the variant position. Also, it possible to zoom in and out via the magnification slider at the top of the window.

Additionally, the tracks displayed can also be modified via the setting options available on right hand side of the viewer . For example:

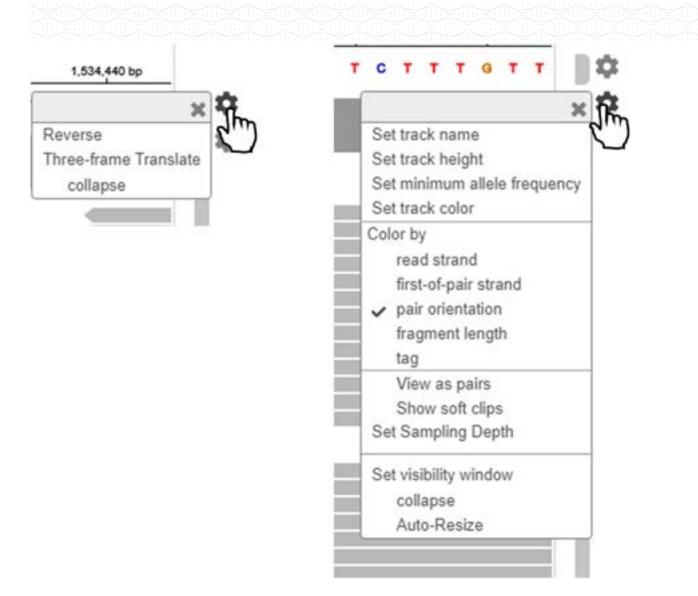


Figure 204: Display options for the integrated IGV browser, firstly using a left click on the mouse and secondly using a right click



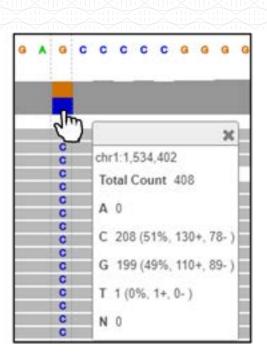


Figure 205: Display options available for sample reads, firstly with a left click and secondly with a right click

Using Tracks

Users can add or remove data tracks to the IGV view. This can be from publicly available sources or from proprietary internal or subscription-based sources.

Tracks can be added in the Software section of the Admin Controls (Admin Controls > Software > Annotation) and documentation of how to do this is in this section of the user guide.

To use this functionality, users need to access the Tools tab of IGV

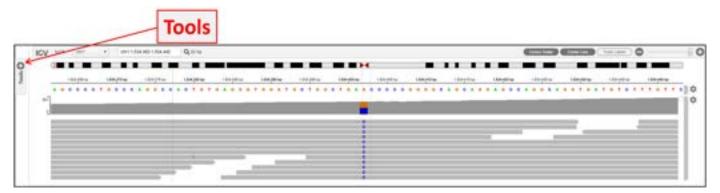


Figure 206: The Tools tab for adding data tracks to annotate an alignment displayed in IGV

Once accessed selecting the drop-down arrow will list the available tracks.

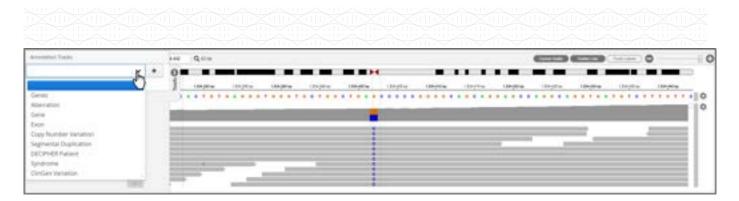


Figure 207: The drop-down list of data tracks available

Select the data track to be added.

Figure 208: Selection of a track

And then click on the 🛨 icon to add it to the set of tracks for the software to display

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Gereil V 🐈	0		
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		the second se	

Figure 209: Click on the + icon to add the data track to the display

The selected track will be displayed. It can be removed by clicking on the minus icon

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Genes	 +	0=			-	 (-	-	_	00
eded Tracks.		-	100 pt 1	 		 	 	100,000		 		0
								÷.	a .			
					1							

Figure 210: The new track is now loaded

Select any further tracks to add to the view

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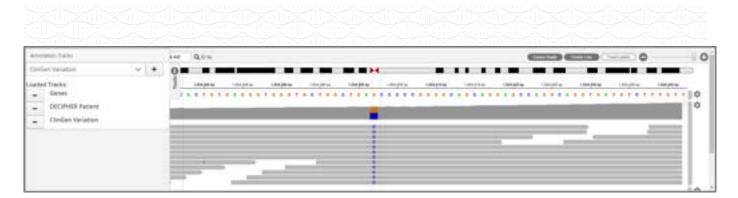


Figure 211: Addition of the required tracks

Finally, close the Tools tab and the data tracks will be displayed.

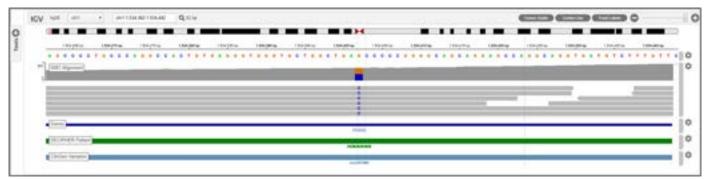


Figure 212: Display of the selected data tracks following closure of the tools tab

'Popping out' of the IGV display

There is, potentially, a substantial amount of information that can be displayed in the IGV view. To accommodate the information and make it easier for the user it is possible to 'pop out' the IGV view into a new browser tab.

This is accomplished using the button in the display



Figure 213: Button to allow display of IGV into a new tab in the browser

Selecting Multiple Samples

As discussed above users can opt to view multiple samples simultaneously by selecting them in the batch view.

for talacted, plant	Comple	ord famples										
SWo/odels	10	Sample	Vest				# 5800	# CMIs	#104	Report	00	
CWULDH Gally		3481				-	2754		10		-	
						-					100	
	10	7408	=		4.00	8 top:	2,740	7	12		1999	
	- 81-	8210			4.07	1 m	2,666	10	16			
		1004	(H)	an II (1996)	4.00	8 top:	2,650	4	32		(100)	
	- 6	10647		w meinen	410	A 10	2,689	3	10			
	10.	11518		-	4.0	A 141	2,571	7	36			
	. 10	12878			4.00	8 tor.	2,427	14	18			
	-18	14130		-	4.10	Birg.	2,614	18	14			

Figure 214: Selecting multiple samples to view in the Variants page

When multiple samples are selected there will be separate tracks for each sample in IGV. This makes it possible to compare the same variant in different samples.

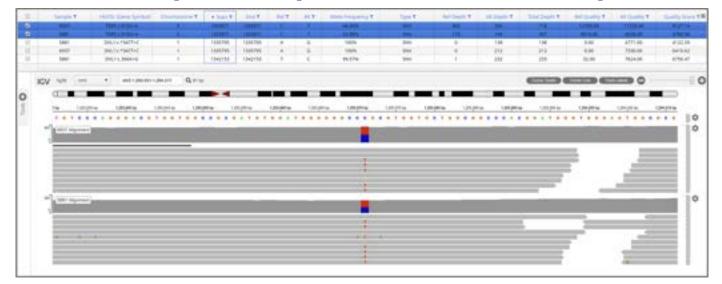


Figure 215: An example of two samples sharing a variant as displayed in the integrated IGV browser

Variant Table Options

There are options within the Variant table accessed using the Actions drop down menu.

				ins a Page 1 of	101-10010 + 101	Page Son: 10 -	C Action (10			
*	Sample .	Donor Gene W	Recipters Gene #	Dover Chromosome T	Done Breakpoint V	Recipiered Chris	Generate Report	inter T	Dever Letter Rends V	Rectilent Loous
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Figure 216: Accessing the options in the variant table

Reporting

Results can be exported by clicking on the Generate Report button below the variant table.

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10	1887	04840-71070-0		261491	383491	1.	7	1029	NW .		327	877	6.00	19147-06	11796.05
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Figure 217: Selecting the Generate Report option from the variant table header menu

Interpret provides multiple types of report and for each of these types there are templates. These are highly customisable and updates can be easily applied in Admin Controls-Analysis-Reports.

When selecting to generate a report, the initial window allows users to select the type of report to generate.

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Report Type		~		
		rt shortlisted variants		
Columns		ill columns ude displayed columns		
	📥 Genera	ate Report		
62				

Figure 218: Initial report option

Default reports supplied with the software are listed.

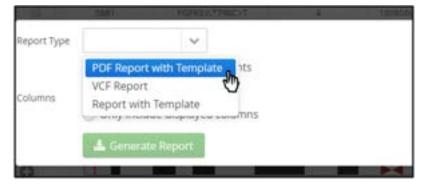


Figure 219: Selection of PDF report type

Once the report type has been selected the user needs to specify the template to use.



Figure 220: Selection of the template to use with the PDF report type

Once all options are chosen, pressing Generate Report will create the PDF file and the web browser will download it.

Sample	Chromosome	Start	End	Length	Genome Build	Ref	Alt	Type
5881	6	1801977	1801977	06	GRCh38	Τ	с	SNF
5881	4	1806167	1806167	06	GRCh38	G	А	SNV
5881	4	1807400	1807402	26	GRCh38	CGT	с	Deletion
5881	4	1808060	1808060	06	GRCh38	с	r	SNT
5881	4	2820740	2820740	66	GRCh38	G	Т	SNF
5881	1	2824673	2824673	06	GRCh38	T	с	SNF
5881	1	2834468	2834468	06	GRCh38	с		SNF

Figure 221: An example of the PDF report generated

Other options are included, for instance an HTML based report.

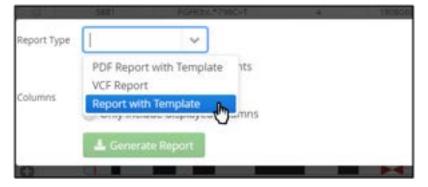


Figure 222: Selecting a template type report

Again, a template needs to be chosen

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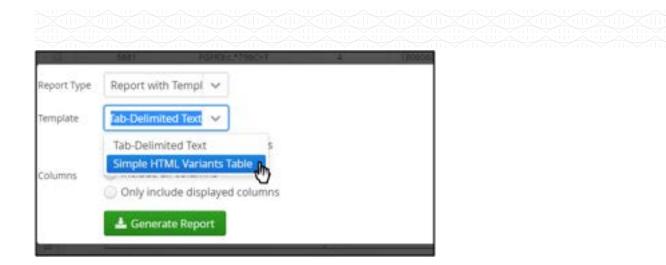


Figure 223: Selection of a HTML format report

The HTML formatted report is then generated and available,

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Figure 224: An example of the HTML report

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Figure 225: Options available for configuring the view in IGV

Display Flanking

Users can choose whether or not to display flanking sequence in the IGV display.

				in a Page 1 of	thread a mi	Page Sept. 13 9	Actions -			
*	Sample	Donor Gene #	Recipient Gene T	Debit Oramiane #	Dance Breakpoint #	Recipient Chris	Generate Report	DOT N	Donor Local Reads #	Recipiers Laton
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							HE With selected	101	Artage Tracks	-

Figure 226: Selecting the Display flanking option in the Actions menu

Manage Tracks

Users can add or remove data tracks to the IGV view. This can be from publicly available sources or from proprietary internal or subscription-based sources.

Tracks can be added in the Software section of the Admin Controls (Admin Controls > Software > Annotation) and documentation of how to do this is in this section of the user guide.

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	(P8 P	4 943 • •		eas		-	H With selected		Mariage Tracks 👌	

Figure 227: Selecting the manage tracks options

The available tracks will be displayed in a pop-up window and users can select the tracks that they want to add to the display.

Genes Compressed BED (*.bed.gz) RefSeq	a	Name	File Type	Source
	1	Genes	Compressed BED (*.bed.gz)	RefSeq

Figure 228: Tracks available to display

Once the required tracks are selected, users can press Update to update the IGV display.

Viewing Analysis Results by Variant

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8	Name	File Type	Source	=
	Genes	Compressed BED (* bed.gz)	RefSeq	

Figure 229: Selecting tracks to add to the IGV display

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Figure 230: Displaying of tracks in the IGV display

Viewing Analysis Results by Variant

As results of samples are generated, they are stored in the Interpret database and can be analysed from a variant-centric point of view

Accessing of this viewpoint is via the Variants button on the dashboard menu bar shown in the figure below.



Figure 231: Selection of Variants from the Dashboard menu bar

Selecting the Variants tab in the menu bar opens up a new page to display all the variants recorded in the database.

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Viewing Analysis Results by Variant

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Figure 232: The start page for viewing variants

There is a substantial amount of information available in the variants page and the different sections are highlighted in the figure below.

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No. Vector V VMM VM		 2		Contractor Broom	 · · · · · · · · · · · · · · · · · · ·			_
		 	 Control Control of C			10-1-1MUW1		ju j rij
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Figure 233: The different sections of the variant page

There a number of active regions

• Filter Pane

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The filter pane allows for dynamic filtering of the variants. By default the filter is set to All Variants, so all variants are displayed in the variants table, however these can be refined according to your specific requirements.

• Variant Table

This displays all variants in the database that meet the filtering requirements of the dynamic filter. By default this is for displaying all variants.

• Sample Table

When a row in the variant table is selected all samples that contain the selected variant will be displayed in this table.

• IGV Pane

Selection of samples in the

• Notes

Users can add notes to variants.

• Quick Filters

These are selection of options that allow users to rapidly filter variants on the basis of some general conditions.

Clicking on the icon on far right of the column headers in the variant table will display all the columns that can be selected for display in the variant table.

HGVSc (Gene Symbol)	HGVSc
Chromosome	Canonical?
Start	rsID
End	Minor Allele Frequency
Ref	Minor Allele
Туре	gnomAD - Total
Genome Build	gnomAD - African
Alt	gnomAD - Latino
Context Length	gnomAD - Ashkenazi Jewish
Genomic Context	gnomAD - East Asian
HGVSp	gnomAD - European (Finnish)
HGVSc	gnomAD - European (non-Finnish)
Classification	gnomAD - South Asian
# Samples	gnomAD - Other
% Samples	ClinVar Significance
#	Gene ID
Most Severe Consequence	Gene Symbol
Impact	Transcript ID
Consequence Terms	Exon Number
PolyPhen Prediction	Protein ID
PolyPhen Score	Length
SIFT Prediction	Transcript Resolution Method
SIFT Score	Exon ID

Figure 234: Columns available to select for display in the variant table

Similarly clicking on the same icon on the sample table provides a series of column options.

Sample ID	
Allele Frequency	
Ref Depth	
Alt Depth	
Total Depth	
Quality Score	
Ref Quality	
Alt Quality	
Genotype	
Zygosity	
Inheritance	
Log Ratio	
# Ref Reads (+)	
# Ref Reads (-)	
# Alt Reads (+)	
# Alt Reads (-)	

Ref Strand Blas	
Alt Strand Bias	
Reads Placed Left	
Reads Placed Right	
Sex	
Homozygosity	
Read 1	
Read 2	
Read 1 Size	
Read 2 Size	
Batch Name	
Batch Date	
User	
Protocol	
Panel	

Figure 235: Columns available to select for display in the sample table

Dynamic Filtering

The dynamic filters are the same set of options discussed in the previous section. They can be accessed from the Actions drop down menu shown below.



Figure 236: Accessing the dynamic filtering options

The dynamic filtering window provides a detailed set of options for investigating the variants stored within the Interpret database.

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Figure 237: Full filtering options available to filter variants

Filtering by Quick Filters

Selecting the Quick Filters tab on the side of the variants page opens a tab that provides some options for quickly drilling down into variants of interest. The options currently available are to select based on a classification type, the NGS panel or gene.

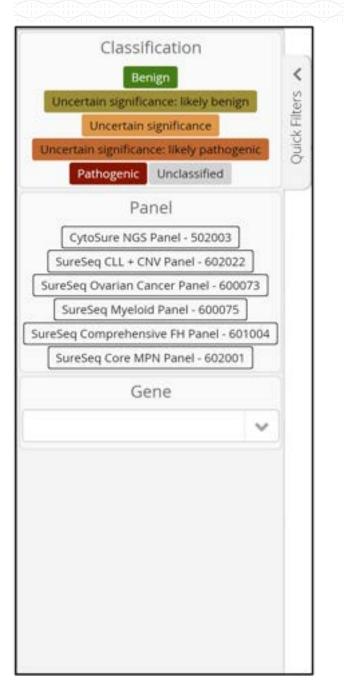


Figure 238: Quick filter options

Classification or Panel can be selected by pressing the corresponding buttons with multiple selections allowed. To filter by genes start typing the gene name in the text box matching values will be displayed.

Ger	ne
SKI	~
SKI - ENSG0000015	7933
SKI	
SKINTL - ENSGOOD	00242267
SKINTL	
SKIL - ENSG000001	36603
SKIL	
SK/V2L2	
SKIV2L2 - ENSGOOD	00039123
SKIV2L	
SKIV2L - ENSG0000	0204351

Figure 239: Using quick filters to select for the gene SKI

Once a gene is selected it will be displayed as below and can be removed by clicking on the x next to the gene name.

 Gene	
	~
SKI ×	

Figure 240: Using quick filters to filter by the SKI gene

The dynamic filter is now updated and shows that, from the input of 8432 variants, there are only 10 found within the SKI gene.



Figure 241: Displaying only variants in the SKI gene

Additional genes can be selected and these will be displayed in the same way.

	Gen	ie
		~
	SKI × D	VL1 ×

Figure 242: Using quick filters to select variants in the SKI or DVL1 genes

When the 2-gene filter is applied the output now increases to 19 variants being displayed.

T Dynamic Hiters: In 201 or 201,1		Crede \$ Charlen
8117 >	In 10 ar DVL1	5 10
	A M M A RELEASED FROM A M Apple 21 - BANK	

Figure 243: Displaying only variants in the SKI or DVL1 genes

Displaying Variants

Each row of the variant table represents a variant that has been detected in at least one sample. Selecting a variant displays all the samples in which the variant is present. In the figure below, the variant DVL1:c.*347T>C is present in 10 samples.

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Figure 244: Selecting a variant displays all samples in which it is present

Subsequently, selecting any of the sample or samples rows will display the alignment for the variant in the corresponding sample.

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Figure 245: Display of the alignment for 3 samples in IGV

Adding Notes to Variants

It is possible to for users to add annotations to variants through the notes function. When a variant has been selected and there are rows populated in the sample table, the user can make a right click on one of these. From the popup menu select the Notes > Add Note options

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Figure 246: Selecting the Add Note option

A window is displayed with a text box where up to 250 characters can be used. Any other pre-existing notes will also be shown.

ating Notes (0)

Figure 247: Note creation template

The user can enter the required notation.

Add Note Te Variant		+ ×
Notes for DVL1:c.*347T>C		
Characters remaining 154	E Existing Notes (C)	
This is an important variant that needs to be monitored.		
Cancel + Add Note		

Figure 248: Note creation

Then selecting + Add Note completes the process and the existing notes section is updated to include the newly created note.

+ Add Note To Variant	s for DVL1:c.*347T>C				
Notes for DVL1:c.*347T>C	2				
Characters remaining 250	Existing Notes (1)				
	Admin User 2021-01-19 21:25:00.0	/ 1			
	This is an important variant that need	s to be monitored.			
Cancel + Add N	iote .				

Figure 249: Note generation

Once all changes have been made, the notes window can be closed and the view will return the normal variant display with the note now being displayed in the Notes panel.

					B	The second secon		Apr 10 10 10 10							
And the second s	Address of the	 	 -Parameter 1	Index 7	and the second	And A	Income T	and the	and strength	damping the	Technold 1	- second -	second .	Inches and	
The A photographic service that we also be consid-				10.000	- not include	(managers)		100		10000		-14			
ty advect & recard pays on prove	1000	10		100.00	iter tonis	inclusion (1.00	100	10	100.00		100		10.010	1004
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		 100		100.010	and height	increase in		4/06-		2000	-	1.00		100	1418
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	1.1	100		100.000	And Rocks		1.1	476		·	14				-
		 10	 +	100.048	Any factors	increase in the		1.00		ALC: NO.	100		+	- 100	- 100
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				10.714	feer factors	the second second	1.0	1000		101.0	1.0	14		1978	inter a
		 	 	100.000	and looks	-		- 100		1000					-

Figure 250: Displaying a note in the note panel

A note can be deleted by pressing the red rubbish bin icon. If the Confirm Delete option is then selected the note will be removed.

 Add Note To Variant: 		-+-:X
Notes for DVL1:c.*347T>C		
Characters remaining 250		
Character's remaining 250	Existing Notes (1)	
	Admin User 2021-01-19 21:25:00.0	
	Cancel A Confirm Delete	
Cancel + Add N	lote	

Figure 251: Deleting a note

Users can also edit a note by clicking on the pen icon; which will show the note in a text box where changes can be made. The update is confirmed by the pressing the Apply button.

+ Add Note To Variant:			+
Notes for DVL1:c.*347T>C			
Characters remaining 250	Existing Notes (1)		
	Admin User 2021-01-19 21:25:00.0	+ Revert	ED Apply
	Characters remaining 140		
	This is an important variant that need Update: this variant is now classified of		
Cancel + Add Note			

Figure 252: Update an existing note

The notes panel is subsequently updated to show the revised note.

										Pres 2							10000 (T
And and a second s	CONTRACTOR /	CONTRACT OF	A DECKER AND A	The Design R	the state of some P and	and the set of	And in case of the local division of the	manufact P	And in case of the	manual ?	transfer 4	And And Address of The Owner of	Although a fill	A REPORT OF	The second second	or there has TW	
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	1014			14	100.00		1.04		interaction of the	the fitness i	101/11						
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Figure 253: Display of an updated note

Administration Controls

These are accessed by selecting the Admin Controls options in the user drop down menu.





There are 4 main parts to the admin controls:

- 1. Overview
- 2. User Controls
 - Current Users
 - Add Users
- 3. Analysis
 - Manage Samples
 - Current Analyses
 - Protocols
 - Panels
 - Region Lists
 - Variant Lists
 - Classifications
 - Metric Sets
 - Manage Links
 - Filters
 - Preferred Transcripts
 - Reports

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• Guidelines

- 4. Software
 - Software Overview
 - Annotation
 - Advanced SettingsPlug-ins

Attribute Definitions

Attribute	Variant Type	Description
#	SNV/Indel	Variant Database Identifier
# Alt Reads (-)	SNV/Indel	Number of alternative alleles on negative strand
# Alt Reads (+)	SNV/Indel	Number of alternative alleles on positive strand
# Markers	CNV/LOH	Number of bins (markers) used to identify CNVs
# Ref Reads (-)	SNV/Indel	Number of reference alleles on negative strand
# Ref Reads (+)	SNV/Indel	Number of reference alleles on positive strand
# Samples	SNV/Indel	Number of samples the variant is present in the database
% Samples	CNV/LOH	Percentage of samples the variant is present in the database
% Samples (Similar CNVs)	CNV/LOH	Number of samples in the database with an overlapping CNV
Allele Frequency	SNV/Indel	Allele Frequency
Alt	SNV/Indel	Alternate allele
Alt Depth	SNV/Indel	Number of reads supporting the alternative allele at the position
Alt Quality	SNV/Indel	Sum of alternative base qualities at the position
Alt Strand Bias	SNV/Indel	Sequencing bias in which one DNA strand is favoured over the other in the reads containing the alternative allele (Percentage)
Bands	CNV/LOH	Location of the variant on the chromosome
Batch Date	SNV/Indel	Date the batch was performed
Batch Name	SNV/Indel	Name of the batch containing the run

Attribute	Variant Type	Description
Canonical?	SNV/Indel	A flag indicating if the transcript is denoted as the canonical transcript for this gene
Chromosome	CNV/LOH	Chromosome of the CNV/LOH
Chromosome	SNV/Indel	Chromosome of the variant
Classification	CNV/LOH	User-assigned classification of the variant
ClinVar Significance	SNV/Indel	Clinical significance of variant according to ClinVar (e.g. benign, pathogenic, uncertain significance etc.)
Confidence	CNV/LOH	Confidence that the call is correct (e.g. High, Low) dependant on the Standard Error of Mean
Consequence Terms	SNV/Indel	Most severe outcome caused by the specific variant (e.g Frameshift variant, Stop gained, Synonymous variant etc.)
Context Length	SNV/Indel	Length of the genomic context overlapping the variant
Copy Number	CNV/LOH	Number of copies of the CNV event
Depth	CNV/LOH	Depth of coverage of the sample at position of the CNV
Description	Translocation	Donor and recipient gene symbol pair
Donor Breakpoint	Translocation	Position on the donor chromosome of the translocation
Donor Chromosome	Translocation	The chromosome number of the donor gene
Donor Gene	Translocation	Donor gene symbol where the translocation originated
Donor Locus Reads	Translocation	Read depth at the donor breakpoint position
Donor Orientation	Translocation	Orientation (strand) of the donor gene
Donor Reads Position	Translocation	Which side of the breakpoint the donor read lies (left or right)
End	CNV/LOH	Genomic position of end of CNV
End	SNV/Indel	Genomic position of end of variant

Estimated tumour content (only used in cancer Estimated Tumour Content CNV/LOH panels) Unique ID for the exon SNV/Indel

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Exon ID

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Attribute	Variant Type	Description
Exon Number	SNV/Indel	The number of the exon in the gene the variant is present
Frequency	CNV/LOH	Average Allele Frequency of common SNP's overlapping the CNV
Fusion Type	Translocation	The method of detection that highlighted the fusion, either: Over-expression of the gene (Expression), detection of a known fusion by sufficient supporting reads (Canonical), detection of an unknown fusion by sufficient supporting reads (Non-canonical)
Gene ID	SNV/Indel	Unique ID of the gene where the variant is
Gene Symbol	SNV/Indel	Gene symbol where the variant is
Genes	CNV/LOH	List of genes overlapping the CNV
Genome Build	CNV/LOH	Genome assembly version
Genomic Context	SNV/Indel	Genomic context the variant is overlapping (Low Complexity, Homopolymer, Simple Repeat)
Genotype	SNV/Indel	Genotype (Heterozygous/Homozygous)
gnomAD - African	SNV/Indel	The frequency variant appears on the Genome Aggregation Database from people of African decent
gnomAD - Ashkenazi Jewish	SNV/Indel	The frequency variant appears on the Genome Aggregation Database from people of Ashkenazi Jewish decent
gnomAD - East Asian	SNV/Indel	The frequency the variant appears on the Genome Aggregation Database from people of East Asian decent
gnomAD - European (Finnish)	SNV/Indel	The frequency the variant appears on the Genome Aggregation Database from people of Finnish decent
gnomAD - European (non- Finnish)	SNV/Indel	The frequency the variant appears on the Genome Aggregation Database from people of European (Non-Finnish) decent
gnomAD - Latino	SNV/Indel	The frequency variant appears on the Genome Aggregation Database from people of Latino decent

Attribute	Variant Type	Description
gnomAD - Other	SNV/Indel	The frequency the variant appears on the Genome Aggregation Database from people of another decent
gnomAD - South Asian	SNV/Indel	The frequency the variant appears on the Genome Aggregation Database from people of South Asian decent
gnomAD - Total	SNV/Indel	The frequency variant appears on the Genome Aggregation Database from all reference genomes
HGVSc	SNV/Indel	The HGVS coding sequence name
HGVSc (Gene Symbol)	SNV/Indel	The HGVS coding sequence name with the Transcript identifier replaced with its Gene Symbol
HGVSp	SNV/Indel	The HGVS protein sequence name
Homozygosity	SNV/Indel	Proportion of the genome covered by LOH regions larger than 5Mb
Impact	SNV/Indel	The Impact score according to Ensembl VEP of the genetic variation in the genetic sequence (e.g. LOW, MODERATE, HIGH etc.)
Inheritance	CNV/LOH	Estimated inheritance of the variant based on the presence of the variant in parental results, if available.
Inheritance	SNV/Indel	Estimated inheritance of the variant based on the presence of the variant in parental results, if available.
Inheritance	Translocation	Estimated inheritance of the variant based on the presence of the variant in parental results, if available.
ISCN	CNV/LOH	CNV/LOH variant encoded according to ISCN (International System for Human Cytogenomic Nomenclature)
ISCN	Translocation	Translocation variant encoded according to ISCN (International System for Human Cytogenomic Nomenclature)
Length	CNV/LOH	Length of CNV
Log Ratio	CNV/LOH	Mean log2 ratio of sample/reference of the CNV
Mean	CNV/LOH	Rescaled mean log2 of sample/reference of the CNV (only used in cancer panels)

Attribute	Variant Type	Description					
Mean Standard Error	CNV/LOH	Standard Error of the Mean					
Minor Allele	SNV/Indel	Base of the minor allele					
Minor Allele Frequency	SNV/Indel	Rate at which the second most common allele occurs					
Mosaicism	CNV/LOH	Estimate of the percentage of mosaicism observed in CNV region					
Mosaicism Lower Bound	CNV/LOH	Estimate of the lower bound of mosaicism observed in the CNV region					
Mosaicism Range	Mosaicism Range CNV/LOH Estimate of the range of mosaicism the sample						
Mosaicism Upper Bound	observed in the CINV region						
Most Severe Consequence	SNV/Indel	Most severe outcome caused by the specific variant (e.g Frameshift variant, Stop gained, Synonymous variant etc.)					
Normalised Expression	Translocation	The expression of the baited gene relative to the housekeeping genes and normalised by total read count					
Overlap	CNV/LOH	Genomic context of the CNV					
P Value	Translocation	Probability of observing the translocation					
Panel	SNV/Indel	Panel used for the analysis					
PolyPhen Prediction	PolyPhen Prediction SNV/Indel The prediction of how dama based off the PolyPhen Sco						
PolyPhen Score	SNV/Indel	The probability that a substitution is damaging (e.g. 0.25 benign, 0.5 possibly damaging, 0.95 probably damaging)					
Proportion	Translocation	Proportion of split reads over total reads at the donor breakpoint					
Protein ID	SNV/Indel	Unique ID for the protein					
Protocol	SNV/Indel	OGT Interpret software protocol used to analyse the run					
Quality	CNV/LOH	(Not implemented)					
Quality Score	SNV/Indel	Phred Quality score of the variant					

Attribute	Variant Type	Description					
Ratio	SNV/Indel	Ratio of depth observed in duplicated PTD exons compared to the exons in the rest of the gene					
Read 1	SNV/Indel	File name of the FASTQ from R1 reads					
Read 1 Size	SNV/Indel	Size of the FASTQ file from R1 reads					
Read 2	SNV/Indel	File name of the FASTQ from R2 reads					
Read 2 Size	SNV/Indel	Size of the FASTQ file from R2 reads					
Reads Placed Left	SNV/Indel	Number of reads with supporting evidence to th left of the variant					
Reads Placed Right	SNV/Indel	Number of reads with supporting evidence to the right of the variant					
Recipient Breakpoint	Translocation	Position on the recipient chromosome of the translocation					
Recipient Chromosome	Translocation	The chromosome of the recipient gene					
Recipient Gene	Translocation	Recipient gene symbol where the translocation ended up					
Recipient Locus Reads	Translocation	Read depth at the recipient breakpoint position					
Recipient Orientation	Translocation	Orientation (strand) of the recipient gene					
Recipient Reads Position	Translocation	Which side of the variant the donor read lies (left or right)					
Ref	SNV/Indel	Reference nucleotide base					
Ref Depth	SNV/Indel	Number of reads supporting the alternative allele at the position					
Ref Quality	SNV/Indel	Sum of alternative reference qualities at the position					
Ref Strand Bias	SNV/Indel	Sequencing bias in which one DNA strand is favoured over the other in the reads containing reference allele (Percentage)					
rsID	SNV/Indel	SNP id from NCBI dbSNP					
Sample	CNV/LOH	ID of the sample containing the CNV					
Sample	SNV/Indel	ID of the sample containing this variant					
Sample	Translocation	ID of the sample containing this variant					
Sample ID	SNV/Indel	ID of the sample containing this variant					

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Attribute	Variant Type	Description					
Score	CNV/LOH	LOH score (Higher scores >30 indicate a higher confidence in the call)					
Sex	SNV/Indel	Inferred chromosomal sex of the sample (Male, Female, Unknown)					
SIFT Prediction	SNV/Indel	Prediction of how detrimental a variant will be to protein function (The opposite of Polyphen in terms of numbering)					
SIFT Score	SNV/Indel	A score that predicts whether a variant will affect protein function (O = deleterious , 1 = tolerated)					
Source	CNV/LOH	Tool used for CNV identification					
Start	CNV/LOH	Genomic position of start of CNV					
Start	SNV/Indel	Genomic start position of variant					
Supporting Reads	Translocation	The sum of split and discordant reads in support of the fusion call					
Total Depth	SNV/Indel	Depth of coverage at the position					
Transcript ID	SNV/Indel	Unique ID of the specific selected transcript					
Transcript Resolution Method	SNV/Indel	Method used to determine which transcript to use					
Туре	CNV/LOH	Variant type (e.g. CNV, LOH)					
Туре	SNV/Indel	Variant type (e.g. SNV, ITD, PTD, etc.)					
Туре	Translocation	Variant type (e.g. Translocation)					
User	SNV/Indel	Login name of user which ran the batch					
VEP Version	SNV/Indel	Version of Ensembl Variant Effect Predictor					
Zygosity	SNV/Indel	The degree at which both copies of the chromosome have the same genetic sequence (e.g. Homozygous or Heterozygous)					

Table 3: Definitions of attributes displayed in various tables in Interpret

Product-specific Guidance

Minimal Residual Disease

Overview

Detection and monitoring of Minimal Residual Disease (MRD) with the SureSeq Myeloid MRD Panel is made possible in Interpret through:

- 1. The ability to specify "Hotspots" (variants) which should be specifically interrogated by the pipeline for their presence at very low frequency.
- 2. The ability to visualise the change in allele frequency of these hotspots in multiple sequencing runs over time.

Discovery Mode

In order to identify candidate variants for use in "Monitoring Mode", where specific variants are interrogated by the pipeline in order to determine their allele frequency at very low depth, it may be necessary to process samples in "Discovery Mode". Discovery Mode uses the standard SNV, Indel and ITD detection algorithms built into OGT's NGS analysis pipeline to report all variants present in a sample above a specific allele frequency and according to other quality-related criteria. To process samples in Discovery Mode:

- 1. Click on the **Batches** button in the toolbar and select **Run Batch**.
- 2. Enter a name for the batch in the Batch Name field.
- 3. Select the SureSeq Myeloid MRD Panel from the Panel drop-down list.
- 4. Select **Discovery Mode** from the **Protocol** drop-down list.
- 5. Select the samples to be processed from the list of available samples such that they are displayed in the **Selected Samples** table.
- 6. Click Run Analysis.
- 7. Click OK.

Once the batch has been started, the **Batch** page will be displayed showing the current status of the processing of the samples in the batch. The status of each sample will be updated automatically (unless the web interface is shut down automatically due to inactivity – see <u>Automatic Shutdown</u> above), and, on completion, the **Completed Samples** table, displaying a summary of the results and relevant QC metrics, will appear.



Minimum Allele Frequency

By default, Discovery Mode is configured to detect variants at a minimum allele frequency of 1%. To reduce this value in order to increase the sensitivity, modify the Discovery Mode protocol as follows:

- 1. In the top-right corner of the screen, click on the user icon and select Admin Controls.
- 2. In the menu on the left-hand side, select Analysis -> Protocols.
- 3. In the Protocols list, select Discovery Mode.
- 4. Click the Edit button at the bottom of the screen.
- 5. Scroll down and select the Advance Pipeline Configuration tab.
- 6. In the SNV Detection section, modify the value of Minimum Alt Fraction as required.
- 7. Click Save.

Hotspot Monitoring

In order to visualise the results of hotspot monitoring:

1. Select**Tools** -> **Hotspot Monitoring Report**, and select the sample/source to be reported.

Select 5	ampie And Runs			+ X
Select S	ample			
	MCWT58			v
	s of MRDEWCWTSR			
0	Collection Owe-	Analysis Debr	Ratifi Alame	
O	(2)	2.)ui 2023	Batch 123	
D	1	30 May 2023	Batch 87	
	2	19 Mar 2023	Batch 54	

Figure 255: Selecting the sample(s) to be reported

2. If necessary, enter the **Collection Date** of the sample(s). This only needs to be carried out once for each sample and will be remembered for future reports. Click **N** ext.

elect Sa	ample	And	Run	S							+ ×
elect Sa	mple										
MRDEN	NCW	TS8									~
nalyses	of MR	DEM	CWTS	8							
	Col	lectio	n Date	e -	A	nalys	is Date	6	Batch Name		
32	60	07/0)4/22	2	2	Jul 2	023		Batch 123		
	«	ć	Ap	ril 20	22		> >>			Save C	ancel
(1)	Mon	Tue	Wed	Thu	Fri	Sat	Sun	3	Batch 87		
	28	29	30	31	1	2	З				
	4	5	6	7	8	9	10		Cancel		
	11	12	13	14	15	16	17	P -			
	18	19	20	21	22	23	24				
= 4	25	26	27	28	29	30	1	0			
17.5	2	3	4	5	6	7	8	0			

Figure 256: Entering the collection date of the sample

3. Select the hotspots to be reported using the same method described in <u>step 4e-f in</u> <u>the Selecting Hotspots</u> section and click View.

List	0	Variant	>		Selected Variants
My hotspot list		NRAS:c.182A>T	<		JAK2:c.1849G>T
Another variant list	0	DNMT3A:c.2644C>T		*	FLT3:c.2503G>T
		SF3B1:c.2219G>A		~	JAK2:c.1611_1616del
		IDH1:c.394C>T			
	0	GATA2:c.599del			
		TET2:c.3782G>A			
		NPM1:c.860_863dup			
		EZH2:c.1253G>A			

Figure 257: Selecting Hotspots to include in the report

4. A graph containing the allele frequencies of all selected hotspots in all selected sample runs will be displayed, along with tabs allowing the user to view the results for individual hotspots. Graph images may be exported by right-clicking of the graph and selecting "Save Image". The table containing the data underlying the graph may be exported as a CSV via the **Export Data**button.

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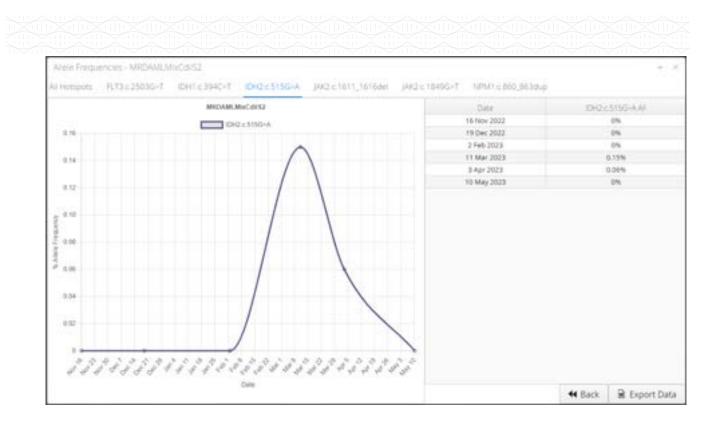


Figure 258: An example of a report

Monitoring Mode

To determine the allele frequency of hotspots in a batch of samples at very low depth, the samples should be processed using the "Monitoring Mode" protocol:

- 1. Upload the FASTQ files for the batch using the method described in the <u>Uploading</u> <u>FASTQ Files</u> section above.
- 2. Click on the **Batches** button in the toolbar and select **Run Batch**.
- 3. Enter a name for the batch in the **Batch Name** field.
- 4. Select the SureSeq Myeloid MRD Panel from the Panel drop-down list.
- 5. Select Monitoring Mode from the Protocol drop-down list.
- 6. Select the samples to be processed from the list of available samples such that they are displayed in the **Selected Samples** table.
- 7. Click Run Analysis.
- 8. Click OK.

Once the batch is complete, allele frequencies of hotspots selected in the Monitoring Mode protocol for a specific sample may be viewed by clicking on the **SNVs** button in the **Completed Samples**.

Selecting Hotspots

To select hotspots for use in Monitoring Mode, they must first be selected from variants identified in Discovery Mode:

1. In the **Batch** page for the Discovery Mode batch, click on the **SNVs** button in

- the Completed Samples table for a sample that may contain potential hotspots.If necessary, filter the list of variants in the Variants page in order to identify potential hotspots more quickly.
- 3. For each variant to be monitored in Monitoring Mode:
 - a. Right-click on the variant in the table.
 - b. Select Add to...
 - i. If no Variant List has been created yet:
 - 1. Click New List
 - 2. Enter a Name for the list (e.g. "Hotspots")
 - 3. Click Create
 - ii. Otherwise, click on the name of the Variant List.
- 4. Once all required hotspots have been added to the Variant List, modify the "Monitoring Mode" protocol to use those hotspots:
 - a. In the top-right corner of the screen, click on the user icon and select Admin Controls.
 - b. In the menu on the left-hand side, select Analysis -> Protocols.
 - c. In the Protocols list, select Monitoring Mode.
 - d. Click the Edit button at the bottom of the screen.
 - e. Scroll down until the **Hotspots** table is displayed, and click on the name of the Variant List created in step 3 in the **List** column.
 - f. In the **Variant** column, select all variants whose allele frequencies should be monitored, and click on the > button to add them to the **Selected Variants** table.
 - g. When all variants have been added to the **Selected Variants** table, click the **Save** button.



Batch hotspot selection

The list of variants included in the Hotspots list for the Monitoring Mode protocol should cover all variants to be monitored in all samples in a batch. If different variants are relevant to different samples, (preferably) create the super-set of these variants in the protocol, or create separate protocols for each set of variants, and run the samples in different batches using the appropriate protocol.



Hotspots not detected in Discovery Mode

If the variant required for monitoring has not been detected in Discovery Mode, contact OGT for assistance to add the variant to a variant list.

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