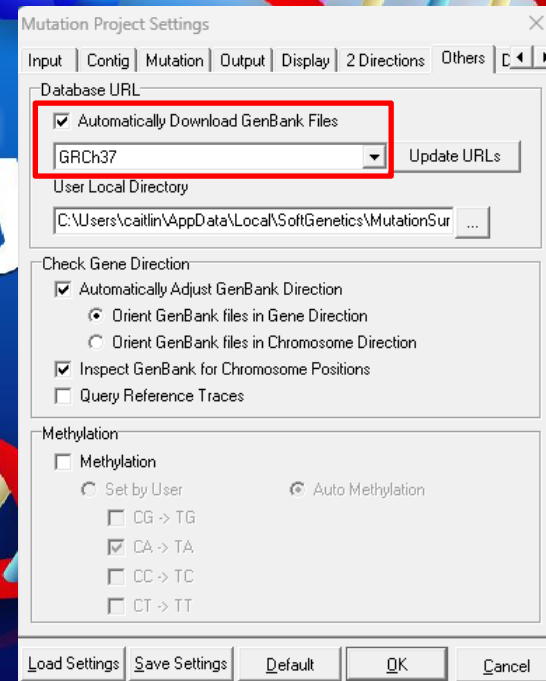


Importing Mutation Surveyor Data into Geneticist Assistant

GenBank Files Used for Analysis in Mutation Surveyor Must Contain Chromosomal Coordinates

GenBank files automatically downloaded by Mutation Surveyor using either the GRCh37 or GRCh38 selection will include chromosomal coordinates.

This setting can be accessed in the Project Settings under the Others tab



Create a Custom Report of the Analysis (Reports -> Custom Report)

The screenshot displays the Mutation Surveyor interface. The main window shows a list of sample files and a table of mutations. The 'Custom Report Settings' dialog box is open, showing the 'Format' tab. The 'Table Format' section has 'Generate Group Consensus' selected. The 'Grouping' section has '1st Order' set to 'Sample ID' from character 1 to 6. The 'Group Summary' section has 'Do Not Show Group Summary Information' selected.

No.	Sample File	Reference File	Dir	Gene	Exon	RF	Start	End	Size	Quality	Mut#	Mutation1	Mutation2	Mutation3
1	185729_BRCA1_10	BRCA1_AmplD_201	1-F	BRCA1	10	2	21054	21392	239	25	0			
2	264381_BRCA1_10	BRCA1_AmplD_201	1-F											
3	185729_BRCA1_10	BRCA1_AmplD_201	1-F											
4	264381_BRCA1_10	BRCA1_AmplD_201	1-F											
5	185729_BRCA1_10	BRCA1_AmplD_201	2-F											
6	264381_BRCA1_10	BRCA1_AmplD_201	2-F											
7	185729_BRCA1_10	BRCA1_AmplD_201	2-F											
8	264381_BRCA1_10	BRCA1_AmplD_201	2-F											
9	185729_BRCA1_11	BRCA1_AmplD_201	3-F											
10	264381_BRCA1_11	BRCA1_AmplD_201	3-F											
11	185729_BRCA1_11	BRCA1_AmplD_201	3-F											
12	264381_BRCA1_11	BRCA1_AmplD_201	3-F											
13	185729_BRCA1_11	BRCA1_AmplD_201	4-F											
14	264381_BRCA1_11	BRCA1_AmplD_201	4-F											
15	185729_BRCA1_11	BRCA1_AmplD_201	4-F											
16	264381_BRCA1_11	BRCA1_AmplD_201	4-F											
17	185729_BRCA1_12	BRCA1_AmplD_201	5-F											
18	264381_BRCA1_12	BRCA1_AmplD_201	5-F											
19	185729_BRCA1_12	BRCA1_AmplD_201	5-F											
20	264381_BRCA1_12	BRCA1_AmplD_201	5-F											
21	185729_BRCA1_12	BRCA1_AmplD_201	6-F											
22	264381_BRCA1_12	BRCA1_AmplD_201	6-F											
23	185729_BRCA1_12	BRCA1_AmplD_201	6-F											
24	264381_BRCA1_12	BRCA1_AmplD_201	6-F											
25	185729_BRCA1_13	BRCA1_AmplD_201	7-F											
26	264381_BRCA1_13	BRCA1_AmplD_201	7-F											
27	185729_BRCA1_13	BRCA1_AmplD_201	7-F											
28	264381_BRCA1_13	BRCA1_AmplD_201	7-F											
29	185729_BRCA1_14	BRCA1_AmplD_201	8-F											
30	264381_BRCA1_14	BRCA1_AmplD_201	8-F											
31	185729_BRCA1_14	BRCA1_AmplD_201	8-F											
32	264381_BRCA1_14	BRCA1_AmplD_201	8-F											
33	185729_BRCA1_15	BRCA1_AmplD_201	9-F											
34	264381_BRCA1_15	BRCA1_AmplD_201	9-F											
35	287531_BRCA1_15	BRCA1_AmplD_201	9-F											

Use the *Table Format* and *Grouping* Settings on the Format Tab to output the consensus for an entire sample. In this example, Generate Group Consensus is needed because more than one paired sample is included per Sample ID and Sample ID from character 1 to 6 can be used to identify the location of the Sample ID name.

Create a Custom Report of the Analysis

Custom Report Settings

Format | Filters | Display | **Mutation** | Color | Nomenclature | Other

Show Mutation Call Information

Show	Show	Filter	Lower Limit	Upper Limit
<input type="checkbox"/> Chromosome Position	<input checked="" type="checkbox"/> Score (Individual)	<input type="checkbox"/>	5.00	20.00
<input checked="" type="checkbox"/> Nucleotide Position	<input type="checkbox"/> Total Score (2-Directional)	<input type="checkbox"/>	20.00	30.00
<input checked="" type="checkbox"/> Reference Nucleotide	<input type="checkbox"/> Mutation Peak Intensity	<input type="checkbox"/>	500	900
<input checked="" type="checkbox"/> Mutation Nucleotide	<input type="checkbox"/> Dropfactor	<input type="checkbox"/>	0.10	0.30
<input checked="" type="checkbox"/> Reference Amino Acid	<input type="checkbox"/> Overlapfactor	<input type="checkbox"/>	0.20	0.50
<input checked="" type="checkbox"/> Amino Acid Position	<input type="checkbox"/> S/N Ratio	<input type="checkbox"/>	1.00	2.00
<input checked="" type="checkbox"/> Mutation Amino Acid	<input type="checkbox"/> Noise	<input type="checkbox"/>	300.00	1000.00
<input type="checkbox"/> Confidence (!)				
<input type="checkbox"/> Comments				

Show Non-Mutation Call Information

Show "n.a." When Data Is Not Available

Show Normal Alleles

- Show All Positions
- Show Negative SNP
- Show Negative Mutations

Show Normal Allele Information

- Nucleotide Position
- Reference Nucleotide
- Normal Nucleotide
- Reference Amino Acid
- Amino Acid Position
- Normal Amino Acid

General Information

- Reported Variant Annotation
- Genotype
- Nucleotide Intensity
- Phred Score
- NM_Score

Display Information

One Field

- Report the Deletion and Insertion as DelIns
- Mutation Call Nucleotides Relative to Gene Direction
- Show Advanced Display

20 # of Nucleotides Displayed Before/After Mutation

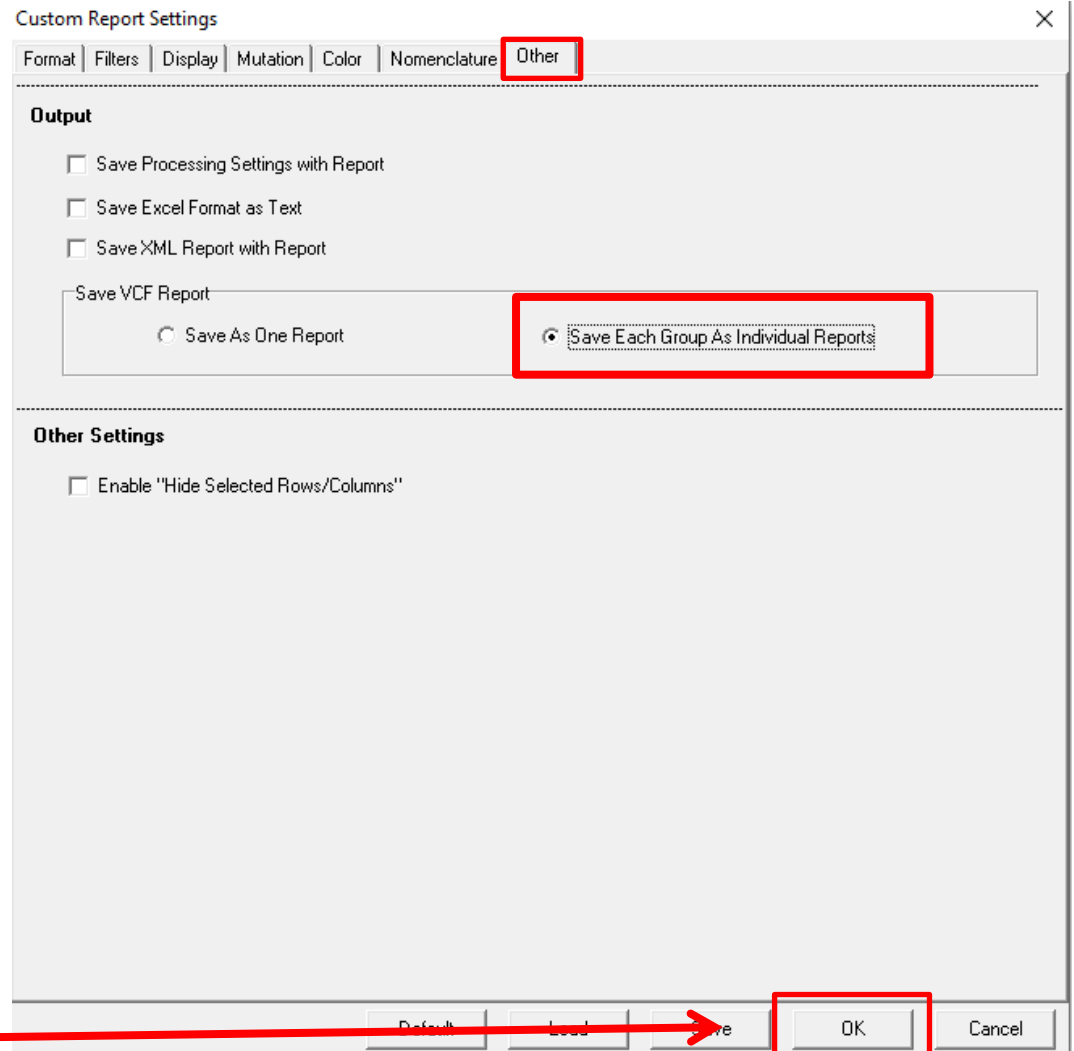
One Field Multiple Fields Display Position

Default Load Save OK Cancel

If "Genomic" Nomenclature is selected for the report, make sure "Chromosome Position" is **deselected** in the *Show Mutation Call Information* located on the Mutation Tab.

Create a Custom Report of the Analysis

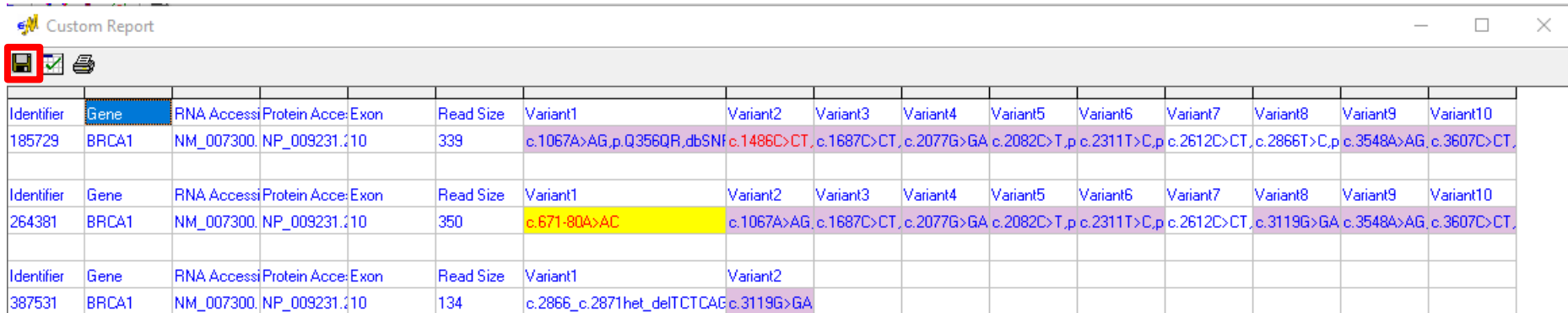
If more than 1 Sample ID exists in project, then make sure to set *Save VCF Report* option to “Save Each Group As Individual Reports” located on the Other Tab.



The screenshot shows the 'Custom Report Settings' dialog box with the 'Other' tab selected. The 'Output' section contains three unchecked checkboxes: 'Save Processing Settings with Report', 'Save Excel Format as Text', and 'Save XML Report with Report'. Below these is the 'Save VCF Report' section, which has two radio button options: 'Save As One Report' (unchecked) and 'Save Each Group As Individual Reports' (checked). The 'Other Settings' section has one unchecked checkbox: 'Enable "Hide Selected Rows/Columns"'. At the bottom, there are buttons for 'Default', 'Load', 'Save', 'OK', and 'Cancel'. The 'Other' tab label, the 'Save Each Group As Individual Reports' radio button, and the 'OK' button are highlighted with red boxes. A red arrow points from the text 'Click OK when done' to the 'OK' button.

Click OK when done

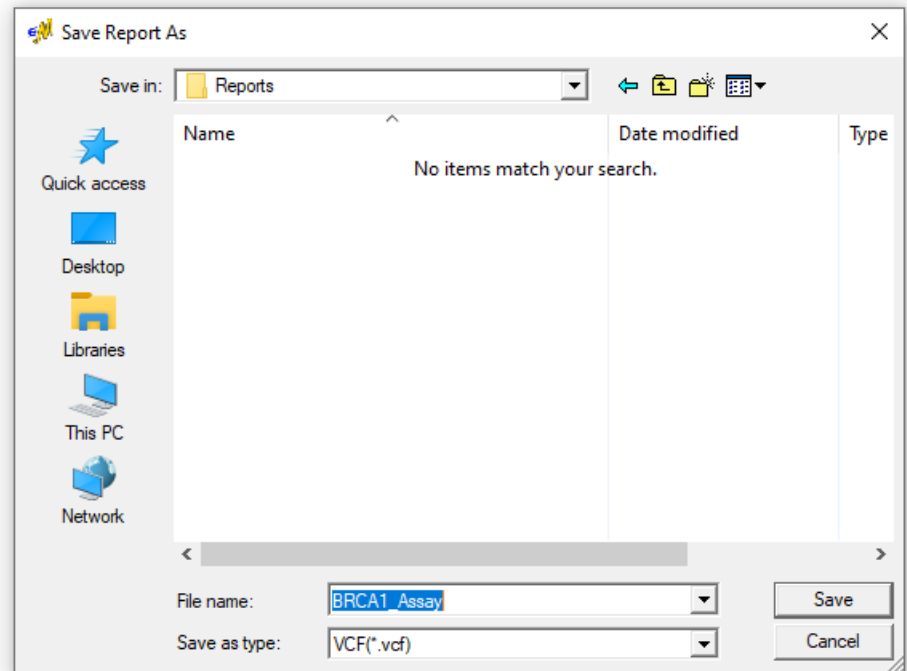
Create a Custom Report of the Analysis



Identifier	Gene	RNA Accessi	Protein Acce:Exon	Read Size	Variant1	Variant2	Variant3	Variant4	Variant5	Variant6	Variant7	Variant8	Variant9	Variant10
185729	BRCA1	NM_007300	NP_009231.10	339	c.1067A>AG,p.Q356QR,dbSNF	c.1486C>CT	c.1687C>CT	c.2077G>GA	c.2082C>T,p	c.2311T>C,p	c.2612C>CT	c.2866T>C,p	c.3548A>AG	c.3607C>CT
264381	BRCA1	NM_007300	NP_009231.10	350	c.671-80A>AC	c.1067A>AG	c.1687C>CT	c.2077G>GA	c.2082C>T,p	c.2311T>C,p	c.2612C>CT	c.3119G>GA	c.3548A>AG	c.3607C>CT
387531	BRCA1	NM_007300	NP_009231.10	134	c.2866_c.2871het_delTCTCAG	c.3119G>GA								

Confirm 1 line of mutation calls per Sample

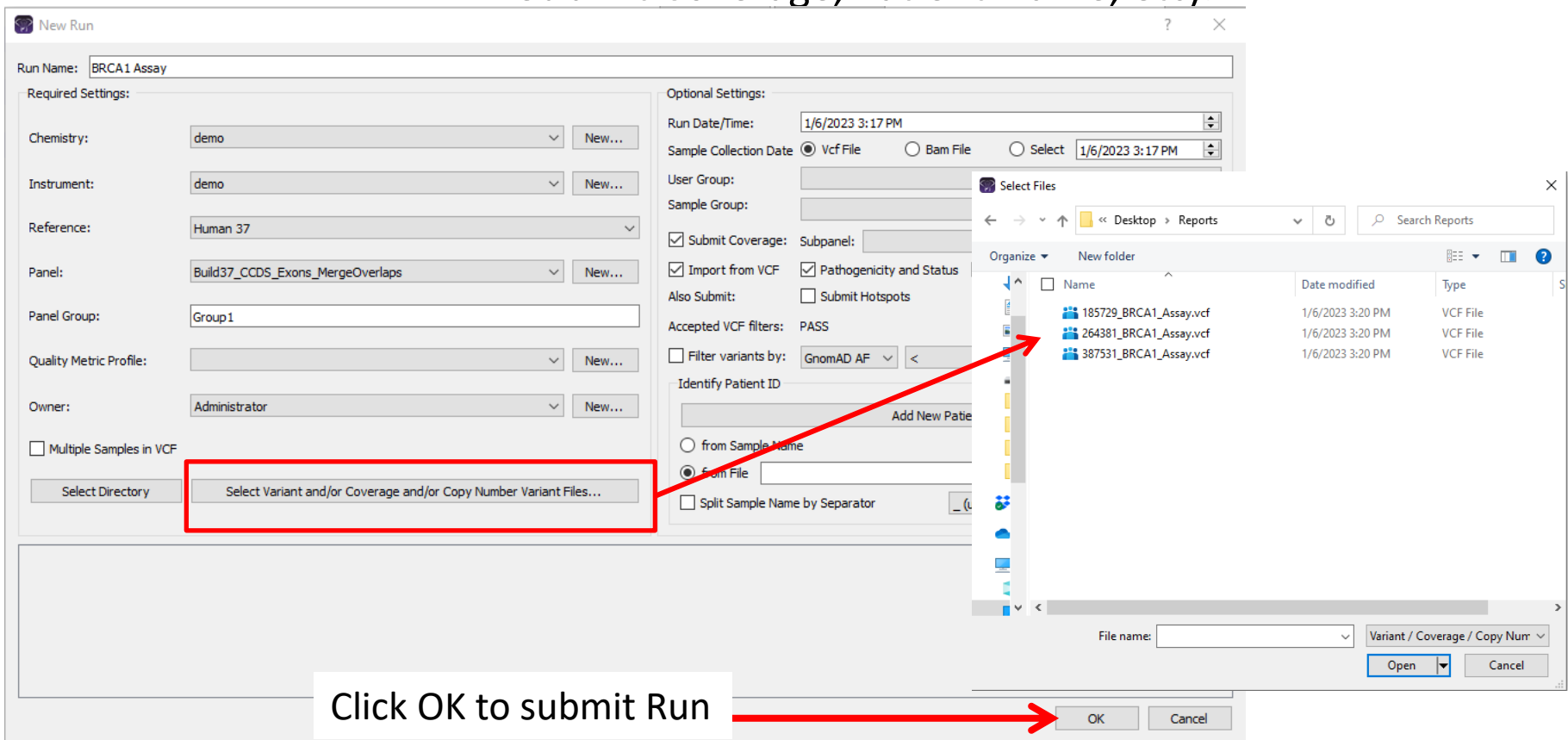
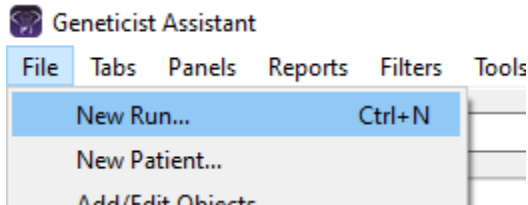
When ready, use the Save icon to save the Report. The default file type is VCF.




Prepare a New Run in Geneticist Assistant

Enter a name for the Run, select appropriate panel (should be same as any corresponding NGS data) and load VCF Files.


Both Sanger and NGS data can be submitted in same run. Change other preferences as desired (Sample name, Submit Coverage, Patient Name, etc).



Review Submitted Data in Geneticist Assistant

 Geneticist Assistant

File Tabs Panels Reports Filters Tools Backup Help

Refresh 

Current Jobs Runs Run 'BRCA1_Assay'

Samples of 'BRCA1_Assay':

ID	Name	Run Date Time	Add Date Time	Run	Build:
37	387531_BRCA1_Assay	1/6/2023 3:34 PM	1/6/2023 3:35 PM	BRCA1_Assay	Build:
36	264381_BRCA1_Assay	1/6/2023 3:34 PM	1/6/2023 3:35 PM	BRCA1_Assay	Build:
35	185729_BRCA1_Assay	1/6/2023 3:34 PM	1/6/2023 3:35 PM	BRCA1_Assay	Build:

Run's Status Changes:

Type	Value	User	Date	Comment
Run Status Change	New	Administrator	1/6/2023 3:35 PM	

Comments of "':
No Data

Reports of 'BRCA1_Assay':
No Data

Use Geneticist Assistant to Help Validate NGS Data

Sample Comparison of 2 Samples:

Patient External ID	HGVS Genomic	1:Zygosity	2:Zygosity	Chr : ChrPos	Rs	Ref	Alt	Ref AA	Alt AA	Type	Pathogenicity	ClinVar Significance	Gene	Exon Number	CDS Number	HG
264381	g.41243941G>A	heterozygous	heterozygous	17:41243941	rs62625308	G	A	Arg	Ter	nonsense	Deleterious	Pathogenic	BRCA1	10	9	NM_007294
264381	g.41244000T>C	heterozygous	heterozygous	17:41244000	rs16942	T	C	Lys	Arg	missense	Likely Benign	Benign	BRCA1	10	9	NM_007294
264381	g.41244429C>T	heterozygous	heterozygous	17:41244429	rs4986852	C	T	Ser	Asn	missense	Unassigned	Benign	BRCA1	10	9	NM_007294
264381	g.41244936G>A	heterozygous	heterozygous	17:41244936	rs799917	G	A	Pro	Leu	missense	Unassigned	Benign	BRCA1	10	9	NM_007294
264381	g.41245237A>G	homozygous	homozygous	17:41245237	rs16940	A	G	Leu	Leu	synonymous	Benign	Benign	BRCA1	10	9	NM_007294
264381	g.41245466G>A	homozygous	homozygous	17:41245466	rs1799949	G	A	Ser	Ser	synonymous	Benign	Benign	BRCA1	10	9	NM_007294
264381	g.41245471C>T	heterozygous	heterozygous	17:41245471	rs4986850	C	T	Asp	Asn	missense	Unassigned	Benign	BRCA1	10	9	NM_007294
264381	g.41245861G>A	heterozygous	heterozygous	17:41245861	rs80356898	G	A	Gln	Ter	nonsense	Deleterious	Pathogenic	BRCA1	10	9	NM_007294
264381	g.41246481T>C	heterozygous	heterozygous	17:41246481	rs1799950	T	C	Gln	Arg	missense	Unassigned	Benign	BRCA1	10	9	NM_007294

Use the “Shared by” setting to show only the mutation calls that are found in both samples



Select your NGS sample and matching Sanger sample from Mutation Surveyor for Sample Comparison



Sample Comparison ? X

Show columns: AF Cov RB Zygosity GT

Filter variants by panel: ▶ None selected

Filter variants by pathogenicity: ▶ None selected

Shared by = ▾ 2 ▾ samples

AF difference ≤ ▾ 1.00 ▾ Exclude missing variants

AF between 0.00 ▾ and 1.00 ▾

Id	Name
18	264381_MSv5.0.1_BRCA1_2Samples_MS_CR_ForGA
17	264381_BRCA1_Output_Mutation_Report1