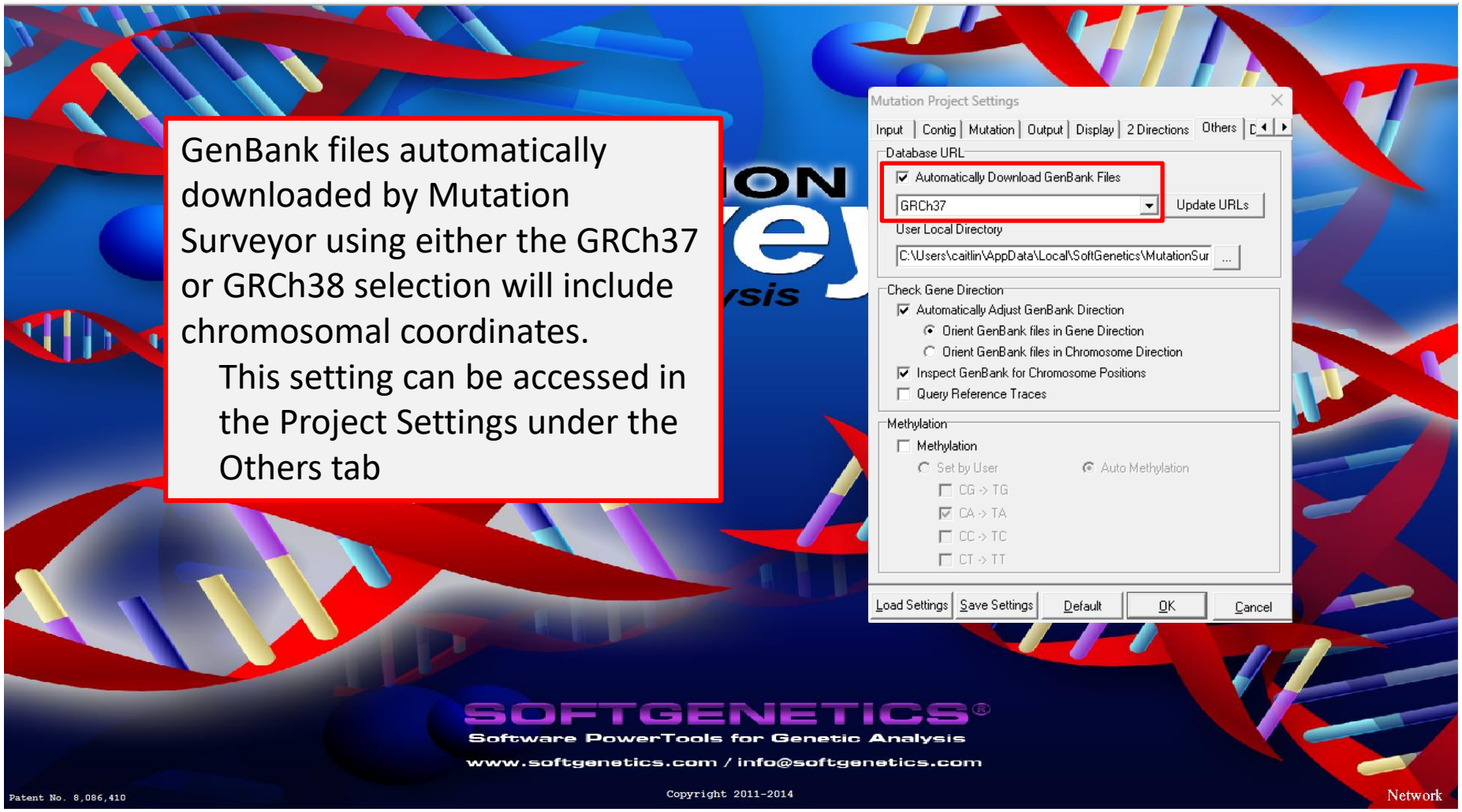
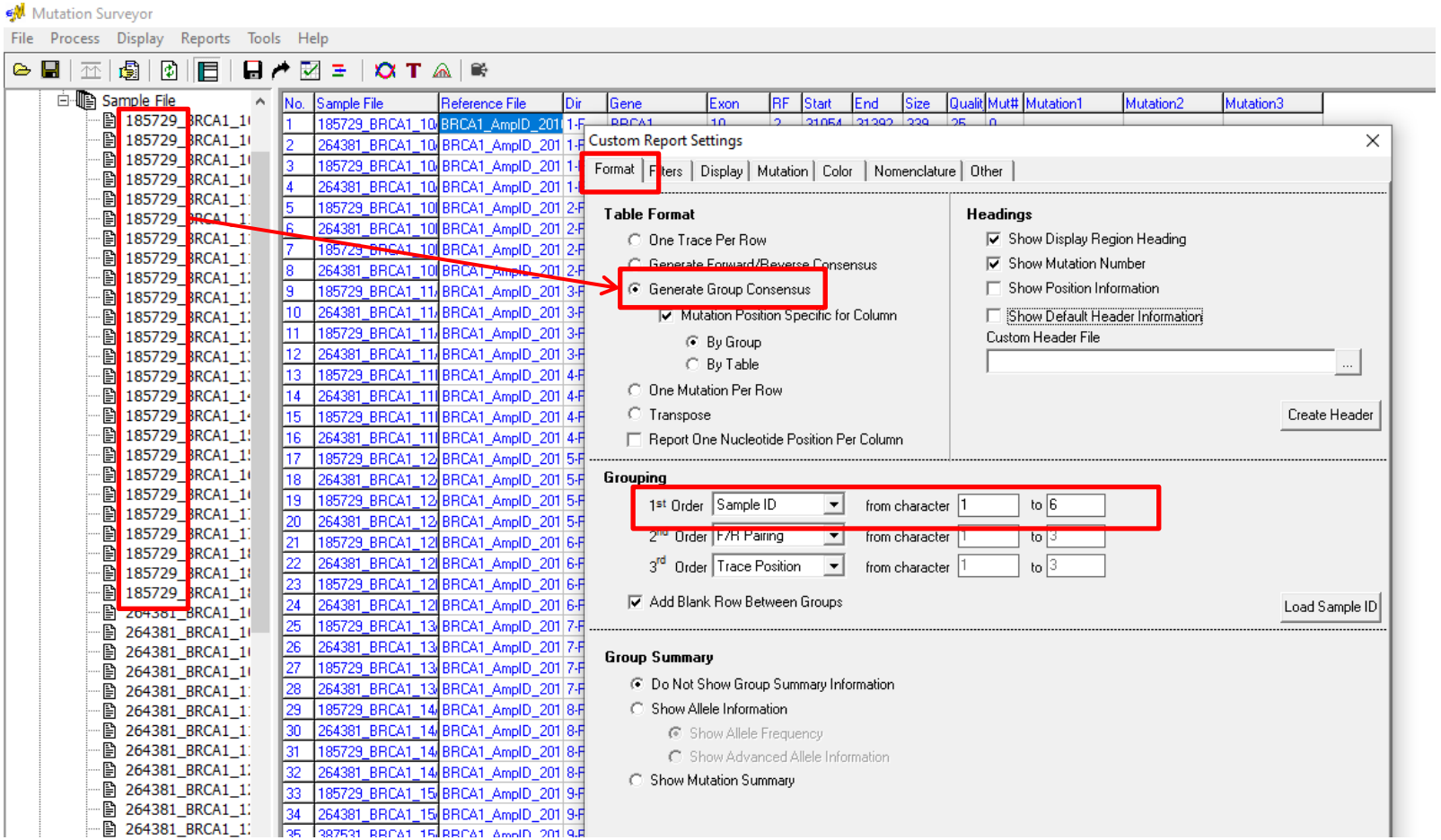
Importing Mutation Surveyor Data into Geneticist Assistant

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

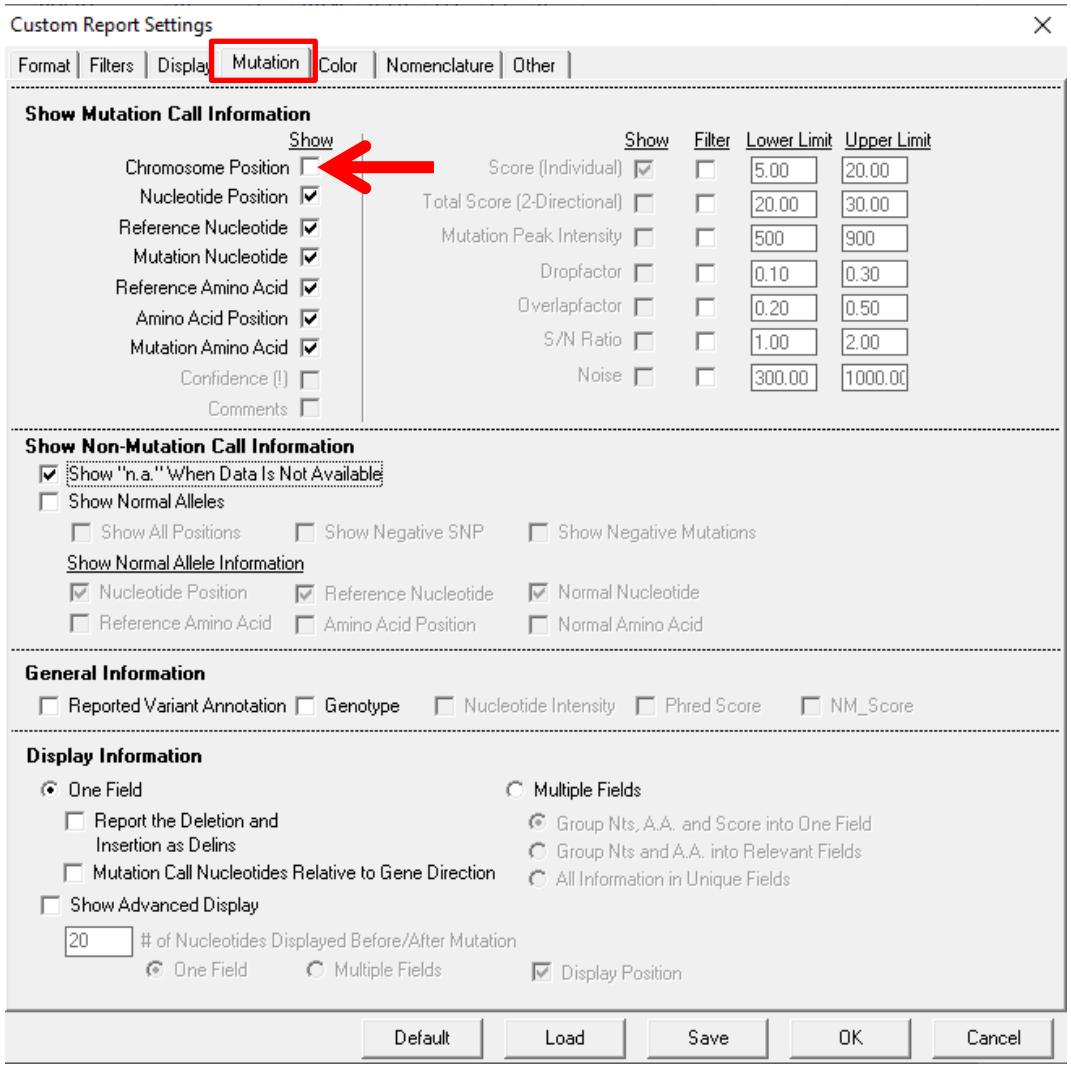


Create a Custom Report of the Analysis (Reports -> Custom Report)



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.

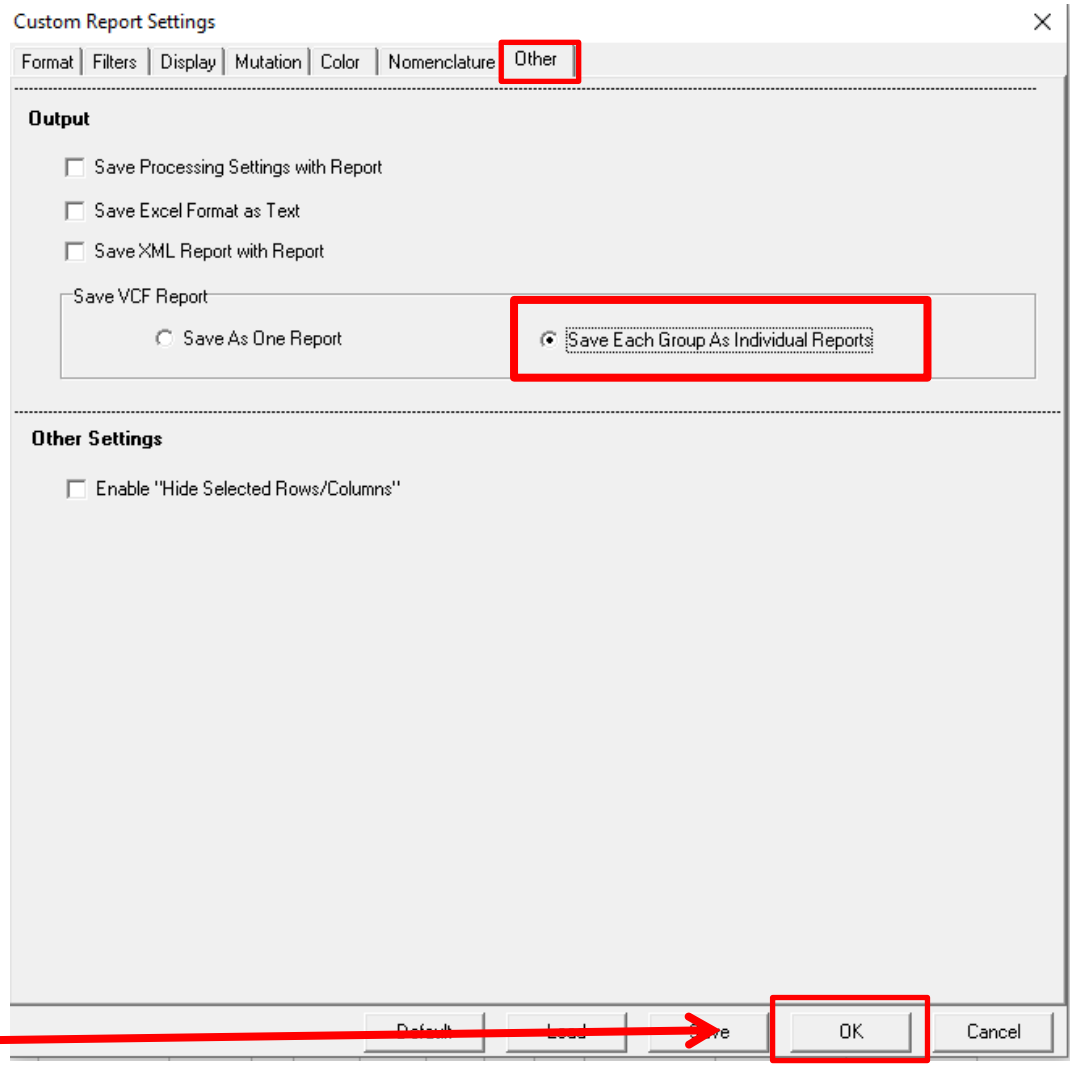
Create a Custom Report of the Analysis



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.



Click OK when done

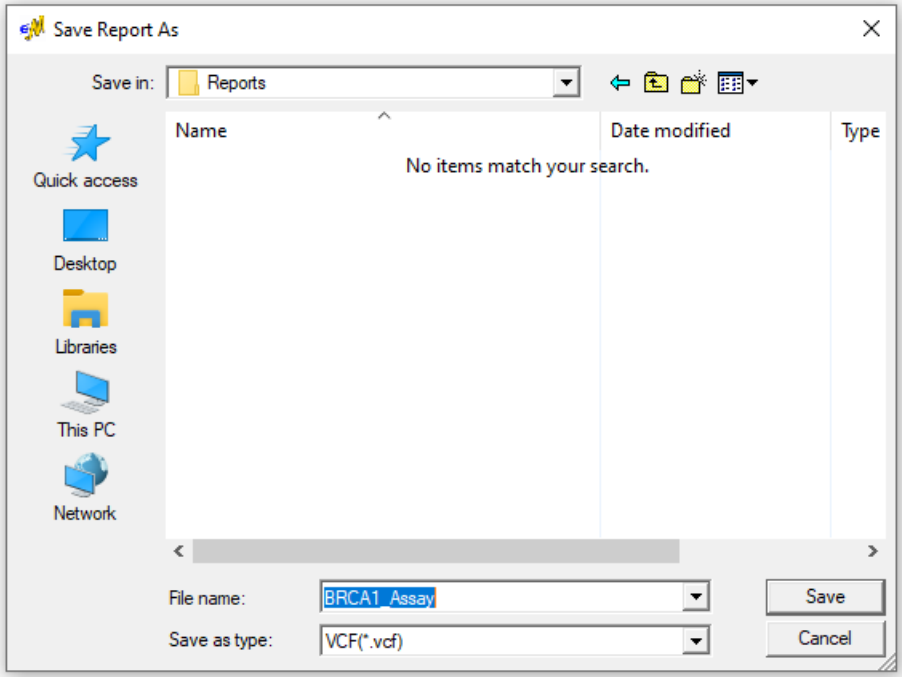
Create a Custom Report of the Analysis

Custom Report

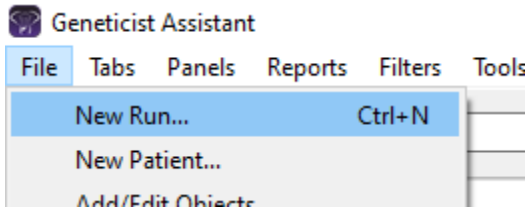
Identifier	Gene	RNA Accessi	Protein Acce	Exon	Read Size	Variant1	Variant2	Variant3	Variant4	Variant5	Variant6	Variant7	Variant8	Variant9	Variant10
185729	BRCA1	NM_007300.	NP_009231.	10	339	c.1067A>AG,p.Q356QR,dbSNI	c.1486C>CT,	c.1687C>CT,	c.2077G>GA	c.2082C>T,p	c.2311T>C,p	c.2612C>CT,	c.2866T>C,p	c.3548A>AG,	c.3607C>CT,
Identifier	Gene	RNA Accessi	Protein Acce	Exon	Read Size	Variant1	Variant2	Variant3	Variant4	Variant5	Variant6	Variant7	Variant8	Variant9	Variant10
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,
Identifier	Gene	RNA Accessi	Protein Acce	Exon	Read Size	Variant1	Variant2								
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.



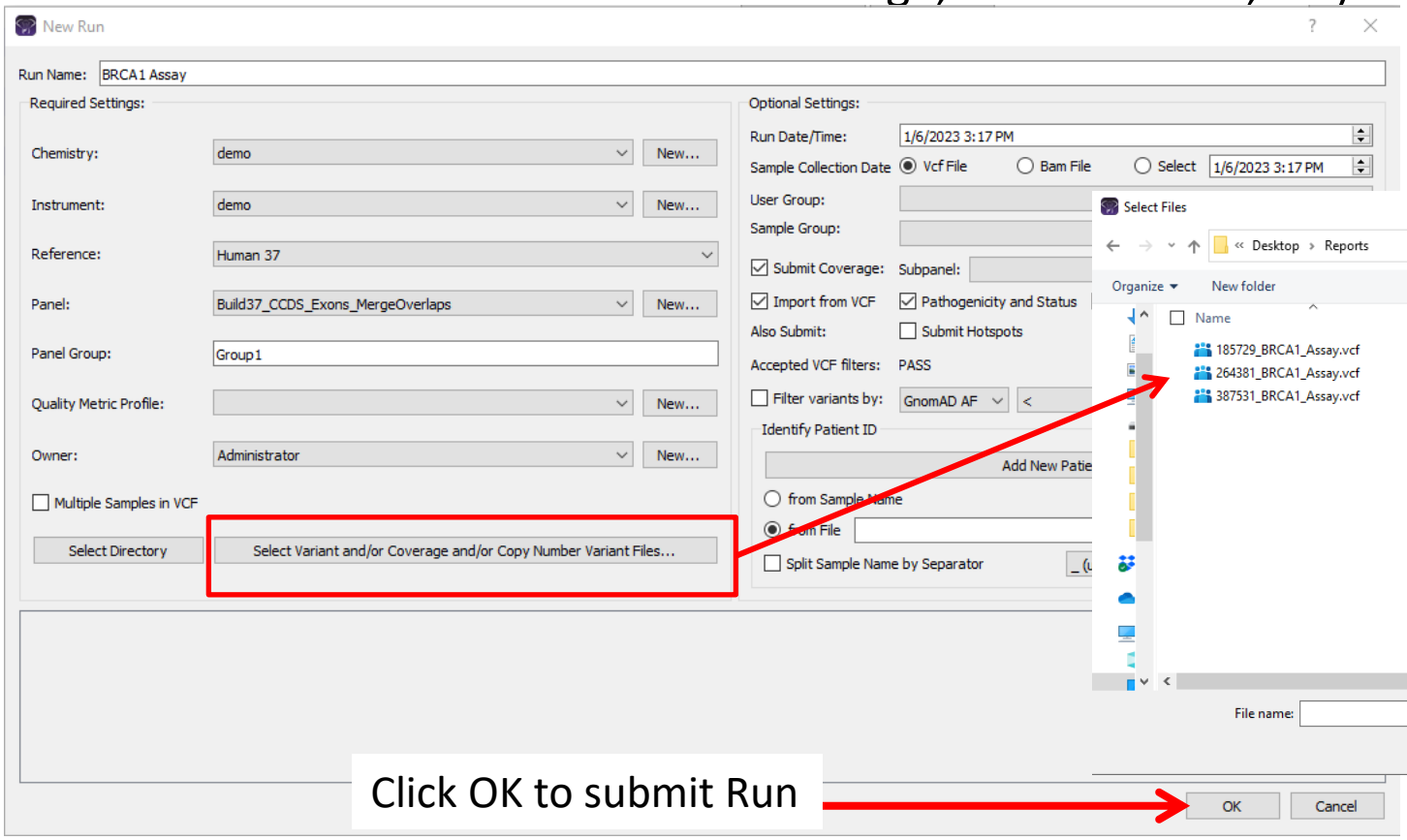
Prepare a New Run in Geneticist Assistant



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

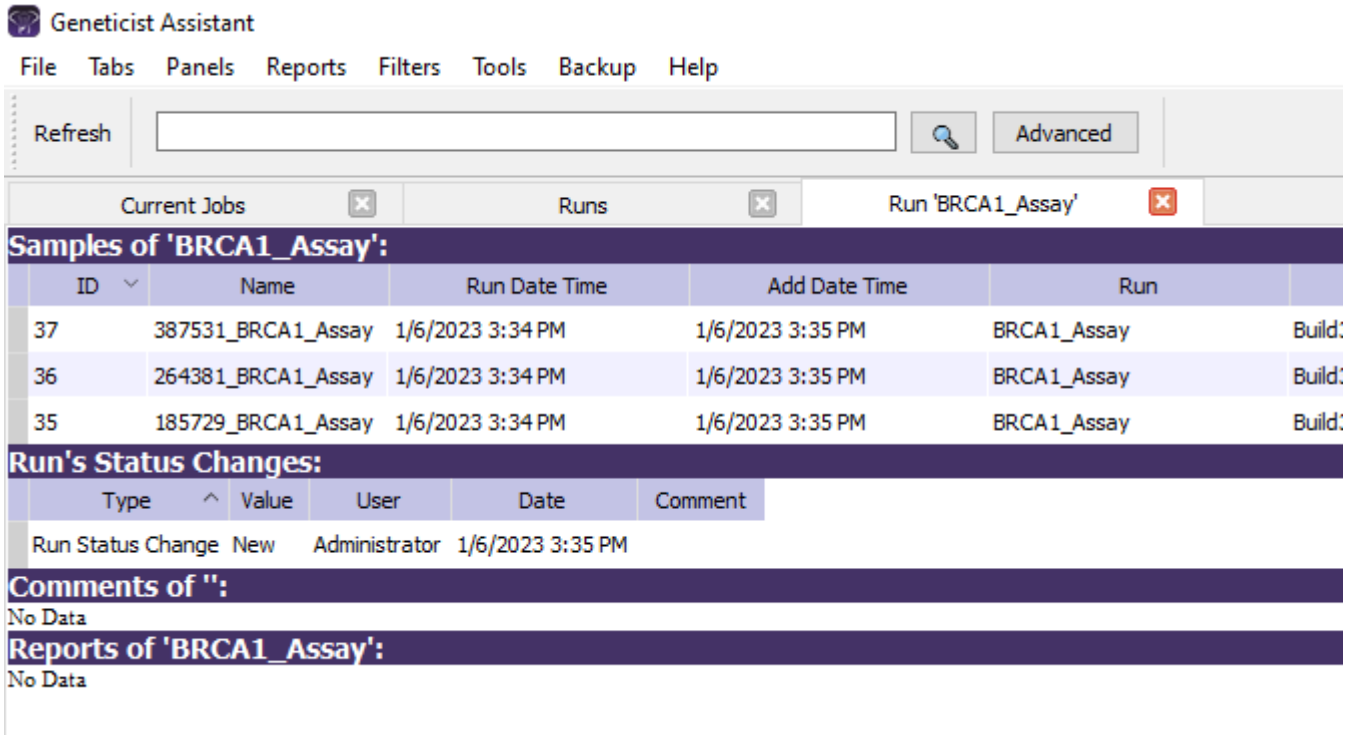
Both Sanger and NGS data can be submitted in same run.

Change other preferences as desired (Sample name, Submit Coverage, Patient Name, etc).



Click OK to submit Run

Review Submitted Data in Geneticist Assistant



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