

Technical Product Report

For Research Use Only; Not for use in Diagnostic Procedures

Product Description:	SeraSeq gDNA BRCA1/2 LGR Inherited Mutation Mix		
Material Number:	0730-0568	Batch Number:	10652943
Material Description:	A genomic DNA mutation mix containing deletions, insertions, single nucleotide variants, and large genomic rearrangements (LGRs) in BRCA1 and BRCA2 genes blended with GM24385 DNA as reference wild-type background		
Fill Volume:	25 μ L		
Date of Manufacture:	23 JAN 2023	Expiration Date:	23 JAN 2025
Concentration Test Method:	Invitrogen™ Qubit™ dsDNA BR Assay		
Nominal Concentration:	15ng/ μ L		
Concentration:	21.9 ng/ μ L		
dPCR Test Method:	Allele-specific digital PCR assays on Bio-Rad QX200 QX200 Droplet Digital™ System		
NGS Assay Test Method:	153 ng of DNA was used to prepare NGS libraries using the Agilent SureSelect ^{XT} Low Input Target Enrichment System with Dual Indexing protocol and enriching with SureSelect Human All Exon v8 exome probes. Variant Allele Frequencies (VAFs) were calculated from mutant and reference reads generated from Whole Exome Sequencing analysis.		

Gene	Nucleotide change	Protein change	% Allele Frequency (dPCR)	% Allele Frequency (NGS)
BRCA1	c.4487_4675+2del	Splice variant	51.5	ND
BRCA1	c.4186_4357dup	p.R1397Yfs*2	49.5	45.7
BRCA1	c.2071_2171del	p.R691*	50.4	40.7
BRCA1	c.4987_5074del	p.V1665Sfs*8	49.9	ND
BRCA1	c.5279_5332del	p.I1760_D1778delinsN	50.8	13.2
BRCA1	c.5209_5248delinsTC	p.R1737Sfs*80	46.1	23.4
BRCA1	c.2820_2830delinsAAGATAAGCCAGTTTGATAA	p.D940_C944delinsER*	50.4	45.9
BRCA1	c.1961del	p.K654Sfs*47	50.4	37.6
BRCA1	c.4327C>T	p.R1443*	49.5	39.0
BRCA1	c.441+2T>G	Splice variant	48.9	47.0
BRCA2	c.8757-2_9023del	Splice variant	50.3	25.0
BRCA2	c.2886_3144del	p.H962Qfs*6	51.3	46.8
BRCA2	c.68_316del	p.D23_L105del	47.8	ND
BRCA2	c.5150_5226delinsTACTTAATACTTATTAAGTATTA	p.E1717_N1742delinsVLNTY*	51.3	42.4
BRCA2	c.891_899delinsGATACTTCAG	p.T298Ifs*7	48.5	39.3
BRCA2	c.5436del	p.E1812Dfs*3	51.3	47.3
BRCA2	c.8167G>C	p.D2723H	52.9	51.6
BRCA2	c.8331+2T>A	Splice variant	52.9	51.5
BRCA2	c.910G>T	p.E304*	48.5	41.0
BRCA2	c.2407dup	p.Y803Lfs*2	51.3	52.7

ND – Not detected

Approval:



08 FEB 2023

Prepared By

Date