Seraseq® BRCA1/2 LGRs Reference Materials

For Research Use Only. Not for use in diagnostic procedures.

This product contains purified fragmented genomic DNA from GM24385 cells and DNA plasmids of BRCA variants.

This file lists the genomic coordinates for the DNA sequences included in the material numbers 0730-0564, 0730-0567 and 0730-0568.

Gene	Nucleotide change	Protein change	Transcript	GRCh37	GRCh38	dbSNP ID	Type of nucleotide alteration	Variant size (bp)	Exon Coverage
BRCA1	c.4487_4675+2del	Splice variant	NM_007294.4	Chr17:41226346_41226536	Chr17:43074329_43074519	rs1555581778	Deletion	191	Whole exon 12
BRCA1	c.4186_4357dup	p.R1397Yfs*2	NM_007294.4	Chr17:41234421_41234592	Chr17:43082404_43082575		Insertion	172	Whole exon 14
BRCA1	c.2071_2171del	p.R691*	NM_007294.4	Chr17:41245377_41245477	Chr17:43093360_43093460	rs1555590294	Deletion	101	Part of exon 10
BRCA1	c.4987_5074del	p.V1665Sfs*8	NM_007294.4	Chr17:41219625_41219712	Chr17:43067608_43067695		Deletion	88	Whole exon 16
BRCA1	c.5279_5332del	p.I1760_D1778delinsN	NM_007294.4	Chr17:41203080_41203133	Chr17:43051063_43051116	rs1555575677	Deletion	54	Part of exon 19
BRCA1	c.5209_5248delinsTC	p.R1737Sfs*80	NM_007294.4	Chr17:41209098_41209137	Chr17:43057081_43057120	rs273901753	Indel	40	
BRCA1	c.2820_2830delinsAAGATAAGCCAGTTTGATAA	p.D940_C944delinsER*	NM_007294.4	Chr17:41244718_41244728	Chr17:43092701_43092711	rs1555588883	Indel	11	
BRCA1	c.1961del	p.K654Sfs*47	NM_007294.4	Chr17:41245587	Chr17:43093570	rs80357522	Deletion	1	
BRCA1	c.4327C>T	p.R1443*	NM_007294.4	Chr17:41234451	Chr17:43082434	rs41293455	SNV	1	
BRCA1	c.441+2T>G	Splice variant	NM_007294.4	Chr17:41256137	Chr17:43104120	rs397509173	SNV	1	
BRCA2	c.2407dup	p.Y803Lfs*2	NM_000059.4	Chr13:32910899	Chr13:32336762	rs2072457050	Insertion	1	
BRCA2	c.2886_3144del	p.H962Qfs*6	NM_000059.4	Chr13:32911378_32911636	Chr13:32337241_32337499	rs2072467220	Deletion	259	Part of exon 11
BRCA2	c.8757-2_9023del	Splice variant	NM_000059.4	Chr13:32953452_32953956	Chr13:32379315_32379819		Deletion	505	Whole exon 22 and part of exon 23
BRCA2	c.68_316del	p.D23_L105del	NM_000059.4	Chr13:32893214_32893462	Chr13:32319077_32319322		Deletion	249	Whole exon 3
BRCA2	c.5150_5226delinsTACTTAATACTTATTAAGTATTA	p.E1717_N1742delinsVLNTY*	NM_000059.4	Chr13:32913642_32913718	Chr13:32339505_32339581		Indel	77	
BRCA2	c.891_899delinsGATACTTCAG	p.T298Ifs*7	NM_000059.4	Chr13:32906506_32906514	Chr13:32332369_32332377	rs276174914	Indel	9	
BRCA2	c.5436del	p.E1812Dfs*3	NM_000059.4	Chr13:32913927	Chr13:32339790	rs397507351	Deletion	1	
BRCA2	c.8167G>C	p.D2723H	NM_000059.4	Chr13:32937506	Chr13:32363369	rs41293511	SNV	1	
BRCA2	c.8331+2T>A	Splice variant	NM_000059.4	Chr13:32937672	Chr13:32363535	rs398122602	SNV	1	
BRCA2	c.910G>T	p.E304*	NM_000059.4	Chr13:32906525	Chr13:32332388	rs1593891461	SNV	1	

NOTE: Above list does not include variants present in the GM24385 background.

Substitution refers to single nucleotide variant; Indels are defined as deletion/insertions less than 10 base pairs, and LGRs (deletions or insertions) are larger than 10 base pairs.

The annotations of nucleotide and protein changes refer to transcripts NM_007294.4 (BRCA1) and NM_000059.4 (BRCA2).

* [#] indicates total length of entry, e.g., [24] indicates total length of 24 bases.