



Next-generation sequencing (NGS) is performed to test for the presence of single nucleotide variations, deletions, and insertions in coding regions and intron/exon boundaries of the genes listed. When appropriate, alterations detected are confirmed by an independent reference method, such as Sanger sequencing. Default reportable range offset is +/-2 base pairs around each targeted exon region.

Genomic Build: GRCh37 (hg19) unless otherwise specified

As a result of technical limitations of the assay (including regions of homology, high GC content, and repetitive sequences), there are regions of some genes that cannot be effectively evaluated. Refer to gene regions table below for complete gene coverage information. To verify if a specific region/exon/variant is covered by this assay, contact the laboratory at 800-533-1710.

Gene	Exon	Chromosome	Genomic Start	Genomic Stop	Reference Transcript
<i>BRCA1</i>	Ex2	chr17	41276032	41276115	NM_007294
<i>BRCA1</i>	Ex3	chr17	41267741	41267798	NM_007294
<i>BRCA1</i>	Ex4	chr17	41258471	41258552	NM_007294
<i>BRCA1</i>	Ex5	chr17	41256883	41256975	NM_007294
<i>BRCA1</i>	Ex6	chr17	41256137	41256280	NM_007294
<i>BRCA1</i>	Ex7	chr17	41251790	41251899	NM_007294
<i>BRCA1</i>	Ex8	chr17	41249259	41249308	NM_007294
<i>BRCA1</i>	Ex9	chr17	41247861	41247941	NM_007294
<i>BRCA1</i>	Ex10	chr17	41243450	41246879	NM_007294
<i>BRCA1</i>	Ex11	chr17	41242959	41243051	NM_007294
<i>BRCA1</i>	Ex12	chr17	41234419	41234594	NM_007294
<i>BRCA1</i>	Ex13	chr17	41228503	41228633	NM_007294
<i>BRCA1</i>	Ex14	chr17	41226346	41226540	NM_007294
<i>BRCA1</i>	Ex15	chr17	41222943	41223257	NM_007294
<i>BRCA1</i>	Ex16	chr17	41219623	41219714	NM_007294
<i>BRCA1</i>	Ex17	chr17	41215889	41215970	NM_007294
<i>BRCA1</i>	Ex18	chr17	41215348	41215392	NM_007294
<i>BRCA1</i>	Ex19	chr17	41209067	41209154	NM_007294
<i>BRCA1</i>	Ex20	chr17	41203078	41203136	NM_007294
<i>BRCA1</i>	Ex21	chr17	41201136	41201213	NM_007294
<i>BRCA1</i>	Ex22	chr17	41199658	41199722	NM_007294
<i>BRCA1</i>	Ex23	chr17	41197693	41197821	NM_007294
<i>BRCA2</i>	Ex2	chr13	32890596	32890666	NM_000059
<i>BRCA2</i>	Ex3	chr13	32893212	32893464	NM_000059
<i>BRCA2</i>	Ex4	chr13	32899211	32899323	NM_000059
<i>BRCA2</i>	Ex5	chr13	32900236	32900289	NM_000059
<i>BRCA2</i>	Ex6	chr13	32900377	32900421	NM_000059
<i>BRCA2</i>	Ex7	chr13	32900634	32900752	NM_000059
<i>BRCA2</i>	Ex8	chr13	32903578	32903631	NM_000059
<i>BRCA2</i>	Ex9	chr13	32905054	32905169	NM_000059

Targeted Genes and Methodology Details
for BRCA1/2 Mutation Analysis (continued)

Gene	Exon	Chromosome	Genomic Start	Genomic Stop	Reference Transcript
<i>BRCA2</i>	Ex10	chr13	32906407	32907526	NM_000059
<i>BRCA2</i>	Ex11	chr13	32910400	32915335	NM_000059
<i>BRCA2</i>	Ex12	chr13	32918693	32918792	NM_000059
<i>BRCA2</i>	Ex13	chr13	32920962	32921035	NM_000059
<i>BRCA2</i>	Ex14	chr13	32928996	32929427	NM_000059
<i>BRCA2</i>	Ex15	chr13	32930563	32930748	NM_000059
<i>BRCA2</i>	Ex16	chr13	32931877	32932068	NM_000059
<i>BRCA2</i>	Ex17	chr13	32936658	32936832	NM_000059
<i>BRCA2</i>	Ex18	chr13	32937314	32937672	NM_000059
<i>BRCA2</i>	Ex19	chr13	32944537	32944696	NM_000059
<i>BRCA2</i>	Ex20	chr13	32945091	32945239	NM_000059
<i>BRCA2</i>	Ex21	chr13	32950805	32950930	NM_000059
<i>BRCA2</i>	Ex22	chr13	32953452	32953654	NM_000059
<i>BRCA2</i>	Ex23	chr13	32953885	32954052	NM_000059
<i>BRCA2</i>	Ex24	chr13	32954142	32954284	NM_000059
<i>BRCA2</i>	Ex25	chr13	32968824	32969072	NM_000059
<i>BRCA2</i>	Ex26	chr13	32971033	32971183	NM_000059
<i>BRCA2</i>	Ex27	chr13	32972297	32972909	NM_000059