



Next-generation sequencing 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.

As a result of technical limitations of the assay (including regions of homology, high guanine-cytosine [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.

Genomic Build: GRCh37 (hg19) unless otherwise specified

| 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 |

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

| Gene | Exon | Chromosome | Genomic Start | Genomic Stop | Reference Transcript |
|--------------|------|------------|---------------|--------------|----------------------|
| <i>BRCA2</i> | Ex9 | chr13 | 32905054 | 32905169 | NM_000059 |
| <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 |