BRCA1 and BRCA2 Full Gene Sequence Analysis and Full Deletion/Duplication Variants

This is a 2nd tier test for uncommon duplications and deletions in BRCA1. Preferred initial test is the sequencing and common deletion/duplication test.

Testing includes full gene sequencing of exons and 25 base pairs of introns of BRCA 1 and 2 and exon level deletion and duplications.

**Indication:** This testing is indicated for individuals that meet the criteria for Hereditary Breast and Ovarian Cancer listed in the National Comprehensive Cancer Network (NCCN) guidelines. For assistance with interpreting guidelines, please contact us or refer to genetic counseling for evaluation. Testing on minors is not indicated.

BRCA1 and BRCA2 are genes that code for proteins that help repair DNA damage. Inherited mutations in BRCA 1 or 2, in combination with additional acquired mutations, can result in increased risk for developing cancer. Specific mutations in BRCA1 and BRCA2 increase the risk of breast and ovarian cancers, and have been associated with increased risk of several additional types of cancer. BRCA1 and BRCA2 mutations account for about 20 to 25 percent of hereditary breast cancers and about 5 to 10 percent of all breast cancers. In addition, BRCA1 and BRCA2 account for about 15 percent of ovarian cancers overall. Breast and ovarian cancers associated with BRCA1 and BRCA2 mutations tend to develop at younger ages than their nonhereditary counterparts.

**Testing method:** Next Generation Sequencing (NGS) for whole gene analysis of single base pair (point) mutations and small (<25bp) deletions and duplications. The TruSight Cancer Sequencing Panel targets 94 genes suspected to play a role in predisposing to cancer, including genes associated with both common (eg, breast, colorectal) and rare cancers. In addition, the panel includes 284 SNPs suspected to be associated with cancer through genome-wide association studies (GWAS). Content selection was based on expert curation of the scientific literature and other high-quality resources. The assay provides coverage of exonic and noncoding DNA in exon-flanking regions, on average 50 bp.

Multiplex ligation-dependent probe amplification (MLPA) PCR assay is used to evaluate copy number variations.

**Turnaround time:** 7-10 business days

**Sample Requirements:** 3 ml peripheral blood in EDTA (lavender) top tube

Specimen stability: Ambient - 72 hours; Refrigerated - 1 week

**CPT codes:** 81162 (complete) or 81213 if performed as reflex to 81211 (BRCA ½ full sequence with common deletions and duplications)

**References:**