Deletion duplication analysis of BRCA1 and BRCA2 (BReast CAncer genes 1 and 2) by MLPA

**Description:** Exon level deletion duplication analysis is analyzed by Multiplex Ligation-dependent Probe Amplification. This test is an adjunct test for sequencing assays that do not include complete deletion duplication testing and should be considered if ordering the Hereditary Breast/Ovarian Cancer-related Gene Sequence Analysis.

**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 and either have already tested negative for BRCA1 and BRCA2 mutations by sequence analysis or have been referred for the Hereditary Breast/Ovarian Cancer-related Gene Sequence Analysis.

Indications include
- A known mutation in a family member
- Personal or family history of breast cancer with onset ≤45 years
- Personal or family history of ovarian cancer or male breast cancer at any age
- Ashkenazi Jewish heritage with breast, ovarian or pancreatic cancer at any age

Please refer to the NCCN Clinical Practice Guidelines in Oncology for Genetic/Familial High-Risk Assessment: Breast and Ovarian for a complete list of indications (https://www.nccn.org/professionals/physician_gls/PDF/genetics_screening.pdf) For assistance with interpreting guidelines, please contact Henry Ford Precision Genomic Diagnostics, or refer to genetic counseling for evaluation. Testing on minors is not indicated.

**Protein Description:** BRCA1 and BRCA2 are genes that code for proteins that help repair DNA damage. Inherited mutations in BRCA1 or BRCA2, 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:** Multiplex ligation-dependent probe amplification (MLPA) PCR assay to evaluate copy number variations in the exons of BCRA1 and BRCA2.

**Turnaround time:** 5-10 business days

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

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

**CPT codes:** 81406
References:


