**Alternate Name(s):**

Hereditary Cancer - Breast/Ovarian

**Specimen:**

Whole blood - 4 mL Lavender EDTA top Vacutainer tube

**Collection Information:**

Sample may be transported at room temperature. Alternative tissues may be accepted after consult with laboratory.

**Reference Ranges:**

See report

**Interpretive Comments:**

A subset (5-10%) of breast/ovarian cancers are familial, and a predisposition to develop malignancy in these tissues has been found to segregate with (autosomal dominant) mutations in either the BRCA1 gene (Chr.17) or the BRCA2 gene (Chr.13). Mutations in both BRCA1 and BRCA2 are associated with a markedly elevated lifetime risk of breast cancer (BRCA1: 65% or greater, BRCA2: 45% or greater) as well as an increased lifetime risk of ovarian cancer (BRCA1: 39% or greater, BRCA2: 11% or greater); PMID:12677558.

**Comments:**

For more information click on: [MOLECULAR DIAGNOSTIC LABORATORY](#)
BREAST CANCER (BRCA1 AND BRCA2 SCREENING)

Sanger sequencing and MLPA copy number analysis. All variants interpreted as either ACMG category 1, 2, or 3 (pathogenic, likely pathogenic, VUS; PMID: 25741868) are confirmed using Sanger sequencing, MLPA, or other assays. ACMG category 4 and 5 variants (likely benign, benign) are not reported, but are available upon request.

Critical Information Required:

Pedigree required.

Storage and Shipment:

Must be received in testing laboratory within 48 hours of collection, shipped at room temperature by courier/overnight delivery.

Test Schedule:

As required,
Monday to Friday 0800-1600 hours