BREAST CANCER (BRCA1 AND BRCA2 SCREENING)

London Health Sciences Centre | St. Joseph's Health Care London

Pathology and Laboratory Medicine

Orderable – E-order/Requisition

Turnaround Time: 4-6 weeks **STAT: 4 weeks**

Laboratory:
Molecular Diagnostics Lab

Requisition:
MOLECULAR DIAGNOSTIC REQUISITION

Method of Analysis:
All coding exons and 20 bp of flanking intronic sequence are enriched using an LHSC customtargeted hybridization protocol (Roche Nimblegen), followed by high throughput sequencing (Illumina). Sequence variants and copy number changes are assessed and interpreted using clinically validated algorithms and commercial software (SoftGenetics: Nextgene, Geneticist Assistant, Mutation Surveyor; and Alamut Visual). All exons have >500x mean read depth coverage, with a minimum 200x coverage at a single nucleotide resolution. This assay meets the sensitivity and specificity of combined

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)

London Health Sciences Centre
St JOSEPH'S HEALTH CARE LONDON
Pathology and Laboratory Medicine

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.

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.

Test Schedule:
As required,
Monday to Friday 0800-1600 hours