BRCA1/BRCA2 Full Sequencing and Full Deletions/Duplications

This hereditary panel detects mutations in genes BRCA1 and BRCA2 from DNA isolated from a blood specimen. Inherited mutations in BRCA1 or BRCA2 are associated with autosomal dominant hereditary breast and ovarian cancer (HBOC) syndrome.

Testing Method and Background

This test utilizes **Next Generation Sequencing (NGS) technology** which provides coverage of all coding exons and noncoding DNA in exonflanking regions (on average 50 bp) enriched using hybrid capture Illumina TruSight Cancer Sequencing Panel. Single base pair (point) mutations, small insertions/deletions (1-25 bp), complex insertions and deletions, or larger deletions and duplication. All reportable copy number variants are confirmed by independent methodology using gene-specific Multiplex Ligation-dependent Probe Amplification (MLPA) assay.

Mutations in BRCA1 and BRCA2 are responsible for approximately 20 to 25% of hereditary breast cancers and about 5 to 10% of all breast cancers. In addition, mutations in BRCA1 and BRCA2 cause around 15% of ovarian cancers. Specific cancer risks are different between BRCA1 and BRCA2. BRCA1 pathogenic mutations are associated with increased lifetime risk for breast cancer (40-87%), ovarian cancer (16-54%), male breast cancer (1-2%), prostate cancer (up to 20%), and pancreatic cancer (1-3%). BRCA2 pathogenic mutations are associated with increased lifetime risk for breast cancer (up to 84%), ovarian cancer (up to 27%), male breast cancer (6%), prostate cancer (up to 20%), pancreatic cancer (2-7%), and melanoma.

Highlights of BRCA1&BRCA2 Full Sequencing and Full Deletions/Duplications

Targeted Region

BRCA1, BRAC2

- **Wide-ranging Coverage of Variants**
  Detects and provides coverage of all coding exons and noncoding DNA in exon flanking regions.
- **Accurate Results Using Clinically Validated Computational Data Analysis**
  A variety of mutation types (point, indels and duplications) are confirmed using computational data analysis for sequence variant calling, filtering and annotation.

Ordering Information

Get started (non-HFHS): Print a Hereditary Cancer Panels requisition form online at www.HenryFord.com/HFCPD

Get started (HFHS): Order through Epic using test "BRCA1/2 Full Sequencing and Full Deletions/Duplications" (DNA210008)

Specimen requirements:

- Peripheral Blood - 1-3ml in lavender top tube (EDTA) **Specimen stability: Ambient - 72 hours; Refrigerated - 1 week**
- Saliva specimen - Oragene self-collection kit
- Extracted DNA - from a CLIA-certified Laboratory

Cause for Rejection: Clotted, hemolyzed, or frozen specimens, improper anticoagulant, tubes not labeled with dual patient identification, non-dedicated tubes.

TAT: 5-10 business days (after Prior Authorization obtained)

Mail test material to:
Henry Ford Center for Precision Diagnostics
Pathology and Laboratory Medicine
Clinic Building, K6, Core Lab, E-655
2799 W. Grand Blvd., Detroit, MI 48202

CPT Codes: 81162, G0452

Contact us: Client Services, Account and Billing Set-up, and connect with a Molecular Pathologist at (313) 916-4DNA (4362)

For more information on Comprehensive Molecular Services, visit our website
www.HenryFord.com/HFCPD

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