BRCA1 and BRCA2 Ashkenazi Jewish 3-Mutation Panel

This hereditary panel detects mutations associated in certain ethnic groups 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.

This panel analyzes BReast CAncer genes 1 and 2 (BRCA1 and BRCA2), that code for proteins that help repair DNA damage. Inherited mutations in BRCA1 or BRCA2 are associated with autosomal dominant hereditary breast and ovarian cancer (HBOC) syndrome (OMIM ID: 604370) which is characterized by increased lifetime risk for developing breast, ovarian and other types of cancer. Members of certain ethnic groups, such as individuals of Ashkenazi Jewish ancestry, have an increased risk for carrying specific types of pathogenic mutations in BRCA1 or BRCA2, known as founder mutations. Per NCCN guidelines, any woman of Ashkenazi Jewish ancestry who has been diagnosed with breast or ovarian cancer meets criteria for BRCA founder variant testing that includes three known founder mutations: BRCA1 c.68_69delAG, BRCA1 c.5266dupC, and BRCA2 c.5946delT.

Highlights of BRCA1 and BRCA2 Ashkenazi Jewish 3-Mutation Panel

Targeted Region

Founder Mutations: BRCA1 c.68_69delAG, BRCA1 c.5266dupC, and BRCA2 c.5946delT

- **Wide-ranging Coverage of Variants**
  Detects and provides coverage of all coding exons and noncoding DNA in exonflanking 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 "BRCA Ashkenazi Jewish Panel" (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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