HENRY FORD HEALTH
CENTER FOR PRECISION DIAGNOSTICS

BRCA1/BRCA2 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 exon flanking regions (on average 50 bp) enriched using hybrid capture methodology. This assay can detect >99% of described mutations in the included genes, when present, including single nucleotide variants (point mutations), small insertions/deletions (1-25 bp), larger deletions and duplication (<100 bp), complex insertions/deletions, splice site mutations, whole-gene deletions/duplications and exon-level intragenic deletions/insertions in each gene targeted for analysis. All reportable copy number variants are confirmed by independent methodology.

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**
- Extracted DNA - from a CLIA-certified Laboratory

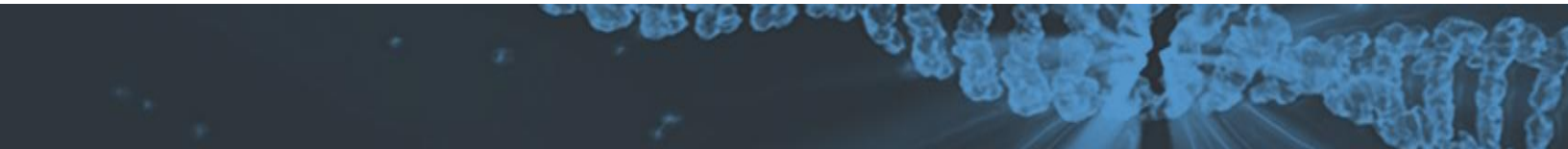
Cause for Rejection: Clotted, hemolyzed, or frozen specimens, improper anticoagulant, tubes not labeled with dual patient identification, non-dedicated tubes.

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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