

HFCI Hereditary Cancer Risk Workup - Breast & Related Cancers

This workup is a comprehensive >100-gene analysis panel that identifies inherited risks for hereditary cancers across several major organ systems including cancers of breast, ovary, uterus, gastrointestinal system (colorectal, gastric, pancreatic), thyroid, kidney, urinary tract, prostate, brain and nervous system, skin, sarcoma and hematologic cancers using genomic DNA. The genes included are in concordance with (but not limited to) NCCN guideline for high-risk assessment for cancers and ACMG variant classification guidelines.

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.

Inherited genetic mutations in *BRCA1* and *BRCA2* account for about 20 to 25% of hereditary breast cancers and about 5 to 10% of all breast cancers. Additionally mutations in *BRCA1* and *BRCA2* cause around 15% of ovarian cancers, and are associated with increased risk of multiple other cancer types including melanoma, pancreatic and prostate cancer in males. Panel also includes genes responsible for rare hereditary cancer syndromes that have been associated with increased lifetime risk for multiple cancer types and are characterized by other clinical features specific for each syndrome, as well as many other genes linked to hereditary cancer predisposition.

Highlights of the HFCI Hereditary Cancer Risk Workup - Breast & Related Cancers

Targeted Region

Genes: *Primary analysis:* *BRCA1 & BRCA2* (Note: this report is issued separately from the expanded secondary analysis)

Secondary analysis: *ABRAXAS1, AIP, ALK, ANKRD26, APC, ATM, AXIN2, BAP1, BARD1, BLM, BMPR1A, BRIP1, BUB1B, CDC73, CDH1, CDK4, CDKN1B, CDKN1C, CDKN2A, CEBPA, CHEK2, CTNNA1, CYLD, DICER1, DIS3L2, EGFR, EGLN1, EPCAM, ETV6, EXT1, EXT2, FAN1, FANCA, FANCC, FANCM, FH, FLCN, GALNT12, GATA2, GPC3, GREM1, HOXB13, HRAS, KIF1B, KIT, LZTR1, MAX, MEN1, MET, MITF, MLH1, MLH3, MRE11, MSH2, MSH3, MSH6, MUTYH, NBN, NF1, NF2, NTHL1, PALB2, PALLD, PDGFRA, PHOX2B, PIK3CA, PMS2, POLD1, POT1, PRKAR1A, PTCH1, PTCH2, PTEN, RAD50, RAD51C, RAD51D, RB1, RECQL4, RET, RNF43, RUNX1, SAMD9, SDHA, SDHAF2, SDHB, SDHC, SDHD, SMAD4, SMARCA4, SMARCB1, SMARCE1, SRP72, STK11, SUFU, TERC, TERT, TMEM127, TP53, TRIP13, TSC1, TSC2, VHL, WRN, WT1, XRCC2*

- **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: Order in Epic using test "HFCI Hereditary Cancer Risk - Breast & Related Cancer Workup (> 100 genes)" (OC210008)

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: 10-14 business days

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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