Hereditary Endometrial Cancer Risk Panel (17 genes)

This Hereditary Endometrial Cancer Risk Panel detects mutations in genes associated with inherited risks for hereditary endometrial (uterine) cancer using DNA isolated from a blood specimen.

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.

Most cases of endometrial cancer are sporadic. However, a small proportion of individuals have hereditary endometrial cancer (approximately 2-5% of cases). The two most common inherited syndromes known to increase a woman's lifetime risk of endometrial cancer are Lynch syndrome caused by inherited mutations in MLH1, MSH2, MSH6, PMS2, or EPCAM and Cowden syndrome caused by inherited mutations in PTEN. This panel also includes genes responsible for other rare hereditary cancer syndromes, such as Li-Fraumeni syndrome (TP53), hereditary breast and ovarian cancer syndrome (BRCA1, BRCA2), hereditary diffuse gastric cancer (CDH1), and Peutz-Jeghers syndrome (STK11). In addition, this panel includes several other genes associated with hereditary predisposition to with breast or ovarian cancer (ATM, BRIP1, CHEK2, NBN, PALB2, RAD51C).

Highlights of Hereditary Endometrial Cancer Risk Panel (17 genes)

Targeted Region

ATM, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, EPCAM, MLH1, MSH2, MSH6, NBN, PALB2, PMS2, PTEN, RAD51C, STK11, TP53

- **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 "Hereditary Endometrial Cancer Risk Panel (17 genes)" (DNA210004)

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: 10-14 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: 81432, 81433, 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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