HENRY FORD HEALTH
CENTER FOR PRECISION DIAGNOSTICS

# Hereditary Colorectal/HNPCC Cancer Risk Panel (21 genes)

This panel is a comprehensive screen of 21 genes linked to increased risk of inherited forms of colorectal 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.

Genetic susceptibility to polyposis or non-polyposis colorectal cancer can be caused by several inherited genetic syndromes. Lynch syndrome accounts for approximately 2-5% of colorectal cancers and is caused by germline mutations in mismatch repair genes: MLH1, MSH2, MSH6, PMS2, or EPCAM. This panel also includes genes associated with moderately increased risk for hereditary colorectal cancer (ATM, CHEK2) as well as genes responsible for rare hereditary cancer syndromes, such as Li-Fraumeni (TP53), Peutz-Jeghers (STK11), Cowden (PTEN), and hereditary diffuse gastric cancer (CDH1) syndrome. These syndromes are associated with increased lifetime risk for multiple cancer types including colorectal cancer and are also characterized by other clinical features specific for each syndrome. Identifying individuals with genetic predisposition to colorectal cancer can allow earlier detection of cancer through increased frequency and younger age of initiating colonoscopy and other cancer screening; consideration of prophylactic colectomy or other risk-reducing measures; availability of targeted therapy options for cancer treatment (e.g., Pembrolizumab for mismatch repair deficient and microsatellite instability (MSI)-high tumors in individuals with Lynch syndrome); and identification of at-risk family members.

## Highlights of Hereditary Colorectal/HNPCC Cancer Risk Panel (21 genes)

### Targeted Region

***APC, ATM, AXIN2, BMPR1A, CDH1, CHEK2, EPCAM, GREM1, MLH1, MSH2, MLH3, MSH3, MSH6, MUTYH, NTHL1, PMS2, PTEN, POLD1, SMAD4, 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 Colorectal/HNPCC Cancer Risk Panel" (DNA210006)

**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:** 81435, 81436, 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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