

Hereditary Melanoma Risk Panel (10 genes)

This Expanded Hereditary Melanoma Panel is a comprehensive screen of genes associated with hereditary predisposition to melanoma and other cancers using isolated genomic DNA.

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.

A small proportion of individuals (approximately 10%) have a family history of the disease typically caused by inherited genetic mutation that increase the risk of developing this type of cancer. Several genes have been associated with hereditary melanoma, including CDKN2A and CDK4. Individuals with inherited pathogenic variant in CDKN2A have 28-67% risk for developing melanoma. This panel also includes genes responsible for rare hereditary cancer syndromes with increased risk for melanoma, such hereditary breast and ovarian cancer syndrome (BRCA1, BRCA2), Li-Fraumeni syndrome (TP53), Cowden syndrome (PTEN), BAP1 hereditary cancer predisposition syndrome (BAP1), and hereditary retinoblastoma syndrome (RB1). Identifying individuals with genetic predisposition to melanoma can allow increased frequency and younger age of initiating screening for cancer, close monitoring of dysplastic nevi for signs of malignant transformation, risk-reducing clinical management options and lifestyle changes, and identification of at-risk family members.

Highlights of Hereditary Melanoma Risk Panel (10 genes)

Targeted Region

Genes: *BAP1, BRCA1, BRCA2, CDK4, CDKN2A, MITF, POT1, PTEN, RB1, 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 Melanoma Risk Panel" (DNA210002)

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)

CPT Codes: 81404, G0452

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

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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