Hereditary Cancer Syndrome: Deletion/Duplication Panel

**Test Code:** MD200  
**Turnaround time:** 2 weeks  
**CPT Codes:** 81203 x1, 81211 x1, 81213 x1, 81294 x1, 81300 x1, 81323 x1, 81403 x1, 81404 x1, 81405 x1

### Condition Description

Approximately 5-10% of all cancers are inherited, meaning that pathogenic variants in a single cancer susceptibility gene can predispose an individual to develop cancer and these pathogenic variants can be passed down in families. The risk for developing cancer can vary dramatically from syndrome to syndrome, from about a 55% risk of developing breast cancer in Peutz-Jeghers syndrome to as high as a 100% risk for colon cancer for familial adenomatous polyposis syndrome. Accurate and timely diagnoses are necessary to provide proper medical surveillance and treatment to affected and at-risk individuals.

Many of the inherited cancers syndromes and cancer susceptibility genes are phenotypically heterogeneous, making molecular testing necessary to confirm a clinical diagnosis. The traditional tiered, single gene approach to genetic testing for inherited cancer syndromes can be costly and time consuming. The Hereditary Cancer Syndrome Panel is designed to detect germline pathogenic variants in individuals with a suspected inherited cancer syndrome. This panel includes sequencing analysis for syndromes such as, Lynch syndrome, Cowden syndrome, Multiple Endocrine Neoplasia syndrome, Birt-Hogg Dube syndrome, and Li-Fraumeni syndrome. Additional syndromes are also tested for by this panel.

Reference:

### Genes

ALK, APC, ATM, BAP1, BARD1, BLM, BMPR1A, BRCA1, BRCA2, BRIP1, BUB1B, CDC73, CDH1, CDKN1C, CDKN2A, CHEK2, EPCAM, FH, FLCN, GPC3, MAX, MEN1, MET, MLH1, MRE11, MSH2, MSH6, MUTYH, NBN, NF2, PALB2, PHOX2B, POLD1, PRKAR1A, PTCH1, PTEN, RAD50, RAD51C, RAD51D, RET, SDHAF2, SDHB, SDHC, SDHD, SMAD4, SMARCB1, STK11, SUFU, TMEM127, TPS3, TSC1, TSC2, VHL, WT1, XRCC2

### Indications

The test is indicated for:
- Individuals with a clinical or suspected diagnosis of an inherited cancer syndrome based on personal or family history of cancer.

### Methodology

**Deletion/Duplication Analysis:** DNA isolated from peripheral blood is hybridized to a gene-targeted CGH array to detect deletions and duplications. The targeted CGH array has overlapping probes that cover the entire genomic region. Please note that a “backbone” of probes across the entire genome are included on the array for analytical and quality control purposes. Rarely, off-target copy number variants causative of disease may be identified that may or may not be related to the patient's phenotype. Only known pathogenic off-target copy number variants will be reported. Off-target copy number variants of unknown clinical significance will not be reported.

**Detection**

**Deletion/Duplication:** Detection is limited to duplications and deletions. The CGH array will not detect point or intronic pathogenic variants. Results of molecular analysis must be interpreted in the context of the patient's clinical and/or biochemical phenotype.

### Specimen Requirements

Submit only 1 of the following specimen types

**Type: Whole Blood**

Specimen Requirements:

In EDTA (purple top) tube:
- Infants (2 years): 3-5 ml
- Older Children & Adults: 5-10 ml
Specimen Collection and Shipping: Ship in appropriate tissue transport medium, overnight, at room temperature.

**Type: Isolated DNA**

Specimen Requirements:

In microtainer: 10 ug

Isolation using the Qiagen™ Puregene kit for DNA extraction is recommended.

Specimen Collection and Shipping: Refrigerate until time of shipment in 100 ng/ul of TE buffer. Ship sample at room temperature with overnight delivery.

**Special Instructions**

This test is for germline mutation analysis. DNA isolated from FFPE tumor samples is not suitable for this test.

**Related Tests**

- Hereditary Cancer Syndrome: Sequencing Panel