Hereditary Cancer Syndrome: Deletion/Duplication Panel

**Test Code:** MD200  
**Turnaround time:** 2 weeks  
**CPT Codes:** 81297 x1, 81300 x1, 81323 x1, 81403 x1, 81404 x1, 81205 x1, 81203 x1, 81164 x1, 81294 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, BRI1, BUB1B, CDC73, CDH1, CDKN1G, CDKN2A, CHEK2, EPCAM, FH, FLCN, GPC3, MAX, MEND1, MLH1, MRE11, MSH2, MSH6, MUTYH, NBN, NF2, PALB2, PHOX2B, POLD1, PRKAR1A, PTCH1, PTEN, RAD50, RAD51C, RAD51D, RET, SDHAF2, SDH4, SDH6, SDHD, SMAD4, SMARC1, STK11, SUFU, TMEM127, TP53, 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.

**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: DNA, Isolated

**Specimen Requirements:**

- Microtainer
- 3µg

Isolation using the Perkin Elmer™Chemagen™ Chemagen™ Automated Extraction method or Qiagen™ Puregene kit for DNA extraction is recommended.

**Specimen Collection and Shipping:**

Refrigerate until time of shipment in 100 ng/µL in TE buffer. Ship sample at room temperature with overnight delivery.

#### Type: Whole Blood (EDTA)

**Specimen Requirements:**

- EDTA (Purple Top)

  - Infants and Young Children (2 years of age to 10 years old): 3-5 ml
  - Older Children & Adults: 5-10 ml

  Autopsy: 2-3 ml unclotted cord or cardiac blood

**Specimen Collection and Shipping:**

Ship sample at room temperature for receipt at EGL within 72 hours of collection. Do not freeze.

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## 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