High Risk Colorectal Cancer: Deletion/Duplication Panel

**Test Code:** MD205  
**Turnaround time:** 2 weeks  
**CPT Codes:** 81436 x1

### Condition Description

EGL Genetics (EGL) High Risk Colorectal Cancer Panel include the well-described hereditary cancer predisposition syndromes; Lynch syndrome, familial adenomatous polyposis, and MYH-associated polyposis. Lynch syndrome, familial adenomatous polyposis, and MutY homolog (MYH)-associated polyposis are three major known types of inherited colorectal cancer, which accounts for up to 5% of all colon cancer cases. Lynch syndrome is most frequently caused by mutations in the mismatch repair genes MLH1, MSH2, MSH6, PMS2, and EPCAM, and is inherited in an autosomal dominant manner.

Familial adenomatous polyposis is manifested as colonic polyposis caused by mutations in the APC gene and is also inherited in an autosomal dominant manner. Finally, MYH-associated polyposis is caused by mutations in the MUTYH gene and is inherited in an autosomal recessive manner but may or may not be associated with polyps. There are variants of both familial adenomatous polyposis (Gardner syndrome—with extracolonic features—and Turcot syndrome, which features medulloblastoma) and Lynch syndrome (Muir-Torre syndrome features sebaceous skin carcinomas, and Turcot syndrome features glioblastomas). Although a clinical diagnosis of familial adenomatous polyposis can be made using colonoscopy, genetic testing is needed to inform at-risk relatives. Because of the overlapping phenotypes between attenuated familial adenomatous polyposis, MYH-associated polyposis, and Lynch syndrome, genetic testing is needed to distinguish among these conditions. This distinction is important, especially for women with Lynch syndrome, who are at increased risk for gynecological cancers.

**Reference:**  

### Genes

APC, ATM, BLM, BMPR1A, CDH1, CHEK2, EPCAM, MLH1, MSH2, MSH6, MUTYH, PTEN, SMAD4, STK11, TP53

### Indications

The test is indicated for:

- Individuals with a clinical or suspected diagnosis of high risk colorectal 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 Analysis:** Detection is limited to duplications and deletions. The CGH array will not detect point or intronic mutations. 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 sample at room temperature with overnight delivery.

**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.
- High Risk Colorectal Cancer: Sequencing Panel.