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.

**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: 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

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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.

**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.