

Gastrointestinal and Colorectal Cancer: Deletion/Duplication Panel

Test Code: MD209

Turnaround time: 2 weeks

CPT Codes: 81436 x1

Condition Description

Gastrointestinal cancer affects the gastrointestinal tract and accessory organs of digestion, including the esophagus, stomach, biliary system, pancreas, small intestine, large intestine, rectum and anus. Gastric cancer is a manifestation of a number of inherited cancer predisposition syndromes. The Gastrointestinal and Colorectal Cancer Panel targets genes known to be associated with various cancers that can develop in these organs and along the gastrointestinal tract.

The Gastrointestinal and Colorectal Cancer Panel also includes testing for the well-described hereditary cancer predisposition syndromes; Lynch syndrome, familial adenomatous polyposis (FAP), and MYH-associated polyposis (MAP). Lynch syndrome, FAP, and MutY homolog MAP are three major known types of inherited colorectal cancer, which account 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:

- Hegde et al. ACMG technical standards and guidelines for genetic testing for inherited colorectal cancer (Lynch syndrome, familial adenomatous polyposis, and MYH-associated polyposis). *Genet Med.* 16 1, 101–116.

Genes

[APC](#), [ATM](#), [BLM](#), [BMPR1A](#), [BRCA1](#), [BRCA2](#), [CDH1](#), [CDKN2A](#), [CHEK2](#), [EPCAM](#), [MLH1](#), [MSH2](#), [MSH6](#), [MUTYH](#), [POLD1](#), [PTEN](#), [SMAD4](#), [STK11](#), [TP53](#)

Indications

The test is indicated for:

- Individuals with a clinical or suspected diagnosis of gastrointestinal or 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

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.
- Pancreatic Cancer: Sequencing Panel.
- Gastrointestinal and Colorectal Cancer: Sequencing Panel.