

Early Onset Inflammatory Bowel Disease: Deletion/Duplication Panel

Test Code: MD160

Turnaround time: 2 weeks

CPT Codes: 81323 x1, 81403 x1, 81228 x1

Condition Description

Inflammatory bowel disease (IBD), encompassing Crohn's disease, ulcerative colitis, and unclassified IBD (IBDU) is characterized by chronic intestinal inflammation and has multi-factorial etiology with complex interactions between genetic and environmental factors. Although the genetics of IBD is believed to be common and complex, over 150 genetic loci have been described to be associated with IBD. The genetic contribution of the majority of those common loci towards explained heritability or their effect sizes are low. Recent studies have revealed an increasing spectrum of human monogenic diseases with high effect sizes/penetrance that can present with IBD or IBD-like intestinal inflammation. A substantial proportion of patients with those genetic defects present with very early onset intestinal inflammations, particularly if the onset of IBD occurs in subjects less than 10 years of age. There is also considerable overlap between primary immunodeficiency and very early onset IBD. Over 20 monogenic defects/genetic loci have been selected in this genetic diagnostic panel to test for very early onset IBD or IBD-like diseases. In addition to IBD or IBD-like diseases, these monogenic disorders also overlap with immunodeficiency affecting granulocyte and phagocyte activity, hyper- and autoinflammatory disorders, defects with disturbed T and B lymphocyte selection and activation, and defects in immune regulation affecting regulatory T cell activity and interleukin (IL)-10 signaling.

The Inflammatory Bowel Disease Panel will:

- Identify mutations associated with very early onset IBD (onset in less than 10 years) or IBD-like diseases
- Make a molecular diagnosis with the basis of pathogenesis
- Obtain rationale for patient-specific early intervention with emerging or experimental therapeutics and cell-based approaches
- Screen family members for carrier detection and genetic counseling

References:

- Uhling, (2013), *Gut*, 62: 1795-1805.
- Zhang and Li, (2014), *World J Gastroenterol*, 20: 91-99.
- OMIM

Genes

[AICDA](#), [BTK](#), [CD40LG](#), [CYBA](#), [CYBB](#), [DCLRE1C](#), [FOXP3](#), [HPS1](#), [HPS4](#), [HPS6](#), [ICOS](#), [IL10RA](#), [IL2RA](#), [LRBA](#), [MEFV](#), [MVK](#), [NCF2](#), [NCF4](#), [PTEN](#), [RET](#), [SH2D1A](#), [SLC37A4](#), [STXBP2](#), [TTC37](#), [WAS](#), [XIAP](#)

Indications

This test is indicated for:

- Confirmation of a clinical diagnosis of inflammatory bowel diseases (IBD).
- Carrier testing in adults with a family history of inflammatory bowel diseases (IBD).

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: 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.

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.

Related Tests

- Early Onset Inflammatory Bowel Disease: Sequencing Panel