**Hereditary Hemorrhagic Telangiectasia: Deletion/Duplication Panel**

**Test Code:** MD390  
**Turnaround time:** 2 weeks  
**CPT Codes:** 81405 x1, 81228 x1

### Condition Description

Hereditary hemorrhagic telangiectasia (HHT) is an autosomal dominant genetic disorder that leads to abnormal blood vessel formation in the skin, mucous membranes, and often in organs such as the lungs, liver, and brain. Of the five types of HHT identified, three have been linked to specific genes. More than 80% of all cases of HHT are due to mutations in either *ENG* or *ACVRL1*. *SMAD4* mutations, which cause colonic polyposis in addition to HHT, comprise about 2% of disease-causing mutations.

**Reference:**


### Genes

*ACVRL1, ENG, GDF2, RASA1, SMAD4*

### Indications

The test is indicated for:

- Individuals with a clinical or suspected diagnosis of hereditary hemorrhagic telangiectasia.

### 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: 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

- Hereditary Hemorrhagic Telangiectasia: Sequencing Panel