## 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. 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: 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

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/μl of TE buffer. Ship sample at room temperature with overnight delivery.

### Related Tests

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• Hereditary Hemorrhagic Telangiectasia: Sequencing Panel