Pulmonary Hypertension: Deletion/Duplication Panel

**Test Code:** MD098  
**Turnaround time:** 2 weeks  
**CPT Codes:** 81228 x1, 81405 x1

### Condition Description

Pulmonary hypertension (PH) is increased pulmonary pressure in the absence of common causes such as lung, heart, or thromboembolic chronic diseases. It is thought that both genetic and environmental factors that alter vascular structure and function contribute to the pathogenesis of PH.

Familial cases of PH are usually inherited in an autosomal dominant manner. With the identification of pathogenic variants in genes known to cause PH, what was previously thought to be idiopathic PH is now known to be genetic. A pathogenic variant in the **BMPR2** gene causes ~70% of hereditary cases of pulmonary arterial hypertension (PAH) and in 10-40% of idiopathic PAH. Other genes with pathogenic variants implicated in PH include: **CAV1, GDF2, RASA1, SMAD4, and SMAD9**.

Heterozygous pathogenic variants in the **ENG** and **ACVRL1** (previously known as **ALK1**) genes cause hereditary hemorrhagic telangiectasia (HHT). HHT is an autosomal dominant vascular disorder characterized by acquired cutaneous telangiectasias and arteriovenous malformations that can lead to the development of PAH.

### References:

- OMIM

### Genes

**ACVRL1, BMPR2, CAV1, ENG, GDF2, RASA1, SMAD4, SMAD9**

### Indications

This test is indicated for:

- Confirmation of a clinical diagnosis of pulmonary hypertension.
- Carrier testing in adults with a family history of pulmonary hypertension.

### Methodology

**Deletion/Duplication Analysis:** DNA isolated from peripheral blood is hybridized to a CGH array to detect deletions and duplications. The targeted CGH array has overlapping probes which 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.

Related Tests

- Pulmonary Hypertension: Sequencing Panel
- Comprehensive cardiomyopathy panel