

Pulmonary Fibrosis and Hermansky-Pudlak Syndrome: Deletion/Duplication Panel

Test Code: MD242

Turnaround time: 2 weeks

CPT Codes: 81228 x1

Condition Description

Hermansky-Pudlak syndrome (HPS) is an autosomal recessive, multisystemic disorder. The main clinical features of HPS include oculocutaneous albinism, which presents as hypopigmentation of the skin and hair; reduced iris and retinal pigments; foveal hypoplasia; nystagmus; increased crossing of optic fibers; bleeding diathesis due to a platelet storage pool deficiency; and deposition of lysosomal ceroid, which may cause pulmonary fibrosis (onset in the early thirties), granulomatous colitis (severe presentation in ~ 15% of all cases), and cardiomyopathy in some cases.

The clinical features of HPS are caused by the disruption of lysosome-related organelles in different tissue types. The incidence of HPS is approximately 1 in 500,000-1,000,000. HPS has an increased incidence, up to 1 in 1800, in Puerto Rico. Locus heterogeneity has been associated with HPS and nine causative genes (HPS1-HPS9) have been identified to date.

Pulmonary fibrosis is a condition in which the lung tissue becomes thickened and scarred over time making the lungs incapable of transporting oxygen into the bloodstream effectively. The most common signs and symptoms of idiopathic pulmonary fibrosis are shortness of breath and a persistent dry, hacking cough. Many affected individuals also experience a loss of appetite and gradual weight loss. It is reported that about 0.5-3.7% of idiopathic pulmonary fibrosis is familial.

References:

- GeneReviews.
- Wei & Li (2013). *Pigm Cell Melanoma R*. 26:176-192
- OMIM #203300: HPS.

Genes

[ABCA3](#), [AP3B1](#), [BLOC1S3](#), [BLOC1S6](#), [CSF2RA](#), [DTNBP1](#), [ELMOD2](#), [HPS1](#), [HPS3](#), [HPS4](#), [HPS5](#), [HPS6](#), [SFTPB](#), [SFTPC](#), [SFTPD](#), [TERT](#)

Indications

This test is indicated for:

- Confirmation of a clinical diagnosis of pulmonary fibrosis.
- Confirmation of a clinical diagnosis of Hermansky-Pudlak syndrome.

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.

Related Tests

- Pulmonary Fibrosis and Hermansky-Pudlak Syndrome: Sequencing Panel
- Pulmonary Disease: Sequencing Comprehensive Panel