Interstitial Lung Disease: Deletion/Duplication Panel

Test Code: MD590
Turnaround time: 2 weeks
CPT Codes: 81228 x1

Condition Description

Interstitial lung disease refers to a group of disorders that cause progressive scarring of the lung tissue, or interstitium. The interstitium provides support to the lungs and contains blood vessels that allow for proper transport of oxygen. This irreversible scarring leads to breathing difficulties including chronic cough, rapid breathing, and shortness of breath. Although there are various non-genetic causes, such as environmental exposures and autoimmune disorders, improved understanding of genetics has created the possibility of including genetic evaluation in the clinical work-up.

Genes

ABCA3, CSF2RA, SFTPB, SFTPC, TERT

Indications

This test is indicated for:

- Confirmation of a clinical diagnosis of suspected interstitial lung disease.

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 ElmerChemagenChemagen Automated Extraction method or QiagenPuregene 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

- Interstitial Lung Disease: Sequencing Panel
- Pulmonary Disease: Sequencing Panel