Interstitial Lung Disease: Sequencing Panel

**Test Code:** MM590  
**Turnaround time:** 6 weeks  
**CPT Codes:** 81479 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, CSF2RB, NKX2-1, SFTPB, SFTPC, TERT

### Indications

This test is indicated for:

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

### Methodology

**Next Generation Sequencing:** In-solution hybridization of all coding exons is performed on the patient's genomic DNA. Although some deep intronic regions may also be analyzed, this assay is not mean to interrogate most promoter regions, deep intronic regions, or other regulatory elements, and does not detect single or multi-exon deletions or duplications. Direct sequencing of the captured regions is performed using next generation sequencing. The patient's gene sequences are then compared to a standard reference sequence. Potentially causative variants and areas of low coverage are Sanger-sequenced. Sequence variations are classified as pathogenic, likely pathogenic, benign, likely benign, or variants of unknown significance. Variants of unknown significance may require further studies of the patient and/or family members.

### Detection

**Clinical Sensitivity:** Unknown. Pathogenic variants in the promoter region, some pathogenic variants in the introns and other regulatory element pathogenic variants cannot be detected by this analysis. Large deletions will not be detected by this analysis. Results of molecular analysis should be interpreted in the context of the patient's clinical and/or biochemical phenotype.

**Analytical Sensitivity:** ~99%.

### 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.
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