Cystic Lung Disease: Sequencing Panel

Test Code: MM243  
Turnaround time: 6 weeks  
CPT Codes: 81406 x1, 81407 x1

**Condition Description**

Certain hereditary lung diseases cause cystic lesions in the lung leading to symptoms such as breathing difficulties, pain and pneumothorax. These cystic lesions are sacs filled with air, fluids, or semi-solid material.

Conditions which present with lung cystic lesions include alpha-1-antitrypsin deficiency, Birt-Hogg-Dube syndrome, tuberous sclerosis, complex-associated lymphangioleiomyomatosis (LAM), cutis laxa, and emphysema.

Reference:
- Genereviews

**Genes**

EFEMP2, ELN, FBLN5, FLCN, LTBP4, SERPINA1, TSC1, TSC2

**Indications**

This test is indicated for:

- Confirmation of a clinical diagnosis in patients with cystic lung lesions.

**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 meant 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**

**Next Generation Sequencing**: 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: 60 ug

Isolation using the Qiagen™ Puregene kit for DNA extraction is recommended.

Specimen Collection and Shipping: Refrigerate until time of shipment in 100 ng/ul of TE buffer. Ship sample at room temperature with overnight delivery.
Related Tests

- Pulmonary Disease: Comprehensive Sequencing Panel
- Bronchiectasis Panel
- Pulmonary Fibrosis - Hermansky-Pudlak Syndrome Panel
- Pulmonary Arterial Hypertension Panel
- Congenital Central Hypoventilation Panel
- Cystic Lung Disease: Deletion/Duplication Panel