Cystic Lung Disease: Deletion/Duplication Panel

Test Code: MD243
Turnaround time: 2 weeks
CPT Codes: 81228 x1, 81405 x1, 81406 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**

**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.

Please note that a "backbone" of probes across the entire genome are included on the array for analytical and quality control purposes. Rarely, off-target copy number variants causative of disease may be identified that may or may not be related to the patient's phenotype. Only known pathogenic off-target copy number variants will be reported. Off-target copy number variants of unknown clinical significance will not be reported.

**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**

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/ul of TE buffer. Ship sample at room temperature with overnight delivery.
Related Tests

- Cystic Lung Disease: Sequencing Panel
- Pulmonary Disease: Comprehensive Sequencing Panel