Cystic Lung Disease: Deletion/Duplication Panel

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

Summary

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
- FLTP4
- 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

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