

Cystic Lung Disease: Deletion/Duplication Panel

Test Code: MD243

Turnaround time: 2 weeks

CPT Codes: 81228 x1, 81406 x1, 81405 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 lymphangioliomyomatosis (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.

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: DNA, Isolated

Specimen Requirements:

Microtainer

3µg

Isolation using the Perkin Elmer™ Chemagen™ Chemagen™ Automated Extraction method or Qiagen™ Puregene 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.

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.

Related Tests

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