Bronchiectasis: Deletion/Duplication Panel

Test Code: MD241  
Turnaround time: 2 weeks  
CPT Codes: 81222 x1, 81228 x1

Condition Description

Bronchiectasis is the abnormal and irreversible dilatation of the bronchi and is frequently associated with inflammation. Genetic diseases which predispose patients to recurrent or chronic lung infections, such as cystic fibrosis and primary ciliary dyskinesia, have been identified in a large proportion of those with bronchiectasis.

Reference:

Genes

CCDC39, CCDC40, CFTR, DNAAF1, DNAAF2, DNAH11, DNAH5, DNAI1, DNAI2, DNA1, NME8, RSPH4A, RSPH9, SCNN1A, SCNN1B, SCNN1G

Indications

This test is indicated for:
- Confirmation of a suspected hereditary respiratory disease in patients with bronchiectasis.

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: 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.

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.

Related Tests

- Bronchiectasis: Sequencing Panel
- Pulmonary Disease: Comprehensive Sequencing Panel
- Pulmonary Hypertension: Sequencing Panel
- Cystic Lung Disease: Sequencing Panel
- Pulmonary Fibrosis and Hermansky-Pudlak Syndrome: Sequencing Panel

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• Congenital Central Hypoventilation: Sequencing Panel