Bronchiectasis: Sequencing Panel

Test Code: MM241
Turnaround time: 6 weeks
CPT Codes: 81223 x1, 81406 x1, 81479 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, DNAL1, NME8, RSPH4A, RSPH9, SCN1A, SCN1B, SCN1G

Indications

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

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