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:

- Notarangelo et al. (2007) *Respiration. 74*(3); 264-275.

### Genes

CCDC39, CCDC40, CFTR, DNAAF1, DNAAF2, DNAH11, DNAH5, DNAI1, DNAI2, DNAI1, 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. 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: 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

- 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
- Congenital Central Hypoventilation: Sequencing Panel