**Condition Description**

A number of genes influence lung formation and function at birth. Defects in these genes cause hereditary syndromic and non-syndromic pulmonary diseases such as cystic lung disease, bronchiectasis, idiopathic pulmonary fibrosis, and cystic lung disease.

Pathogenic changes in genes associated with these diseases cause severe, and sometimes lethal, lung malformations including those in the sonic hedgehog, fibroblast growth factor and thyroid transcription factor-1 pathways.

Reference:

**Genes**

- ABCA3, ACVRL1, AP3B1, ASCL1, BDNF, BLOC1S3, BLOC1S6, BMPR2, CCDC39, CCDC40, CFTR, CFSP2RA, DNAAF1, DNAAF2, DNAH11, DNAH5, DNAI1, DNAI2, DNAJ, DOC5, DONBP1, EDN3, EFEMP2, ELMOD2, ELMN, ENG, FBLN5, FLCN, GDNF, HPS1, HPS3, HPS4, HPSS, HPS6, LTBP4, NME8, PHOX2B, RET, RSPH4A, RSPH9, SCN1A, SCN1B, SCN1G, SERPINA1, SFTPB, SFTPQ, SFTPD, SMAD9, STAT3, TERT, TSC1, TSC2

**Indications**

This test is indicated for:

- Confirmation of a clinical diagnosis of suspected hereditary respiratory disease.

**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**

- Pulmonary Disease: Sequencing Panel
- Bronchiectasis: Sequencing Panel
- Cystic Lung Disease: Sequencing Panel
- Pulmonary Fibrosis - Hermansky-Pudlak Syndrome: Sequencing Panel
- Pulmonary Hypertension: Sequencing Panel
- Congenital Central Hypoventilation: Sequencing Panel