Test Code: MM240
Turnaround time: 6 weeks
CPT Codes: 81223 x1, 81332 x1, 81404 x1, 81405 x1, 81406 x1, 81479 x1

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

<table>
<thead>
<tr>
<th>Genes</th>
</tr>
</thead>
<tbody>
<tr>
<td>ABCA3, ACVR1L, AP3B1, ASCL1, BDNF, BLOC1S3, BLOC1S6, BMP2, CCDC39, CCDC40, CFTR, CSF2RA, DNAAF1, DNAAF2, DNAH11, DNAH4, DNAI1, DNAI2, DNAJ1, DOCK8, DTPBP1, EDN3, EFEMP2, ELMOD2, ELN, ENG, FBLN5, FLCN, GDNF, HPS1, HPS3, HPS4, HPS5, HPS6, LTBP4, NME8, PHOX2B, RET, RSPH4A, RSPH9, SCNN1A, SCNN1B, SCNN1G, SERPINA1, SFTPB, SFTPC, SFTPD, SMAD9, STAT3, TERT, TSC1, TSC2</td>
</tr>
</tbody>
</table>

Indications

This test is indicated for:
- Confirmation of a clinical diagnosis of suspected hereditary respiratory disease.

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.

Disclaimer: This information is confidential and subject to change without notice. It may not be reproduced in whole or part unless authorized in writing by an authorized EGL representative.
Related Tests

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