The \textit{ABCA3} gene contains 30 exons and spans around 11 kb mapped to 16p13.3, and encodes a membrane-associated protein that is a member of the superfamily of ATP-binding cassette (ABC) transporters. It is expressed in alveolar type II pneumocytes and localizes predominantly to the limiting membrane of lamellar bodies. The \textit{ABCA3} protein plays an important role in the proper formation of lamellar bodies and pulmonary surfactant, probably by transporting lipids such as cholesterol. The \textit{ABCA3} mutations are passed on in an autosomal dominant fashion. Diseases associated with \textit{ABCA3} include pulmonary surfactant metabolism dysfunction 3 and abca3-related pulmonary surfactant metabolism dysfunction. Definitive genotype/phenotype correlations have not been described.

The John Welsh Cardiovascular Diagnostic Laboratory offers molecular genetic testing for \textit{ABCA3} mutations. Individuals are tested by DNA sequencing of the coding exons of the \textit{ABCA3} gene. We strongly recommend initial testing of a clearly affected individual, if available, in order to provide the greatest test sensitivity and clearest interpretation of results for subsequent family members. Genetic counseling is recommended for all individuals.

**REASONS FOR REFERRAL**

Molecular confirmation of the diagnosis of IPF, pulmonary surfactant metabolism dysfunction 3 and abca3-related pulmonary surfactant metabolism dysfunction.

**METHODOLOGY**

Genomic DNA is analyzed for \textit{ABCA3} mutations by DNA sequencing of the coding exons of the \textit{ABCA3} gene, as well as the exon/intron junctions and a portion of the 5’ and 3’ untranslated regions. Patient DNA is sequenced in both the forward and reverse orientations. If a mutation is identified, additional family members are analyzed only for the familial mutation by automatic fluorescent DNA sequencing.

**SERVICE FEES**

<table>
<thead>
<tr>
<th>Index Case (Male or Female)</th>
<th>Direct and Institutional Billing</th>
<th>CPT Codes</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>$1,200 per sample</td>
<td>81407</td>
</tr>
<tr>
<td>Additional Family Members</td>
<td>$300 per sample; known familial mutation only</td>
<td>81403</td>
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</tbody>
</table>

**SENSITIVITY**

DNA Sequencing Analysis: Approximately 99 percent detection of mutations in the coding exons 1-30 of \textit{ABCA3}.

**SPECIMEN REQUIREMENTS**

- **Blood** (preferred): EDTA (purple-top) tubes: \textit{Adult}: 5 cc \textit{Child}: 5 cc \textit{Infant}: 2-3 cc
- **Tissue**: Frozen (preferred) or RNAlater
- **Other Body Fluids or Formalin-Fixed, Paraffin-Embedded Tissue**: Call to inquire

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