The *SFTPD* gene contains 7 exons and spans around 11 kb mapped to 10q22.3, and encodes surfactant protein D. Pulmonary surfactant consists of a complex mixture of phospholipids and several proteins essential to normal respiratory function. Pulmonary surfactant proteins contribute to the lung's defense against inhaled microorganisms, organic antigens and toxins. It interacts with compounds such as bacterial lipopolysaccharides, oligosaccharides and fatty acids and modulates leukocyte action in immune response. The SFTPD protein has 355 amino acid residues with a short noncollagen-like N-terminal section of 25 residues, followed by a collagen-like region of 177 residues and a C-terminal C-type lectin domain of 153 residues. Diseases associated with SFTPD include idiopathic pulmonary alveolar proteinosis and Wilson-Mikity syndrome. Definitive genotype/phenotype correlations have not been described.

The John Welsh Cardiovascular Diagnostic Laboratory offers molecular genetic testing for *SFTPD* mutations. Individuals are tested by DNA sequencing of the coding exons of the *SFTPD* 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 idiopathic pulmonary alveolar proteinosis and wilson-mikity syndrome.

**METHODOLOGY**

Genomic DNA is analyzed for *SFTPD* mutations by DNA sequencing of the coding exons of the *SFTPD* 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>$700 per sample</td>
<td>81405</td>
</tr>
</tbody>
</table>

| Additional Family Members  | $300 per sample;               | 81403     |
|                           | known familial mutation only    |           |

**SENSITIVITY**

DNA Sequencing Analysis: Approximately 99 percent detection of mutations in the coding exons 1-7 of *SFTPD*.

**SPECIMEN REQUIREMENTS**

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