SFTPC gene encodes pulmonary-associated surfactant protein C (SPC) which is produced and secreted by type II alveolar epithelial cells. Due to its extremely hydrophobic nature, pulmonary-associated surfactant protein C assists in the prevention of lung collapse by lowering surface tension of fluids that coat the lung. SFTPC gene consists of six exons that span 3.5-kb genomic distance mapped to chromosome 8p21.3. Alternatively spliced transcript variants encoding different protein isoforms have been previously identified. It is believed that SFTPC gene mutations cause alteration of pulmonary-associated surfactant protein C metabolism or complete pulmonary-associated surfactant protein C deficiency leading to diffuse lung disease. SFTPC mutations appear to demonstrate autosomal dominant inheritance with reduced penetrance. Definitive genotype/phenotype correlations have not yet been described.

The John Welsh Cardiovascular Diagnostic Laboratory offers molecular genetic testing for SFTPC mutations. Individuals are tested by DNA sequencing of the coding exons of the SFTPC 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 fibrosis (IPF) or familial pulmonary fibrosis (FPF).

### METHODOLOGY

Genomic DNA is analyzed for SFTPC mutations by DNA sequencing of the coding exons of the SFTPC gene, as well as the exon/intron junctions and a portion of the 5’ and 3’ untranslated region. 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>
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<th>Direct and Institutional Billing</th>
<th>CPT Codes</th>
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<tbody>
<tr>
<td>Index Case (Male or Female)</td>
<td>$600 per sample</td>
<td>81404</td>
</tr>
<tr>
<td>Additional Family Members</td>
<td>$300 per sample; known familial mutation only</td>
<td>81403</td>
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### SENSITIVITY

DNA Sequencing Analysis: Approximately 99 percent detection of mutations in the coding exons 1-5 of SFTPC.

### SPECIMEN REQUIREMENTS

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