SFTPA2 gene encodes surfactant, pulmonary-associated protein A2 (SFTPA2), a protein that not only plays a role in innate host defense in the lung, but also assists with maintaining alveolar stability. Transfection studies in human alveolar epithelial cells have shown that mutant SFTPA2 proteins are not secreted but are instead confined to the endoplasmic reticulum, leading to a decreased concentration of mature SFTPA2 protein in the alveolar space. It is likely a result of disruption to the protein structure due to missense mutations in the SFTPA2 gene which lead to a change in the amino acid sequence at highly conserved regions within the carbohydrate-recognition domain of the SFTPA2 protein. SFTPA2 gene consists of five exons mapped to chromosome 10q22.3 and appears to demonstrate autosomal dominant inheritance. Definitive genotype/phenotype correlations have not yet been described.

The John Welsh Cardiovascular Diagnostic Laboratory offers molecular genetic testing for SFTPA2 mutations. Individuals are tested by DNA sequencing of the coding exons of the SFTPA2 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 SFTPA2 mutations by DNA sequencing of the coding exons of the SFTPA2 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**

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<th>Direct and Institutional Billing</th>
<th>CPT Codes</th>
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<td>Index Case (Male or Female)</td>
<td>$600 per sample</td>
<td>81404</td>
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<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 2-5 of SFTPA2.

**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