The *ELMOD2* (ELMO/CED-12 domain containing 2) gene contains 8 exons and spans around 30 kb mapped to 4q31.1. *ELMOD2* encodes one of six engulfment and motility (ELMO) domain-containing proteins. *ELMOD2* functions as a GTPase-activating protein (GAP) for members of the ARL and ARF families of small GTPases and is thought to play a role in antiviral responses. Mutations in this gene may be involved in the cause of familial idiopathic pulmonary fibrosis. Definitive genotype/phenotype correlations have not been described.

The John Welsh Cardiovascular Diagnostic Laboratory offers molecular genetic testing for *ELMOD2* mutations. Individuals are tested by DNA sequencing of the coding exons of the *ELMOD2* 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 familial idiopathic pulmonary fibrosis (IPF).

### METHODOLOGY

Genomic DNA is analyzed for *ELMOD2* mutations by DNA sequencing of the coding exons of the *ELMOD2* 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>Direct and Institutional Billing</th>
<th>CPT Codes</th>
</tr>
</thead>
<tbody>
<tr>
<td>Index Case (Male or Female)</td>
<td>$750 per sample</td>
</tr>
<tr>
<td>Additional Family Members</td>
<td>$300 per sample; known familial mutation only</td>
</tr>
</tbody>
</table>

### SENSITIVITY

DNA Sequencing Analysis: Approximately 99 percent detection of mutations in the coding exons 1-8 of *ELMOD2*.

### 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