The COA5 gene is located at 2q11.2 and encodes an assembly factor for mitochondrial cytochrome c oxidase (complex IV). Diseases associated with COA5 include cytochrome-c oxidase deficiency disease and leigh syndrome with cardiomyopathy. COA5 mutations are passed on in an autosomal recessive fashion. Definitive genotype/phenotype correlations have not been described.

The John Welsh Cardiovascular Diagnostic Laboratory offers molecular genetic testing for COA5 mutations. Individuals are tested by DNA sequencing of the coding exons of the COA5 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 cytochrome-c oxidase deficiency disease and leigh syndrome with cardiomyopathy.

**METHODOLOGY**

Genomic DNA is analyzed for COA5 mutations by DNA sequencing of the coding exons of the COA5 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>
<th>Index Case (Male or Female)</th>
<th>Direct and Institutional Billing</th>
<th>CPT Codes</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>$500 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>
</tr>
</tbody>
</table>

**SENSITIVITY**

DNA Sequencing Analysis: Approximately 99 percent detection of mutations in the coding exons 1-3 of COA5.

**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