The SDHA gene encodes succinate dehydrogenase subunit A, flavoprotein subunit (SDHA) which is one of the 4 components of complex II of the mitochondrial respiratory chain. The SDHA gene contains 16 exons that were mapped to chromosome 5p15. Defects in this gene are the cause of dilated cardiomyopathy (DCM). DCM is a type of cardiomyopathy in which the heart becomes weakened and enlarged and cannot pump blood efficiently. About 25–35% of patients have familial forms of the disease. The disease is genetically heterogeneous, but the most common form of its transmission is autosomal dominant. Definitive genotype/phenotype correlations have not been described.

The John Welsh Cardiovascular Diagnostic Laboratory offers molecular genetic testing for SDHA mutations. Individuals are tested by DNA sequencing of the coding exons of the SDHA 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 dilated cardiomyopathy (DCM).

### METHODOLOGY

Genomic DNA is analyzed for SDHA mutations by DNA sequencing of the coding exons of the SDHA 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>
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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>$1100 per sample</td>
<td>81406</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 1-16 of SDHA.

### 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 and Formalin-fixed, Paraffin-embedded Tissue:** Call to inquire