**NEXN MUTATION ANALYSIS**

**JOHN WELSH CARDIOVASCULAR DIAGNOSTIC LABORATORY**

*NEXN* (nexilin) gene encodes a filamentous actin-binding protein that may function in cell adhesion and migration. Nexilin also has an essential role in the maintenance of Z line and sarcomere integrity. *NEXN* gene contains 12 coding exons and spans a genomic distance of about 55.38 kb that has been mapped to chromosome 1p31.1. Alternatively spliced transcript variants have been described. Mutations in this gene have been associated with dilated cardiomyopathy 1cc (CMD1CC) and familial hypertrophic cardiomyopathy type 20. *NEXN* mutations demonstrate autosomal dominant inheritance with a broad range of clinical severity both within and between families. Definitive genotype/phenotype correlations have not been described.

The John Welsh Cardiovascular Diagnostic Laboratory offers molecular genetic testing for *NEXN* mutations. Individuals are tested by DNA sequencing of the coding exons of the *NEXN* 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 1cc (CMD1CC) and familial hypertrophic cardiomyopathy type 20.

**METHODOLOGY**

Genomic DNA is analyzed for *NEXN* mutations by DNA sequencing of the coding exons of the *NEXN* 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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<thead>
<tr>
<th></th>
<th>Direct and Institutional Billing</th>
<th>CPT Codes</th>
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<tbody>
<tr>
<td>Index Case (Male or Female)</td>
<td>$1,000 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 2-13 of *NEXN*.

**SPECIMEN REQUIREMENTS**

**Blood (preferred):** EDTA (purple-top) tubes: **Adult:** 5 cc  **Child:** 5 cc  **Infant:** 2-3 cc

**Tissue:** Frozen (preferred), RNA*Later*

**Other Body Fluids and Formalin-fixed, Paraffin-embedded Tissue:** Call to inquire