Emerin is an inner nuclear membrane protein that is expressed in cardiac, skeletal and smooth muscle. Multiple functions for emerin, including the stabilization of the nuclear membrane during muscle contraction and organizational maintenance of the nuclear membrane during cell division, have been proposed. Emerin is encoded by the gene EMD (STA), which is composed of six exons and is located at Xq28.

Mutations in EMD have been identified in males with X-linked recessive Emery-Dreifuss muscular dystrophy, which is characterized by early contractures of the Achilles tendons, elbows, and post-cervical muscles with humero-peroneal weakness and muscle wasting. Affected individuals also have cardiac conduction defects and may have a generalized cardiomyopathy. X-linked recessive EMD mutations identified in affected individuals include missense, nonsense, splicing, small deletion or insertion, and larger genomic DNA deletion mutations.

The John Welsh Cardiovascular Diagnostic Laboratory offers molecular genetic testing for EMD mutations. Symptomatic males are tested by DNA sequencing of all 6 exons of the EMD gene. We strongly recommend initial testing of an affected male, if available, in order to provide the greatest test sensitivity and clearest interpretation of results for subsequent carrier female testing. If an affected male is unavailable for testing, testing of females at high risk is offered. Genetic counseling is recommended for all individuals in order to identify additional at-risk family members and to discuss reproductive issues.

**REASONS FOR REFERRAL**
- Molecular confirmation of the diagnosis of Emery-Dreifuss muscular dystrophy in males
- Carrier testing in females with a family history of Emery-Dreifuss muscular dystrophy in males
- Carrier testing is not offered for asymptomatic minor females. Please call for additional information.

**METHODOLOGY**
Genomic DNA is analyzed for EMD mutations by DNA sequencing of all 6 exons of the EMD 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>Service Description</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>$450 per sample</td>
<td>81405</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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</tbody>
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**SENSITIVITY**
- Males - DNA Sequencing Analysis: Approximately 97.5% detection of mutations in exons 1-6 of EMD
- Females - DNA Sequencing Analysis: Approximately 93.5% detection of mutations in exons 1-6 of EMD

**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