Emerin is an inner nuclear membrane protein that is expressed in cardiac, skeletal, and smooth muscle. Multiple functions for emerin have been proposed including the stabilization of the nuclear membrane during muscle contraction and organizational maintenance of the nuclear membrane during cell division. Emerin is encoded by the gene \textit{EMD} (\textit{STA}), which is composed of 6 exons and is located at Xq28.

Mutations in \textit{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 \textit{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 \textit{EMD} mutations. Symptomatic males will be tested by automatic fluorescent DNA sequencing of all 6 exons of the \textit{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.

**METHODODOLOGY**
Genomic DNA will be analyzed for \textit{EMD} mutations by automatic fluorescent DNA sequencing of all 6 exons of the \textit{EMD} gene, as well as the exon/intron junctions and a portion of the 5` and 3` untranslated regions. Patient DNA will be sequenced in both the forward and reverse orientations. If a mutation is identified, additional family members will be 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>$450 per sample</td>
<td>83891, 83898x3, 83904x6, 83912</td>
</tr>
<tr>
<td>Additional Family Members</td>
<td>$200 per sample; known familial mutation only</td>
<td>83891, 83898, 83904x2, 83912</td>
</tr>
</tbody>
</table>

**SENSITIVITY**
- Males - DNA Sequencing Analysis: Approximately 97.5% detection of mutations in exons 1-6 of \textit{EMD}
- Females - DNA Sequencing Analysis: Approximately 93.5% detection of mutations in exons 1-6 of \textit{EMD}

**SPECIMEN REQUIREMENTS**
- **Blood (preferred):** EDTA (purple-top) tubes: \textit{Adult}: 5 cc \textit{Child}: 5 cc \textit{Infant}: 2-3 cc
- **Tissue:** Frozen (preferred), RNAlater, Formalin-fixed, Paraffin embedded
- **Other Body Fluids:** Call to inquire