The EFEMP2 gene encodes EFEMP2, a member of the fibulin family of proteins, characterized by tandem arrays of EGF-like domains and a C-terminal fibulin-type module. The EFEMP2 gene contains 11 exons that were mapped to chromosome 11q13. Mutations in this gene are associated with cutis latexit type 1B (AR: autosomal recessive) and lethal arteriopathy syndrome. Definitive genotype/phenotype correlations have not been described.

The John Welsh Cardiovascular Diagnostic Laboratory offers molecular genetic testing for EFEMP2 mutations. Individuals are tested by DNA sequencing of the coding exons of the EFEMP2 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 valvar and vascular disorders, lethal arteriopathy syndrome.

**METHODOLOGY**

Genomic DNA is analyzed for EFEMP2 mutations by DNA sequencing of the coding exons of the EFEMP2 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></th>
<th>Direct and Institutional Billing</th>
<th>CPT Codes</th>
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</thead>
<tbody>
<tr>
<td>Index Case (Male or Female)</td>
<td>$1,100 per sample</td>
<td>81406</td>
</tr>
<tr>
<td>Additional Family Members</td>
<td>$300 per sample; Known familial mutation only</td>
<td>81403</td>
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</tbody>
</table>

**SENSITIVITY**

DNA Sequencing Analysis: Approximately 99 percent detection of mutations in the 11 coding exons of EFEMP2.

**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

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