Elastic fibers are the major structural protein of tissues which must expand rapidly and recover completely, such as aorta and nuchal ligament. They contain two distinct components, a more abundant amorphous component (elastin) and the microfibrillar component. *ELN* gene encodes the elastin protein which is rich in glycine, proline and other hydrophobic amino acids. Multiple lysine-derived crosslinks link the individual polypeptide chains into a rubberlike network and the hydrophobic regions of the chains between the crosslinks are highly mobile. *ELN* gene contains 33 coding exons and spans around 49 kb of genomic distance on chromosome 7q11.23. Multiple transcript variants encoding different isoforms have been reported. Mutations in this gene are associated with supravalvular aortic stenosis (SVAS) and autosomal dominant cutis laxa. *ELN* mutations demonstrate both autosomal dominant and recessive 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 *ELN* mutations. Individuals are tested by DNA sequencing of the coding exons of the *ELN* 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 CHD, valvar and vascular disorders.

**METHODOLOGY**
Genomic DNA is analyzed for *ELN* mutations by DNA sequencing of the coding exons of the *ELN* 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**

<table>
<thead>
<tr>
<th></th>
<th>Direct and Institutional Billing</th>
<th>CPT Codes</th>
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<tr>
<td>Index Case (Male or Female)</td>
<td>$1,500 per sample</td>
<td>81407</td>
</tr>
<tr>
<td>Additional Family Members</td>
<td>$300 per sample; known familial mutation only</td>
<td>81403</td>
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</table>

**SENSITIVITY**
DNA Sequencing Analysis: Approximately 99 percent detection of mutations in the coding exons 1-33 of *ELN*.

**SPECIMEN REQUIREMENTS**

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

**Tissue:** Frozen (preferred), RNAlater

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

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