COL2A1 (collagen, type II, alpha 1) gene encodes the alpha-1 chain of type II collagen, a fibrillar collagen found in cartilage and the vitreous humor of the eye. It is essential for the normal embryonic development of the skeleton, for linear growth and for the ability of cartilage to resist compressive forces. COL2A1 gene contains 54 coding exons and spans over 31.5 kb, and has been mapped to chromosome 12q13.11. Mutations in this gene are associated with CHD, Valvar and Vascular Disorders. COL2A1 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 COL2A1 mutations. Individuals are tested by DNA sequencing of the coding exons of the COL2A1 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 COL2A1 mutations by DNA sequencing of the coding exons of the COL2A1 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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<tbody>
<tr>
<td>Index Case (Male or Female)</td>
<td>$1,600 per sample</td>
<td>81408</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

### 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