COL1A1 MUTATION ANALYSIS
JOHN WELSH CARDIOVASCULAR DIAGNOSTIC LABORATORY

The COL1A1 gene encodes collagen type I, alpha I (COL1A1), one of the major components of collagen of skin, tendon and bone. The COL1A1 gene contains 51 exons that were mapped to chromosome 17q21.31-q22.05. Mutations in this gene are associated with Caffey Disease, Osteogenesis Imperfecta and Ehlers-Danlos Syndromes I and VIIa. Definitive genotype/phenotype correlations have not been described.

The John Welsh Cardiovascular Diagnostic Laboratory offers molecular genetic testing for COL1A1 mutations. Individuals are tested by DNA sequencing of the coding exons of the COL1A1 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, Caffey Disease, Osteogenesis Imperfecta and Ehlers-Danlos Syndromes I and VIIa.

METHODOLOGY

Genomic DNA is analyzed for COL1A1 mutations by DNA sequencing of the coding exons of the COL1A1 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

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<tr>
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<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>81407</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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SENSITIVITY

DNA Sequencing Analysis: Approximately 99 percent detection of mutations in the 51 coding exons of COL1A1.

SPECIMEN REQUIREMENTS

Blood (preferred): EDTA (purple-top) tubes: Adult: 5 cc  Child: 5 cc  Infant: 2-3 cc
Tissue: Frozen (preferred), or RNA later
Other Body Fluids or Formalin-fixed, Paraffin-embedded tissue: Call to inquire