The \textit{CHST14} gene encodes dermatan-4-sulfotransferase-1 (D4ST1), which catalyzes the 4-O-sulfation of N-acetylgalactosamine (GalNAc) residues in dermatan sulfate, a stereoisomeric form of chondroitin sulfate that contains varying proportions of iduronic acid in place of glucuronic acid. The \textit{CHST14} gene contains 1 exon that was mapped to chromosome 15q14. Mutations in this gene are associated with Ehlers-Danlos syndrome, musculocontractural type 1. Definitive genotype/phenotype correlations have not been described.

The John Welsh Cardiovascular Diagnostic Laboratory offers molecular genetic testing for \textit{CHST14} mutations. Individuals are tested by DNA sequencing of the coding exons of the \textit{CHST14} 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 and musculocontractural Ehlers-Danlos syndrome type 1.

**METHODOLOGY**

Genomic DNA is analyzed for \textit{CHST14} mutations by DNA sequencing of the coding exons of the \textit{CHST14} 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>Description</th>
<th>Direct and Institutional Billing</th>
<th>CPT Codes</th>
</tr>
</thead>
<tbody>
<tr>
<td>Index Case (Male or Female)</td>
<td>$500 per sample</td>
<td>81403</td>
</tr>
<tr>
<td>Additional Family Members</td>
<td>$300 per sample; Known familial mutation only</td>
<td>81403</td>
</tr>
</tbody>
</table>

**SENSITIVITY**

DNA Sequencing Analysis: Approximately 99 percent detection of mutations in the coding exon of \textit{CHST14}.

**SPECIMEN REQUIREMENTS**

\textbf{Blood (preferred):} EDTA (purple-top) tubes: \textit{Adult}: 5 cc \hspace{1cm} \textit{Child}: 5 cc \hspace{1cm} \textit{Infant}: 2-3 cc

\textbf{Tissue:} Frozen (preferred), or RNAlater

\textbf{Other Body Fluids or Formalin-fixed, Paraffin-embedded tissue:} Call to inquire