The *JUP* gene encodes the junction plakoglobin, a major cytoplasmic protein which is the only known constituent common to submembranous plaques of both desmosomes and intermediate junctions. The *JUP* gene contains 14 exons and spans around 167.5 kb genomic distance that was mapped to chromosome 17q21.1. Mutations in *JUP* gene are associated with familial arrhythmogenic right ventricular dysplasia/cardiomyopathy type 12 (ARVD/C12) and Naxos disease. ARVD/C is an autosomal dominant disease characterized by partial degeneration of the myocardium of the right ventricle, electrical instability and sudden death. Naxos disease is an autosomal recessive disorder that combines palmoplantar keratoderma and other ectodermal features with cardiac disorders suggesting arrhythmogenic right ventricular dysplasia/cardiomyopathy. Definitive genotype/phenotype correlations have not been described.

The John Welsh Cardiovascular Diagnostic Laboratory offers molecular genetic testing for *JUP* mutations. Individuals are tested by DNA sequencing of the coding exons of the *JUP* 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 familial arrhythmogenic right ventricular dysplasia/cardiomyopathy type 12 (ARVD/C12) and Naxos disease.

**METHODOLOGY**

Genomic DNA is analyzed for *JUP* mutations by DNA sequencing of the coding exons of the *JUP* 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>Index Case (Male or Female)</th>
<th>Direct and Institutional Billing</th>
<th>CPT Codes</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>$1100 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 coding exons 1-14 of *JUP*.

**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 and Formalin-fixed, Paraffin-embedded Tissue:** Call to inquire

John Welsh Cardiovascular Diagnostic Laboratory • Section of Cardiology • Department of Pediatrics  
Baylor College of Medicine • 1102 Bates Avenue, Suite 480.02 • Houston, TX 77030  
PHONE: (832) 824-4155 • FAX: (832) 825-5159 • E-MAIL: yuxinf@bcm.edu  
Web Site: www.bcm.edu/pediatrics/welsh