**ARHGAP31 MUTATION ANALYSIS**

**JOHN WELSH CARDIOVASCULAR DIAGNOSTIC LABORATORY**

*ARHGAP31* gene encodes for a GTPase-activating protein (GAP). A variety of cellular processes are regulated by Rho GTPases which cycle between an inactive form bound to GDP and an active form bound to GTP. Research has shown that mutations in this gene result in Adams-Oliver Syndrome 1. Mapped to chromosome 3q13.33, *ARHGAP31* gene spans a genomic distance of approximately 126 kb, is composed of 12 exons, and has two splice variants. *ARHGAP31* mutations demonstrate autosomal dominant inheritance; however, definitive genotype/phenotype correlations have not yet been described.

The John Welsh Cardiovascular Diagnostic Laboratory offers molecular genetic testing for *ARHGAP31* mutations. Individuals are tested by DNA sequencing of the coding exons of the *ARHGAP31* 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, Adams-Oliver syndrome 1.

### METHODOLOGY

Genomic DNA is analyzed for *ARHGAP31* mutations by DNA sequencing of the coding exons of the *ARHGAP31* 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

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<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>$1,100 per sample</td>
<td>81406</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 coding exons 1-12 of *ARHGAP31*.

### 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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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