The *KCNA5* gene encodes the potassium voltage-gated ion channel, shaker-related subfamily, member 5 protein (KCNA5), also known as Kv1.5. This member contains six membrane-spanning domains with a shaker-type repeat in the fourth segment. It belongs to the delayed rectifier class, the function of which could restore the resting membrane potential of beta cells after depolarization and thereby contribute to the regulation of insulin secretion. This gene consists of 1 exon and spans about 2.8 kb of genomic distance that has been mapped to 12p13.32. Mutations in *KCNA5* gene are associated with familial atrial fibrillation type 7 and pulmonary arterial hypertension (PAH). *KCNA5* 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 *KCNA5* mutations. Individuals are tested by DNA sequencing of the coding exons of the *KCNA5* 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 pulmonary arterial hypertension (PAH) and familial atrial fibrillation type 7.

### METHODOLOGY

Genomic DNA is analyzed for *KCNA5* mutations by DNA sequencing of the coding exons of the *KCNA5* 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>
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<tr>
<th>Category</th>
<th>Direct and Institutional Billing</th>
<th>CPT Codes</th>
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<td>Index Case (Male or Female)</td>
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<tr>
<td>Additional Family Members</td>
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<td></td>
<td>Known familial mutation only</td>
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### SENSITIVITY

DNA Sequencing Analysis: Approximately 99 percent detection of mutations in the coding exon 1 of *KCNA5*.

### 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