The type I cGMP-dependent protein kinase gene consists of 18 exons spanning more than 220-kb genomic distance on chromosome 10 at 10q11.2-q21.1. The first 2 exons are used alternatively and encode the soluble alpha isoform and beta isoform. PRKG1 is highly expressed in smooth muscle cells and plays a central role in regulating cardiovascular and neuronal functions in addition to relaxing smooth muscle tone, preventing platelet aggregation, and modulating cell growth. Mutations in PRKG1 have been identified in patients with aortic aneurysm and dissection. Definitive genotype/phenotype correlations have not been described.

The John Welsh Cardiovascular Diagnostic Laboratory offers molecular genetic testing for PRKG1 mutations. Symptomatic individuals will be tested by DNA sequencing of all 18 exons of the PRKG1 gene. Genetic counseling is recommended for all individuals and their families.

**REASON FOR REFERRAL**

- Molecular confirmation of the diagnosis of aortic aneurysm and dissection.

**METHODOLOGY**

Genomic DNA will be analyzed for PRKG1 mutations by DNA sequencing of all 18 exons of the PRKG1 gene, as well as the exon/intron junctions and a portion of the 5′ and 3′ untranslated regions. Patient DNA will be 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>
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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>$1300 per sample</td>
<td>81406</td>
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<tr>
<td>Additional Family Members</td>
<td>$300 per sample; Known familial mutations only</td>
<td>81403</td>
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</table>

**SENSITIVITY**

DNA Sequencing Analysis: Approximately 99% detection of mutations in the 18 exons of PRKG1

**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

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