The *GDF2* gene encodes bone morphogenetic protein 9 (BMP9), a member of the highly conserved transforming growth factor-beta (TGFβ). The *GDF2* gene contains 2 exons and spans around 3.8 kb genomic distance that was mapped to chromosome 10q11.22. Defects in this gene are the cause of hereditary hemorrhagic telangiectasia (HHT). HHT is an autosomal-dominantly inherited vascular-malformation syndrome characterized by telangiectases and arteriovenous malformations (AVMs). *GDF2* gene is also associated with pulmonary artery hypertension (PAH), which is a devastating disease characterized by elevated pulmonary artery pressure. Definitive genotype/phenotype correlations have not been described.

The John Welsh Cardiovascular Diagnostic Laboratory offers molecular genetic testing for *GDF2* mutations. Individuals are tested by DNA sequencing of the coding exons of the *GDF2* 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 hereditary hemorrhagic telangiectasia (HHT) and pulmonary artery hypertension (PAH).

**METHODOLOGY**

Genomic DNA is analyzed for *GDF2* mutations by DNA sequencing of the coding exons of the *GDF2* 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>$500 per sample</td>
<td>81404</td>
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</tbody>
</table>

| Additional Family Members        | $300 per sample; Known familial mutation only |

| CPT Codes | 81403 |

**SENSITIVITY**

DNA Sequencing Analysis: Approximately 99 percent detection of mutations in the coding exons of *GDF2*.

**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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