**APOA2 MUTATION ANALYSIS**  
**JOHN WELSH CARDIOVASCULAR DIAGNOSTIC LABORATORY**

**APOA2** gene encodes apolipoprotein (apo-) A-II, which is the second most abundant protein of the high density lipoprotein particles. The human gene encodes a product containing 100 amino acids and encompasses 3 exons. It is located at 1q23.3. The APOA2 protein is found in plasma as a monomer, homodimer, or heterodimer with apolipoprotein D. Defects in this gene may result in apolipoprotein A-II deficiency or hypercholesterolemia. APOA2 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 APOA2 mutations. Individuals are tested by DNA sequencing of the coding exons of the APOA2 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 apolipoprotein A-II deficiency and hypercholesterolemia.

**METHODOLOGY**
Genomic DNA is analyzed for APOA2 mutations by DNA sequencing of the coding exons of the APOA2 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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<tr>
<th>Direct and Institutional Billing</th>
<th>CPT Codes</th>
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<tbody>
<tr>
<td><strong>Index Case (Male or Female)</strong></td>
<td>$500 per sample</td>
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<tr>
<td><strong>Additional Family Members</strong></td>
<td>$300 per sample; Known familial mutation only</td>
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**SENSITIVITY**
DNA Sequencing Analysis: Approximately 99 percent detection of mutations in the coding exons 1-3 of APOA2.

**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