The RPSA gene encodes laminin receptor which is a cell-surface receptor with high affinity for its primary ligand. Laminin receptor is an important molecule both in cell adhesion to the basement membrane and in signalling transduction following this binding event. The protein also plays critical roles in the metastasis of tumour cells. The RPSA gene contains 7 exons that were mapped to chromosome 3p21.3. Mutations in this gene are associated congenital asplenia and arrhythmogenic right ventricular dysplasia (ARVD). Definitive genotype/phenotype correlations have not been described.

The John Welsh Cardiovascular Diagnostic Laboratory offers molecular genetic testing for RPSA mutations. Individuals are tested by DNA sequencing of the coding exons of the RPSA 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.

**METHODOLOGY**

Genomic DNA is analyzed for RPSA mutations by DNA sequencing of the coding exons of the RPSA 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></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>$600 per sample</td>
<td>81405</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 7 coding exons of RPSA.

**SPECIMEN REQUIREMENTS**

**Blood (preferred):** EDTA (purple-top) tubes: **Adult:** 5 cc  **Child:** 5 cc  **Infant:** 2-3 cc  
**Tissue:** Frozen (preferred), or RNAlater  
**Other Body Fluids or Formalin-fixed, Paraffin-embedded tissue:** Call to inquire