The **RBM20** gene encodes RNA-binding motif protein 20 (RBM20) which contains an RNA recognition motif-1 (RRM1), an arginine-serine (RS)-rich domain, and a U1-type zinc finger domain. The combination of RRM1 and RS domain is characteristic of spliceosomal proteins. The **RBM20** gene contains 14 exons that was mapped to chromosome 10q25.2. Defects in this gene are the cause of dilated cardiomyopathy 1DD. DCM is a type of cardiomyopathy in which the heart becomes weakened and enlarged and cannot pump blood efficiently. About 25–35% of patients have familial forms of the disease. The disease is genetically heterogeneous, but the most common form of its transmission is autosomal dominant. Definitive genotype/phenotype correlations have not been described.

The John Welsh Cardiovascular Diagnostic Laboratory offers molecular genetic testing for **RBM20** mutations. Individuals are tested by DNA sequencing of the coding exons of the **RBM20** 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 dilated cardiomyopathy 1DD.

**METHODOLOGY**

Genomic DNA is analyzed for **RBM20** mutations by DNA sequencing of the coding exons of the **RBM20** 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>$1100 per sample</td>
<td>81406</td>
</tr>
<tr>
<td>Additional Family Members</td>
<td>$300 per sample; Known familial mutation only</td>
<td>81403</td>
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</tbody>
</table>

**SENSITIVITY**

DNA Sequencing Analysis: Approximately 99 percent detection of mutations in the coding exons 1-14 of **RBM20**.

**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