ANKRD1 is located on chromosome 10q23.31. It encodes 9 exons. ANKRD1 belongs to the conserved muscle ankyrin repeat protein (MARP) family. It is localized to the nucleus of endothelial cells and is induced by IL-1 and TNF-alpha stimulation. Studies in rat cardiomyocytes suggest that this gene functions as a transcription factor. Interactions between this protein and the sarcomeric proteins myopalladin and titin suggest that it may also be involved in the myofibrillar stretch-sensor system. ANKRD1 mutations are passed on in an autosomal dominant fashion. Diseases associated with ANKRD1 include ankrd1-related dilated cardiomyopathy, and diastolic heart failure. Definitive genotype/phenotype correlations have not been described.

The John Welsh Cardiovascular Diagnostic Laboratory offers molecular genetic testing for ANKRD1 mutations. Individuals are tested by DNA sequencing of the coding exons of the ANKRD1 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.

Molecular confirmation of the diagnosis of dilated cardiomyopathy and diastolic heart failure.

Genomic DNA is analyzed for ANKRD1 mutations by DNA sequencing of the coding exons of the ANKRD1 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.

DNA Sequencing Analysis: Approximately 99 percent detection of mutations in the coding exons 1-9 of ANKRD1.

**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

**SERVICE FEES**

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<tr>
<th>Description</th>
<th>Direct and Institutional Billing</th>
<th>CPT Codes</th>
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<tr>
<td>Index Case (Male or Female)</td>
<td>$800 per sample</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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**REVIEWS**

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