The CTNNA3 gene is located at 10q21.3, contains 17 exons and spans about 1,776 kb. It encodes a protein that belongs to the vinculin/alpha-catenin family. Ctnna3 plays a role in cell-cell adhesion in muscle cells and may be involved in formation of stretch-resistant cell-cell adhesion complexes. Mutations in this gene are associated with familial isolated arrhythmogenic ventricular dysplasia, biventricular form. These mutations are passed on in an autosomal dominant fashion. Definitive genotype/phenotype correlations have not been described.

The John Welsh Cardiovascular Diagnostic Laboratory offers molecular genetic testing for CTNNA3 mutations. Individuals are tested by DNA sequencing of the coding exons of the CTNNA3 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 familial isolated arrhythmogenic ventricular dysplasia, biventricular form.

Genomic DNA is analyzed for CTNNA3 mutations by DNA sequencing of the coding exons of the CTNNA3 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 1-17 of CTNNA3.

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