Arrhythmogenic Right Ventricular Dysplasia/Cardiomyopathy: Deletion/Duplication Panel

**Test Code:** MD096  
**Turnaround time:** 2 weeks  
**CPT Codes:** 81228 x1, 81479 x1

### Condition Description

Arrhythmogenic right ventricular dysplasia/cardiomyopathy (ARVD/C) is an autosomal dominant condition characterized by abnormalities in cardiac structure and rhythm. The fibrofatty replacement of myocardium can predispose affected individuals to ventricular tachycardia and sudden death in young individuals and athletes. Common presenting features include heart palpitation, syncope, and death. Other diagnostic criteria include right ventricular dilation and reduction of right ventricular function, and right ventricular aneurysms. The phenotype of ARVD/C is highly variable and while it primarily affects the right ventricle, it may involve the left ventricle as well.

Reference:
- GeneReviews

### Genes

DSC2, DSG2, DSP, JUP, PKP2, RYR2, TMEM43

### Indications

This test is indicated for:
- Confirmation of a clinical diagnosis of arrhythmogenic right ventricular dysplasia/cardiomyopathy (ARVD/C).
- Carrier testing in adults with a family history of arrhythmogenic right ventricular dysplasia/cardiomyopathy (ARVD/C).

### Methodology

**Deletion/Duplication Analysis:** DNA isolated from peripheral blood is hybridized to a gene-targeted CGH array to detect deletions and duplications. The targeted CGH array has overlapping probes that cover the entire genomic region.

### Detection

**Deletion/Duplication Analysis:** Detection is limited to duplications and deletions. The CGH array will not detect point or intronic mutations. Results of molecular analysis must be interpreted in the context of the patient's clinical and/or biochemical phenotype.

### Specimen Requirements

**Submit only 1 of the following specimen types**

**Type: Whole Blood (EDTA)**

**Specimen Requirements:**
- EDTA (Purple Top)  
- Infants and Young Children (2 years of age to 10 years old): 3-5 ml  
- Older Children & Adults: 5-10 ml  
- Autopsy: 2-3 ml uncotted cord or cardiac blood

**Specimen Collection and Shipping:**
- Ship sample at room temperature for receipt at EGL within 72 hours of collection. Do not freeze.

**Type: DNA, Isolated**

**Specimen Requirements:**
- Microtainer  
- 3µg  
- Isolation using the Perkin Elmer™Chemagen™ Chemagen™ Automated Extraction method or Qiagen™ Puregene kit for DNA extraction is recommended.

**Specimen Collection and Shipping:**
- Refrigerate until time of shipment in 100 ng/µL in TE buffer. Ship sample at room temperature with overnight delivery.

### Related Tests

- Comprehensive Cardiomyopathy Panel.
- Arrhythmogenic Right Ventricular Dysplasia/Cardiomyopathy: Sequencing Panel.