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/cardio myopathy (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/cardio myopathy (ARVD/C).
- Carrier testing in adults with a family history of arrhythmogenic right ventricular dysplasia/cardio myopathy (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. Please note that a “backbone” of probes across the entire genome are included on the array for analytical and quality control purposes. Rarely, off-target copy number variants causative of disease may be identified that may or may not be related to the patient's phenotype. Only known pathogenic off-target copy number variants will be reported. Off-target copy number variants of unknown clinical significance will not be reported.

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

Specimen Requirements:
- In EDTA (purple top) tube:
  - Infants (2 years): 3-5 ml
  - Older Children & Adults: 5-10 ml

Specimen Collection and Shipping: Ship sample at room temperature with overnight delivery.

Type: Isolated DNA

Specimen Requirements:
- In microtainer: 10 ug

Isolation using the Qiagen™ Puregene kit for DNA extraction is recommended.

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

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Related Tests

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