

## Using Genetic Testing To Diagnose Arrhythmogenic Right Ventricular Dysplasia/Cardiomyopathy (ARVD/C)

### Indications:

- Clinical diagnosis of Arrhythmogenic Right Ventricular Dysplasia/Cardiomyopathy (ARVD/C)
- Unexplained cardiovascular symptoms such as palpitations and/or history of syncopal episodes in young adults, especially athletes
- Family history of ARVD/C
- Family history of sudden cardiac death in individuals under age 45

### Benefits:

Genetic testing for ARVD/C can:

- confirm a clinical diagnosis of ARVD/C.
- identify at-risk family members who should undergo regular cardiac screening for ARVD/C.
- distinguish between different forms of ARVD/C including more highly penetrant forms and those with pronounced left ventricular involvement
- help to identify candidates for implantable cardioverter defibrillator (ICD) intervention.

### Background:

- Arrhythmogenic Right Ventricular Dysplasia/Cardiomyopathy (ARVD/C) is characterized clinically by right ventricular (RV) arrhythmia and histologically by replacement of normal myocardial tissue in the RV by fibrotic adipose tissue.
- ARVD/C occurs at a prevalence of as high as 1 in 1000 (0.1%) individuals and typically shows a dominant mode of inheritance.<sup>1,2</sup>
- ARVD/C is the second leading cause of SCD in young adults, including competitive athletes.<sup>3,4</sup>
- Extensive cardiac screening at regular intervals can identify patients at high risk for SCD, who may benefit from ICD implantation.<sup>5</sup>
- About 50% of ARVD/C cases are familial and are associated with mutations in any one of at least 8 different genes.<sup>2,6</sup>
- Most familial ARVD/C cases are due to mutations in genes encoding components of the cardiac desmosome, a complex of proteins forming intercellular junctions in the myocardium.<sup>7,8</sup>

**References:** 1. Peters S, et al. (2004) *Int Journal Cardiology* 97:499-501. 2. Awad MM, et al. (2008) *Nature Clin Practice Cardio Med* 5: 258-67. 3. Thiene G, et al. (1988) *NEJM* 318: 129-33. 4. Corrado D, et al. (1998) *NEJM* 339: 364-69. 5. Corrado D, et al. (2003) *Circulation* 108: 3084-91. 6. Hamid MS, et al. (2002) *JACC* 40: 1445-50. 7. Van Tintelen JP, et al. (2007) *Curr Opin in Cardio* 22:185-92.. 8. Garrod D, et al. (2007) *BBA* 1778: 572-87.

**Ordering Information:** Please see other side.

## Ordering Information for Arrhythmogenic Right Ventricular Dysplasia/Cardiomyopathy (ARVD/C) Testing

### Indications for Testing

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### Ordering Information for Single Gene Tests

Gene(s)	CPT Codes	Test Code
<b>PKP2</b>	83891(1) 83892(1) 83898(15) 83904(30) 83909(30) 83912(1)	190701
<b>DSP</b>	83891(1) 83892(1) 83898(38) 83904(76) 83909(76) 83912(1)	190702
<b>DSC2</b>	83891(1) 83892(1) 83898(17) 83904(34) 83909(34) 83912(1)	190703
<b>DSG2</b>	83891(1) 83892(1) 83898(17) 83904(34) 83909(34) 83912(1)	190704
<b>TMEM43</b>	83891(1) 83892(1) 83898(12) 83904(24) 83909(24) 83912(1)	190705

### Ordering Information for Multi-Gene Panels\*

<b>PKP2, DSP, DSC2, DSG2, TMEM43</b>	83891(1) 83892(1) 83898(99) 83904(198) 83909(198) 83912(5)	190799
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### Family Testing (single amplicon)

<b>(applies to all genes)</b>	83891(1) 83892(1) 83898(1) 83904(2) 83909(2) 83912(1)	use single-gene test code
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### Test Methodology and Sample Requirements

- Amplification by polymerase chain reaction (PCR); sequencing of entire protein-coding region
- For blood samples:
  - 2 mL whole blood in EDTA tube (lavender top)
  - Samples can be stored briefly at 4°C, but should be shipped on day of collection.
- For buccal swab samples: (only accepted for family testing)
  - Please contact client services at 1-866-647-0735 for instructions.
- All sample types should be shipped overnight at room temperature.
- To request a sample shipping kit, please call 1-866-647-0735.

For more information, please contact Correlagen Diagnostics, Inc., at 1-866-647-0735 or visit us on the web at [www.correlagen.com](http://www.correlagen.com).