Correlagen Diagnostics, Inc.
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Molecular Diagnostics for Genes that Matter™…

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.[1,2]
- ARVD/C is the second leading cause of SCD in young adults, including competitive athletes.[3,4]
- Extensive cardiac screening at regular intervals can identify patients at high risk for SCD, who may benefit from ICD implantation.[5]
- About 50% of ARVD/C cases are familial and are associated with mutations in any one of at least 8 different genes.[2,6]
- 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. [7,8]

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

- Clinical diagnosis of Arrhythmogenic Right Ventricular Dysplasia/ Cardiomyopathy (ARVD/C)
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Ordering Information for Single Gene Tests

Gene(s)	Test Code
PKP2	190701
DSP	190702
DSC2	190703
DSG2	190704
TMEM43	190705

Ordering Information for Multi-Gene Panels

PKP2, DSP, DSC2, DSG2,TMEM43	190799

Family Testing (single amplicon)

Family Testing is available for all genes. Please contact Client Services at 1-866-647-0735 for requirements.

Test Methodology

- Amplification by polymerase chain reaction (PCR); sequencing of entire protein-coding region

NOTE: Specimens must be accompanied by a completed consent form. In the case of family tests (ie, known mutations), a copy of the result of the first patient tested in the family (the index case) must be submitted unless that test was performed at Correlagen. Other family members are subsequently tested for the specific mutation found in the first patient tested.

For test information, sample requirements, or to request a sample shipping kit, please contact Client Services at 1-866-647-0735 or visit us on the web at www.correlagen.com.