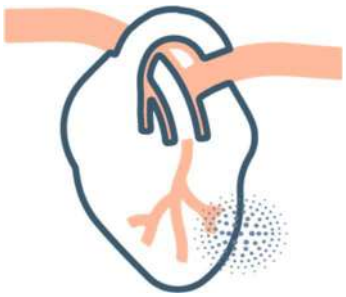
Arrhythmogenic Right Ventricular Dysplasia

Precision Panel



Overview

Arrhythmogenic Right Ventricular Dysplasia (ARVD) is an inherited cardiomyopathy characterized by structural and functional abnormalities in the right ventricle, resulting in ventricular arrhythmias. It is a rare but important cause of sudden arrhythmic death in young and healthy personas, as well as a subtle cause of congestive heart failure, which can lead to temporary incapacitation with severe consequences. Structurally, although less prominent than right ventricle, left ventricle can be also affected by myocyte loss and fibrosis, increasing the likelihood of this condition with age. This fact suggests that ARVD is a progressive disease. Affected individuals do not have evidence of the disease at birth, and typically the disease starts to manifest clinically at approximately 12 or 13 years of age, with the mean age at diagnosis in the early 30s. The mode of inheritance is mainly autosomal dominant.

The Igenomix Arrhythmogenic Right Ventricular Dysplasia Precision Panel can be used to make an accurate and directed diagnosis as well as a differential diagnosis of cardiomyopathy, ultimately leading to a better management and prognosis of the disease. It provides a comprehensive analysis of the genes involved in this disease using next-generation sequencing (NGS) to fully understand the spectrum of relevant genes involved.

Indications

The Igenomix Arrhythmogenic Right Ventricular Dysplasia Precision Panel is indicated for those patients with a clinical diagnosis or suspicion presenting with or without the following manifestations:

- Palpitations
- Arrhythmias during exercise
- Syncope
- Atypical chest pain
- Dyspnea
- Right Ventricular Failure

Clinical Utility

The clinical utility of this panel is:

- The genetic and molecular confirmation for an accurate clinical diagnosis of a symptomatic patient.
- Early initiation of multidisciplinary treatment to reduce mortality, prevent disease progression, improve symptoms and quality of life, limit heart failure symptoms and improve functional capacity. Pharmacological treatment includes Beta-blocker therapy and antiarrhythmic agents such as flecainide, propafenone, sotalol and amiodarone, to prevent ventricular tachycardia. In most severe cases, an implantable cardioverter-defibrillator or even a heart transplantation might be needed. Lifestyle changes will always help getting a better prognosis.
- Risk assessment and genetic counselling of asymptomatic family members according to the mode of inheritance.
- Improvement of delineation of genotype-phenotype correlation.

Genes & Diseases

Gene	OMIM Diseases	Inheritance*	% Gene Coverage (20x)	ClinVar**	HGMD**
BAG3	Cardiomyopathy	AD	100	49 of 49	83 of 85
CDH2	Agenesis Of Corpus Callosum, Arrhythmogenic Right Ventricular Dysplasia	AD	99.98	9 of 9	16 of 16
CTNNA3	Arrhythmogenic Right Ventricular Dysplasia	AD	99.97	2 of 2	14 of 17
DES	Cardiomyopathy, Scapuloperoneal Syndrome, Desminopathy	AD,AR	99.97	66 of 66	133 of 134
DSC2	Arrhythmogenic Right Ventricular Dysplasia	AD,AR	100	31 of 31	123 of 124
DSG2	Arrhythmogenic Right Ventricular Dysplasia, Cardiomyopathy	AD	99.38	51 of 52	167 of 169
DSP	Arrhythmogenic Right Ventricular Dysplasia, Cardiomyopathy, Keratosis Palmoplantaris Striata, Skin Fragility-Woolly Hair Syndrome, Carvajal Syndrome, Pulmonary Fibrosis	AD,AR	99.91	221 of 222	366 of 369
FLNC	Cardiomyopathy, Filaminopathy, Myopathy	AD	100	102 of 102	185 of 186
JUP	Arrhythmogenic Right Ventricular Dysplasia, Naxos Disease, Lethal Acantholytic Epidermolysis Bullosa	AD,AR	100	12 of 12	56 of 56
LDB3	Cardiomyopathy, Left Ventricular Noncompaction	AD	100	5 of 5	60 of 60
LMNA	Cardiomyopathy, Charcot-Marie-Tooth Disease, Emery-Dreifuss Muscular Dystrophy, Heart-Hand Syndrome, Hutchinson-Gilford Progeria Syndrome, Malouf Syndrome, Werner Syndrome, Lipodystrophic Laminopathy, Hypergonadotropic Hypogonadism	AD,AR	100	287 of 287	619 of 620
MYH7	Cardiomyopathy, Ebstein Malformation, Scapuloperoneal Muscular Dystrophy	AD,AR	99.95	322 of 322	1053 of 1054
NKX2-5	Atrial Septal Defect, Conotruncal Heart Malformations, Hypoplastic Left Heart Syndrome, Hypothyroidism, Tetralogy Of Fallot, Ventricular Septal Defect, Athyreosis, Bicuspid Aortic Valve, Cardiac Conduction Defect, Thyroid Ectopia	AD,AR	99.98	45 of 45	112 of 116
PKP2	Arrhythmogenic Right Ventricular Dysplasia, Brugada Syndrome	AD	100	151 of 151	306 of 307
PLN	Cardiomyopathy	AD	100	3 of 3	26 of 33

RYR2	Arrhythmogenic Right Ventricular Dysplasia, Ventricular Tachycardia, Atrial Dysfunction, Dilated Cardiomyopathy, Catecholaminergic Polymorphic Ventricular Tachycardia	AD	99.2	126 of 129	466 of 472
SCN5A	Atrial And Ventricular Fibrillation, Brugada Syndrome, Cardiomyopathy, Long Qt Syndrome, Progressive Familial Heart Block, Sick Sinus Syndrome, Sudden Infant Death Syndrome, Progressive Cardiac Conduction Defect, Romano-Ward Syndrome	AD,AR,MU	99.45	245 of 246	929 of 942
TGFB3	Arrhythmogenic Right Ventricular Dysplasia, Loeys-Dietz Syndrome, Thoracic Aortic Aneurysm And Aortic Dissection	AD	100	25 of 26	34 of 35
TMEM43	Arrhythmogenic Right Ventricular Dysplasia, Emery-Dreifuss Muscular Dystrophy	AD	99.98	4 of 4	26 of 26
TTN	Cardiomyopathy, Limb Girdle Muscular Dystrophy	AD,AR	97.93	1033 of 1081	1153 of 1219

*Inheritance: AD: Autosomal Dominant; AR: Autosomal Recessive; X: X linked; XLR: X linked Recessive; Mi: Mitochondrial; Mu: Multifactorial.; G: Gonosomal Inheritance; D: Digenic Inheritance
**Number of clinically relevant mutations according to ClinVar and HGMD

Methodology

Call +34 963 905 310 or send an email to supportspain@igenomix.com for any of the following objectives:

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