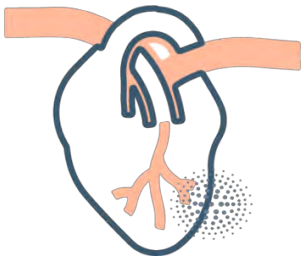
Arrythmias Precision Panel



Overview

Arrythmias are a group of conditions in which there is an alteration in the rate or rhythm of the heart. The heart can beat too fast, too slow or with an irregular rhythm. Arrythmias are caused by changes in heart tissue and activity or in the electrical signals that control your heartbeat. These changes can be caused by damage from disease, injury or genetics including cardiac channelopathies. Cardiac channelopathies are a group of inherited conditions that are associated with a defect in the cardiac ion channel function. These problems cause an increased susceptibility to abnormal heart rhythm (dysrhythmia or arrythmia), most often ventricular tachycardia or ventricular fibrillation that ultimately can lead to sudden cardiac death (SCD). The differential diagnosis between ion channel disease and cardiomyopathies can be challenging on occasion as severe ventricular dysrhythmias can manifest in patients with cardiopathies or with structurally normal hearts.

The Igenomix Arrythmia Precision Panel serves as a diagnostic and screening tool 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.

Indications

The Igenomix Arrythmia Precision Panel is indicated in those cases where there is a clinical suspicion of arrythmia with the following manifestations:

- Shortness of breath
- Palpitations
- Sweating
- Dizziness
- Fainting or nearly fainting
- Fluttering of the chest
- Chest pain
- Light-headedness
- Sudden weakness
- Blurry vision

Clinical Utility

The clinical utility of this panel is:

- The genetic and molecular diagnosis for an accurate clinical diagnosis.
- Early initiation of treatment with a multidisciplinary team for appropriate preventive ICD placement, pacemaker, pharmacologic therapy, or interventional procedures.
- Prevent complications including stroke, heart failure or sudden cardiac death.
- Risk assessment and genetic counselling of asymptomatic family members according to the mode of inheritance.

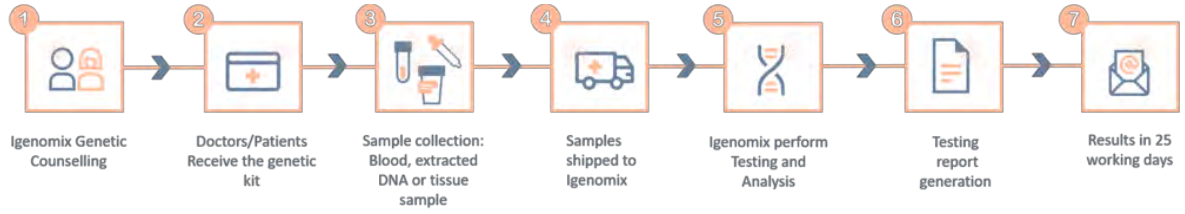
Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
ABCC9	Familial Atrial Fibrillation, Dilated Cardiomyopathy, Brugada Syndrome	AD	100	51 of 51
ACTN2	Dilated Cardiomyopathy With Or Without Left Ventricular Noncompaction, Congenital Myopathy With Structured Cores And Z-line Abnormalities	AD	100	56 of 56
AKAP9	Long QT Syndrome, Brugada Syndrome , Romano-Ward Syndrome	AD	98.34	43 of 46
ANK2	Cardiac Arrhythmia, Ankyrin-B-Related, Romano-Ward Syndrome	AD	99.98	130 of 130
ASPH	Ectopia Lentis, Spontaneous Filtering Blebs, And Craniofacial Dysmorphism	AR	98.45	7 of 8
BAG3	Dilated Cardiomyopathy, Myofibrillar Myopathy	AD	100	83 of 85
CACNA1C	Brugada Syndrome, Timothy Syndrome, Romano-Ward Syndrome	AD	99.8	85 of 85
CACNA1D	Sinoatrial Node Dysfunction And Deafness	AD,AR	100	18 of 18
CACNA2D1	Brugada Syndrome, Familial Short QT Syndrome	-	99.96	12 of 12
CACNB2	Brugada Syndrome	AD	99.84	32 of 34
CALM1	Long QT Syndrome, Catecholaminergic Polymorphic Ventricular Tachycardia, Romano-Ward Syndrome	AD	100	12 of 12
CALM2	Long QT Syndrome, Catecholaminergic Polymorphic Ventricular Tachycardia, Romano-Ward Syndrome	AD	98.71	11 of 11
CALM3	Long QT Syndrome, Catecholaminergic Polymorphic Ventricular Tachycardia, Romano-Ward Syndrome	AD	100	5 of 5
CASQ2	Catecholaminergic Polymorphic Ventricular Tachycardia, With Or Without Atrial Dysfunction And/Or Dilated Cardyomyopathy	AD,AR	100	39 of 40
CAV3	Familial Hypertrophic Cardiomyopathy, Long QT Syndrome, Romano-Ward Syndrome	AD	100	50 of 50
CDH2	Famililal Arrhythmogenic Right Ventricular Dysplasia	AD	99.98	16 of 16
DES	Dilated Cardiomyopathy, Myofibrillar Myopathy	AD,AR	99.97	133 of 134
DPP6	Paroxysmal Familial Ventricular Fibrillation	AD	97.03	23 of 28
DSC2	Familial Arrhythmogenic Right Ventricular Dysplasia	AD,AR	100	123 of 124
DSG2	Familial Arrhythmogenic Right Ventricular Dysplasia, Dilated Cardiomyopathy	AD	99.38	167 of 169
DSP	Familial Arrhythmogenic Right Ventricular Dysplasia, Dilated Cardiomyopathy	AD,AR	99.91	366 of 369
EMD	X-linked Emery-Dreifuss Muscular Dystrophy	X,XR,G	99.92	NA of NA
FLNC	Familial Hypertrophic Cardiomyopathy, Autosomal Dominant Filaminopathy, Familial Isolated Restrictive Cardiomyopathy	AD	100	185 of 186
GATA4	Atrioventricular Septal Defect, Testicular Anomalies With Or Without Congenital Heart Disease, Tetralogy Of Fallot, Ventricular Septal Defect, Atrial Septal Defect	AD	94.69	108 of 130
GATA5	Congenital Heart Defects, Familial Bicuspid Aortic Valve, Tetralogy Of Fallot	AD,AR	87.02	26 of 32
GJA5	Familial Atrial Fibrillation, Tetralogy Of Fallot	AD	99.88	13 of 13
GPD1L	Brugada Syndrome	AD	100	14 of 14
HCN4	Brugada Syndrome, Sick Sinus Syndrome	AD	98.01	40 of 41
JUP	Familial Arrhythmogenic Right Ventricular Dysplasia	AD,AR	100	56 of 56
KCNA5	Familial Atrial Fibrillation	AD	99.99	33 of 33
KCND3	Brugada Syndrome	AD	100	32 of 32

KCNE1	Jervell And Lange-Nielsen Syndrome, Long QT Syndrome, Romano-Ward Syndrome	AD,AR	100	53 of 53
KCNE2	Familial Atrial Fibrillation, Long QT Syndrome, Romano-Ward Syndrome	AD	100	23 of 24
KCNE3	Brugada Syndrome	AD	100	7 of 7
KCNE5	Brugada Syndrome	-	99.66	NA of NA
KCNH2	Long QT Syndrome, Short QT Syndrome, Familial Short Qt Syndrome , Romano-Ward Syndrome	AD	98.69	908 of 930
KCNJ2	Andersen Cardiodysrhythmic Periodic Paralysis, Familial Atrial Fibrillation, Familial Short Qt Syndrome	AD	100	93 of 93
KCNJ5	Long Qt Syndrome, Romano-Ward Syndrome	AD	99.52	21 of 21
KCNJ8	Brugada Syndrome	-	100	8 of 8
KCNQ1	Familial Atrial Fibrillation, Jervell And Lange-Nielsen Syndrome, Long QT Syndrome, Short QT Syndrome, Romano-Ward Syndrome	AD,AR	93.23	600 of 624
LAMP2	Danon Disease, Glycogen Storage Disease Due To LAMP-2 Deficiency	X,XD,G	99.96	NA of NA
LMNA	Dilated Cardiomyopathy, Emery-Dreifuss Muscular Dystrophy, Heart-hand Syndrome	AD,AR	100	619 of 620
MYL4	Familial Atrial Fibrillation	AD	100	2 of 2
NKX2-5	Atrial Septal Defect With Or Without Atrioventricular Conductiondefects, Conotruncal Heart Malformations, Truncus Arteriosus Communis, Hypoplastic Left Heart Syndrome, Congenital Hypothyroidism, Tetralogy Of Fallot, Ventricular Septal Defect, Atrial Septal Defect, Familial Bicuspid Aortic Valve, Familial Progressive Cardiac Conduction Defect, Tetralogy Of Fallot	AD,AR	99.98	112 of 116
NPPA	Familial Atrial Fibrillation	AD,AR	99.61	7 of 8
PKP2	Familial Arrhythmogenic Right Ventricular Dysplasia, Brugada Syndrome	AD	100	306 of 307
PLN	Dilated Cardiomyopathy, Familial Hypertrophic Cardiomyopathy	AD	100	26 of 33
PPA2	Alcohol-Induced Sudden Cardiac Failure, Infantile Sudden Cardiac Failure	AR	99.95	9 of 9
PRKAG2	Familial Hypertrophic Cardiomyopathy, Lethal Congenital Glycogen Storage Disease Of Heart, Wolff-Parkinson-White Syndrome	AD	99.98	61 of 61
RANGRF	Brugada Syndrome	-	100	5 of 5
RBM20	Dilated Cardiomyopathy	AD	96.83	73 of 75
RYR2	Familial Arrhythmogenic Right Ventricular Dysplasia, Catecholaminergic Polymorphic Ventricular Tachycardia	AD	99.2	466 of 472
SCN10A	Brugada Syndrome, Romano-Ward Syndrome	AD	99.89	96 of 96
SCN1B	Familial Atrial Fibrillation, Brugada Syndrome, Familial Progressive Cardiac Conduction Defect	AD,AR	99.67	46 of 48
SCN2B	Familial Atrial Fibrillation, Brugada Syndrome	AD	100	8 of 8
SCN3B	Brugada Syndrome	AD	100	7 of 7
SCN4B	Long QT Syndrome, Romano-Ward Syndrome	AD	100	11 of 11
SCN5A	Familial Atrial Fibrillation, Brugada Syndrome, Dilated Cardiomyopathy, Long QT Syndrome, Progressive Familial Heart Block Type 1A, Sick Sinus Syndrome, Sudden Infant Death Syndrome, Ventricular Fibrillation During Myocardial Infarction, Familial Progressive Cardiac Conduction Defect , Romano-Ward Syndrome	AD,AR,MU	99.45	929 of 942
SLMAP	Brugada Syndrome		99.8	4 of 4
SNTA1	Long QT Syndrome, Romano-Ward Syndrome	AD	95.66	18 of 18
TMEM43	Familial Arrhythmogenic Right Ventricular Dysplasia, Emery-Dreifuss Muscular Dystrophy	AD	99.98	26 of 26
TNNI3	Dilated Cardiomyopathy, Familial Hypertrophic Cardiomyopathy, Familial Restrictive Cardiomyopathy	AD,AR	100	139 of 139
TNNT2	Dilate Cardiomyopathy, Familial Hypertrophic Cardiomyopathy, Familial Restrictive Cardiomyopathy	AD	100	169 of 169
TRDN	Catecholaminergic Polymorphic Ventricular Tachycardia With Or Without Atrial Dysfunction And/Or Dilated Cardiomyopathy, Romano-Ward Syndrome	AD,AR	98.72	10 of 12
TRPM4	Progressive Familial Heart Block, Type IB, Brugada Syndrome, Familial Progressive Cardiac Conduction Defect	AD	99.98	44 of 44
TTN	Dilated Cardiomyopathy, Familial Hypertrophic Cardiomyopathy, Early-Onset Myopathy With Fatal Cardiomyopathy , Myofibrillar Myopathy	AD,AR	97.93	1153 of 1219

* Inheritance: AD: Autosomal Dominant; AR: Autosomal Recessive; X: X linked; XLR: X linked Recessive; Mi: Mitochondrial; Mu: Multifactorial
** HGMD: Number of clinically relevant mutations according to HGMD

Methodology



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

- Get more information about the test.
- Request your kit.
- Request a pick up of the kit after collecting the sample.

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