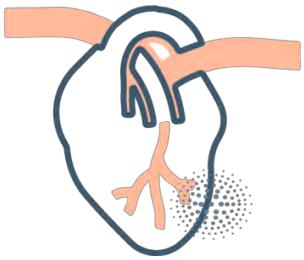
Sudden Cardiac Death Precision Panel



Overview

Sudden Cardiac Death (SCD) is an unexpected death due to cardiac causes that takes place in a short time period in a person with known or unknown underlying cardiac disease. It is an important public-health problem with multiple etiologies, risk factors and changing temporal trends. The overwhelming sadness suffered by families is heightened by the risk many of these deaths confer upon surviving relatives. For those with known cardiac disease, disease-specific therapy and risk stratification are key to reducing sudden cardiac death. Uncovering a definitive cause of death can help relieve the uncertainty as a first step in screening surviving relatives. Increasing knowledge about the molecular mechanisms and genetic drivers of malignant arrythmias have become a key component in achieving a risk stratification system to optimize and personalize patient care.

The Igenomix Sudden Cardiac Death Precision Panel serves as a diagnostic and screening tool ultimately leading to a preventive approach of the disease by risk stratification. 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 Sudden Cardiac Death Precision Panel is indicated in those cases where there are a series of risk factors that could precipitate SCD including:

- Family history of premature coronary artery disease
- Family history of cardiomyopathy
- Family history of malignant arrythmia
- Family history of SCD
- Personal history of cardiovascular risk factors
    - Smoking
    - Dyslipidemia
    - Hyeprtension
    - Diabetes
    - Obesity
    - Sedentary lifestyle

Clinical Utility

The clinical utility of this panel is:

- The genetic and molecular diagnosis for an accurate clinical diagnosis.
- Early initiation of treatment in case risk factors are present (smoking, diabetes, dyslipidemia, hyerptension etc)
- Early prevention with a multidisciplinary team in the form of preventive ICD placement, pacemaker, pharmacologic therapy, or interventional procedures.
- 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**
ABCA12	Ichthyosis Congenita	AR	100	151 of 153
ABCC6	Generalized Arterial Calcification Of Infancy	AD,AR	99	346 of 349
ABCG5	Homozygous Familial Hypercholesterolemia		99.81	57 of 57
ABCG8	Homozygous Familial Hypercholesterolemia	AR,MU,P	100	64 of 64
ACAD9	Familial Acyl-CoA Dehydrogenase Deficiency	AR	100	62 of 62
ACADL	Long Chain Acyl-CoA Dehydrogenase Deficiency		100	1 of 1
ACADVL	Acyl-CoA Dehydrogenase Deficiency, Very Long Chain Acyl-CoA Dehydrogenase Deficiency	AR	100	329 of 329
AKAP10	Cardiac Conduction Defect, Sudden Cardiac Death	AD	92.93	1 of 1
AKAP9	Long Qt Syndrome, Brugada Syndrome, Romano-Ward Syndrome	AD	98.34	43 of 46
AKT1	Proteus Syndrome, Cowden Syndrome, Meningioma	AD	100	6 of 6
ALG10B	Long Qt Syndrome	AD	99.97	2 of 2
ANK2	Cardiac Arrhythmia, Ankyrin-b-Related, Romano-ward Syndrome	AD	99.98	130 of 130
APOB	Familial Hypobetalipoproteinemia, Homozygous Familial Hypercholesterolemia	AD,AR	99.62	369 of 375
BAZ1B	Williams Syndrome		99.05	5 of 5
CACNA1C	Brugada Syndrome, Timothy Syndrome, Romano-ward Syndrome	AD	99.8	85 of 85
CACNA2D1	Brugada Syndrome, Familial Short Qt Syndrome		99.96	12 of 12
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 Cardiomyopathy	AD,AR	100	39 of 40
CAV3	Familial Hypertrophic Cardiomyopathy, Long Qt Syndrome, Rippling Muscle Disease, Romano-Ward Syndrome	AD	100	50 of 50
CLCF1	Cold-Induced Sweating Syndrome, Crisponi Syndrome	AR	100	4 of 4
CLIP2	Williams Syndrome		99.99	1 of 1
CPT1A	Carnitine Palmitoyltransferase I Deficiency	AR	100	50 of 50
CRLF1	Cold-Induced Sweating Syndrome, Crisponi Syndrome	AR	91.53	31 of 33
CSRP3	Dilated Cardiomyopathy, Familial Hypertrophic Cardiomyopathy	AD	100	36 of 36
DES	Dilated Cardiomyopathy, Myofibrillar Myopathy	AD,AR	99.97	133 of 134
DNAJC19	3-a Methylglutaconic Aciduria Type V, Dilated Cardiomyopathy With Ataxia	AR	100	6 of 6
DPP6	Familial Paroxismal 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, Carvajal Syndrome	AD,AR	99.91	366 of 369

DTNA	Left Ventricular Noncompaction	AD	97	10 of 10
ELN	Supravalvular Aortic Stenosis, Williams-Beuren Syndrome, Familial Thoracic Aortic Aneurysm And Aortic Dissection, Williams Syndrome	AD	99.99	95 of 96
EMD	X-linked Emery-Dreifuss Muscular Dystrophy	X,XR,G	99.92	NA of NA
ENPP1	Generalized Arterial Calcification Of Infancy, Non Insulin-Dependent Diabetes Mellitus, Obesity	AD,AR,MU,P	96.59	73 of 75
EYA4	Dilated Cardiomyopathy	AD	100	32 of 32
FHL1	X-Linked Reducing Body Myopathy, X-Linked Emery-Dreifuss Muscular Dystrophy	X,XR,XD,G	99.98	NA of NA
GNAI2	Familial Ventricular Tachycardia	AD	100	3 of 3
GPD1L	Brugada Syndrome	AD	100	14 of 14
GTF2I	Williams Syndrome		63.79	NA of NA
GTF2IRD1	Williams Syndrome		99.98	1 of 1
HLA-B	Takayasu Arteritis	MU	99.55	1 of 1
IKZF1	Common Variable Immunodeficiency	AD	99.98	43 of 43
JUP	Familial Arrhythmogenic Right Ventricular Dysplasia	AD,AR	100	56 of 56
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
KCNH2	Long QT Syndrome, Short QT Syndrome, Romano-Ward Syndrome	AD	98.69	908 of 930
KCNJ2	Andersen Cardiodysrhythmic Periodic Paralysis , Familial Atrial Fibrillation, Short QT Syndrome	AD	100	93 of 93
KCNJ5	Long QT Syndrome, Romano-Ward Syndrome	AD	99.52	21 of 21
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
LDB3	Dilated Cardiomyopathy With Or Without Left Ventricular Noncompaction Myofibrillar Myopathy, Familial Isolated Dilated Cardiomyopathy	AD	100	60 of 60
LDLR	Homozygous Familial Hypercholesterolemia	AD	99.89	1921 of 1996
LDLRAP1	Familial Hypercholesterolemia	AR	91.83	18 of 27
LIMK1	Williams Syndrome		100	2 of 2
LMNA	Dilated Cardiomyopathy, Emery-Dreifuss Muscular Dystrophy, Familial Dilated Cardiomyopathy With Conduction Defect Due To LMNA Mutation	AD,AR	100	619 of 620
LRP6	Autosomal Dominant Coronary Artery Disease	AD	100	44 of 44
MYH7	Dilated Cardiomyopathy, Left Ventricular Noncompaction, Familial Hypertrophic Cardiomyopathy, Familial Isolated Dilated Cardiomyopathy	AD,AR	99.95	1053 of 1054
MYL2	Familial Hypertrophic Cardiomyopathy	AD	100	67 of 67
MYL3	Familial Hypertrophic Cardiomyopathy	AD,AR	100	42 of 42
NOS1AP	Romano-Ward Syndrome		100	4 of 4
PCSK9	Familial Hypercholesterolemia	AD	100	96 of 98
PKP2	Familial Arrhythmogenic Right Ventricular Dysplasia, Brugada Syndrome	AD	100	306 of 307
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
PTEN	Bannayan-Riley-Ruvalcaba Syndrome, Lhermitte-Duclos Disease, Proteus Syndrome	AD	99.97	609 of 629
PTPN22	Insulin-Dependent Diabetes Mellitus, Rheumatoid Arthritis, Systemic Lupus Erythematosus, Granulomatosis With Polyangiitis , Vogt-Koyanagi-Harada Disease	AD	99.67	5 of 5
RBM20	Familial Isolated Dilated Cardiomyopathy	AD	96.83	73 of 75
RFC2	Williams Syndrome		100	3 of 3
RYR2	Familial Arrhythmogenic Right Ventricular Dysplasia, Catecholaminergic Polymorphic Ventricular Tachycardia With Or Without Atrial Dysfunction	AD	99.2	466 of 472
SCN10A	Brugada Syndrome, Romano-Ward Syndrome	AD	99.89	96 of 96
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 Ia, Sick Sinus Syndrome, Sudden Infant Death Syndrome, Ventricular Fibrillation During Myocardial Infarction, Romano-Ward Syndrome	AD,AR,MU	99.45	929 of 942
SNTA1	Long QT Syndrome, Romano-Ward Syndrome	AD	95.66	18 of 18

SYNE1	Emery-Dreifuss Muscular Dystrophy	AD,AR	99.99	193 of 193
SYNE2	Emery-Dreifuss Muscular Dystrophy	AD	99.94	12 of 12
TBL2	Williams Syndrome		96.14	NA of NA
TECRL	Catecholaminergic Polymorphic Ventricular Tachycardia	AR	99.48	4 of 4
TGFB3	Familial Arrhythmogenic Right Ventricular Dysplasia, Loeys-Dietz Syndrome, Familial Thoracic Aortic Aneurysm And Aortic Dissection	AD	100	34 of 35
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
TRDN	Catecholaminergic Polymorphic Ventricular Tachycardia With Or Without Atrial Dysfunction And/Or Dilated Cardiomyopathy	AD,AR	98.72	10 of 12
WAS	Wiskott-Aldrich Syndrome, X-linked Severe Congenital Neutropenia	X,XR,G	100	NA of NA
WIPF1	Wiskott-Aldrich Syndrome	AR	99.79	3 of 3

* 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

Contact us

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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