

CardioGeneScan

Testing for over 100 genetic causes of familial cardiac disease

Proven Relevance

Familial cardiac diseases have a combined prevalence of about 1:100 and are associated with a large percentage of sudden cardiac death in adolescents and younger adults.

Simple Indications

Symptoms of any of the following cardiac conditions, in the presence of a positive family history:

- Cardiomyopathy
- Arrhythmia
- Aortopathy
- Noonan-like disease
- Congenital heart disease
- Early-onset coronary artery disease/hypercholesterolemia



Clear Benefits

- Confirm a clinical diagnosis, inform the prognosis, guide treatment
- Identify the familial mutation, to help prevent sudden cardiac death in family members with the mutation, and to reassure family members without the mutation

Lasting Currency

- New genes will be added as their association with cardiac disease is reported
- Interpretations are updated as new information on variants becomes available

For the most current list of genes in the CGS, please visit us at www.correlagen.com.

Clear

Comprehensive

Current



For more information, please call us at 1-866-647-0735 or visit us on the web at www.correlagen.com

Six indications – available separately or combined

Familial Cardiomyopathy

>65% of Hypertrophic Cardiomyopathy	<i>ACTC1, APOA1, CAV3, CSRP3, GLA, LAMP2, MTTG, MTTI, MTTK, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, PRKAG2, TNNI3, TNNT2, TPM1, TTR</i>
>50% of Arrhythmogenic Right Ventricular Dysplasia/Cardiomyopathy	<i>DSC2, DSG2, DSP, JUP, PKP2, RYR2, TGFB3, TMEM43</i>
>40% of Dilated Cardiomyopathy	<i>ABCC9, ACTC1, ACTN2, ALMS1, APOA1, CSRP3, CTF1, DES, DNAJC19, EMD, EYA4, FKTN, HOPX, LAMP2, LDB3, LMNA, MTND1, MTND5, MTND6, MTHH, MTTK, MTTL1, MTTQ, MTTTS1, MTTTS2, MYBPC3, MYH7, PLN, SCN5A, SGCD, TAZ, TCAP, TMPO, TNNC1, TNNI3, TNNT2, TPM1, TTR, VCL</i>
>10% of other Cardiomyopathies (restrictive, left ventricular non-compaction)	<i>ACTC1, DTNA, MYH7, TAZ, TNNI3, TNNT2</i>

Familial Arrhythmia

>75% of Long QT syndrome/ Brugada syndrome	<i>AKAP9*, ANK2, ATP1B1, CACNA1C, CACNB2, CAV3, CNOT1, GINS3, GPD1L, KCNE1, KCNE2, KCNH2, KCNJ2, KCNQ1, LIG3, NOS1AP, PLN, SCN1B, SCN4B, SCN5A, SNTA1</i>
>50% of Arrhythmogenic Right Ventricular Dysplasia/Cardiomyopathy	<i>DSC2, DSG2, DSP, JUP, PKP2, RYR2, TGFB3, TMEM43</i>
>50% of Catecholaminergic Polymorphic Ventricular Tachycardia	<i>CASQ2, KCNJ2, RYR2</i>
>7% of Atrial Fibrillation	<i>KCNE2, KCNQ1, NPPA</i>

Familial Aortopathy

>90% of Marfan Syndrome, Loeys-Dietz Syndrome, vascular-type Ehlers-Danlos Syndrome	<i>COL3A1, FBN1, TGFB1, TGFB2</i>
>15% of Thoracic Aortic Aneurysms and Dissections	<i>ACTA2, MYH11, TGFB1, TGFB2</i>

Noonan Syndrome and Related Disorders

>70% of Noonan Syndrome, Cardiofaciocutaneous Syndrome, Costello Syndrome, LEOPARD syndrome	<i>BRAF, HRAS, KRAS, MAP2K1, MAP2K2, PTPN11, RAF1, SHOC2*, SOS1</i>
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Familial Congenital Heart Disease

>12% of Atrial Septal Defects	<i>GATA4, NKX2-5, TBX5</i>	>75% of CHARGE Syndrome	<i>CHD7</i>
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Early-onset Coronary Artery Disease / Familial Hypercholesterolemia

>90% of Familial Hypercholesterolemia / Coronary Artery Disease	<i>ABCA1, APOA2, APOB*, APOC3, LDLR, PCSK9, PON2</i>
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* For these genes, only a single exon is sequenced, which covers all disease-causing variants known to date.

References are available on our web site at www.correlagen.com

For pricing and ordering information please contact Correlagen Client Services at (781) 647-0604 or email testing@correlagen.com

The CardioGeneScan is enabled through use of single-molecule sequencing on a Helicos Genetic Analysis System.