

CardioGeneScan Using Genetic Testing to Diagnose Familial Cardiomyopathy

Indications:

- Clinical diagnosis of Cardiomyopathy
- Family history of Cardiomyopathy
- Family history of sudden cardiac death, especially in individuals under age 45

Benefits:

Genetic testing for Familial Cardiomyopathy can:

- Confirm a clinical diagnosis of Cardiomyopathy
- Differentiate between different forms of Cardiomyopathy
- Identify family members of an index patient who harbor the familial mutation and should undergo extensive cardiac screening at regular intervals
- Identify family members of an index patient who do not harbor the familial mutation and do not need to undergo extensive cardiac screening at regular intervals
- Facilitate accurate genetic counseling for family members
- In some cases, predict the type/severity of manifestations based on the exact mutation found

Background:

- Cardiomyopathy, with an estimated prevalence of about 1 in 1000, is generally characterized by impaired contractile function of the myocardium, putting patients at high risk of arrhythmias, embolic stroke, and sudden cardiac death (SCD).¹
- Prevention of stroke and SCD can be achieved through treatment with drugs, implantable cardioverter defibrillator therapy, and lifestyle changes.¹
- Different forms of cardiomyopathy may have different prognosis.¹
- Familial forms of cardiomyopathy account for between 30% and 100% of all cases.²⁻⁵ Different forms of cardiomyopathy are often associated with different genes or different variants within the same genes.¹
- Genetic testing can help to identify familial forms of cardiomyopathy and allow timely diagnosis and treatment in family members.²⁻⁹
- Genetic causes of cardiomyopathy tested for in this Cardiomyopathy evaluation account for about 60% of familial cardiomyopathy cases.¹⁻⁹

For References and Ordering Information: please see other side



Ordering Information for Familial Cardiomyopathy Testing

Indications for Testing

- Clinical diagnosis of Cardiomyopathy
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Ordering Information

Test Code 190860

Test Methodology

- Protein coding regions are selectively enriched by hybridization to specific nucleic-acid probes or by PCR and sequenced using next-generation sequencing technology. All variants known or predicted to be pathogenic are confirmed by uni-directional Sanger sequencing.

Sample Requirements

- For blood samples:
 - 2 mL whole blood in EDTA tube (lavender top)
 - Samples can be stored briefly at 4°C, but should be shipped on day of collection
- For DNA samples:
 - Minimum requirement of 15 µg DNA, preferably in 10 mM TE (if different, please specify)
 - DNA concentration should be clearly indicated
- All sample types should be shipped overnight at room temperature
- For all sample types, a minimum of 2 sample identifiers are required on every sample sent to Correlagen
- To request a sample shipping kit, please call 1-617-851-5798

References

1. Maron BJ, Towbin JA, Thiene G, Antzelevitch C, Corrado D, Arnett D, Moss AJ, Seidman CE, and Young JB (2006) Contemporary definitions and classification of the cardiomyopathies: an American Heart Association Scientific Statement from the Council on Clinical Cardiology, Heart Failure and Transplantation Committee; Quality of Care and Outcomes Research and Functional Genomics and Translational Biology Interdisciplinary Working Groups; and Council on Epidemiology and Prevention. *Circulation* 113:1807-1816.
2. Van Driest SL, Ommen SR, Tajik AJ, Gersh BJ, and Ackerman MJ (2005) Yield of genetic testing in hypertrophic cardiomyopathy. *Mayo Clin Proc* 80:739-744.
3. Hershberger RE, Kushner JD, and Parks SB (2009) Dilated Cardiomyopathy Overview. In GeneReviews: NIH:<http://www.ncbi.nlm.nih.gov/bookshelf/br.fcgi?book=gene&part=dcmm-ov>
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6. Cirino AL, and Ho C (2009) Familial Hypertrophic Cardiomyopathy Overview. In GeneReviews: NIH:<http://www.ncbi.nlm.nih.gov/bookshelf/br.fcgi?book=gene&part=hyper-card>
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8. Mogensen J, and Arbustini E (2009) Restrictive cardiomyopathy. *Curr Opin Cardiol* 24:214-220.
9. Sekijima Y, Yoshida K, Tokuda T, and Ikeda S (2009) Familial Transthyretin Amyloidosis. In GeneReviews: NIH:<http://www.ncbi.nlm.nih.gov/bookshelf/br.fcgi?book=gene&part=tfap>

**For more information, please contact Correlagen Diagnostics, Inc., at 1-617-577-0152
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