

Dilated Cardiomyopathy: Sequencing Panel

Test Code: MM092

Turnaround time: 6 weeks

CPT Codes: 81406 x1, 81407 x1, 81404 x1, 81405 x1

Condition Description

Hereditary dilated cardiomyopathy (DCM) may be inherited in an autosomal dominant, autosomal recessive, or X-linked manner, depending on the gene involved. DCM is characterized by left ventricular enlargement and reduced myocardial contraction force. Typically, DCM presents with one of three features: heart failure, thromboembolic disease, or arrhythmias and/or conduction system disease. Approximately 20-50% of idiopathic dilated cardiomyopathy (those cases not due to acquired causes) are thought to have a genetic cause.

Note: This test does not detect the retrotransposon insertion in the 3' UTR of the *FKTN* gene common in some Asian populations. For patients with suspected Fukuyama congenital muscular dystrophy, testing for the *FKTN* insertion is recommended. Analysis for the *FKTN* insertion is available as a separate assay.

Reference:

- GeneReviews

Genes

[ABCC9](#), [ACTC1](#), [ACTN2](#), [ANKRD1](#), [BAG3](#), [CRYAB](#), [CSRP3](#), [DES](#), [DMD](#), [DSG2](#), [DSP](#), [EMD](#), [FKTN](#), [GATAD1](#), [LAMA4](#), [LAMP2](#), [LDB3](#), [LMNA](#), [MYBPC3](#), [MYH6](#), [MYH7](#), [MYPN](#), [NEBL](#), [NEXN](#), [PDLIM3](#), [PLN](#), [PRDM16](#), [RBM20](#), [SCN5A](#), [SGCD](#), [TAZ](#), [TCAP](#), [TNNC1](#), [TNNI3](#), [TNNT2](#), [TPM1](#), [TTN](#), [TTR](#), [VCL](#)

Indications

This test is indicated for:

- Confirmation of a clinical diagnosis of hereditary dilated cardiomyopathy (DCM).
- Carrier testing in adults with a family history of hereditary dilated cardiomyopathy (DCM).

Methodology

Next Generation Sequencing: In-solution hybridization of all coding exons is performed on the patient's genomic DNA. Although some deep intronic regions may also be analyzed, this assay is not meant to interrogate most promoter regions, deep intronic regions, or other regulatory elements, and does not detect single or multi-exon deletions or duplications. Direct sequencing of the captured regions is performed using next generation sequencing. The patient's gene sequences are then compared to a standard reference sequence. Potentially causative variants and areas of low coverage are Sanger-sequenced. Sequence variations are classified as pathogenic, likely pathogenic, benign, likely benign, or variants of unknown significance. Variants of unknown significance may require further studies of the patient and/or family members.

Detection

Next Generation Sequencing: Clinical Sensitivity: Unknown. Mutations in the promoter region, some mutations in the introns and other regulatory element mutations cannot be detected by this analysis. Large deletions/duplications will not be detected by this analysis. Results of molecular analysis should be interpreted in the context of the patient's clinical/biochemical phenotype.

Analytical Sensitivity: ~99%.

Specimen Requirements

Submit only 1 of the following specimen types

Type: Saliva

Specimen Requirements:

Oragene™ Saliva Collection Kit

Oragene™ Saliva Collection Kit used according to manufacturer instructions. Please contact EGL for a Saliva Collection Kit for patients that cannot provide a blood sample.

Specimen Collection and Shipping:

Please do not refrigerate or freeze saliva sample. Please store and ship at room temperature.

Type: DNA, Isolated

Specimen Requirements:

Microtainer

8µg

Isolation using the Perkin Elmer™ Chemagen™ Chemagen™ Automated Extraction method or Qiagen™ Puregene kit for DNA extraction is recommended.

Specimen Collection and Shipping:

Refrigerate until time of shipment in 100 ng/μL in TE buffer. Ship sample at room temperature with overnight delivery.

Type: Whole Blood (EDTA)**Specimen Requirements:**

EDTA (Purple Top)

Infants and Young Children (2 years of age to 10 years old): 3-5 ml

Older Children & Adults: 5-10 ml

Autopsy: 2-3 ml unclotted cord or cardiac blood

Specimen Collection and Shipping:

Ship sample at room temperature for receipt at EGL within 24 hours of collection. Do not refrigerate or freeze.

Related Tests

- Comprehensive Cardiomyopathy Sequencing and Deletion/Duplications Panels.
- Dilated Cardiomyopathy: Deletion/Duplication Panel.