Dilated Cardiomyopathy: Deletion/Duplication Panel

Test Code: MD092
Turnaround time: 2 weeks
CPT Codes: 81404 x1, 81616 x1, 81228 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.

Reference:
- GeneReviews

Genes

ABC9, ACTN2, ACTC1, ACTN4, ANKRD1, BAG3, CRYAB, CSRP3, DES, DMD, DSG2, DSP, EMD, FKTN, GATA4, LAMA4, LAMP2, LDB3, LMNA, MYBPC3, MYH6, MYH7, MYH8, MYPN, NEBL, NEXN, PLN, RBM20, SCN5A, SGCD, TAZ, TCAP, TNN1, TNN1C, TNN1C1, 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

For Deletion/Duplication Analysis: DNA isolated from peripheral blood is hybridized to a CGH array to detect deletions and duplications. The targeted CGH array has overlapping probes which cover the entire genomic region.

Detection

Deletion/Duplication Analysis: Detection is limited to duplications and deletions. The CGH array will not detect point or intronic mutations. Results of molecular analysis must be interpreted in the context of the patient's clinical and/or biochemical phenotype.

Specimen Requirements

Submit only 1 of the following specimen types

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 72 hours of collection. Do not freeze.

Type: DNA, Isolated

Specimen Requirements:
Microtainer
3µg
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.

Related Tests

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

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