Dilated Cardiomyopathy: Deletion/Duplication Panel

Test Code: MD092
Turnaround time: 2 weeks
CPT Codes: 81161 x1, 81228 x1, 81404 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, ACTC1, ACTN2, ANKRD1, BAG3, CRYAB, CSRP3, DES, DMD, DSG2, DSP, EMD, FKTN, GATAD1, LAMA4, LAP2, LDB3, LMNA, MYH8PC3, MYH6, MYH7, MYPN, NEBL, NEPX, PLN, 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

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. Please note that a “backbone” of probes across the entire genome are included on the array for analytical and quality control purposes. Rarely, off-target copy number variants causative of disease may be identified that may or may not be related to the patient's phenotype. Only known pathogenic off-target copy number variants will be reported. Off-target copy number variants of unknown clinical significance will not be reported.

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

Specimen Requirements:
- In EDTA (purple top) tube:
  - Infants (2 years): 3-5 ml
  - Older Children & Adults: 5-10 ml

Specimen Collection and Shipping: Ship sample at room temperature with overnight delivery.

Type: Isolated DNA

Specimen Requirements:
- In microtainer: 10 ug

Isolation using the Qiagen™ Puregene kit for DNA extraction is recommended.

Specimen Collection and Shipping: Refrigerate until time of shipment in 100 ng/μl of TE buffer. Ship sample at room temperature with overnight delivery.

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Related Tests

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