Cardiomyopathy: Deletion/Duplication Panel

Test Code: MD520
Turnaround time: 2 weeks
CPT Codes: 81228 x1, 81479 x1

<table>
<thead>
<tr>
<th>Condition Description</th>
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<tr>
<td>Dilated Cardiomyopathy</td>
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<tr>
<td>Hypertrophic Cardiomyopathy</td>
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<td>Left Ventricular Noncompaction</td>
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<tr>
<td>Restrictive Cardiomyopathy</td>
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<tr>
<td>Arrhythmogenic Right Ventricular Dysplasia/Cardiomyopathy</td>
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<tr>
<td>Catecholaminergic Polymorphic Ventricular Tachycardia</td>
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electrical instability. This instability can be exacerbated by acute activation of the adrenergic nervous system, such as during exercise or extreme emotional events. These episodes have an underlying cause of ventricular tachycardia, which may progress into ventricular fibrillation.

The Cardiomyopathy Panel offered at Emory Genetics Laboratory includes genes that cause each of the above cardiomyopathies, as well as genes that cause genetic syndromes, which have cardiomyopathy as a clinical feature. Syndromic conditions on this panel include Duchenne/Becker muscular dystrophy, Emery-Dreifuss muscular dystrophy, Pompe disease, Fabry disease, Danon disease, Charcot-Marie Tooth, congenital muscular dystrophy, limb girdle muscular dystrophy, Wolff-Parkinson-White syndrome, cardiac glycogenosis, Barth syndrome, familial transthyretin amyloidosis, myofibrillar myopathy, total anomalous venous return, rippling muscle disease, long QT syndrome, skin fragility and wooly hair syndrome, lethal acantholytic epidermolysis bullosa, Naxos disease, and progeria.

References:

- GeneReviews

Genes

ABC9, ACTC1, ACTN2, ANKR1D1, BAG3, BRAF, CASQ2, CAV3, CRYAB, CSRP3, DES, DMD, DSC2, DSG2, DSP, DTNA, EMD, FKTN, GAA, GATAD1, GLA, HRAS, JPH2, JUP, Kras, LAMA4, LAMP2, LB63, LMNA, MAP2K1, MAP2K2, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYOZ2, MYOPN, NEBL, NEXN, NRAS, PKP2, PLN, PRKAG2, PTPN11, RAF1, RBBM90, RIT1, RYR2, SCN5A, SGCD, SOS1, TAZ, TGRAP, TMEM43, TNNC1, TNNT2, TPM1, TTN, TTR, VCL

Indications

This test is indicated for:

- Individuals with a cardiomyopathy.

Methodology

Deletion/Duplication Analysis: DNA isolated from peripheral blood is hybridized to a gene-targeted CGH array to detect deletions and duplications. The targeted CGH array has overlapping probes that 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/ul of TE buffer. Ship sample at room temperature with overnight delivery.

**Related Tests**

- Individual gene sequencing analysis is available for CAV3, DES, DMD, EMD, GAA, GLA, LAMP2, LMNA, RYR2, SGCD, and TCAP.
- Custom diagnostic mutation analysis (KM) is available to family members if mutations are identified by targeted mutation testing or sequencing analysis.
- Prenatal testing is available only for known familial mutations to individuals who are confirmed carriers of mutations. Please contact the laboratory genetic counselor to discuss appropriate testing prior to collecting a prenatal specimen.
- Cardiomyopathy: Sequencing Panel.