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Comprehensive Cardiomyopathy Panel

Next Gen Sequencing Test Specifications

Cardiomyopathy refers to a disease of the heart which is usually characterized by an enlargement or thickening or greater rigidity of the heart muscle. Although cardiomyopathies can be acquired, in many cases genetic factors can be ultimately attributed to the disease. Cardiomyopathy often goes undiagnosed, but it has been estimated that as many as 1 of 500 adults may have this condition. ApolloGen's Comprehensive Cardiomyopathy Panel detects target genes associated with inherited cardiomyopathies, including Hypertrophic Cardiomyopathy (HCM), Dilated Cardiomyopathy (DCM), Restrictive Cardiomyopathy (RCM), Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC), Catecholaminergic Polymorphic Ventricular Tachycardia (CPVT), and Left Ventricular Non-Compaction (LVNC). The causal genes for Danon Syndrome, Fabry disease, Barth Syndrome, and Transthyretin Amyloidosis are also included in this panel.

Indications for Testing

1. Molecular confirmation of a clinical diagnosis of cardiomyopathy in symptomatic patients.
2. Assessment for at-risk family members

Requisition Form: Cardiac Test Requisition Form or General Test Requisition Form (www.apollogen.com)

Genes (44): *ABCC9, ACTC1, ACTN2, BMPR2, CAV3, DES, GLA, LAMA4, LAMP2, LDB3, LMNA, MT-TD, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TQ, MT-TS1, MT-TS2, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYPN, PLN, PRKAG2, PSEN1, PSEN2, RBM20, SCN5A, SGCD, TAZ, TCAP, TMPO, TNNC1, TNNI3, TNNT2, TPM1, TTR, and VCL*

CPT Codes: 81401x2, 81403x2, 81404x2, 81405x11, 81406x7, 81407x3, 81408x1, 81479x16

Turnaround Time: 4 weeks

Specimen Requirement: 3-5 mL Blood (EDTA) – Lavender Top Tube (preferred);

Saliva samples are optional

Other Specimen Types: Contact ApolloGen Diagnostic Laboratory

Pricing: Please contact us at (949) 916-8886 or at inquiries@apollogen.com for current pricing

Testing Methodology

Genomic DNA is extracted from the patient's specimen and fragmented via sonication. All of the exons, flanking intronic (at least 10 nucleotides into the introns), and untranslated regions (5' and 3') of the targeted genes are enriched using capture-based hybridization. Massively parallel sequencing is applied to the enriched target DNA regions to detect mutation. The region <10- fold coverage is regarded as a low coverage region and will be rescued by Sanger Sequencing. Variants with an allele frequency > 1% are considered likely benign polymorphisms, and are not included in the final report. Interpretation of rare alterations with allele frequency <1% is based on ACMG guidelines. All pathogenic and likely pathogenic variants are verified by Sanger Sequencing.

Massively parallel sequencing can reliably detect insertion/deletion mutations smaller than 10 base pairs. However, larger insertion, deletion, duplication due to rearrangement, and mutations in regulatory and deep intronic regions cannot be detected by this technology. Rare primer site variants may lead to erroneous results that may need further investigation.

Analytical Sensitivity:

This test can detect >95% of the small variants in the examined regions. Please contact us for detailed information regarding coverage for specific genes of interest.

Related Tests

Dilated Cardiomyopathy Panel

Simultaneously analyzes 33 critical genes associated with an increased risk for dilated cardiomyopathy

Hypertrophic Cardiomyopathy Panel

Analyzes 18 critical genes that have been associated with an increased for hypertrophic cardiomyopathy

Atrial Fibrillation Panel

Analyzes 47 critical genes that have been associated with atrial fibrillation.

Variant Classification

Sequencing results will be interpreted and reported following the recommendations of the American College of Medical Genetics (www.acmg.net). Sequence variations will be analyzed and classified into the following categories based on current scientific knowledge. Variants found in categories 1-3 (pathogenic, likely pathogenic, and variants of unknown clinical significance) will be reported.

1. **Pathogenic:** Pathogenic variants include nonsense mutations and frame shift mutations that are predicted to result in premature protein truncation, splice site mutations, and previously reported missense mutations that are recognized as disease-causing by databases and the scientific literature.
2. **Likely Pathogenic:** Likely pathogenic variants are those variants that are likely to adversely affect gene function, but for which there is no conclusive evidence to strongly support pathogenicity.
3. **Variant of Unknown Clinical Significance (VUS):** VUSs are sequence variations for which there is insufficient evidence to either confirm or exclude pathogenicity.
4. **Likely Benign:** Likely benign variants are sequence variations for which there is significant, but not conclusive evidence supporting that the variant is not disease-causing.
5. **Negative:** A negative classification is reported when no disease-causing variant is found, or a variant is classified as a benign polymorphism based on the population data, or if there is no clinical significance based on review of the literature and mutation databases.

References

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