

Client: Example Client ABC123
123 Test Drive
Salt Lake City, UT 84108
UNITED STATES

Physician: Doctor, Example

Patient: Patient, Example

DOB: 1/23/1984
Gender: Male
Patient Identifiers: 01234567890ABCD, 012345
Visit Number (FIN): 01234567890ABCD
Collection Date: 00/00/0000 00:00

Cardiomyopathy and Arrhythmia Panel, Sequencing and Deletion/Duplication

ARUP test code 2010183

Cardiomyopathy/Arrhythmia Panel Specimen whole Blood

Cardiomyopathy/Arrhythmia Panel Interp

Negative

INDICATION FOR TESTING
Clinical diagnosis of dilated cardiomyopathy.

RESULT
No pathogenic variants were detected in any of the genes tested.

INTERPRETATION
No pathogenic variants were identified by massively parallel sequencing of the coding regions and exon-intron boundaries of the genes tested. No large exonic deletions and duplications were identified in the genes tested. This result decreases the likelihood of, but does not exclude, a heritable form of cardiomyopathy or arrhythmia. Please refer to the background information included in this report for a list of the genes analyzed and limitations of this test.

RECOMMENDATIONS
Medical screening and management should rely on clinical findings and family history. Genetic consultation is recommended.

COMMENTS
Likely benign and benign variants are not reported.

This result has been reviewed and approved by [REDACTED]

BACKGROUND INFORMATION: Cardiomyopathy and Arrhythmia Panel, Sequencing and Deletion/Duplication

CHARACTERISTICS: Inherited cardiomyopathy and arrhythmia disorders are genetically and phenotypically heterogeneous. Phenotypes include hypertrophic cardiomyopathy (HCM), dilated cardiomyopathy (DCM), arrhythmogenic right ventricular cardiomyopathy (ARVC), left ventricular noncompaction (LVNC), catecholaminergic polymorphic ventricular tachycardia (CPVT), Brugada syndrome (BrS), long QT syndrome (LQTS), and short QT syndrome (SQTS).

EPIDEMIOLOGY: The prevalence of HCM is 1 in 500, DCM is greater than 1 in 500, ARVC is 1 in 1000, LQTS is 1 in 3,000, CPVT is 1 in 10,000, and it is unknown for BrS, LVNC, and SQTS.

CAUSE: Pathogenic germline variants in genes associated with cardiomyopathy and arrhythmia such as nuclear genes encoding

H=High, L=Low, *=Abnormal, C=Critical

sarcomeric or desmosomal proteins, cardiac ion channel components and cytoskeletal proteins, or pathogenic variants within the mitochondrial genome.

INHERITANCE: Is gene dependent and can be autosomal recessive, autosomal dominant, X-linked or mitochondrial.

PENETRANCE: Variable; dependent on gene and variant.

CLINICAL SENSITIVITY: Dependent on clinical phenotype. Estimated at 50 percent for ARVC, 15-30 percent for BrS, 60 percent for CPVT, 30-40 percent for DCM, 50-60 percent for non-syndromic familial HCM, and 60-75 percent for LQTS.

GENES TESTED: ABCC9, ACTC1, ACTN2, ANK2, ANKRD1, BAG3**, CACNA1C, CACNB2, CASQ2, CAV3, CRYAB**, CSRP3, DES, DMD, DSC2, DSG2, DSP, DTNA, EMD**, EYA4, FHL1**, FKRP, FKTN, GAA, GATAD1**, GLA, GPD1L, JPH2, JUP, KCNE1, KCNE2, KCNE3, KCNH2, KCNJ2, KCNQ1, LAMA4, LAMP2, LDB3, LMNA, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOT, MYOZ2, MYPN, NEXN, PKP2, PLN, PRKAG2, RBM20, RYR2, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SGCA, SLC6A1, SLC6A4, SLC6A6, SLC6A7, SLC6A8, SLC6A9, SLC6A10, SLC6A11, SLC6A12, SLC6A13, SLC6A14, SLC6A15, SLC6A16, SLC6A17, SLC6A18, SLC6A19, SLC6A20, SLC6A21, SLC6A22, SLC6A23, SLC6A24, SLC6A25, SLC6A26, SLC6A27, SLC6A28, SLC6A29, SLC6A30, SLC6A31, SLC6A32, SLC6A33, SLC6A34, SLC6A35, SLC6A36, SLC6A37, SLC6A38, SLC6A39, SLC6A40, SLC6A41, SLC6A42, SLC6A43, SLC6A44, SLC6A45, SLC6A46, SLC6A47, SLC6A48, SLC6A49, SLC6A50, SLC6A51, SLC6A52, SLC6A53, SLC6A54, SLC6A55, SLC6A56, SLC6A57, SLC6A58, SLC6A59, SLC6A60, SLC6A61, SLC6A62, SLC6A63, SLC6A64, SLC6A65, SLC6A66, SLC6A67, SLC6A68, SLC6A69, 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following exons:
ACTN2(NM_001103) 17; CACNB2(NM_001167945) 7; DMD(NM_000109) 1;
DSC2(NM_024422) 1; DTNA(NM_001390) 16,21; KCNH2(NM_000238) 13;
KCNQ1(NM_000218) 16; KCNQ1(NM_181798) 1; MYBPC3(NM_000256)
10,24; MYH6(NM_002471) 24,28; MYH7(NM_000257) 29;
PKP2(NM_004572) 6; PRKAG2(NM_001304527) 1; PRKAG2(NM_001304531)
2; PRKAG2(NM_016203) 5; SGCB(NM_000232) 1; SGCG(NM_000231) 8;
TGFB3(NM_003239) 7; TRPM4(NM_001321285) 4; TRPM4(NM_017636) 7;
TTN(NM_001267550) 158,173,176,185,191,194; TTN(NM_133378)
114,148,155,156,167,170

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Cardiomyopathy/Arrhythmia Panel Specimen	21-085-402602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Cardiomyopathy/Arrhythmia Panel Interp	21-085-402602	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00

END OF CHART

H=High, L=Low, *=Abnormal, C=Critical