

Client: ARUP Example Report Only  
500 Chipeta Way  
Salt Lake City, UT 84108  
UNITED STATES

Physician: ARUP,

**Patient: REPORT, NEGATIVE**

**DOB:** 4/21/2021  
**Gender:** Female  
**Patient Identifiers:** 29771  
**Visit Number (FIN):** 30076  
**Collection Date:** 4/22/2021 13:59

**Dilated Cardiomyopathy Panel, Sequencing**

ARUP test code 3001581

Dilated Cardiomyopathy Specimen      whole Blood

**Dilated Cardiomyopathy Interp**

Negative

INDICATION FOR TESTING  
Family history 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. This result decreases the likelihood of, but does not exclude, a heritable form of dilated cardiomyopathy. 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 of this individual should rely on clinical findings and family history. Cardiology and genetic consultations are recommended.

COMMENTS  
Likely benign and benign variants are not included in this report, but are available upon request.

This result has been reviewed and approved by [REDACTED]

BACKGROUND INFORMATION: Dilated Cardiomyopathy Panel, Sequencing

CHARACTERISTICS: Dilated cardiomyopathy (DCM) is characterized by left ventricular enlargement with systolic dysfunction. DCM is a leading cause of symptoms requiring heart transplantation in children and adults. Familial DCM is defined as two or more individuals in single family with DCM, or an individual with DCM with a relative with unexplained sudden death less than 35 years of age. Affected individuals are at risk for heart failure, arrhythmias or conduction disease, pregnancy-related cardiomyopathy, stroke, and sudden cardiac death. Symptoms may include dyspnea, chest pain, palpitations, fatigue, fainting or edema. Syndromic forms of DCM include extracardiac manifestations.

EPIDEMIOLOGY: Prevalence of DCM is estimated at 1:250 to 1:2500; 20-50 percent of cases are familial.

CAUSE: Pathogenic germline variants in genes associated with

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

Unless otherwise indicated, testing performed at:

**ARUP LABORATORIES | 800-522-2787 | aruplab.com**  
500 Chipeta Way, Salt Lake City, UT 84108-1221  
Tracy I. George, MD, Laboratory Director

Patient: REPORT, NEGATIVE  
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familial DCM.

**INHERITANCE:** Typically autosomal dominant for familial DCM. Genes with X-linked, autosomal recessive, and mitochondrial inheritance also associated. De novo variation, compound heterozygous, or digenic heterozygous variants have been reported.

**PENETRANCE:** Variable.

**CLINICAL SENSITIVITY:** 25-40 percent for familial DCM, 10-25 percent for isolated DCM.

**GENES TESTED:** ABCC9, ACTC1, ACTN2, ALMS1, BAG3, CRYAB, CSR3, DES, DMD, DOLK, DSC2, DSG2, DSP, EMD, FKTN, FLNC\*, GLA, JUP, LAMP2, LDB3, LMNA, MYBPC3, MYH6, MYH7, MYL2, MYL3, PKP2, PLN, PRDM16, PRKAG2\*, RAF1, RBM20, RYR2, SCN5A, SGCD, TAZ, TCAP, TMEM43, TNNC1, TNNI3, TNNT2, TPM1, TTN\*, TTR, VCL.

\* - One or more exons are not covered by sequencing for the indicated gene; see limitations section below.

**METHODOLOGY:** Capture of all coding exons and exon-intron junctions of the targeted genes, followed by massively parallel sequencing. Sanger sequencing was performed as necessary to fill in regions of low coverage and to confirm reported variants. Human genome build 19 (Hg 19) was used for data analysis.

**ANALYTICAL SENSITIVITY/SPECIFICITY:** The analytical sensitivity of this test is approximately 99 percent for single nucleotide variants (SNVs) and greater than 93 percent for insertions/duplications/deletions from 1-10 base pairs in size. Variants greater than 10 base pairs may be detected, but the analytical sensitivity may be reduced. Variants greater than 10 base pairs may be detected, but the analytical sensitivity may be reduced.

**LIMITATIONS:** A negative result does not exclude a heritable form of DCM. This test only detects variants within the coding regions and intron-exon boundaries of the targeted genes. Mitochondrial (mtDNA) genes are not interrogated. Regulatory region variants, large deletions/duplications/inversions and deep intronic variants will not be identified. Deletions/duplications/insertions of any size may not be detected by massive parallel sequencing. Diagnostic errors can occur due to rare sequence variations. In some cases, variants may not be identified due to technical limitations in the presence of pseudogenes, repetitive, or homologous regions. This assay may not detect low-level mosaic or somatic variants associated with disease. Interpretation of this test result may be impacted if this patient has had an allogeneic stem cell transplantation. Non-coding transcripts are not analyzed.

The following regions are not sequenced due to technical limitations of the assay:

FLNC(NM\_001458) exons 47, 48

PRKAG2(NM\_016203) exons 10, 13

TTN(NM\_001267550) exons 172, 173, 175, 176, 177, 178, 179, 180, 183, 184, 185, 186, 187, 188, 189, 190, 191, 192, 193, 194, 195, 196, 197, 215

TTN(NM\_133378) exons 153, 154, 155

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.

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VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Dilated Cardiomyopathy Specimen	21-112-115658	4/22/2021 1:59:00 PM	4/22/2021 1:59:35 PM	4/22/2021 2:01:00 PM
Dilated Cardiomyopathy Interp	21-112-115658	4/22/2021 1:59:00 PM	4/22/2021 1:59:35 PM	4/22/2021 2:01:00 PM

END OF CHART

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