ARUP LABORATORIES | www.aruplab.com
500 Chipeta Way, Salt Lake City, Utah 84108-1221
phone: (801) 583-2787, toll free: (800) 522-2787
Julio C. Delgado, MD, MS, Director of Laboratories

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

Physician: Doctor, Example

Patient: Patient, Example

DOB	3/9/2019
Gender:	Female
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	00/00/0000 00:00

Rapid Mendelian Genes Sequencing Panel, Trio

ARUP test code 2012849

Rapid Sequencing Specimen	Whole Blood
Rapid Sequencing Interpretation	Negative

Section 79-1 of New York State Civil Rights Law requires informed consent be obtained from patients (or their legal guardians) prior to pursuing genetic testing. These forms must be kept on file by the ordering physician. Consent forms for genetic testing are available at www.aruplab.com. Incidental findings are not reported unless clinically significant but are available upon request.

Test Performed: Rapid Mendelian Genes Sequencing Panel, Trio

Next generation sequencing was performed on 4900+ genes in the patient and this patients unaffected parents. Parental sequencing was performed to aid in variant analysis and result interpretation.

Indication for Testing: [Individual medical history and indication for testing.]

Result: Negative; no pathogenic variants were identified that provide an explanation for this individual's phenotype.

Interpretation: No pathogenic variants were identified in any of the 4900+ genes analyzed that provide an explanation for this individual's symptoms. No rare de novo variant was identified.

Please refer to the background information included in this report for limitations of this test.

Recommendations: Medical management and screening should rely on clinical findings. Genetic consultation is recommended.

No secondary pathogenic variants were detected in the American College of Medical Genetics and Genomics (ACMG) recommended list of genes. This result does not exclude the possibility this individual may have a variant in one of these genes as not all of the ACMG recommended genes are adequately covered by this method, or in another gene not included on this list. Note that single pathogenic variants in recessive genes from this list are not reported.

This result has been reviewed and approved by Rong Mao, M.D.

BACKGROUND INFORMATION: Rapid Mendelian Genes
Sequencing Panel, Trio

CHARACTERISTICS: This gene panel was designed for diagnostic testing in critically ill infants suspected of having an underlying genetic diagnosis. Massively parallel sequencing was performed on the DNA coding regions and intron/exon boundaries of genes identified in the Human Genome Mutation Database (HGMD) that are confirmed to have a human disease association. Variants of uncertain significance in recessive genes will be reported when one pathogenic variant has also been identified in the same gene. Parental samples are required for interpretation of results.

CLINICAL SENSITIVITY: Varies based on clinical symptoms, family history, inheritance pattern, previous evaluations and receipt of parental samples.

GENES TESTED: Refer to Additional Technical Information located at http://ltd.aruplab.com/Tests/Pub/2012849.

METHODOLOGY: Targeted capture of all (or selected) coding exons and exon-intron junctions of the targeted genes, followed by massively parallel sequencing. Sanger sequencing was performed as necessary 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.

LIMITATIONS: A negative result does not exclude all genetic diagnoses. This test only detects variants within the coding regions and intron-exon boundaries of the targeted genes. Regulatory region variants and deep intronic variants will not be identified. Deletions/duplications/insertions of any size may not be detected by massively 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 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 were not analyzed. Refer to Additional Technical Information for complete list of genes tested and gene-specific limitations, located at http://ltd.aruplab.com/Tests/Pub/2012849.

Test developed and characteristics determined by ARUP Laboratories. See Compliance Statement C: aruplab.com/CS

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Rapid Sequencing Specimen	19-070-151132	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Rapid Sequencing Interpretation	19-070-151132	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