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

Physician: Doctor, Example

Patient: Patient, Example

DOB	9/11/1968
Gender:	Female
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	01/01/2017 12:34

Citrullinemia, Type I (ASS1) Sequencing

ARUP test code 2007069

Citrullinemia, Type I (ASS1)Seq Specimen	Whole Blood

Citrullinemia, Type I (ASS1) Seq Interp

Negative

TEST PERFORMED - 2007069
TEST DESCRIPTION - Citrullinemia, Type I (ASS1) Sequencing
INDICATION FOR TEST - Not Provided

RESULT
No pathogenic variants were detected in the ASS1 gene.

INTERPRETATION
No pathogenic variants were detected in the ASS1 gene by sequencing all coding regions and intron-exon boundaries. This result significantly decreases the likelihood that this individual is affected with, or a carrier of, citrullinemia type I (CTLN1). Please refer to the background information included in this report for the clinical sensitivity and limitations of this test.

RECOMMENDATIONS
Medical and dietary management should rely on clinical and biochemical findings. Genetic consultation is recommended. If clinical suspicion for CTLN1 remains high, consideration should be given to ASS1 deletion/duplication analysis.

COMMENTS
Reference Sequence: GenBank # NM_000050.4 (ASS1)
Nucleotide numbering begins at the "A" of the ATG initiation codon.
Likely benign and benign variants are not reported.

This result has been reviewed and approved by [redacted]

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

BACKGROUND INFORMATION: Citrullinemia, Type I (ASS1) Sequencing:

Characteristics: Classic citrullinemia type I is a urea cycle disorder characterized by hyperammonemia, lethargy, vomiting, coma and neonatal death if not treated. There is also a milder, late-onset form and a form in which women have onset of severe symptoms during pregnancy or postpartum.
Incidence: Approximately 1 in 57,000.
Inheritance: Autosomal recessive.
Penetrance: Variable.
Cause: Pathogenic ASS1 gene mutations.
Clinical Sensitivity: Approximately 96 percent.
Methodology: Bidirectional sequencing of the entire ASS1 coding region and intron/exon boundaries.
Analytical Sensitivity and Specificity: 99 percent.
Limitations: Diagnostic errors can occur due to rare sequence variations. Regulatory region mutations, deep intronic mutations, and large deletions/duplications will not be detected. Mutations in genes other than ASS1 are not evaluated.

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

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Citrullinemia, Type I (ASS1)Seq Specimen	19-281-401007	10/8/2019 5:55:00 AM	10/9/2019 6 29:53 PM	10/29/2019 8:43:00 AM
Citrullinemia, Type I (ASS1) Seq Interp	19-281-401007	10/8/2019 5:55:00 AM	10/9/2019 6 29:53 PM	10/29/2019 8:43:00 AM

END OF CHART