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

Physician: Doctor, Example

Patient: Patient, Example

DOB	12/31/1752
Gender:	Female
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	01/01/2017 12:34

Familial Mutation, Targeted Sequencing, Fetal

ARUP test code 2001980

SEQ FSM FE Specimen	Amniotic fluid
Targeted Sequencing Gene	OTC
Targeted Sequencing Interpretation	Negative

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

TEST PERFORMED - 2001980
TEST DESCRIPTION - Familial Mutation, Targeted Sequencing, Fetal
INDICATION FOR TEST- Predictive Testing

RESULT
Negative for the requested variant in the OTC gene.

INTERPRETATION
The pathogenic familial OTC gene variant, c.844delC; p.Gln282fs, was not detected in this prenatal sample by targeted sequencing. This variant was previously reported to be associated with ornithine transcarbamylase (OTC) deficiency in the family. This male fetus is predicted to be unaffected with OTC deficiency.

RECOMMENDATIONS
Genetic consultation is recommended. For quality assurance purposes, ARUP Laboratories will confirm the above result at no charge following delivery. Order Confirmation of Fetal Testing and include a copy of the original fetal report (or the mother's name and date of birth) with the test submission. Please contact an ARUP genetic counselor at (800) 242-2787 extension 2141 prior to specimen submission.

COMMENTS
Reference Sequence: GenBank # NM_000531.5
Nucleotide numbering begins at the "A" of the ATG initiation codon.
Benign variants are not included in this report but are available upon request.

Note: A positive familial control was tested.

This result has been reviewed and approved by [redacted]

Background Information for Ornithine Transcarbamylase Deficiency (OTC), Familial Mutation, Targeted Sequencing:
Characteristics: Classic OTC deficiency is a urea cycle disorder characterized by hyperammonemia, cyclical vomiting, seizures, lethargy, coma and neonatal death if not treated. Clinical presentation varies widely in females, and some males can have non-classical forms.
Incidence: Approximately 1 in 14,000.
Inheritance: X-linked.
Penetrance: Variable depending on sex and mutation.
Cause: Pathogenic OTC gene mutations.
Clinical Sensitivity: Approximately 80 percent.
Methodology: Bidirectional sequencing of a specific OTC exon.
Analytical Sensitivity and Specificity of Sequencing: 99 percent.
Limitations: Only the requested OTC familial mutation will be tested. Rare diagnostic errors can occur due to primer site mutations.

BACKGROUND INFORMATION: Familial Mutation, Targeted Sequencing

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

Maternal Contamination Study Fetal Spec

Fetal Cells

Single fetal genotype present; no maternal cells present. Fetal and maternal samples were tested using STR markers to rule out maternal cell contamination.

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

	INTERPRETIVE INFORMATION: Maternal Cell Contamination, Fetal Specimen Please refer to fetal report for interpretation.
Maternal Contam Study, Maternal Spec	Whole Blood For quality assurance purposes, ARUP Laboratories will confirm the above result at no charge following delivery. Order Confirmation of Fetal Testing and include a copy of the original fetal report (or the mother's name and date of birth) with the test submission. Please contact an ARUP genetic counselor at (800) 242-2787 extension 2141 prior to specimen submission.

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
SEQ FSM FE Specimen	17-011-118790	1/11/2017 6:36:00 PM	1/11/2017 6:37:09 PM	1/18/2017 6:01:00 PM
Targeted Sequencing Gene	17-011-118790	1/11/2017 6:36:00 PM	1/11/2017 6:37:09 PM	1/18/2017 6:01:00 PM
Targeted Sequencing Interpretation	17-011-118790	1/11/2017 6:36:00 PM	1/11/2017 6:37:09 PM	1/18/2017 6:01:00 PM
Maternal Contamination Study Fetal Spec	17-011-118790	1/11/2017 6:36:00 PM	1/11/2017 6:37:09 PM	1/18/2017 6:01:00 PM
Maternal Contam Study, Maternal Spec	17-011-118790	1/11/2017 6:36:00 PM	1/11/2017 6:37:09 PM	1/18/2017 6:01:00 PM

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