ARUP LABORATORIES
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Tracy I. George, MD, Chief Medical Officer

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

Physician: Doctor, Example

Patient: Patient, Example

DOB	9/2/1990
Gender:	Male
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	01/01/2017 12:34

Guanidinoacetate Methyltransferase (GAMT) Deficiency Sequencing

ARUP test code 2011140

GAMT Sequencing Specimen	See Note *
GAMT Sequencing Interpretation	Negative

TEST PERFORMED - 2011140
TEST DESCRIPTION - Guanidinoacetate Methyltransferase (GAMT) Deficiency Sequencing
INDICATION FOR TEST - Not Provided

RESULT
No pathogenic variants were detected in the GAMT gene.

INTERPRETATION
No pathogenic GAMT gene variants were detected by sequencing of all coding regions and intron-exon boundaries. This result significantly reduces, but does not eliminate, the likelihood that this individual is affected with guanidinoacetate methyltransferase (GAMT) deficiency. Please refer to the background information included in this report for the clinical sensitivity and limitations of this test.

RECOMMENDATIONS
The diagnosis and management of creatine deficiency syndromes should rely on clinical symptoms and biochemical/functional assay results. Additional molecular testing may be helpful depending on biochemical results. Genetic consultation is recommended.

COMMENTS
Reference Sequence: GenBank # NM_000156.5 (GAMT)
Nucleotide numbering begins at the "A" of the ATG initiation codon.

This result has been reviewed and approved by [redacted]

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

Background Information for Guanidinoacetate Methyltransferase (GAMT) Deficiency Sequencing:
Characteristics: Intellectual disability and seizure disorder of variable severity. May also include speech / language delays, movement disorder, and behavioral disorders such as autism, hyperactivity, and self-injury.
Incidence: Unknown. More than 50 cases have been described.
Inheritance: Autosomal recessive.
Cause: Pathogenic GAMT gene mutations.
Clinical Sensitivity: Based on limited data, may be as high as 99 percent.
Methodology: Bidirectional sequencing of the entire coding region and intron/exon boundaries of the GAMT gene.
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 GAMT are not evaluated.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online at www.aruplab.com.

See Compliance Statement C: www.aruplab.com/CS

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
GAMT Sequencing Specimen	17-304-401020	10/26/2017 12:01:00 AM	10/31/2017 11:12:00 AM	11/9/2017 12:57 00 PM
GAMT Sequencing Interpretation	17-304-401020	10/26/2017 12:01:00 AM	10/31/2017 11:12:00 AM	11/9/2017 12:57 00 PM

END OF CHART

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