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	7/8/2019
Gender:	Female
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	01/01/2017 12:34

Galactosemia (GALT), Sequencing

ARUP test code 2006697

Galactosemia (GALT) Sequencing Specimen	Whole Blood
Galactosemia (GALT) Sequencing Interp	Negative

TEST PERFORMED - 2006697
TEST DESCRIPTION - Galactosemia (GALT) Sequencing
INDICATION FOR TEST - Not Provided

RESULT
No pathogenic variants were detected in the GALT gene.

INTERPRETATION
No pathogenic GALT gene variants were detected by sequencing of the coding region, intron/exon boundaries and 5'UTR. This reduces, but does not eliminate the possibility of galactosemia as this test does not detect all pathogenic GALT gene variants (e.g., large deletions and deep intronic variants). Rare forms of galactosemia caused by pathogenic variants in other genes will not be detected.

RECOMMENDATIONS
Medical and dietary management should rely on clinical and biochemical findings. Genetic consultation is recommended.

COMMENTS
Reference Sequence: GenBank Ref #NM_000155.2 (GALT)
Nucleotide numbering begins at the "A" of the ATG initiation codon.
Likely benign and benign variants are not included in this report.

This result has been reviewed and approved by [redacted]

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

BACKGROUND INFORMATION: Galactosemia (GALT), Sequencing

CHARACTERISTICS: Vomiting, diarrhea, feeding problems, failure to thrive, hepatocellular damage, bleeding, sepsis, mental retardation, and neonatal death. If treated early, most symptoms resolve, although speech, motor problems, developmental delay and premature ovarian failure may persist.
INCIDENCE: Approximately 1 in 30,000.
INHERITANCE: Autosomal recessive.
PENETRANCE: 100 percent for severe mutations.
CAUSE: Pathogenic galactose-1-phosphate uridyl transferase (GALT) gene mutations.
CLINICAL SENSITIVITY: 98 percent.
METHODOLOGY: Bidirectional sequencing of the entire GALT coding region, intron/exon boundaries and partial 5'UTR.
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 GALT 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
Galactosemia (GALT) Sequencing Specimen	19-239-402539	8/27/2019 2:38:00 PM	8/28/2019 11:52:00 AM	9/13/2019 7:07:00 PM
Galactosemia (GALT) Sequencing Interp	19-239-402539	8/27/2019 2:38:00 PM	8/28/2019 11:52:00 AM	9/13/2019 7:07:00 PM

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