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

Physician: Doctor, Example

Patient: Patient, Example

DOB	6/18/2002
Sex:	Female
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	01/01/2017 12:34

Galactosemia (GALT) Enzyme Activity and 9 Mutations (Extended TAT as of 11/20/20-no referral available)

ARUP test code 0051175

Galac-1-Phos Uridyltransferase	16.0 U/g Hb L (Ref Interval: >=19.4)

INTERPRETIVE INFORMATION: Galac-1-Phos Uridyltransferase

One U/g Hb is equivalent to one umol/hour/gram of hemoglobin (umol/hr/g Hb).

Genotype	Galac-1-Phos Uridyltransferase activity(umol/hr/g Hb)
Classic galactosemia(G/G)	Less than or equal to 0.7
Duarte galactosemia(D/G)	3.1-7.8
Classic galactosemia carrier(G/N)	6.5-16.2
Duarte homozygous(D/D)	6.4-16.5
Duarte carrier(D/N)	12.0-24.0
Normal(N/N)	Greater than or equal to 19.4

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.

Galactosemia (GALT) DNA Pan,G1PUT Spec	Whole Blood
Galactosemia (GALT) Allele 1	Negative
Galactosemia (GALT) Allele 2	Negative
Galactosemia - Ethnicity	Unknown
Galactosemia - Symptoms	Unknown
Galactosemia - Abnormal Newborn Screen	Unknown
Galactosemia - Family History	Unknown
Galactosemia (GALT) DNA Pan,G1PUT Interp	See Note

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

Possible Galactosemia Carrier; Reduced Enzyme Activity/No Mutations Detected: This sample has partially reduced galactose-1-phosphate uridyltransferase (GALT) activity and is negative for the seven common GALT gene mutations and 2 variants tested. This individual is not affected with classic galactosemia, but may carry a mutation in the GALT gene not detected by this assay. Consideration should be given to full gene sequencing to detect the rare severe GALT mutation that was not identified by this assay (Galactosemia (GALT) Sequencing, ARUP test #2006697).Genetic consultation is recommended.

This result has been reviewed and approved by [redacted]

One U/g Hb is equivalent to one umol/hour/gram of hemoglobin (umol/hr/g Hb).

Background Information for Galactosemia (GALT) Enzyme Activity and 9 Mutations:
Characteristics: Affected infants present at 3-14 days old with poor feeding, vomiting, diarrhea, jaundice, lethargy progressing to coma, and abdominal distension with hepatomegaly usually followed by progressive liver failure. Patients with galactosemia are also at increased risk for E. coli or other gram-negative neonatal sepsis. Diagnosis is made by measuring GALT enzyme activity in red blood cells.
Incidence: Approximately 1 in 30,000 to 60,000 of classic galactosemia in Caucasian, varies in other populations.
Inheritance: Autosomal recessive
Penetrance: 100 percent for severe GALT mutations
Cause: Mutations in the GALT gene.
Mutations Tested: Seven GALT gene mutations (Q188R, S135L, K285N, T138M, L195P, Y209C, and IVS2-2 A>G) and two variants (N314D and L218L).
Clinical Sensitivity DNA: Approaches 80 percent in Caucasians but reduced in other ethnic groups.
Methodology DNA: Polymerase chain reaction followed by single nucleotide extension (SNE) and capillary electrophoresis.
Analytical Sensitivity DNA: 99 percent for mutations listed.
Limitations DNA: GALT gene mutations, other than the 9 targeted, will not be detected. Diagnostic errors can occur due to rare sequence variations.
Methodology Enzymatic: Spectrophotometric quantitation of enzyme activity in red blood cells.

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VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Galac-1-Phos Uridyltransferase	21-088-402027	3/27/2021 11:38:00 AM	3/29/2021 9:37:58 PM	4/7/2021 5:08:00 PM
Galactosemia (GALT) DNA Pan,G1PUT Spec	21-088-402027	3/27/2021 11:38:00 AM	3/29/2021 9:37:58 PM	4/7/2021 5:08:00 PM
Galactosemia (GALT) Allele 1	21-088-402027	3/27/2021 11:38:00 AM	3/29/2021 9:37:58 PM	4/7/2021 5:08:00 PM
Galactosemia (GALT) Allele 2	21-088-402027	3/27/2021 11:38:00 AM	3/29/2021 9:37:58 PM	4/7/2021 5:08:00 PM
Galactosemia - Ethnicity	21-088-402027	3/27/2021 11:38:00 AM	3/29/2021 9:37:58 PM	4/7/2021 5:08:00 PM

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

Galactosemia - Symptoms	21-088-402027	3/27/2021 11:38:00 AM	3/29/2021 9:37:58 PM	4/7/2021 5:08:00 PM
Galactosemia - Abnormal Newborn Screen	21-088-402027	3/27/2021 11:38:00 AM	3/29/2021 9:37:58 PM	4/7/2021 5:08:00 PM
Galactosemia - Family History	21-088-402027	3/27/2021 11:38:00 AM	3/29/2021 9:37:58 PM	4/7/2021 5:08:00 PM
Galactosemia (GALT) DNA Pan,G1PUT Interp	21-088-402027	3/27/2021 11:38:00 AM	3/29/2021 9:37:58 PM	4/7/2021 5:08:00 PM

END OF CHART

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