

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

Physician: Doctor, Example

Patient: Patient, Example

DOB: 3/17/2021
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

11.6 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	Q188R *
Galactosemia (GALT) Allele 2	Negative
Galactosemia - Ethnicity	Unknown
Galactosemia - Symptoms	Unknown
Galactosemia - Abnormal Newborn Screen	Yes
Galactosemia - Family History	Unknown
Galactosemia (GALT) DNA Pan,G1PUT Interp	See Note

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

Unless otherwise indicated, testing performed at:

ARUP LABORATORIES | 800-522-2787 | aruplab.com
500 Chipeta Way, Salt Lake City, UT 84108-1221
Jonathan R. Genzen, MD, PhD, Laboratory Director

Patient: Patient, Example
ARUP Accession: 21-097-148120
Patient Identifiers: 01234567890ABCD, 012345
Visit Number (FIN): 01234567890ABCD
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Galactosemia Carrier Reduced Enzyme Activity/ One Mutation Detected: This sample has partially reduced galactose-1-phosphate uridylyltransferase (GALT) activity. One mutation was identified in the GALT gene. Therefore this individual is predicted to be a carrier of galactosemia. 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	21-097-148120	4/7/2021 3:00:00 PM	4/9/2021 5:48:44 AM	4/15/2021 12:51:00 PM
Galactosemia (GALT) DNA Pan,G1PUT Spec	21-097-148120	4/7/2021 3:00:00 PM	4/9/2021 5:48:44 AM	4/15/2021 12:51:00 PM
Galactosemia (GALT) Allele 1	21-097-148120	4/7/2021 3:00:00 PM	4/9/2021 5:48:44 AM	4/15/2021 12:51:00 PM
Galactosemia (GALT) Allele 2	21-097-148120	4/7/2021 3:00:00 PM	4/9/2021 5:48:44 AM	4/15/2021 12:51:00 PM
Galactosemia - Ethnicity	21-097-148120	4/7/2021 3:00:00 PM	4/9/2021 5:48:44 AM	4/15/2021 12:51:00 PM
Galactosemia - Symptoms	21-097-148120	4/7/2021 3:00:00 PM	4/9/2021 5:48:44 AM	4/15/2021 12:51:00 PM
Galactosemia - Abnormal Newborn Screen	21-097-148120	4/7/2021 3:00:00 PM	4/9/2021 5:48:44 AM	4/15/2021 12:51:00 PM

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Galactosemia - Family History	21-097-148120	4/7/2021 3:00:00 PM	4/9/2021 5:48:44 AM	4/15/2021 12:51:00 PM
Galactosemia (GALT) DNA Pan,G1PUT Interp	21-097-148120	4/7/2021 3:00:00 PM	4/9/2021 5:48:44 AM	4/15/2021 12:51:00 PM

END OF CHART

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