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

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

Physician: Doctor, Example

Patient: Patient, Example

DOB	3/31/2023
Gender:	Male
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	00/00/0000 00:00

Galactosemia (GALT) Enzyme Activity and 9 Mutations

ARUP test code 0051175

Galac-1-Phos Uridyltransferase	25.0 U/g Hb (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

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

Galactosemia - Family History	Unknown
Galactosemia (GALT) DNA Pan,G1PUT Interp	See Note

Normal: This sample has normal galactose-1-phosphate uridyltransferase (GALT) activity and is negative for the seven common GALT gene mutations and 2 variants tested. Therefore this individual appears to be neither a carrier of, nor affected with, classic galactosemia.

This result has been reviewed and approved by Yuan Ji, Ph.D.

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	23-102-155942	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Galactosemia (GALT) DNA Pan,G1PUT Spec	23-102-155942	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Galactosemia (GALT) Allele 1	23-102-155942	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Galactosemia (GALT) Allele 2	23-102-155942	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Galactosemia - Ethnicity	23-102-155942	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Galactosemia - Symptoms	23-102-155942	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Galactosemia - Abnormal Newborn Screen	23-102-155942	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Galactosemia - Family History	23-102-155942	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Galactosemia (GALT) DNA Pan,G1PUT Interp	23-102-155942	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00

END OF CHART

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