

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

Physician: Doctor, Example

Patient: Patient, Example

3/10/2023
Male
01234567890ABCD, 012345
01234567890ABCD
00/00/0000 00:00

Galactosemia (GALT) Enzyme Activity and 9 Mutations

ARUP test code 0051175

Galac-1-Phos Uridyltransferase	9.9 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).			
	GenotypeGalac-1-Phos Uridyltransferase activity(umol/hr/g Hb)Classic galactosemia(G/G)Less than or equal to 0.7Duarte galactosemia(D/G)3.1-7.8Classic galactosemia carrier(G/N)6.5-16.2Duarte homozygous(D/D)6.4-16.5Duarte carrier(D/N)12.0-24.0Normal(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	S135L *			
Galactosemia (GALT) Allele 2	Negative			
Galactosemia - Ethnicity	Unknown			
Galactosemia - Symptoms	Unknown			
Galactosemia - Abnormal Newborn Screen	Unknown			

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

Unless otherwise indicated, testing performed at:



Galactosemia - Family History Unknown Galactosemia (GALT) DNA Pan,G1PUT Interp See Note Galactosemia Carrier; Reduced Enzyme Activity/ One Mutation Detected: This sample has partially reduced galactose-1-phosphate uridyltransferase (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 consultation is recommended. This result has been reviewed and approved by 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 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. Indentifiance: Autosomal recessive Jana Closenta III Caucastali, 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. 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.

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Unless otherwise indicated, testing performed at:



VERIFIED/REPORTED DATES					
Procedure	Accession	Collected	Received	Verified/Reported	
Galac-1-Phos Uridyltransferase	23-095-401360	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00	
Galactosemia (GALT) DNA Pan,G1PUT Spec	23-095-401360	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00	
Galactosemia (GALT) Allele 1	23-095-401360	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00	
Galactosemia (GALT) Allele 2	23-095-401360	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00	
Galactosemia - Ethnicity	23-095-401360	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00	
Galactosemia - Symptoms	23-095-401360	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00	
Galactosemia - Abnormal Newborn Screen	23-095-401360	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00	
Galactosemia - Family History	23-095-401360	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00	
Galactosemia (GALT) DNA Pan,G1PUT Interp	23-095-401360	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

Unless otherwise indicated, testing performed at: