

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

Physician: Doctor, Example

Patient: Patient, Example

DOB: 3/3/2025
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

13.5 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 **S135L ***

Galactosemia (GALT) Allele 2 Negative

Galactosemia - Ethnicity Unknown

Galactosemia - Symptoms Unknown

Galactosemia - Abnormal Newborn Screen Yes

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

Unless otherwise indicated, testing performed at:

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Galac-1-Phos Uridyltransferase	25-071-124311	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Galactosemia (GALT) DNA Pan,G1PUT Spec	25-071-124311	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Galactosemia (GALT) Allele 1	25-071-124311	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Galactosemia (GALT) Allele 2	25-071-124311	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Galactosemia - Ethnicity	25-071-124311	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Galactosemia - Symptoms	25-071-124311	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Galactosemia - Abnormal Newborn Screen	25-071-124311	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Galactosemia - Family History	25-071-124311	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Galactosemia (GALT) DNA Pan,G1PUT Interp	25-071-124311	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:

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: 25-071-124311
Patient Identifiers: 01234567890ABCD, 012345
Visit Number (FIN): 01234567890ABCD
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