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	Unknown
Gender:	Unknown
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	00/00/0000 00:00

Galactosemia, (GALT) 9 Mutations

ARUP test code 0051176

Galactosemia (GALT) DNA Panel Specimen	Whole Blood	
Galactosemia (GALT) Allele 1	**Q188R**	*
Galactosemia (GALT) Allele 2	**N314D**	*
Galactosemia - Ethnicity	Unknown	
Galactosemia - Symptoms	Unknown	
Galactosemia - Family History	Unknown	
Galactosemia (GALT) DNA Panel Interp	See Note	

One Mutation and One Duarte Variant: This sample is positive for one severe mutation and one Duarte (D) variant in the GALT gene, consistent with D/G variant galactosemia. This individual is not predicted to have classic galactosemia. Medical management should rely on clinical and biochemical findings. Genetic and metabolic consultations are recommended.

This result has been reviewed and approved by [redacted]

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

BACKGROUND INFORMATION: Galactosemia (GALT) 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 for classic galactosemia in Caucasian, varies in other populatons.
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 SENSTIVITY: Approaches 80 percent in Caucasians but reduced in other ethnic groups.
METHODOLOGY: Polymerase chain reaction followed by single nucleotide extension (SNE) and capillary electrophoresis.
ANALYTICAL SENSITIVITY: 99 percent for mutations listed.
LIMITATIONS: GALT gene mutations, other than the 9 targeted, will not be detected. Diagnostic errors can occur due to rare sequence variations.

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.

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Galactosemia (GALT) DNA Panel Specimen	23-065-103370	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Galactosemia (GALT) Allele 1	23-065-103370	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Galactosemia (GALT) Allele 2	23-065-103370	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Galactosemia - Ethnicity	23-065-103370	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Galactosemia - Symptoms	23-065-103370	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Galactosemia - Family History	23-065-103370	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Galactosemia (GALT) DNA Panel Interp	23-065-103370	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