ARUP LABORATORIES
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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	3/10/2023
Gender:	Male
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	Negative
Galactosemia (GALT) Allele 2	Negative
Galactosemia - Ethnicity	Hispanic
Galactosemia - Symptoms	Unknown
Galactosemia - Family History	Unknown
Galactosemia (GALT) DNA Panel Interp	See Note

Negative: This sample is negative for the seven common GALT gene mutations and two GALT variants tested. This individual may still be a carrier of, or affected with, galactosemia since not all GALT mutations are detected by this assay. Correlation with GALT enzyme activity is recommended. Genetic consultation is 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	23-094-402985	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Galactosemia (GALT) Allele 1	23-094-402985	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Galactosemia (GALT) Allele 2	23-094-402985	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Galactosemia - Ethnicity	23-094-402985	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Galactosemia - Symptoms	23-094-402985	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Galactosemia - Family History	23-094-402985	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Galactosemia (GALT) DNA Panel Interp	23-094-402985	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