

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

Physician: Doctor, Example

Patient: Patient, Example

DOB: 12/31/1978
Gender: Male
Patient Identifiers: 01234567890ABCD, 012345
Visit Number (FIN): 01234567890ABCD
Collection Date: 00/00/0000 00:00

Galactosemia (GALT) 9 Mutations, Fetal

ARUP test code 0051270

Galactosemia (GALT) DNA Pan, Fetal Spec Amniotic fluid

Galactosemia (GALT) Allele 1 Negative

Galactosemia (GALT) Allele 2 Negative

Galactosemia - Ethnicity Caucasian

Galactosemia - Symptoms No

Galactosemia - Family History No

Galactosemia (GALT) DNA Panel Interp See Note

Section 79-1 of New York State Civil Rights Law requires informed consent be obtained from patients (or their legal guardians) prior to pursuing genetic testing. These forms must be kept on file by the ordering physician. Consent forms for genetic testing are available at www.aruplab.com. Incidental findings are not reported unless clinically significant but are available upon request.

This result has been reviewed and approved by ██████████

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

Unless otherwise indicated, testing performed at:

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 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: 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.

Maternal Contamination Study Fetal Spec

Fetal Cells

Single fetal genotype present; no maternal cells present. Fetal and maternal samples were tested using STR markers to rule out maternal cell contamination.

Maternal Contam Study, Maternal Spec

whole Blood

For quality assurance purposes, ARUP Laboratories will confirm the above result at no charge following delivery. Order Confirmation of Fetal Testing and include a copy of the original fetal report (or the mother's name and date of birth) with the test submission. Please contact an ARUP genetic counselor at (800) 242-2787 extension 2141 prior to specimen submission.

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

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Galactosemia (GALT) DNA Pan, Fetal Spec	22-346-400885	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Galactosemia (GALT) Allele 1	22-346-400885	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Galactosemia (GALT) Allele 2	22-346-400885	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Galactosemia - Ethnicity	22-346-400885	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Galactosemia - Symptoms	22-346-400885	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Galactosemia - Family History	22-346-400885	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Galactosemia (GALT) DNA Panel Interp	22-346-400885	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Maternal Contamination Study Fetal Spec	22-346-400885	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Maternal Contam Study, Maternal Spec	22-346-400885	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: 22-346-400885
Patient Identifiers: 01234567890ABCD, 012345
Visit Number (FIN): 01234567890ABCD
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