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** | 5/14/2018 |
| **Gender:** | Unknown |
| **Patient Identifiers:** | 01234567890ABCD, 012345 |
| **Visit Number (FIN):** | 01234567890ABCD |
| **Collection Date:** | 00/00/0000 00:00 |

## Galactosemia (GALT) 9 Mutations, Fetal

ARUP test code 0051270

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|---|---|---|
| Galactosemia (GALT) DNA Pan, Fetal Spec | Amniotic fluid | |
| Galactosemia (GALT) Allele 1 | Q188R | * |
| Galactosemia (GALT) Allele 2 | L195P | * |
| Galactosemia - Ethnicity | Caucasian | |
| Galactosemia - Symptoms | Yes | |
| Galactosemia - Family History | Yes | |
| Galactosemia (GALT) DNA Panel Interp | See Note | |

Two Mutations: This sample is positive for two GALT gene mutations. Thus, this individual is predicted to be affected with classic galactosemia. Life-long dietary restriction of lactose and galactose is necessary. Genetic and metabolic consultations are strongly recommended.

This result has been reviewed and approved by [redacted]

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

| | |
|---|---|
| | BACKGROUND INFORMATION: Galactosemia (GALT) 9 Mutations<br><br>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.<br>INCIDENCE: Approximately 1 in 30,000 to 60,000 for classic galactosemia in Caucasian, varies in other populatons.<br>INHERITANCE: Autosomal recessive.<br>PENETRANCE: 100 percent for severe GALT mutations.<br>CAUSE: Mutations in the GALT gene.<br>MUTATIONS TESTED: Seven GALT gene mutations (Q188R, S135L, K285N, T138M, L195P, Y209C, and IVS2-2 A>G) and two variants (N314D and L218L).<br>CLINICAL SENSTIVITY: Approaches 80 percent in Caucasians but reduced in other ethnic groups.<br>METHODOLOGY: Polymerase chain reaction followed by single nucleotide extension (SNE) and capillary electrophoresis.<br>ANALYTICAL SENSITIVITY: 99 percent for mutations listed.<br>LIMITATIONS: GALT gene mutations, other than the 9 targeted, will not be detected. Diagnostic errors can occur due to rare sequence variations.<br><br>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.<br><br>Counseling and informed consent are recommended for genetic testing. Consent forms are available online. |
| Maternal Contamination Study Fetal Spec | Fetal Cells<br><br>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 |

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

VERIFIED/REPORTED DATES

| Procedure | Accession | Collected | Received | Verified/Reported |
|---|---|---|---|---|
| Galactosemia (GALT) DNA Pan, Fetal Spec | 23-026-116760 | 00/00/0000 00:00 | 00/00/0000 00:00 | 00/00/0000 00:00 |
| Galactosemia (GALT) Allele 1 | 23-026-116760 | 00/00/0000 00:00 | 00/00/0000 00:00 | 00/00/0000 00:00 |
| Galactosemia (GALT) Allele 2 | 23-026-116760 | 00/00/0000 00:00 | 00/00/0000 00:00 | 00/00/0000 00:00 |
| Galactosemia - Ethnicity | 23-026-116760 | 00/00/0000 00:00 | 00/00/0000 00:00 | 00/00/0000 00:00 |
| Galactosemia - Symptoms | 23-026-116760 | 00/00/0000 00:00 | 00/00/0000 00:00 | 00/00/0000 00:00 |
| Galactosemia - Family History | 23-026-116760 | 00/00/0000 00:00 | 00/00/0000 00:00 | 00/00/0000 00:00 |
| Galactosemia (GALT) DNA Panel Interp | 23-026-116760 | 00/00/0000 00:00 | 00/00/0000 00:00 | 00/00/0000 00:00 |
| Maternal Contamination Study Fetal Spec | 23-026-116760 | 00/00/0000 00:00 | 00/00/0000 00:00 | 00/00/0000 00:00 |
| Maternal Contam Study, Maternal Spec | 23-026-116760 | 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