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PATIENT REPORT

Patient Age/Sex: 36 years Female

Specimen Collected: 5/6/2025 08:16 MDT

Galactosemia (GALT) 9 Mutations, Fetal | **Received: 5/6/2025 08:16 MDT** | **Report/Verified: 5/7/2025 19:12 MDT**

Procedure	Result	Units	Reference Interval
Galactosemia (GALT) DNA Pan, Fetal Spec	Cultured Amnio		
Galactosemia (GALT) Allele 1	**Q188R** *		
Galactosemia (GALT) Allele 2	**L195P** *		
Galactosemia -Ethnicity	Caucasian		
Galactosemia -Symptoms	Unknown		
Galactosemia -Family History	Yes		
Galactosemia (GALT) DNA Panel Interp	See Note f1 i1		
Maternal Contamination Study Fetal Spec	Fetal Cells f2		
Maternal Contam Study,Maternal Spec	Whole Blood		

Result Footnote

f1: Galactosemia (GALT) DNA Panel Interp

Two Mutations: According to information provided to ARUP, the mother of this fetus carries the GALT gene variant Q188R and the father of this fetus carries the GALT gene variant L195P. This prenatal sample is positive for the two familial GALT gene variants. Thus, this fetus is predicted to be affected with classic galactosemia. Life-long dietary restriction of lactose and galactose is necessary in individuals with classic galactosemia. Genetic consultation is recommended.

This result has been reviewed and approved by [redacted]

f2: Maternal Contamination Study Fetal Spec

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

Test Information

i1: Galactosemia (GALT) DNA Panel Interp

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.

*=Abnormal, #=Corrected, C=Critical, f=Result Footnote, H-High, i-Test Information, L-Low, t-Interpretive Text, @=Performing lab

Test Information

i1: Galactosemia (GALT) DNA Panel Interp

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.