

Specimen Collected: 29-Oct-25 14:15

Galactosemia (GALT) 9 Mutations, Fetal | Received: 29-Oct-25 14:15 Report/Verified: 04-Nov-25 10:54

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

Indication: Prenatal diagnosis

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. Both the Q188R and the L195P familial variants were identified in this prenatal sample. Thus, this fetus is predicted to be affected 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 populations.

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

Unless otherwise indicated, testing performed at:

ARUP Laboratories

500 Chipeta Way, Salt Lake City, UT 84108

Laboratory Director: Jonathan R. Genzen, MD, PhD

ARUP Accession: 25-302-900288

Report Request ID: 20887774

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Patient Age/Sex: 22 years Female

Test Information

il: 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 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.

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