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PATIENT REPORT

Patient Age/Sex: 21 years Female

Specimen Collected: 29-Oct-25 14:14

Galactosemia (GALT) 9 Mutations |Received: 29-Oct-25 14:14 Report/Verified: 30-Oct-25 11:10

Procedure	Result	Units	Reference Interval
Galactosemia (GALT) DNA Panel Specimen	Whole Blood		
Galactosemia (GALT) Allele 1	**Q188R** *		
Galactosemia (GALT) Allele 2	**N314D** *		
Galactosemia -Ethnicity	Caucasian		
Galactosemia -Symptoms	Yes		
Galactosemia -Family History	Unknown		
Galactosemia (GALT) DNA Panel Interp	See Note f1 i1		

Result Footnote

f1: Galactosemia (GALT) DNA Panel Interp

One Mutation and One Duarte Variant: This sample is positive for one severe mutation and one Duarte (D) variant in the GALT gene, consistent with D/G variant galactosemia. This individual is not predicted to have classic galactosemia. Medical management should rely on clinical and biochemical findings. Genetic and metabolic consultations are recommended.

This result has been reviewed and approved by [redacted]

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.
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.

Test Information

i1: Galactosemia (GALT) DNA Panel Interp

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.