
Lab Dept: **Anatomic Pathology**

Test Name: **GALT FULL GENE ANALYSIS**

General Information

Lab Order Codes: GALTZ

Synonyms: GALT; Galactose-I-phosphate uridyltransferase Sequencing

CPT Codes: 81406 – GALT (galactose-I-phosphate uridyltransferase) full gene sequence

Test Includes: All detected alterations will be evaluated according to the American College of Medical Genetics and Genomics (AMCG) recommendations.(1) Variants will be classified based on known, predicted, or possible pathogenicity and reported with interpretive comments detailing their potential or known significance.

Logistics

Test Indications: Identifying mutations in individuals who test negative for the common mutations and who have a biochemical diagnosis of galactosemia or galactose-1 phosphate uridyltransferase activity levels indicative of carrier status.

Lab Testing Sections: Chemistry - Sendouts

Referred to: Mayo Medical Laboratories (Mayo Test: GALTZ)

Phone Numbers: MIN Lab: 612-813-6280

STP Lab: 651-220-6550

Test Availability: Daily, 24 hours

Turnaround Time: 14 – 20 days, performed weekly

Special Instructions: Submit a [Molecular Genetics Congenital Inherited Diseases Information form](#) and submit to lab to be sent with the sample.

Specimen

Specimen Type: Whole blood

Container:	Preferred: Lavender top (EDTA) tube Alternate: All anticoagulants are acceptable
	Note: Blood spots are also acceptable. Contact the lab for more information. (Mayo Supply T493)
Draw Volume:	3 mL (Minimum: 1 mL) blood
Processed Volume:	Same as Draw Volume
Collection:	Routine blood collection
Special Processing:	Lab Staff: Do Not centrifuge. Specimen should remain in original collection tube. Store and ship at ambient- preferred (refrigerated or frozen are ok) temperatures. Include Molecular Genetics Congenital Inherited Diseases Information form . Forward promptly. Specimen is preferred to arrive within 96 hours of draw.
Patient Preparation:	A previous bone marrow transplant from an allogenic donor will interfere with testing. Consult with Mayo for instructions for testing patients who have received a bone marrow transplant.
Sample Rejection:	Mislabeled or unlabeled specimens

Interpretive

Reference Range: An interpretive report will be provided.

Critical Values: N/A

Limitations: A small percentage of individuals who are carriers or have a diagnosis of galactosemia may have a mutation that is not identified by the methods described above (eg, large genomic deletions, promoter mutations). The absence of a mutation(s), therefore, does not eliminate the possibility of positive carrier status or the diagnosis of galactosemia. For carrier testing, it is important to first document the presence of galactose-1-phosphate uridylyltransferase (GALT) gene mutation in the affected family member.

In some cases, DNA alterations of undetermined significance may be identified. Rare polymorphisms exist that could lead to false-negative or false-positive results. If results obtained do not match the clinical and biochemical findings, additional testing should be considered.

Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Errors in our interpretation of results may occur if information given is inaccurate or incomplete. This is not recommended for carrier screening or diagnosis in individuals with a positive newborn screen.

Methodology: Polymerase chain reaction (PCR) amplification/DNA sequencing

References: [Mayo Medical Laboratories](#) Web Page (April 2016)

Updates: 4/18/2018: Updated container options.