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:	Lavender top (EDTA) tube Alternate: Yellow top (ACD) tube

Draw Volume:	3 mL (Minimum: 1 mL) blood
Processed Volume:	Same as Draw Volume
Collection:	Routine venipuncture
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.
Patient Preparation:	None
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 uridyltransferase (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 (November 2016)

Updates:

11/21/2006: MML has removed the upper normal range value for this test. The reference range previously was stated as 18.5-28.5 U/g of hemoglobin.
2/14/2014: Test down at MML, temporary move to ARUP.
4/28/2014: Test back up at MML with new method, previously listed as enzymatic. Please note new reference range.