Lab Dept:	**Anatomic Pathology**
Test Name:	**GALACTOSEMIA GENE ANALYSIS (14 MUTATION PANEL)**

General Information

Lab Order Codes:	GAL14
Synonyms:	GALT; Galactose-I-phosphate uridyltransferase Mutation Panel
CPT Codes:	81401 – GALT (galactose-I-phosphate uridyltransferase) common variants
Test Includes:	Tests for the presence of the following 14 mutations in the GALT gene: -119_-116delGTCA, D98N, S135L, T138M, M142K, F171S, Q188R, L195P, Y209C, K285N, N314D, Q344K, c.253-2A>G, and 5 kb deletion.

Logistics

Test Indications:	Second-tier test for confirming a diagnosis of galactosemia (indicated by enzymatic testing or newborn screening) Carrier testing family members of an affected individual of known genotype (has mutations included in the panel) Resolution of Duarte variant and Los Angeles (LA) variant genotypes.
Lab Testing Sections:	Chemistry - Sendouts
Referred to:	Mayo Medical Laboratories (Mayo Test: GAL14)
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. Results should be interpreted within the context of biochemical results.
Critical Values:	N/A
Limitations:	This assay will not detect all of the mutations of galacosemia. Therefore, the absence of a detectable mutation does not rule out the possibility that an individual is a carrier of or affected with this desease. 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. Rare polymorphisms exist that could lead to false-negative or false-positive results. If results obtained do not match the clinical findings, additional testing should be considered. In rare cases, DNA alterations of undetermined significance may be identified. A previous bone marrow transplant from an allogenic donor will interfere with testing. Call Mayo Medical Laboratories for instructions for testing patients who have received a bone marrow transplant. Many disorders may present with symptoms similar to those associated with galactosemia. Therefore, biochemical testing is recommended to establish the diagnosis prior to DNA analysis.
Methodology:	Multiplex Polymerase Chain Reaction (PCR) – Based Assay Utilizing the Agena Mass ARRAY Platform

References: Mayo Medical Laboratories Web Page (November 2016)