Lab Dept: Chemistry

Test Name: GALACTOKINASE, BLOOD

General Information

Lab Order Codes:	GALK
Synonyms:	Galactokinase (GALK) Deficiency; Galactosemia
CPT Codes:	82759 – Galactokinase, RBC
Test Includes:	Galactokinase level reported in nmol/h/mg of hemoglobin.

Logistics

Test Indications:	Diagnosis of galactokinase deficiency, the second most common cause of galactosemia.
Lab Testing Sections:	Chemistry - Sendouts
Referred to:	Mayo Medical Laboratories (Mayo Test: GALK)
Phone Numbers:	MIN Lab: 612-813-6280 STP Lab: 651-220-6550
Test Availability:	Daily, 24 hours
Turnaround Time:	8 - 15 days, performed weekly on Mondays
Special Instructions:	N/A

Specimen

Specimen Type:	Whole blood
Container:	Lavender top (EDTA) tube Alternate: Green top (NaHep), Green top (LiHep) or Yellow top (ACD) tube
Draw Volume:	4 mL (Minimum: 2 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 refrigerated temperatures. Forward promptly.
Patient Preparation:	None
Sample Rejection:	Mislabeled or unlabeled specimens; gross hemolysis

Interpretive

Reference Range:	≥0.7 nmol/h/mg An interpretive report will be provided.
Critical Values:	N/A
Limitations:	This assay is not useful for monitoring dietary compliance. See Galactose-1-Phosphate, Erythrocytes This assay will not detect epimerase (GALE) deficiency or galactose-1-phosphate uridyltransferase (GALT) deficiency. It is important to notify the laboratory if the patient has been transfused prior to specimen collection. The results of testing performed in erythrocytes are invalid following a transfusion, including analysis of enzymes, biochemical phenotyping, or galactose-1-phosphate. The most common cause of galactosemia is GALT. In most cases, GALT deficiency should be ruled out prior to evaluating for GALK deficiency.
Methodology:	Enzyme Reaction followed by Liquid Chromatography-Tandem Mass Spectrometry (LC-MS/MS)
References:	Mayo Medical Laboratories Web Page (November 2016)