Lab Dept: Chemistry

Test Name: UDP-GALACTOSE 4' EPIMERASE (GALE),

BLOOD

General Information

Lab Order Codes: GALE

Synonyms: Galactosemia; Uridine diphosphate galactose-4-epimerase

CPT Codes: 82542 – Column chromatography, includes mass spectrophotometry, if

performed, non-drug analytes not elsewhere specified, qualitative or

quantitative, each specimen

Test Includes: UDP-Galactose 4' epimerase level reported in nmol/h/mg of

hemoglobin.

Logistics

Test Indications: Diagnosis UDP-galactose 4' epimerase deficiency.

Lab Testing Sections: Chemistry - Sendouts

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

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 Wednesdays

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: 5 mL (Minimum: 2 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 refrigerated temperatures. Forward

promptly.

Patient Preparation: None

Sample Rejection: Mislabeled or unlabeled specimens; gross hemolysis

Interpretive

Reference Range: >=3.5 nmol/h/mg of hemoglobin

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 uridyltransfererase (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 December 2017

Updates: 12/14/2017: Updated CPT and reference range.

2/6/2018: Updated reference range, previously listed as >5.0 nmol/h/mg

hemoglobin