Galactosemia: Galactose-1-Phosphate, Quantitative, Red Blood Cells

Test Code: GP
Turnaround time: 7 days - 10 days
CPT Codes: 84378 x1

Condition Description

Galactosemia is an autosomal recessive disorder of galactose metabolism that often presents in the newborn period with poor suck, vomiting, diarrhea, bleeding diathesis, lethargy, jaundice, and sepsis. If left untreated, an individual may progress to irreversible liver disease and mental retardation. The prevalence of galactosemia is approximately 1 in 30,000 newborns.

Most often galactosemia is caused by a deficiency of the galactose-1-phosphate uridylyltransferase (GALT) enzyme, due to mutations in the GALT gene located on chromosome 9p13. The galactose-1-phosphate uridylyltransferase (GALT) enzyme catalyzes the production of glucose-1-phosphate and UDP-galactose from gal-1-P and UDP-glucose. Deficiency results in the accumulation of galactose-1-phosphate, galactose, and urine galactitol.

There are other biochemical defects in the galactosemia pathway, such as epimerase and galactokinase deficiencies. Galactokinase deficiency should be considered in patients with cataracts and galactosemia who have normal gal-1-P and normal GALT activity. Epimerase deficiency should be considered in patients with liver disease, sensorineural deafness, failure to thrive, and elevated gal-1-P but normal GALT activity.

Please click here for the GeneReviews summary on this condition.


Indications

This test is indicated for:

- Monitoring the effectiveness of therapy in individuals with a biochemical diagnosis of classical, Duarte, or epimerase deficient galactosemia.

Please note that this test alone is NOT recommended for galactosemia screening or diagnosis in persons with a positive newborn screen. Please refer to the Galactosemia Comprehensive Panel (GS).

Methodology

Measures the concentration of galactose-1-phosphate in red blood cells, using a coupled enzymatic reaction detected by a spectrophotometric assay.

Detection

Results should be interpreted in the context of the patient’s biochemical diagnosis and treatment plan.

Reference Range

Normal: 0-1.0 mg/100 ml red blood cells.

Classic Galactosemia Therapeutic range: <3.5 mg/100 ml red blood cells.

Specimen Requirements

Additional Specimen Collection/Handling Instructions Required for this Test
If whole blood specimen cannot be received by EGL within 24 hours of collection, send 2-3 ml washed RBC’s. (Click here for protocol.)

Ship washed RBC’s frozen on dry ice with overnight delivery. Please indicate on the requisition that the specimen type is washed RBC’s.

Type: Whole Blood

Specimen Requirements:

In sodium heparin (green top) tube: 3-5 ml

Specimen Collection and Shipping: Refrigerate until time of shipment. Ship sample overnight at room temperature for receipt at EGL within 24 hours of collection.

Related Tests

- Galactitol (test code GL) - biochemical screening test for diagnosis and monitoring or patients with galactosemia.
- Galactosemia: GALT Enzyme Activity (test code GT) - Gal-1-P uridylyltransferase enzyme activity.
- Galactosemia: Comprehensive Panel (test code GS) - GALT activity and GAL-1-P.
- Galactosemia: Nonclassic Galactosemia Panel (GALK & GALE Enzymes and Galactose-1-Phosphate) (test code BGKEP)
- Galactosemia: (Galactokinase Deficiency): GALK Enzyme Activity (test code GK).