Galactosemia, Classic: Panel (GALT Enzyme Activity & Galactose-1-Phosphate Quantitative), Red Blood Cells

Test Code: GS
Turnaround time: 7 days - 14 days
CPT Codes: 82775 x1, 84311 x1, 84378 x1, 85018 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. Approximately 10% of individuals also have cataracts. If left untreated, an individual may progress to irreversible liver disease and mental retardation.

Many affected infants are detected through state newborn screening programs near the time that they may become clinically symptomatic. For this reason, lactose should be removed from the diet of an affected infant while results of biochemical tests are pending. Infants born with classic galactosemia require lifelong dietary restriction of galactose, while variant galactosemia patients may not. Despite current treatment, affected individuals remain at risk for developmental delays, speech dyspraxia, and premature ovarian failure (females). The prevalence of galactosemia is approximately 1 in 30,000 newborns.

Galactosemia is most often caused by a deficiency of the galactose-1-phosphate uridyltransferase (GALT) enzyme, which catalyzes the production of glucose-1-phosphate and UDP-galactose from gal-1-P and UDP-glucose. The deficiency results in the accumulation of galactose-1-phosphate, galactose, and urine galactitol.

Comprehensive testing (GS) for galactosemia is accomplished by determining the GALT activity and the concentration of galactose-1-phosphate (gal-1-p).

Please note that if GALT gene sequencing (test code SG) is needed due to results from this test, DNA can be obtained from the sodium heparin (green top) tube submitted for this test.

There are two other enzymes in this biochemical pathway that when deficient can cause nonclassic (GALK) and UDP-galactose 4-epimerase (GALE). GALK and GALE deficiencies should be considered in patients who have an elevated gal-1-p concentration but 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.


Click here for the GeneReviews summary on this condition.

Genes

GALT

Indications

This test is indicated in the case of:

- Follow up to abnormal newborn screening results
- Clinical symptoms of galactosemia
- Testing for siblings of an individual with galactosemia

For carrier screening, please order Galactosemia: Carrier Panel (GR).

Methodology

GALT enzyme activity is performed using liquid chromatography – tandem mass spectrometry. Gal-1-P concentration is performed using a coupled enzymatic reaction detected by a spectrophotometric assay.

Detection

Diagnosis of galactosemia is made by measurement of GALT activity. Detection of reduced GALT activity is diagnostic for GALT deficient galactosemia.

Reference Range

Ranges of Gal-1-P uridylytransferase (GALT) activity (µmol/Hr/g Hb):

- Normal (N/N) 22.1 – 46.3
- Galactosemia carrier (G/N) 8.5 – 23.0
- Duarte galactosemia (D/G) 1.9 – 13.4
Classic galactosemia (G/G) 0 – 0.3

Ranges for Galactose-1-Phosphate (RBC’s):

- Normal: 0-1.0 mg/100 ml red blood cells
- Classic Galactosemia Therapeutic Range: <3.5 mg/100 mg 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 is washed RBC’s. Send an additional 1-2 ml of whole blood in an EDTA (purple top) tube. (Do not wash and do not freeze the whole blood. Refrigerate until shipment. Send at room temperature with overnight delivery.)

**Type: Whole Blood**

Specimen Requirements:

In sodium heparin (green top) tube:

Infants (Related Tests)

- Galactitol analysis is available and is the biochemical screening test for diagnosis and monitoring of patients with galactosemia.
- Galactosemia Carrier Testing is available through GALT enzyme activity.
- If the proband has a biochemical phenotype of galactosemia, GALT full gene sequencing is offered.
- GALT deletion/duplication analysis is offered when mutations are not detected by full gene sequencing.
- Prenatal testing is available by molecular methods. Please contact the laboratory genetic counselor to determine the availability of prenatal testing.