Galactosemia: Galactitol Quantitative, Urine

Test Code: GL
Turnaround time: 7 days - 10 days
CPT Codes: 82570 x1, 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. Galactosemia is most often caused by a deficiency of the galactose-1-phosphate uridylyltransferase (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.

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. Urinary galactitol excretion is increased in all forms of galactosemia. Quantitation of urinary galactitol is another biochemical screening test for monitoring patients with galactosemia.


Please click here for the GeneReviews summary on this condition.

Indications

This test is indicated in the case of:

- Monitoring effectiveness of therapy in individuals with biochemical diagnosis of classical, Duarte, or variant galactosemia due to galactokinase or epimerase deficiencies.

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 Comprehensive Galactosemia Panel (GS).

Methodology

Quantitation of galactitol in urine, using gas chromatography/mass spectrometry (GCMS).

Detection

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

Reference Range

Click here for reference range.

Specimen Requirements

Additional Specimen Collection/Handling Instructions Required for this Test

For diagnostic purposes, specimens should ideally be collected during time of acute illness as abnormal metabolite levels may decrease, sometimes to near normal concentrations, when patient is well.

Type: Urine

Specimen Requirements:

In a clean container without preservatives: 2-5 ml. Freeze.

Fasting or first void sample is preferable.

Specimen Collection and Shipping: Ship frozen sample on dry ice with overnight delivery.

Related Tests

- Galactose-1-Phosphate (test code GP) - 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)
- Galactosemia (Epimerase Deficiency): GALE Enzyme Activity (test code EP)