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 uridyltransferase (GALT) enzyme, due to mutations in the GALT gene located on chromosome 9p13.

Galactosemia is detected by (GALT) enzyme activity. Individuals with galactosemia have decreased enzyme levels.

Please click here for Gene Reviews Clinical Summary.


**Indications**

This test is indicated for:

- Individuals with a molecular diagnosis of galactosemia or for GALT carrier status. For comprehensive carrier testing, refer to Galactosemia: Carrier Panel (GR).

Please note that this test is NOT intended to provide a comprehensive diagnosis. For clinical diagnosis, refer to Galactosemia: Comprehensive Panel (GS).

**Methodology**

GALT enzyme activity is performed using liquid chromatography – tandem mass spectrometry.

**Detection**

Detection of classic galactosemia is highly reliable by measurement of GALT activity.

**Please note that enzyme characterization by isozyme analysis has been discontinued.**

Determination of the Duarte variant will require sequencing of the GALT gene to identify the Duarte associated changes.

**Reference Range**

Ranges of Gal-1-P uridyltransferase (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

**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 - biochemical screening test for diagnosis and monitoring or patients with galactosemia.
- Galactose-1-Phosphate - biochemical screening test for diagnosis and monitoring of patients with galactosemia.
- Galactosemia: Comprehensive Panel - GALT activity and GAL-1-P.
- Galactosemia: Carrier Testing - GALT enzyme activity.
- Galactosemia: GALT Full Gene Sequencing - biochemical phenotype of galactosemia.
- Custom Diagnostic Mutation Analysis - family members of individuals with GALT mutations.
- GALT deletion/duplication analysis is available in cases when a mutation is not detected by full gene sequencing.
- Prenatal testing is available by molecular methods. Please contact a laboratory genetic counselor to determine the availability of prenatal testing.