Galactosemia, Classic (Galactose-1-Phosphate Uridyltransferase Deficiency): GALT Enzyme Activity, Red Blood Cells

Test Code: GT  
Turnaround time: 7 days - 10 days  
CPT Codes: 82775 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. 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):

<table>
<thead>
<tr>
<th>Condition</th>
<th>Activity (µmol/Hr/g Hb)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Normal (N/N)</td>
<td>22.1 – 46.3</td>
</tr>
<tr>
<td>Galactosemia carrier (G/N)</td>
<td>8.5 – 23.0</td>
</tr>
<tr>
<td>Duarte galactosemia (D/G)</td>
<td>1.9 – 13.4</td>
</tr>
<tr>
<td>Classic galactosemia (G/G)</td>
<td>0 – 0.3</td>
</tr>
</tbody>
</table>

Specimen Requirements

Submit only 1 of the following specimen types

Type: Washed Blood Cells

Specimen Requirements:
Sodium Heparin
3-5 ml
Washed Red Blood Cell Preparation:
Step 1: Centrifuge the whole blood at 3000rpm (1800rcf) for 5 minutes.
Step 2: Remove plasma and buffy coat layer.
Step 3: Resuspend the red cells in normal saline (0.9% NaCl) with approximately 2 times the volume of the red cells, and invert the tube to mix.
Step 4: Centrifuge for 5 minutes at 2000 rpm and discard the supernatant.
Step 5: Repeat Steps 3 & 4 twice for a total of 3 washes or until the supernatant is clear.
Step 6: Completely remove the supernatant and freeze the red cells at -20°C, preferably at -80°C.

Specimen Collection and Shipping:
Ship frozen

Disclaimer: This information is confidential and subject to change without notice. It may not be reproduced in whole or part unless authorized in writing by an authorized EGL representative.
Type: Whole Blood (Sodium Heparin)

Specimen Requirements:
Sodium Heparin (Green Top)
3-5 ml

Specimen Collection and Shipping:
Ship sample at room temperature for receipt at EGL within 24 hours of collection. Do not refrigerate or freeze. Not accepted on Saturday. (Late Friday collections may be stored at room temperature over the weekend for Monday receipt.)

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.