**Galactosemia**

Newborn screening (NBS) panels in all 50 states include galactosemia. For patients identified through NBS as at risk for galactose-1-phosphate uridylyltransferase (GALT) deficiency, EGL Genetics recommends measuring the activity of red blood cell (RBC) GALT and the metabolite galactose-1-phosphate (GAL-1-P). If deficient GALT activity (the most common form of galactosemia) and/or elevated Gal-1-P are detected, confirmatory molecular analysis by GALT gene sequencing and testing for the common 5kb deletion can be initiated using DNA extracted from the same sample, upon request.

If NBS reveals normal GALT activity but elevated galactose, EGL Genetics offers enzymatic panels to detect deficiencies in galactokinase (GALK) activity. Confirmatory molecular testing is also recommended to identify mutations in patients diagnosed with either enzymatic deficiency.

NBS methods vary by state, and may not detect all defects in galactose metabolism. If galactosemia is suspected, particularly a variant form, clinical enzyme analysis and molecular confirmation is available for patients of any age. The combination of biochemical and molecular testing assures an almost 100% detection rate for galactosemia.

If a diagnosis of galactosemia is made, clinical monitoring is available through measurement of RBC GAL-1-P and urinary galactitol levels. EGL Genetics can interpret these results in the context of the individual’s known enzyme deficiency.

EGL Genetics also offers a targeted CGH array with deletion/duplication analysis of the GALT gene, when mutations are not detected by gene sequencing. Combined enzymatic and full-gene sequencing or if appropriate, targeted sequencing analysis for carrier testing of family members, is also recommended to provide families with reproductive risk information.

<table>
<thead>
<tr>
<th>Test Code</th>
<th>Test Name</th>
<th>CPT®®® Codes</th>
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<tr>
<td>GL</td>
<td>Galactosemia: Galactitol Quantitative, Urine</td>
<td>82570 (x1), 84378 (x1)</td>
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<td>GR</td>
<td>Galactosemia, Classic: Carrier Testing (GALT Enzyme Activity), Red Blood Cells</td>
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<td>GS</td>
<td>Galactosemia: Classic Galactosemia Panel</td>
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<td>GT</td>
<td>Galactosemia: GALT Enzyme Activity</td>
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<td>GP</td>
<td>Galactose-1-Phosphate Profile</td>
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<td>GK</td>
<td>Galactosemia (Galactokinase Deficiency): GALK Enzyme Activity</td>
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<td>SG</td>
<td>Galactosemia: GALT Full Gene Sequencing</td>
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<td>DGALT</td>
<td>Galactosemia: GALT Gene Deletion/Duplication</td>
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<td>JU</td>
<td>Galactosemia (Epimerase): GALE Full Gene Sequencing</td>
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<td>JV</td>
<td>Galactosemia (Epimerase): GALE Gene Deletion/Duplication</td>
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<td>IQ</td>
<td>Galactosemia (Galactokinase Deficiency): GALK1 Full Gene Sequencing</td>
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<td>JA</td>
<td>Galactosemia (Galactokinase Deficiency): GALK1 Gene Deletion/Duplication</td>
<td>81228 (x1)</td>
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</tbody>
</table>

**CPT® is a registered trademark of the American Medical Association.**
Galactosemia

Galactosemia Panel
(GALT Enz., Gal-1-P)
Comprehensive 
Report Issued 
(Molecular and Biochemical)

GALE Enzyme 
Analysis*
GALK Enzyme 
Analysis*

GALT Sequencing and 
5kb Deletion Analysis

Biochemical 
Report Issued

NBS: ↓ GALT Enz. 
(Total Gal Not Determined)

If requested, 
full gene sequencing 
will be initiated 
from the same sample

Galactosemia Panel 
(GALT Enz., Gal-1-P)

GALT Enz. <25%, 
nl or ↑ Gal-1-P

Biochemical 
Report Issued

GALT Enz. >25%, 
nl Gal-1-P

Biochemical 
Report Issued

NBS: nl GALT Enz. 
(↑ Total Gal)

If GALT enz. is nl with nl or 
↑ Gal-1-P and galactosemia 
is still suspected

If requested, 
full gene sequencing 
will be initiated 
from the same sample

Gal-1-P

nl GALT Enz., 
↑ Gal-1-P

Biochemical 
Report Issued

nl GALT Enz., 
nl Gal-1-P

Biochemical 
Report Issued

GALE Enzyme 
Analysis*

GALK Enzyme 
Analysis*

comprehensive 
report issued 
(Molecular and Biochemical)

NBS: nl GALT Enz. 
(↑ Total Gal)

If GALT enz. is nl with nl or 
↑ Gal-1-P and galactosemia 
is still suspected

If requested, 
full gene sequencing 
will be initiated 
from the same sample

GALE or GALK 
Sequencing

Comprehensive 
Report Issued 
(Molecular and Biochemical)

*These tests may be ordered as a panel.

For more information about EGL Genetics:

CALL 470.378.2200
WEB eglgenetics.com

About EGL Genetics
EGL Genetics specializes in genetic diagnostic testing, with nearly 50 years of clinical experience and board-certified laboratory directors and genetic counselors reporting out cases. EGL Genetics offers a combined 1000 molecular genetics, biochemical genetics, and cytogenetics tests under one roof and custom testing for all medically relevant genes, for domestic and international clients.

Equally important to improving patient care through quality genetic testing is the contribution EGL Genetics makes back to the scientific and medical communities. EGL Genetics is one of only a few clinical diagnostic laboratories to openly share data with the NCBI freely available public database ClinVar (>35,000 variants on >1700 genes) and is also the only laboratory with a free online database (EmVClass), featuring a variant classification search and report request interface, which facilitates rapid interactive curation and reporting of variants.

EGL Genetics Galactosemia Diagnosis Algorithm