

## Galactosemia (GALT)

### Use

Quantitative determination of total galactose (galactose and galactose-1-phosphate) in blood specimens dried on filter paper as an aid in screening newborns for classic galactosemia (GALT), galactokinase deficiency (GALK), and galactose epimerase deficiency (GALE). Newborns identified as receiving lactose-free formula also undergo GALT enzyme testing.

### Clinical Significance

Galactosemia is an autosomal recessive disorder that is characterized by elevated concentrations of galactose in the blood resulting from the absence or dysfunction of any of the three enzymes responsible for the transformation of galactose to glucose, i.e., D-galactose-1-phosphotransferase, D-galactose-1-phosphate-uridyltransferase, or UDP-glucose-4-epimerase. If not diagnosed and treated within the newborn period, galactosemia can lead to diarrhea, dehydration, jaundice, hepatic failure, hypoglycemia, cataracts, developmental retardation and death within a few weeks. Sepsis due to *Escherichia coli* seems to be particularly frequent among galactosemic neonates and is usually the cause of death. Treatment of the disease consists of withdrawal of all foods containing lactose and galactose from the diet.

Further information and ACT Sheets can be found at the OSDH Newborn Screening Program [website](#).

### Methodology

- GSP solid phase fluoroimmunoassay (screen) - The GSP Total Galactose Neonatal kit quantitatively measures total galactose, i.e., both galactose and galactose-1-phosphate, in dried blood spot specimens based on a fluorescent galactose oxidase method.
- Semi-quantitative enzymatic assay (reflex) - Specimens with elevated total galactose are reflexed to analysis of galactose-1-phosphate uridyl transferase (GALT) enzyme activity.

### Specimen Type

See [Guidance for Collection of NBS Dried Blood Spots](#)

### Minimum Volume/Size

See [Guidance for Collection of NBS Dried Blood Spots](#)

### Collection Instructions

See [Guidance for Collection of NBS Dried Blood Spots](#)

### Common Causes for Rejection

See [Guidance for Collection of NBS Dried Blood Spots](#)

### Shipping

See [Guidance for Collection of NBS Dried Blood Spots](#)

### Turn-around Time

Within 5 working days from receipt

### Reference Range

- Total Galactose: < 12 mg/dL
- GALT Enzyme: > 3.5 mg/mL

### Reportable Results

- Within Normal Limits:
  - Total Galactose: Within Normal Limits (< 12 mg/dL)
  - GALT Enzyme: Within Normal Limits (> 3.5 mg/mL)
- Outside Normal Limits
  - Total Galactose: (< 12 mg/dL)
  - GALT Enzyme
    - Decreased (2.5 to 3.5 mg/mL)
    - Low (< 2.5 mg/mL)

### Interpretation

- Within Normal Limits
  - Not consistent with classic galactosemia
- Outside Normal Limits
  - Total Galactose High <value> mg/dL but GALT Enzyme Within Normal Limits: Not consistent with classic galactosemia; repeat testing at provider's discretion
  - GALT Enzyme Decreased: Repeat screen unless on lactose free formula, family history, or symptomatic then consult
  - GALT Enzyme Low or Repeat GALT Enzyme Decreased: Possible Galactosemia; Immediate confirmatory testing, start baby on lactose free formula

### Limitations/Interferences

- This is a screening test only. This test is not designed to screen for GALT heterozygosity. A diagnostic procedure should be used to confirm a diagnosis of galactosemia.
- Infants that have not ingested breast milk or formula containing lactose prior to the sample collection may have lower total galactose values.
- Conjugated bilirubin concentrations greater than 16.6 mg/dL and acetaminophen concentrations greater than 2.75 mg/dL in the blood of infants may decrease measured TG concentrations, which may cause false negative results.
- Exchange transfusions may also lead to a false negative screening test.
- Specimens improperly collected, processed or transported may result in erroneous results.

### CPT Code

82760

### Notes

This test is approved for in vitro diagnostic use by the U.S. Food and Drug Administration.