

# Galactosemia

Galactosemia is a hereditary disease that is caused by the lack of a liver enzyme required to digest galactose. The genetic disorder is transmitted as an **autosomal recessive** disease. Galactose is a breakdown product of lactose, which is most commonly found in milk products. When galactose cannot be broken down, it builds up in the cells and becomes toxic. If not diagnosed and treated it can lead to diarrhea, dehydration, jaundice, hepatic failure, hypoglycemia, cataracts, developmental retardation and death. Sepsis due to *E. 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.

**Prevalence in Missouri:** 1:50,000

**Analyte Measured:** Galactose-1-Phosphate Uridyl Transferase

## Feeding Effect:

- **Transferase:** The transferase test should be abnormal in all severe (classical) galactosemic infants even if the specimen is obtained before lactose is ingested, **unless the infant has been transfused.**

**Timing Effect:** Infants who need a transfusion or antibiotics should be screened prior to instituting these therapies.

- **Pre-transfusion:** at any hour of age results are valid
- **Post-transfusion:**  $\geq 30$  days post transfusion results are valid

## Reporting Results:

- **Normal:** Normal transferase ( $> 4.0$  U/gHb).  
The final newborn screening reports are mailed to the submitter and physician of record.
- **Borderline Risk:** Decreased transferase (3.1 - 4.0 U/gHb).  
The final newborn screening report is mailed to the submitter and physician of record.
- **High Risk:** Decreased transferase ( $\leq 3.0$  U/gHb).  
Final galactosemia results are phoned and faxed to physician/health care provider and appropriate follow-up center. Final newborn screening report is mailed to the submitter and physician of record.

## Interpretation of Newborn Screening Results:

Final Result	Test Results		Likely Causes
	Transferase	Reference Range	
Normal	Normal	$> 4.0$ U/gHb	No indication of a defect in galactose metabolism.
Borderline Risk	Abnormal (Decreased)	3.1 - 4.0 U/gHb	- Variant galactosemia - Other enzyme defects in red blood cells - Improperly handled sample (heat damage or transit delay)
High Risk	Abnormal (Decreased)	$\leq 3.0$ U/gHb	- Classical galactosemia - Variant galactosemia - Other enzyme defects in red blood cells - Improperly handled sample (heat damage or transit delay)

## Confirmation:

Decreased transferase levels require prompt follow up testing to determine if the baby has classical galactosemia or is a carrier for the disease. It is highly recommended that the baby's physician/health care provider contact one of the **Metabolic Treatment Centers** (see below) for follow up. Follow up testing usually includes a quantitative Galactose-1-Phosphate Uridyl Transferase (also known as GPUT or GALT) and electrophoresis testing that determines the genotype of the baby.

## Metabolic Treatment Centers:

- [St. Louis Children's Hospital](#) 314-454-6093
- [Cardinal Glennon Hospital For Children](#) 314-577-5639
- [Children's Mercy Hospital](#) 816-234-3290
- [University of Missouri](#) 573-882-6991

## Variant Forms of Galactosemia:

- There are several genetic variants characterized by less severe reduction in the enzyme activity (e.g. Duarte variant). Although most of these individuals are asymptomatic, all should be evaluated, as some will require management and monitoring.
- **Galactokinase Deficiency:** This rare enzymatic defect is also recessively inherited. It results in cataracts in infancy and possibly mild mental retardation. The life-threatening symptoms of severe galactosemia do not occur.
- **Galactose Epimerase Deficiency:** Caused by defective UDP glucose 4-epimerase, results in accumulation of galactose-1-phosphate in the red blood cells and is usually benign.

## Treatment:

The galactosemia syndromes are treated by exclusion of lactose and galactose from the diet and should be done in consultation with a pediatric metabolic specialist. Soy formula is used. Parents require education in assessing food labels and in food preparation. Families with galactosemia should be referred for genetic counseling.

## Avoiding False Positive and False Negative:

Infants with galactosemia may have false negative screening results if they:

- are being treated with antibiotics
- have been recently transfused

Infants with galactosemia may have false positive screening results if they:

- had an improperly handled sample (heat damage or transit delay)