Galactosemia

Galactosemia is an inherited disorder of galactose metabolism. The main dietary source of galactose is lactose, which is found in milk. Clinical features of Galactosemia are failure to thrive (most common initial symptom), vomiting and diarrhea seen within a few days of birth after ingestion of milk. There is also an increased incidence of E.coli sepsis in untreated neonates. Early detection, proper diet and monitoring for signs of infection are essential to prevent death in infants with Galactosemia.

Prevalence:	1:60,000 –80,000
Analyte Measured:	Free Galactose
Reporting Ranges:	Elevated Galactose-1-Phosphate Reduced Uridyl-1-transferase deficiency (GALT)
Feeding Effect:	Minimal – Classical galactosemic babies have elevated galactose-1-phosphate levels
Timing Effect	**<** 24 hours of age: Repeat within 7 days > 24 hours of age: Results are valid
Other Effect:	Potential for false positives due to the stability of transferase decreases in hot humid months. Blood transfusions may have a negative Beutler test for as long as 2-3 months**.**
Confirmation:	Quantitative measurement for galactose, galactose1- phosphate and starch gel electrophoresis for transferase enzyme. Check for galactosuria
Treatment:	Referral made to a Metabolic Specialist. Elimination of dietary lactose, including breast milk, cow's milk and /or lactose based infant formula.
Comment:	Symptoms may occur before receiving the results of the Newborn screening. A galactose-free diet and supportive care for E. coli sepsis, liver failure, and coagulation problems should be considered pending confirmation of the diagnosis.

Maine Newborn Screening Program
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