FACT SHEET
Healthcare Provider

Galactosemia (GALT)

Description:
The most common form of galactosemia, an autosomal recessive metabolic disorder, is the result of very little or absence of the enzyme galactose-1-phosphate uridyl transferase. This enzyme is involved in the digestion of galactose, a breakdown product of lactose. Lactose is present in milk and most infant formulas, and it naturally occurs in organ meats, legumes, fruits, and vegetables. Deficiency of galactose-1-phosphate uridyl transferase quickly raises the galactose content in the blood to dangerous levels.

Incidence in General Population:
1:50,000 live births

Symptoms:
Infants with galactosemia may appear normal at birth; within a few days to 2 weeks after initiating milk feedings, the symptoms of untreated galactosemis can become severe.

Severity

- Physical disabilities: cerebral palsy, ataxia, seizures, cataracts, and liver disease.
- Developmental disabilities: mental retardation.
- Mortality: liver failure, sepsis, or bleeding can cause severe morbidity and death.

Symptomatic diagnosis

- Symptoms can occur even before receiving the results of newborn screening. Early symptoms include jaundice, vomiting, lethargy, hepatosplenomegaly, cataracts, failure to thrive, hypoglycemia, and sepsis.

Variants

- Duarte Galactosemia is a milder form where activity of galactose-1phosphate uridyl transferase is about 25-50% of the enzyme's normal activity. Research has not revealed medical or other developmental complications associated with Duarte Galactosemia.

Diagnosis:
Newborn screening—Quantitative Flurometric testing is used to determine the presence of the galactose-1-phosphate uridyl transferase enzyme. In the absence of the enzyme, florescence (activity) does not occur. If there is no activity, a test for total galactose is performed. Several dried-blood-spot filter paper cards may be requested by the Newborn Screening Laboratory if the initial screening results are not within normal range. Infants with presumptive positive screening (critical) results require prompt follow up. If this occurred, the clinician would be contacted by the Metabolic Treatment Center. When notified of these results, the clinician should immediately check on the clinical status of the baby and facilitate referral to the Metabolic Treatment Center. The Metabolic Treatment Center will provide consultation and assistance with diagnostic testing.

Monitoring:
Individuals diagnosed with classical galactosemia require life-long medical management and dietary modification coordinated by nutrition and metabolic specialists. Clinical observation is important for healthcare providers caring for patients with classical galactosemia. It is important for primary care provider and the Metabolic Treatment Center to develop an ongoing collaborative relationship in caring for these patients.

Treatment:
Infants with galactosemia are started on milk-substituted formula, most likely a lactose-free soybean protein formula. Galactose is a non-essential nutrient, and individuals diagnosed with classical galactosemia require lactose-restricted diets for life. Endogenous production of galactose can complicate dietary treatment of galactosemia and may result in some developmental delays.

Illness and Immunizations:
Immunizations should be kept current. Consult with the Metabolic Treatment Center within 24 hours of the onset of an illness or at the time of hospitalization.

Surgical/Surgical Procedures:
Caution concerning administration of anesthesia and certain drugs that may contain lactose is necessary.

Growth and Development:
Monitor child for normal growth and developmental milestones.

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