

Physician ACT SHEET for POSITIVE Newborn Screening Result for GALACTOSEMIA

Disease Category: Inborn error in metabolism of galactose

POTENTIAL NEONATAL EMERGENCY YOU SHOULD TAKE THE FOLLOWING ACTIONS:

- **Contact the family to inform them of the newborn screening result and arrange an immediate office visit for physical exam. Advise the family to switch to powder based soy formula, after the confirmatory specimen is collected.**
- **If infant is lethargic or not feeding well or demonstrating any signs of failure to thrive, emergency care is warranted.**
- **Immediately consult a metabolic specialist.**
- **Undertake definitive investigations in consultation with metabolic specialist.**
- **Report findings back to state newborn screening program.**

Pediatric metabolic specialists are available through the metabolic centers at Children's Hospital and the Nebraska Medical Center. The pediatric metabolic specialist on service/call today is:

_____ MD and can be paged at: _____

Meaning of the Screening Result: Possible severe galactosemia, improper specimen handling (heat damage or transit delay), mild galactosemia variant, other enzyme defect in RBC's, false positive.

Metabolic Description: The severe form of this disease is due to almost total deficiency of galactose-1-phosphate uridyl transferase enzyme activity in all cells of the body. There are several genetic variants characterized by a less severe reduction in enzyme activity (e.g. Duarte Variant). Most of these cases are asymptomatic and are detected because of a persisting abnormality in the enzyme test. However, some of these cases may benefit from dietary therapy.

Confirmation of Diagnosis: **Order collection of 2 ml. of whole blood, EDTA (lavendar top) and order a confirmatory test for galactosemia, specifically a quantitative galactose 1 phosphate uridyl transferase. Specimen should be sent cold (e.g. use ice pack, not frozen or on dry ice).**

Clinical Expectations: If untreated, results in liver dysfunction manifest as jaundice and hypoglycemia; neurological findings of irritability and seizures; and gastrointestinal findings of poor feeding, failure to thrive, vomiting, and diarrhea. Death may result from gram-negative sepsis within one to two weeks of birth. If the infant is untreated and survives the neonatal period, cataracts, cirrhosis, Fanconi syndrome, and mental retardation are usual developments.

Additional Information:

New England Metabolic Consortium – Emergency Protocols www.childrenshospital.org/newenglandconsortium

Gene Tests/Gene Clinics - www.genetests.org

U.S. National Newborn Screening & Genetics Resource Center www.genes.us.uthscsa.edu