

Physician Action Sheet for Abnormal Newborn Screen – D/G Galactosemia, Galactosemia Variants

Newborn Screen Findings: Deficient or decreased galactose-1-phosphate uridylyltransferase (GALT) & possibly increased galactose (GAOS) on newborn screen. *Because environmental factors (such as heat) can affect screening results, the Indiana University Newborn Screening (NBS) Lab may request a repeat newborn screen before requesting the next step in confirmatory testing.*

The Indiana University NBS Lab should have already contacted your office & the infant's birthing facility about the abnormal results & recommended confirmatory testing. The IU NBS Lab has also notified the Metabolic Specialist. *Please note that the IU NBS Lab does not contact the patient's family.*

Required Actions

1. **Contact the Metabolic Specialist (317) 274-3966 (ask for Newborn Screening Nurse or Genetic Counselor) to discuss abnormal results, follow-up recommendations & confirmatory testing.**
2. **Contact the family as soon as possible.**
 - a. Inform the family of the abnormal NBS result & the need for additional confirmatory testing.
 - b. Determine the condition of the infant. *See the infant in your office immediately if there are concerns of poor feeding, vomiting or lethargy. These symptoms require immediate evaluation & treatment.*
 - c. Educate the family about the need to *immediately switch the infant to soy-based formula* while awaiting confirmatory test results.
 - d. Review the list of symptoms that require immediate contact to your office (see above).
3. **Arrange for confirmatory laboratory testing at the infant's birthing facility.**
4. **Plan for follow-up with the Metabolic Specialist if diagnosis of D/G or variant galactosemia is confirmed.**

Clinical Summary: Classical galactosemia is an autosomal recessive disorder of galactose metabolism resulting from defective activity of galactose-1-phosphate uridylyltransferase (GALT) enzyme. GALT deficiency results in accumulation of galactose-1-phosphate and galactose, causing multi-organ disease. Symptoms of the disorder include poor feeding, vomiting, lethargy, prolonged hyperbilirubinemia, poor growth & developmental delay with language problems. Infants with classic galactosemia may present in early infancy with *E. coli* sepsis & liver dysfunction. Galactosemia carriers (N/G) & individuals with milder galactosemia variants (D/G) are almost always asymptomatic. *All infants with a positive newborn screen should switch from lactose formula/breast milk to soy formula while awaiting the results of follow-up testing.*

Treatment: Metabolic Specialist will recommend treatment if diagnosis of galactosemia variant or D/G galactosemia is confirmed. Treatment of D/G galactosemia usually involves soy formula & lactose/galactose restriction for at least six months, at which point a galactose challenge test will be done to determine if the infant can be transitioned to a normal diet. *The galactose challenge test is never done with classic galactosemia.*

Incidence: 1/3,400 in Indiana (D/G galactosemia only). Genetic testing available.

Contact Information:

- Regular business hours: (317) 274 – 3966; ask for the Newborn Screening Nurse or Genetic Counselor.
- Emergency night/weekend contact: (317) 944 – 5000; have the on-call Metabolism physician paged.

Resources:

- Newborn Screening ACT sheets – www.acmg.net (select "Resources," click on "ACT Sheets")
- Information for clinicians - <http://www.ncbi.nlm.nih.gov/sites/GeneTests/review>