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Protocol for Evaluation: Classic Galactosemia Newborn Screen
(Florida GALT screening level of equal to or less than 2.5 units/gram Hgb)

1. This baby is a risk for CLASSIC GALACTOSEMIA which results in accumulation of galactose-1-phosphate and galactose, causing multi-organ disease. Immediately stop breast or cow's milk feeding and start non-lactose feeding with powdered soy-based formula such as Good Start Soy, Enfamil Prosobee, or Similac Isomil. The breastfeeding mother should pump and store breast milk until confirmatory testing has been completed. If the baby has classic galactosemia, he/she will not be able to breast feed or consume lactose-containing formula.

2. The primary physician should examine the baby as soon as possible. Evaluate for jaundice, poor feeding, vomiting, lethargy, bulging fontanel and bleeding. An infant with Classic Galactosemia is at risk for E.Coli sepsis. Refer to the ACMG ACT sheet (attached).

3. Order confirmatory testing and have blood sent to Mayo Medical Laboratories, ARUP Labs OR Quest Diagnostics. A hospital lab with infant experience will advise on what reference lab is used.
   a. **Galactosemia (GALT) Enzyme activity (Galactose-1-Phosphate Uridyltransferase)** This test verifies low or absent enzyme activity
   b. **Galactosemia (GALT) gene mutation analysis.** This test confirms the genotype.
   c. **Galactose-1-Phosphate (Gal1P), RBC.** This test measures a substrate of galactose metabolism. It does not confirm diagnosis but is used to monitor treatment.

   Lab protocols for the above tests are available on websites for:
   1) Mayo Medical Laboratories, 2) ARUP Laboratories and 3) Quest Diagnostics

4. Interpretation of results: GALT enzyme activity and mutation analysis determines the enzyme activity and allows for correlation with the DNA genotype. Generally, the GALT enzyme activity correlates with the following genotypes:

   100% = N/N (normal status)
   ~75% = N/D (Duarte carrier)
   ~50% = N/G (galactosemia carrier) or D/D ("double" Duarte carrier)
   ~5-25% = D/G (Duarte [variant] galactosemia)
   ~0-5% = G/G (classic galactosemia)

   If the baby is D/G, Duarte galactosemia, a benign variant form of galactosemia, is present. We can provide appropriate management protocols. In addition, a short video is available website at:
   www.peds.ufl.edu/peds2/divisions/genetics/caw/ programs_newborn_ screening . htm

   If the baby is N/N, N/G or D/D, regular formula and breast feeding are allowed and no biochemical monitoring is needed. The baby can be treated as a normal infant.

   If the baby is G/G, classic galactosemia is present. This infant requires lifelong galactose restriction and Management by a genetic metabolic physician and dietitian. Genetic counseling is advised.

   Please contact us with any questions. Any of our clinical physicians or our metabolic dietitians is available to provide consultation. Call the Division of Genetics at 352-294-5050 to make a referral.

   Edited 9.14.12