

Protocol for Evaluation: Borderline Galactosemia Result
(Florida GALT screening level of >2.5-3.5 units/gram Hgb)

1. A borderline galactosemia result suggests that this baby has normal or near normal ability to metabolize lactose in breast milk or milk-based formula. Unless there are symptoms such as jaundice, poor feeding, vomiting, diarrhea or lethargy, a diet change is not needed. Preservation of breastfeeding is optimal. If the baby is on (or supplemented with) milk-based formula, changing to soy-based infant formula is an option but not necessary.
2. The primary physician should examine the baby and order confirmatory testing. We recommend the following tests. Please fax all results to our division.

- 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. (If sufficient blood); Galactose-1-Phosphate, RBC.** This test measures a substrate of galactose metabolism. It is NOT diagnostic-it is used for monitoring treatment.

Lab protocols for the above tests are available on websites for

1) Mayo Medical Laboratories, 2) ARUP Laboratories and 3) Quest Diagnostics

3. 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 G/G, classic galactosemia is present and lifelong galactose restriction and specialist follow-up are required.

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.

4. Genetic counseling is advised for families who have babies with the G/G genotype. Call the Division of Genetics at 352-294-5050 to make a referral.

Please contact us with any questions. Any of our clinical physicians or our metabolic dietitians is available to provide consultation.

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