

Protocol for Evaluation: Borderline Galactosemia Result

(Florida GALT screening level of 2.11-3.1 units/gram Hgb)

1. This baby has normal or near normal ability to metabolize lactose containing formula, so no change in diet is required.
2. The primary physician should examine the baby and collect blood for confirmatory testing. Blood can be sent to either Mayo Medical Laboratories or Emory Biochemical Genetics Program (addresses below) for the following tests:

a. Galactose-1-Phosphate, RBC level. This is a substrate not an enzyme level.

b. Transferase (GALT) enzymatic activity level. This test verifies low or absent enzyme activity. Generally, the 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/galactosemia carrier)

~0-5% = G/G (classic galactosemia)

c. DNA genotyping. The DNA test confirms the genotype (eg. G/G, D/G or N/G) and is more accurate than the enzyme activity level.

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 on our 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.

Mayo Medical laboratories

3050 Superior Drive NW
Rochester, MN 55901
1-800-533-1710

Emory Diagnostic Laboratory

Dept. of Genetics and Molecular Medicine
Emory Clinic, Rm. 412, 1262 Clifton Road
Atlanta, GA 30322
(800) 727-8308 FAX (404) 727-8310

3. Send a copy of the initial laboratory results to the Regional Genetics Program at the University of Florida (Fax 352-392-3051).
4. Genetic counseling is advised for families who have babies with the G/G or D/G genotype. Call the Division of Genetics at 352-392-4104 to make a referral.

Please contact us with any questions. Any of our clinical physicians, Dr. Charles Williams, Dr. Roberto Zori, Dr. Bryce Hess or Dr. Daniel Driscoll or our newborn screening coordinator, Penny Edwards MS, RD is available to provide consultation.