

Protocol for Evaluation: 0% Fluorescence Test Result
(Florida GALT screening level of ≤ 2.1 units/gram Hgb)

1. When the 0% result is reported, immediately place the baby on a soy-based (powder) formula such as Prosobee, Isomil or Good Start Soy. The breastfeeding mother should pump and store breast milk until confirmatory testing is completed and diagnosis is confirmed. A baby with classic galactosemia will not be able to breast feed or consume lactose-containing formula.
2. The primary physician should examine the baby as soon as possible. Blood should be collected and sent to either Mayo Medical Laboratories or Emory Biochemical Genetic Program (addresses below) for the following tests.
 - a. **Galactose-1-Phosphate (Gal-1-P) level:** Gal-1-P is the substrate for the galactose metabolizing transferase enzyme.
 - 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)

b.DNA genotyping: The DNA test confirms the genotype (e.g., G/G, D/G or N/G).

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

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 http://www.peds.ufl.edu/peds2/divisions/genetics/caw/programs_newborn_screening.htm

If the baby is N/N, N/G or D/D, dietary changes and restrictions are not required and biochemical monitoring is not necessary. The baby can be treated as a normal infant.

Mayo Clinic Medical Laboratories
Mayo Clinic Rochester
200 First Street, Southwest
Rochester, Minn 55905
1-800-533-1710 Fax 507-266-2888

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. Please call the Division of Genetics to make a referral (352-392-4104).

Please call with any questions. Our Newborn Screening Coordinator, Penny Edwards MS, RD, or any of our clinical physicians, Dr. Charles Williams, Dr. Bryce Heese, Dr. Roberto Zori and Dr. Daniel Driscoll are available for consultation as needed.