

Suggested Follow-up for Classic Galactosemia (Deficient GALT with Elevated Total Galactose)

Possible Causes:

Classic galactosemia is a condition of abnormal galactose metabolism caused by deficient functioning of galactose-1-P-uridyl transferase (GALT). When GALT is moderately deficient, the infant may have Duarte galactosemia or maybe a carrier for a form of galactosemia.

GALT can also be falsely deficient if specimen is exposed to extremes of heat and/or humidity.

Next Steps if Abnormal:

Potential medical emergency when GALT is deficient and total galactose is significantly elevated. See infant as soon as possible to ascertain health status. Consult pediatric metabolic specialist and initiate diagnostic evaluation and treatment as recommended.

If the mother is breastfeeding, discuss feeding with a metabolic specialist to determine if a switch to soy based infant formula is necessary.

Repeat GALT and total galactose screening on filter paper and send to the DHEC laboratory. If GALT is still deficient in repeat specimen, further testing is necessary to clarify the diagnosis.

Neonatal Presentation:

Classic galactosemia - hypoglycemia, jaundice, sepsis, failure to thrive.
Duarte galactosemia - none.

Emergency Treatment:

Feeding change to soy based infant formula when GALT is low and total galactose is significantly elevated. See "Next Steps" section for other feeding recommendations.

Standard Treatment:

Classic galactosemia - galactose restricted diet for life.
Duarte galactosemia - usually none past infancy.

Advice for Family:

Provide basic information about classical galactosemia. The handout, *When Baby Needs a Second Test for Classic Galactosemia*, may be used for this purpose. Stress the importance of seeking immediate medical attention if the infant shows any signs of illness.

Internet Resources:

<http://ghr.nlm.nih.gov/condition/galactosemia>

<http://www.newbornscreening.info/Parents/otherdisorders/Galactosemia.html>