Suggested Follow-up for Variant Galactosemia
(Elevated Total Galactose with Normal GALT)

Possible Causes:

Variant galactosemia can be caused by deficient functioning of galactokinase (GALK) or UDP galactose-4-epimerase (GALE).

Next Steps if Abnormal:

Repeat galactosemia screening on filter paper and send to the DHEC Public Laboratory. No formula/feeding changes until result of repeat testing known. If total galactose is still elevated in repeat specimen, refer to metabolic specialist.

Further evaluation/testing is necessary to clarify diagnosis. Switch infant to soy-based formula as recommended by metabolic specialist.

Neonatal Presentation: Usually none.

Emergency Treatment: None.

Standard Treatment:

GALK - galactose restricted diet for life. GALE - if the GALE deficiency is localized in the red blood cell, the infant does not have any symptoms of disease and no treatment is necessary. If the GALE deficiency involves other tissues, galactose restricted diet for life.

Advice for Family:

Provide basic information about galactosemia. The handout, *When Baby Needs a Second Test for Galactosemia, Elevated Total Galactose with Normal GALT*, may be used for this purpose.

Internet Resources:

http://ghr.nlm.nih.gov/gene=gale

http://ghr.nlm.nih.gov/gene=galk1

https://www.babysfirsttest.org/newborn-screening/conditions/galactokinase-deficiency

https://www.babysfirsttest.org/newborn-screening/conditions/galactoepimerase-deficiency