



North Carolina Department of Health and Human Services
State Laboratory of Public Health

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MEMORANDUM

To: Hospitals, Birthing Centers, Health Departments, and Physician Offices

From: Scott J. Zimmerman, DrPH, MPH, HCLD (ABB), Laboratory Director
Shu Chaing, Ph.D., Manager, Newborn Screening *SJC*

Date: November 5, 2014

Re: Change in Methodology for Total Galactose to Identify Classic Galactosemia and Modifications to Follow-Up Recommendations

In mid-September 2013, the Newborn Screening (NBS) Laboratory at the North Carolina State Laboratory of Public Health (NCSLPH) began evaluation of a new testing algorithm for galactosemia. This new testing algorithm tests all NBS specimens to determine total galactose concentration and galactose-1-phosphate uridytransferase (GALT) enzyme activity. These assays are used to identify infants with Classic Galactosemia. Specimens with abnormalities in GALT activity are further analyzed to detect 3 DNA mutations that are associated with Classic Galactosemia. The three DNA mutations tested for by NCSLPH are as follows:

- Q188R (the most common mutation in the Caucasian population),
- S135L (the most common mutation in those of African descent), and
- N314 D (the most common mutation in patients with Durant variant Galactosemia)

NCSLPH is planning to implement the new testing algorithm and the new methodology for total galactose beginning November 5, 2014. The new methodology will employ the use of Fluorescent Galactose Oxidase manufactured by Perkin Elmer.

The total galactose reference range will change and be dependent on GALT enzyme activity. Elevated total galactose values may require additional patient follow up and genetic counseling. The DNA mutation analysis, if required, will supplement total galactose and GALT results. The results and follow up recommendations will be provided on a second page of the laboratory report.

With addition of the methodology change for total galactose, the recommended follow up actions are also being included on the report.

If you have any questions, please contact Dr. Shu Chaing (919-807-8880), Radish Persaud (919-807-8829) or Lara Perenti (919-707-5634).

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