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

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

From: Scott J. Zimmerman, DrPH, MPH, Director, NC State Laboratory of Public Health

Date: September 8, 2015

Re: New Addition to Newborn Screening Panel

The Newborn Screening Program of the North Carolina State Laboratory of Public Health (NCSLPH) is pleased to announce new additions to the newborn screening (NBS) panel. Beginning with specimens received Thursday, October 1, 2015, NCSLPH will screen for additional disorders: **Tyrosinemia Type 1 (TYR 1), Malonic Acidemia (MAL),** and **Argininemia (ARG). Also, a new marker, Agrininosuccinic Acid, will replace Citrilline as the primary marker,** for **Argininosuccinic Aciduria (ASA)**. No additional specimen is required as NCSLPH will use the dried blood spot specimen collected from the initial newborn heelstick specimen.

These genetic disorders that will be added to the NC NBS panel are part of the US Department of Health and Human Services' Recommended Uniform Screening Panel (RUSP) Core and Secondary Conditions. Tyrosinemia Type 1 is an amino acid disorder where affected infants exhibit symptoms such as failure to thrive, neurological crises, liver and kidney problems. Untreated infants can suffer liver and kidney failure, bleeding abnormalities and even death. Malonic Acidemia is a very rare organic acid disorder; symptoms may include seizures, developmental delay, and metabolic acidosis. Symptoms of Argininemia, an amino acid disorder, include failure to thrive, movement disorders, and seizures. Infants diagnosed with Argininosuccinic Aciduria may exhibit chronic symptoms of developmental delay, and acute symptoms of lethargy, seizures, and coma.

Results for the analytes Succinylacetone (Suac) to screen for Tyrosinemia Type 1 and Arginine (Arg) to screen for Argininemia will be reported as normal, borderline, abnormal, or unsatisfactory. Results for the analytes Malonylcarnitine (C3DC) the marker for Malonic Acidemia and Arginosuccinic Acid (Asa) the primary marker for Argininosuccinic Aciduria will be either normal, abnormal, or unsatisfactory. Borderline results will require a repeat heelstick specimen collected and submitted as soon as possible. Abnormal laboratory results will be immediately faxed to medical follow-up personnel, who will contact the healthcare provider with directions for further testing and/or evaluation.

If you have questions about this update to the newborn screening panel, please contact Mr. Hari Patel, MS/MS Supervisor at (919) 807-8891 or the Newborn Screening Manager, Dr. Shu Chaing, at (919) 807-8880.