

STATE OF CONNECTICUT

DEPARTMENT OF PUBLIC HEALTH

Manisha Juthani, MD
Commissioner



Ned Lamont
Governor
Susan Bysiewicz
Lt. Governor

May 10, 2023

Re: CT Newborn Screening Program Updates-Correction Highlighted Below.

Dear Provider,

Addition of Disorder to the CT Newborn Screening Panel

The Connecticut Newborn Screening (CT NBS) Program has completed validation of a blood spot screening method for Guanidinoacetate Methyltransferase Deficiency (GAMT Deficiency). It is anticipated that the CT NBS Program will go-live GAMT Deficiency screening around May 1, 2023. At that time, every newborn blood spot sample received at the State Public Health Laboratory will be screened GAMT Deficiency. Initial samples that screen borderline out-of-range for GAMT Deficiency will be reported by the CT NBS Program to the newborn's health care provider with a request to submit blood spot sample for repeat screening. Presumptive positive screening results will be reported by the CT NBS Program to the newborn's healthcare provider and to the CT NBS Network (the Network) to begin the diagnostic work-up. A nurse from the Network will follow up with the newborn's health care provider with a specific recommendations for care and diagnostic testing. When indicated, the newborn will be evaluated by a Pediatric Geneticist through Yale New Haven Health or the CT Children's Medical Center.

GAMT Deficiency is a rare genetic disorder with an estimated incidence rate of 1:100,000 people in the United States. Left untreated, low levels of creatine and high levels of guanidinoacetate create serious damage to the organs including the brain and muscles. According to the Federal Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC), experts believe that screening all newborns in the United States for GAMT Deficiency will identify approximately 7 newborns each year with the condition. There is no cure for this condition, but diagnosis in the newborn period enables early monitoring and treatment. Treatment includes lifelong supplements and possible dietary changes. Treatment may help alleviate some symptoms and may prevent new symptoms.

Implementation of Age-Related Reference Ranges for Thyroid Stimulating Hormone

Thyroid Stimulating Hormone (TSH) measurement is the target of newborn Congenital Hypothyroidism (CH) screening in CT. For most infants, TSH levels peak at within the first 24 hours of life and then rapidly drop within the first 3-4 days of life. There was a concern the existing TSH reference range could fail to identify infants at risk for CH when a bloodspot sample is collected ≥ 4 days of life. This led to the analysis of over 10 years of CT CH screening data and the development of age-related reference ranges for TSH. Age related



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reference ranges are particularly useful when an initial NBS sample is collected late and in the analysis of repeat CH screening results. These age-related reference ranges are effective April 20, 2023.

Implementation of Repeat Congenital Hypothyroidism Screening for Very Low Birthweight Newborns

Many infants born preterm, ill and low birthweight have lower serum thyroid hormone levels as compared to term infants, a reflection of reduced TSH surge following birth, immature postnatal pituitary-thyroid function, and loss of the maternal contribution¹. Rescreening of very low birthweight (VLBW) infants (<1500 grams) for CH closer to a normal corrected gestational age has been demonstrated effective in detecting CH in some infants whose TSH level was within normal limits on the initial NBS. Effective April 20, 2023, the CT NBS Program will request collection of a blood spot sample at **28 to 30** days of age for repeat CH screening for all VLBW infants. CT NBS Program Follow-up staff will call your facility/practice to request this sample for VLBW infants and will follow up with a written request.

Please direct any questions regarding GAMT Deficiency screening, age-related TSH reference ranges or repeat CH screening for VLBW infants to the CT NBS Program at 860.920.6628.

Sincerely,

Marie Burlette, RN, BSN, MPH
Supervising Nurse Consultant
Connecticut Newborn Screening Program

¹ LaFranchi SH (2021) Thyroid Function in Preterm/Low Birth Weight Infants: Impact on Diagnosis and Management of Thyroid Dysfunction. *Front. Endocrinol.* 12:666207.doi: 10.3389/fendo.2021.666207



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