STATE OF CONNECTICUT
DEPARTMENT OF PUBLIC HEALTH

Deidre S. Gifford, MD, MPH
Acting Commissioner

Ned Lamont
Governor
Susan Bysiewicz
Lt. Governor

January 24, 2021

Dear Hospital and Primary Care Medical Providers,

The Connecticut Newborn Screening (CT NBS) Program officially implemented universal screening for Pompe Disease and Mucopolysaccharidosis Type 1 (MPS-I) on January 1, 2021. All NBS samples received on or after October 1, 2020 have been screened as part of the validation process. Pompe and MPS-I are autosomal recessive lysosomal storage disorders. Pompe results from a deficiency in the Acid Alpha-Glucosidase (GAA) enzyme and MPS-I from a deficiency in the Alpha-L-Iduronidase (IDUA) enzyme. There is a high possibility of heterozygous (carriers) and pseudodeficiency (false positive) results associated with these enzymes. In order to decrease the impact of pseudodeficiencies on CT's NBS system, samples that screen positive for Pompe or MPS-I through the CT NBS Program are sent to Mayo Clinical Laboratories for second tier screening. Results that remain positive after second tier screening are immediately reported by the CT NBS Program to healthcare provider (HCP) caring for the infant and the CT Newborn Diagnostic and Treatment Network (the Network). Network staff will contact the HCP and coordinate follow-up of the newborn and evaluation by the appropriate specialty care provider (genetic or neurology) as necessary.

The estimated incidence rate for Pompe Disease is approximately 4:100,000 with varying age and severity of onset. Pompe Disease consists of two major forms, the classical infantile form and the non-classical late onset form. Both forms can cause problems in the heart and lung and muscle in general. The classical infantile form begins in early infancy, is the most severe and can worsen quickly, potentially causing death within the first year of life. Non-classical late onset Pompe Disease varies for age of onset and severity of disease. While there is no cure for Pompe Disease, early diagnosis and treatment, including enzyme replacement therapy, can prevent symptoms from getting worse. Infants with Pompe disease look healthy at birth making Newborn Screening an important tool to help to identify newborns with Pompe Disease so that treatment can begin right away. According to the federal Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC), it is anticipated that NBS will identify 144 babies with Pompe Disease each year (nationally), will prevent up to 28 people with the disease from requiring mechanical ventilation and may prevent up to 19 deaths due to the disease each year.

The estimated incidence rate for MPS-I is approximately 1:100,000 with clinically variable onset and a wide range of symptoms that are classified into two major forms of the disease: severe MPS-I and attenuated MPS-I. The severe form of MPS-I can cause problems with the heart, airways, eyes and ears, muscles, bones, joints, and brain and can often lead to death without treatment. Early identification of MPS-I through newborn screening allows for early monitoring and treatment when necessary. Not all children with MPS-I require treatment, but for those who do, enzyme replacement therapy and bone marrow transplant can prevent symptoms from getting worse. According to the ANHDNC, it is anticipated that NBS will identify 44 babies with MPS-1 each year (nationally) and will prevent up to 2 deaths before age 5 years due to the disease each year. Please direct any questions regarding Pompe Disease or MPS-1 screening to the CT NBS Program at 860.920.6628.

Sincerely,

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Connecticut Newborn Screening Program

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