

STATE OF CONNECTICUT

DEPARTMENT OF PUBLIC HEALTH

Manisha Juthani, MD
Commissioner



Ned Lamont
Governor
Susan Bysiewicz
Lt. Governor

June 6, 2024

Re: CT Newborn Screening Program Update

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 Mucopolysaccharidosis Type II (MPS II). Screening for MPS II went live in CT on April 26, 2024. Since that time, every newborn blood spot sample received at the State Public Health Laboratory has been screened for MPS II. 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 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.

MPS II is a rare genetic disorder with an estimated incidence rate of 1.6:100,000 people in the United Statesⁱ. Left untreated, the disease can cause problems with organs and body systems, including the liver, spleen, heart, airways, lungs, bones, joints, abdomen, head and neck, ears, mouth, nose, skin, throat brain, movement and behavior. There are two types of MPS II. The severe type causes serious brain and behavior problems, while the attenuated type causes less severe issues with the brain but may have the same impact on other body systems as severe MPS II. 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 MPS II will identify approximately 59 newborns each year with the conditionⁱ. There is no cure for this condition, but diagnosis in the newborn period enables early monitoring and treatment. Treatment includes enzyme replacement therapy (ERT) and/or hematopoietic stem cell transplantation (HSCT) which may slow the progression of the disease.

Sincerely,

Heidi Elsinger, MPH
Health Program Associate
Connecticut Newborn Screening Program

ⁱ [Newborn Screening for MPS II: A summary of the Evidence and Advisory Committee Decision \(hrsa.gov\)](https://www.hrsa.gov/newborn-screening/mcps2/)



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