

# STATE OF CONNECTICUT

## DEPARTMENT OF PUBLIC HEALTH

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### Connecticut Newborn Screening Program News Implementation of Screening for Spinal Muscular Atrophy

Rocky Hill, CT, 12/31/19. The Connecticut Department of Public Health's Newborn Screening Program (CT NBS) announced today that screening for spinal muscular atrophy (SMA) will be implemented statewide as of January 1, 2020.

SMA is a rare genetic disorder. An estimated 1 out of every 11,000 people has SMA. People with SMA have a change in a specific gene that prevents it from making enough of the protein that nerve cells need to survive. There are different types of SMA. Most children have SMA Type 1, which causes weakness and, without treatment, can worsen quickly and lead to death. While there is no cure for SMA, new treatments can slow or even prevent SMA symptoms from getting worse, improve muscle function and lower the risk of death from SMA<sup>1</sup>.

Legislation was passed during the last session that mandates the CT Department of Public Health to start screening for SMA, a disorder recently added to the Federal Advisory Committee on Heritable Disorders in Newborns and Children's Recommended Uniform Screening Panel (RUSP), by January 1, 2020. Additionally, legislation was passed that allows the CT Department of Public Health to add other disorders to the CT NBS screening panel once they have been added to the RUSP with approval from the Connecticut Office of Policy and Management.

The RUSP is a list of disorders that the Secretary of the Department of Health and Human Services (HHS) recommends for states to screen as part of their state universal newborn screening (NBS) programs. Disorders on the RUSP are chosen based on evidence that supports the potential net benefit of screening, the ability of states to screen for the disorder, and the availability of effective treatments. It is recommended that every newborn be screened for all disorders on the RUSP<sup>2</sup>.

The CT NBS Program works to ensure that every newborn, who is born or resides in Connecticut, has a valid newborn screening on record and that those infants with abnormal screening results are promptly referred to a specialty treatment center for further evaluation and treatment when needed. These comprehensive efforts help prevent unnecessary disability and premature death. A small amount of blood is taken from the newborn's heel shortly after birth and submitted to the state laboratory to be screened for over 60 conditions.

When a newborn is identified through NBS at being at risk of having SMA, the CT NBS Program will quickly report the result to the Connecticut Newborn Diagnostic and Treatment Network which works to immediately connect the infant, family and the infant's primary care provider with a specialist with expertise in SMA to either rule out or confirm the disorder.

Currently, eleven states screen for SMA.

For more information on SMA go to <https://www.babysfirsttest.org/newborn-screening/conditions/spinal-muscular-atrophy>.

For more information about the CT NBS Program go to <https://portal.ct.gov/newbornscreening>

<sup>1</sup> <https://www.hrsa.gov/sites/default/files/hrsa/advisory-committees/heritable-disorders/rusp/previous-nominations/sma-consumer-summary.pdf>

<sup>2</sup> <https://www.hrsa.gov/advisory-committees/heritable-disorders/rusp/index.html>



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