

Public Health Committee JOINT FAVORABLE REPORT

Bill No.: HB-7282

AN ACT CONCERNING NEWBORN SCREENING FOR SPINAL MUSCULAR

Title: ATROPHY.

Vote Date: 3/22/2019

Vote Action: Joint Favorable

PH Date: 3/13/2019

File No.:

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SPONSORS OF BILL:

Public Health Committee

REASONS FOR BILL:

Beginning January 1, 2020, this legislation requires all health care institutions caring for newborns to test for spinal muscular atrophy, unless a parent objects on religious grounds. Currently in Connecticut, the newborn screening program for genetic and metabolic disorders includes testing for cystic fibrosis, severe combined immunodeficiency disease and critical congenital heart disease. This legislation would add spinal muscular atrophy (SMA) to the newborn screening panel (NBS).

RESPONSE FROM ADMINISTRATION/AGENCY:

Raul Pino, MD, MPH, Commissioner of the Department of Public Health (DPH): The Department supports the intent of the bill. Spinal Muscular Atrophy (SMA) is one of the most common lethal genetic disorders and the most common cause of infant death. This condition results from a specific and identifiable mutation that can be readily detected through newborn screening. Without treatment, it is fatal within the first few months to two years of life. The addition of this disorder to the Public Health Laboratory's NBS panel would provide another tool to identify possibly affected infants in order to provide beneficial follow-up treatment. The Department respectfully requests that the Committee review the language in Section 1. This language would allow DPH to test for any diseases on the Recommended Uniform Screening Panel (RUSP) declared by the U.S. Department of Health and Human Services (HHS) Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC). RUSP disorders are chosen based on evidence that supports the potential benefits of screening, the ability to screen for the disorder, and the availability of effective treatments. The RUSP list of disorders is recommended by the Secretary of the DHHS to be part of a state's newborn

screening panel. The addition of SMA to the Public Health Laboratory's NBS panel would further enhance the ability of the NBS program to identify affected infants for necessary and beneficial follow-up treatment.

NATURE AND SOURCES OF SUPPORT:

Dr. Gyula Acsadi, Division Head of Neurology of Connecticut Children's Medical

Center: Dr. Acsadi supports this legislation because early diagnosis and treatment of Spinal Muscular Atrophy (SMA) provides the most effective outcomes for children with SMA. The most common form of this condition manifests early in life and is the leading cause of death in infants and toddlers before the age of two, unless invasive ventilator support is provided. SMA is caused by a defect in the Survival Motor Neuron 1 (SMN1) gene. This gene is critical to the health and survival of large nerve cells in the spinal cord which controls muscle contraction for voluntary movements. Since SMA is a recessive genetic disease, an individual could be a carrier with no obvious symptoms. In a situation where both parents are carriers of the SMA gene, the potential for their children to have the disease increases. It is also important to note that brain cells are not affected by this disease. Following decades of no specific treatment for SMA, extensive research has led to several effective therapeutic options. One treatment currently under review by the FDA is gene therapy. However, in December of 2016, the FDA approved nusinersen (Spinraza) to treat children with SMA. Several clinical trials and at least two years of experience show that this treatment prevents decline, delays death and improve function in patients. It is critical to note that this treatment is most effective if it is used very early in life, even before the onset of symptoms. To achieve the best outcomes for children with this disease, Connecticut must implement a clinical screening mechanism to diagnose this progressive disease at birth.

Jamie Vickery, Vice President, Advocacy and Policy, CURE SMA: Mr. Vickery submitted testimony on behalf of CURA SMA, the largest nonprofit organization dedicated to finding a cure for SMA. CURE supports this legislation that would provide early screening for the SMA disease. This genetic disease, caused by a mutation in a motor neuron gene, is the most common cause of death among infants in the United States. In a healthy child, this gene produces a protein that is critical for the nerves that control muscle movement. The most effective way to control this disease is through early detection and treatment. The FDA approved a drug, SPINRAZA, which must be administered as soon as possible to be the most effective. It is encouraging to note that in clinical trials of SPINRAZA, pre-symptomatic infants with genetic markers causing SMA are reaching age –appropriate developmental benchmarks. In his testimony, Mr. Vickery notes that Health and Human Services Secretary Alex Azar added SMA to the Recommended Uniform Screening Panel in July of 2018. Also, 20 states have approved adding SMA screening to their newborn panel, and 5 states are already using this test.

Leslie Bennett, Connecticut Rare Action Network CT-RAN, Volunteer State

Ambassador: CT-RAN supported a similar bill which was introduced last year but removed from the Governor's budget following negotiations. This critical issue is back before the legislature and CT-RAN strongly supports the bill. While CT-RAN understands the concerns of budgeting, it is important to note that more than 90% of SMA cases can be detected shortly after birth with a simple blood test. Many of the children and adults currently living with severe forms of SMA, require wheelchairs, feeding tubes to eat and machines to help

them breathe. The cost of caring for SMA is a huge burden not only on families, but also, our local communities for special education costs and the state Medicaid program. CT-RAN urges the committee to pass this legislation which will alleviate suffering and financial stress for all involved.

Wildon Farwell, MD, MPH, Senior Director, Clinical Development, Biogen: Dr. Farwell testified on behalf of Biogen in support of this bill. Biogen is a biotech company that develops and distributes world-wide innovative therapies for people living with serious neurological and neurodegenerative diseases. Biogen introduced the first and only FDA-approved treatment for SMA called SPINRAZA which has been available since 2016. For the last several years, Biogen with the support of CureSMA and the CDC developed and validated a cost-effective newborn screening assay for SMA and opened a clinical trial to test the hypothesis that beginning to treat babies affected by SMA at, or near birth, would result in better outcomes compared to waiting until the onset of symptoms. Now that a treatment option is available, and the data supports that beginning in the pre-symptomatic period improves clinical outcomes. Babies with SMA who receive SPINRAZA after symptom onset appear to achieve more motor milestones and live longer without the need for respiratory support than babies who do not receive this drug. Babies genetically diagnosed with SMA and treated with SPINRAZA before symptoms appear achieve even greater improvement. Biogen urges the Connecticut legislature to add SMA to the state's newborn screening panel as soon as possible. Included in Dr. Farwell's testimony is a graph with the results of this clinical study.

Maria Eleni Kaloidis, Advisory Chair for the Connecticut Chapter of CureSMA: In her testimony, Ms. Kaloidis shared her experience having SMA. She began showing symptoms when she was 9 months old. Her diagnosis was drawn out and consisted of numerous hospital stays and misdiagnoses. She was described as being a "lazy baby" which often happens in cases of SMA. These difficult months of searching could have been easily avoided by a simple blood test and her life could have been greatly and positively impacted. After her diagnosis, Ms. Kaloidis was given a prognosis of 2 years, and only a 5% chance of living past that age. She is currently 22 years old. The FDA approval of the first-ever treatment (SPINRAZA) has completely revolutionized the expectations of those living with this disease. Ms. Kaloidis shared the story of an individual who very early in life received this treatment and lives now, walking and dancing and free from the debilitating effects of untreated SMA. SMA is now part of the Recommended Uniform Screening Panel (RUSP), added on February 8, 2018. Ms. Kaldoidis hopes that we can see a similar result in Connecticut very soon. She strongly supports this bill.

NATURE AND SOURCES OF OPPOSITION:

None submitted.

Reported by: Kathleen Panazza

Date: April 2, 2019