

Public Health Committee JOINT FAVORABLE REPORT

Bill No.: HB-6919

AN ACT REQUIRING NEWBORN SCREENING FOR DUCHENNE MUSCULAR

Title: DYSTROPHY.

Vote Date: 3/5/2025

Vote Action: Joint Favorable

PH Date: 2/21/2025

File No.:

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SPONSORS OF BILL:

The Public Health Committee.

REASONS FOR BILL:

This bill came at the request of many members of the Public Health Committee seeking to add Duchenne Muscular Dystrophy (DMD) to the state's newborn screening program. The screening would start July 2027 and cost \$600,000. Currently, the newborn screening program requires health care institutions to perform newborn screenings using blood spot specimens between 1 or 2 days after the infant's birth.

RESPONSE FROM ADMINISTRATION/AGENCY:

Manisha Juthani, MD, Commissioner, Connecticut Department of Public Health (DPH):

The Commissioner stated that DMD is not on the Recommended Uniform Screening Panel (RUSP). Conditions listed on the RUSP are part of the comprehensive preventive health guidelines supported by the Health and Human Services (HHS) federal agency for infants and children and are required to be covered by health plans with no charge to the individual. DMD has been nominated for inclusion on the RUSP however a vote on its inclusion has yet to be scheduled. The department can't support the bill at this time as it would cost at least \$660,125 over two years, and this cost is not included in the Governor's proposed budget.

NATURE AND SOURCES OF SUPPORT:

Debra Ellis, Newborn Screening Program Manager, Connecticut Children's:

Ms. Ellis commented that on May 2018 the DPH partnered with Connecticut Children's to launch the Connecticut Newborn Screening Network. The first three years after the network was established there was a 71% decrease in the time between screening and diagnosis, from an average of 140 days down to 40 days. She mentioned that newborn screening for DMD would impact roughly 1 in 3,500 to 5,000 families and early detection will allow the start of treatment at a stage when it is most effective. She added that DMD is currently not on the RUSP. However, should it be added to RUSP, cautioned that careful planning will be necessary regardless of where the funding comes from. She strongly supports the addition of DMD once it is included in the RUSP.

Kathleen Cardinale, Physician, Yale University:

Ms. Cardinale has been a pediatric neurologist for 7 years and she has encountered several children with progressing motor delays which turned out to be DMD. She stated that children are typically between 3-5 years old at time of presentation and are already struggling to walk upstairs or stand up from a seated position. If left untreated, the condition would require a child to use a wheelchair full-time and decrease life expectancy. She stated that currently there are several treatment options for DMD, tailored to the particular genetic mutation. They are proven to significantly slow down the rate of muscle deterioration, but they cannot bring back lost muscle. She supports this bill that would allow children to be treated as soon as possible instead of waiting to present when the child displays deterioration of muscle function.

Melanie Kelly, Mother:

Ms. Kelly shared the story of her two sons who were diagnosed with DMD. With her first son, the provider initially said not to worry because children develop differently and that he was just a little behind. She ended up pregnant with her second son and when they took their second son to another provider, they were told that he might have Muscular Dystrophy. They were told over the phone the test results had come back abnormally high which is how they found out that their son had DMD. She added that DMD screening will allow parents to make an informed decision when it comes to having another child and that it reduces stress. The cost of the test is minimal compared to treatment once the condition has progressed.

Jennifer Handt, Founder, Charlie's Cure:

Ms. Handt shared her story of how her son was diagnosed with DMD. She pointed out that adding DMD to the RUSP is currently stalled, so states such as Minnesota, Ohio, New York, and Massachusetts have moved independently to adopt DMD screening.

The following submitted testimony in support of the bill similar to that expressed above:

- Maria Davila, Mother
- Christopher Curran, Co-Executive Director, Kindness over Muscular Dystrophy
- Bhaskar Roy, MBBS, American Academy of Neurology
- David Weinstein, MD, OBGYN Associates

NATURE AND SOURCES OF OPPOSITION:

None Expressed.

Reported by: Piotr Kolakowski

Date: 3/6/25