



Connecticut Department of Public Health

Testimony Presented Before the Public Health Committee

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House Bill # 5527 - An Act Concerning Globoid Cell Leukodystrophy

The Department of Public Health (DPH) provides the following information in regards to House Bill # 5527.

HB 5527 would require screening for globoid cell leukodystrophy, also known as Krabbe disease, as part of the Newborn Screening (NBS) panel conducted by the Public Health Laboratory (PHL).

Krabbe disease is a genetic disorder caused by an enzyme deficiency which affects nervous system development. In the infantile form, symptom onset is typically before one year of age, and includes muscular weakness, feeding difficulties, vision loss, seizures, and slowed mental and physical development. As the muscular wasting progresses, the infant's ability to move, chew, swallow, and breathe lessens. The infantile form of the disease is generally fatal before age 2. Incidence in the United States is approximately 1:100,000.

Although we understand the interest in addressing this devastating disease, the available science is not yet in place to support universal newborn screening. The panel of experts which comprise the Discretionary Advisory Committee on Heritable Disorders in Newborns and Children, within the federal Department of Health and Human Services, was petitioned to include Krabbe disease in the recommended uniform screening panel (RUSP). Following a careful review of available evidence, the Committee concluded that, while there could be a benefit of early identification and intervention, substantial harm is also possible, either from testing/identification, from treatment/other interventions, or both. The Committee determined not to recommend adding the condition to the recommended panel, noting that there are significant gaps in evidence that must be addressed before further consideration of its inclusion. Treatment for Krabbe disease requires stem cell transplantation and carries significant risks, which must be weighed against the current difficulty predicting which babies will be affected by the severe infantile form of the disease.

A very small minority of states are offering Krabbe testing within their NBS panel. Their experience is that screening is problematic for a variety of reasons, including the generation of high numbers of false positive results that require further confirmatory testing. This significantly increases the cost of screening, tracking, and follow-up testing. The situation is further complicated by the identification of infants with mutations of unknown clinical significance.

The Department does not support the addition of any new disorder to Connecticut's screening panel until such time as the Discretionary Advisory Committee includes this disorder in its RUSP. The Committee was petitioned to include Krabbe disease but has not done so at this time. Implementation of this screening would have significant costs as well as policy implications. Should this screening be mandated, the responsibility could not be conducted without additional resources for instrumentation, reagents, and staff. Consequently, due to the additional resources that would be needed to implement the requirements associated with this bill, the Department is not supportive at this time.

Thank you for your consideration of the Department's views on this bill.