



**Connecticut Department of Public Health**

**Testimony Presented before the Committee on Children  
March 6, 2014**

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**Senate Bill No. 285: An Act Concerning Newborn Screening for Krabbe Disease**

The Department of Public Health (DPH) would like to provide the following information regarding Senate Bill 285.

Senate Bill 285 would require screening for Krabbe disease as part of the Newborn Screening (NBS) panel tested 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 unfortunately not yet in place to support universal newborn screening.

The Secretary's 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. Following a careful review of available evidence, the Committee concluded that, while there could be a benefit from 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.

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