Hunter's Hope Foundation

Krabbe ~ Leukodystrophies ~ Newborn Screening

Newborn Screening for Krabbe and other Lysosomal Storage Disorders (LSD)

Bill Number: SB 284
Sponsor: Senator Boquist
Hearing Date: February 14, 2013

Please support SB 284, to add Krabbe, Pompe, Gaucher, Fabry, Niemann Pick or any other metabolic disorder resulting from a defect in lysosomal functionality to Oregon's Newborn Screening Panel.

In 2006, New York became the first state to begin screening for Krabbe disease. In August of 2012, Missouri began screening for Krabbe disease and one other LSD. New Jersey, Illinois, and New Mexico have all passed legislation for LSD screening and Illinois plans to implement statewide LSD screening in 2013. Although implementation dates vary by state, important progress is being made, as these diseases are thought to have a collective incidence of 1:5,000.

Screening is available for Krabbe and five similar LSDs (Fabry, Gaucher, Pompe, MPS I and Niemann-Pick A/B) in a single assay. Although the assays would need to be validated in the particular state as the cutoffs may vary, there is experience in the states previously mentioned for the Oregon lab to build upon.

Effective treatment for many of the LSDs is now available and supported by data gathered during clinical trials and published in the medical literature. Umbilical cord blood transplantation has been shown to significantly extend life and to improve the quality of life for children with LSDs. If the procedure is performed early in the course of disease, 80-90% of patients survive with good quality of life. This treatment has been shown to be effective in ameliorating the course of Krabbe disease and MPS I, and it is clear that treatment must be initiated at a very early age to be effective. In Krabbe disease, it must be initiated before any clinical manifestations appear. Enzyme replacement therapy is effective in the treatment of Gaucher disease, Fabry disease, Pompe disease, MPSI and MPS II and has been approved in each case by the Food and Drug Administration.

Should Oregon begin screening for these diseases, a quality assurance program for LSD newborn screening is in place at the CDC and would be available to the laboratory. Additionally, some reduction in costs can be anticipated – certainly the care of children with Krabbe disease and other LSDs, would be much less costly when children are functional and do not require continuous nursing care. There would also be a reduction in costs related to the birth of second affected children before the first child was diagnosed. Additionally, it is estimated that the medical cost of an untreated child affected by Krabbe and similar diseases costs a state at least $700,000 annually.

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