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### **Support for Senate Bill 465: An Act Requiring Newborn Screening For Adrenoleukodystrophy**

X-linked adrenoleukodystrophy (ALD) is a genetic disorder with an estimated incidence of 1:17,000 and it is known to affect all ethnic groups. The disorder has an adrenal gland component and a neurological presentation. The disorder in the adrenal gland is primary adrenal insufficiency or Addison's disease. This occurs in 90% of affected males. Individuals with adrenal insufficiency are at risk for severe decompensation at the time of infections or other physiologic stresses. The Addisonian crisis may result in substantial morbidity or even death. Because of the protean nature of adrenal insufficiency there is often a substantial delay in diagnosis and many individuals will have repeated events before being appropriately diagnosed and treated.

In terms of ALD neurologic presentation, 35% of boys will develop an inflammatory demyelinating condition referred to as childhood cerebral disease (CCALD) between the ages of 4 and 10 years of age. Boys with initially normal development will begin to manifest neuropsychiatric features such as changes in attention, learning, and behavior. Often misdiagnosed as ADHD, learning disorders, or autism, boys eventually go on to have progressive neurologic findings. They are usually diagnosed by evidence of widespread abnormalities on MRI. The disease is strikingly aggressive and affected individuals die within 2-3 years of initial presentation.

It needs to be highlighted that while the ALD clinical disease is very rapid in progression, the MRI abnormalities precede disease onset by months. The only available treatment for cerebral disease is allogeneic bone marrow transplant. This therapy can arrest the progressive demyelination, but is of no benefit when the disease is well-advanced. To be effective, a bone marrow transplant must be performed when there are early changes on MRI, but prior to neurologic findings. The optimal candidates are those who have undergone serial monitoring with MRI from an early age.

Because of the ability to intervene both for the endocrine (Addison's) and neurologic issues, it is critical to identify pre-symptomatic individuals. Newborn screening of dried blood spots is a near universal method of testing individuals and allows the diagnosis of a wide variety of genetic disorders. A newborn screen for ALD has been developed and piloted, and over 50,000 newborn dried blood spot samples have been screened to validate the test.

My son, Spencer, was born with ALD, but is now a healthy 13-year-old boy who is a loving young man, top student, and star of his school's swim team. After many years of misdiagnosis, Spencer's cousin Oliver was diagnosed with ALD. Spencer was one year old when this occurred. This led to Spencer being tested and learning that he too was born with ALD. When he was

two years old he had a cord blood transplant that stopped the disease. If newborn screening were available at the time his cousin Oliver was born, Oliver would be alive today. It took years of Oliver going to doctors to find out what was going on and by then it was too late to help Oliver, since transplants do not work at that stage of the disease. Oliver died at the age of 12, a few years after he was diagnosed. Spencer is alive and healthy because Oliver was the ALD screen for our family. Given an accurate and affordable newborn screen now exists and early diagnoses will allow life saving treatment, families should never again have to suffer the painful losses that our family did. All babies born with ALD should be identified at birth, so they too can be saved as Spencer was.

In summary, there exists over 30 years of experience in the diagnosis and evaluation of ALD. It is well established that the outcomes are vastly improved by the early identification and clinical monitoring of affected individuals. Therapies are available that can improve the quality of life for affected individuals. In fact, the most common manifestation of ALD, adrenal insufficiency, can be treated with steroid replacement, a simple and inexpensive medication. Finally, there is a methodology that allows us to accurately identify individuals off of the newborn blood spot.

Based on the information that we already have, we can estimate screening newborns for ALD will have a significant impact on the acute and sometimes catastrophic adrenal presentation and will alter the population of boys who are judged ineligible for bone marrow transplant because they are too advanced when diagnosed with ALD. It is for these reasons that we ask you to require that all newborns are screened for x-ALD.

Thank you for your time and consideration of this important, life-saving request.