



# STATE OF CONNECTICUT

DEPARTMENT OF PUBLIC HEALTH

## **TESTIMONY PRESENTED BEFORE THE JOINT COMMITTEE ON PUBLIC HEALTH February 27, 2013**

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### **Senate Bill 465 – AN ACT REQUIRING NEWBORN SCREENING FOR ADRENOLEUKODYSTROPHY**

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The Department of Public Health is opposed to Senate Bill 465.

Adrenoleukodystrophy (ALD) is an X-linked inherited disorder of very long chain fatty acid metabolism which results in a build-up of these fatty acids in the nervous system, adrenal glands and testes. The incidence of ALD is approximately 1/17,000 births, predominately affecting boys. The disease manifests itself in three forms: a cerebral form affecting children between the ages of four to eight; adrenomyelopathy, affecting men in their twenties; and adrenal gland failure. Other than treatment of adrenal gland failure with steroids, treatment for the other forms of ALD is still experimental and involves diet very low in very long chain fatty acids, supplemented with other oils (Lorenzo's oil). Bone marrow transplant is also being considered.

In October 2012, the Secretary of Health and Human Services' Advisory Committee on Heritable Disorders in Newborns and Children responded to a letter from The Stop ALD Foundation indicating that they would not nominate the disorder for external review because there was not sufficient data to support inclusion on the recommended Newborn Screening (NBS) panel. However, they also noted that the Mayo Clinic is undergoing a large pilot study testing for ALD, to be completed at the end of 2013, the results of which will be used by the Committee to decide whether to move ALD into the nomination and prioritization process.

Currently, there are no public health laboratories that screen newborns for ALD. As such, there are no standard methods available for the test. Consequently, it would take an enormous effort for the Connecticut Newborn Screening program to develop and validate an in-house method to test for ALD. Time will be required for method development and validation to enable routine screening to be implemented by the NBS laboratory. A minimum of 5,000 dried blood spots would have to be tested before the method could be validated. The Laboratory tests all newborns in Connecticut for over 40 disorders on its NBS panel. The addition of another disorder for routine screening, including method development and validation, would require additional staff, equipment and supplies. These resources are not included in the Governor's proposed budget.

Analysis for this disorder also presents additional problems with the identification of many individuals with one normal gene and one gene for the disorder, as well as secondary target disorders that will be inevitably identified through screening. This carrier and secondary disorder identification may prove problematic for treatment and follow-up at the level of the overseeing physician.

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

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