brian's hope
Stopping the progression of ALD

Jean Kelley DIRECTOR

Screening Newborns For Adrenoleukodystrophy

ADRENOLEUKODYSTROPHY

Adrenoleokodystrophy (ALD) is a life threatening disorder that often causes adrenal gland failure, as well as neurologic dysfunction affecting the spinal cord and/or brain.

The disease is passed down from parents to their children as an X-linked genetic trait.[1] It therefore affects mostly males, although some women who are carriers can have milder forms of the disease. It affects approximately 1 in 17,000 people from all races. The condition results in the buildup of very-long-chain fatty acids in the nervous system, adrenal gland, and testes, which disrupts normal activity. There are three major categories of disease:

- *Childhood cerebral form*—appears in mid-childhood (at ages 4–8)
- *Adrenomyelopathy*—occurs in men in their 20s or later in life
- *Impaired adrenal gland function (called Addison disease or Addison-like phenotype)*—adrenal gland does not produce enough steroid hormones

NEWBORN SCREENING

Connecticut law requires newborns to be screened in the hospital for a number of diseases including cystic fibrosis, severe combined immunodeficiency, critical congenital heart disease, HIV, sickle cell and other tests that determine if the newborn has inborn metabolic or other disorders (Section 19a-55).

The Federal government is currently monitoring a study underway at the Mayo Clinic's Biochemical Genetics Laboratory to develop the best possible screening test using dried blood spots from anonymous newborns to test for ALD. We expect the results of this work could be completed and forwarded to the federal panel that will consider newborn screening protocols in May 2013. A larger study would then occur before a final recommendation is made to the Uniform Screening Panel in 2014.

- ALD met all the criteria to be included in the uniform newborn screening panel and the initial preliminary results from the Mayo clinic were 12 positives out of 42,000 samples.
- Due to the extensive time this will take for each state to implement, too many children will be put at significant risk, many of them dying.
- The legislation we are seeking will add screening for Adrenoleukodystropy to the existing newborn screening panel.

LIFE-SAVING TEST:

- It is an inexpensive and sensitive screening test.
- It is an opportunity to detect complications from the disease before there are symptoms.
- There are treatments, which if given in the early phase, dramatically improve the outcome.
- If treatment is delayed until the condition is apparent clinically, the outcome is much worse.
- Up to this point, early, often life-saving treatment has been available only to those diagnosed because a relative suffered the disease.

1. www.nih.gov

BRIAN KELLEY

Brian Kelley is a 24-year old, young man from Branford who was diagnosed with ALD at the age of 6. Unfortunately, Brian was already symptomatic. There were no red flags in his family. Had the newborn screening for ALD been in place at the time of Brian's birth, he would not be facing the great challenges he does today. With great courage and tenacity, Brian has taught much to many in his very quiet way for the past 18 years. We hope we can honor Brian by making a difference in the lives of others diagnosed with ALD.

CONNECTICUT FACTS

- Connecticut Birth Rate: 37,708 per year
- Price of test: $1.50-2.00 per test
- Cost per year to add ALD newborn screening: $56,500-74,000

MISSION STATEMENT OF THE CONNECTICUT DEPARTMENT OF PUBLIC HEALTH

To protect and improve the health and safety of the people of Connecticut by:

- Assuring the conditions in which people can be healthy
- Preventing disease, injury, and disability, and
- Promoting the equal enjoyment of the highest attainable standard of health, which are a human right and a priority of the state

The time is right.... there is an accepted method and treatments.

Let's make this happen.

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