



THE EVANOSKY
FOUNDATION

We Win in the End

The Evanosky Foundation Supports Expanded Newborn Screening Within the State of Illinois To Include Lysosomal Storage Disorders

Fact Sheet prepared by The Evanosky Foundation

Background on Newborn Screening

- Newborn Screening is a mandated public health practice performed within the first 48 hours of an infant's life for the early identification of certain genetic, metabolic, hormonal and/or functional conditions.
- These conditions often have no immediate visible effects, but unless detected and treated early, can cause physical problems, mental retardation and even death. Early diagnosis and proper treatment can make the difference among death, lifelong disability or healthy development.
- The newborn screening test is performed within 2 days of birth. The baby's heel is pricked to obtain a few drops of blood, which are then sent to a laboratory for analysis. Parents of babies who test positive are contacted by the state's Department of Public Health.
- Each state can choose which tests it will administer in its newborn screening program. Because of this, significant disparities exist which can result in late diagnosis and inadequate treatment. Approximately four million infants are born annually in the United States, and of these, an estimated 150,000 are born with serious birth defects (3.75%).

About Lysosomal Storage Disorders (LSD)

- Lysosomal storage disorders are inherited genetic diseases in which a person's lysosomes—the microscopic recycling bins in his or her cells—do not function properly, causing unrecycled material to build up inside the lysosomes. Lysosomes contain many different enzymes that help them break down and recycle complex materials that the body can no longer use, but a person with an LSD is missing one of those enzymes.
- Lysosomal storage disorders have been recognized as one of the major groups of genetic disorders affecting children. With over 40 different disorders and a combined prevalence of up to one in 5,000 births, this group of disorders is a major public health problem and places an enormous burden on affected individuals and their families as well as the public and private health systems. Early identification and diagnosis is essential since the most serious and debilitating symptoms—particularly neurological and skeletal—often do not respond to therapy.

Testing for Lysosomal Storage Disorders (LSD) in Illinois

- According to the Illinois Department of Public Health, in 2007, Illinois will screen all newborns for the 29 disorders recommended by the March of Dimes and the American College of Medical Genetics (ACMG). Approximately 180,000 babies are born each year in Illinois.
- Due to technological advances in chemistry and tandem mass spectrometry, one test can be used to detect a number of lysosomal storage disorders, thereby reducing cost and effort. In addition, a number of LSD newborn test reagents will be available at no charge through the Centers of Disease Control (CDC) in early 2008.
- If Lysosomal Storage Disorder testing was implemented in Illinois, approximately 37 babies per year could ultimately be identified as having one of the 43 known lysosomal storage disorders. This is the equivalent of nearly two kindergarten classes each year. Currently, 5 of these disorders have validated newborn screening tests. Those disorders are Krabbe, Pompe, Niemann-Pick, Gaucher and Fabry.
- The equipment and resources needed to add these 5 LSD's would be purchased through available monies in the Metabolic Screening and Treatment Fund. In addition, it is anticipated that there would be a slight increase to the newborn screen fee to implement these five LSD disorders. **There would be no impact to the general revenue fund and no additional cost to taxpayers.**
- If it chooses to move forward with LSD testing, the State of Illinois can procure up to \$100,000 in private funding to support an initial LSD implementation study.

Recommendations by The Evanosky Foundation

- Contact your local Illinois State legislators and ask them to support SB1566. This bill will benefit the citizens of Illinois in three ways:
 1. SB1566 introduces Lysosomal Storage Disorder (LSD) Newborn Screening to the State of Illinois by screening newborns for Krabbe, Pompe, Gaucher, Fabry and Niemann-Pick diseases, and requires that screening begin by July 1, 2008.
 2. SB1566 protects the monies in the Metabolic Screening and Treatment Fund. In 2004, \$3.4 million was taken from this fund, which is used to provide newborn screening services, and appropriated into the general fund.
 3. SB1566 calls for the protection of the Genetic and Metabolic Diseases Advisory Committee, which advises the Illinois Department of Public Health on which future disorders should be included in its newborn screening test profile.
- Contact the Office of Rod Blagojevich, Governor of Illinois, to ask for his support of SB1566.

Supporting Agencies Include:

- Children's Memorial Hospital, Chicago
- The Evanosky Foundation
- Hunter's Hope Foundation
- Liam Hammonds Memorial Foundation
- PKU Organization of Illinois
- United Pompe Foundation
- Propionic Acidemia Foundation
- Acid Maltase Deficiency Association (AMDA)
- Illinois Academy of Family Physicians (IAFP)
- Save Babies Through Screening Foundation
- Chicago Center for Jewish Genetic Disorders

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