

Written comments to the Advisory Committee on Heritable Disorders and Genetic Disease in Newborns and Children

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Thank you for the opportunity to testify on this very important issue Newborn Screening.

I became involved in the important concept of early detection as a result of my son Hunter having Krabbe disease. Krabbe disease is an inherited neurodegenerative lysosomal enzyme disease affecting the peripheral and central nervous system. Without early detection, children like my son Hunter suffer through the rapid progression of this disease. My son Hunter is currently on oxygen 24 hours a day, cannot move or speak, is fed through a JG-Tube, receives treatments every four hours around the clock of chest therapy with medication, CPT and the VEST, and takes multiple medications. In contrast, children born with Krabbe who receive early identification have access to effective treatment with hematopoietic stem cells using umbilical cord blood. This Cord Blood Transplant prevents neurological damage, halts or alters the disease process, reverses the manifestations of the disease in the central and peripheral nervous systems, saves lives and preserves quality of life. It is because of the need for early identification that I am now involved with improving newborn screening!

If Hunter had received early identification, he would have had access to the effective treatment of hematopoietic stem cell transplant using umbilical cord blood. Children born with Krabbe disease who are identified pre-symptomatically (currently only possible in case-index families) have had their lives saved, are now growing up and are expected to live productive adult lives.

Our Foundation, the Hunter's Hope Foundation, which my wife Jill and I started in 1997 to increase awareness and accelerate the pace of research about Krabbe and related Leukodystrophies has already awarded more than \$3.8 million dollars in grants. Last weekend our 7th Annual Scientific and Family Symposium was attended by more than 30 families and a number of distinguished basic and clinical researchers. During the symposium I had the opportunity to spend time with many children born with Krabbe, including the children who have been treated pre-symptomatically. It is because of the dramatic difference between children like my son Hunter, who did not receive early identification and those who did, that I am speaking today. With a newborn screening test for four leukodystrophies (**Li Y, Brockmann K, Turecek F, Scott CR, Gelb, MH: Tandem mass spectrometry for the direct assay of enzymes in dried blood spots: application to newborn screening for Krabbe disease. Clin Chem. Mar; 50(3):638-40 2004.**) due to become available within the next few years I am here to share with you our commitment to ensuring that all children in all states receive all existing newborn screening tests possible.

Today, in the United States, thousands of our children are suffering and dying needlessly. I have heard appallingly large numbers, that thousands of infants in the United States, with treatable diseases, go untreated each year and die due to inequities in the current newborn screening system.

I looked into why this is happening. And what I found out is that medical research and Tandem Mass Spectrometry (MS/MS) technology have advanced more quickly than our implementation of them. Today, there are more than 60 diseases that can be tested, using MS/MS, and other technologies, all of which are treatable. Most treatments are effective only before symptoms are present. This means children MUST be diagnosed and treated as early in life as possible.

The current Newborn Screening (NBS) system is legislated by state. The range in number of diseases tested is Alabama screening for 4 diseases and Hawaii 48. Twenty-nine states currently screen for 10 diseases or less. New York tests for 12 diseases. Children are suffering and dying needlessly because they are born in the wrong state. A child's chances for life should not be dependent on where he/she is born. No child should be denied the right to a healthy life. Nor should parents' rights be denied to know that their children are at risk due to these inequities from state to state newborn screening. It is impossible to fully express the devastation these illnesses bring to the entire family...

There is a cost for freedom from disease, but the cost of the alternative is much, much greater. It extends far beyond our comprehension. I can't help but think that if we received a terrorist threat that thousands of our infants were going to be killed by the end of 2004, our Nation would use all the money and power we had to stop it and we would stop it! We have a worse threat right here, today, in our midst that is "silently" killing our children. It is within the very systems (NBS) that we established to help our children.

Why are the state public health departments NOT USING ALL AVAILABLE RESOURCES, including private sector resources to screen infants at birth? I DO NOT UNDERSTAND HOW THIS CAN BE IN THE BEST INTEREST OF PUBLIC HEALTH?

I know that once our legislators understand the importance of and need for immediate action on this issue, we are confident they will respond. We recommend that this Committee encourage Congress to require states to inform parents in writing of the potential for their children to receive additional newborn screening tests that may not be required under state law. We must start by mandating that the hospitals educate parents on the availability of supplemental NBS tests. This parental notification must be meaningful and informed and require consent. We must immediately put a plan in place for adding all testable/treatable diseases to every state in the nation's mandatory NBS list.

The solution seems so simple. Screening tests, technology, and treatments are all available today. We just need to use them. We need to fix our current NBS system so that currently available resources are used to give every child the right to a healthy life.