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On

"Newborn Screening Saves Lives: The Past, Present, and Future of the
Newborn Screening System"

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Chairman Hagan, Ranking Member Enzi, and Members of the Subcommittee – good morning. Thank you for the opportunity to testify today at this important hearing about newborn screening.

I am Natasha Bonhomme, Director of Baby's First Test, the nation's Newborn Screening Clearinghouse, the premier resource for newborn screening. We offer families and healthcare providers support throughout the newborn screening experience. In addition to being there when families need resources most and providing up to date information in an accessible manner, we bring family and public perspectives to the newborn screening dialogue. By increasing awareness, Baby's First Test offers millions of newborns and their families a chance at a healthy and informed start.

Imagine it is 2010 and you are a new parent. You receive a call from the pediatrician on a Friday afternoon and learned that your baby's newborn screening results were not normal. During the 7 years I have worked in newborn screening, countless parents have described this scenario to me, and they explain the anguish they felt as they had nowhere to turn until the following week when the doctor's office was open. Fast forward to today, and now 24 hours a day, 7 days a week, we are there for them.

What Does the Public Think?

It is critical to ask: "What do parents want and need?" when considering newborn screening. In 2008, Genetic Alliance, the parent organization of Baby's First Test, and partners conducted surveys and focus groups with more than 2,000 women about their attitudes and perspectives on newborn screening. This group was representative of the nation at that time in regards to race/ethnicity and socio-economic status. Some key findings of this survey are:

- 98% believed that newborns should be screened for conditions where early diagnosis can make a difference.
- More than 95% believed that newborn screening was important to help families prepare to care for a child with a condition.
- More than 94% believed that newborn screening was important to improve the health of babies.

There are few programs we can turn to that have this level of public support. It is clear to the vast majority of people that because newborn screening has the ability to save and improve lives, it should receive strong support. While 1 in 300 babies are identified with a treatable condition found through newborn screening, this program reaching nearly all of the 4 million babies born in this country annually.

Communication

From the research we conducted, a key need of parents and the public is actionable information.

- 86% wanted information on newborn screening either while planning a pregnancy or during the pregnancy. Only 44% remembered receiving information during this timeframe.
- 93% wanted information on what happens if there is an abnormal result.

- 89% wanted to know what specific conditions their baby was screened for.
- 88% wanted to know how they would be told of the results.

During our focus groups with families who had experienced an out of range result, yet had a healthy child (also know as a false positive result), parents told us about receiving a phone call from their pediatrician's office saying that something was wrong with the newborn screening results. Countless times parents said that they received little to no information about the condition and no resources on where they could learn more. On multiple occasions, these calls came before the weekend, leaving sleep deprived new parents to find information on next steps on their own. Now, 24 hours a day, 7 days a week, parents have a place to turn.

Newborn Screening Clearinghouse

During the same period of time we were conducting our research, the Newborn Screening Saves Live Act became law. This law provides a national framework to support educational programs for parents and grant initiatives for follow up care. The information gathered from parents, healthcare professionals, state newborn screening programs, and other experts in the field helped us formed the basis for the structure and content of the Newborn Screening Clearinghouse, also supported by the Newborn Screening Saves Lives Act. Fully launched 2 years ago, nearly 80% of the visitors to Baby's First Test are new to the site. This is to be expected as everyday thousands of babies are born and screened and parents must learn all they can about their new baby. Baby's First Test averages more than 15,000 visits per month. This grows steadily as we get the word out.

Key components of the online Clearinghouse include:

- Comprehensive and specific information on the variety of conditions screened in all 50 states, Washington DC, Puerto Rico, the U.S. Virgin Islands, and Guam. The vast majority of parents and providers find this offering indispensable.
- Guidance on what this experience will involve: when does it take place (most parents don't know) how parents will receive results, what to do if there is an abnormal result, and how to obtain additional testing.
- Detailed information on all conditions screened including condition descriptions, immediate follow-up steps, treatments, expected outcomes, and support services/organizations.
- Information for health professionals including links to diagnostic protocols, trainings and toolkits for nurses, and communication guides on how to speak with families about newborn screening.
- Information on living with a condition found through newborn screening including sections that shows stories of children identified through newborn screening and the healthy lives they lead.

Majority of the requests we get from parents have to do with obtaining results of their child's newborn screening, how to receive additional testing, and how can they share the story of their child being saved by this screening program.

The Clearinghouse also invests in local and national programs to support newborn screening through its annual Challenge Awards. These awards support sustainable newborn screening educational efforts throughout the country including but not limited to:

- Developing nurse education and public awareness campaigns (Iowa).
- Incorporating newborn screening education into a home visiting program (Virginia).
- Evaluating the experiences of nurse-midwives to better understand their barriers and improve their skill level (Michigan).
- Designing outreach programs to raise awareness on newborn screening amongst WIC program participants (Illinois).
- Creating parent and provider videos on screening for Critical Congenital Heart Disease, which have been used throughout the country as states implement this new screening policy.

Due to Genetic Alliance's 27-year history of bringing individuals, families, and communities into the dialogue about health, it is important to us that we provide an on ramp for families who want to become more involved in their communities. We provide training for parents interested in learning more about and making an impact in newborn screening. The Consumer Task Force on Newborn Screening was created to engage relevant stakeholders with an interest in newborn screening policies, activities, and current events. Members are chosen through a competitive application process to participate in a one-year program. The three components of this program are training, project development, and project execution. We train members of the Task Force on issues relevant to newborn screening and implement projects targeting groups who typically are under-informed about the importance of newborn screening. After serving on the Task Force, members are equipped with the skills and knowledge to continue work on newborn screening programs or other maternal and child health-related issues.

Through this program, members of the Consumer Task Force have been able to:

- Present comments to the Secretary's Advisory Committee on Heritable Disorder in Newborns and Children.
- Share their experiences at national and international conferences.
- Expand their project management skill set.
- Join their state newborn screening advisory committees.

While the Clearinghouse has made great strides since its launch, we are eager to do more. Baby's First Test will undertake these additional projects this year:

- Develop a Spanish-language version of the Baby's First Test site that not only provides a translation of all 100,000 pages of newborn screening information but also addresses specific issues and concerns of the Latino community in a culturally competent fashion.
- Conduct a follow up national survey on attitudes and perspectives on newborn screening to evaluate newborn screening awareness initiatives and to track the needs of parents.

Cost savings of newborn screening

Newborn screening not only saves lives but it also saves money. Information available through the Association of Public Health Laboratories indicate that the cost of treating severe combined immunodeficiency (SCID) also known as 'bubble boy' disease can reach over \$2 million. This fatal disease can be cured if a baby is identified early and given a bone marrow transplant. If this transplant is done within the first 3.5 months of life it typically costs around \$100,000. Another example of cost savings is in congenital hypothyroidism, one of the most common conditions detected by newborn screening. It is estimated that nearly \$400 million per year is saved by identifying babies early and providing them treatment, preventing devastating IQ loss.

The Newborn Screening Saves Lives Act has been instrumental in educating parents and providing support for newborn screening, both through the Clearinghouse and through other programs. However, there is still much to be accomplished. Even though this year we celebrate 50 years of newborn screening, a program that the Centers for Disease Control and Prevention named one of the great public health achievements in the nation, we know that most expecting or new parents do not know what newborn screening is, or what their states do or do not screen for. This needs to change. As the data shows, newborn screening is a first step for a healthy start for our nation's youngest citizens. The parents mentioned earlier are grateful for your support.

Thank you for this opportunity to speak to the Subcommittee. I hope my testimony has been informative and thought provoking and I look forward to answering questions.