

**NEWBORN SCREENING SAVES LIVES: THE PAST,
PRESENT, AND FUTURE OF THE NEWBORN
SCREENING SYSTEM**

HEARING

BEFORE THE

SUBCOMMITTEE ON CHILDREN AND FAMILIES
OF THE

COMMITTEE ON HEALTH, EDUCATION,
LABOR, AND PENSIONS

UNITED STATES SENATE

ONE HUNDRED THIRTEENTH CONGRESS

FIRST SESSION

ON

EXAMINING HOW NEWBORN SCREENING SAVES LIVES, FOCUSING ON
THE PAST, PRESENT, AND FUTURE OF THE NEWBORN SCREENING
SYSTEM

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**NEWBORN SCREENING SAVES LIVES: THE
PAST, PRESENT, AND FUTURE OF THE NEW-
BORN SCREENING SYSTEM**

THURSDAY, SEPTEMBER 26, 2013

U.S. SENATE,
SUBCOMMITTEE ON CHILDREN AND FAMILIES,
COMMITTEE ON HEALTH, EDUCATION, LABOR, AND PENSIONS,
Washington, DC.

The subcommittee met, pursuant to notice, at 10:03 a.m. in Room SD-430, Dirksen Senate Office Building, Hon. Kay Hagan, chairman of the subcommittee, presiding.

Present: Senators Hagan, Casey, and Enzi

OPENING STATEMENT OF SENATOR HAGAN

Senator HAGAN. I want to welcome everyone to this morning's hearing in the HELP committee's subcommittee on Children and Families.

I want to thank all of our witnesses. Thank you for being here today, thank you for your work, and thanks for taking the time to come from all across the country. I really look forward to hearing your testimony.

I want to especially thank our Ranking Member, Senator Enzi, for his work and for his staff's work on this hearing. I am sincerely looking forward to working with my colleague to move the reauthorization of the Newborn Screening Saves Lives Act throughout the Senate this Congress, and I am proud to have you as a cosponsor of this bipartisan bill. Thank you, Senator Enzi.

This morning, we are here to discuss the past, the present, and the future of the newborn screening system in the United States. I sit not just as chairman of this subcommittee, but as a chair mom, because as a mother of three, I know from personal experience that when you have a child, your first hope and prayer is that your child is healthy. "Let our child be healthy," every family says. That is the one thing that every parent is praying for.

And thanks to advances to medical technology, the vision of medical professionals, and the daily work of nurses, doctors, and lab technicians, we now have the ability to detect and to treat dozens of life threatening conditions before they are able to cause serious harm. But it was not always this way.

Our system has developed over the course of decades. In fact, this month, we recognize the 50th anniversary of newborn screening. In 1963, Massachusetts, Delaware, and Oregon became the

first States in the Nation to mandate universal newborn screening, and the first condition that we screened for was PKU.

About 1 baby in 19,000 is born with PKU in the United States every year. These babies appear normal for the first few months of life, but unprocessed proteins will build up in their bloodstream and cause developmental delays if no action is taken. Thanks to the dried blood spot test that Dr. Guthrie developed so many years ago—and that we still use today—babies can avoid that fate with simple changes to their diet; just amazing.

Later, screens were developed for new conditions like sickle cell disease and cystic fibrosis, and new technology, like tandem mass spectrometry and DNA extraction, drastically expanded our ability to quickly and accurately screen newborns for many more conditions with shorter waiting times for results. These advantages are lifesaving, but only for those who lived in the States where they were actually implemented.

Some States lagged behind others in adopting new methods and technologies. As a result, a baby born with a condition that is inherited might receive the proper treatment in one State, but go undetected in another.

In fact, in this very room, 11 years ago in the last hearing that the Senate held on newborn screening 11 years ago, Senator Chris Dodd, who was then the chairman of this subcommittee said, “There is an enormous disparity in the newborn screening between the various States in our country. Only two States,” at that time, “Only two States will test for all 30 disorders. The vast majority test for 8 or fewer.” That was 11 years ago.

The situation cried out for Federal leadership. Thanks to the work of Dr. Rodney Howell—who was the first chairman of the Secretary’s Advisory Committee on Heritable Disorders—Dr. Howell is with us today, and I am so pleased. I thank you for your work over so many years. In addition, thanks to the work of the American College of Medical Genetics, the March of Dimes, the Department of Health and Human Services, and many others, a consensus document was developed that recommended to the States which conditions to screen for.

Congress also recognized the problem and passed the Newborn Screening Saves Lives Act of 2008, which cemented the role of the advisory committee in reviewing new conditions, and established Federal support for educating parents, researching new screening technologies, and ensuring the validity of existing screening tests. Today, all the States in the United States screen for at least 27 out of 31 recommended conditions. This is a dramatic improvement and a triumph for the American people.

In 2011, the CDC recognized the advances in newborn screening as 1 of the 10 great public achievements in the United States for the decade 2001 to 2010. I think that is something that we, in America, have to really be proud of.

That is why I am proud to take the lead with Senator Hatch on this reauthorization in building on the progress we have made so far by reauthorizing the Newborn Screening Saves Lives Act. Our bipartisan bill, of which Senator Enzi is a sponsor, focuses on: ensuring followup care for all newborns, expanding research on the long-term health impacts of newborn screening, establishing time-

lines for the review of new conditions to recommended States for screening, and continuing NIH research aimed at identifying new treatments for conditions that can be detected through newborn screening, and developing new screening technologies.

I look forward to working with the cosponsors of this bill—Senator Hatch, Senator Casey, Senator Enzi, and hopefully many others—to pass this bill this Congress. Simply put, newborn screening saves lives.

To tell us how this system works from a variety of perspectives, we have a great panel of witnesses today. I ask each of our witnesses to keep your opening statements to less than 5 minutes, and I thank you for your written statements, which have been submitted for the record.

I now want to turn to my colleague, Senator Enzi, for his opening remarks.

OPENING STATEMENT OF SENATOR ENZI

Senator ENZI. Well, thank you, Chairman Hagan. I appreciate the great work of you, and Senator Hatch, in coming up with this bill, and the history that you just covered. It is very helpful.

I appreciate the witnesses who have taken valuable time out of their time to help educate us. And the record will definitely do that with all of the Senators so that hopefully we can get this brought up before the full committee, get it onto the floor, and get it taken care of.

I have said before, and I think it is even truer today, that we need to spend more time listening to the thoughts and ideas of our constituents rather than presuming that we, here in Washington, have all the answers.

Screening every new baby for these serious health conditions—many of which would be otherwise undetectable for months or even years—is an important public health priority for States and the Federal Government. I think Mrs. Mullis' testimony will underscore just how meaningful these screenings programs are for the children and families whose lives are affected by these terrible health problems.

Therefore, I am particularly pleased to see that Chairman Hagan's bill improves the process for Health and Human Services, and the advisory committee, to review the evidence on potential new screening tests, and places a priority on screening for conditions where new treatments or therapies might already be in the works. We must continue to support medical and scientific innovation along with basic research if there is to be hope that we can further improve the lives of babies and children afflicted with these conditions.

Again, I look forward to hearing from all of the witnesses about the significant health benefits that newborn screening programs have provided for the last 50 years, as well as what they envision for the future of newborn screening.

I want to thank you all for being here and I know that afterwards, the record will be open for additional questions too for those who are not able to make it to the hearing. Thank you very much for participating and thank you, Chairman Hagan.

Senator HAGAN. Thank you, Ranking Member Enzi. I am so glad we can work together on this bipartisan bill. It is always a pleasure to work with my colleague, Senator Enzi.

Our first witness today is Ms. Natasha Bonhomme, the director of Baby's First Test, our Nation's clearinghouse of newborn screening information and education for parents and healthcare professionals.

Miss Bonhomme.

**STATEMENT OF NATASHA BONHOMME, DIRECTOR,
BABY'S FIRST TEST, WASHINGTON, DC**

Ms. BONHOMME. Chairman Hagan, Ranking Member Enzi, good morning.

Thank you for the opportunity to testify today on this important hearing about newborn screening. I am Natasha Bonhomme, director of Baby's First Test, the Nation's newborn screening clearinghouse. We offer families and healthcare professionals support throughout the newborn screening experience. In addition to being there when families need resources most, we bring family and public perspectives to the newborn screening dialog.

Imagine it is 2008 and you are a new parent. You receive a call from the pediatrician on a Friday afternoon and learn 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. They explain the anguish they felt as they had nowhere to turn to 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.

It is critical to ask: what do parents want and need in regards to newborn screening? In 2008, Genetic Alliance, the parent organization of Baby's First Test, and our partners, conducted surveys and focus groups with over 2,000 women to understand their attitudes and perspectives on newborn screening.

Some of the key findings of this survey were that 98 percent believe that newborns should be screened for conditions where early diagnosis can make a difference. More than 94 percent believe 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, this program reaches nearly all of the 4 million babies born in this country annually.

A key need of parents and the public is actionable information. Of those surveyed, 93 percent wanted information on what happens if there is an abnormal result and 89 percent wanted to know what specific conditions their baby would be screened for.

During the same period of time, we were conducting our research. The Newborn Screening Saves Lives Act became law. This law provides a national framework to support education and evaluation programs. The information gathered from parents, healthcare professionals, State newborn screening programs, and other experts in the field helped us design the structure and the content

of the newborn screening clearinghouse, also supported by the Newborn Screening Saves Lives Act.

Fully launched 2 years ago, nearly 80 percent of our visitors are new to the site. This is to be expected as every day, thousands of babies are born and screened, and parents must learn all that they can about their new baby. We average more than 15,000 visits per month, and this grows steadily as we are able to get the word out about this resource.

Some of the key components of the online clearinghouse include: comprehensive and specific details on all 50 State and territory newborn screening programs. The vast majority of parents and providers find this offering indispensable.

We also provide information on what exactly newborn screening is, what to do if there is an abnormal result, as well as condition-specific information such as description, followup care, as well as support services. We also have information for health professionals, including links to diagnostic protocols, trainings and tool kits, and communication guides on how to speak to families about screening.

While the clearinghouse has made great strides since its launch, we are eager to do more. This year, we plan to develop a Spanish language version of the site that not only provides a translation of all 100,000 pages of newborn screening content that we have, but also addresses specific issues and concerns of the Latino community in a culturally competent fashion.

We also plan to conduct a followup national survey to evaluate newborn screening awareness initiatives and to track the needs of parents.

The Newborn Screening Saves Lives Act has been instrumental in educating parents and providing support for newborn screening. This year, we celebrate 50 years of newborn screening. However, we know that most expecting and new parents still do not know what newborn screening is and what their State screens for. We are working to change that. As our data shows, newborn screening is the first step in a healthy start for our Nation's youngest citizens.

Thank you for this opportunity to speak to the subcommittee. I look forward to answering any of your questions.

[The prepared statement of Ms. Bonhomme follows:]

PREPARED STATEMENT OF NATASHA F. BONHOMME

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 dialog. 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 percent believed that newborns should be screened for conditions where early diagnosis can make a difference.
- More than 95 percent believed that newborn screening was important to help families prepare to care for a child with a condition.
- More than 94 percent 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 reaches 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 percent wanted information on newborn screening either while planning a pregnancy or during the pregnancy. Only 44 percent remembered receiving information during this timeframe.
- 93 percent wanted information on what happens if there is an abnormal result.
- 89 percent wanted to know what specific conditions their baby was screened for.
- 88 percent 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 Lives Act became law. This law provides a national framework to support educational programs for parents and grant initiatives for followup 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 percent 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 followup steps, treatments, expected outcomes, and support services/organizations.
- Information for health professionals including links to diagnostic protocols, trainings and tool kits 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 dialog 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 1-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 followup 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 know 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.

Senator HAGAN. Thank you very much for your testimony.

Our next witness is Dr. Rodney Howell, currently a Professor of Pediatrics at the University of Miami School of Medicine, but also one of the leading researchers and advocates in the history of newborn screening. Dr. Howell, we are certainly honored to have you with us today.

STATEMENT OF R. RODNEY HOWELL, M.D., PROFESSOR OF PEDIATRICS, UNIVERSITY OF MIAMI SCHOOL OF MEDICINE, MIAMI, FL

Dr. HOWELL. Thank you very much, Madam Chairman Hagan, Ranking Member Enzi, and members of this committee.

Thank you very much for inviting me here today to talk to this important committee.

I have had the opportunity to see, firsthand for nearly 50 years, the remarkable accomplishments of the newborn screening program in the United States. The current panel of conditions implemented by the States will, this year, identify 5,000 children with hearing loss, 2,100 with hypothyroidism, 1,775 children with sickle cell disease, 1,250 children with cystic fibrosis, and additional serious conditions for a total of 12,500 children whose lives will either be profoundly altered or saved due to newborn screening.

As a physician and a geneticist, I am very encouraged by the therapeutic pipelines in development that represent great promise of new science and hold potential that we may help many more families and children.

Two examples of how advances in sciences will impact newborn screening in the coming years involve Duchenne muscular dystrophy and spinal muscular atrophy. Both of these devastating conditions have drug therapies currently under development, which will likely be of great benefit, but they will require to be administered very soon after birth. Therefore, this will require the availability of newborn screening for these conditions.

The NIH Hunter Kelly component of the Newborn Screening Saves Lives Reauthorization of 2013 is really essential. It is a very important part of the legislation that will support the research needed to develop new therapy for conditions for which we currently lack treatment, and there are a considerable number of those.

Now that we have treatments for conditions that can be diagnosed and treated as a result of newborn screening, we will also need to have continuing support for large pilot programs for the study of the long-term outcomes of children and infants diagnosed as a result of newborn screening.

Newborn screening also has a potential of actually saving money in our challenged healthcare system. SCID, or Severe Combined Immunodeficiency as it is officially known, is a condition where infants are born lacking an immune system. If a baby with SCID is not diagnosed at birth, the outcome is death in infancy, but usually

only after weeks or months in a hospital intensive care unit battling life threatening infections.

In addition to the enormous emotional burdens to the family, there are medical bills that routinely exceed hundreds of thousands of dollars. On the other hand, if a baby with SCID undergoes newborn screening as we currently recommend, and is identified at birth at a cost of well under \$20, the baby can receive lifesaving umbilical transplant in the outpatient clinic over a period of days, and that transplant will cost under \$50,000.

I am extremely proud of the committee's work and their thoroughness, and believe that Senate bill 1417 builds on the accomplishments of the newborn screening program; will allow the committee to continue to deliver the latest evidence-based diagnoses and treatments for now and in the future; and holds tremendous promise for genetically based therapies that will benefit our Nation's children and families.

I greatly appreciate the support of each member of this committee for your continuing interest and support in this important legislation.

Thank you very much.

[The prepared statement of Dr. Howell follows:]

PREPARED STATEMENT OF R. RODNEY HOWELL, M.D.

Chairwoman Hagan, Senator Enzi and members of this subcommittee thank you very much for inviting me to testify today on Newborn Screening Saves Lives: The Past, Present and Future of the Newborn Screening System. I am a pediatrician who specializes in genetic disorders that produce serious biochemical abnormalities in children and was beginning my career at Johns Hopkins when newborn screening was beginning in Maryland in the 1960s. I have had the opportunity to see firsthand for over nearly 50 years the remarkable accomplishments of our newborn screening programs in the United States.

Children with an inherited condition known as phenylketonuria, or PKU, if untreated have profound developmental delay with an average IQ of less than 20. This means that such untreated children, who have a normal life-span, are unable to speak or care for even simple needs, and require full-time care. They are robbed of many of life's opportunities. Over 50 years ago, it was shown that babies with PKU identified at birth and treated with a very special diet could grow into adults with normal abilities. Dr. Robert Guthrie at the State University of New York in Buffalo solved a key problem, and developed a reliable, inexpensive test that could be done on all babies born in this country. This led to the beginning of newborn screening, which is carried out in every State under the leadership of the individual State health departments.

The use of the Guthrie test, or the PKU test, fairly quickly spread throughout the United States. And this week, we are celebrating the 50th anniversary of our Newborn Screening program. Since the beginning, newborn screening has been carried out under the aegis of the State Health Departments and has always been among the most successful preventive health programs in this country. And today, we have thousands of adults, treated for PKU from infancy functioning well in all the walks of life.

Since the benefit of the early diagnosis and treatment of PKU was so very dramatic, individual States, which are responsible for newborn screening, began to add tests for other conditions, using the same blood sample, to their newborn screening programs. Such conditions as congenital hypothyroidism were among the more common additions since early diagnosis and treatment of this condition also can prevent substantial developmental delay. But since each State has its own advisory panels, there developed considerable variation among the States. This variation was not only in the specific conditions being tested, but also the numbers of conditions included in the screening panel. In other words, whether your child would be identified to have a serious medical condition and receive the necessary life-saving medical intervention simply depended on the State in which your baby was born. This became a big problem for at-risk families who moved to another State between preg-

nancies. It was a lottery that the public health system never intended and consistency between the States needed to be established.

Early efforts at harmonization of screening panels between States began when the Maternal and Child Health Bureau/HRSA charged the American College of Medical Genetics to evaluate the scientific and medical information related to screening for specific conditions, and to make recommendations based on this evidence. They convened an expert group which produced a report which recommended a uniform screening panel and system.

Then Title XXVI of the Children's Health Act of 2000 enacted sections of the Public Health Service Act which established the Advisory Committee on Heritable Disorders in Newborns and Children (Committee), which held its first meeting in 2004.

The Advisory Committee on Heritable Disorders in Newborns and Children was established to provide advice to the Secretary of Health and Human Services on newborn screening. It was my privilege to serve as the founding chairman of this committee and continue in this role for the committee's first 8 years. When the committee first began its work, there was extraordinary variation among the States in screening programs. In the year 2000, 35 percent of the States were testing for fewer than 5 conditions, and 65 percent were testing for 5–10 conditions—none were testing above this number. Early in its work, the committee after careful review and study accepted the report of the American College of Medical Genetics and recommended that the more than 4,000,000 babies born each year in the United States be tested for 29 specific disorders including certain metabolic, and hearing deficiencies in early 2005.

It has been most gratifying to see how the various States have responded to recommendations from the Advisory Committee. Although States are responsible for their own screening programs, and virtually every State has an advisory committee that oversees decisions for that individual State, it is extremely difficult (even for large States) to have the extensive expertise required in the evaluation of these individually rare inherited conditions. The Advisory Committee membership contains or has access to all the required expertise. The legislation under which the Advisory Committee works also requires that all recommendations for inclusion in the newborn screening panel be evidence-based. As the committee has made recommendations, the States have been extremely responsive in reviewing these recommendations in light of their own needs, and in virtually every situation has adopted the recommendations of the Advisory Committee.

The committee has established a program for the recommendation of other conditions to be added to the recommended uniform screening panel, or the RUSP. It is felt that any individual, group, or organization should be able to submit a nomination to the committee for a condition to be added to the recommended RUSP. In order to accomplish this the committee (<http://www.hrsa.gov/advisorycommittees/mchbadvisory/heritabledisorders>) has developed a form outlining the exact information needed and directions for presenting such a nomination. To date, 10 completed nominations for new conditions to be considered for addition have been submitted to the committee. After careful review by the committee, and evidence review that would be necessary for consideration for newborn screening, three additional conditions have been recommended by the committee for addition to the RUSP. The Secretary of HHS has approved two of these (severe combined immune deficiency and critical congenital heart disease) and is currently considering the recommendation of the third, Pompe Disease.

It is important to emphasize that the conditions that are included on the newborn screening panel all result in serious medical complications (e.g., developmental delay) and/or death if not recognized early. All children with these conditions benefit from early diagnosis and treatment.

Since the passage of Public Law 110–204 in 2008 (Newborn Screening Saves Lives Act of 2008) there has been great harmonization among the States, and at the end of 2010, 100 percent of U.S. births were screened for over 30 conditions. And as a result of these expanded screening programs lives have clearly been saved.

The current implementation by the States of the core panel of conditions (not including severe combined immune deficiency and critical congenital heart disease both of which are in the process of being implemented across the country) will identify 5,064 children with hearing loss, 2,156 with primary congenital hypothyroidism, 1,775 children with sickle cell disease, 1,248 children with cystic fibrosis, and 239 children with medium-chain acyl-CoA dehydrogenase deficiency, and other important conditions for a total of 12,500 children yearly whose lives will be either profoundly altered or saved due to newborn screening.

The Secretary's Advisory Committee has worked tirelessly to meet the Nation's public health needs and the needs of our children. I am particularly proud of the rigor that it has applied to the evidence review of conditions that have been nomi-

nated for consideration to the committee. As a physician and a geneticist, I am equally encouraged by the therapeutic pipelines in development that represent great promise of new science and hold potential that we may help many more families and children. Certain of the mucopolysaccharide storage diseases are well-positioned, with new approved therapies, to be considered for addition to the newborn screening panels.

There are many new opportunities on the horizon but two come to mind. Two examples of how advances in science will impact newborn screening in coming years are Duchenne Muscular Dystrophy and spinal muscular atrophy. Both of these disorders result in profound and devastating health consequences for the affected children. In both these conditions, drug therapies are currently under development which will likely be of the greatest benefit if administered, presymptomatically, which will be very soon after birth. The availability of newborn screening programs for these disorders will be essential to benefit maximally from any new treatments.

The NIH Hunter Kelly component of the Newborn Screening Saves Lives Reauthorization of 2013 is an essential part of the legislation that will support research needed to develop new therapies for conditions for which we currently lack treatment. Some of our vexing conditions in the newborn, which we could readily detect through newborn screening, currently lack safe and effective treatment.

Now that we have treatments for conditions that can be diagnosed and treated as a result of newborn screening, we need additional support for the study of the long-term outcomes of infants treated as a result of newborn screening.

As other conditions are recommended for addition to the RUSP, we will need to identify funding and partners for large pilot research projects prior to the implementation of a program throughout the country. Prior to the full implementation of the newborn screening for severe combined immune deficiency, a large pilot study was carried out that was a great example of cooperation between the public sector organizations, and a not-for-profit Foundation.

Public information about newborn screening has been recognized for a long time as not only important but lacking. Some public concern about the use of residual blood samples has in my opinion been linked to a lack of understanding about the program itself. The HRSA Clearing House for Newborn Screening Information and the National Newborn Screening and Genetic Resource Center will go a long way to address these needs.

The CDC Newborn Screening Quality Assurance Program is known throughout the world for its excellent work. This program has been, and remains, vital to the entire newborn screening program. As I travel the United States as well as Europe, Asia and the Middle East to meet with local leaders dealing with newborn screening, this distinguished program is routinely identified as vital. And this group's provision of quality assurance materials is essential to the development of new tests, and the assurance that our testing procedures are working well.

It is critical to the health of our infants that the Nation's newborn screening programs be reauthorized with the passage of the Newborn Screening Saves Lives Reauthorization Act of 2013.

In consideration of the life-altering potential advances on the near horizon for so many of our Nation's children, I want to call particular attention to the new "Priority Review" section of the legislation which serves to strengthen the Federal newborn screening program. In our current newborn screening programs, we are regularly concerned with delays of days during which an affected infant, if not identified, can die or be damaged. And in considering new treatments, if there is a beneficial treatment to be considered, delays mean lost lives.

Under the reauthorization, there will be consistent and predictable time periods allowed completing the evidence review process. It will be most important that we work to ensure that sufficient funds are available for these costly and intense evidence reviews required by the committee. I believe that the impact of these timelines will encourage nominees to develop and submit more complete nomination packages and will provide the review committee an appropriate period of time to thoroughly and completely review the nomination to determine whether the condition meets all of the critical scientific standards necessary to warrant addition to the RUSP. It will require a lot of hard work, and of course we cannot afford any shortcuts since babies lives are at stake.

Equally important, this legislation will encourage the committee to more closely align its activities with the development of new and emerging interventions to narrow the gap between the approval of new treatments and the ability to identify the babies who could be saved if identified through newborn screening—again without undermining or diminishing the role of science in the committee process.

The individuals who serve on the Secretary's Advisory Committee do an incredible job of balancing limited public health resources with the goal of identifying babies

who could benefit from newborn screening. Not only does newborn screening save lives, the program actually represents overall cost savings to the American healthcare system especially important at this time of extraordinary restricted funds. Medical interventions following newborn screening can prevent or ameliorate severe, childhood-onset diseases and reduce the financial burden of intensive care hospitalizations.

SCID (severe combined immune deficiency) where infants are born lacking an immune system provides a very clear case study demonstrating the importance of newborn screening. If a baby with SCID is not diagnosed at birth, the outcome is death in infancy but only after weeks or months in a hospital intensive care unit battling life-threatening infections. In addition to the enormous emotional burdens to families as well as lost time at work for parents there are medical bills that routinely exceed hundreds of thousands of dollars. Unfortunately, in the end it is common that a baby with SCID doesn't survive this long hospital ordeal, so the devastating loss of a child is added to the family's burden. On the other hand, if a baby with SCID undergoes newborn screening and is identified at birth at a cost of no more than \$20, the baby can receive a life-saving umbilical blood transplant in the outpatient clinic over a period of days at a total cost of around \$50,000.

We are at a unique point in history. The mapping of the Human Genome is now complete. Genetics has moved out of the laboratory and into the clinic, where its applications can save lives every day. The current progress in the development pipeline of genetically targeted therapies is tremendous.

I am very proud of the committee's work and thoroughness and believe that S. 1417 builds on the accomplishments of the newborn screening program and will allow the committee to continue to deliver the latest evidence-based diagnoses and treatments for now and in the future which holds tremendous promise for genetically based therapies that will benefit our Nation's children and their families.

Senator HAGAN. Thank you, Dr. Howell. And certainly, as a mom, thank you for all the research you have done on pediatrics for so many years, and for the difference this has made in the lives of so many children nationwide.

Our next witness is Dr. Jennifer Howse, president of the March of Dimes which, as an organization, has been involved in the development and spread of newborn screening for more than 50 years.

Dr. Howse, thank you on behalf of all the work you do for the March of Dimes, and for being here today.

**STATEMENT OF JENNIFER L. HOWSE, Ph.D., PRESIDENT,
MARCH OF DIMES, WASHINGTON, DC**

Ms. HOWSE. Thank you very much and good morning, Madam Chair and Ranking Member Enzi.

I want to begin just by commending both of you for representing States that are currently offering 30 out of the 31 recommended conditions with a great deal of attention on the remaining test; so just commendations to both of you.

As president of the March of Dimes, I have the privilege of representing a very unique partnership of scientists, clinicians, parents, and volunteers who work together to prevent birth defects, pre-term birth, and infant mortality. So I very much appreciate the opportunity to testify before you today on newborn screening, which is truly one of the great public health victories of the early 21st Century.

Newborn screening is critically important. It is a highly effective public health program that tests every newborn for certain genetic, metabolic, hormonal, and functional conditions that are not otherwise apparent at their birth. Approximately 1 in every 300 newborns has a condition that can be detected through screening.

Newborn screening detects conditions that, if left untreated, can cause disability, developmental delay, prolonged illness, or even

death. But if diagnosed early through screening, these disorders can be managed successfully, thus reducing not only the physical burden of the disease, but also the associated economic burden on families, communities, and our Nation.

This year, we celebrate the 50th anniversary of newborn screening. The March of Dimes is very proud of our decade-long involvement in the history, and funding, and research that has helped to lead to contributions for the development of new screening tests.

The progress of newborn screening over the past two decades did persuade the Congress to pass the Newborn Screening Saves Lives Act in 2008. The law renewed and updated various programs that underpin the States' newborn screening efforts and the Federal Secretary's Advisory Committee on Heritable disorders. The law is now due for its regular 5-year renewal. Passage of the Newborn Screening Saves Lives Reauthorization Act is essential to the continued success of the newborn screening programs across our Nation.

Very importantly, reauthorization will ensure the uninterrupted continuation of the Secretary's Advisory Committee on Heritable Disorders and its vital work to maintain and update the recommended uniform screening panel that States use to consider, to adopt, and to implement new conditions. And so, that committee provides also ongoing planned evidence reviews and its work should be able to be continued uninterrupted.

The Newborn Screening Saves Lives Reauthorization Act also extends very, very important programs at HRSA, CDC, and NIH which include seven genetic and newborn screening regional collaborative groups, and a national coordinating center to support States' capacity. The Critical Congenital Heart Disease Newborn Screening Demonstration Program, which is improving protocols for point of care screening; Baby's First Test, which you have already heard about, a wonderful national education resource for parents; the Newborn Screening Technical Assistance and Evaluation Program, which serve to help States evaluate the effectiveness of their screening programs; a quality assurance program, which continues to upgrade the accuracy of newborn screening tests; and the very important Hunter Kelly Research Program at NIH, which supports grants and contracts to improve technology related to newborn screening.

Today, 42 States and the District of Columbia require screening for at least 29 of the recommended 31 conditions. Millions of babies have been screened for dozens of disorders, and in thousands of cases, the health and well-being of these children has been preserved. Newborn screening also represents a model, we believe, of Federal, State, public health partnership that has produced extraordinary improvements in child health.

So we urge you not to let this vital public health program falter. On behalf of 3 million March of Dimes volunteers and countless other organizations and families, we urge you and we urge the Senators to cosponsor, and to support the Newborn Screening Saves Lives Reauthorization Act. We quite respectfully request that you report this bill out of committee.

We look forward to working closely on this bill with you, with chamber leadership to ensure that it can be passed as soon as possible by both the Senate and the House.

We thank you so much for your considered attention to this vital health issue, and we stand ready to assist you in ensuring efforts for newborn screening programs to continue so that they may protect the health and well-being of newborns for many years to come.

Thank you very much.

[The prepared statement of Ms. Howse follows:]

PREPARED STATEMENT OF JENNIFER L. HOWSE, PH.D.

Good morning Chairwoman Hagan, Ranking Member Enzi, and members of the Subcommittee on Children and Families. My name is Dr. Jennifer Howse, and I'm proud to serve as president of the March of Dimes Foundation, a unique partnership of scientists, clinicians, parents, members of the business community and other volunteers affiliated with 52 chapters and over 200 divisions in every State, the District of Columbia and Puerto Rico. I appreciate this opportunity to testify today on newborn screening, one of the great public health victories of the 20th century, and one which continues to save infants' lives every day.

The March of Dimes is a national voluntary health agency founded in 1938 by President Franklin D. Roosevelt to support research and services related to polio. Today, the Foundation works to improve the health of women, infants and children by preventing birth defects, premature birth and infant mortality through research, community services, education and advocacy. In 1998, the March of Dimes established its Global Programs division to extend its mission overseas through partnerships with countries to deliver interventions directed at reducing birth defects and pre-term birth.

BACKGROUND

Newborn screening is a critically important and highly effective public health program for testing every newborn for certain genetic, metabolic, hormonal and functional conditions not otherwise apparent at birth. Approximately 1 in every 300 newborns has a condition that can be detected through screening. Newborn screening detects conditions that, if left untreated, can cause disabilities, developmental delays, illnesses or even death. If diagnosed early, many of these disorders can be managed successfully, which not only reduces the physical burden of disease but can also help to reduce the associated economic burden on families, communities, and government.

Since the mid-1960s, the success of newborn screening programs has led to routine testing for the over 4 million infants born in the United States each year. The Centers for Disease Control and Prevention (CDC) estimates that each year over 6,000 newborns are diagnosed as having a treatable metabolic condition and another 12,000 are found to have hearing impairment that requires followup. The majority of newborn screen tests are performed using a single sample of a few drops of blood from the newborn's heel, usually taken in the hospital 24 to 48 hours after birth. Hearing screening and screening for critical congenital heart disease (CCHD) are performed with non-invasive devices; hearing screening utilizes a handheld device held near the infant's ear, while pulse oximetry is used to test for CCHD by way of a small probe that clips onto a newborn's hand or foot.

HISTORY OF NEWBORN SCREENING

This year, our Nation is celebrating the 50th anniversary of newborn screening; however, the program's origins reach back much earlier. In 1959, after the March of Dimes had led our Nation to the successful development of the Salk and Sabin polio vaccines and refocused our mission on birth defects prevention, we initiated discussions about newborn screening on a large scale as a means to detect and prevent the catastrophic consequences of metabolic conditions such as phenylketonuria (PKU). This led to a grant to Dr. Robert Guthrie to support his development of a simple and effective population-based screening test for PKU. Dr. Guthrie's work demonstrated conclusively that identifying infants with PKU and immediately beginning a low-protein diet could completely avert the otherwise devastating developmental disabilities PKU causes. These results were so dramatic that the State of Massachusetts mandated PKU screening for all infants in 1968, beginning the modern era of newborn screening.

Subsequently, the March of Dimes funded research into tests for other genetic and metabolic diseases in newborns as we promoted newborn screening as a central component of newborn medical care. The Foundation is deeply proud of our decades-long history of funding research that has led or contributed to the development of numerous newborn screening tests, including those for congenital adrenal hyperplasia, biotinidase deficiency, and others. Together, these tests have allowed us to preserve the health and well-being of thousands of children.

As more tests became available, however, a patchwork developed in which some States screened for numerous disorders and others very few. In 2000, the March of Dimes led the way in proposing a national standard for newborn screening which included a core list of 9 disorders, with provisions for expanding the list as science and technology evolved. At the same time, the March of Dimes and others in the policy community began working with Congress to bring new attention and focus to this rapidly developing field. We worked to identify policy changes that would allow the Federal Government to assist States in evaluating new tests and determining whether to include them in their screening panels. The landmark Children's Health Act of 2000 (P.L. 106-310) included two vital provisions that advanced newborn screening policies. The law created the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children to provide expert evaluations of new tests and consideration of challenges in the field. It also established Federal grants to enhance and evaluate State newborn screening programs, allowing them to develop and implement best practices.

In August 2004, the American College of Medical Genetics (ACMG) submitted a report requested by the Health Resources and Services Administration (HRSA) setting out proposed nationwide standards for State newborn screening programs. The report listed 29 core treatable disorders that should be targeted directly and an additional 25 secondary conditions for which test results should be reported. These secondary disorders were not directly targeted by newborn screening because they did not yet have documented treatments or because there was limited knowledge of their natural history. Their presence would be revealed, however, in the course of screening for the core conditions. The ACMG recommendation to screen all newborns for 29 core conditions was endorsed by the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children as well as the March of Dimes in 2005.

The Federal Recommended Uniform Screening Panel (RUSP) gave advocates a powerful tool to press State legislatures to adopt this consistent set of tests. The March of Dimes led a grassroots advocacy campaign to secure adoption of the recommended uniform panel in every State, issuing annual report cards to document progress. And it was spectacularly effective: in 2004, only 21 States screened for at least nine of the recommended conditions, but just 4 years later all but two States were screening for at least 21.

Since 2010, the Advisory Committee, with the Secretary's approval, has added two new conditions to the Recommended Uniform Screening Panel: severe combined immunodeficiency (SCID) and critical congenital heart disease (CCHD). A third condition, Pompe Disease, is currently awaiting a decision by the Secretary. This year alone, the March of Dimes and allies like the American Heart Association have advocated successfully for 24 States to add CCHD to their newborn screening panels. This system of review and recommendations by the expert Advisory Committee, approval and dissemination by the HHS Secretary, and adoption by the States continues to work effectively to ensure that tests are evaluated appropriately and then adopted in a timely fashion to protect the health of our Nation's infants.

THE NEWBORN SCREENING SAVES LIVES ACT

The remarkable progress of newborn screening over the past two decades persuaded Congress to pass the Newborn Screening Saves Lives Act in 2008. The law renewed and updated various programs that underpin States' newborn screening efforts as well as the Secretary's Advisory Committee. Most notably, it codified the authority of the Secretary of Health and Human Services to establish the Recommended Uniform Screening Panel and to accept or reject the Advisory Committee's recommendations to add conditions to the RUSP. The law is now due for its regular 5-year renewal.

The March of Dimes is deeply grateful to Subcommittee Chairwoman Kay Hagan and Senator Orrin Hatch and Representatives Lucille Roybal-Allard and Mike Simpson for introducing S. 1417 and H.R. 1281, the Newborn Screening Saves Lives Reauthorization Act. Reauthorization is critical to ensuring we continue to provide the most accurate and comprehensive screening available to our Nation's children.

Passage of the Newborn Screening Saves Lives Reauthorization Act is essential to the continued success of newborn screening programs across our Nation. Most importantly, reauthorization will ensure the uninterrupted continuation of the Secretary's Advisory Committee on Heritable Disorders and its work. The Advisory Committee's charter expired in April of this year, and it was only through the timely action of Health and Human Services Secretary Kathleen Sebelius that it was extended on a discretionary basis for up to an additional 2 years. Maintaining and updating the Recommended Uniform Screening Panel that States use to adopt and implement new conditions is vital, and ongoing and planned evidence reviews should not be delayed.

The Newborn Screening Saves Lives Reauthorization Act also extends important grant programs at the Health Resources and Services Administration, Centers for Disease Control and Prevention and National Institutes of Health, including:

- Seven Genetics and Newborn Screening Regional Collaborative Groups (RCs) and a National Coordinating Center (NCC) funded by HRSA, which strengthen and support the genetics and newborn screening capacity of States using a regional approach to addressing mal-distribution of genetic services and resources. Special emphasis is given to underserved populations and those families and providers in rural areas. The RCs include all States, U.S. Territories and the District of Columbia.

- The Critical Congenital Heart Disease (CCHD) Newborn Screening Demonstration Program, a 3-year HRSA grant designed to support the development, dissemination and validation of screening protocols and newborn screening infrastructure for point of care screening specific to CCHD. CCHD presents special challenges to implementation since it is not tested with the blood spot.

- *Baby's First Test*, a national educational resource center for newborn screening presently operated by Genetic Alliance under a HRSA grant. *Baby's First Test* informs and empowers families and healthcare providers throughout the newborn screening experience.

- The Newborn Screening Technical Assistance and Evaluation Program (NewSTEPs) funded by HRSA, which serves as a technical assistance program for State newborn screening systems.

- Newborn Screening Quality Assurance Program (NSQAP), a comprehensive CDC program devoted to ensuring the accuracy of newborn screening. NSQAP is the only comprehensive program in the world devoted to ensuring the accuracy of newborn tests. In 2012, the program guaranteed the quality of newborn testing in more than 550 laboratories worldwide, and assured identification of between 5,000 and 6,000 infants with treatable diseases who might have otherwise died or become severely disabled.

- The Hunter Kelly Research Program, which supports numerous grants and contracts to develop and improve technologies related to newborn screening. Through the Hunter Kelly Newborn Screening Research Program, the Eunice Kennedy Shriver National Institute of Child Health and Human Development also funds the Newborn Screening Translational Research Network, a resource for investigators engaged in newborn screening-related research.

CONCLUSION

Today, 42 States and the District of Columbia require screening of at least 29 of the 31 treatable core conditions. Millions of babies have been screened for dozens of disorders, and in thousands of cases, the health and well-being of those children has been preserved. Newborn screening represents a model Federal-State public health partnership that has produced extraordinary improvements in child health.

We must not allow this vital public health effort to falter. Our most immediate challenge is to preserve and renew the Newborn Screening Saves Lives Act. On behalf of over 3 million March of Dimes volunteers and countless other organizations and families, I urge Senators to cosponsor and support S. 1417 and the committee to report the legislation. We look forward to working closely with the committee and chamber leadership to ensure it can be passed as soon as possible in both the Senate and the House. Furthermore, although beyond the jurisdiction of this committee, I urge Congress and the Administration to agree on a balanced approach to deficit reduction that protects investments in programs such as newborn screening. Authorization bills are only effective insofar as funding is appropriated to implement their provisions.

Newborn screening has improved and saved the lives of countless thousands of affected children. Thank you for your attention to this vitally important child health issue. The March of Dimes stands ready to assist you in ensuring that newborn screening programs will continue to preserve the health and well-being of newborns for many years to come.

Senator HAGAN. Thank you, Dr. Howse, and thank you to the March of Dimes, to your staff and to your many, many volunteers all across the country for the work that they do.

And now, we have our last witness, Mrs. Joye Mullis, of Raleigh, NC. I certainly want to welcome her husband Jeremy and her son Ethan to the hearing today. Mrs. Mullis has some personal experiences with newborn screening that, I hope, will remind all of us here today why this is so important.

Mrs. Mullis.

STATEMENT OF JOYE MULLIS, RALEIGH, NC

Mrs. MULLIS. Good morning, Madam Chairman Hagan, Ranking Member Enzi.

Thank you for the opportunity to share our story with you today.

As with all children, my son's story began well before his birth. I believe that every heart has a story and this is his.

My husband and I learned that we were expecting our first child on a hot July morning in 2008. Our joy and celebration were tempered quickly by some early complications. However, we were overjoyed to see our baby's strong heartbeat on the screen in front of us during our first ultrasound.

From that point forward, my husband and I weathered the ups and downs of a complex pregnancy with optimism and hope. In all, I had five ultrasounds, and ultimately we learned that our baby would be born with two noncritical birth defects that would require surgical intervention. Our physician assured us, however, that his heart was strong and his prognosis was good.

On March 8, 2009 our precious baby boy Ethan was born. My husband and I spent the first 8 hours of his life with him, surrounded by friends and family. We prayed that he would be OK, but we had no idea that in just a short time, new challenges would be coming our way.

As the postpartum nurse was bringing Ethan back to our room after his newborn screening, she noticed that, in her words, "He just did not look right." She immediately wheeled him back into the nursery and hooked him up to a pulse oximeter. Through that noninvasive screening, she discovered that Ethan's oxygen saturation level, which should have been at least 95 percent, was in the mid-60's. I will never forget hearing the words, "We have reason to believe there is something wrong with your baby's heart."

Ethan was diagnosed with pulmonary atresia with a ventricular septal defect. While a pediatric cardiologist explained in great detail what that meant, the bottom line was that our son would need to be rushed to Duke University Medical Center in Durham for open heart surgery. It was not until he was 9 weeks old that Jeremy and I walked out of Duke as a family of three and into our home with our son.

Four years later, Ethan's health is now stable. He has endured 14 surgeries and procedures, and he has survived full cardiac and pulmonary arrest. Despite his rocky start, we now have a boy on our hands who loves bugs, and cars, and playing with his preschool friends. We know that there will be more surgeries ahead, but we cherish the time that we have together with him now.

Every heart has a story. The story of Ethan's is one of strength and resilience. While pulse oximetry screening cannot take away the heartache of surgeries and complications, it can be the start of a lifetime of success for a baby born with a congenital heart defect.

It has taken a lot of work to get Ethan to where he is today, and it all began with an observant nurse. However, babies should not have to rely on a doctor's or a nurse's intuition to diagnose potentially fatal conditions.

Ethan's story exemplifies the importance of comprehensive newborn screening, and the Newborn Screening Saves Lives Reauthorization Act will help ensure that infants throughout our Nation are screened for treatable conditions like Ethan's at birth.

I urge all members of the committee to support this legislation, and I sincerely hope it will be passed by the full Senate this fall.

Thank you again for listening to our story today, and may God bless you.

[The prepared statement of Mrs. Mullis follows:]

PREPARED STATEMENT OF JOYE MULLIS

Good morning Madam Chairman Hagan, Ranking Member Enzi and distinguished members of the subcommittee.

Thank you for the opportunity to share my story with you today.

As with all children, my son's story began well before his birth. I believe that every heart has a story and this is his.

My husband and I learned that we were expecting our first child on a hot July morning in 2008.

Our joy and celebration was tempered quickly by some early complications; however, we were overjoyed to see our baby's strong heartbeat on the screen in front of us during our first ultrasound.

From that point forward, my husband and I weathered the ups and downs of a complex pregnancy with optimism and hope.

In all, I had five ultrasounds and ultimately we learned that our baby would be born with two non-critical birth defects that would require surgical intervention. Our physician assured us, however, that our baby's heart was strong and the prognosis was good.

On March 8, 2009, our precious baby boy Ethan was born. My husband and I spent the first 8 blissful hours of his life with him, surrounded by family and friends. We prayed that he would be OK, but we had no idea that in just a short time, new challenges would be coming our way.

As the post-partum nurse was bringing Ethan back to our room after his newborn screening, she noticed that "he just didn't look right" and immediately wheeled him back into the nursery and hooked him up to a pulse oximeter.

Through that non-invasive screening, she discovered that Ethan's oxygen saturation level, which should have been at least 95 percent, was in the mid-60s percentile.

I will never forget hearing the words, "We have reason to believe there is something wrong with your baby's heart." Questions raced through my mind and fear coursed through my veins.

I asked myself, "How could there be something wrong with his heart? How did this go unnoticed before now?" I was angry and very, very scared.

Ethan was diagnosed with pulmonary atresia with a ventricular septal defect. While a pediatric cardiologist explained in great detail what that meant, the bottom line was that my hours old son would need to be rushed to Duke University Medical Center for his first open-heart surgery.

It wasn't until Ethan was 9½ weeks old that my husband and I walked out of Duke as a family of three and into our home with our son.

Four years later, Ethan's health is now stable. He has endured 14 surgeries and procedures, and an incident in which he went into full cardiac and pulmonary arrest taking a team of about 30 doctors and nurses, and 11 minutes of CPR to bring him back to us.

Despite his rocky start, we now have a boy on our hands who loves bugs, cars, and playing with his preschool friends. There may be more surgeries ahead, and we cherish the time we have together now.

Every heart has a story. The story of Ethan's is one of strength and resilience. While pulse oximetry screening can't take away the heartache of surgeries and complications, it can be the start of a lifetime of success for a baby born with a congenital heart defect.

It has taken a lot of work to get Ethan to where he is today, and it all began with an observant nurse.

However, babies should not have to rely on a doctor's or nurse's intuition to diagnose potentially fatal conditions.

Ethan's story exemplifies the importance of comprehensive newborn screening, and *The Newborn Screening Saves Lives Reauthorization Act* will help ensure that infants throughout our Nation are screened for treatable conditions—like Ethan's—at birth.

I urge all members of the committee to support this legislation and I sincerely hope it will be passed by the full Senate this fall.

Thank you for listening to my story today, and may God bless you.

Senator HAGAN. Thank you, Mrs. Mullis, and thank you Jeremy and Ethan for being here today, and thanks for your personal experience of how important newborn screenings really are to each and every family who has a child.

The hearing record will remain open for 10 business days for the Senators to submit questions.

So we are going to go ahead and start with the questions now, and let us just do a round of 5 minutes, and then we can continue.

Mrs. Mullis, I wanted to ask you a question and just thank you so much for sharing your story and Ethan's story. We are glad to hear that he is now a happy 4-year-old, experiencing all the things, as you said, from bugs, and cars, and trains, and planes.

You noted earlier in your statement that your physician detected noncritical birth defects during your pregnancy. I am curious if, at that point or later during your pregnancy, if your physician or any other healthcare provider, shared information with you about newborn screening? And if not, is that information that now, looking back, you think you and your family would have found helpful and/or comforting?

Mrs. MULLIS. Sure. The physician took us through all the details of those two birth defects. He assured us that Ethan would be screened at birth. At that time, pulse oximetry was not mandated by North Carolina, so that was hospital-specific. And so, he did not share that specific test with us, but did inform us that he would be well checked and looked over after his birth.

Senator HAGAN. I know how important it would be, I would think, for pregnant moms to understand newborn screening. I think one of the questions we have is: how much of that is actually being done in the offices?

One other question, since Ethan was born, as you said, North Carolina has required the pulse oximetry testing for all newborns in the State to detect these critical congenital heart defects like the one that Ethan had. I understand that you have been active in the community, and that you have been meeting with other parents whose children might have a heart defect or other condition detected from the newborn screening.

Can you share with us what mandating the CCHD screening, and/or not letting it remain optional, has meant for other parents in the State that you have spoken with?

Mrs. MULLIS. Absolutely, and your question is very timely.

I just learned of a family in North Carolina—two families actually—who, since the screening of their babies, have been screened

at their community hospital. Congenital heart defect was detected right away, and the baby was able to get to a larger hospital to receive the care that he needed.

So it is already making a difference in North Carolina, and I am very honored to have played a small role in that.

Senator HAGAN. Thank you.

Miss Bonhomme, as director of Baby's First Test, you are regularly engaged in trying to educate parents who may not know much about newborn screening.

What are the most common misperceptions that you encounter from parents about the newborn screening system? And are there education efforts targeted for specific communities that we know have a higher incidence of conditions that would be detected by newborn screening? For example, we know that sickle cell anemia is obviously prevalent among African-Americans, and that certain heart defects are more common in certain areas of Alaska.

Can you just go over some of the kinds of questions and educational issues that you encounter?

Ms. BONHOMME. Absolutely. One of the main questions that we receive about newborn screening is, first, why they did not hear about it earlier. Parents always wonder,

"Oh, why did I not hear about this when I was planning my pregnancy, or when we were talking about all the different screenings that were going to take place?"

You hear about breast feeding. You hear about all these other activities that take place in that first few days of life, but oftentimes, parents do not hear about newborn screening.

In terms of programs targeted toward specific communities, there are a number of different activities, both at the State and national level to target those different communities. One activity that actually Baby's First Test is helping to fund is a PSA in the Atlanta region targeted toward African-American families about newborn screening and to make sure they know what their status is, particularly their sickle cell status, but also wrapping newborn screening into that.

I know that in Alaska, there have been efforts to create some DVD's so that the local birthing centers there can show information about newborn screening, and the particular conditions that affect that particular population.

Senator HAGAN. Thank you.

Senator Enzi.

Senator ENZI. Thank you, Madam Chairman.

And for the Mullis family, I cannot even imagine what you have been through. My wife and I had a daughter that was born 3 months premature, and so we went through some of those daily ritual things.

But something that is much more common is the doctor talking to you about something as simple as a tonsillectomy and explaining that there is no problem in 99.9 percent of the cases. And you think, "But what if I am the 1/10th? It is my kid's life." So thank you for sharing your story. It makes a tremendous impact.

I would like to ask Dr. Howell about this legislation that has been introduced by the Chairman and Senator Hatch. It has new timelines for that Advisory Committee to be able to review the evi-

dence for the new conditions under a priority review status, and has shorter deadlines for the Secretary to adopt or reject the recommendation of the Advisory Committee.

Can you explain why these new timelines are needed and would be helpful, and whether you think they improve the process of evaluating the conditions nominated for screening?

Dr. HOWELL. Thank you, Senator Enzi.

I think that the timelines are introduced, basically, to try to ensure that the process goes forward briskly. Particularly, for instance, when a drug has been approved for a condition such as a newborn screening condition, we think that the FDA has reviewed that and has proven that it is a valuable drug. And so, we think that the evaluation of newborn screening should proceed quickly and perhaps a little more rapidly than we would like.

I think in newborn screening, it is very interesting, we are always working on a short timeline because the conditions that we are looking at and screening for commonly are fatal conditions. So any delay that we see that we have, we are losing lives, and so, we really would encourage that.

I think that the timeline, I have looked at them pretty carefully. They are aggressive; I agree with that, but I think on the other hand, they clearly can be accomplished.

I think the one thing that will be important as the community works on this legislation is to ensure that the committee has adequate resources to do the evidence reviews; the evidence reviews are expensive. And so, if we have a couple of conditions that need rapid review, we need to be sure that the funding is there to have the evidence reviewed. But I think that in the bureaucratic world we live in, we want these conditions to move rapidly, and I think the advantage of having timelines is great.

The Secretary is always very busy, as you obviously know, and there is a timeline. During the latter portion of my time as chair of the committee, having the Secretary have timelines to respond to the committee was effective; we got letters back more quickly.

So I think they are realistic. I think that they will require a little effort, but I think that is fine. I think it is a good idea.

Senator ENZI. Thank you.

Dr. HOWELL. And let me add one other thing.

Senator ENZI. Sure.

Dr. HOWELL. I think the fact that you do have these timelines, I believe it will encourage the person making nominations to be a little more complete and a little clearer so they know that they can go quickly rather than having a lot of loose ends out there. It is a good thing to do.

Senator ENZI. Thank you. Does anyone else want to comment on that?

So we will move to the science of genetics, which is one of the more rapidly changing fields of research in the United States. Scientists and researchers are learning more and more about the code that makes each of us who we are. To that end, the field of genetic medicine is also continually evolving and innovating, and the Newborn Screening Saves Lives Reauthorization Act does include a new priority review pathway that we have just talked about for the Ad-

visory Committee to review conditions for screening where there are pending applications for new drugs or breakthrough therapies.

You talked about it a little bit already, Dr. Howell, but can you discuss the significance of the new pathway in terms of medical research into treatments for these conditions? What are some of the new things that are coming down the road that we may not know about and how that will affect the development of the new therapies for these conditions?

Dr. HOWELL. Let me elaborate just briefly on the two conditions that I quickly passed over and so forth.

Duchenne muscular dystrophy is a condition that many people are aware of. It affects boys. It is a devastating disease. We have known about it for a century, and we have actually known the genetic defect for 20 years; it is a genetic absence of a protein. But it has been really tough to figure out how to get that protein back, and there are all sorts of studies going on.

But there is one study going on using antisense oligonucleotide—a very unfortunate long term—but basically that is a compound that attaches to the DNA, and where you have a deletion or an absence of a portion of the DNA, you have these special compounds that basically jump over these deletions. It is like a bridge; so you have a part of the gene here, you have a deletion/a hole, and then you have the rest of the gene. And so these drugs that are antisense oligonucleotides have some very encouraging results.

We really would like to be certain of that as we move ahead. There is a lot of conversation between the people doing newborn screens to people working on drugs, and that is encouraging this bill. The FDA would be working with the committee so the drugs are coming.

If a drug indeed does show great results, and is lifesaving, it would be very important for that newborn screening program to be rapidly moving ahead. In other words, you have a new drug. It is lifesaving. You do not want to sit and think about it for a long time. You want to move quickly. And so, I am a big advocate of that, and I think it makes sense.

And the number, there are similar drugs for spinal muscular atrophy that are very exciting. Again, spinal muscular atrophy is the most common fatal neurologic disease of childhood; a devastating disease. Again, encouraging and exciting results are out there. And we want to be ready to take care of them once these drugs hit the market.

Senator ENZI. Thank you for your ability to explain something very difficult very well.

Dr. HOWELL. I expect you will be explaining antisense oligonucleotides now to your colleagues.

[Laughter.]

Senator ENZI. And talking about SCID's. Thank you.

Senator HAGAN. I am glad that Senator Enzi is going to be doing that explanation.

[Laughter.]

Dr. HOWELL. Well, you are going to be busy talking about SCID, right?

Senator HAGAN. Senator Casey.

STATEMENT OF SENATOR CASEY

Senator CASEY. Thank you, Madam Chair.

I want to thank you for calling this hearing and grateful that you and Senator Enzi are here with us today to talk about what is such an important issue, but sometimes an issue we do not spend enough time on.

I will have a longer statement for the record, and I will pose a few questions here.

Dr. Howell, some of your testimony reminds me of a story, but I want to first thank all of our witnesses, especially Mrs. Mullis, for taking the time to be here, and to being not just an advocate, but a personal witness to how important newborn screening is, and how important it is to reauthorize the legislation.

Dr. Howell, I am sorry I missed your testimony, but we are lucky to have a copy of all of your testimonies. Dr. Howell, when you said in the second paragraph of your testimony about PKU and how a lot of these screening efforts started as long as 50 years ago, you said that,

“Children with an inherited condition known as PKU, if untreated, have profound developmental delay with an average IQ of less than 20. This means that such untreated children, who have a normal life-span, are unable to speak or care for even simple needs and require full-time care. They are robbed of many of life’s opportunities.”

That was your testimony, and it just so happens that I have a little bit of a personal connection here. My father was a State senator in the early 1960s in Pennsylvania—I know that now U.S. Senator Hagan was a State senator at one point in her career—but he saw this information that was available at that time. This would have been, for him, 1963 or 1964. He had a law passed in the senate of Pennsylvania which at least led to a new policy as it relates to children in Pennsylvania. Just a very inexpensive screening at the time would save a lot of lives.

I will put in the record his recollection of that. I will not go into that today, but he passed away more than 13 years ago. So I want to, as we are paying tribute to our panel and all those who are great advocates, I want to pay tribute to him and put a reminder in the record, which I will include in my statement for the record.

But I wanted to ask about one of the challenges we still have with all of the Internet access, all of the technology that is available to folks. We know in just the last couple of days, we have reports about folks who do not have regular access to the Internet.

Miss Bonhomme, I wanted to ask you about if we are focused on Baby’s First Test and the work that you are doing that relies on Internet access to disseminate information, what steps have been taken to ensure that parents who lack reliable Internet access, that they can also obtain this information?

Ms. BONHOMME. I thank you for that question, Senator.

While Baby’s First Test has a very public, online presence, we also do a number of locally based, community-based activities. So we have a series of challenge awards that really are targeted toward where there are educational gaps, and not having Internet ac-

cess or reliable Internet access is a great gap that a number of our citizens are faced with. So we do invest in that fashion.

So we will partner with community-based organizations to make sure that they can disseminate information and we have had programs who have gone and worked with their local public library system to make sure that there is information there. But really, we partner with communities so that they can say, "We know that there are a lot of moms and families that go to this particular part of town," for whatever reason, oftentimes it is a library or a church. "Let us make sure we have materials there." So we really do try to address that issue of not having a lot of Internet access.

We also work with State public health departments who oftentimes have access to being able to provide materials in, let us say, in the bag that goes home with the mom at the hospital. We partner with them to create materials that will go in there, so that there is something—either a little postcard or a handout—that they can refer to when they go home after the birth of their child.

Senator CASEY. I appreciate that. I know my time is running out, but let me take one more for Dr. Howell while we have a couple of minutes.

I wanted to ask you about the fact that we have had States taking action over time; I mentioned what happened 50 years ago in Pennsylvania. But when States are making decisions about these issues about screening, what factors that you can identify, what factors might lead a State not to screen for a particular illness that the Advisory Committee recommends screening for?

Can you shed some light on the determinations they make or what goes into that?

Dr. HOWELL. Every State has an advisory committee that advises the State health department on newborn screening and they basically will look at a recommendation that comes from the committee.

One of the things that has been extremely gratifying to me is that since the committee has been established, it has very broad representation and a tremendous number of experts, public members, scientists, ethicists, et cetera. When that committee does a detailed evidence review, one of the things that has been exciting is the States have looked at that and have overwhelmingly adopted it, which has been very gratifying.

I might point out that adoption has not been out of the blue. It has been helped tremendously by advocates, the March of Dimes being a leading advocate in the State. But fundamentally, the advocates in the States now have an absolute criteria to go on.

Virtually all the recommendations have been adopted, but occasionally, some recommendation will come down and the State will decide, "Well, this would be very difficult for us to do," et cetera. But again, I think that even when they initially decide not to do it, they do move along and do it.

I would like to comment briefly also that when the committee has recommended the implementation of what I would call a rather complicated new technology such as Severe Combined Immuno-deficiency, or SCID, where you are looking at a test that is new, the committee had recommended and the NIH had funded, a national pilot program.

So you tried it in three or four States, et cetera, and did a lot of babies to see how it worked: what are the problems out in the field? That has also been invaluable. If you look today at what is happening, still States are implementing Severe Combined Immunodeficiency and States are starting to implement critical congenital heart disease.

In the State of Florida, for example, Florida is in the process of implementing the cardiac screening. But again, to get that going, they need to meet with the various people around the State and decide exactly what is going to happen when a baby in Pensacola needs to be followed up. And so, you have organizational changes and so forth.

But there will always be a committee that will say, "No, we are not going to do this," and so forth, but that is rare and it has been the exception.

When the committee was first established, and I had the privilege of working with it, a number of colleagues, thoughtful colleagues said,

"I do not know why you are going to waste your time on that because you are going to sit in Washington, you are going to make all these wonderful recommendations with all these very bright people, and the people are not going to pay any attention to it."

That did not happen largely because of the advocates that basically once the data were available, they made it happen.

Senator CASEY. I want to thank the panel and the advocates who are here with us today, and others like you, for bringing light to a terrible darkness, and we are grateful for the time.

And Madam Chair, thanks for another 3 minutes and 30 seconds. [The prepared statement of Senator Casey follows:]

PREPARED STATEMENT OF SENATOR CASEY

Thank you Senator Hagan for holding this hearing today.

Providing the foundation for our children to lead healthy and productive lives is the most important thing we can do for our children. More than 1 in 300 newborns have a condition that is treatable through newborn screening. This means that the newborn screening system plays a vital role in ensuring that a multitude of diseases are caught early, when they are easier to treat. My office has heard from constituents who have benefited, or whose children have benefited, from newborn screening. Children's lives have been saved and greatly improved because medical conditions were promptly identified.

The newborn screening system in our country has developed over the past 50 years thanks to the vision and hard work of countless researchers, medical professionals, and patient advocates. The Newborn Screening Saves Lives Act in 2008 was a crucial step forward for that system. I am proud to have cosponsored that legislation, especially in light of how standardized newborn screening has become since its passage. A decade ago, the newborn screening system differed significantly from State to State. Today, as noted by one of this panel's witnesses, 100 percent of U.S. births were screened for over 30 conditions at the end of 2010. The additional

resources and access to information provided by the Newborn Screening Act helped make this progress possible.

Although individual States must maintain flexibility to address their own needs, we must also work to ensure that children born in different States do not have substantially unequal access to medical screening. The death of any child is a tragedy, but there is no death more heartbreaking than one that could have been prevented by a simple screening test followed by prompt treatment.

The Advisory Committee on Heritable Disorders in Newborns and Children provides all States with accurate, scientifically based recommendations for their newborn screening programs. The Newborn Screening Act took the important step of codifying this committee, and the reauthorization bill that Senator Hagan has introduced will help to improve and streamline the process under which it considers conditions for the Recommended Uniform Screening Panel.

I happen to have a personal connection to this subject. My father was a Pennsylvania State Senator in the early 1960s when he heard that a test was available to diagnose children with phenylketonuria, or PKU. I would like to enter into the record an excerpt from his memoir, detailing his successful efforts to mandate a State test for PKU:

My first cause as a State senator was helping children. Shortly after taking office, I heard about a problem known at the time as PKU. It was an acronym for the long, clinical name of a birth defect which prevented an infant from metabolizing certain foods, including milk. Undetected, the ingestion of such food would cause normal babies to become [intellectually disabled]. For some reason the State of Pennsylvania had no law on its books requiring the simple test needed to detect the presence of PKU. Once detected, a simple change in diet could correct the problem. Babies throughout Pennsylvania who might have been spared were instead being born with that defect going undetected.

The test cost practically nothing, I learned. So why weren't we requiring it by law? It did not strike me as a complex problem. So we passed a simple law—just a few lines on paper—requiring that Pennsylvania's babies be given the PKU test. Thirty years later I still count it as among the best things I ever did. A simple change in diet can protect a child from a lifetime of [intellectual disability], thanks to the passage of that simple law. I remember reading a small article a few months afterward about the first baby who had been found in Pennsylvania with PKU since the new law had taken effect. A healthy, beautiful baby, saved from [intellectual disability].

We have made too much progress over the past 50 years to stop now. We must continue to invest in our newborn screening system, and we must continue to support groundbreaking new research at institutions such as the NIH. I look forward to continuing to work with Senator Hagan and the other cosponsors of the Newborn Screening Saves Lives Act to ensure that we are building on the success of this program.

Senator HAGAN. Any time. Thank you. Thank you, Senator Casey.

This is a question for Dr. Howse and Miss Bonhomme about babies born outside of the hospital setting. While most babies are born in hospitals today, some parents do choose to have their babies born at home or in other settings. In fact, since 2004, I understand that the rate of births occurring at home has risen nearly 30 percent in the United States.

These newborns and their families should not miss out on the lifesaving opportunities presented by screening just because the provider setting in which they are born is not in the hospital.

Dr. Howse, can you tell us how the newborn screening system covers babies that are not born in a hospital and does that occur?

Ms. HOWSE. Well, I am going to defer to my Baby's First Test colleague for the particulars of outreach.

The quick answer to your question is it is really very unfortunate for any of the babies who are born at home not to have a link into the hospital so that those tests can be taken care of. I think in many cases, there is active partnership between the home birth attendant or the midwife. There is active partnership with an obstetrician and with a nearby hospital so the baby can be brought in and be tested. You know, it is quite important that that happen in the first 48 hours. The first 24 hours would be even better. So I think the problem is recognized and there are bridges for outreach and connection of the baby.

But clearly you put your finger on a potential problem and that is the babies that are born at home that do not have that kind of linkage and they go without testing. And it is something that we need to continue to be very vigilant about.

Thank you.

Senator HAGAN. Miss Bonhomme.

Ms. BONHOMME. Great. Thank you for that question.

We have done a lot of work to really try to understand, yes, most babies are born in the hospital setting, but what about all the other babies that are born? And we have done a lot of work with nurse midwives to understand both how do they do newborn screening and it really does depend on the State.

In some States, the midwife can actually do the filter paper blood collection, and they will do it, and they are kind of that link. In other States, the family will go and see the pediatrician at 48 hours at their very first pediatric visit. But it does vary by State.

One thing that we have done is really to understand what are the perspectives of nurse midwives in terms of newborn screening because that is going to tell us what they are saying to the families whose babies they are delivering.

We have actually done focus groups with birthing centers here in Washington, DC to get a better sense of what their educational needs are. Generally, it is just that they know that newborn screening is supposed to happen. They know they are supposed to collect this blood on this filter paper, but they are not exactly quite sure why, or where the information goes, and all of that.

It is one reason that Baby's First Test—not only do we educate parents, but also all those health professionals, including nurse

midwives because they are a very strong link between families and this public health program.

Senator HAGAN. Dr. Howell, if these children are not born in a hospital setting, and if a midwife does the test, what is the best time to actually do that test? Can it be done right after birth?

Dr. HOWELL. It should be done and we recommend between 24 and 48 hours.

Senator HAGAN. But it could be done at like 2 minutes.

Dr. HOWELL. It should not be done at 2 minutes.

Senator HAGAN. That is what I thought.

Dr. HOWELL. In other words, most States quite properly, if a test is done within the first 24 hours will require a repeat because there are so many changes happening that soon after birth, so that that would be early.

And I think as Jennifer and Natasha have pointed out, many of the midwives have connections so that the baby might go to a site within 24 to 48 hour. They should not be delayed because certain of the conditions that we screen for, notably galactosemia, you need a very rapid diagnosis and to delay for a week is much too late. You like to have the data back to the baby in under a week.

Senator HAGAN. Miss Bonhomme, in your example, the midwife would have to either come back or the child would have to be taken.

Ms. BONHOMME. Right. And oftentimes, the midwife is already planning to come back to visit the family. They will do the delivery, and leave making sure everything is OK. But then the next day, they will come back, particularly in rural areas to make sure that everything is still going along as planned.

Because of that, it is pretty easy to be able to fit it into the already scheduled appointments, if you will, with the midwife.

Senator HAGAN. Thank you. Thank you.

The Newborn Screening Saves Lives Act put into law significant support for the Nation's newborn screening system including grants to expand State programs, technical assistance, and quality assurance for State labs, and then researched into additional conditions that may be screened by the States.

Authorization for these programs expires at the end of this fiscal year, which is next Tuesday. Senator Hatch and I introduced a bill in July to reauthorize and make important improvement to this law, which is what we are talking about.

Aside from the changes included in our bill, Dr. Howell, Dr. Howse, Miss Bonhomme, can any of you describe generally just a statement or so about why this reauthorization of this law is so important?

Dr. Howse.

Ms. HOWSE. Yes, first of all, I thank you very much for that excellent question.

I really want to underscore the importance of the deadlines that are in the law, the timelines for the action by the Secretary's committee because you heard Dr. Howell speak very eloquently about the successful candidates out of the discovery pipeline. Those need to be linked very quickly with the process that is put together by the Secretary's committee so those recommendations can be quickly evaluated and responded to by the Secretary, and States can get

that information quickly. So I think that is a very, very important part of the bill.

Then simply to continue the programs that are established. They are good programs. They are well-formed; they are well-accepted by States. They allow for the continual improvement of newborn screening. The various parties accept the way the program works as a Federal-State partnership. So I think we should take the program that is well working and continue it in an uninterrupted fashion.

We are very concerned about the potential for interruption in a program that has a lot of moving pieces, but the pieces are a well-connected through the legislation. This is a proven program. So we are very, very concerned, really. We so appreciate your effort, and we are just very concerned that the program continue in an uninterrupted fashion.

Senator HAGAN. Dr. Howell.

Dr. HOWELL. I think several things that we might comment about is that the States are always very, very squeezed for funds. I think that the funding for the States to add and innovate is really very important, and without the legislation that is not going to happen. The State labs tend to not have new money, and so they are conditioned to be added, and so that their resources are tremendously stretched. So this is really very, very important.

Other things that are in the legislation, that are really critical to making the whole system work, is that the new legislation requires that the committee meet at least four times a year and at least two of those meetings must be in-person. I think the meetings of this committee are very essential to be in-person because when they are in-person, it provides an opportunity for a variety of people, advocates and other people, to come and talk to the committee, and that makes a huge difference. So I think that the requirement of meeting in-person and having four meetings a year to move things along is really essential.

Again, the research efforts are just so important. There are so many. For example, some of the conditions that we can screen for, we do not really have very effective treatments for, and we will not get those treatments without NIH funding. It has just got to be there. Again, HRSA is responsible for supporting this committee, and they need to have the money to do evidence reviews. So I think that the whole spectrum of things that are included in this bill are just absolutely essential.

The United States is unquestionably the world leader in newborn screening and that is a great thing to brag about, but it also means that we are saving lives in the United States. And without this legislation, we cannot continue to be that.

I think that those would be a quick summary of why we should really urgently pass this legislation.

Senator HAGAN. Thank you.

Miss Bonhomme.

Ms. BONHOMME. The issue with going last, everyone has hit all the key points very nicely, but what I will say is that while newborn screening is a State program, the funding at the State level is still limited, and this reauthorization would really allow the national dialog around newborn screening to continue.

Oftentimes States do not really have enough money to even do all the educational efforts that they would want to that today we have talked about are so important. Being able to have the reauthorization will allow for the programs, such as Baby's First Tests, but also the evaluation programs that fall underneath the law to continue to go further. That is really key because if there is a disruption in that, we really will be lost. We will lose data and potentially lose lives around that. So this reauthorization is really key.

Senator HAGAN. Mrs. Mullis, you and your family have personally experienced the fact that newborn screening does save lives. Do you have any comments you want to share on this reauthorization?

Mrs. MULLIS. I would just like to reiterate what the rest of the panel has said. I feel that it is very urgent so that the conversation can continue.

I cannot speak enough to how important this is to our family. We were very fortunate that we were in a place, in a hospital that had the capability to do this. We are between Duke and UNC. We have a lot of very good medical professionals in our area. So the newborn screening was talked about and shared with us when we first found out about Ethan's original birth defects, not including his heart.

I just feel very strongly that other families and other babies should be afforded that same opportunity.

Senator HAGAN. Thank you. Thank you.

Let me talk about the financing of newborn screening. I understand that screening programs, and obviously we have heard in the testimony it varies from State to State, but I think one of the questions that parents may have about newborn screening is whether they will be able to pay for more tests or whether the baby will not be tested if they do not have health insurance.

Can the panel, can you explain how most State newborn screening programs are financed and whether the insurance status of the parents has any effect on whether the newborns are screened?

I know that in North Carolina, the screening fee is \$19. In Wyoming, the fee is \$70 and in Utah, the fee is \$103. The amount of the fees varies widely from State to State. Are the newborn screening fees set by each State and are they typically covered by insurance?

Miss Bonhomme.

Ms. BONHOMME. Sure, I will start with that.

Senator HAGAN. Obviously, when you look at these fees compared to any sort of treatment, we know that the screening must take place.

Ms. BONHOMME. Correct. Each State determines its own newborn screening fees. I think a lot of that plays into the lab and what tests are included.

Now, one thing that we make very clear to parents is even if they do not have insurance, they will be able to get newborn screening. That is something that when we were building Baby's First Test, and asking about information about fees and putting that on the Web site, every State that I spoke to said,

"Please make it clear that no matter what the dollar amount we list, we will find a way to cover the newborn screening for each baby."

So typically, there are the fees that we mentioned, but also some States have a fund that comes from different taxes that help support the newborn screening program.

Dr. HOWELL. It is a potpourri of funding and it is very interesting. Some States do not charge anything. For instance, New York State pays for its entire newborn screening laboratory out of general funds and a variety of things. But it is the one program that I can point to that is universal. It is a public health program and every baby is screened regardless of the ability to pay.

The way it works in most States that charge a fee is that, let us say, you are born at a given hospital, the hospital receives a bill, and they then decide how they get the money. The patient ordinarily does not, out of his or her own pocket, pay any of those fees.

Newborn screening is arguably the best bargain in the United States. It is estimated that it costs a little over \$100 to do the initial blood spot and the initial screen, which means that the total program in the United States costs a little over \$400 million a year.

Now you say, "That is a lot of money," and it is a lot of money, but it is less than what we spend in 1 week on drugs for hypercholesterolemia to put things in perspective, so it is an enormous bargain. And again, all babies will get screened regardless of their ability to pay, and so that is something. It probably is the only healthcare item I can think of that you can say that about. There may be others, but it is certainly fairly unique.

Senator HAGAN. Dr. Howse.

Ms. HOWSE. Just connecting that to a question that Senator Casey had asked about why States might not have the full panel in place or why there might be delays when a new condition is recommended. Well, part of that does tie to the question of funding because it costs additional resource to implement a new test. To add that to the laboratory's responsibility, to make sure that the followup and specialty services are in place, et cetera.

But the bottom line is that the burden does not come to the families. States have a number of methods about how they pay for newborn screening. Many of them now have special funds that are a combination of fees and money that is appropriated by the legislature. I would really commend the States, Senator, for the manner in which they have determined how they are going to put together the funding packages to make this program work.

I think that despite enormous pressures that have faced the States, State legislatures, and Governors, they have gone to great lengths to make sure that this program is in place, that it expands properly when new screening conditions are recommended, and that the burden does not fall to families.

Thank you.

Senator HAGAN. Miss Bonhomme, in your testimony, you stated that parents expressed a desire to learn more about newborn screening earlier in their pregnancy, but that most parents actually do not remember getting that information.

What are the basic things that you would advise parents that are expecting to do during the prenatal period to actually learn about or to prepare for the newborn screening?

Ms. BONHOMME. What we have heard from parents is that they do not want to hear about newborn screening after the fact. They want to feel that they are a part of the health decisions around their baby even before the baby is here.

So we really do encourage parents to ask their doctors, either ask their prenatal physician or a prenatal nurse. Also a number of people speak to the pediatrician before the baby is born, so really bringing it up in that initial meeting with the pediatrician to see, "When will I hear about the newborn screening results?" those types of key questions so that they can start the dialog early.

A lot of times we are all busy, including physicians. But it really is, if parents know the right questions to ask, they can put it on the radar of their health professional and start the dialog early.

Senator HAGAN. We talked a little bit about some of the new technological developments. Originally, it was the development of the dried blood spot test that allowed us to engage in the newborn screening in the first place which, to me, is just an outstanding scientific feat.

But then the development of the tandem mass spectrometry has allowed us to significantly expand the number of conditions that can be screened. And then the use of the DNA extraction and molecular testing has greatly improved the accuracy of newborn screening, as well as made the screening of these new conditions possible.

You have covered some of this, but if you could expand on some of the other new technological developments that you think are just over the horizon. How will they shape the future of newborn screening? And as the Secretary's Advisory Committee, NIH, and the medical community, how they consider the implications of the new technological advancements like whole genome sequencing. Will there be formal opportunities for parents' voices to be heard in those debates?

Dr. HOWELL. Tandem mass spectroscopy has, of course, been the hallmark of the technology that has permitted us to expand. And again, this technology continues to be very useful and there are other things you can do. In other words, you get many, many compounds at once.

But I think that the technology that is on the horizon that will be shaping the future is whole genome, whole exome sequencing.

Senator HAGAN. What was the other one?

Dr. HOWELL. Whole exome or whole genome, in other words, with exomes you look at the active part of the gene and when you do the whole genome, you look at every little base pair. And so, most of those studies will be looking at the functional genes or the exome.

The National Institutes of Health has recently awarded four grants that look at the impact of whole genome, whole exome sequencing on newborn screening. And you can get out of the dried blood spot, you can extract a sample of DNA that is adequate enough to look at the whole exome or the whole genome.

This will be, indeed, an extremely powerful technology as far as the ability to look at conditions that we currently do not screen for, because you will be able to look at any gene of interest, et cetera.

I think that that technology will clearly be a driving force of the future.

Now, the question of will folks have an opportunity? All four grants that the NIH has recently awarded has a required section within them on the ethical, legal, and social issues of this technology. So that all of those grantees—be it at the University of California San Francisco, Chapel Hill, or Missouri, et cetera—they will all be having very careful looks at the ethical, legal, and social aspects of using this technology in newborn screening.

I think the technology will be extremely important in helping us understand some of our newborn screening findings. For example, when we have a baby born with certain conditions like Krabbe Disease, for example, one of the conditions screened in New York State, you have a low enzyme activity on the blood spot. But you cannot predict reliably whether that baby is going to have a really serious outcome or not so bad. And by looking at the whole genome, in other words you basically are looking at the whole environment.

By looking at the whole genome, the whole exome, you are going to be able to decide, “Well, goodness. I think this baby is going to do pretty good.” Or, “We have some really serious problems.”

I think to answer your question, the whole genome, whole exome sequencing will be the wave of the future. It is just now in some important pilot studies recently funded jointly by Child Health and the Genome Institute, and all of those have big time efforts to look at the implications for the family, and the community, and the public at large. And I think there will be tremendous care exerted as such technology advances to the public.

Senator HAGAN. Thank you.

I was pleased to hear that UNC Chapel Hill also got one of those grants too, to be a part of this study.

Dr. HOWELL. Yes.

Senator HAGAN. Before I became a U.S. Senator, I served on the ethics committee of a local hospital. And so I have been a witness to many discussions within family members on issues concerning many of the ethical concerns in a hospital setting. So it is something that, I know, will be quite a bit of research, scientific, and ethical debate.

Dr. HOWELL. Those are very important debates.

Senator HAGAN. They are. They are very, very important.

A question on followup assessment. This bill expands the scope of the current HRSA grants to ultimately ensure that followup care for newborns and families occur.

Dr. Howse, Dr. Howell, can you describe what the appropriate followup care from a nurse or a doctor receiving the newborn screening results should be, and what the common gaps are in providing the followup care?

Ms. HOWSE. I will make a couple of overview comments and then invite my colleague, Dr. Howell, to speak very particularly about pediatric care.

The key in newborn screening is that this is a program for which a test is linked to an effective treatment. That is really the heart and soul of the program. A test is linked to an effective treatment. And there is urgency, there is a great deal of urgency in terms of timely intervention and timely treatment for those newborns. The

key to followup is rapid-fire notification between the lab, the parents, the hospital, and the physician of record.

Often there are challenges when a baby is released from the hospital to make sure that pediatric care is immediately available to the child. So that is why there is an emphasis in the bill, because that was sensed as an area that needed to be strengthened, frankly.

One of the key questions has to do with the parents' ability to be connected to a primary physician, a pediatrician, and how that gets managed between the time of release from the hospital to the parents going back home. Insurance coverage matters, whether the parents have Medicaid or some other form of insurance. There have been, frankly, some issues there about continuity of care. So I do think that is an area that we need to look at.

From a clinical standpoint, though, I would really invite Dr. Howell to speak about how that connection gets made and how vital and important it is.

Dr. HOWELL. Let me make a couple of comments—the followup starts with the initial test. And what happens is that the State laboratory has an abnormal test that they confirm in the lab and so forth. They contact the primary care person who is usually a pediatrician or a family practitioner.

The initial problem at that point has been addressed fairly aggressively because of the following problem. Each of these conditions is individually rare. So that on Friday afternoon, if the lab in Raleigh calls a pediatrician in Cary on Friday—and this always happens on Friday afternoon about 4 o'clock—and says, "We have just had an abnormality in a fatty acid oxidation defect like Medium-Chain acyl-CoA dehydrogenase." And so the pediatrician or the primary care doctor probably has never heard of this condition. It would be unusual to have heard of it.

So one of the things that has been done to do this with regard to the panel, every condition on the panel, the American College of Medical Genetics has prepared a 1-page document that is called an ACT sheet, Immediate Action Sheet and it summarizes the name of the condition. It tells what the immediate problems are, and what you should do, and it has some references. And most State labs now are faxing that to the physician at that point so that when he or she calls the family, they will have a little bit of information already. They will know a little bit about it so that they say, "We need to repeat this." So that is the first, immediate followup. And then, obviously, depending on the condition, you need to institute the therapy.

The highly specialized therapy is usually coordinated through a referral center. For instance, if this baby is born in Cary, they would either go to Chapel Hill or to Duke, very likely, for followup. And the diet would be instituted and then they would be followed up long term with their doctor.

Some conditions require really aggressive, long continuing treatment such as Pompe Disease where you have to have regular infusions, and those would also be done.

Now, the other thing that is in this legislation that is very important is that we have never had a systematic way of following up data on these children.

Senator HAGAN. Right.

Dr. HOWELL. So we do not know a lot about some of these conditions except PKU, which had a Federal study.

So one of the things that is very important is that we really need to get more of these babies into followup programs so that we enter data on them at 6 months, 1 year so that when we come back in 10 years, we can say, "This is what has happened." Or, "We need to do this," so you can develop new therapies and new modifications.

But basically, the long-term followup is done, usually, by the primary care doctor and in conjunction with a regional medical center. And the State laboratories have been very, very experienced over the years. They know who to call.

For example, they will call. For instance, like this baby born in Cary that I just brought up, they will also call the lab or they will call the places at Duke and at Chapel Hill, and give them a heads up that there is a baby in their region that will likely be calling, so that they do not drop through the cracks. And I might point out the States are aggressive in finding these children.

I began my career at Johns Hopkins near here and the State patrol would go out and find a baby if the family could not be located because you need to find them and get them in. And the States have really been aggressive in discovering these early sick babies.

Senator HAGAN. Thank you for that background. I think that is very important and it certainly does paint a picture as to how the followup is actually done.

I think what you have stated too is how important the provision is in this bill, after 50 years of the screenings, that we really do need to have these long-term studies done, and followup on the children that have had abnormal screenings and, obviously, treatment.

This is an interesting thought. If Ethan had been born during the height of a hurricane, what would have taken place? I know that when you look at these natural disasters like Hurricane Katrina, what does that do to disrupt the newborn screening program?

The law required the CDC to write a national contingency plan with instructions for how to react in those instances. Our bill would require that plan to be updated at least every 5 years because we all know that babies do not stop being born just because all of a sudden there is a fire, or a hurricane, or a flood, or a power outage.

So Dr. Howse, do you know how the contingency plan was helpful when Hurricane Sandy hit up in the northeast last year, and how the affected States were able to maintain their newborn screening program?

Ms. HOWSE. Well, first of all, every State does have a contingency plan in place, and we did not receive any reports of interruptions to the newborn screening program in New York as a result of Hurricane Sandy.

We know that many of the medical facilities, particularly in New York City, were definitely affected very adversely. But there seems to have been a very supreme effort made in New York and New Jersey to make sure that vital medical services continued uninterrupted.

We know there were many emergency pregnancy labor and delivery situations that were handled. We know NICU babies were evacuated often in the arms of their nurses and doctors to be brought to more safe and secure locations. We also received, in addition to those kinds of reports, no indications that there were interruptions in this vital testing and followup. So I would, again, really commend the health professionals and the officials responsible.

That being said, no plan survives its first contact with reality. So I think it is very important for this provision to continually and regularly update those contingency plans, and make sure that particularly these babies do not fall between the cracks when Mother Nature comes to us in such a difficult way.

Senator HAGAN. Miss Bonhomme, do you have any examples of actually getting out and speaking with parents that have had deliveries during natural disasters?

Ms. BONHOMME. I have not spoken to any parents directly who had experience during the disasters, but I was in quite a bit of contact with the New Jersey Department of Health during that time.

And really from their experience, the fact that there was this plan in place, which actually forced different agencies to speak to each other beforehand and build the relationship before there was a disaster. When the Hurricane hit, that newborn screening program in New Jersey was able to pick up the phone and say, "Hey, we need help. We need to put this plan into place." And what actually happened in New Jersey is that there were State troopers that went and picked up the blood spots from hospitals and brought them to the lab.

And so I think it is the fact that there was a plan in place and people had met each other before. As you know, relationships are so important, so that there were these State troopers who knew what newborn screening was and why it was really important. I think that is just such a great example of why having this type of plan in place is so key, and why we need to keep those efforts going, and make sure that those plans are updated every 5 years.

Senator HAGAN. You know, it is great. I had not even thought about such a situation, but obviously, children do continue to be born during these natural disasters. So it certainly shows the efficacy of planning and then carrying it out, and building those relationships that you said. That certainly is very worthwhile.

Before the meeting started, Dr. Howell, you and I were speaking about how we are at the 50th Anniversary for this newborn screening in the United States. But what are other countries doing, looking at what I would call the best practices happening in the United States?

Can you just share a couple of thoughts before we end our hearing?

Dr. HOWELL. I think that there is interest throughout the world, really, at the current time in newborn screening.

As I mentioned briefly to you in the hall, Europe has been really interested in trying to harmonize its efforts between the members of the EU, the European Union, and that is even more difficult than it is to do between the States.

But in Europe, there is tremendous variation between the countries. You have countries like Austria and Germany that have programs that are very similar to ours. The United Kingdom has a very modest number of conditions on their program and they are very slowly moving ahead on that, but there is considerable discussion about trying to move ahead. And in all circumstances, they are very interested in how the United States has moved ahead and harmonized that. And so, we are invited to talk with them.

I think the National Institutes of Health and some other groups have also sponsored some meetings in the Middle East and North Africa where newborn screening, for example, is extremely well developed in oil-rich countries such as Saudi Arabia and Qatar. But in the more modestly funded countries, they have very little newborn screening.

It can be particularly beneficial in many countries of Africa because you have a high degree of first-cousin marriages. And when you have intermarriage, it dramatically increases the frequency of rare recessive conditions. So that a condition here that might occur in 1 in 20,000 in a country with intermarriage, it might occur in 1 in 5,000.

So I think that there is a great deal of interest in working with these countries to identify important conditions that can be identified and treated simply because they also lack the infrastructure. So there are a lot of discussions there.

China has a spotted screening program. They, again, are trying to move ahead in that. Other places like Australia and New Zealand have fairly well developed programs, but there is a potpourri around the country.

I think that one of the interesting things is that everyone is interested in what is happening in the United States, which is kind of always nice when someone thinks that the United States is doing something well.

Senator HAGAN. I think it is important that newborn screening does save lives.

I do think it is really important to reiterate, again, that in 2011, the CDC recognized the advances in newborn screening as one of the 10 great public achievements in the United States during the decade of 2001 to 2010.

For all of you here today, I really do, in particular Dr. Howell, thank you for all the past work that you, and the March of Dimes, Dr. Howse, have done to make this such a fundamental important health aspect in our country. Then also knowing, we have got to build on that. We certainly do need to expand it and to get this reauthorization done.

I want to thank all of you today for your testimony. The hearing record, as I stated earlier, will remain open for 10 business days for the other Senators to submit their statements or questions for the record.

And we will now adjourn this hearing.
[Additional Materials follow.]

ADDITIONAL MATERIALS

PREPARED STATEMENT OF MARCIA BOYLE, PRESIDENT AND FOUNDER,
IMMUNE DEFICIENCY FOUNDATION

Chairwoman Hagan, Ranking Member Enzi and members of the committee, thank you for convening this hearing to focus on the importance of newborn screening. I submit this testimony for the record on behalf of the Immune Deficiency Foundation (IDF). Founded in 1980, the Immune Deficiency Foundation (IDF) is the national patient organization dedicated to improving the diagnosis, treatment and quality of life of persons with Primary Immunodeficiency (PI) diseases through advocacy, education, and research. These diseases occur in persons born with an immune system that is either absent or hampered in its ability to function. These diseases are caused by hereditary or genetic defects and can affect anyone, regardless of age or sex. The World Health Organization recognizes more than 185 primary immunodeficiency diseases. My comments today will focus on Severe Combined Immune Deficiency (SCID), one of the rarest and the most devastating of these diseases.

SCID screening in newborns became possible just a few years ago with the development of the T-cell receptor excision circles (TREC) test that can detect SCID using the same dried blood spot filter cards that are currently collected from all babies to screen for a variety of inborn conditions. Infants affected by SCID lack T-lymphocytes, the white blood cells that help resist infections due to a wide array of viruses, bacteria and fungi. These genetic defects lead to extreme susceptibility to serious illness. As a result, the condition is fatal in infancy unless treated, usually with bone marrow transplantation. Transplants done in the first months of life have the highest success rate. A survey of more than 150 patients, commissioned by IDF, found that SCID patients who were diagnosed early and treated by 3.5 months had a 91 percent survival rate; those treated after 3.5 months had a 76 percent survival rate. If diagnosis is late, even a successful bone marrow transplant can still leave a patient with persistent health problems.

IDF was very pleased that Dr. Rebecca Buckley of Duke University served on the *Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children*. Dr. Buckley, Chair of the IDF Medical Advisory Committee, has spent most of her career addressing genetic disorders of the immune system and has been a strong proponent of newborn screening for these types of diseases. She has been a pioneer in the use of bone marrow transplantation to provide immune reconstitution to all infants with SCID.

The diagnosis of SCID very early in life is a true pediatric emergency, and the decision to screen for SCID will literally save the lives of infants. We are very pleased that this has been recognized at the Federal level. In May 2010, SCID was added to the Recommended Uniform Screening Panel. Since that time, we have been working to ensure that States implement newborn screening for SCID. To date, 16 States and the territory of the Navajo Nation have already implemented newborn screening for SCID. Based on the screening done in these States, SCID is estimated to occur in approximately 1 in 40,000 to 1 in 70,000 births. Without newborn screening for SCID, these children have little chance at an early diagnosis and treatment. Newborn screening has led to the identification and treatment of dozens of infants with SCID and many more with other kinds of T-lymphocyte deficiencies in those States that are screening. Successful screening ensures that these babies will have the opportunity for early treatment and the chance of a normal, healthy life because of early detection.

This fall, the Center for Disease Control and Prevention's (CDC) National Center for Environmental Health, Newborn Screening, and Molecular Biology Branch publicized that three more States—Georgia, Oklahoma, and Virginia—will be funded for a total for \$1,800,000 under the "Program to Support New Implementation of State of Territorial Public Health Laboratory Capacity of Newborn Bloodspot Screening of SCID." These States will be compensated between \$250,000 and \$300,000 per year for 2 years, which will help support the implementation of SCID to their newborn screening panels. CDC previously funded Wisconsin, Massachusetts, Michigan, and Minnesota to include SCID on their newborn screening panels and now, all four States maintain active NBS SCID screening programs. Today, 44 States and the District require screening of at least 29 of the 31 treatable core conditions.

Newborn screening has a profound impact on children with SCID and their families. Therefore, IDF urges Congress to pass S. 1417, "the Newborn Screening Saves Lives Reauthorization Act of 2013" introduced by Chairwoman Hagan and Senator Hatch. Importantly, this legislation would reauthorize Health Resources and Services Administration (HRSA) grants to States to expand and improve their screening programs, educate parents and healthcare providers, and improve followup care for

infants with a condition detected through newborn screening and continue to support the Advisory Committee on Heritable Disorders in Newborns and Children, which provides States with a Recommended Uniform Screen Panel to help ensure every infant is screened for conditions which have a known treatment. The legislation includes a number of provisions that will strengthen current efforts, ensure timely review of new conditions, promote quality assurance and support research in this area.

As we celebrate the 50th anniversary of Newborn Screenings this year, on behalf of IDF, we hope Congress will support the facilitation of a comprehensive newborn screening program in every State to save the lives of thousands of newborns.

Thank you for your serious consideration of this critical issue.

RESPONSE TO QUESTIONS OF SENATOR WARREN BY NATASHA BONHOMME

Question 1. Newborn screening is carried out on a State level by public health departments, and their labs are doing some of the research that leads to new tests. State budgets, and public health departments' in particular, are under considerable pressure. Can you tell us about the States' need for Federal funding for screening, and the impact of funding reductions on public health labs, screening programs, and our ability to invest in further research?

Answer 1. While newborn screening is run at the State level, Federal funds are used to maintain the integrity of these programs from both a laboratory and followup perspective. As State budgets contract, there is a growing need for Federal support to maintain the basic functions of the newborn screening programs. The Centers for Disease Control and Prevention (CDC) assesses the quality of newborn screening programs. This ensures that State laboratories are compliant with the Federal Clinical Laboratory Improvement Amendments (CLIA). Without the support of CDC, States would have to find other ways to meet CLIA requirements. Even if this were possible, there would be a great deal of uncertainty regarding the validity of a new assessment process. Through CDC, Federal funds are also used to help newborn screening programs improve the quality of tests as well as provide training on new testing methodologies. Without this work, progress in detecting treatable conditions would greatly diminish and could lead to negative outcomes for children who otherwise would have been diagnosed early. Without the Federal funding, States would not be in a position to invest in updating their approaches to screening, causing newborn screen programs to regress.

In recent years, Federal funds have also gone to support the implementation of and education around conditions newly added to screening panels. The Health Resources and Services Administration (HRSA) currently runs the Critical Congenital Heart Disease (CCHD) Newborn Screening Demonstration Program. This program focuses on increasing the number of newborns screened for CCHD before discharge from newborn nurseries. The grantees of this program utilize validated screening protocols and enhance State newborn screening infrastructure; as well as create or build upon their State's infrastructure to collect and utilize information from various hospitals within a health information network for the detection of CCHD and related patient followup and outcomes. HRSA also oversees a number of cross-State/cross-regional programs to encourage sharing of information and support for public education, including *Baby's First Test*, the Nation's newborn screening clearinghouse.

Question 2. As science is rapidly advancing, some parents have concerns about research on their baby's blood samples. Massachusetts has a mandatory screening panel, and optional tests that the Department of Public Health is researching to determine whether there is enough evidence to require them. Parents must give informed consent before these optional screens. This strikes a good balance between making sure that all babies are screened, and making sure that parents are informed about what is research and what is clinical care. Are there national efforts underway to better inform parents about research that may take place with their children's blood samples?

Answer 2. Many States will have an optional or pilot panel. Typically, newborn screening programs use these panels to determine if there is strong enough evidence to include certain conditions on their mandatory panel. Because every State has a different protocol adding conditions to the required list, the process of consent does vary. There are national efforts to have State newborn screening programs share their process so best practices can be developed. Because not every State at all times has a pilot program or optional list of conditions running, it is important that educational efforts not only continue but also expand. This will ensure that parents in all States are aware of newborn screening and what conditions are included as well

as how the bloodspots potentially may be used in research and quality control for the State programs.

There are also specific efforts in many States to inform parents and the public about the different types of research that may take place with a bloodspot. Particularly, Michigan and Minnesota have done extensive work on educating the public about how bloodspots are stored and the potential uses in regards to improving the newborn screening programs and helping to detect life-threatening conditions. The National Institutes of Health funded a group from the University of Utah to better understand what the public, nationwide, knew about this topic and to determine public options and educational needs around bloodspot usage and research.

Question 3. Some parents have concerns about the disclosure of their children's genetic information. It's extremely important that providers and public health departments are transparent about how samples are used and stored. Can you tell us how patient information is protected, and how any genetic information is kept safe?

Answer 3. There is a national push through the work of organizations such as the Association of Public Health Laboratories, to encourage all State programs to have and periodically update their policies on newborn screening information. *Baby's First Test* works closely with these entities on having these policies be transparent and easily accessible to the public. While there are great barriers to State programs due to budget cuts, there are efforts to build upon existing infrastructure to keep patient information protected. This includes protected databases that follow standard procedures for public health data, limited access to data, and employee training on data practices and State statutes. All data is kept behind protected firewalls. Newborn screening programs also comply with their State's Government Data Practices Act. When research is done, any identifiable information is removed from the sample in accordance with the Office of Human Research and Protection guidelines. If for some reason identifiable information is needed, consent from parents is required ahead of time.

RESPONSE TO QUESTION OF SENATOR WARREN BY R. RODNEY HOWELL, M.D.

Question. Mandatory newborn screening saves lives, however some parents have concerns with the mandatory nature of newborn screening. These concerns stem from whether these tests are necessary, cost-effective, or appropriate for widespread administration. Can you describe the process and factors considered by the Advisory Committee to determine whether new tests should be added to the list of recommended conditions, and the importance of mandatory screening?

Answer. The Advisory Committee (SACHDNC) has developed a lengthy, rigorous process but fully transparent, process for nominating, reviewing, and recommending new conditions to the Secretary of Health and Human Services. Any individual or group can nominate a condition to be considered for addition to the recommended uniform panel. The process and the nomination form is described in detail with directions on the Advisory Committee Web site (<http://www.hrsa.gov/advisorycommittees/mchbadvisory/heritabledisorders/>). The nomination form to be completed by the nominating group includes details about the affiliation of the nominator and organizations making the nomination.

Section I of the nomination form describes the condition nominated (type of disorder, screening method, genetic information, case definition, incidence, timing of clinical onset, and severity of disease). Only conditions which are very serious (with regards to morbidity, disability, mortality), and have reliable screening tests available are considered. Later in this section of the form an outline of the treatment is required: modality, urgency, benefits, availability, and potential harms of treatment.

Section II, part A of the nomination form requires evidence of a validated laboratory test, evidence of widely available confirmatory testing, and a prospective population-based pilot study. Extensive laboratory quality information about the screening test is required. Part B of this section of the form requires information about the confirmatory testing (validity, type of sample required, is test FDA-approved, and a list of the CLIA-approved labs in the United States offering confirmatory testing) Section II, part C requires the detailed information from the population-based pilot study, including false positive and negative rates, and number of infants with confirmed diagnosis).

Section III requires a series of the key references supporting the nomination.

The nomination form is then sent to HRSA, where it is reviewed to ensure it is complete, and then sent to the Advisory Committee. The exact details of how the Advisory Committee handles nominated conditions has been published in order to ensure transparency of the process (Committee Report: Method for evaluating condi-

tions nominated for population-based screening or newborns and children, Calonge, N, et al. *Genetics in Medicine* 12:153–159, 2010). Once complete, the nomination package is studied by the committee’s internal nomination and prioritization workgroup to ensure that it is likely that there is sufficient information to permit a systematic evidence review of the natural history and severity of the condition, the analytical and clinical validity of the screening tests, and the effectiveness of treatments. If this workgroup feels it is appropriate the nomination is then moved to an external workgroup for a systematic evidence review.

The external workgroup was established through a competitive contract through HRSA, and is comprised of an independent group of experts in evidence review. Since all recommendations by the committee must be evidence-based, this group conducts a structured, detailed evidence review of all the issues involved. They have also published how they do the evidence review, so again that is transparent (An evidence development process for newborn screening. Perrin, JM, et al. *Genetics in Medicine*, 12:131–34, 2010). The current charge to the Advisory Committee includes cost-effectiveness analysis as one category of evidence to be considered by the committee. A recent publication by members of the external workgroup (with the addition of some experts in cost analysis) has outlined how this information can be gathered (Decision analysis, economic evaluation, and newborn screening: challenges and opportunities. Prosser, LA, et al, *Genetics in Medicine*, 14:703–12, 2012).

After the external workgroup finishes its detailed evidence review (all of which are published verbatim on the SACHDNC Web site), the entire committee reviews and discusses all the evidence, asks for input from the public, and then finally votes on its recommendation. The committee recommendations are sent to the Secretary of Health and Human Services who makes the final determination as to its being added to the Recommended Uniform Screening Panel (the RUSP).

Laws and regulations regarding mandatory newborn screening of newborns are in force in virtually all States. In all circumstances States make these rules. Some States have historically had regulations that required asking permission to perform newborn screening, but from the information I have these have in general not been observed. The conditions on the newborn screening panel have been identified as being very serious, life-threatening, or life-altering conditions. All have treatments. Since these conditions are inherited in a recessive fashion, families would not have any way to know that their children were at risk. And since they are so very serious (some cause profound retardation, and other sudden death without treatment), it is my strong feeling that no competent adult could decline a test, minimally invasive, which could be life saving for their infant.

It is certainly the practice currently that if a newborn screening test is being performed for a research purpose, permission is asked from the family. It is interesting that in some recent work where research studies were being performed on infants in certain hospitals, the overwhelming majority of parents agreed to the study, although the condition being studied had, at that time, no treatment. There needs to be a great effort to better inform parents about newborn screening, since most at this time are poorly informed. It is most difficult to include this information at the time of birth since the usual mother stays only 24 hours in the hospital after birth, and these few post-partum hours are filled with many activities, and hopefully some rest.

Since the conditions on the recommended uniform screening panel are individually rare, there are many areas of research needed, such as long-term outcomes and treatment followup of screened newborns and how best to obtain informed consent for future newborn screening research.

RESPONSE TO QUESTIONS OF SENATOR WARREN BY JENNIFER L. HOWSE, PH.D.

Question 1. Newborn screening is carried out on a State level by public health departments, and their labs are doing some of the research that leads to new tests. State budgets and public health departments’ in particular, are under considerable pressure. Can you tell us about the States’ need for Federal funding for screening, and the impact of funding reductions on public health labs, screening programs, and our ability to invest in further research?

Answer 1. The March of Dimes recognizes that budget limitations present many challenges at the State level, and funding cuts can pose difficult choices for newborn screening programs. For example, shrinking budgets hinder the ability of States to update their newborn screening panels with new conditions added to the Recommended Uniform Screening Panel. Today, 44 States and the District of Columbia require screening for at least 29 of the 31 treatable core conditions. Federal funding supports efforts to implement pilot studies that can assist States to more quickly adopt screening for new conditions. For example, the Centers for Disease Control

and Prevention provided pilot funding to Minnesota and Michigan to assist with the implementation of severe combined immunodeficiency (SCID) screening and determine best practices for other States to implement. While additional Federal resources would speed implementation of SCID and other conditions in States, reductions in Federal funding coupled with limitations at the State level could completely halt the important progress being made.

Cuts to Federal and State funding not only affect the number of conditions screened, they also affect the quality of the screening itself. The Centers for Disease Control and Prevention's Newborn Screening Quality Assurance Program (NSQAP) is a voluntary, non-regulatory program to assist State health departments and their laboratories in maintaining and enhancing the quality of test results. The program provides services to more than 85 domestic newborn screening laboratories, 31 manufacturers of diagnostic products, and laboratories in 67 countries. NSQAP has been the only comprehensive source of essential quality assurance services for dried bloodspot testing for more than 33 years. In partnership with State laboratories, NSQAP continues to make improvements in services offered and to meet the growing and changing needs for newborn screening in the public health community. Reductions in funding would hamper this progress and prohibit NSQAP from assisting public health laboratories in developing and refining screening tests, conducting pilot studies, and implementing new methods to improve detection of treatable disorders. Funding cuts would impact the quality of these critical tests that prevent death and disability.

The March of Dimes would be pleased to provide you with more information about the impact of budget cuts on specific research initiatives or related undertakings.

Question 2. As science is rapidly advancing, some parents have concerns about research on their baby's blood samples. Massachusetts has a mandatory screening panel, and optional tests that the Department of Public Health is researching to determine whether there is enough evidence to require them. Parents must give informed consent before these optional screens. This strikes a good balance between making sure that all babies are screened, and making sure that parents are informed about what is research and what is clinical care. Are there national efforts underway to better inform parents about research that may take place with their children's blood samples?

Answer 2. The March of Dimes recognizes that Federal and State newborn screening laws must strike a careful balance between advancing public health and protecting individual privacy. Historically, each State has retained the authority to determine which tests are included in its newborn screening panel, how those tests are offered to families, and the operation of systems for storage and future use of newborn bloodspots. There are no Federal laws or rules about how these important decisions should be made; however, the U.S. Secretary of Health and Human Services' Advisory Committee on Heritable Disorders in Newborns and Children issued a report in July 2011 titled, "Considerations and recommendations for national guidance regarding the retention and use of residual dried bloodspot specimens after newborn screening." This report was compiled in order to provide basic guidance for State policies related to protecting an individual's privacy and to allow for the important public health uses of the residual bloodspots.

In addition, the Federal Government supports a comprehensive source of information about newborn screening, known as Baby's First Test. This online information clearinghouse is maintained by the non-profit Genetic Alliance and funded by the Genetic Services Branch of the Maternal and Child Health Bureau of the Health Resources and Services Administration (HRSA). The clearinghouse provides current educational and family support and services information, materials, and resources about newborn screening at the local, State, and national levels. This resource is dedicated to educating parents, family members, health professionals, industry representatives, and other members of the public about the newborn screening system, including what happens with the residual bloodspots.

More specifically, *Baby's First Test* provides parents with detailed State-specific newborn screening program overviews for all 50 States and the District of Columbia, including information on all screened conditions, any "opt-out" actions available, and the rules and procedures in each State that govern the storage and use of dried bloodspots.

The March of Dimes would be pleased to share further information with you about any aspect of Federal or State laws or guidelines related to research on newborn bloodspots.

Question 3. Some parents have concerns about the disclosure of their children's genetic information. It's extremely important that providers and public health de-

partments are transparent about how samples are used and stored. Can you tell us how patient information is protected, and how any genetic information is kept safe?

Answer 1. The March of Dimes believes in the importance of patient privacy protections related to the use of newborn screening information. Each State has laws governing the storage and use of newborn bloodspots and the associated information. State laws related to privacy of personal or medical information generally apply to any unauthorized access to or malicious use of newborn screening information. Protecting the interests of the infants from whom the dried bloodspots are obtained is of the utmost importance to State public health programs, and States continue to refine guidelines for the use of residual samples.

Most State newborn screening programs routinely use post-screening residual samples for the purpose of laboratory quality assurance (i.e., comparing results of the tests from the screening laboratory with those of the reference laboratory) and for the development of new screening methods. This ensures the ongoing accuracy of laboratory equipment and methods and ensures that results will be correct for all newborns.

Beyond the protections afforded by States, any research undertaken with newborn bloodspots must be conducted in an ethical manner that respects and protects the rights of children and their families. Federal regulations on the protection of human subjects, known as the Common Rule, apply to all research that is conducted or supported by any U.S. Federal agency or department. These rules would require studies that use newborn screening bloodspots be reviewed and approved by an Institutional Review Board (IRB). Additionally, this research is governed by protections provided by executive agencies such as the Food and Drug Administration and laws that govern medical privacy such as the Health Insurance Portability and Accountability Act (HIPAA).

The March of Dimes would be pleased to supply you with further information on privacy laws in any specific State or region, or examples of laws that deal with certain aspects of the permissible uses of newborn screening bloodspots.

[Whereupon, at 11:28 a.m., the hearing was adjourned.]

