ADVISORY COMMITTEE ON HERITABLE DISORDERS IN NEWBORNS AND CHILDREN

WASHINGTON MARRIOTT AT METRO CENTER
WASHINGTON, D.C.
JANUARY 21-22, 2010

THURSDAY, JANUARY 21, 2010

7:30 A.M. – 8:30 A.M. **CONTINENTAL BREAKFAST** (COMMITTEE MEMBERS AND PRESENTERS)

8:30 A.M. – 8:45 A.M. **WELCOME**

- APPROVAL OF MINUTES FROM THE SEPTEMBER 2009 MEETING
- COMMITTEE CORRESPONDENCE

R. RODNEY HOWELL, M.D.
COMMITTEE CHAIR

8:45 A.M. – 9:45 A.M. **REPORTS FROM THE NOMINATION AND PRIORITIZATION WORKGROUP**

- HYPERBILIRUBINEMIA
- CRITICAL CONGENITAL HEART DISEASE

PIERO RINALDO, M.D., PH.D.
COMMITTEE MEMBER

9:45 A.M. – 10:30 A.M. **CLEARINGHOUSE FOR NEWBORN SCREENING INFORMATION**

SHARON TERRY, M.A.
PRESIDENT, GENETIC ALLIANCE

10:30 A.M. –11:00 A.M. **BREAK**

11:00 A.M. – 12 NOON **COMMITTEE REPORT ON THE RETENTION AND USE OF RESIDUAL DRIED BLOOD SPOT SPECIMENS AFTER NEWBORN SCREENING — COMMITTEE DISCUSSION**

ALISSA JOHNSON, M.A.
JOHNSON POLICY CONSULTING

12 NOON – 1:00 P.M. **LUNCH** (COMMITTEE MEMBERS AND PRESENTERS)

1:00 P.M. – 1:45 P.M. **NEWBORN SCREENING AND HEALTH CARE REFORM**

ALISSA JOHNSON, M.A.
JOHNSON POLICY CONSULTING

1:45 P.M. – 2:15 P.M. **T-LYMPHOCYTE DEFECTS/SEVERE COMBINED IMMUNODEFICIENCY (SCID)**

JENNIFER PUCK, M.D.
PROFESSOR OF PEDIATRICS
UNIVERSITY OF CALIFORNIA

2:15 P.M. – 2:30 P.M. **PUBLIC COMMENTS**

2:30 P.M. – 3:00 P.M. **COMMITTEE DISCUSSION**

3:00 P.M. – 5:30 PM **SUBCOMMITTEE MEETINGS**

- **FOLLOW-UP AND TREATMENT**
- **EDUCATION AND TRAINING**
- **LABORATORY STANDARDS**

5:30 P.M. **ADJOURN**

FRIDAY, JANUARY 22, 2010

7:30 A.M. – 8:30 A.M. **CONTINENTAL BREAKFAST** (COMMITTEE MEMBERS AND PRESENTERS)

8:30 A.M. – 9:30 P.M. **SUBCOMMITTEE REPORTS**

- **SUBCOMMITTEE ON LABORATORY STANDARDS AND PROCEDURES**

  GERARD VOCKLEY, M.D., PH.D.
  COMMITTEE MEMBER

- **SUBCOMMITTEE ON EDUCATION AND TRAINING**

  JANA MONACO AND TRACY L. TROTTER, M.D.
  COMMITTEE MEMBERS

- **SUBCOMMITTEE ON FOLLOW-UP AND TREATMENT**

  COLEEN BOYLE, PH.D., M.S.
  COMMITTEE MEMBER

9:30 A.M. – 10:30 A.M. **CARRIER SCREENING FOR SICKLE CELL DISEASE**

- **REPORT FROM THE SICKLE CELL DISEASE ASSOCIATION OF AMERICA WORKSHOP ON CARRIER SCREENING**

  LANETTA JORDAN, M.D.
  SICKLE CELL DISEASE ASSOCIATION OF AMERICA

  KWAKU OHENE-FREMPONG, M.D.
  COMMITTEE MEMBER

10:30 A.M. – 10:45 A.M. **RESPONSE TO COUNCIL ON BIOETHICS' REPORT ON NEWBORN SCREENING – UPDATE**

TRACY L. TROTTER, M.D.
COMMITTEE MEMBER

10:45 A.M. –11:15 A.M. **BREAK**

11:15 A.M. – 11:30 A.M. **WORKGROUP ON DATA – UPDATE**

COLEEN BOYLE, PH.D., M.S. AND PIERO RINALDO, M.D., PH.D.
COMMITTEE MEMBERS

11:30 A.M. – 12 NOON **NEWBORN SCREENING INTEROPERABILITY SPECIFICATIONS**

ALAN ZUCKERMAN, M.D.
GEORGETOWN UNIVERSITY

12 NOON – 1:00 P.M. **LUNCH** (COMMITTEE MEMBERS AND PRESENTERS)

1:00 P.M. – 2:00 P.M. **EVIDENCE REVIEW WORKGROUP REPORT: LITERATURE REVIEW FOR HEMOGLOBIN H DISEASE**

ALEX KEMPER, M.D., M.P.H., M.S.
EVIDENCE REVIEW WORKGROUP

2:00 P.M. – 2:30 P.M. **PUBLIC COMMENTS**

2:30 P.M. – 3:00 P.M. **COMMITTEE BUSINESS**

- CALENDAR FOR 2010 COMMITTEE MEETINGS
- AGENDA ITEMS FOR MAY 2010 MEETING

R. RODNEY HOWELL, M.D.
COMMITTEE CHAIR

3:00 P.M. **ADJOURN**

APPENDIX A: WRITTEN PUBLIC COMMENTS

COMMENTS PRIOR TO THE ADVISORY COMMITTEE'S VOTE ON SCID

1. **Fred and Vicki Modell, Jeffrey Modell Foundation**

2. **Missy and Mike Bornheimer, Parents of a Baby Recently Born with SCID/Rac 2 Mutation in Wisconsin Who Was Cured**

3. **Stacey and James Barrett, Parents of a Baby Recently Born with SCID in Oregon Who Did Not Survive**

4. **Barbara Ballard, SCID Family Network and Immune Deficiency Foundation**

OTHER COMMENTS

5. **Sylvia Au, M.S., C.G.C., Newborn Metabolic Screening Program, Hawaii Department of Health**

6. **Annamarie Saarinen, Parent of a Child with Congenital Heart Disease**

7. **Andrea Williams, Children's Sickle Cell Foundation, Inc.**

8. **Micki Gartzke, VP, Save Babies Through Screening & Parent of a Child Who Died from Krabbe Disease**

9. **Susan Gallagher, Parent of a Child with Phenylketonuria (PKU) (comments submitted by e-mail)**

1. Fred and Vicki Modell
Jeffrey Modell Foundation
Statement to the HHS Advisory Committee
on Heritable Disorders in Newborns and Children
January 21, 2010

Let me first say to you Mr. Chairman and members of the Advisory Committee, thank you for this opportunity. Most of you know Vicki and I established the Jeffrey Modell Foundation in memory of our son, who lost his battle with one of the primary immunodeficiency diseases at the age of 15. Since our earliest days with the foundation, we have had a very close collaboration with the Centers for Disease Control and Prevention (CDC) and with the National Institutes of Health (NIH) on biomedical research and the CDC on education and awareness activities. Our work with the Appropriations Committee in both houses of Congress has had a profound impact. In the areas of research, public awareness, and physician education, these efforts have actually led to extraordinary results for these often-undiagnosed disorders.

But in recent days we have directed our efforts and resources to implementing population-based screening for severe T-cell lymphopenia including SCID. We have always stressed the need for earliest possible diagnosis. And we actually believe, as most of you do, that newborn screening is the ultimate path to reaching that goal.

This Committee has an opportunity today to make history. The evidence-based review that Dr. Puck talked about was brought before this committee a year ago, raised some very important and very relevant questions about screening for SCID. Now, with general population screening of all births well established in Wisconsin and Massachusetts, and with programs ready to launch. Ready to launch in New York, California, Louisiana, Texas, Minnesota, and Connecticut, we can be assured that those questions raised have been adequately addressed.

- First, the review questioned the prevalence of SCID. The NIH estimates prevalence at 1 in 100,000. There are other experts in the field that believe it's closer to 1 in 40,000 once we started screening. Without screening, newborns with this disease will develop overwhelming infections. With intervention, morbidity and mortality is greatly reduced and many babies are in fact totally cured.
- Second, the review questioned the accuracy of the screening. And Dr. Routes, who made an eloquent presentation and also in the December 2009 Journal of the American Medical Association article, addressed that issue. And there is specificity and sensitivity that was reached with this test.
- Third, the review questioned the feasibility of conducting this screening. To date, Wisconsin and Massachusetts have screened nearly 200,000 babies. Both states have indicated that their respective laboratories can handle three to four times that number. And they're willing to make their protocols and their laboratories available to the states. So as the states gear up, feasibility is no longer an issue.
- Fourth, the review raised the issue of public acceptance of the screen. In Massachusetts as an example, where families can opt out, such requests are less than 1 percent. There is nothing in this test that would generate controversy or otherwise offend the overwhelming majority of American parents.
- Fifth, the review raised the issue of cost-effectiveness. Wisconsin and Massachusetts have reported the cost at about $5.00, $5.50. And the CDC, Newborn Screening Laboratory in Atlanta has developed and even simpler method to run the TREC assay, further lowering the per unit cost and the capital investment. Wider application of screening will drive down the cost even more.

- Sixth, and finally, the review questioned the adequacy of available treatment centers. The Jeffrey Modell Center's network consists of 79 research, diagnostic, and referral centers at leading academic teaching hospitals throughout the United States. They have skilled and 15 experienced experts, and teams in place and are fully prepared to respond.

Now let's think about it. Each day about 11,000 babies are born in the United States. But only, as of today, only about 300 to 400 are born in those states that screen for SCID. They will be the lucky ones. They will be diagnosed. They will be treated, often cured, and have a good chance at life. If on the other hand, they are among the unlucky ones. If they live in 48 out of the 50 states that do not currently screen for SCID, they will be sick throughout their entire lives. And they'll be short lives.

I know that this Advisory Committee does not mandate the states to adopt these tests. But we can tell you from our experience, with meetings we have held in states across the country, that your actions, your actions are critical in implementing this screening. When this test is added to the core panel, states will move forward. Screening programs for SCID will be routine and precious babies will be saved. All of us in this room know that screening for SCID is not only the right thing to do, it's the smart thing to do. And in this case, the word smart is a great acronym for all of the essential elements required for a successful newborn screening program.

SMART—specific, measurable, achievable, realistic, and timely. Mr. Chairman and members of this committee, this is our moment. We have the technology to screen for SCID with 99 percent plus accuracy. We have a success rate of over 95 percent to treat these babies. The cost for this life saving screen is $5.00 or $5.50. The investment for the laboratory equipment, personnel, and supplies at the state level has been addressed. It has been resolved and it does not pose a problem. And our foundation continues to commit funding to jumpstart population screening in the states using the TREC assay.

Tomorrow, another 11,000 babies will be born in this country. Your decision today can give great comfort and hope to those new mothers and fathers who will not have to risk a tragedy and a loss of their child to severe combined immunodeficiency or lymphopenia.

Vicki and I accept the fact that science and discovery did not come in time for Jeffrey. But we are dedicated and committed to working with you to help all of the Jeffrey's in the future. This is our wish. This is our hope. This is our dream. Let us go forward on this journey together, beginning today.

Thank you.

2. Missy and Mike Bornheimer
Parents of a Baby Recently Born with SCID/Rac2 Mutation in Wisconsin Who Was Cured
Statement to the HHS Advisory Committee
on Heritable Disorders in Newborns and Children
January 21, 2010

Mr. Chairman and members of the Advisory Committee, my name is Missy Bornheimer and I am here today with my family. We come from Edgar, Wisconsin. It's a small town in central Wisconsin with about 1,400 people. And I would like to thank you for this opportunity to tell our story. We were thrilled at the prospect of welcoming home our second child in June of 2008.

We were so excited and felt blessed to have a new baby brother for Dylan. Mike and I would say to each other, life was good. Our son, Dawson, was born on June 12, 2008. When the pediatrician called us 12 days later with the news that our newborn baby Dawson may have a life-threatening condition called severe combined immunodeficiency, our life was changed overnight. Our dreams were shattered and we were devastated. We learned that SCID, or "boy in the bubble" disease, was a condition in which most babies do not make it to their first birthday.

But fortunately, we were blessed. Just a few months earlier, Wisconsin had started screening every newborn baby in Wisconsin for this disease in a program funded by the Children's Hospital of Milwaukee, Wisconsin Public Health Laboratory, and the Jeffrey Modell Foundation. The doctors at Children's Hospital told us that for Dawson to have a chance at life he would need to have a bone marrow transplant. On the day of his transplant, every single person in our Edgar School District wore a t-shirt that said, Dawson has big dreams. And with lots of prayers and support from family and friends, the transplant was successful and his life was saved. All of this because Dawson was born in Wisconsin, the first state in the nation to screen for primary immune deficiencies.

And today, Dawson is the first baby in the world born with a combined immunodeficiency who was cured as a result of this newborn screening. It is scary to think that if Dawson had been born just six months earlier, he might not be with us today. We give thanks every day that we live in Wisconsin. A drive from our home takes only about two hours to go to Minnesota, Michigan, Iowa, or Illinois; none of which currently test for SCID. What if we chose to live just two hours away? We would not have our beautiful son, Dawson.

Mr. Chairman, I would personally like to thank you and each of the members of the Advisory Committee for giving Dawson and our family at a chance at life. You have played a huge role in saving my baby's life. My days are filled with smiles, laughter, and happiness because of you. And I hope your days are filled with the same knowing that. Because of you, I get to be a mom to one of the most wonderful babies in the world. And how do you express thanks for something like that.

Our only wish is that young families like ours in Minnesota, Michigan, Iowa, Illinois, and all of the states can feel secure knowing that if any one of them gets a call from their pediatrician like we did, a program of newborn screening can turn a devastating tragedy into the kind of joy that Dawson gives us every single day.

Thank you.

3. Stacey and James Barrett
Parents of a Baby Recently Born with SCID in Oregon Who Did Not Survive
Statement to the HHS Advisory Committee
on Heritable Disorders in Newborns and Children
January 21, 2010

Stacey & James Barrett January 21, 2010

Testimony for Universal Newborn Screening for SCID

On behalf of Liam, our family and all the families living with the affects of SCID, I thank you for giving me the opportunity to speak. My name is Stacey Barrett and this is my husband, James. We are Liam's parents. Our son was diagnosed and passed away from SCID. Liam would have been one on the 30th of this month.

Liam was born on January 30, 2009 in Oregon, in the wrong state. If we had been in Massachusetts or Wisconsin, Liam would have been tested at birth for SCID. If that had been the case, his journey, our journey may have had a different ending.

Our family's journey with SCID began on June 1st when Liam was admitted to the hospital for Failure to Thrive. That was 8 months after this committee voted to delay acceptance of universal newborn screening for SCID, 10 years after the American Academy of Pediatrics (AAP) called for national newborn screening standards, 6 years after an expert on SCID, Dr. Rebecca Buckley, testified at the first meeting of this committee saying that SCID was a pediatric emergency and should be included in the uniform panel, 2 years after SCID was nominated, and 18 months after Wisconsin began screening for SCID.

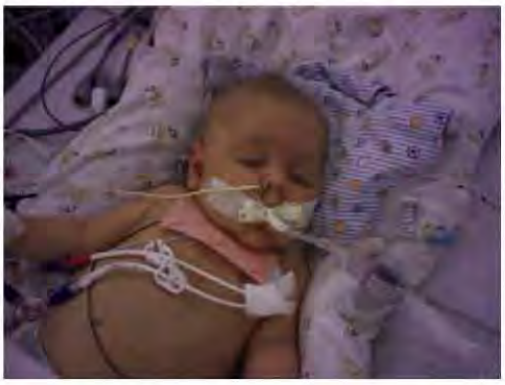
At four months old he was 5 pounds below the weight for his age. During our hospital stay, the doctors ran several tests for genetic diseases; all the while Liam was gaining weight at a steady rate. Because every test came back negative, the conclusion was that Liam was behind on weight because of a common cold. After 19 days in the hospital, we were sent home with Liam on a feeding tube, antibiotics and physical therapy. We were told this would be a long haul, but he would eventually fall back into the correct percentile for his weight.

After 5 days at home and several more trips to the doctor's office, we received the call for us to take Liam back to the hospital to be admitted. His blood count was low. A few days later we received the news that he had SCID. My husband and I were numb. How could something like this happen to us? We had no genetic trace of SCID in our family. We have three healthy children that were born before Liam that did not have SCID. We started going through the process blind. We had no idea on where to take our son for care. Little did we realize that this was only the first step in our journey.

During his second hospital stay, Liam was diagnosed with three more infections. All together, he had four infections but no immune system. He was only five months old. His diagnoses was 3 months later than published articles have stated a SCID child could be successfully treated with bone marrow transplant after diagnosis at birth.

We then traveled to Seattle Children's Hospital to await a bone marrow transplant, which we hoped would come from a sibling match. Unfortunately, being diagnosed with four infections prior to admission in Seattle, the doctors were extremely cautious.

Good news came when we were told that Liam's 3-year-old sister, Rylee was a perfect match. The only obstacle in our way was the infections, which were now down to only two. But the two left, PCP and Para Influenza III were the most serious and life threatening.



Although the bone marrow transplant was a success and he was engrafting well with his sister's marrow; Liam suddenly took a turn for the worse. The infections in his lungs were getting worse. On August 16th, Liam's CO2 levels had reached over 100, more than twice the amount of an average baby. His heart rate was decreasing and he was completely sedated into a coma. As we watched his vitals decline, we believe this was his way of telling us he was tired. On the 17th of August, my husband and I, with help from Liam, made one of the hardest decisions in our life… to let him go.

If our family were living in Wisconsin or Massachusetts at the time Liam was born, Liam would have been diagnosed with an Immune Deficiency. Shortly after birth he could have had a transplant with no infections. If that were the case, statistically my son would have had a higher success rate if diagnosed at birth, over 97% as Dr. Buckley testified before this committee in 2004. Statistics indicate our son would still be alive.

To many times our society's political infighting creates delay in progress. My son is a casualty in bureaucracy. If we have the means to test a child for a disease, the means to have a successful survival rate, what stops us from doing it? With immune deficiency, we cannot afford to wait for this board to decide whether it can be statistically proven that screening for SCID is cost effective and meets other rigid rules that focus on a population of newborns instead of on each newborn as an individual. Action needs to be taken now. While we wait for numbers and testimonies, countless children have lost their lives to this condition. It is incredible that we don't know the numbers lost to this disease because there is no national database to collect this information and the stories of these vulnerable newborns. Liam's story being one of the most recent and too familiar.

It is society's duty to protect and nourish the young children in our lives. It is the responsibility of this board to utilize its power to save lives. What are we waiting for? The statistics in Wisconsin may have shown that a classically defined SCID baby was not diagnosed in the pilot, but they identified other forms of immune deficiency that required treatment. And we know that in Oregon, it has been statistically shown that my son has died from not being diagnosed soon enough. I guess that statistic is one up on yours.

As you consider the updated nomination for SCID and other immune deficiencies, please remember that infants like Liam are born every day in the United States and around the world. We have the technology to screen and diagnose and we have a treatment that is amazingly successful. But we have not time to delay further. It may take several years to start screening in all 50 states. How many more stories like Liam's can we bear?

When I learned I could have the opportunity to speak to this committee, I thought what a wonderful way to honor my son's life and death by helping to see universal newborn screening for SCID and other immune deficiencies become a reality. Please help celebrate what would have been Liam's 1st birthday this month and support universal newborn screening for SCID. Thank you for your time.

The Barrett Family
James & Stacey
Alexis, Grace, Rylee & Liam

4. Barbara Ballard
SCID Family Network and Immune Deficiency Foundation
Statement to the HHS Advisory Committee
on Heritable Disorders in Newborns and Children
January 21, 2010

My name is Barbara Ballard. And this Committee has heard me speak before. Many of you may remember, I'm the mother of a child with X-linked SCID. I run a support network for families with SCID. I'm also on the board of trustees for the Immune Deficiency Foundation.

I found it very apropos that we were able to hear this morning a presentation on morality in regards to newborn screening, because I wanted to bring up that subject today myself. It was the philosopher Peter Singer who queried society's morals by asking the question, “If you see a child drowning in a pond and you can save that child without any risk to yourself, other than you would ruin a $200 pair of shoes, would you save that child?” Basically everyone asked that question almost incredulously answered, “Of course.” But when asked if they would write a $200 check to save 100 children, significantly fewer people say they would write such a check.

The human psyche does not grasp the same feeling of loss and grief on a large scale. We cannot feel it viscerally. Even if the loss is of 10 children, we do not feel 10 times the grief and loss we would feel watching one child. We do not even feel it twice as much. In fact, when studied, we learned that the higher the number of children lost, the less we feel it because it no longer is a visceral feeling that you can see and touch and realize. We all need to remember that Liam Barrett was that drowning child. And that you, the members of this Committee, stood on the edge of that pond looking at your shoes.

When you next vote on whether or not to recommend the testing for SCID as a universal newborn test, I want you all to take a good look at your shoes. And I want you to remember Liam Barrett's face. And I want you to hopefully grant him his birthday wish by casting your vote to recommend universal newborn screening for SCID.

Thank you.

5. Sylvia Au, M.S., C.G.C.
Newborn Metabolic Screening Program
Hawaii Department of Health
Statement to the HHS Advisory Committee
on Heritable Disorders in Newborns and Children
January 21, 2010

Good afternoon. I wasn't planning to make public comment. But on some of the discussion that you've had today on newborn screening, I just wanted to really come from a state perspective. I'm speaking on behalf on the Hawaii Department of Health and not the Western States Genetic Services Collaborative.

I think that we really need to make the Secretary recognize that newborn screening programs do the best job that they possibly can. I don't know any newborn screening programs that don't try to do the best job that they can. And I think that some of the things that are happening with newborn screening programs aren't being recognized.

We have a lot of pressure at the state level right now. We have reduced budgets; we have furloughs. You can throw all the money you want to our programs, but we can't hire people. So some of the recommendations to add this disorders, add new programs would be great. Totally support them, love families, want to help them.

But we are really in a situation where we're having a tough time. And you have to recognize the workload of the newborn screening programs. And to say that you can just add a disorder or add a program, it's not that easy. And I come from a state that went from being you know, last in the country at screening two disorders in the mid-90's to screening 32 disorders now. And we're doing two furlough days a month. We've got lots of pressures on us. You can't hire new staff.

So I just want the committee to be sensitive to the newborn screening programs that really work hard to do a good job for their families. And your recommendations are going to impact us, because things like, if you have minimum standards; I spend a lot of time arguing why we pay for certain things. We pay in Hawaii for all the treatment, confirmation. We pay for DNA mutation analysis. And they ask us why we're doing that because that's a lot to pay for.

And if you come up with minimum standards, I mean our administration wants to dive down. So they're going to get rid of all the extra stuff that we do. So you have to be careful for the states that actually do more than we're required to do, because we love our families and want to do good for them.

So you have to make sure that you're politically sensitive to what's really happening at the state level and not dismantle what we have to advocate for every day. So that's all I had to say.

Thank you.

6. Annamarie Saarinen
Parent of a Child with Congenital Heart Disease
Statement to the HHS Advisory Committee
on Heritable Disorders in Newborns and Children
January 21, 2010

Thank you Dr. Howell and Committee. My heart goes out to the families that are here today. And I'm feeling a little challenged in speaking after you to be honest. So bear with me, I'll do the best I can.

The good news is, I came here to sort of lobby a little bit. Put on my lobbying hat and convince all of you how important newborn screening for critical congenital heart disease (CCHD) is. And gratefully, I have to do a little less of that thanks to Dr. Rinaldo's very astute report and to the work that's been done thus far. So I'm grateful there were a lot of head nodding around the table after Piero spoke. Because this is such an entrenched belief for me that this is the right thing to do.

So I'll give you just sort of a little bit of background. I'm the mother of three. I have a real job in public policy, so poor Sharon Terry has had to see me twice during this trip on health IT issues. But my daughter, Eve, was diagnosed at two days old with a severe mitral valve heart defect and an enlarged heart. She was very nearly sent home. Was in complete heart failure at four days old. In other words, she would have never made her one-week well visit. One of many babies I soon found out are in that boat.

One in 100 babies are diagnosed with a heart defect. That is the most common of all birth defects. And building on Dr. Rinaldo's comments, less then a third of these heart defects are diagnosed prenatally. That leaves two-thirds of them that are not. I was in the two-thirds, obviously because I had a daughter diagnosed at two days old. But of these, the data indicates that routine newborn exams fail to detect 25 percent, conservatively; Dr. Rinaldo and some reports go up to 40 or 50 percent, depending on what you're looking at.

So the pediatricians in this room, thank you for your diligence in you know—when you hear that murmur not always saying let's check it again at the one-week well visit. If we have the option to explore further testing, going ahead to do that. Murmurs often indicate the heart defect, but serious defects, many of them don't present with murmur immediately after birth.

And even with a murmur and a careful exam, additional measures can help increase early detection. Pulse oximetry—a simple, noninvasive test, which can be done at an interval of 28 to 48 hours after birth—can detect those otherwise silent heart defects. Pulse oximetry increases the detection of congenital heart disease over exams alone.

The important thing here is that, as with many of the things you look at on this Committee, the earlier a congenital heart defect is detected and treated, the more likely the child will survive and have fewer developmental delays and long-term health complications. A baby coming back to the hospital in heart distress is proven to have an increased chance of death and a worse neurological outcome then those diagnosed before discharge.

Obviously, there are ripple effects on the economy with kids that aren't diagnosed soon enough and come back in that acute situation end up in a longer term care situation. Or if they just don't make it, the families are forced to relocate often for treatment; there are job losses; there's divorce. There are all sorts of horrible things that go along with you know, severe illnesses in children. And I think it's important to think kind of outside just the single case of a child just being sick to what the real impact on society is.

There are many fine institutions in this country that are already screening for congenital heart disease without mandate using pulse oximetry, including Regents Hospital in St. Paul, Mary Bridge Heart Center in Tacoma, and Children's National Medical Center right here in Washington. We are actually in the process in Minnesota of launching a very well-planned pilot study. It's going to be rather brief and rather concise—3,000 babies in about 12 weeks. So compared to the huge numbers that you've seen on some of the other material presented today it's a small group.

But so many pilot tests have been done domestically and around the world that the data is clearly there to help your evidence review board. And hopefully our data coming out of Minnesota will be helpful in that regard as well. And in the fact that it's very current and very well thought through. Our evidence has taken into account many of the existing studies. So we've kind of tried to poke holes in the things that have been problems in other studies.

And we also have been thinking a lot about the number of deliveries outside of major medical centers. I'm a farm girl. A lot of my friends are still in outlying parts of Minnesota. We have a lot of deliveries in our state, as many do, that are outside of major medical centers. So we've been very careful about thinking through what happens with those families if they do indeed test low on a pulse oximeter screening, that they won't be having to wait for the echocardiogram or the echo read so that they can get a quick diagnosis. Not always will there be someone who's maybe well attuned to doing a pediatric echocardiogram. But they do have access to the machinery and an echo technician in the major medical centers, and with the collaboration with the Minnesota Department of Health, these centers have committed to using telemedicine to make sure there are no outstanding wait times for diagnosis so that parents aren't left to worry and wonder whether their child does indeed have a heart issue or something else. I mean perhaps another respiratory or a lung issue which is the other great thing about pulse ox—that it can identify things for these babies outside of CCHD.

So I believe a one-year challenge is an evidence review. Most of the textbooks identify more than 40 different defects. Many cardiologists would not that there are probably more than 100 different variants. Our daughter's was very rare indeed. So many congenital heart problems are different to identify by fetal and neonatal ultrasound. And I think the reach that you'll have in implementing a pulse ox screening will have exponentially greater impact in areas outside of those major medical centers. And hopefully it's going to be a lot easier and actually more cost-effective to implement as a physical screening then even hearing screening was several years back.

I understand the role of this Committee is ensuring that suitable newborn screening tests are developed and safe, effective treatments are available for implementation. Congenital heart disease accounts for the majority of deaths for congenital defects in children—six times more than common then chromosomal abnormalities. By any standard, when we have in 100 kids affected by a defect it's a public health need. In the past three months alone, I personally know of six families have had to bury their babies to undiagnosed heart defects. Eve's surgery happened within about a week of her heart stopping. It was not going to work anymore. I believe she's proof that medical professionals can work their magic on these babies if they are given the opportunity to do so. They need to know there's a problem before they can fix it. So on behalf of the 40,000 U.S. families whose newborn will be diagnosed with heart disease this year, and the 4,000 newborns will not live to see their first birthday, I sincerely thank you for your commitment to the health of newborns and for considering moving forward to the next phase—screening for congenital heart defects.

Thank you.

7. Andrea Williams
Children's Sickle Cell Foundation, Inc.
Statement to the HHS Advisory Committee
on Heritable Disorders in Newborns and Children
January 22, 2010

My comments today are regarding sickle cell carrier trait testing. As a research assistant to Dr. Lakshmanan Krishnamurti, I have been involved with the follow-up of families with children identified as sickle cell trait carriers by the newborn screening program (NBS) since 2005. The program has been successful in providing genetic counseling (using a certified genetic counselor) via phone to more than 97% of those families who were able to be contacted. A much smaller number of them come in for confirmatory testing and further counseling. In 2009, there were approximately 700 children with the sickle cell trait born in Western Pennsylvania. The 17- and 18-year-olds that are leaving high school depending on their birthday and its relationship to September 1992 the start of NBS in Allegheny County may not have been screened via the NBS program.

As the executive director of a community-based organization, I have been afforded the opportunity to collaborate with the sickle cell providers in western Pennsylvania; we have established a community outreach program that focuses on awareness, education and screening in many venues; schools, universities, health fairs, community events, religious organizations and churches. At present, we are establishing collaboration with the blood bank to offer screening with blood donation. I am sure that there are clusters of providers and community efforts across this great nation, but still, there is MORE to be done…

As a parent and consumer advocate, I am compelled to share with you my perspective around the issues surrounding the National Collegiate Athletic Association (NCAA) recommendations for screening athletes for sickle cell trait. I am grateful for the platform that this issue raises for the sickle cell disease community. I stand here advocating for your continued attention to resources around sickle cell trait awareness, genetic counseling and education, proper screening and coordinated follow-up for everyone. A starting point may be to those persons that have been identified through the NBS program as having sickle cell trait carrier status and moving into education and screening for everyone via proper awareness and educational campaigns should be carefully crafted and launched across the nation.

We must remember that there is a growing population that are in and/or entering their childbearing years that are likely to be ignorant of their sickle cell trait carrier status. To neglect to properly design and fund the education, screening and follow-up for everyone is to neglect the next generation of parents who will have children with sickle cell disease that will undoubtedly feel the shock that accompanies the diagnosis when one or both parents lack the knowledge of their sickle cell carrier trait status. They will feel the pain that I felt as a parent when my son was diagnosed at birth, learning later that my (former) husband is a sickle cell trait carrier.

I understand the urgency of this recommendation for screening and more research. It is fitting to screen athletes and educate them of their risk for an adverse event and how to protect themselves. However, I feel constrained to give a voice to all of the other students on college and university campuses that aren't aware of their sickle cell carrier trait status and their possible risk for having a child with SCD. When both partners are sickle cell trait carriers, the risk is 25% for having a child with SCD, 25% the child will be unaffected, and the remaining 50% that the child will have sickle cell trait. These risks are much higher than exist with known risks of having sickle cell trait.

Further, what about the rest of the population? There are thousands that choose to attend two-year colleges and post secondary training or job corps where sports aren't offered? Where is the intervention

for them? What about the working poor who labor in occupations that require only a diploma or GED? Last, but certainly not least and may indeed be the largest population at risk, the unemployed and underemployed; approximately 80% of the families that we have Medicaid as their primary insurance.

My comment is to recommend that a funded system of awareness, education, and screening that is carefully designed for successful implementation to include the athlete as a part of the message and service for everyone. I understand the complexity of the system that I propose and the history of what has been attempted in the past. The time is now and here is the reason… we have never before had the technology to systemically bring about awareness and education with screening, knowledge of the proper screening processes, protections that of (GINA) the Genetic Information Nondiscrimination Act – which we know is law! This system will become the model for other genetic diseases as we move forward. I am confident that you will make recommendations that give a voice to everyone and serve to provide for and protect us all for generations to come.

Thank you for your time.

Andrea M. Williams, BA
Executive Director

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8. Micki Gartzke
VP, Save Babies Through Screening
& Parent of a Child Who Died from Krabbe Disease
Statement to the HHS Advisory Committee
on Heritable Disorders in Newborns and Children
January 22, 2010

Hi. As always, thank you very much for the opportunity to present public comments to the Committee and the chair Dr. Howell, Dr. Lloyd-Puryear, and Dr. van Dyck. I will be very expeditious.

Thank you for nominating the SCID group of disorders to be added to the core panel. It's what the consumers are looking for, and I think you deserve to be applauded for the work that you've done to accomplish that.

My second comment is you earlier today asked for possibilities of recommendations of who might else be invited to sit at the table. And I've thought for a while as a consumer, that with all the education that the genetic counselors do, that they might be given that opportunity with their valuable service that they provide to consumers.

So thanks for adding SCID.

9. Susan Gallagher
Parent of a Child with Phenylketonuria
Letter e-mailed to the HHS Advisory Committee
on Heritable Disorders in Newborns and Children
January 21, 2010

First and foremost, thank you for all your efforts! I understand a significant exposure and funds have been allocated to ACHDNC for the 2010 budget.

I am writing on behalf of my son, DECLAN, who will be turning 2 on Jan. 29th; he has PKU. Please continue to support the efforts of our newly found National Alliance: NPKUA in their research/ scholarship proposals. Also, our legislation for MEDICAL FOODS BILL S2766 currently in the House.

Thank you for your time and continued work on behalf of All of our Heritable Disorder infants/children, and our PKU Community.

Sincerely,
Susan Gallagher
Wilmington, Delaware