



Issues in Newborn Genetic Screening in California

On March 11, 2004, in Sacramento, national and local scientific experts, parent advocates, policymakers, afflicted children, biotechnology industry representatives and public health professionals convened. Organized by the Public Health Institute, and co-sponsored by the US Centers for Disease Control and Prevention and the March of Dimes, the topic of conversation was newborn genetic screening in California.

Speakers shared perspectives on the importance of expanding the number of screening tests performed on California newborns. Experts from Atlanta, Texas, New Jersey and across California shared information about the need for such programs, their costs and potential for cost-savings over the lives of children with inborn metabolism errors, the comparison between California's program scope and those of other states, and the real-life impact on children and their families of having and not having functional screening programs in place.

Symposium binders provided detailed background material, supplemented by the speakers' excellent presentations, all of which were taped. From the transcripts of those tapes, we are delighted to provide highlights of the California Policymakers' Symposium on Newborn Genetic Screening.

One immediate result of the Symposium was the passage of SB 142 by the Assembly Health Committee, a bill that would mandate newborn screening for all detectable metabolic and genetic disorders. The bill was authored by Senators Dede Alpert (a speaker at the Symposium), Jackie Speier and Sheila Kuehl. If passed, the bill would make permanent California's 18-month tandem mass spectrometry pilot and would bring California's newborn screening program into line with 42 other states by July 1, 2005.

For further information, see box on page 8.

Financial support graciously provided by Perkin-Elmer, Inc., Bio-Rad and Pediatrix.

We save anywhere from \$2 to \$4 for every dollar we invest in screening

— George Cunningham, MPH, MD

EXECUTIVE SUMMARY

Screening Program Overview

A comprehensive newborn screening program encompasses:

- Testing that is fast, flexible, confidential and that produces few false positives.
- Follow-up with families to ensure compliance with interventions and treatment.
- Education for physicians, nurses, hospital staffs and families, about the need for, and availability of, newborn screening.
- The production of statistics for ongoing research and improvement.

Newborn screening is a complex system of testing, evaluation and treatment, dependent on the dedication of the persons working within the system. It includes primary health care professionals, laboratory personnel, administrators, specialty care centers, payors, family members and policymakers.

Mandatory screening varies by state, with the following number of states mandating it for:

Galactosemia, hypothyroidism and PKU	50
Maple syrup urine disease	31
Sickle cell	47
Homocystinuria	28
Congenital adrenalhyperplasia	36
MCAD	26
Biotinidase deficiency	31
Cystic fibrosis	6

California's Newborn Screening Program

California uses a central laboratory system for newborn screening and five Area Service Centers to follow up with the families. At the Stanford ASC, the model is: Find the babies. Treat the babies. Save the babies.

California screens for PKU (since 1966); hypothyroidism and galactosemia (1980); and sickle cell disease and other hemoglobin disorders (1990).

California conducted a pilot project using tandem mass spectrometry* from January 2002 to June 2003. Approximately 400,000 children have been born in California since the pilot program ended in 2003. Based on the population incidences, about 25 of those children have MCAD deficiency.

California is the only tandem mass spectrometry pilot study worldwide which has not been converted into a permanent newborn screening program.

Since 1980, California has screened 12 million newborns, 7,000 of whom were put into treatment with a significant clinical condition as a result of this effort.

Ethnic mix is a challenge to screening in California. Tests developed in the non-Anglo population or at East Coast medical centers may not have the same results here. Our Asian population has a lot of hemoglobin-H; our black population has a lot of sickle cell; our Hispanic population a lot of hypothyroidism.

For many decades, California was considered the leader in newborn screening. Because of budget constraints, California has continued to screen for only four conditions and has fallen abysmally behind other states.



Lisa and Gonzo Jaquez

Parent Advocates, March of Dimes

Lisa and Gonzo Jaquez are the parents of Ysabel, a four-year-old whose L-CHAD was not identified at birth because California did not do tandem mass spectrometry screening when she was born.

Lisa: At birth, Ysabel was perfect. She had good lungs, a good Apgar score and the sweetest set of lips. Three days later, she almost died in my husband's arms. Her heart rate went down to 20, CPR was done twice and she was intubated. Ysabel's diagnosis came at the expense of her brain and almost death. Ysabel's NICU stay at Stanford ran \$10,000 to \$14,000 per day, and she was there for 30 days. There were follow-up neurology procedures and physical therapy. We had a social worker from the Golden Gate Regional Center and a high-risk infant nurse. She attends a special-needs preschool and will need speech therapy. Consider the number of children who could have been screened for the cost of Ysabel's special care.

Gonzo: Children who haven't been screened are, for lack of a better term, ticking time bombs waiting to explode. And when the explosion happens and the dust settles, the questions will be: Have they survived? What quality of life will they have? Will they be able to make a difference? Will they wonder if anyone will hear them?

The Economics of Screening

Newborn screening is one of the largest disease prevention programs in the U.S., reaching approximately four million babies each year. In California, about 530,000 infants are born annually—one in eight of all the children born in the U.S. Virtually all are screened for certain birth defects. California's genetic disease testing program budget, which includes prenatal screening, is about \$67 million annually. Since 1980, over 7,000 cases of metabolic disorders have been detected.

California's screening program costs \$62,000 per case detected. This compares to the estimated \$1 million lifetime cost if disorders such as these are not identified early. We save \$2 to \$4 for every dollar we invest in screening.

Screening fees are attached to the hospital bill, and money is collect directly from the hospital. The only tax money that goes into it is the money that pays for the Medicaid babies. The tandem mass spectrometry screening program could be implemented by charging a \$20 fee.

** Mass spectrometry is a technology that analyzes compounds by electronically "weighing" the molecules in a sample to determine how much of a single compound is present. Tandem mass spectrometry uses two chambers, first to sort and weigh the entire sample, then to analyze separate pieces of the sample in the second chamber. Thus, a great number of substances can be screened using a single sample.*

INTRODUCTION

Jose Cordero, MD, MPH

Assistant US Surgeon General & Director,

National Center on Birth Defects and Developmental Disabilities

One area being looked at by the Centers for Disease Control in our Future Initiatives is prevention at different stages of the life cycle and in infancy. Because the newborn period is so important, it is critical to recognize what conditions a child may have that, if gone undetected, may have an impact. PKU and newborn screening are primary examples of the importance of early recognition and treatment. Today, our discussion is how to ensure that children grow healthy and have optimal development.

WHY IS COMPREHENSIVE NEWBORN SCREENING IMPORTANT?

Joe Valenzano, Jr.

President, CEO & Publisher, Exceptional Parent

We would never put our children in harm's way, so it is unclear why there is debate about newborn screening. Today's technology can screen for more than 55 inborn errors of metabolism, yet there is wide disparity amongst the states in what is screened and the level of communication provided to families and physicians.



Why screen?

First of all, screening is the right thing to do. It should be done for all known conditions, no exceptions, no more debates about how families can deal with information or about the effectiveness of interventions. Identifying disease states earlier provides a greater opportunity for positive outcomes. That is absolute fact.

Screening is economically justified. Every study shows that newborn screening is cost effective and provides a return on investment, especially when the cost savings for treating children with profound developmental delays and profound disorders are factored in.

What is needed?

Beyond the capabilities of laboratories to screen efficiently and effectively for these disorders, we need a system*

- Whose speed and turnaround time for identifying these disorders is unquestioned and unparalleled.
- That ensures every family is informed about the availability of newborn screening. The literature says that 12 states let families know that additional expanded screening is available. Of 250 expectant mothers we interviewed in the Northeastern US, not one had been informed of newborn screening.
- That has few false positives and maintains confidentiality.
- That is flexible to add additional screening capabilities, such as for lysosomal storage disorders.
- That believes and acts on the idea that children are our future and that we need to keep them out of harm's way.
- That provides the ability for quality research on how we are doing and what we can do better.

Finance and business executives ask three questions in every investment decision:

- **Is it real?** Yes, newborn screening is real.
- **Can we win?** What is winning? What is the value of a life? Of a thousand lives? What's the value of identifying disease states earlier?
- **Is it worth it?** Measure the costs associated with caring for one child who has not been diagnosed with gluteric acidemia or any other disorder against the investment needed to diagnose that child—there is the answer in terms of return on investment.

* Editor's note: Because many of the elements Mr. Valenzano listed are dealt with by other speakers, this list has been edited to eliminate repetition.

California is the only tandem mass spectrometry pilot study in the world which has not been converted into a permanent newborn screening program.

— Gregory Enns, MD

INTRODUCTION TO NEWBORN SCREENING

Gregory Enns, MD

Director, Biochemical Genetics Program,
Division of Medical Genetics, Stanford University

What is screening?

The American Academy of Pediatrics defines screening as: "The early detection of conditions for which early and timely interventions can lead to the elimination or reduction of associated mortality, morbidity and disabilities."

Newborn screening is far more than a simple blood test. It is an integrated program with a large number of components:

- The screen itself.
- Follow-up with the babies and children.
- Diagnosis.
- Treatment and management of the disorder.
- Evaluation and education of the community, the family and other caretakers.

How is screening accomplished?

Every child born in California is mandated to have a heel stick test. Blood is taken 12-to-24 hours after birth and spotted on a collection card, which is sent to a State-approved laboratory to be screened for four conditions: phenylketonuria, hypothyroidism, galactosemia and for hemoglobin disorders, like sickle cell disease.

Testing takes place up and down the state at regional labs. The central State laboratory coordinates the accumulation of the data from the tests. It also:

- Establishes uniform methodologies.
- Ensures quality results by training, proficiency testing and inspection.
- Manages inventory and utilization.
- Performs emergency testing.
- Prepares service contracts.

Until recently, four separate machines screened for the four conditions. Now, thanks to the tandem mass spectrometer, we can screen for a large number of conditions, virtually simultaneously. Most programs that use tandem mass spectrometry screen for 15-to-30 different biochemical disorders.

It requires tremendous expertise to know what the data show, how to interpret them, where to set cutoffs and how to do this for a given population. Today, California has a great wealth of information and expertise from its pilot program.

Once a screen is performed and the State has come up with a result that looks like a positive, California has five organizations called Area Service Centers (ASCs) to do the follow-up with families. At the Stanford ASC, our model is: Find the babies. Treat the babies. Save the babies.

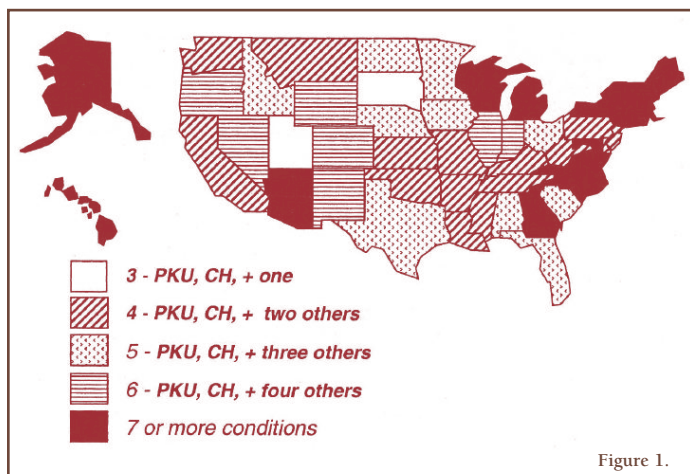
The ASCs are staffed by nurses and community liaisons. Administrators track down where the baby was born, talk to the family and the pediatrician. They coordinate the specialists who take care of these kids, and make sure that follow up, re-testing and treatment happen in a timely manner.

What diseases are identified by screening?

In California, the program started in 1966 with PKU screening. Hypothyroidism and galactosemia were added in 1980. Sickle cell disease and other hemoglobin disorders like thalassemia were added in 1990.

More than 500,000 children are born in California each year. That is about one-eighth of the babies born in the United States. More than 99 percent of those California newborns are screened.

This map (Figure 1) shows that states march to their own drummers in what they test, how and what is done afterwards. Depending on where your child is born, and where your family might have moved, one child in your family may have been screened one way and another child, differently.



The disorders detected by tandem mass technology are those that you wouldn't necessarily know a child has. A child can appear completely normal for days, hours or years. The medical literature reports adults aged 40 to 70 with a metabolism disorder, having never presented symptoms until they had a severe flu or other infection. Suddenly, they become mentally disabled or die from the underlying condition.

Most of these disorders can be treated or ameliorated by simple dietary therapies. Individually, these disorders are quite rare, but in aggregate, about one in 5,000 children is detected using the current tandem mass spectrometry screening.

What happens after the screen?

California has detected thousands of children with inborn errors of metabolism and other congenital disorders that can severely affect them. Approximately 400,000 children have been born in California since the pilot program stopped last year. Based on the population incidences, about 25 kids with MCAD deficiency have already been born in California.

You can't put a cost on a child's life, but every cost-effectiveness paper demonstrates the clear benefit and cost savings of doing newborn screening by tandem mass spectrometry. California is the only tandem mass spectrometry pilot study worldwide which has not been converted into a permanent newborn screening program.

CALIFORNIA NEWBORN SCREENING DATA

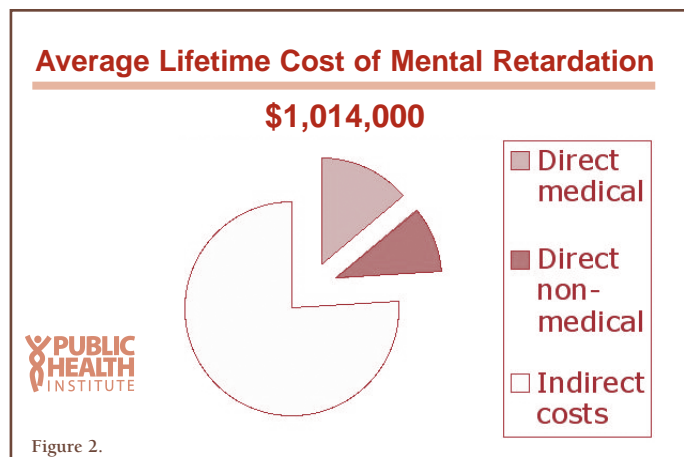
Carmen Rita Nevarez, MD, MPH
Medical Director & VP of External Relations, Public Health Institute

This is information collected in California during fiscal years 2001/02 and 2002/03.

Latino	509,086
Asian	93,060
Other	71,137
White	288,558
Black	56,432
Unknown	14,467
Total	1,032,740

In terms of ethnicity, approximately half of California births are Latino and about half of the babies screened were Latino. A look at the statistics shows that all racial and ethnic groups were screened.

In those 24 months, 832 disorders were detected. Knowing the number of babies identified with disorders, which can then be treated, starts to give us an indication of the cost benefit of newborn screening. In January 2004, the CDC estimated the economic costs associated with a number of developmental disorders. To care for someone with mental retardation over the course of a lifetime costs more than \$1 million, taking into consideration direct medical care, indirect non-medical care, such as education and indirect costs, including loss of income. (See Figure 2)



ACCOMPLISHMENTS OF THE CALIFORNIA PROGRAM

George Cunningham, MPH, MD
Chief, California Genetic Disease Branch

California started screening for PKU in 1966, sending the tests to 175 private clinical laboratories. This proved to be an extremely cumbersome and expensive system, and 15 cases of PKU were missed. In 1980, the program was centralized to establish standards, to coordinate laboratory quality assurance and to ensure effective and immediate follow-up.

Since 1980, we have screened 12 million newborns. We have identified 7,000 babies who have been put into treatment with a significant clinical condition:

- More than 444 babies with PKU.
- 4,254 with hypothyroidism.
- 153 with galactosemia.
- 1,836 babies with sickle cell disease.
- 269 with hemoglobin H disease.

In California, one problem with screening is the ethnic mixture. Tests developed in the non-Anglo population or at East Coast medical centers may not have the same results in California. For example, our Asian population has a lot of hemoglobin-H; our black population has a lot of sickle cell; our Hispanic population has a lot of hypothyroidism.

Our screening program costs \$62,000 per case detected. Compare that to the estimated \$1 million lifetime cost when disorders such as these are not identified early. **We save anywhere from \$2 to \$4 for every dollar we invest in screening.** That's a very cost-beneficial program.

In 2001, the Legislature approved a bill authored by Senator Sheila Kuehl, authorizing \$3.9 million for a tandem mass spectrometry screening pilot, which ran from January 7, 2002 through June 13, 2003. It was a voluntary program and about half the state's hospitals decided not to participate, primarily due to concerns about individual IRB* approval and additional nursing time costs. As a result, we ended up with 402,566 babies who were not screened.

Amongst the screened babies:

- We had 705 positive results, some of them normal variations.
- We referred 468 to one of California's 14 metabolic centers. Of those, 332 were resolved as normal. We're waiting for results on the rest.
- We diagnosed 51 cases of disorder.

The net result was that one out of 6,500 newborns had one of these conditions.

* Institutional Review Board – a panel required by law to assure no harm comes to participants in research or pilot programs.



Malathy Ramanujam

Parent Advocate, Southern California
PKU Network

Ms. Ramanujam's 21-year-old son Bhirat is afflicted with homocystinuria, which was not detected early. To help her son, she continued her education and expects to be awarded a Master's of Science degree in Nutritional Science in the summer of 2004. She has developed various low-protein recipes for metabolic disorders and has conducted numerous low-protein cooking workshops.

At age two, following a bout of severe seizures, Bhirat was admitted to the hospital in London, where it took 45 days before he was diagnosed with homocystinuria and treatment was started. If Bhirat's condition had been caught as soon as he was born, he wouldn't have had to go through all the health complications before and since his diagnosis.

Because he was diagnosed at age two, his brain was already affected. Without realizing it, because of our vegetarian diet, we were partially treating him before he was diagnosed. The diet helps and he could have led a normal life, if he was diagnosed at birth. Luckily for us, even though my son had seizures and more complications, he is able to function pretty well.

Nobody is born perfect. I'm not saying that if babies are screened at birth, they will not have any problems. I'm just saying they don't have a chance to a better life. Everybody has a right to a better life.

Of these 51 cases:

- 13 were medium chain fatty oxidation disorder, which constitutes 3 percent of Sudden Infant Deaths.
- 8 were short chain acylCoAdehydrogenase deficiency.
- 11 were methylmalonic and propionic aciduria, and a variety of other disorders.

Of the babies not screened in the pilot program, 11 cases were picked up clinically by physicians. We would expect that number to be 50, an indication that these disorders are not being identified by physicians in the absence of screening.

Fees for newborn screening are attached to the hospital bill and collected from the hospital. The only tax money that goes into screening pays for the Medicaid babies (approximately 220,000 annually). We estimate a tandem mass spectrometry screening program could be implemented by charging a \$20 fee.

Supported by a Health Resources and Services Administration (HRSA) grant through the Public Health Institute, our economic analysis shows the cost to detect a single case was \$85,000.



In aggregate, about one in 5,000 children is detected using the current tandem mass spec screening.

— Gregory Enns, MD

Audience Question

Of all the disorders screened for, how many are treatable?

Dr. Cunningham

The majority are treatable. The higher incidence disorders screened by tandem mass are treatable: MCAD homocystinuria, maple syrup urine disease. Even for the rare conditions without treatments,



Carol Cook

Parent Advocate, Southern California
PKU Network

Carol Cook is the mother of three sons, two of whom have PKU. Her son Michael's test was performed too early and did not pick up the condition. Her next son, Jack, was diagnosed appropriately and functions at a neurologically higher level.

When Michael was born, we thought we had another bouncing baby boy. I noticed that at about three months, Michael wasn't holding his head up. At six months, he wasn't rolling over. At Michael's one-year visit, I insisted that some kind of workup be done. Michael's two-year visit was with a new pediatrician who sent us to a neurologist, who said Michael had cerebral palsy.

When Michael was five, we went to a new neurologist who asked what became of the metabolic screening mentioned in the record. I said I didn't remember that test. He said, "Well, it's just a urine test. Do you mind going down to the lab?" We had poked and prodded and put this child through the wringer, and he was asking us if minded a urine test?

A month later, Michael was diagnosed with PKU. I asked how that could happen. I have since learned that it was convenient for the staff to do the blood draws between midnight and 6:00 a.m. when the babies were in the nursery, too soon for Michael's test results to be accurate.

Things that we take for granted—sports, birthday parties, sleepovers—don't happen with my son Michael. I was three months pregnant with Jack when I found out Michael had PKU. Jack also has PKU. But I tell you, Jack was the greatest gift God ever gave Michael, because Jack loves Michael and will be there for him for the rest of his life, even when I can't be.

we're never going to have treatments unless we find people with these disorders and accumulate information about them.

Dr. Enns

As a critical care physician, standing at the bedside of a baby who may be dying of an untreatable disorder, it really is important to know the cause, to minimize guilt or remorse, to reassure parents that it's not their fault. That's a very important contribution to clinical care.

A MODEL PROGRAM FOR CALIFORNIA

Senator Dede Alpert

39th Senatorial District (San Diego);

Chair, Senate Select Committee on Genetics and Genetic Testing

Newborn screening is one of the largest disease prevention programs in the US, reaching approximately four million babies each year. In California, about 530,000 infants are born annually—one in eight of all the children born in the US. Virtually all are screened for certain birth defects. California's genetic disease testing program budget, which includes newborn and prenatal screening, is about \$67 million annually. Since 1980, over 7,000 cases of metabolic disorders have been detected.

California's pilot program in tandem mass spectrometry screening ended last June due to lack of funding. Along with Senator Sheila Kuehl, who authored the legislation authorizing the pilot program, I am committed to seeing what we can do to work with the Administration this year and will offer legislation to expand newborn screening.

For many decades, California was considered the leader in newborn screening. And the work that our people do is exemplary. But the truth is we're now only screening for four conditions. We have fallen abysmally behind.

A REVIEW OF THE NATION'S PROGRAMS

Brad Therrell, PhD

Director, National Newborn Screening and Genetics Resource Center, University of Texas, Austin

Organizational Features

Newborn screening can be defined as an essential public health program that prevents catastrophic health consequences through early detection, diagnosis, and treatment.

It's a system that includes primary health care professionals, laboratory personnel, administrators, specialty care centers, payors, family members, policymakers and other interested people like Senator Alpert.



Every state has a law giving it the authority to do newborn screening. Three programs (Maryland, Wyoming and Washington, DC) require informed consent. Everybody else does these programs under an informed dissent model: you can opt out, but you don't have to opt in.

Every state screens for PKU, hypothyroidism, and galactosemia. Hemoglobinopathies are screened for in almost every state. Eight states mandate two screens on every baby—one at birth in the hospital and one at two weeks of age. In these states, they actually report finding some cases of hypothyroidism, CAH and other disorders on the second test, when the first test was normal.

Some states combine their laboratory testing into one centralized laboratory. Others also use commercial laboratories. In California, eight private laboratories contract with the State to do testing.

Screening Choices

The number of tests done varies across the country. (See Figure 3) Eight states test for four disorders, including California. Texas is among the four states that test for five disorders. Combining the births in Texas and California, you get more than 20 percent of the births in the country. That's 900,000 babies.

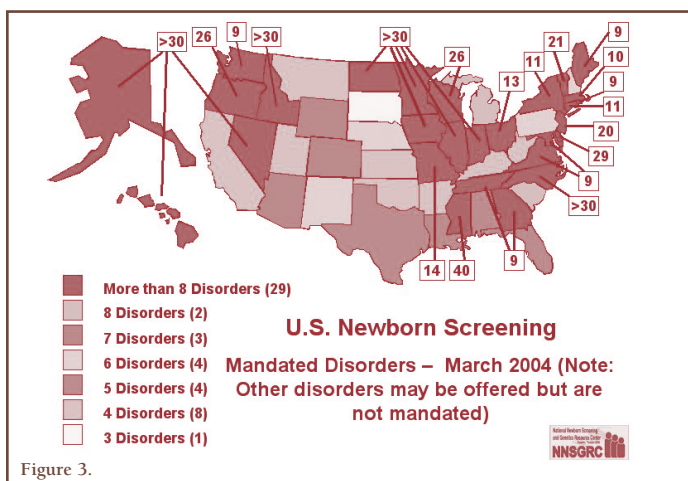


Figure 3.

Here's a recap of how many states mandate screening for certain disorders.

Galactosemia, hypothyroidism and PKU	50
Maple syrup urine disease	31
Sickle cell	47
Homocystinuria	28
Congenital adrenalhyperplasia	36
MCAD	26
Biotinidase deficiency	31
Cystic fibrosis	6

Every state, plus Washington, DC, has some sort of tandem mass spectrometry program, whether mandatory, optional or pilot. More than half have mandated tandem mass spectrometry programs.

State legislators decide what is screened and how it is done, guided by public, professional and political interest, health care cost savings, and sometimes, scientific evidence. A 1968 World Health Organization paper is often used to define the criteria for what should be screened. (Figure 4)

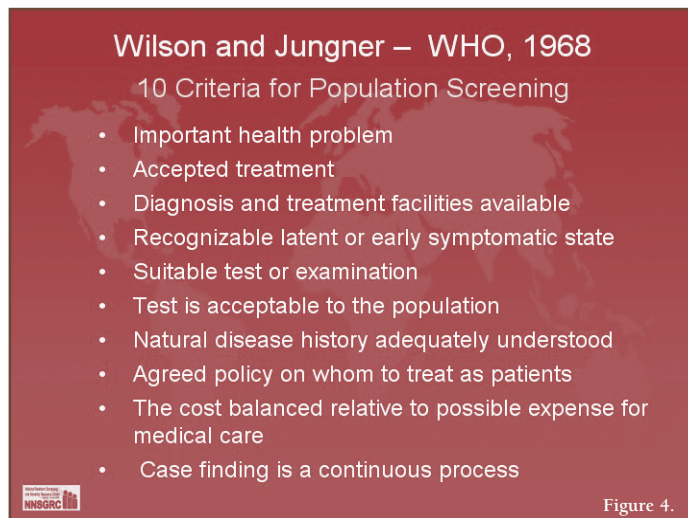


Figure 4.

The HRSA and the American College of Medical Genetics are working out national recommendations on how states should consider which disorders to add to their program. Their recommendations are expected in June and are intended to give states guidance and to eliminate some of the inequities in screening programs.

Kelly Leight, JD

Director, CARES Foundation

Our stories of pain, suffering, mental retardation and death are particularly poignant, considering that the devastation could have been prevented with an inexpensive test from a simple drop of blood. We must reach out to administrators, legislators, service providers and health care professionals, people who need to hear us and to listen to our recommendations. We need to present a single message to be effective. That message is universal comprehensive newborn screening.

We have to work with:

- The March of Dimes and genetic disease support groups.
- Medical societies, like the American Academy of Pediatrics, the American College of Obstetrics and Gynecology and the Pediatric Endocrine Nurses Society.
- Groups that care about children's health issues, such as the Junior League.
- Community groups: the PTA, School Nurses Association, religious groups.

Here are a few techniques for reaching out:

- Use e-mail and the Internet to reach targeted and mass audiences.

- Ask people to do something: contact their legislator or make a phone call.
- Drop in on your legislators. When people visit, they listen.
- Use the media. Call your local papers, organize press conferences.
- Be creative. Organize carpools to visit legislators' offices. Have your kids and local organizations make cards and send them to legislators.
- Use disease support groups in e-mail and letter-writing campaigns.

Our pink postcard campaign here in California asks people to sign and send pre-printed postcards to Governor Schwarzenegger that say: "You're doing a not-so-great job with newborn screening. You're not screening for enough. Babies are dying. Screen our babies. Save our babies." Each post card—8,000 so far—represents a constituent.

I cannot emphasize enough the power of one. One person can change the world. Be the squeaky wheel. Hopefully, the State will listen and do the right thing.

Senator Alpert

With tandem mass spectrometry, is there any reason to do only 10 screens, or is it just as cost effective to do 30?

Dr. Cunningham

Our position is that if you buy the machines and run them, you should run them for as many tests as possible. The argument that's often used against screening for more disorders is that some of these disorders are not treatable.

Along those same lines even without tandem mass spectrometry screening, we can screen for cystic fibrosis on the same machine that we use for hypothyroidism, by adding a DNA test for a cystic fibrosis mutation.

Senator Alpert

Screening for untreatable disorders has value too, because that may prompt people to make different choices in their future family planning. It provides a knowledge base.

Audience Question

My son was born in 1994. He was screened and missed in the follow-up. He gets an enormous amount of services from the regional center. Just one of his services costs over \$9,500 a month. Over a year, that's \$114,000.

Dr. Cunningham

Medi-Cal is funded by 50 percent matching federal funds and 50 percent General Funds. The Medi-Cal share for tandem mass screening would be \$20 per baby, or \$2 million. Medi-Cal will save four times as much over the course of the lifetime of what

we detect in 30 years. Therefore, there is no net cost to the state. If we delay for a year or two, the money that is put out in long-term care for those kids born during those years will evaporate most of the proposed budget savings. This should be viewed legitimately as part of our long-range debt reduction.

Speaker Biographies

Senator Dede Alpert represents the 39th Senatorial District in San Diego, after having served three terms in the California State Assembly. She chairs the Senate Select Committee on Genetics and Genetic Testing.

Jose Cordero, MD, MPH, is the Director of the National Center of Birth Defects and Developmental Disabilities at the Centers for Disease Control and Prevention and is Assistant Surgeon General of the U.S. Public Health Service.

George Cunningham, MD, MPH, is Chief of the Genetic Disease Branch of the California Department of Health Services. A leader in major genetic organizations, he currently serves on the Advisory Group for the American College of Medical Genetics and FDA panel on genetics testing.

Gregory Enns, MD, directs the Biochemical Genetics Program at Stanford University, where he is an Assistant Professor of pediatrics. He co-directs the UCSF Stanford Lysosomal Disease Center and is Director of the Metabolic Special Care Center at Lucile Packard Children's Hospital.

Kelly Leight, JD is the founder and Executive Director of the Congenital Adrenal Hyperplasia Research, Education and Support (CARES) Foundation. The mother of a daughter with CAH, she serves on the Public Affairs Committee of the March of Dimes in New Jersey.

Carmen Rita Nevarez, MD, MPH, is the Medical Director and Vice President of External Relations of the Public Health Institute. A practicing physician, she previously served as Director of the Department of Health and Human Services for the City of Berkeley, California.

Brad Therrell, PhD is the Director of the National Newborn Screening and Genetics Resource Center, based at the University of Texas Health Sciences Center in Austin. He served as President of the International Society for Neonatal Screening and directed the Texas Department of Health Services' Chemical Services Division.

Joe Valenzano, Jr. is CEO and President of EP Global Communications, and publisher of Exceptional Parent magazine. He chairs the Advisory Board of the American Academy of Developmental Medicine and Dentistry.



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