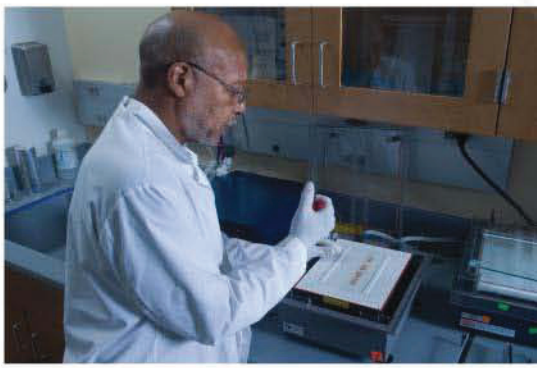
Newborn Screening

LABORATORY BULLETIN



Newborn Screening Laboratory Bulletin

October 2008

U.S. Department of Health and Human Services
Centers for Disease Control and Prevention
National Center for Environmental Health

Centers for Disease Control and Prevention
National Center for Environmental Health
Division of Laboratory Sciences
Atlanta, Georgia 30341-3724

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The Centers for Disease Control and Prevention (CDC) protects people's health and safety by preventing and controlling diseases and injuries; enhances health decisions by providing credible information on critical health issues; and promotes healthy living through strong partnerships with local, national, and international organizations.




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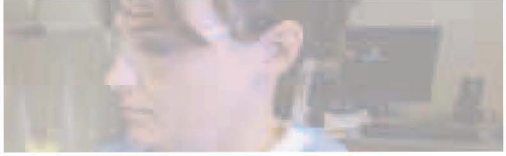
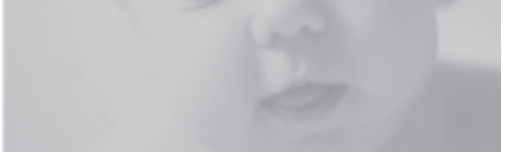




I. Newborn Screening

Overview of Newborn Screening

Newborn screening is one of the nation's most successful public health programs (1). Newborn screening programs test babies for disorders that are often not apparent at birth. Such disorders may be inherited, infectious, or caused by a medical problem of the mother. If these disorders are not detected and treated soon after birth, they may cause mental retardation, severe illness, or premature death. More than 4 million newborns are screened annually in the United States, and thousands of infants are rescued from disability and death.

Newborn screening begins within 24 to 48 hours of a child's birth when a few drops of blood are obtained from a heel stick. The blood spots are sent to a laboratory that is a part of the state or territorial public health department. The spots are analyzed by several different laboratory methods to test for biochemical and genetic markers that reveal hidden congenital (present at birth) disorders. If such markers are found, the newborn screening follow-up program notifies the parents and physicians so that the baby can receive immediate attention. Follow-up programs arrange for diagnostic tests to confirm the newborn screening results. Follow-up programs also refer the child to a treatment center to provide access to the essential medical services needed to minimize the effects of the underlying disorder.

Newborn screening offers life without physical disability

Photo courtesy of Dr. Holmes Morton

The three children in the photograph all have an inherited metabolic disorder called glutaric aciduria type I, which occurs when the body is unable to degrade certain amino acids (the building blocks of proteins) completely. Excessive levels of organic acids accumulate in the brain and cause degeneration in a region of the brain that controls movement (2). In the past, children with brain damage, like the boy in the picture, were often said to have cerebral palsy. He was born in a hospital where newborn screening for glutaric aciduria was not being done. The sisters (not his siblings) on each side of him were tested for glutaric aciduria as part of a regional newborn screening program to develop new methods to diagnose and treat the disorder. Early detection and intervention, which included a special diet and intravenous glucose treatments, prevented the sisters from becoming disabled. The different health outcomes for these children and reports about success in identifying and treating glutaric aciduria throughout the United States and Europe have led to widespread screening of newborns for this treatable disorder.

II. CDC's Laboratory Role in Newborn Screening

Overview of the Newborn Screening Quality Assurance Program (NSQAP)

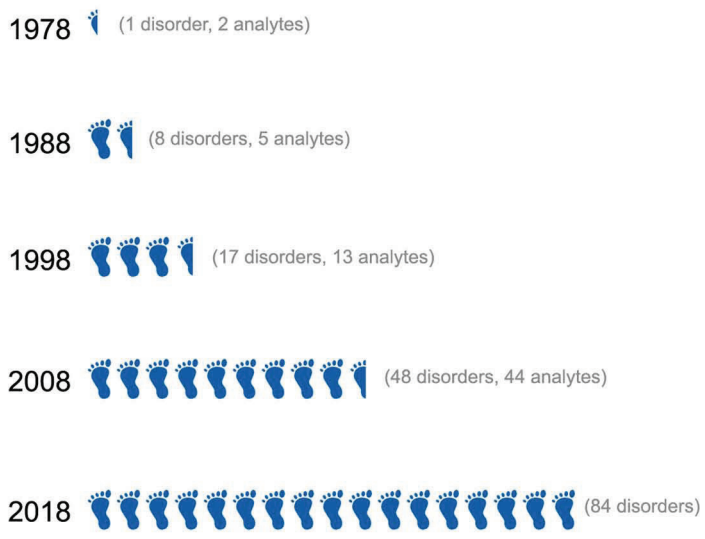
In 1978, CDC established the Newborn Screening Quality Assurance Program (NSQAP) to enhance and maintain the quality of newborn screening tests performed in the United States. NSQAP—housed in CDC's Environmental Health Laboratory—has grown to become the only comprehensive program in the world devoted to quality assurance of newborn screening tests. Since its inception, NSQAP has steadily added disorders and analytes* (Figs. 1 and 2) to the program and continues to expand the program.† NSQAP provides training, consultation, proficiency testing, guidelines, and reference materials to state public health laboratories and other laboratories responsible for newborn screening in the United States and in several other countries. Because of NSQAP, parents and doctors in the United States can trust the results of newborn screening tests.

Figure 1
Sample listing of disorders added to NSQAP

- 1978 Congenital Hypothyroidism
- 1980 Phenylketonuria
- 1988 Galactosemia & HIV Seroprevalence
- 1990 Congenital Adrenal Hyperplasia
- 1991 Sickle Cell Disorders
- 1992 Maple Syrup Urine Disease
- 1995 Homocystinuria
- 1997 Biotinidase
- 2000 Fatty Acid Oxidation & Organic Acid Disorders
- 2002 Cystic Fibrosis/IRT & Diabetes (type 1)
- 2005 Toxoplasmosis
- 2006 2nd Tier Congenital Adrenal Hyperplasia
- 2007 Cystic Fibrosis DNA Mutation Panel
- 2008 Succinylacetone (Tyrosinemia type 1)

Future Years: Lysosomal Storage Disorders, Severe Combined Immune Deficiency, Wilson's Disease, Cytomegalovirus, and others.

Figure 2
Growth of NSQAP: Disorders (and analytes) in NSQAP



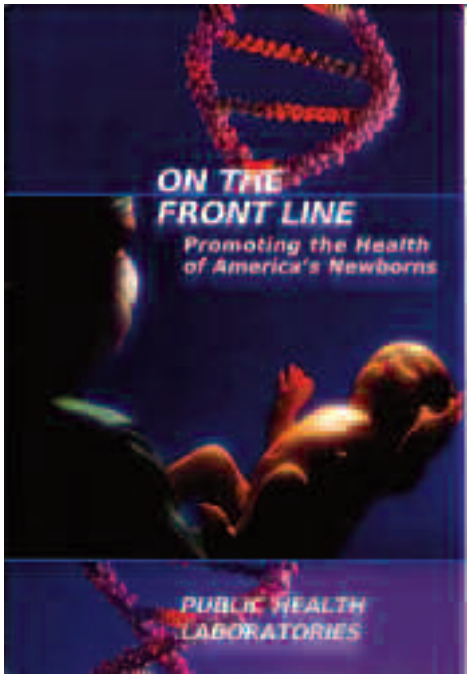
**An analyte is a substance that can be measured in the laboratory, and its presence or absence can show abnormal processes resulting in disease. The number of analytes and the number of disorders in newborn screening tests are not identical numbers. The newborn screening tests conducted in laboratories look for analytes that are associated with disorders. Some analytes identify more than one disorder, and some disorders are related to more than one analyte.*

†The American College of Medical Genetics (ACMG) recommends that states conduct newborn screening tests for a core panel of 29 disorders (including hearing screening, which does not routinely involve blood spot analysis). ACMG has noted that screening for the core panel will provide data that enable states to include a secondary set of 25 disorders. CDC's goal is to include primary analytes for the 53 disorders that can be screened using dried blood spots in NSQAP so that it can help the state newborn screening programs meet the ACMG recommendations with confidence and accuracy. NSQAP is currently at 48 disorders plus toxoplasmosis and HIV.

NSQAP's Partnership with Laboratories

As NSQAP has developed, so have its relationships with public health partners. One of NSQAP's most important partners is the Association of Public Health Laboratories (APHL), which serves as a dynamic interface between CDC and local, state, and territorial public health laboratories. For the past 30 years, APHL has worked closely with NSQAP to assure the highest standards of performance for newborn screening nationwide for public and private laboratories.

Through its Newborn Screening and Genetics in Public Health Committee, APHL is involved in a broad range of issues—including training in laboratory methods using advanced technology, development of policy statements on newborn screening issues, and contingency planning for continued newborn screening in the event of a disaster or other public health emergency. APHL promotes the scientific and technologic expertise of NSQAP to public health officials at the state and federal levels. APHL also provides valuable strategic guidance and expertise to NSQAP. With APHL's assistance, NSQAP is recognized worldwide and serves as a model program of quality assurance for newborn screening for many other countries.



APHL ASSOCIATION OF PUBLIC HEALTH LABORATORIES

Preventing Life-Long Disability

Tayla Cunzenheim

The birth of Tayla Cunzenheim in Wisconsin on December 23, 2002, was an early Christmas present for her happy parents, Sheryl and Jim. Shortly after they brought Tayla home on Christmas Day, they received a call from the hospital telling them that Tayla's newborn screening test showed that she had a congenital disorder. Tayla had phenylketonuria (PKU), an inherited and treatable metabolic disorder. With PKU, the body cannot process the amino acid called phenylalanine (Phe), which is in almost all foods that contain protein. If left untreated, the Phe level can get too high in the blood, resulting in mental retardation (3).

When parents are told that their baby has a disorder like PKU, they need to know for certain that the tests are accurate. NSQAP works with all state public health laboratories, including the Wisconsin State Laboratory of Hygiene that analyzed Tayla's test, to assure that all test results are accurate and that no disorders are missed. In many cases, newborn screening makes the difference between life and death for newborns; in other instances, identifying babies with a disorder means that they can be treated and thus not face life-long disability or cognitive impairment.

The good news for the Cunzenheims is that today Tayla is thriving. The treatment for Tayla involves keeping her on a special diet and special formula. The Cunzenheims worked with a dietician from the Waisman Center at the University of Wisconsin to develop this diet plan. Her parents describe Tayla as "smart as a whip." She attends school and loves to draw and color. What a gift!

In many cases, newborn screening makes the difference between life and death for newborns; in other instances, identifying babies with a disorder means that they can be treated and thus not face life-long disability or cognitive impairment.

A Snapshot of NSQAP

Newborn screening begins with a heel stick that is done within 24-48 hours of a baby's birth. A health-care professional collects blood from the baby's heel onto a filter-paper card, which is the blood-collection device (card). This card is sent to a newborn screening laboratory for testing.

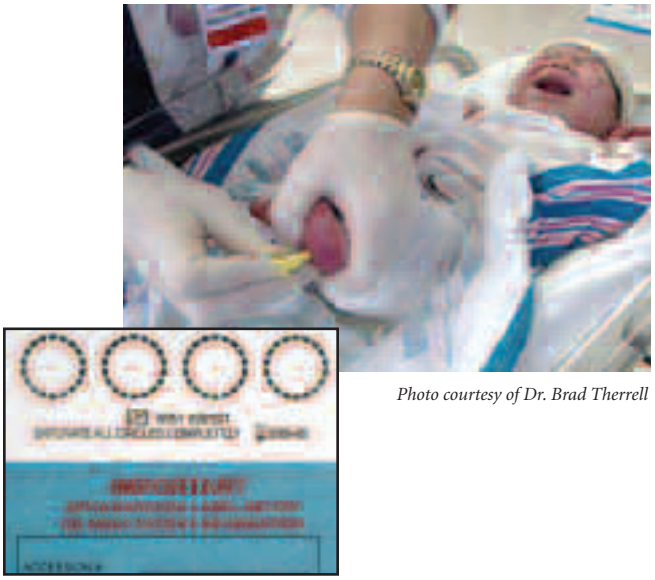
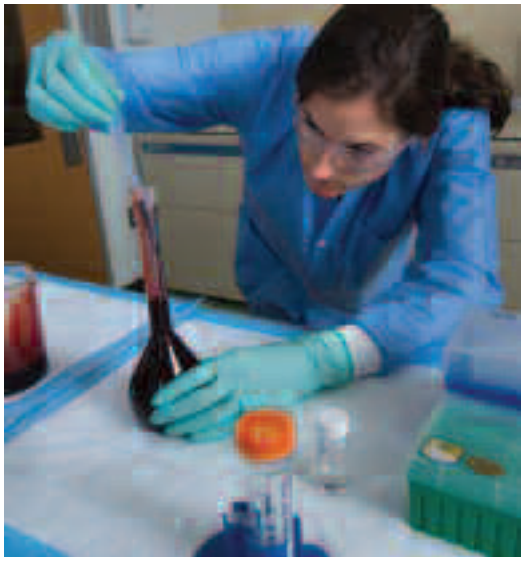


Photo courtesy of Dr. Brad Therrell

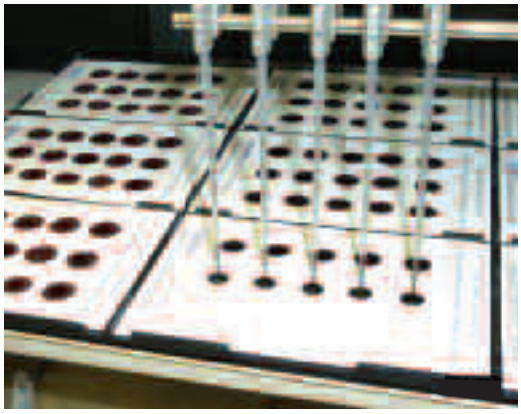
Working with manufacturers, NSQAP tests all of the filter paper used in the blood-collection devices before they are released for sale. Filter-paper manufacturers provide CDC with sample sets of paper from the beginning, middle, and end of their production lots. The amount of paper sent to CDC varies with the size of the production lots; however, thousands of strips of randomly selected paper are tested each year. The filter paper needs to adhere to strict specifications so that the blood can be collected and analyzed correctly.

NSQAP provides quality assurance products, including training, consultation, proficiency testing, guidelines, and reference materials, to state public health laboratories and other laboratories responsible for newborn screening. Currently, NSQAP works with laboratories in every state and U.S. territory as well as with laboratories worldwide.

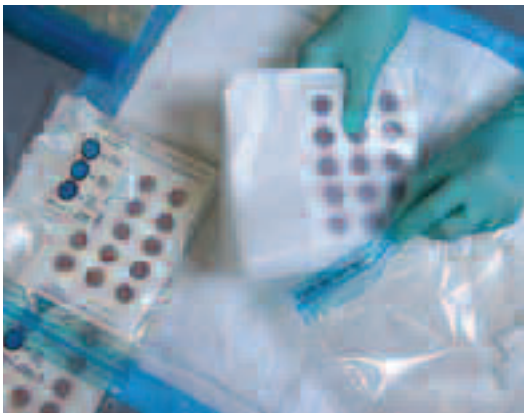
One of NSQAP's primary efforts is to prepare dried blood spot reference materials for distribution to laboratories participating in the program's quality assurance tests. Each year, NSQAP creates and sends out nearly a million dried blood spot reference specimens. CDC scientists start with prescreened blood from blood banks. They enrich the blood with selected biomarkers (i.e., chemical substances associated with the specific disorders in newborn screening tests).



The blood is then spotted—either by hand or through use of a robot—onto filter paper.

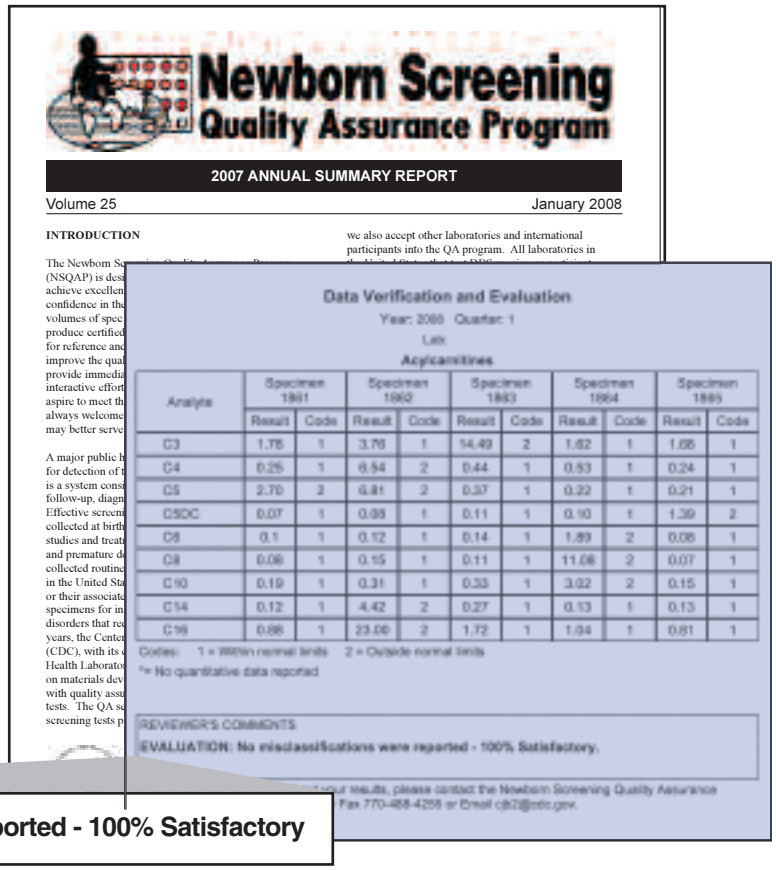


The spots are carefully dried and packed to avoid any degradation and then stored in a freezer until needed for quality control and proficiency tests.



The quality assurance efforts at CDC involve a continual interaction between NSQAP and the participating laboratories. NSQAP prepares panels of dried blood-spot specimens and sends them out to participating labs.

The laboratories analyze the specimens and return their assessments to NSQAP for review. NSQAP then compiles and reports the results to help laboratories maintain accurate and reliable testing practices. The Data Verification and Evaluation reports, along with NSQAP's dedicated efforts to work with laboratories, help newborn screening laboratories get the right answer every time.



Newborn Screening
Quality Assurance Program

2007 ANNUAL SUMMARY REPORT

Volume 25 January 2008

INTRODUCTION

we also accept other laboratories and international participants into the QA program. All laboratories in

Data Verification and Evaluation

Year: 2000 Quarter: 1

Lab:

Acylcarnitines

Analyte	Specimen 1991		Specimen 1992		Specimen 1993		Specimen 1994		Specimen 1995	
	Result	Code	Result	Code	Result	Code	Result	Code	Result	Code
C3	1.76	1	3.76	1	14.49	2	1.62	1	1.66	1
C4	0.25	1	6.54	2	0.44	1	0.53	1	0.24	1
C5	2.70	2	6.81	2	0.37	1	0.22	1	0.21	1
C5DC	0.07	1	0.08	1	0.11	1	0.10	1	1.39	2
C8	0.1	1	0.12	1	0.14	1	1.89	2	0.06	1
C8	0.06	1	0.15	1	0.11	1	11.06	2	0.07	1
C10	0.19	1	0.31	1	0.33	1	3.02	2	0.15	1
C14	0.12	1	4.42	2	0.27	1	0.13	1	0.13	1
C16	0.66	1	23.06	2	1.72	1	1.04	1	0.81	1

Codes: 1 = Within normal limits 2 = Outside normal limits

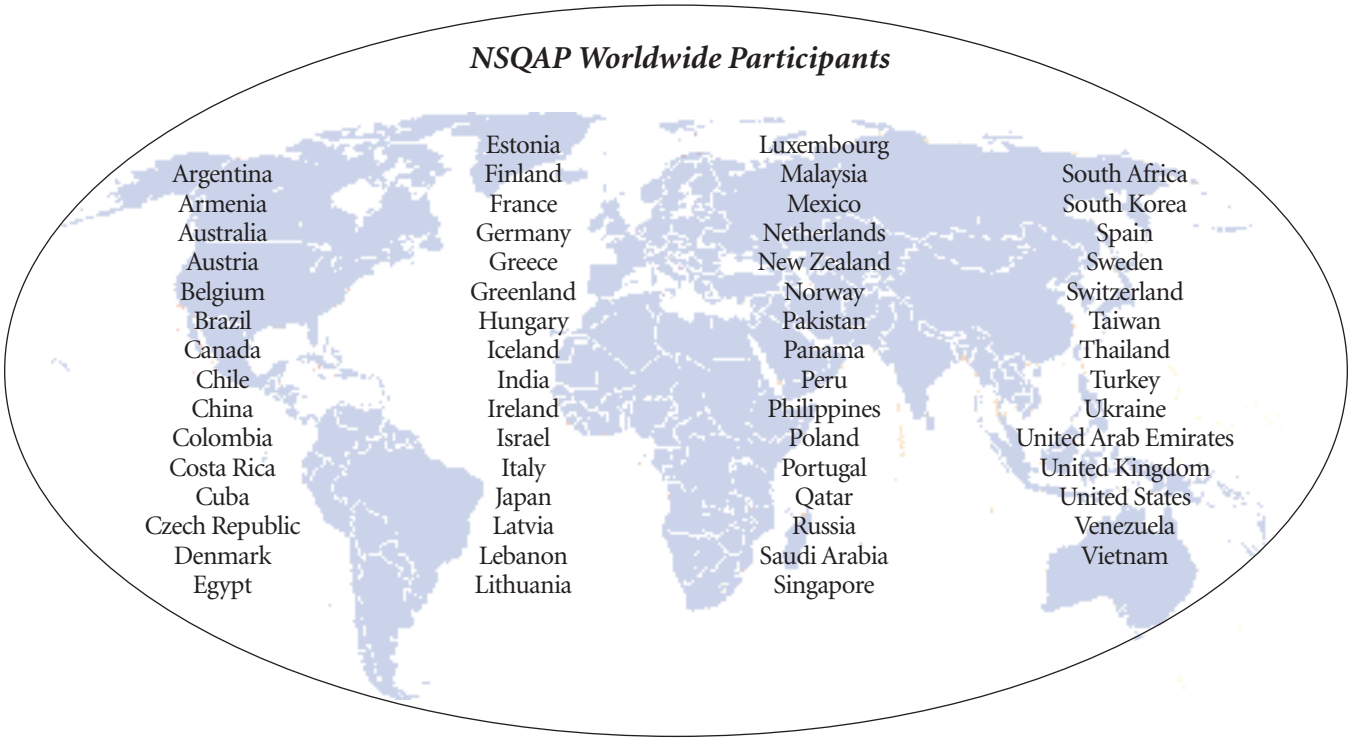
* No quantitative data reported

REVIEWER'S COMMENTS

EVALUATION: No misclassifications were reported - 100% Satisfactory.

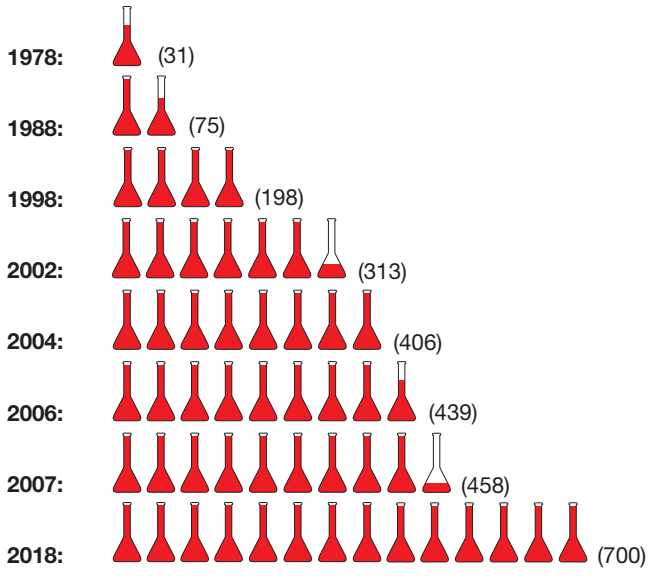
EVALUATION: No misclassifications were reported - 100% Satisfactory

NSQAP monitors newborn screening worldwide so that it can stay on top of new technology and tests. The graphic below lists countries with one or more labs participating in NSQAP.



Since 1978, NSQAP has steadily added laboratories to the program. Whereas only 31 laboratories participated in 1978, nearly 500 labs were enrolled in NSQAP by 2007 (Fig. 3).

Figure 3
Number of active labs in NSQAP (by year)



NSQAP makes sure that there is sustained quality in newborn screening so that a diagnosis such as Gabriel's is not missed.

Obtaining accurate results

Gabriel George

Gabriel George was diagnosed with sickle cell disease when he was just three weeks old. Sickle cell disease is a group of inherited red blood cell disorders. Healthy red blood cells are round, and they move through small blood vessels to carry oxygen to all parts of the body. In sickle cell disease, the red blood cells become hard and sticky, and consequently clog the blood flow in small blood vessels. This condition can cause pain and other serious problems, such as anemia and infections (4). NSQAP makes sure that there is sustained quality in newborn screening so that a diagnosis such as Gabriel's is not missed.

Since his diagnosis, Gabriel's parents, Charles and Cheryl George, have been actively involved in the Marc Thomas Sickle Cell Foundation in Austin, Texas. Gabriel enjoys reading, riding his bike, and spending time with his brother Nathaniel. Gabriel was named the 2007-2009 poster child for the Sickle Cell Disease Association of America, Inc.

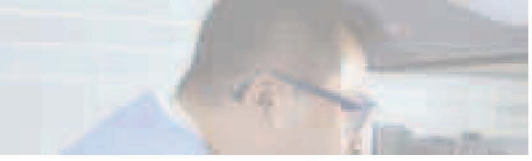
Improving health outcomes

Matthew Fisch

When Jill Levy-Fisch's third child, Matthew, was born, she knew something was not quite right. He was developmentally delayed, and he never smiled. At age one, Matthew weighed 22 pounds. By the time he reached his second birthday, Matthew still weighed 22 pounds. During the first two and a half years of Matthew's life, Jill went from doctor to doctor searching for an answer in what is sometimes called a medical diagnostic odyssey. Matthew endured test after test, and the experience was an emotional strain on everyone in the family. At age three-and-a-half, Matthew was finally diagnosed with short chain acyl-CoA dehydrogenase deficiency (SCADD). SCADD is a rare condition that prevents the body from converting certain fats into energy. Some of the symptoms of SCADD include vomiting, low blood sugar, a lack of energy, poor feeding, and failure to gain weight and grow at expected rates. Other features of this disorder may include poor muscle tone, seizures, and developmental delays (5). While there is no standard treatment for SCADD, Matthew has a feeding tube, takes dietary supplements to restore his metabolism, and requires physical therapy. In addition, because of his developmental delay, he visits a reading tutor four days a week.

Newborn screening for SCADD was not available in Matthew's state at the time of his birth in 2001. Spurred to action, Jill began working for the Save Babies Through Screening Foundation, a volunteer organization that supports newborn screening. She wants to make sure that no family has to endure a medical diagnostic odyssey like her family did. Had Matthew been diagnosed through newborn screening, therapies could have been introduced earlier, and his health outcomes would have been better. While Matthew did not receive the benefit of an accurate, early diagnosis, Jill is thrilled that today her state screens for SCADD and that NSQAP works with her state laboratory to ensure accurate newborn screening tests for SCADD.

Had Matthew been diagnosed through newborn screening, therapies could have been introduced earlier, and his health outcomes would have been better.



III. CDC's Role in Translation Research

Collaborating with State Public Health Laboratories

Cutting-edge technology is available now that can test newborns for more diseases than has ever been possible. CDC is working to harness the latest advances in science and technology so that more disorders can be detected accurately and treated quickly. In 2005, CDC established the Newborn Screening Translation Research Initiative (NSTRI) with the CDC Foundation. Working with corporate, academic, and Foundation partners, NSTRI assures the quality of research methods during both pilot studies and routine screening. Goals for NSTRI include—

- developing new screening tests for specific disorders.
- adapting innovative methods such as DNA testing and nanotechnology for screening and quality assurance.
- transferring new screening technologies to state public health laboratories.
- assisting states with pilot studies related to new screening tests for newborns.
- supporting state laboratory functions when states add disorders to their current panel of tests.

NSTRI has developed laboratory projects focusing on a variety of disorders, including lysosomal storage disorders (LSD) and severe combined immune deficiency (SCID). In 2008, CDC supported three pilot studies for SCID, including one involving the Indian Health Service and the Western Navajo Reservation in Arizona. By 2018, CDC anticipates that it will collaborate on, support, or fund 15 pilot projects for conditions such as Pompe disease, Krabbe disease, Fabry disease, Niemann-Pick, metachromatic leukodystrophy (MLD), DiGeorges disease, and X-linked adrenoleukodystrophy (X-ALD).

Lysosomal Storage Disorders (LSDs): Assuring the quality of new tests

LeA Gartzke

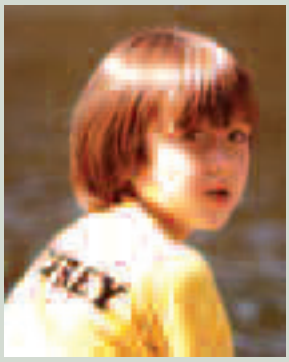
In 1996, Micki Gartzke gave birth to a daughter named LeA. As a newborn, LeA ate and slept like all other newborns and began to notice the world around her. Within a few months, LeA's physical condition changed dramatically. Her body became rigid. She cried constantly, and she would not eat. At 10 months, LeA was diagnosed with Krabbe disease, which is part of a group of approximately 40 rare diseases known as lysosomal storage disorders (LSDs).

LSDs are caused by deficient enzymes that normally eliminate unwanted substances in the cells of the body (6). Because the diagnosis took months, LeA was past the period when she could receive treatment. Her physical condition worsened to where she needed feeding tubes, specialty formulas, 36 doses of medicine a day, oxygen, suctioning, and in-house nursing. After a quarter of a million dollars worth of medical bills, LeA died at the age of two. Her mother says that early diagnosis and intervention could have made the difference in LeA's life, and for this reason, Micki Gartzke became a strong advocate for newborn screening. Today, she works with foundations, researchers, corporations, and legislators to raise awareness about the importance of screening.

One state—New York—now screens for Krabbe disease, and CDC plans to provide the reagents that can be used to perform the Krabbe tests. In addition, CDC has established a program to ensure that the Krabbe test results are accurate. Illinois has passed legislation mandating newborn screening for Krabbe and four other LSDs (Pompe, Fabry, Niemann-Pick, and Gaucher diseases). CDC will provide the reagents and will assure the quality of all five tests.

Severe Combined Immune Deficiency (SCID): Guiding the transition to routine tests

Jeffrey Modell



Jeffrey Modell was a sweet, cheerful, outgoing kid. He was like every other boy his age, except he got sick very often. His life was filled with normal, happy times, but was frequently disrupted with sudden high fevers, bronchitis, sinusitis and other infections that led to extended periods on medication and lengthy hospital stays. Plans for fun activities were made in spite of the strong possibility that Jeffrey might be too sick to attend. Even periods of good health were strained with the ever-present threat of sudden illness caused by Jeffrey's immune deficiency. Three decades ago, no one knew very much about primary immune deficiencies. They were considered so rare, that many doctors knew too little about them and had even fewer treatment options. Lay people had never heard of these disorders, and primary immune deficiencies were not at the top of many research agendas.

At 15, Jeffrey's condition overwhelmed him and took his life. Inspired by Jeffrey's courage and optimism, his parents, Fred and Vicki Modell, started a Foundation to raise awareness about primary immune deficiencies, including severe combined immune deficiency (SCID). Sometimes known as "Bubble Boy Disease," SCID is characterized by an inability to resist infections. Without early diagnosis and treatment, babies with SCID usually die within a year of birth. Newborns with SCID do not generally exhibit signs of infection or illness because during the first few weeks after birth, they are protected by the antibodies passed on to them from their mothers and because they have not yet been exposed to infection. Newborn screening enables diagnosis and treatment before the onset of infection and related medical complications; early detection also improves health outcomes and helps reduce hospitalizations (7).

The Modells recognize that having an accurate test leads to an early diagnosis and the best opportunity for effective treatment. Before newborn screening for SCID can be implemented at the state level, an effective and efficient screening method (that could process hundreds of samples daily) is needed (7). With Congressional funding, CDC is addressing this need through cooperative agreements that encourage states to research, develop, and evaluate newborn blood spot screening tests for SCID.

Lysosomal Storage Disorders (LSDs): Encouraging research and development

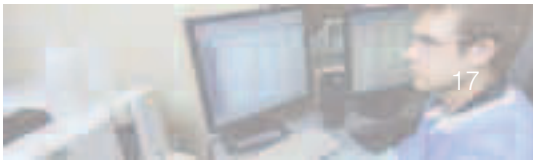
John, Jack, and Christopher Evanosky

Bob and Sonya Evanosky know the benefits of early diagnosis and intervention. Both of their twin boys, John and Christopher, were premature babies when they were born in 2001, and so it was not surprising to their parents that they met growth and development milestones more slowly than their peers. At 15 months of age, the twins were struggling to walk. This is when the Evanoskys' difficult search for answers began. After a battery of tests, John and Christopher were diagnosed with cerebral palsy. The twins began therapy for cerebral palsy; however, they soon began to lose the ability to walk, their speech worsened, and they were struggling to eat. The twins were eventually diagnosed with metachromatic leukodystrophy (MLD), an LSD.

John, Jack and Christopher Evanosky have metachromatic leukodystrophy (MLD), which is a genetic disease.

Although there is no cure for MLD, experimental treatment involving bone marrow transplant or cord blood stem cell transplant can delay the progression of the disease in infants without significant symptoms (8). John and Christopher were not candidates for treatment because they showed significant symptoms. The Evanoskys' youngest child Jack tested positive for MLD one month after the twins, and because he showed no symptoms, he underwent a stem cell transplant. Post-transplant, Jack is doing well, but unfortunately the future for John and Christopher is bleak.

The Evanoskys, through their Foundation, are working with CDC to develop newborn screening for MLD. This collaboration will also establish a dried blood spot card repository for conditions that are part of public health newborn screening programs as well as conditions that may be added in the future. This repository will help CDC enhance quality assurance by providing biologic reference materials for current and newly-implemented screening tests. Although their children cannot benefit from newborn screening for LSDs, the Evanoskys think that newborn screening is the best way to make sure that other children have a chance at a normal life. They recognize that ensuring the accuracy of the tests is critical and believe that families must have the health information they need to make informed decisions.



IV. Future Directions

Newborn Screening National Contingency Plan

When Hurricane Katrina hit New Orleans and the levees were breached, the state's newborn screening laboratory was decimated, and the normal operations of newborn screening, diagnosis, and follow-up were interrupted for several weeks. One of the lessons learned from that experience is that a back-up system or contingency plan is essential to keep this critical service functioning without interruption.

The Newborn Screening Saves Lives Act, which was signed into law in 2008, requires the development of a national contingency plan for newborn screening for use by a state, region, or consortia of states in the event of a public health emergency. CDC is working with the Health Resources and Services Administration, state public health departments, and others to develop a plan that addresses—

- the collection and transport of specimens;
- the shipment of specimens to state newborn screening laboratories;
- the processing of specimens;
- the reporting of screening results to physicians and families;
- the diagnostic confirmation of positive screening results;
- ensuring the availability of treatment and management resources;
- educating families about newborn screening; and
- carrying out other activities determined appropriate by the Secretary of Health and Human Services.

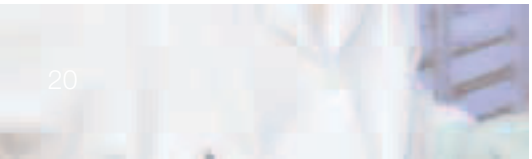
The establishment and continued refinement of a national contingency plan will help ensure that all babies receive the benefits of newborn screening, even under emergency circumstances.

Environmental Uses of Dried Blood Spots

For at least three decades, scientists at CDC have been determining which environmental chemicals people have been exposed to and how much of those chemicals actually gets into their bodies. This technique is known as biomonitoring. CDC is combining its biomonitoring expertise with its newborn screening expertise to examine the possibility of using newborn screening dried blood spots to measure exposure to environmental chemicals, such as pesticides and metals. The benefit of using dried blood spots is that researchers will be able to determine which environmental chemicals are actually in newborns. Such research will improve pediatric studies, for example, that currently depend on interviews and memory to try and reconstruct exposure history (9).

Although this is a promising area of study, a number of significant issues need to be addressed before any kind of widespread use can begin. For example, overcoming sample volume limitations, determining the priority chemicals to measure, and facing analytical/methodological challenges are all valid concerns that can be addressed through further research and collaboration.

CDC can use its laboratory and newborn screening expertise to collaborate with states and other research partners to develop laboratory methods that can take the microliter volume of whole blood available in dried blood spots and still produce precise, accurate measurements of selected environmental chemicals.



V. Next Generation Newborn Screening

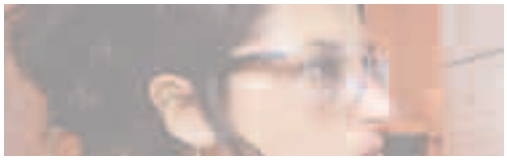
Genomics

CDC strives to stay in the forefront of technological and scientific advancement in order to continually improve its newborn screening-related activities. Recently CDC expanded laboratory capabilities for its Newborn Screening Quality Assurance Program (NSQAP) by including a group of its scientists (molecular biologists) who have extensive expertise in understanding how genetics and changes in DNA are associated with important public health issues such as diabetes, kidney disease, birth defects, and asthma. The expertise of these scientists helps newborn screening research because DNA-based testing—

- can be used to confirm the results when a newborn tests positive for a disorder.
- can be done, to confirm a positive result, on the same blood spot used for the initial test (allowing all screening results to be reported at the same time without worrying families about testing and retesting).
- can be used to add new disorders to panels for newborn screening.

The newly expanded laboratory has focused on cystic fibrosis, a particularly complex disease with more than 1500 known mutations. The collaboration of CDC scientists and the Cystic Fibrosis Foundation (which was instrumental in providing donor case samples) resulted in the development of proficiency testing materials that are now being used by state laboratories to assure high-quality DNA-based confirmatory testing of positive screening results.

As genetic tests become more available, many newborn screening laboratories are considering the use of DNA-based testing to confirm positive results for disorders such as galactosemia, congenital adrenal hyperplasia, severe combined immune deficiency and lysosomal storage disorders. As states move testing in this direction, it will be critical for them to have access to technical expertise as well as the kind of blood spot materials necessary for quality assurance and proficiency testing. As newborn screening laboratory testing evolves, CDC will continue to provide the technical expertise and the materials required as more disorders are added to the states' screening panels. NSQAP—the only comprehensive quality assurance program for newborn screening— has the experience, the infrastructure, and the solid reputation in the newborn screening community to respond to the changing technology and rapid expansion of this field.




Emerging Technologies

Although DNA-based testing is a developing part of newborn screening, biochemical testing is widely used in state newborn screening programs. CDC can use its expertise in both genetics and biochemistry to collaborate with states, research partners, and stakeholders to provide comprehensive data for new methods, new applications, and new blood spot materials for use in newborn screening. In particular, CDC is interested in collaborative research that—

- helps states maintain high standards of proficiency as they expand their newborn screening tests and programs.
- seeks to understand how genetic and biochemical differences that occur within the diverse population of the United States will help improve newborn screening.*
- explores innovative testing technologies (e.g., nanotechnology and "a lab on a chip").†

Ultimately, the knowledge gained from future research and collaborations will lead to improvements and expansion of newborn screening activities that save lives and improve the health and well-being of all babies.

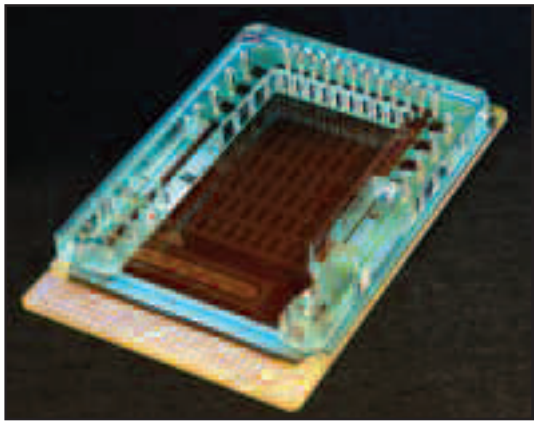


Image courtesy of Dr. David Millington of Duke University Medical Center

**Understanding the genetic and biochemical differences will not only help improve newborn screening but also allow for research that provides a better understanding of the disease process. CDC scientists will collaborate on population studies with research partners at the federal and state level.*

†*DNA microarray or digital microfluidics (e.g., chip technology or "lab on a chip") can be used for detecting disease markers. This technology is appealing to the field of newborn screening because, with additional research and development, it holds the potential to screen simultaneously for hundreds of disorders, improve testing specificity, improve prediction of disease severity (which can guide treatment decisions), enable the inclusion of infectious agents, reduce the amount of blood needed for testing, and produce faster results (10).*

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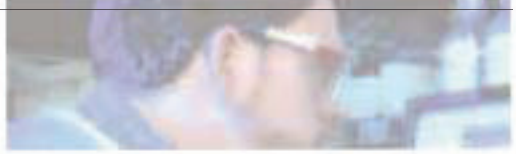



VII. Selected Publications

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