



**1965-2015**

**Michigan**

**Newborn**

**Screening:**

**A Public Health**

**Success Story**



# Foreword

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Half a century is a long time, especially when talking about a public health program. Since the start of the Michigan Newborn Screening Program in 1965, it was evident that the work was not only life saving for the babies identified but also very important to the people who worked persistently on improving the program. The original Michigan screening pioneers, Drs. Richard Allen and K. Stanley Read, with great support from state senator, Dr. Vern Ehlers, developed the foundation for what the program is today. Now, the Michigan Newborn Screening

Program screens for 55 disorders. Five decades of hard work and dedication has led to life altering diagnosis and treatment for over 7,200 Michigan newborns.

To celebrate these fifty years, the Newborn Screening Program has put together a compilation of Michigan's successes. We would like to thank all of those involved in the newborn screening process, from hospital staff to medical management, for making all of this possible. Here's to fifty more years of improving and saving babies' lives!

- Harry Hawkins, *Newborn Screening Laboratory Manager*
- William Young, *Newborn Screening Follow-Up Program Manager*

*January, 2015*





# Acknowledgements

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*Michigan's Newborn Screening Program has a rich history of success made possible by the contributions of many individuals over the last 50 years. We'd like to recognize and thank those who were so instrumental in advocating for, and implementing new screening technologies and treatments that have benefited Michigan babies over the last five decades. Each of the following individuals dedicated more than 25 years of their career to make newborn screening a better program — without them, it would not be what it is today.*

## **Pioneers**

### **Dr. Richard Allen**

Pediatric Neurologist and Director,  
University of Michigan Metabolic Clinic

### **Dr. K. Stanley Read**

Public Health Laboratory Microbiologist

## **Leaders**

### **Harry Hawkins**

Newborn Screening Laboratory Section Manager

### **Dr. Charles Whitten**

Hematologist and Founder, Sickle Cell Detection and Information Program

### **Dr. William Young**

Newborn Screening Follow-up Program Manager

## **Support Team**

### **Karen Andruszewski**

Newborn Screening Quality Assurance Coordinator

### **Denise Archambeault**

Newborn Screening Laboratory Data Technician

### **Janice Bach**

Genomics and Genetic Disorders Section Manager

### **Caron Burns**

Newborn Screening Laboratory Endocrine Unit Manager

### **Catherine Mazzolini**

University of Michigan Metabolic Clinic  
Administrative Assistant

### **Eleanor Stanley**

Newborn Screening Laboratory Metabolic Unit Manager

# Introduction

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**Newborn Screening** is a public health program that touches nearly every Michigan newborn in all corners of the state, from Detroit to Traverse City to the far reaches of the Upper Peninsula. Every baby must be screened because most babies who have a medical condition on the newborn screening (NBS) panel seem healthy at birth but can become very sick in a short time. If not found early, the consequences can include serious and permanent health problems, severe developmental delays and even death.

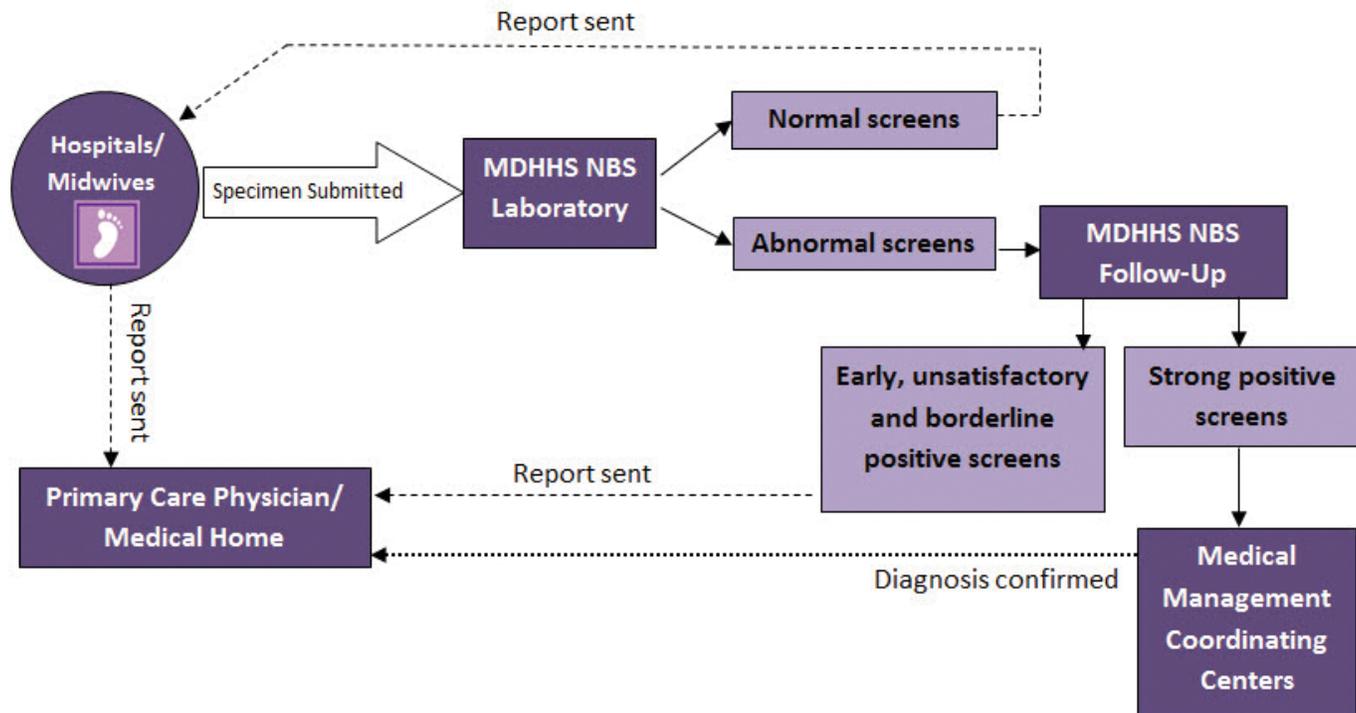
Newborn screening works because a coordinated system of players—hospitals and midwives, couriers, public health laboratory and follow-up staff, primary care providers, medical specialists and families—help to make sure every baby has an opportunity to be tested and treated quickly, if needed.

Between 24 and 36 hours of life, a few drops of blood are drawn from a baby's heel to fill five or six spots on a filter paper card. The card with the dried blood spots is sent to the State Newborn Screening Laboratory for testing. Follow-up with referral to medical management coordinating centers is activated immediately when an abnormal screen is reported.

Thanks to advances in science and hard work by many dedicated individuals over the last 50 years, Michigan blood spot screening now looks for over 50 conditions that may affect blood cells, brain development, how the body breaks down nutrients from food, hormones, lungs and breathing, and how the body fights infection. In addition, babies are screened by hospital and home birth attendants for hearing loss and low oxygen levels that could be a sign of critical congenital heart disease. On the occasion of our golden anniversary in 2015, we stop to reflect on our accomplishments and celebrate lives saved and improved through 50 years of newborn screening.



## Michigan's Blood Spot Screening and Follow-up System



## In the Beginning

A breakthrough in the treatment of inborn errors of metabolism occurred in 1954 when Dr. Horst Bickel and co-workers introduced a dietary therapy for the management of the rare disorder phenylketonuria (PKU), a disease that without treatment can cause damage to the brain and central nervous system. At the same time, it was recognized that the therapy would be most effective if introduced in the newborn period. This prompted the search for an effective screening test for early detection of PKU. The discovery came in 1962 when Dr. Robert Guthrie devised a brilliant method for detecting PKU in large populations of newborn infants. The method was the semi-quantitative Bacterial Inhibition Assay (BIA) that allowed growth of *Bacillus subtilis* on agar plates exposed to elevated levels of phenylalanine in a drop of blood obtained from a newborn's heel. Not only did Dr. Guthrie invent this newborn screening test for PKU, he devised the logistics for obtaining blood on filter paper from newborns in hospital nurseries and sending the specimens to a centralized state public health laboratory for testing, so that babies with PKU could be identified and treated early to improve health outcomes. In retrospect, the simple strategy Dr. Guthrie promoted throughout his career has proved to be one of the most significant achievements in public health over the past 50 years.

*PKU was the first disorder diagnosed through newborn screening. An inborn error of amino acid metabolism resulting from a deficiency in the enzyme phenylalanine hydroxylase, it can lead to severe intellectual disabilities without early treatment.*

# The Michigan NBS Story: From One Disorder to 50+

Michigan's Newborn Screening Program began in **1965** as a result of Dr. Guthrie's screening technology for detection of PKU. The program was pioneered by Dr. Richard Allen, a pediatric neurologist at the University of Michigan, and Dr. K. Stanley Read, a microbiologist at the Michigan Department of Public Health Laboratory. The collaboration of Drs. Allen and Read established state laboratory testing methods and protocols for referral, diagnosis and medical management. These men realized very early the power of this public health strategy in prevention of disability not only for PKU but for other inherited disorders and birth defects. This was demonstrated in **1977** when Michigan became one of the first states to add a second disorder to the NBS panel. By using the same dried blood spot specimens collected for PKU detection, screening for *Congenital Hypothyroidism* (CH) was added.

In **1985**, *Galactosemia* (GALT) was the third disorder added to the screening panel. Over the years, GALT was found to be life threatening in the newborn period and therefore, timely diagnosis and early treatment were vital for saving the lives of affected babies.

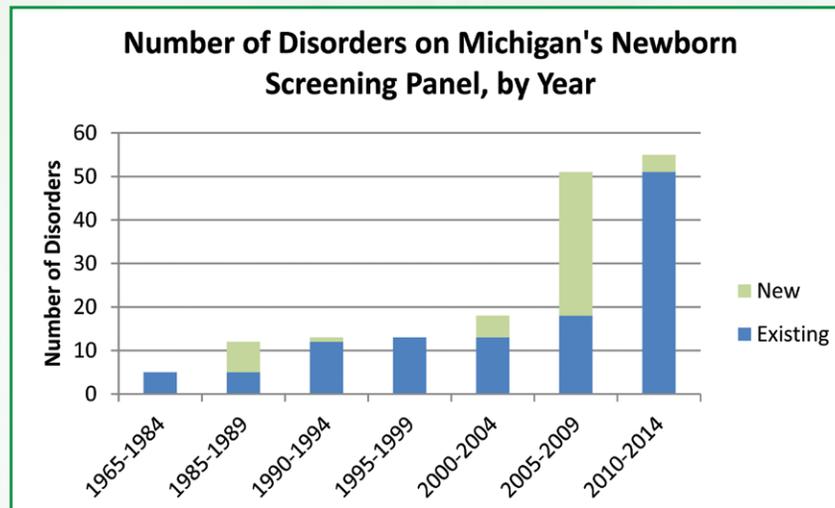
With the continued addition of new diseases to the NBS panel, it became important to develop a system for short-term and long-term follow-up. The NBS Follow-up and medical management programs developed by Dr. Allen and Dr. William Young in the 1980s, remain a fundamental part of newborn screening in order to ensure every baby in Michigan receives a screen, and that those identified receive proper diagnosis and treatment.

Shortly after GALT screening began, it was recognized that a reliable funding source would be necessary to further expand NBS for additional disorders. Fortunately, a far-sighted state senator, Dr. Vern Ehlers, recognized the importance of the



screening program. In 1987, Senator Ehlers, with help from Dr. Allen and Dr. Charles Whitten, introduced and guided a groundbreaking bill through the legislative process. Public Act 14 of 1987 doubled the screening panel from three to six disorders, adding *Biotinidase Deficiency* (BIOT), *Maple Syrup Urine Disease* (MSUD) and *Sickle Cell Disease* (SCD). The legislation also established a fee for each filter paper test card sold, assuring a more stable funding source to cover the costs associated with laboratory testing and follow-up. Senator Ehlers would remain a strong advocate for newborn screening both as a state Senator and later as a member of the U.S. Congress from Michigan's 3rd congressional district.

The introduction of tandem mass spectrometry (MS/MS) allowed for major enhancements in NBS laboratory technology making it possible to detect amino acid, organic acid and fatty acid oxidation disorders. With strong support from family advocates that led to a legislative mandate, Michigan added screening for *Medium-chain acyl-CoA dehydrogenase deficiency* (MCAD) in 2003. Over the next two years, the availability of MS/MS technology allowed the addition of *Homocystinuria* (HCY), *Citrullinemia* (CIT), *Argininosuccinic Acidemia* (ASA) and 31 more conditions.



While the NBS fee enacted in 1987 provided a mechanism for effective maintenance and expansion of the program, adding any new disorder still required legislative approval to amend the public health code. With the ever increasing technical complexities involved in decision making about NBS, Senator Tom George and others sponsored a bill, which later became Act No. 31 of 2006, to establish a legislatively mandated Newborn Screening Quality Assurance Advisory Committee (NBS-QAAC). This panel of experts has met annually since 2006 to review the program and recommend changes to the screening panel for legislature approval, streamlining the process and avoiding the need for introduction and passage of new legislation whenever a disorder is added.

One of the first recommendations made by the NBS-QAAC was to add *Cystic Fibrosis* (CF) to the panel, beginning in **2007**. Testing for CF required additional enhancements in laboratory technology brought on by Michigan Department of Health and Human Services (MDHHS) laboratory scientist Kelly TenEyck. This technology for the first time incorporated the use of molecular testing in the newborn screening environment. A related molecular technology would be used again in **2010** for the application of *Severe Combined Immunodeficiency* (SCID) screening. Laboratory scientist, Heather Wood, played a major role both in Michigan and nationally in perfecting the molecular techniques used for detection of these disorders.

Another enhancement in **2007** was statewide implementation of the point of care screening test for *Hearing Loss*. Although many hospitals had already begun to screen infants several years earlier, universal screening of all infants was not required until 2007. More recently, in **2014**, a second point of care screen was added to the mandated NBS panel, namely pulse oximetry screening for detection of *Critical Congenital Heart Disease* (CCHD). Also in **2014**, *Pompe disease* was the first lysosomal storage disorder approved for the Michigan NBS panel with implementation of statewide screening scheduled to begin by the fall of 2015. Michigan's NBS program has undergone remarkable expansion since its inception—from a single disorder in 1965 to 55 conditions as of **2015**. Approximately 6.9 million Michigan newborns have been screened with more than 7,200 babies identified with disease and treated early for disabling and life threatening conditions.

# Michigan NBS Timeline



1965 – Dr. Richard Allen and Dr. K. Stanley Read implement newborn screening for PKU



1977 – CH

1985 – GALT

1987 –BIOT, MSUD and SCD



1965 – First Michigan newborn is identified with PKU through NBS

### National Milestones in the Last Decade

|  |  |
|--|--|
| <p><b>2005</b> – American College of Medical Genetics determines the Recommended Uniform Screening Panel for NBS, a list of conditions recommended for screening which Michigan follows when adding disorders to the panel</p> | <p><b>2008</b> – Newborn Screening Saves Lives Act is passed</p> |
| <p><b>2011</b> – CDC declares NBS one of the “Ten Great Public Health Achievements”</p>  |  |

1987 – Public Health Code is amended by Act 14 of 1987 to add three disorders and initiate a fee for the NBS test

1990 – 3 millionth baby is screened

1990 – Michigan one of the first states to use automated continuous flow technology for PKU and GALT testing

\* Dates above the timeline signify when new disorders were added to the Michigan Newborn Screening panel.

1993 – Congenital Adrenal Hyperplasia (CAH)



2003 – MCAD

2004 – HCY, CIT and ASA

2005 – MS/MS allows addition of 31 metabolic disorders

2011 – SCID

2014 – CCHD



2015 – 2016 Screening for Pompe disease, a lysosomal disorder, scheduled to begin

2007 – CF

2007 – Hearing Loss



2000 – Public Health Code is amended by Act 33 of 2000 to allow the use of blood specimens for medical research during retention period established by the department

2006 – Public Health Code is amended by Act 31 of 2006 to create NBS-QAAC

2009 – NBS results are displayed on the Michigan Care Improvement Registry (MCIR) for healthcare providers

2015 – Newborn Screening Online (NBSO) Card Order and Inventory System is implemented



2003 – Laboratory Information Management System (LIMS) is developed

2008 – Laboratory operations are extended to six days a week

2003 – MS/MS is first used for MCAD, MSUD and PKU

2008 – Courier system is implemented to reduce specimen transit times

2009 – The Michigan BioTrust for Health initiative is launched



# Laboratory Blood Spot Screening



At the heart of Michigan newborn screening is the blood spot testing performed by the MDHHS Laboratory, where scientists and technicians take great strides to make sure all samples are tested accurately and results are reported as quickly as possible. As described earlier, Michigan NBS started with the “Guthrie test” for detection of PKU. This relatively inexpensive test was done by comparing and measuring bacterial growth zone rings around a ¼ inch disc that was punched out of blood or urine samples on filter paper cards. Many refinements have brought newborn screening a long way due to improvements in laboratory technology and instrumentation over the last half-century. Through the years it became obvious that this vital screening test would best be performed in a high volume, centralized laboratory rather than individual hospitals. To improve testing quality at an affordable cost, the State Public Health Laboratory officially took over the screening duties for all Michigan newborns in 1987.



The laboratory has experienced many changes over the years. In 1978, the Centers for Disease Control and Prevention implemented a Quality Assurance Program to provide proficiency testing and vital support for state laboratories. The Clinical Laboratory Improvement Amendments (CLIA) program was phased in through 1994 to establish quality standards for laboratory testing. The filter paper for specimen collection is now specially manufactured as a controlled filter paper medical device. Many of the assays have become more automated, and a sophisticated laboratory information management system tracks each sample from arrival in the lab through every step of the screening process. Through rigorous quality

control and quality assurance, these changes allow for quicker and more accurate results. In addition, every infant’s screening results are now available to his or her primary care provider on the Michigan

Care Improvement Registry website. With guidance from former laboratory division directors, Dr. Jacqueline Scott (1990-2002) and Dr. Kevin Cavanagh (2003-2013), major advancements were implemented.

The laboratory actively participates in national initiatives to improve screening standards promoted by the Association for Public Health Laboratories and Newborn Screening Technical Assistance and Evaluation Program; and undergoes inspections to meet the College of American Pathologists (CAP) accreditation process. Recently the laboratory has undergone renovations to provide workspace for new instruments. Since 2008, the laboratory operates six days a week including holidays, employs 24 staff members and processes over 122,000 samples a year.



Since 1965, the MDHHS Laboratory has helped identify nearly 5,700 newborns with disease, although many more infants receive an initial positive result requiring follow-up to make sure no problems exist. To minimize the chance of missing a true case of disease, the laboratory works with the MDHHS NBS Follow-up team and medical experts to establish cutoff ranges for abnormal results that balance sensitivity and specificity, making sure an assay is highly likely to identify affected children while minimizing the number of “false positive” test results. For every true case, about 7 babies need additional follow-up to rule out a disorder. That may be as simple as repeating the heel stick screen, or involve a referral for diagnostic evaluation and testing by a medical specialist. But the payoff—finding the one baby with a condition that will benefit from early treatment out of about every 450 children screened—is well worth the effort.

# Continuous Quality Improvement

A key factor to the success of NBS is making sure a specimen is obtained from every baby between 24-36 hours after birth and getting the sample to the state laboratory in Lansing for testing as quickly as possible. Delays in blood spot specimen arrival at the laboratory could contribute to irreversible health problems for infants with a disorder requiring immediate diagnosis and treatment. Prompt specimen collection, pickup and delivery reduce turnaround time from birth to reporting of results and initiation of treatment.

The logistics of sending NBS specimens quickly from across Michigan's large geographic area—83 birthing hospitals in 48 counties—to the state laboratory are complex. In 2006, more than 95% of specimens were sent by U.S. mail, arriving on average 3.4 days after collection. In 2008, the NBS Program began providing a courier service to all Michigan birthing hospitals in an effort to reduce specimen transit time. Currently, all birthing hospitals in the state have Monday through Friday and either Saturday or Sunday courier service. Along with Saturday laboratory operations, these changes have resulted in reducing average specimen transit time to 1.8 days in 2014. A recent innovation is the creation of hospital-specific cutoffs for evaluating specimen transit time based on specimen collection time and each hospital's particular courier pickup days and times. This information allows for better monitoring and identification of hospital or other transit-related factors that contribute to delays.

After specimens arrive in the laboratory, Newborn Screening Follow-up staff links NBS cards with birth certificates to find babies whose blood spot specimen has not been received in the laboratory and may have been missed. Staff also works with hospitals and midwives to reduce the number of specimens drawn that are unsatisfactory for testing, and to reduce

| Hospital Performance Metric                 | Goal |
|---|------|
| Late Screens                                | <2%  |
| Receipt by Appropriate Day                  | >90% |
| Unsatisfactory Screens                      | <1%  |
| NBS Card Number on Birth Certificate        | >95% |
| Completed BioTrust for Health Consent Forms | >90% |
| Reported Pulse Oximetry Screening Results   | >90% |

turnaround time from collection to receipt in the laboratory. Training and technical assistance are provided through quarterly hospital-specific performance reports, newsletters, screening guides, and other materials posted to [www.michigan.gov/newbornscreening](http://www.michigan.gov/newbornscreening) as well as site visits and regional in-service trainings.

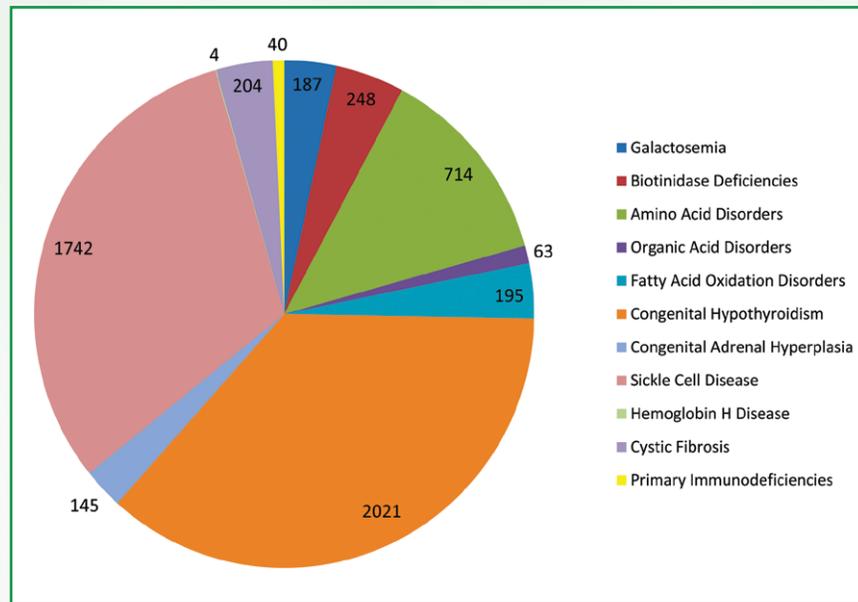
As part of continuing efforts to improve quality and customer service, a Newborn Screening Online (NBSO) ordering system has been developed and will be implemented before the end of 2015. NBSO provides a web-based system for purchasing NBS test cards and will be available 24/7 to hospitals, midwives and homebirth parents. All NBS related pamphlets can also be ordered online. NBSO is expected to expedite the shipping process and allows automated tracking and inventory of all NBS supplies which were formerly manual functions.

Through continual process improvements such as monitoring timely specimen collection and transit, evaluating screening algorithms, and establishing referral and medical management protocols, the Michigan NBS Program has become one of the most comprehensive and effective screening programs in the nation.



# Follow-Up and Medical Management

Currently, the NBS program confirms an average of 255 newborns with diseases on the NBS panel through blood spot screening each year. NBS is no longer just a test for PKU nor is it just a blood screening test. NBS is a comprehensive program that includes blood spot, hearing and critical congenital heart disease screening with assurance of follow-up. If a baby is identified with one of these conditions, treatment is started early and usually continues through life. Referrals of suspected cases are made to designated medical management coordinating centers, and a network of medical specialists has been identified for each group of diseases. This multi-faceted collaboration allows all newborns across the state to benefit from early identification and coordinated comprehensive care.



Distribution of Disorders Identified in Newborns via blood spot screening, Michigan Residents, 1965-2014

## Cystic Fibrosis

Cystic fibrosis (CF) is an inherited chronic disease that primarily affects the respiratory and digestive systems. Newborn screening will identify nearly all infants with CF and some who are carriers (those with one abnormal CF gene but not affected with the disease). Follow-up testing after a positive screen differentiates newborns who have CF from those who are carriers. The Cystic Fibrosis Newborn Screening Coordinating Center at the University of Michigan and MDHHS work closely with the state's five CF care centers in Ann Arbor, Detroit, Lansing, Grand Rapids and Kalamazoo so that all babies receive diagnostic sweat chloride testing, genetic counseling and specialty care following a positive CF newborn screen. These centers are accredited by the CF Foundation in providing expertise in the diagnosis and management of children with CF. While there is wide variation in disease symptoms, early treatment is usually aimed at preventing lung infections and improving nutrition. The Cystic Fibrosis Quality Improvement Committee is actively involved in research collaborations and provides MDHHS guidance on screening protocols. CF newborn screening identifies over 30 babies each year and offers a greater chance for improved quality of life and increased survival for children affected with this disease.

*“Before NBS we would sweat test patients months or years after they exhibited symptoms and that would put them so far behind in therapies and treatment. Now many of them thrive from the beginning and do so well!”*

– Paulette Ratkiewicz,  
CF sweat testing  
technician



## Endocrine Disorders

The Michigan NBS panel includes two endocrine disorders, congenital hypothyroidism (CH) and congenital adrenal hyperplasia (CAH). CH is one of the most commonly detected NBS disorders, accounting for about 90 cases annually or 36% of all newborns identified through screening. It affects the body's ability to produce thyroid hormone, and can lead to intellectual disability and poor growth if not treated with thyroid hormone replacement. CAH affects the adrenal glands and hormones needed to help protect the body during stress or illness. Treatment for classic CAH may include steroids to replace low hormones and surgery for girls born with ambiguous genitalia. Follow-up for babies with positive screens is carried out by the Pediatric Endocrine Coordinating Center at the University of Michigan, and patients are managed by pediatric endocrinologists across the state. A Pediatric Endocrine Advisory Council (PEAC) was formed under the leadership of Dr. Nancy Hopwood and the University of Michigan, Division of Pediatric Endocrinology in 1987 to establish a statewide group of board certified pediatric endocrinologists who now provide oversight and recommendations to MDHHS for the screening, diagnosis and medical management of CH and CAH.

*"Under the direction of Bill Young, the laboratory and follow-up program collaborated to implement the NICU protocol in 2007 which has led to the detection of babies with congenital hypothyroidism who otherwise would have gone undetected by newborn screening."*

– Karen Andruszewski  
and Caron Burns



## Metabolic Disorders

Metabolic Disorders, also called Inborn Errors of Metabolism (IEM), account for the majority (~80%) of conditions on the NBS panel. This category includes the disease that started it all, PKU. The Children's Hospital of Michigan Metabolic Clinic (CHMMC) located in Detroit is the designated medical management coordinating center providing diagnosis and long term follow-up for children identified by NBS who have amino acid, organic acid and fatty acid oxidation disorders as well as biotinidase deficiency and galactosemia. Individually the disorders are relatively rare, but together about 70 children are identified each year by NBS. While the features of IEM disorders vary depending on the specific condition, they typically affect enzymes involved in breaking down food to make energy for the body. If not diagnosed promptly, they can lead to a variety of serious health problems, intellectual disability, coma or death. Lifelong nutritional treatment in the form of a special diet, vitamins and/or supplements is required for most metabolic disorders; and weekly to monthly monitoring of blood levels may be needed to assure proper diet restrictions are in place. The Metabolic Quality Improvement Committee includes biochemical genetics experts from the University of Michigan in addition to CHMMC, and provides MDHHS with guidance on addition of new metabolic disorders to the NBS panel as well as individual case follow-up.



*"Without newborn screening and everyone involved in those first crucial hours, my life would have turned out very differently."*

– Mike Finkel,  
Young Adult with PKU

## Primary Immunodeficiencies

Screening for primary immunodeficiency disorders began in 2010. This group of diseases includes Severe Combined Immunodeficiency Disease (SCID). There are multiple genetic mutations that cause different forms of SCID or other immunodeficiency syndromes, all of which involve abnormal production of antibodies needed to fight infection. About 15 babies are found with some type of primary immunodeficiency each year. Early identification allows for timely treatment leading to better health outcomes. The NBS Primary Immunodeficiency Coordinating Center at Children's Hospital of Michigan helps to assure diagnostic confirmation and referral for treatment which may involve hematopoietic stem cell transplant at CHM, Helen DeVos Children's Hospital, or University of Michigan. The Michigan Primary Immunodeficiency Disorders Quality Improvement Committee was established prior to implementation of screening and currently meets twice a year to review laboratory and clinical services, and propose strategies and policies related to primary and secondary immunodeficiencies that may be detected in Michigan newborns. Specialists representing the fields of allergy and immunology, hematology-oncology and infectious disease serve on the committee.

*"I was shocked, in disbelief. The newborn screening found our daughter had SCID. Due to early detection, she had a bone marrow transplant before ever developing a severe infection. Early detection greatly increased her chance for survival. It saved her life."*

– Jenna Heady,  
Mother of a daughter with  
SCID



## Sickle Cell and Other Hemoglobinopathies

Each year, the NBS Program identifies about 60 newborns with sickling conditions or other hemoglobinopathies and approximately 2,700 infants who are not affected but carry sickle cell trait. Sickle cell disease (SCD) is the most common inherited blood disorder in the United States. The condition affects the shape of red blood cells, leading to anemia and increased susceptibility to infections. Some of the other possible complications include severe episodes of pain, stroke, vision loss, pulmonary embolism and damage to the spleen. Timely identification through screening aims to prevent death from infections in early childhood, and to initiate disease-modifying therapies such as hydroxyurea to reduce the risk of future complications. Founded by Dr. Charles Whitten, the Sickle Cell Disease Association of America, Michigan Chapter (SCDAA-MI) has coordinated confirmatory diagnosis and follow-up since 1987 for newborns through age five with hemoglobinopathies detected by NBS. The SCDAA-MI, located in Detroit with patient advocates serving Ann Arbor, Benton Harbor, Flint, Grand Rapids, Jackson, Kalamazoo, Lansing, Pontiac and Saginaw, helps to assure that all newborns with a confirmed diagnosis of SCD receive penicillin prophylaxis and have access to sickle cell counseling, social work services and a medical home to provide ongoing treatment. Hematologists and other medical experts participate in the Hemoglobinopathy Quality Improvement Committee to advise MDHHS on screening and follow-up for sickle cell and related disorders.

*“From my perspective, the biggest benefit for Michigan families from newborn sickle cell screening is the early initiation of penicillin treatment to prevent life threatening infections. Prior to newborn screening one of the ways sickle disease was diagnosed was when a baby presented to the emergency room dead on arrival from a devastating infection called pneumococcal sepsis. Diagnosing infants at birth also allows for early education and support services. Families are taught by Patient Advocates from the Michigan Chapter of SCDAA how to recognize complications and the steps to take should they arise.”*

– Dr. Wanda Whitten Shurney,  
Chief Executive Officer and  
Medical Director, SCDAA-MI

# Point of Care Screening

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In addition to the disorders detectable through blood spot screening, Michigan newborns are also screened for hearing loss and critical congenital heart disease. These “point of care” screens are performed in the hospital or by midwives attending home births.

Ninety eight percent of infants have a hearing screening completed in the hospital prior to discharge. Hearing in infants can be tested using two different methods; the auditory brainstem response or the otoacoustic emission measures. Both tests are accurate, noninvasive, automated and do not require any observable response from the infant. About 150-160 babies are identified with hearing loss each year through newborn hearing screening.

Hospitals and midwives also administer pulse oximetry screening to detect low oxygen levels in the blood that might indicate certain kinds of congenital heart disease. The procedure uses a small sensor placed on a baby’s right hand and one foot. It is fast, easy and noninvasive.

Even though the hearing and CCHD screens are performed by hospital staff or midwives, all results are submitted to the state Newborn Screening Program. MDHHS plays an important public health assurance function, providing education on proper screening techniques and making sure every baby not only receives both point of care screens but also receives any follow-up that may be needed.

## Hearing Loss

The Michigan Early Hearing Detection and Intervention (EHDI) Program began in 1997, with statewide screening in place by 2007. The EHDI Program goals are to provide better outcomes for Michigan newborns and young children with hearing loss and their families, through early hearing screening, appropriate audiological diagnosis and intervention. The EHDI Program works in collaboration with hospitals, clinics, parents, midwives and audiologists to identify infants with hearing loss and assist families with support services. Once identified, EHDI follows these infants to ensure enrollment in early intervention services to help strive toward achieving the national EHDI 1-3-6 goals. The national EHDI goals are:

- Goal 1:** All newborns will be screened for hearing loss no later than 1 month of age, preferably before hospital discharge;
- Goal 2:** All infants who screen positive for hearing loss will have a diagnostic audiologic evaluation no later than 3 months of age;
- Goal 3:** All infants identified with hearing loss will receive appropriate early intervention services no later than 6 months of age.

Early intervention is important to help each child develop communication and to give families information. Many families choose *Early On*® Michigan to help with family centered coordinated services. Services may be provided in the family's home or professional offices after a hearing loss is found. Other family support is available through the Guide By Your Side™ program from Michigan Hands & Voices™, that provides services for families with infants and young children who are deaf or hard of hearing.

*"My daughter was diagnosed close to birth and has had hearing aids and language support from the get-go, thanks to those wonderful newborn hearing screenings!"*

– Kim Williamson,  
Mother of a child with  
hearing loss



## Critical Congenital Heart Disease

More than 1,700 Michigan babies are born with congenital heart disease each year. Some forms of congenital heart disease in the newborn are detectable by point of care pulse oximetry screening. This screening targets twelve specific anomalies classified as critical congenital heart disease (CCHD). Failure to detect such heart defects while in the hospital puts the baby at risk for serious complications within the first few days or weeks of life. Costly emergency room care, potential permanent disability and even death may be the result of delayed treatment.

In 2012, MDHHS received a 3-year grant from the federal Health Resources and Services Administration to develop a CCHD Newborn Screening Demonstration Program. The goals were to: 1) increase the number of Michigan newborns screened for CCHD using a validated screening protocol; and 2) to develop state infrastructure for collection of CCHD screening data through electronic health information exchange to enable effective public health follow-up, quality assurance and evaluation.



Grant funding enabled expansion of the program to all birthing hospitals, and effective April 1, 2014, CCHD was added to the mandated newborn screening panel so that all Michigan newborns are now screened. The Newborn Screening Program and the CCHD Advisory Committee recommend that newborns be screened as close to 24 hours of age as possible, using the approved MDHHS CCHD Screening Algorithm prior to hospital discharge or following a home birth.

# Michigan BioTrust for Health



The *Michigan BioTrust for Health*, launched in June 2009, is a pioneering program that oversees the storage and use of NBS blood spots. Research using blood spots left-over from newborn screening offers an added public health benefit beyond identifying babies in need of early treatment. The BioTrust allows release of blood spots for medical and public health research after a thorough review and approval process. The samples are important for research because they can provide a population-based snapshot of infants born during a time period and contain over 160 different biomarkers that could provide clues for finding and treating disease. Blood spots from about 5 million individuals dating back to July 1984 have been preserved by the BioTrust program, with over 70,000 samples added each year. After all identifying information is removed and the spots are labelled only with a numerical code, they are stored indefinitely at the Michigan Neonatal Biobank (MNB) in Detroit, managed by Wayne State University. The MNB is a non-profit repository with temperature controls as well as privacy and security protections.

Michigan has led the nation with the first parent consent process for research use of residual NBS blood spots. Since May 2010, parents decide whether their newborn's blood spots can be used in future health research. Blood spots collected before May 2010 are available for research use but can be removed from the BioTrust by a parent or person over the age of 18 years contacting MDHHS to opt-out. Michigan was also the first state to convene a *Community Values Advisory Board* (CVAB), with representation from diverse community and advocacy organizations. Along with the MDHHS Institutional Review Board and BioTrust Scientific Advisory Board, the CVAB provides guidance that helps ensure proper research use of blood spots.

Since 2009, the *BioTrust* has approved approximately 40 studies for use of Michigan dried blood spots. In addition, they have been used by individual families for clinical testing, molecular autopsy and research. They are also used by the laboratory to continue to improve and expand newborn screening so that more babies will benefit from early detection and life-saving treatment.



# The Future: Looking Ahead

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The last half century has brought incredible changes to newborn screening, with many improvements and advances in technology occurring just in the last decade. This recent, quickening expansion is only expected to continue in coming years. At the end of 2015, Pompe disease will be the first lysosomal storage disorder added to the Michigan panel, with others likely to follow.

The use of genome sequencing as part of newborn screening has been discussed and debated nationally. While sequencing could potentially provide a complete molecular picture of the newborn, the clinical and ethical implications of such technology are not known. Therefore, research studies are underway nationally to evaluate the promise—and challenges—of using whole genome or exome sequencing for newborn screening.

The Michigan Newborn Screening Program's five decades of success is due to constant improvements and incorporation of new technologies throughout the years. With the continuation of these efforts, we will strive towards another 50 years of improving health outcomes for newborns. Our hope for the future is that even more babies and their families will reap the benefits of early detection and treatment to prevent disability and death from rare disorders.





# For more information:

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