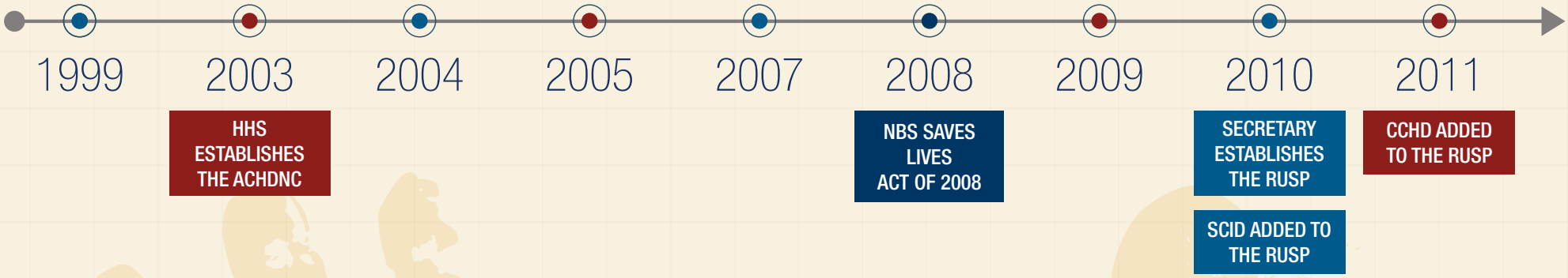
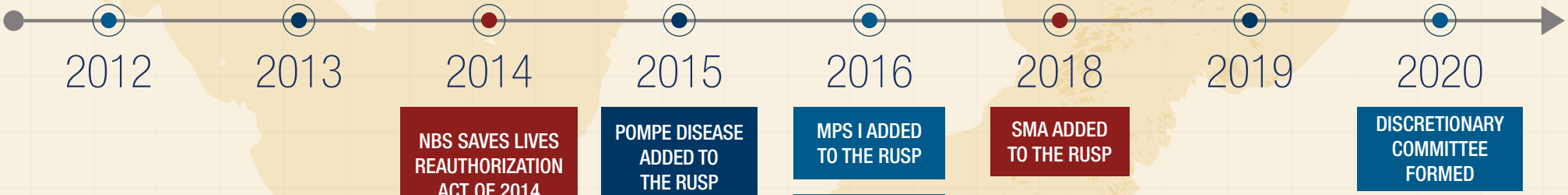


HISTORY OF THE ACHDNC

The Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) advises the Secretary of Health and Human Services on newborn screening conditions, tests, technologies, policies, guidelines, and standards. The ACHDNC also reviews evidence and makes recommendations to the Secretary about which conditions should be included on a uniform screening panel that all states are encouraged to adopt. This document presents the history of the ACHDNC. Click on each year to find out more about the history of the ACHDNC.



ACHDNC
Advisory Committee on Heritable Disorders in Newborns and Children

HISTORY OF THE ACHDNC

1999
NBS TASK FORCE REPORT

The Health Resources and Services Administration's Maternal and Child Health Bureau partnered with the American Academy of Pediatrics to form a national Task Force on Newborn Screening. The purpose of the Task Force was to review issues and challenges for state newborn screening programs. Five workgroups collected information and presented findings to the full Task Force in May 1999. The Task Force made the following four recommendations:

1. Effective newborn screening systems need an adequate public health infrastructure and must be a part of the health care delivery system.
2. Public health agencies must involve health professionals, families, and the general public in the development, operation, and oversight of newborn screening systems.
3. Public health agencies must ensure adequate infrastructure and policies for surveillance and research related to newborn screening.
4. Public health agencies should ensure adequate funding to support a newborn screening program.

2003
ACHDNC ESTABLISHED

In 2003, the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) was created under Section 1111 of the Public Health Service Act, 42 U.S.C. 300b-10 as amended in the Newborn Screening Saves Lives Act of 2007 (Act) to advise the Secretary of Health and Human Services about newborn and childhood screening. ACHDNC comprises up to 15 members, including

- Medical, scientific, and public health experts in heritable disorders
- Experts in ethics
- Members of the public with expertise or concern with heritable disorders
- Representatives from federal agencies, medical societies, and public health groups

2004
FIRST ACHDNC MEETING

The first meeting of the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) was held June 7–8, 2004. Thirteen members attended the first meeting. The members received presentations and briefings covering topics such as the following:

- An overview of state newborn screening programs
- Roles and activities of federal agencies and other Federal Advisory Committees in newborn screening
- Standardization of guidelines and practices for newborn screening programs
- Delivery of genetic services to children in a clinical setting

Eleven individuals from industry, family advocacy organizations, parents, community-based organizations, and research groups delivered public comments.

At the end of the first meeting, ACHDNC identified its three highest priorities:

- Consideration of a uniform panel
- Future research agenda
- Emerging technologies

Related LINK:

- Meeting Notes of the ACHDNC

HISTORY OF THE ACHDNC

2005
ACMG EXPERT PANEL

In response to the recommendations of the 1999 Task Force on Newborn Screening, the Health Resources and Services Administration contracted with the American College of Medical Genetics (ACMG). ACMG was asked to analyze scientific literature and collect expert opinions on newborn screening. The goal was to use the findings to make recommendations for newborn screening, including a uniform panel of conditions.

ACMG released its report in 2005. The proposed Core Condition Panel contained 29 conditions, and 25 more conditions were listed as secondary targets. The report also described the specific steps in the newborn screening process that should be monitored. It also recommended a uniform approach to data collection and program evaluation, to help improve comparison of programs across states.

Related LINKS:

- Newborn Screening: Toward a Uniform Screening Panel and System
- Newborn Screening: Toward a Uniform Screening Panel and System – Executive Summary, as published in Pediatrics 117 (Supplement 3), May 2006

2007
PRENATAL EDUCATION

Educating parents about newborn screening during the prenatal period is important because it gives them time to understand and discuss the benefits of newborn screening prior to birth. The Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) recommended that the Secretary of Health and Human Services develop and fund more studies about what information expectant parents receive about newborn screening. They also recommended studying how parents learn about newborn screening from their health care providers.

Related LINK:

- ACHDNC Chair Letter to the Secretary of Health and Human Services

2008
NBS SAVES LIVES ACT OF 2008

The Newborn Screening Saves Lives Act of 2007 funded federal, state, and local newborn screening activities. It aimed to improve education, outreach, follow-up, and laboratory quality and surveillance in newborn screening. Congress passed the Act in December 2007, and the President signed it in April 2008. The Act required the Advisory Committee on Heritable Disorders in Newborns and Children to make recommendations on conditions for newborn screening. It also required the following:

- An information clearinghouse
- Quality assurance and control mechanisms for laboratories and testing tools
- Establishment of an Interagency Coordinating Committee on Newborn and Child Screening
- Development of a national plan for newborn screening in the event of a public health emergency
- Authorization for continued and expanded newborn screening research

ACMG-RECOMMENDED SCREENING PANEL ADOPTED

On September 9, 2005, the Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) wrote a letter to the Secretary of Health and Human Services to recommend the use of a uniform panel for state newborn screening programs. The ACHDNC also recommended that newborn screening include follow-up, diagnosis, management and treatment, evaluation, and education. On October 21, 2008, the Secretary adopted the ACMG recommendations.

Related LINKS:

- ACHDNC Chair Letter to the Secretary of Health and Human Services
- Response Letter from the Secretary of Health and Human Services

HISTORY OF THE ACHDNC

2009
MEDICAL FOODS NOT COVERED BY INSURANCE

Newborn screening helps to identify children with serious health conditions who require early treatment. Some of these children require special medical foods. These foods are expensive, and it can be hard for parents to afford them. Some government programs help families pay for medical foods. Unfortunately, not all families qualify to receive this support. Health insurance sometimes helps to cover some medical foods, but often, it is not enough.

The Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) recommended that the Secretary of Health and Human Services change the rules on health insurance coverage of medical foods. They also suggested that State Medicaid programs cover medical foods. However, the Department of Health and Human Services did not have the authority to make these changes.

Related LINKS:

- ACHDNC Chair Letter to the Secretary of Health and Human Services
- Enclosure Tab A: List of State Laws
- Enclosure Tab B: Table of Conditions and Medical Foods
- Enclosure Tab C: Review of Treatment of PKU
- Response Letter from the Secretary of Health and Human Services

2010
RUSP ESTABLISHED

In May 2010, the Secretary of Health and Human Services accepted the recommendation of the Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) to create a Recommended Uniform Screening Panel (RUSP). The RUSP is a list of disorders that the Secretary recommends for states to screen as part of their state universal newborn screening programs. The RUSP includes a set of both core and secondary conditions (i.e., disorders that may be found while screening for core conditions). In 2010, the RUSP comprised 29 core conditions and 25 secondary conditions. Since then, additional conditions have been added to the RUSP. For information on how to nominate a condition for inclusion on the RUSP, please reference the ACHDNC's Nominate a Condition page.

The Secretary of Health and Human Services recommends screening every newborn for all heritable disorders on the RUSP, but each state decides which disorders to include in its newborn screening program. Most states screen for the majority of disorders, and some also screen for disorders not included on the RUSP.

Related LINKS:

- ACHDNC Chair Letter to the Secretary of Health and Human Services
- Response Letter from the Secretary of Health and Human Services

2010
SCID ADDED TO THE RUSP

Severe combined immunodeficiency (SCID) is a genetic disorder that makes the body unable to fight off infections. White blood cells are needed to fight off infections. People with SCID have either too few white blood cells or white blood cells that do not work properly. Signs of SCID include diarrhea, repeated infections, poor weight gain, and thrush. If left untreated, SCID can lead to early death. Early diagnosis and treatment can improve outcomes.

On February 25, 2010, the Advisory Committee on Heritable Disorders in Newborns and Children recommended that SCID be added to the Recommended Uniform Screening Panel (RUSP). The Secretary of Health and Human Services accepted the recommendation on May 21, 2010. At the same time, the Secretary requested a report on the status of states' implementation of screening for SCID, which was provided on May 19, 2011.

Related LINKS:

- ACHDNC Chair Letter to the Secretary of Health and Human Services
- Enclosure: Evidence-Based Review: Newborn Screening for SCID
- Response Letter from the Secretary of Health and Human Services

HISTORY OF THE ACHDNC

2010
HEALTH CARE REFORM AND NBS

In March 2010, the Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) wrote a White Paper on how health care reform might improve quality, efficiency, and access to newborn screening programs. As a result, on March 23, 2010, the ACHDNC recommended four actions to address barriers to improving the newborn screening system. The Secretary of Health and Human Services responded on September 23, 2010, and accepted three of the four recommendations. The recommendations included encouraging the Centers for Medicare and Medicaid Services to:

- Streamline the billing process for newborn screening services and standardize health transactions
- Develop a payment method for care coordination through the medical home framework
- Adopt and further define the use of the Newborn Screening Use Case for health information exchange endeavors

The fourth recommendation was not accepted. This recommendation was about closing gaps in insurance coverage for medical foods and foods modified to be low in protein. The topic was addressed in more detail in December 2010.

Related LINKS:

- ACHDNC Chair Letter to the Secretary of Health and Human Services
- Response Letter from the Secretary of Health and Human Services

2010
CLIAC RECOMMENDATIONS

On August 25, 2010, the Centers for Disease Control and Prevention (CDC) wrote a letter to the Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) on the Clinical Laboratory Improvement Advisory Committee (CLIAC) recommendations for good laboratory practices for newborn screening. The recommended practices addressed the following:

- The benefits of using a quality management system approach
- Factors to consider before introducing new tests
- Establishment and verification of test performance specifications
- An overview of the laboratory testing process
- Confidentiality of patient information and test results
- Personnel qualifications and responsibilities for laboratory testing for inherited metabolic diseases

Related LINKS:

- Letter from CDC to the ACHDNC
- ACHDNC Chair Letter to the CDC, Centers for Medicare & Medicaid Services, and Food and Drug Administration
- CLIAC Recommendations for Good Laboratory Practices
- Morbidity and Mortality Weekly Report on Good Laboratory Practices for Biochemical Genetic Testing and Newborn Screening for Inherited Metabolic Disorders

2010
INSURANCE COVERAGE OF MEDICAL FOODS

In 2010, when the Department of Health and Human Services (HHS) was planning the Affordable Care Act, the Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) asked HHS to require that all types of health insurance cover medical foods and foods modified to be low in protein. These products can be expensive for families who need them to treat their child's metabolic disorder. The ACHDNC also wanted people of all ages with one or more of the conditions on the Recommended Uniform Screening Panel (RUSP) to be considered high-risk and to be guaranteed access to comprehensive health care coverage.

At that time, the Secretary of Health and Human Services was in the process of gathering information to determine essential health benefits and could not accept the Committee's recommendation. The Secretary indicated that the information provided would be considered in determining essential health benefits.

Related LINKS:

- ACHDNC Chair Letter to the Secretary of Health and Human Services
- Response Letter from the Secretary of Health and Human Services

HISTORY OF THE ACHDNC

2011
SICKLE CELL TRAIT

People who have one sickle cell gene and one normal gene have sickle cell trait (SCT). It is important for people with SCT to know they have the condition. People with SCT are more likely to experience heat stroke and muscle breakdown during strenuous exercise. The National Collegiate Athletic Association and National Athletes Trainers' Association recommended SCT screening for Division 1 athletes to reduce the number of training-related deaths.

On October 11, 2010, the Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) provided five recommendations related to SCT and athletes. On July 27, 2011, three of the five recommendations were accepted by the Secretary of Health and Human Services:

1. All individuals should have the opportunity to know their risk for various medical disorders, including SCT.
2. Genetic testing should not be a prerequisite for participation in sports, unless medically necessary.
3. All athletes should receive education on prevention of heat-related illnesses.

Related LINKS:

- ACHDNC Chair Letter to the Secretary of Health and Human Services
- Response Letter from the Secretary of Health and Human Services
- Journal article: Screening U.S. College Athletes for Their Sickle Cell Disease Carrier Status

2011
NATIONAL CONTINGENCY PLAN FOR NBS

Many organizations have developed contingency plans to ensure the continuity of operations in case of an emergency. The Newborn Screening Saves Lives Act of 2007 required the development a national contingency plan for newborn screening. The Centers for Disease Control and Prevention, the Health Resources and Services Administration, and state health departments worked together to develop a national newborn screening contingency plan. In May 2010, the Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) reviewed and approved the plan. On August 6, 2010, the ACHDNC recommended that the Secretary of Health and Human Services use the plan to coordinate newborn screening emergency preparedness activities. The Secretary responded on September 2, 2011, with acceptance of the recommendation.

Related LINKS:

- ACHDNC Chair Letter to the Secretary of Health and Human Services
- Response Letter from the Secretary of Health and Human Services

2011
CCHD ADDED TO THE RUSP

Congenital heart disease (CHD) is a general term describing a range of symptoms resulting from heart defects that are present at birth. These varied congenital defects change the normal flow of blood through the heart, leading to a range of conditions and symptoms. Critical congenital heart disease (CCHD) is a group of defects that cause severe and life-threatening symptoms. CCHD requires intervention within the first days or first year of life. Critical congenital cyanotic heart disease (CCCHD) is a type of CCHD that causes babies to have a low level of oxygen in their blood. Blood oxygen levels can be identified using pulse oximetry. CHD is the most common cause of death in the first year of life.

On October 15, 2010, the Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) recommended that CCCHD be added to the RUSP. However, the Secretary of Health and Human Services approved the addition of CCHD on September 21, 2011.

Related LINKS:

- ACHDNC Chair Letter to the Secretary of Health and Human Services
- Enclosure: Evidence-Based Review: Newborn Screening for CCCHD
- Response Letter from the Secretary of Health and Human Services
- Journal Article: Strategies for Implementing Screening for Critical Congenital Heart Disease

HISTORY OF THE ACHDNC

2012
POINT OF CARE SCREENING IMPLEMENTED

Point-of-care screening tests are conducted where patients receive medical care. This screening is different from most newborn screenings, which use a laboratory to perform tests. An example of point-of-care newborn screening is testing for congenital heart disease using a pulse oximeter.

In 2012, the Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) wrote a report providing guidance to state public health agencies, doctors, and hospitals on using point-of-care newborn screening. ACHDNC developed this information to help ensure high-quality, universal access to newborn screening; establish standards of care; and provide mechanisms for effective diagnosis, intervention, and follow-up care.

Related LINKS:

- ACHDNC Chair Letter to the Secretary of Health and Human Services
- Response Letter from the Secretary of Health and Human Services

2013
ACHDNC ADOPTS NEW DECISION MATRIX

The Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) adopted a new approach for recommending new conditions to be added to the Recommended Universal Screening Panel in January 2013. Using the new approach, the ACHDNC would consider the range of expected benefits and risks of screening for each condition, along with the readiness of state newborn screening programs to provide comprehensive screening for the conditions. The range of expected benefits and risks and the readiness to implement each receive a rating, and the two ratings are combined into a single score that serves as the basis for the ACHDNC-recommended action.

Related LINKS:

- Decision Matrix
- Users' Guide to the SACHDNC Decision Matrix

2013
ACHDNC ESTABLISHED AS DISCRETIONARY ADVISORY COMMITTEE

On April 24, 2013, the Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) was re-established as a discretionary committee. The ACHDNC continued to advise the Secretary of Health and Human Services about newborn screening.

LINKING NBS TO BIRTH CERTIFICATES

Matching newborn screening results with birth certificate information is an important quality assurance measure. However, it is often challenging because there may be spelling or recording errors with a baby's name on the hospital records, and newborns' names are often not finalized at the time the screening specimen is submitted. Thus, the matching process can be time consuming and labor intensive.

To help LINK this information, in March 2012, the Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) recommended including the newborn dried bloodspot specimen's serial number on the birth certificate. However, after reviewing the recommendations, the Secretary of Health and Human Services could not endorse them for the following reasons:

- Newborn screening quality assurance is up to each state.
- The opportunity to add a field to the U.S. Standard Certificate of Live Birth would not be open until 2019.
- The ability to use a unique state-specific serial number will vary from state to state.
- There are questions about the security of a serial number and personally identifiable information.
- Endorsing this method might limit the creativity of states in using alternative approaches.

As a result, on August 21, 2013, the recommendation was rejected.

Related LINKS:

- ACHDNC Chair Letter to the Secretary of Health and Human Services
- Journal Article: Improving Data Quality and Quality Assurance in Newborn Screening by Including the Bloodspot Screening Collection Device Serial Number on Birth Certificates
- Response Letter from the Secretary of Health and Human Services

HISTORY OF THE ACHDNC

2013
RESIDUAL DRIED BLOOD SPOTS

A dried bloodspot specimen is blood collected from a person and dried on filter paper for testing and analysis. Laboratories use this process to conduct newborn screening.

In 2010, the Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) wrote a paper describing its concerns with the use and storage of dried bloodspot specimens. The paper had two main purposes: First, the ACHDNC wanted to review the ethical, legal, and social issues related to dried bloodspot specimens; it also wanted to discuss the importance of educating the public about the process for taking a dried blood specimen sample and financial considerations of storing these samples. Second, the ACHDNC hoped the paper would help lead to the creation of national guidance for states about dried bloodspot samples.

The ACHDNC submitted to the Secretary of Health and Human Services this paper and eight recommendations to help guide state newborn screening programs in their use and storage of dried bloodspots. After review in 2013, the Secretary of Health and Human Services accepted four of the ACHDNC's recommendations addressing state and federal initiatives to educate newborn screening stakeholders and start a national dialogue among stakeholders.

Related LINKS:

- ACHDNC Chair Letter to the Secretary of Health and Human Services
- Journal article: Considerations and Recommendations for National Guidance Regarding the Retention and Use of Residual Dried Blood Spot Specimens after Newborn Screening
- Response Letter from the Secretary of Health and Human Services

2014
PUBLIC HEALTH IMPACT ASSESSMENTS CREATED

The Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) established an Expert Advisory Panel to make recommendations for the development of a public health system impact survey instrument. The ACHDNC developed the tool to assess the feasibility and readiness of states to start screening for a new condition. The Office of Management and Budget approved the tool. The tool is administered to states to:

- Assess the state's ability to screen for the condition
- Assess the availability of follow-up diagnostic and clinical referrals
- Provide an estimate of how long it would take to states to start testing once the decision was made to screen for the condition

Related LINK:

- Presentation and Survey: Assessing Public Health System Impact

2014
NBS SAVES LIVES REAUTHORIZATION ACT OF 2014

In 2014, the passage of the Newborn Screening Saves Lives Reauthorization Act of 2014 extended the Newborn Screening Saves Lives Act of 2008. The 2014 Act focused on improving screening, counseling, and other services related to heritable disorders detectable in newborns. The 2014 Act stated that funding should be used to help make the newborn screening process quicker and to train health care professionals to provide results to families in a timely manner.

Some of the other important parts of the 2014 Act included the following:

- Continues the grant program to evaluate the effectiveness of newborn screening
- Authorizes the Advisory Committee on Heritable Disorders in Newborns and Children for another 5 years (until September 2019)
- Continues the Hunter Kelley Newborn Screening Research Program to continue research on disorders that could be added to the Recommended Uniform Screening Panel
- Continues the clearinghouse for newborn screening to maintain information on the number of conditions that each state screens for and to share guidelines for conditions detected by newborn screening
- Continues the requirements for quality assurance of laboratories involved in newborn screening
- Continues the Interagency Coordinating Committee on Newborn and Child Screening
- Requires the Comptroller General within the Government Accountability Office to report on the timeliness of newborn screening and the Secretary of Health and Human Services to report on newborn screening activities and expenditures
- Requires the national contingency plan for newborn screening be updated at least every 5 years
- Directs the Department of Health and Human Services to update the Common Rule and require parental consent before residual dried bloodspots from newborns are used in research

HISTORY OF THE ACHDNC

2015
USPSTF TRANSFERRED NBS TOPICS TO THE ACHDNC

The U.S. Preventive Services Task Force (USPSTF) is an independent, volunteer panel of national experts who make evidence-based recommendations about clinical preventive services such as screenings, counseling services, and preventive medications. In 2015, the USPSTF decided to refer its newborn screening topics, including sickle cell disease, phenylketonuria (PKU), and congenital hypothyroidism, and any future newborn screening topics to the Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC). USPSTF recommendations are based on a rigorous review of existing peer-reviewed evidence, so the decision to refer these newborn screening topics to the ACHDNC recognizes the rigor of the ACHDNC's evidence-based review process.

2015
POMPE DISEASE ADDED TO THE RUSP

Pompe disease is an inherited lysosomal storage disorder. In the body, lysosomes help recycle cell substances. In people with Pompe disease, lysosomes cannot break down certain sugars. These sugars and other substances build up in the cells, causing damage. There are three forms of Pompe disease characterized by severity and age of onset. Symptoms vary based on type, but can include muscle weakness, failure to grow and gain weight, and difficulty breathing. Early diagnosis and treatment can lead to better outcomes.

On June 3, 2013, the Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) recommended adding Pompe disease to the Recommended Uniform Screening Panel (RUSP). The Secretary of Health and Human Services accepted the recommendation to add Pompe disease to the RUSP on March 2, 2015.

Related LINKS:

- ACHDNC Chair Letter to the Secretary of Health and Human Services
- Enclosure: Evidence-Based Review: Newborn Screening for Pompe Disease
- Response Letter from the Secretary of Health and Human Services

2015
SUAC AS PRIMARY MARKER TO DETECT TYR 1

Tyrosinemia is a genetic disorder resulting in the inability of the body to break down the amino acid, tyrosine. Tyrosinemia type 1 (TYR 1) is the most severe form of tyrosinemia, with symptoms appearing within the first few months of life. TYR 1 may affect the brain, liver, and kidneys and can result in early death if left untreated. Research shows that treatment is most effective when started within the first month of life. Thus, TYR 1 is included in the core conditions of the Recommended Uniform Screening Panel. Most newborn screening programs screen for TYR 1.

Commonly, TYR 1 is detected by measuring the level of tyrosine in the blood. However, elevated tyrosine levels are not specific to TYR 1 and can result in false negative and false positive test results. Research has determined that succinylacetone (SUAC) is a better indicator for TYR 1. By using SUAC as a primary marker for TYR 1, false negative and false positive test results can be decreased.

In 2014, the Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) recommended that the Secretary of Health and Human Services facilitate a national dialogue among federal and state stakeholders on the benefits of measuring SUAC in dried bloodspots for TYR 1.

On March 16, 2015, the Secretary of Health and Human Services accepted the recommendation. The Secretary of Health and Human Services also encouraged a discussion on a more consistent implementation of screening for TYR 1.

Related LINKS:

- ACHDNC Chair Letter to the Secretary of Health and Human Services
- Journal article: Succinylacetone as primary marker to detect tyrosinemia type I in newborns and its measurement by newborn screening programs
- Response Letter from the Secretary of Health and Human Services
- Journal article: Newborn screening for Tyrosinemia type 1 using succinylacetone – a systematic review of test accuracy

HISTORY OF THE ACHDNC

2015
TIMELY NEWBORN SCREENING GOALS

Collecting and transporting dried bloodspots and reporting test results in a timely manner are critical steps to newborn screening. In 2005, the Newborn Screening: Toward a Uniform Screening Panel and System Report made four recommendations concerning timeliness for the newborn screening system. By 2015, many states were still not meeting these recommendations.

In 2015, the Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) developed three specific goals to improve the timeliness of the newborn screening system. The ACHDNC proposed that newborn screening programs strive to meet the following timelines:

1. Presumptive positive results for time-critical conditions should be communicated immediately to the newborn's health care provider, but no later than 5 days of life.
2. Presumptive positive results for all other conditions should be communicated to the newborn's health care providers as soon as possible, but no later than 7 days of life.
3. All newborn screening tests should be completed within 7 days of life, with results reported to the health care provider as soon as possible.

To meet these goals, initial newborn screening specimens should be collected no later than 48 hours after birth. The specimens should be received at the laboratory as soon as possible, ideally within 24 hours of collection.

States are not required to meet these goals, but ACHDNC suggested that states aim to meet these goals for 95% or more of newborns by 2017. To support these efforts, the Health Resources and Services Administration awarded funding through a cooperative agreement to support states and help implement strategies to improve timeliness and share best practices.

Related LINKS:

» ACHDNC Chair Letter to the Secretary of Health and Human Services
» Response Letter from the Secretary of Health and Human Services

2015
INFORMED CONSENT FOR NBS

Residual newborn screening dried bloodspots are often stored by state health departments after the initial tests are completed. Recent lawsuits showed that some states used these dried bloodspots without parental consent for more research. Amendment 12 of the Newborn Screening Saves Lives Reauthorization Act of 2014 requires parental consent before residual dried bloodspots from newborn screening are used in research. The Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) determined that more guidance was needed for state newborn screening programs on how to implement this amendment. On June 23, 2015, the ACHDNC provided six recommendations to the Secretary of Health and Human Services. After review, the Secretary of Health and Human Services agreed to one of the recommendations. The recommendation was to create and distribute materials on the importance of newborn screening. The materials would include information on options for parents to take part in newborn screening research.

Related LINKS:

» ACHDNC Chair Letter to the Secretary of Health and Human Services
» Response Letter from the Secretary of Health and Human Services

2016
MPS I ADDED TO THE RUSP

Mucopolysaccaridosis type I (MPS I) is an inherited lysosomal storage disorder. In the body, lysosomes help recycle cell substances. In people with MPS I, lysosomes are not able to break down certain sugars. These sugars and other substances build up in cells. As a result, the cells cannot perform properly and cause damage to the body. There are two forms of MPS I: severe and attenuated. Symptoms include developmental delays and learning disabilities, large head, clouding of the eyes, and swollen abdomen. Early diagnosis and treatment can lead to better outcomes.

On April 13, 2015, the Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) recommended that MPS I be added to the Recommended Uniform Screening Panel (RUSP). The ACHDNC also recommended that federal funding be provided to states to start screening for MPS I. The Secretary of Health and Human Services accepted the recommendation to add MPS I to RUSP on February 16, 2016. However, the Secretary was unable to provide funding to states to implement screening for this condition.

Related LINKS:

» ACHDNC Chair Letter to the Secretary of Health and Human Services
» Enclosure: Evidence-Based Review: Newborn Screening for MPS I
» Final Response Letter from the Secretary of Health and Human Services

HISTORY OF THE ACHDNC

2016
X-ALD ADDED TO THE RUSP

X-LINKed adrenoleukodystrophy (X-ALD) is an inherited disorder. It occurs when the body cannot break down certain fats called very long–chain fatty acids (VLCFA). When VLCFA build up, it affects the nervous system and the adrenal glands. These effects reduce the ability of the nerves to send information to the brain and the adrenal gland to produce certain hormones. There are three types of X-ALD. Symptoms vary based on the type of X-ALD and age of onset. Symptoms can include learning and behavior problems, muscle weakness, seizures, and hearing and vision problems. The condition affects more males than females and can result in early death if not treated early. Available treatments depend on type of X-ALD.

Early diagnosis is important for early treatment of this condition. On September 25, 2015, the Advisory Committee on Heritable Disorders in Newborns and Children (ACHNDC) recommended that X-ALD be added to the Recommended Uniform Screening Panel (RUSP). The ACHDNC also recommended that federal funding be provided to state newborn screening programs to implement screening for this condition. The Secretary of Health and Human Services accepted the recommendation to add X-ALD to the RUSP on February 16, 2016. However, the Secretary was unable to provide funding for states to implement screening for this condition.

Related LINKS:

» ACHDNC Chair Letter to the Secretary of Health and Human Services
» Enclosure: Evidence-Based Review: Newborn Screening for X-ALD
» Final Response Letter from the Secretary of Health and Human Services

2018
SMA ADDED TO THE RUSP

Spinal muscular atrophy (SMA) is an inherited disorder that affects the nerves and muscles. It affects roughly 1 in 11,000 newborns. SMA is divided into five types, based on the age when symptoms start. An estimated 54% of cases are type I and have progressive weakness in the first 6 months of life and death within the first few years of life. An additional 18% of cases are type II, with progressive weakness by 15 months of age and death after the third decade of life. Until recently, only palliative care (nutritional support, physical therapy, and ventilation/respiratory support) has been available for individuals with SMA. In 2016, the Food and Drug Administration approved the first treatment medication for SMA. Studies have shown that, in patients who start having symptoms in the first 15 months of life, the medication can preserve muscular function and help improve motor skills. Also, some research shows that the treatment is more effective when started before symptoms develop.

The Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) reviewed the findings that showed the benefits of newborn screening for infants affected with SMA. On March 8, 2018, the ACHDNC recommended that SMA be added to the Recommended Uniform Screening Panel (RUSP). The Secretary of Health and Human Services reviewed this proposal and accepted the recommendation to expand RUSP to include SMA on July 2, 2018.

Related LINKS:

» ACHDNC Chair Letter to the Secretary of Health and Human Services
» Enclosure: Evidence-Based Review: Newborn Screening for SMA
» Response Letter from the Secretary of Health and Human Services

HISTORY OF THE ACHDNC

2018

ACHDNC PUBLISHES EDUCATIONAL RESOURCES

The Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) published educational resources on its website. These include the following:

- Communication guide: A communication guide for clinicians and providers to help frame the initial notification and discussion with parents about positive/abnormal/out-of-range newborn screening results.
- Educational planning guide: The Newborn Screening Educational Planning Guide helps newborn screening programs to develop and improve their educational resources.

2019

NBS SAVES LIVES REAUTHORIZATION OF 2019

On July 24, 2019, the U.S. House of Representatives passed the Newborn Screening Saves Lives Reauthorization Act of 2019. The Senate referred the bill to the Committee on Health, Education, Labor, and Pensions on July 25, 2019.

2019

MEDICAL FOODS FOR INBORN ERRORS OF METABOLISM

Over the past few years, the Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) recommended that medical foods and foods modified to be low in protein should be included as a health insurance medical benefit for anyone with a metabolic condition. Currently, there are no national standards for insurance coverage of these products. Many families face financial hardship because they cannot afford the medical foods needed for their child or young adult.

The ACHDNC completed a review of medical food reimbursement in the United States. They noted that insurance coverage for medical foods varies among states and saw an urgent need to address this issue. The ACHDNC concluded that it is time to provide stable and affordable access to medical foods required to treat patients who have metabolic conditions.

Related LINKS:

- ACHDNC Chair Letter to the Secretary of Health and Human Services
- Enclosure: ACHDNC Committee Report: Medical Foods for Inborn Errors of Metabolism: The Critical Need to Improve Patient Access
- Response Letter from the Secretary of Health and Human Services
- Medical Foods for Inborn Errors of Metabolism: History, Current Status, and Critical Need

HISTORY OF THE ACHDNC

2019

NEWBORN SCREENING SAVES LIVES ACT EXPIRES

The Newborn Screening Saves Lives Act of 2007 was originally passed in 2008 to improve education, outreach, follow-up, and laboratory quality and surveillance in newborn screening. In 2014, the Newborn Screening Saves Lives Reauthorization Act extended the legislation for another 5 years. It was up for renewal again in 2019 but was not passed by the Senate. As a result, the Act expired on September 30, 2019.

2020

DISCRETIONARY COMMITTEE FORMED

With the expiration of the Newborn Screening Saves Lives Reauthorization Act of 2014, the authorization for Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) lapsed. On November 10, 2020, the ACHDNC was formed as a Discretionary Committee to fulfill the functions previously undertaken by the former ACHDNC.

Related LINK:

» 2020 Charter of the ACHDNC

2020

REVIEW OF NBS IMPLEMENTATION FOR SMA

In February 2018, the Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) recommended the addition of spinal muscular atrophy (SMA) to the Recommended Uniform Screening Panel. On July 2, 2018, the Secretary of Health and Human Services accepted the recommendation. The Secretary also asked for a report on the implementation of newborn screening for SMA. In collaboration with experts, the ACHDNC developed the report. It detailed states' experiences implementing newborn screening for SMA. It also included outcomes of early treatment, including any potential harms, for infants diagnosed with SMA.

Related LINKS:

» ACHDNC Chair Letter to the Secretary of Health and Human Services

» Review of Newborn Screening Implementation for Spinal Muscular Atrophy Final Report