AACC Better health through laboratory medicine.

POSITION STATEMENT

Newborn Screening and Improving Children's Health

July 2014

Introduction

Newborn screening is central to ongoing efforts to improve children's health. More than 4 million babies are tested each year for health conditions that, if left undetected and untreated, could result in death or serious long-term mental and/or physical limitations. (1) Through early detection, more than 12,500 babies in the United States receive life-saving treatments and interventions annually. (2) In addition to improved health outcomes, newborn screening can reduce costs to the healthcare system.

Background

Development of Newborn Screening

Newborn screening began in 1960 when Robert Guthrie, MD, developed a test for phenylketonuria (PKU), an inherited disorder that increases phenylalanine in the blood. "Phenylalanine is found in most foods and can build up in the blood and tissues of a baby…resulting in brain damage" (3). Early diagnosis and intervention, involving a change in the baby's diet, resulted in children growing up with significantly improved intellectual and health outcomes. This discovery led states to create newborn screening programs to test for PKU and later other conditions.

AACC POSITION:
Newborn screening is vital to improving children's health. AACC endorses public-private efforts to maintain, improve, and expand newborn screening programs. The Association also supports evidence-based efforts to identify additional conditions for screening and the use of residual specimens for quality assurance and continuous improvements in pediatric healthcare.

Benefits From Early Screening

Early detection of a disorder is a life-altering experience for affected newborns. It can mean the difference between life and death or whether a child develops physical or mental disabilities. For example, each year more than 2,000 babies in the U.S. are diagnosed with primary congenital hypothyroidism (CH) (4). Early diagnosis and treatment for this condition prevents the baby from developing permanent intellectual limitations while also saving the U.S. economy approximately $400 million annually in healthcare costs (2). Similarly, screening and treating PKU saves an additional $1 billion to $2 billion each year as children with this condition require specialized care (5).

Uniformity of Testing

Although newborn screening became universal after Dr. Guthrie's discovery, there was wide disparity among the states regarding the number of conditions for which newborns were screened. A 2003 study by the General Accounting Office (GAO) indicated that the genetic and metabolic disorders tested by state newborn screening programs ranged from 4 to 36 (6). In response to this study, one of the federal agencies actively involved in newborn screening, the Health Resources and Services Administration (HRSA) requested the American College of Medical Genetics (ACMG) develop an evidence-based list of disorders for which an accurate test and effective treatment were available (7). The ACMG Newborn Screening Expert Group identified 29 core conditions. HRSA endorsed the ACMG list. The list has been expanded to 31 conditions.

Expansion of Newborn Sceening Programs

In response to the GAO findings, Congress enacted the Newborn Screening Saves Lives Act in 2008, which provided financial assistance for states to expand their newborn screening programs. Many states responded positively to the federal initiative. As of 2014, 44 states and the District of Columbia screen for at least 29 of 31 treatable conditions (8). The statute also provided funding for the Centers of Disease Control and Prevention (CDC) and HRSA to educate and train state laboratory personnel regarding new screening tests and technologies and to assist screening programs in improving the assessment, coordination, and treatment of affected newborns. AACC endorsed the 2008 legislation.

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Improvements of Technology

The development of electrospray tandem mass spectrometry (MS/MS) facilitated the rapid expansion of newborn screening. Prior to the introduction of MS/MS it was necessary to take an aliquot of the specimen to test for each disorder. MS/MS is a multi-analyte technique that allows numerous disorders to be detected from a single blood specimen more quickly, accurately, and cost-effectively than previous methods. Physicians can now diagnose more metabolic diseases before the onset of symptoms, thus preventing newborns from suffering death or irreversible adverse health conditions. The use of MS/MS technology has played a vital role in the expansion of newborn screening. Further advances in newer technologies, such as sequencing, should permit laboratories to screen for additional diseases.

Evidence-based Review Process

HRSA and other stakeholders are continually seeking to identify new conditions that can be added to the recommended list of screening tests. The federal Discretionary Advisory Committee on Heritable Disorders in Newborns and Children (DACHDNC) is charged with directing this task. DACHDNC employs a rigorous evidence-based process to determine whether a condition warrants placement on the federal list of recommended conditions for screening. AACC supports the use of evidence-based review to identify future conditions that can lead to improvements in neonate care and assist states in making informed decisions about expanding their programs.

Using Residual Specimens to Improve Newborn Screening

Newborn screening also involves the storage and use of residual specimens. Currently, states collect blood specimens on specialized collection cards (Guthrie cards) from each baby. After the initial testing, these dried blood spots are stored until confirmatory testing is performed. Once the testing process is completed, each state decides whether to maintain the residual specimens and, if they retain them, under what circumstances others may access the blood spots. AACC strongly encourages states to retain residual specimens and permit their use for improvements in pediatric care, including the development of more consistent reference intervals, new screening tests, and improved methods of screening.

Reauthorization of Federal Newborn Screening Initiative

The 2008 Newborn Screening Saves Lives Act included a sunset provision that required congressional reauthorization every five years. Although the five-year window has expired, the government has continued to fund the valuable program in the short-term. AACC strongly supports the continuation of this program, which would continue research on treatments for conditions that can be detected through early testing, assist state screening programs in assessing and coordinating newborn treatments, and ensure the quality of testing provided by newborn screening laboratories.

Considerations

Creating, maintaining, and continually improving a program that screens more than 4 million newborns annually requires significant efforts from public health laboratories, plus the active involvement and cooperation of many stakeholders, including laboratory organizations, Congress, federal and state agencies, private laboratories, hospitals, in vitro diagnostics manufacturers, children's advocates, and many others. These groups are critical to developing new and better diagnostic tests, developing new treatments, educating the public, and ensuring that newborns get the necessary treatment to avert debilitating diseases or death. The specific roles and actions of these various stakeholders are outlined below.

Laboratory Organizations

- Laboratory organizations should educate the public about the value and importance of newborn screening through consumer websites, such as Lab Tests Online.
- Laboratory organizations should work with key stakeholders to improve, expand, and promote newborn screening.
- The laboratory community should consult with ethicists and collaborate with other stakeholders on the appropriate retention and use of valuable specimens and residual newborn blood spots for quality improvement activities and research that could benefit public health.

Congress

- Congress should continue to assist newborn screening programs through the Newborn Screening Saves Lives Act.
- Congress should periodically evaluate US newborn screening efforts to ensure that such programs are meeting their objectives.

Federal and State Agencies

- HRSA, CDC, and the National Institutes of Health, as the lead federal agencies in newborn screening, should continue to assist states in expanding and improving their programs, coordinate follow-up care, and identify new core conditions that can be tested and treated.
- State newborn screening programs must continue to expand the conditions tested as the evidence warrants and ensure that babies testing positive for a condition receive timely and appropriate follow-up care.

Public and Private Laboratories

- Public and private sector laboratories provide critical information needed for diagnosing a newborn's condition. Laboratories must continue to lead in the research and development of new and improved screening tests and provide accurate testing services.

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- Public screening laboratories should publish their screening performance as assessed through participation in the CDC Newborn Screening Quality Assurance Program. This review would ensure the most appropriate screening cut-offs have been used to maximize the benefits to newborns.

- A laboratory performing newborn screening testing needs to ensure that testing is available each day to ensure prompt analysis and timely reporting of critical screening results. Systems of notification need to be developed to expedite physician communication of abnormal results and to expedite follow-up confirmatory testing and clinical treatment.

Hospitals

- Newborn screening starts in hospitals, which collect most of the specimens. Hospitals must ensure that staff are appropriately trained to collect adequate specimens, to accurately complete all necessary forms, and to send specimens to the screening facility in a timely manner.

In Vitro Diagnostics Manufacturers

- Technological advances in high throughput analytical platforms like mass spectrometry and sequencing are rapidly expanding the ability of laboratories to screen for additional diseases. Manufacturers need to continue their ongoing efforts to improve these analytical methods to further reduce false-positive and false-negative results.

Children's Advocates

- Children's advocates should work with prenatal providers to educate expectant mothers regarding state required screening tests, their purpose, and the meaning of the results.

All Stakeholders

- All stakeholders must continue to work with Congress and the federal agencies to raise awareness of the importance of newborn screening and urge continued funding for newborn screening efforts and additional research to identify new conditions where testing and intervention can improve individual care and outcomes.

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