Newborn Screening Controversy
Past, Present, and Future

Newborn screening has been hailed by the US Centers for Disease Control and Prevention as one of the most successful public health programs of the 21st century. The year 2013 marked the 50th anniversary of newborn screening, and the first half century has been heralded by many triumphs. An estimated 4 million infants are screened each year under newborn screening programs that are mandatory in most states, with approximately 12,500 being identified yearly with certain heritable disorders, many of which have devastating effects if not identified and treated prior to the onset of symptoms. As new technology has been developed, newborn screening programs have evolved. Today, 31 conditions are recommended by the Discretionary Advisory Committee on Heritable Disorders in Newborns and Children for inclusion on state newborn screening panels. Despite these successes, newborn screening programs have been fraught with controversy since their inception.

The prevention of the deleterious effects of phenylketonuria (PKU) was the original justification for requiring mandatory newborn screening. As newborn screening has evolved and the controversy surrounding mandatory newborn screening has grown, the justification for requiring mandatory newborn screening has become less compelling. Nevertheless, it is important to remember that the original justification for requiring mandatory screening, namely, the prevention of the morbidity associated with certain heritable disorders, such as PKU, through early identification and treatment remains. The inestimable value of the screening process to patients who have been identified through newborn screening programs and their families should not be overlooked when considering the controversies that continue to plague newborn screening programs.

Pediatricians and others who care for children should understand that the history of newborn screening has been marked by both success and controversy and that newborn screening programs will continue to face new challenges and generate new controversy as they continue their evolution in response to technological advances. It is imperative that care providers understand these controversies so that they can have meaningful conversations with concerned parents and educate parents about the potential value of newborn screening for their children. The following examples illustrate past, present, and potential future controversies associated with the operation of state newborn screening programs.

In 1960, Robert Guthrie developed a bacterial inhibition assay that could be administered shortly after birth to detect elevated levels of phenylalanine in infants’ blood. Population-level newborn screening for PKU began in 1963 when Massachusetts became the first state to mandate screening. At the time, many researchers believed that it was premature to require mandatory screening. The American Academy of Pediatrics and the American Medical Association initially opposed mandatory screening for PKU on the grounds that too little was known about the reliability of the screening test or the efficacy of the treatment to justify mandatory screening.

Similar concerns were raised when the advent of tandem mass spectrometry made possible the detection of a greater number of conditions through expanded newborn screening despite the fact that many of the additional conditions did not meet traditional screening criteria. Discussions about the appropriateness of adding new conditions to state newborn screening panels are ongoing, particularly when a state legislature requires the addition of a condition based on political pressure from child advocates rather than with the full support of the scientific community as was the case when Krabbe disease was added to the New York newborn screening panel.

More recently, controversy surrounding the retention and use of residual newborn screening dried blood samples, the blood that is leftover after newborn screening has been completed, has jeopardized the newborn screening programs in Texas and Minnesota. In both states, parents who objected to the state practice of retaining and releasing deidentified samples for public health research without explicit parental consent sued the state departments of health, which had a profound effect on the operation of the state newborn screening program. In Texas, more than 5 million residual dried blood samples were destroyed. In Minnesota, new legislation was passed to allow the state to retain residual samples and use them for program operations. Both states now require parental consent to release deidentified residual newborn screening dried blood samples for research. These experiences have led to further calls to reconsider whether mandatory newborn screening is appropriate.

Advances in genomics will further challenge state newborn screening programs. Although the performance of genomic sequencing in newborns offers tremendous promise to identify children at risk and potentially alter behavior that may affect their long-term health outcomes, we must consider the potential perils associated with conducting this type of testing at this early stage of life.

Although the question of what genomics results should be returned is a subject of debate, little has been said about the more practical issue of how to return results. Particularly problematic is the question of how to
return genomics results to parents at a population-wide level. If whole genome or whole exome sequencing is to be conducted on newborns at a population level, depending on what results each state newborn screening program chooses to return, every baby may screen positive for something that could endanger his or her future health. The genetics and subspecialty workforces will not be staffed adequately to meet the growing demand. Moreover, coveted appointments with subspecialists may be filled by children whose conditions may not manifest until later in life making access more difficult for those whose needs are more urgent.

It will be important for general pediatricians to have access to resources such as the ACTion (ACT) sheets developed by the American College of Medical Genetics (but for a greater number of conditions) to provide appropriate guidance for parents. Leaving pediatricians and parents to their own devices to interpret the significance of genomics results would be irresponsible and would not maximize the utility of the application of genomic technology in this context.

The application of genomic technology in the newborn screening context will be controversial, but newborn screening programs have proven to be resilient. Over time, state newborn screening programs have had to change in response to advances in technology and in response to political pressure from parents, advocacy organizations, and other stakeholders, but the programs’ commitment to their primary mission of protecting the health of our nation’s children has never wavered. The pediatric community should be aware of these issues and continue to advocate for newborn screening as a means to promote child health.

ARTICLE INFORMATION
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REFERENCES