Newborn Screening and the Obstetrician

Nancy C. Rose MD, and Siobhan M. Dolan, MD, MPH (Obstet Gynecol 2012;120:908–17)

Question 1:

Newborn screening is a state-mandated set of tests performed after the infant’s birth, when the infant is no longer the patient of the obstetrician. Prenatal counseling about newborn screening adds additional expense for a busy obstetric practice. Is there evidence that educating parents prenatally provides greater participation or better outcomes than postpartum counseling?

Response from Drs. Rose and Dolan:

Educating families about newborn screening so that they can effectively participate in this important and successful public health program is challenging. While health care providers view obstetric care as separate and distinct from pediatric care, families usually see the birth of their baby as one integrated experience. Large national advisory organizations, such as the American College/Congress of Obstetricians and Gynecologists, the American Academy of Pediatrics, and the American College of Medical Genetics (ACMG) all endorse newborn screening education in the prenatal period. To facilitate this initiative for practitioners, resources have been developed from these organizations as well as from other groups such as the Health Resources and Services Administration (HRSA) and the March of Dimes. For example, HRSA provides an attractive two-page educational brochure on newborn screening called “These Tests Can Save Your Baby’s Life” (available at: http://mchb.hrsa.gov/pregnancyandbeyond/newbornscreening/newborn_brochure.pdf) that can be printed out and easily added to the packet of educational materials that an obstetric practice provides. Another excellent online resource for families, also funded by HRSA, is “Baby’s First Test,” available at: http://www.babysfirsttest.org/.
Question 2:

What are the medical or public health reasons for state-level newborn screening programs? Would it not be less expensive, more efficient, more equitable, and more consistent to have a federal newborn screening program? What are acceptable thresholds for positive and negative screening results assigned by state programs? What are acceptable rates for false-positive and false-negative screen results? Would it make sense for all states to have the same thresholds?

Response from Drs. Rose and Dolan:

Newborn screening historically has been developed and administered on the state level. Each state considers its own population, disease prevalence, and funding, and works with its laboratory to set thresholds for screening of the individual disorders. The Newborn Screening Quality Assurance Program (NSQAP) was developed by the Centers for Disease Control and Prevention (CDC) and is operated in partnership with the Association of Public Health Laboratories (APHL). This program helps state laboratories and health departments assure quality test results in newborn screening. Federal efforts to support states have been provided by the HRSA and were further enhanced with the Newborn Screening Saves Lives Act of 2007.

Question 3:

If the obstetrician provides prenatal counseling, is he or she obliged to convey the results or assist with arrangement for care from a positive screen? If the obstetrician is not responsible for informing patients of a positive screen, who is responsible? If a diagnostic test is positive, should treatment of the affected child be provided by a general pediatrician or a specialist? Would a general pediatrician be able to provide appropriate care if a specialist was not nearby?

Response from Drs. Rose and Dolan:

Newborn screening is performed by the hospital in which a child is born, and the results are returned to the hospital as well as the pediatric provider for each newborn. Obstetricians do not have responsibility for reviewing results of newborn screening with parents. Pediatricians who receive results can use the ACMG web site to find information known as ACT sheets (Newborn screening ACTion sheets) that clearly explain the steps to be taken for each condition. These algorithms outline when immediate action is necessary, which diagnostic tests are required, and when referral to a specialist is suggested. ACMG ACT sheets are available at: http://www.acmg.net/AM/Template.cfm?Section=NBS_ACT_Sheets_and_Algorithms_Table&Template=/CM/HTMLDisplay.cfm&ContentID=5072.

Question 4:

When is newborn screening performed? When is the diagnostic test performed? What if a parent refuses the screening or diagnostic test? Why would the ACMG identify secondary conditions for which testing is available but for which there are no known effective treatments?

Response from Drs. Rose and Dolan:

Newborn screening is performed within the first 24–48 hours of life for a healthy newborn. Screening test results are usually returned within a few days, and the acuity of each condition dictates the urgency of the follow-up evaluations. For example, conditions such as galactosemia and maple syrup urine disease (MSUD) rapidly can cause severe illness, and thus positive newborn screens require immediate evaluation by a metabolic specialist. However, cystic fibrosis can be followed up within days to weeks. ACT sheets (Newborn screening ACTion sheets) are available from the ACMG as noted in Question 3 above; ACT sheets outline the precise steps to be taken for each condition.
Question 5:
How does society manage the anxiety created when a child understands the implications of a positive screen for an adult onset condition? What if the child would have chosen not to be screened? For those individuals who feel a false-positive screening test creates undue parental stress, would it be reasonable to run the diagnostic test after a positive screen prior to informing the parents? What rights do parents and children have to genomic information?

Response from Drs. Rose and Dolan:
Newborn screening does not currently screen for adult onset conditions. While the technology will soon be available to sequence an entire genome at birth, a careful assessment of what genomic information should be made available will require extensive ethical, legal, and social analysis. These are areas of ongoing debate in the evolution of newborn screening as technology becomes more sophisticated.

Question 6:
In regards to severe combined immunodeficiency (SCID), is the early bone marrow transplant meant to be curative? In regards to critical congenital heart disease (CCHD), what is the yield for pulse oximetry in the neonate with a normal prenatal anatomic survey?

Response from Drs. Rose and Dolan:
Early bone marrow or cord blood transplant is the only known cure for SCID. These procedures prevent later infections that can cause early death. In general, the earlier the treatment, the better the long-term outcome—which is a basic principle of newborn screening. The test for critical congenital heart disease is in development at this time, so a definitive answer to your question is not available.