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Universal State Newborn Screening Programs Can Reduce Health Disparities

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Fifty years after the advent of state newborn screening (NBS) programs for a metabolic condition, there is evidence that the decision to mandate universal screening can reduce health disparities. When in-hospital screening for phenylketonuria began in the early 1960s, most hospitals simply added the procedure to the list of routine clinical practices for newborns, such as giving vitamin K. For a variety of reasons, including fear of missed cases, advocates managed to get state governments involved. By the late 1960s, most states required screening of all or almost all newborns.[1] Although these advocates and state legislators did not describe their actions as addressing population-level health disparities, they believed that it was unfair for some infants to bear the consequences of late diagnosis of phenylketonuria simply because they were born in a hospital that did not provide the test. By making NBS for phenylketonuria universally available, they reduced the impact of unequal access to a new and effective therapeutic intervention– one cause of health disparities based on income, location, education, and race/ethnicity.[2]

Recent reports from states that perform NBS for severe combined immune deficiency (SCID) confirm this hypothesis. This is a rare condition that is typically diagnosed when an infant or young child has 1 or more unusual infections. Bone marrow transplantation is highly effective to treat this condition, and outcomes are better when performed in the newborn period. In 2010 SCID was added to the Recommended Uniform Screening Panel (RUSP), the list of conditions recommended for NBS by the US Secretary of Health and Human Services. Early reports of NBS for SCID have revealed that SCID is much more common in black and Hispanic individuals than previously suggested by clinical referrals to

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transplant centers. Data from the first 2 years of screening for SCID in California, for example, reveal rates of SCID among black, Hispanic, and Asian children that are much higher than would be predicted by birth rates.[3] More importantly, only 2 of the 15 infants who have undergone lifesaving bone marrow transplantation because of the state NBS program begun in 2010 were non-Hispanic white. This contrasts with earlier clinical series in which more than 80% of bone marrow transplantations for SCID were performed in non-Hispanic white children. The difference in the frequency of SCID among various racial/ethnic groups had been thought to be genetic. However, data from the California NBS program suggest that differential access to specialty care is a more likely explanation and that universal screening for SCID reduces health disparities from that condition.

It may seem strange that we are only now confirming what seems like common sense. After all, the whole idea of having an RUSP is to reduce disparities based on geography. If there is good reason to screen for a specific condition, why should infants in one state receive the benefits while infants in neighboring states do not? Similar reasoning also prevailed in the recent decision to add critical congenital heart disease (CCHD) to the RUSP. A point-of-care procedure such as pulse oximetry does not require a state laboratory, and it could have simply been added to best practices in newborn clinical care. Although CCHD can be detected either during prenatal ultrasonography or through postnatal clinical observation, access to high-quality prenatal and postnatal care may vary by race/ethnicity, socioeconomic status, and location–some hospitals are better staffed and equipped to diagnose CCHD than others. In the absence of universal screening, the frequency of late detection of CCHD has been shown to be significantly higher in birth hospitals with a level I nursery only, and universal screening should in principle reduce disparities by birth hospital type.[4]

Discovering a condition in the newborn period is not sufficient to eliminate disparities in outcomes owing to variability in uptake and adherence to follow-up and management by hospitals, clinicians, and families, which is often related to underlying social and environmental factors. Robust state public health programs form part of a system of care that goes beyond the NBS test to include contacting families and their physicians, confirming that diagnostic testing has been performed, providing training to clinicians, and ensuring that a family is connected to clinical resources. Even when such a system is in place, infants of less educated parents can be less likely to receive timely diagnosis and services.[5] Special attention to historically underserved populations, including targeted interventions to improve short-term follow-up, may be needed to ensure that the benefits of early identification are universally obtained.

Choices about which conditions to include in NBS can also alleviate or aggravate health disparities. Sickle cell disease (SCD) primarily affects infants of Hispanic or African American parents, for example, and universal NBS for SCD in combination with parental and clinical awareness and penicillin prophylaxis eliminated the majority of excess mortality resulting from that condition in young children.[6] Although the full potential of SCD NBS was not realized owing to incomplete adherence to prophylaxis, the subsequent introduction of universal immunization with conjugate pneumococcal vaccine further lowered SCD-related deaths.

Choices about NBS implementation procedures made after a condition is added to the RUSP can also potentially affect disparities. For example, the method of single-sample 2-tier screening for cystic fibrosis (CF) used in most US states–immunoreactive trypsinogen followed by testing for selected *CFTR* mutations in samples with elevated immunoreactive trypsinogen–detects fewer non-white infants who have CF because those *CFTR* mutations are less common in people of non-European ancestry who have CF. Other screening approaches for CF that provide comparable sensitivity across ethnic groups could avoid health care disparities among children with CF associated with differences in age at diagnosis.[7]

Like most NBS conditions, SCID is rare–approximately 1 case per 50 000 births–and screening and early intervention for SCID by itself will not reduce the broader disparities in health outcomes for children from Hispanic or African American families compared with children from other racial/ethnic groups. However, when all NBS conditions on the RUSP are combined, including hearing loss and CCHD, approximately 5 in 1000 newborns have a condition detectable by screening that can be addressed. The promise of NBS to improve health outcomes for all children, regardless of location, race, ethnicity, or socioeconomic status, should not be taken for granted. The history of NBS programs is replete with disruptive technologies and difficult political choices,[1] and the future promises to bring many challenging new questions. As NBS programs evolve, we must ensure that they continue to reduce the persistent health disparities among historically underserved populations. Long-term follow-up studies will be needed to monitor use of health care services and health outcomes, including impact on health disparities. More important, NBS programs need to maintain their universal nature and public health follow-up structure to maximize their role in reducing population-based health disparities.

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