A Look at Newborn Screening: Today and Tomorrow

Peter C. van Dyck, MD, MPH, FAAP, E. Stephen Edwards, MD, FAAP

The Federal Maternal and Child Health Bureau (MCHB) of the Health Resources and Services Administration (HRSA) has been involved with newborn screening and genetic testing and services since the 1960s. Although other branches of the federal government, such as the National Institutes of Health and the Centers for Disease Control and Prevention, have played key roles in newborn screening, this report focuses on the issues from the perspective of HRSA and its collaboration with the American Academy of Pediatrics (AAP).

Genetic diseases gained recognition in public health with the introduction of the newborn screening program for phenylketonuria in the early 1960s. This public health program resulted from intense pressure put on state legislatures by advocates such as Dr Robert Guthrie, by parents, and by organizations excited at the prospect of preventing mental retardation. State laws mandating such screening became the foundation of this public health program. Funding from the Children’s Bureau sponsored the early work of Dr Guthrie and the field trial for developing the screening test for phenylketonuria. The field test was expanded eventually to involve 400,000 infants in 29 states. The involvement of HRSA/MCHB in state newborn screening has continued over the past 40 years, with the funding of many initiatives designed to strengthen these important public health systems. HRSA/MCHB thinks it is critical that we continue to apply sound public health principals to newborn screening programs.

In this regard, HRSA/MCHB and the AAP believe that the newborn screening program’s efficiency and effectiveness depend on its coordinated linkages with medical homes. It is with a commitment to this belief that we continue our partnership in the newborn screening domain. Together, our aim is to enhance child health professionals’ ability to provide quality comprehensive care to identified children and their families within the primary care setting. The medical home provides a focal point for the collaboration and coordination necessary for a successful newborn screening program. HRSA/MCHB and the AAP continue to view the medical home as the foundation for optimal health care for all children.

In August 2000, HRSA/MCHB and the AAP published a report from the Newborn Screening Task Force, “Serving the Family From Birth to the Medical Home.” The report served as our most prominent collaboration regarding newborn screening at that time and provided guidance for our future steps. The articles in this supplement to Pediatrics highlight activities that HRSA/MCHB and the AAP have implemented since the report’s recommendations were generated. We look forward to the future of our partnership as we continue to strive for a quality-driven newborn screening program for children and their families nationwide.

Abbreviations: MCHB, Maternal and Child Health Bureau; HRSA, Health Resources and Services Administration; AAP, American Academy of Pediatrics

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www.pediatrics.org/cgi/doi/10.1542/peds.2005-2633A
doi:10.1542/peds.2005-2633A
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*Pediatrics* 2006;117;S193

DOI: 10.1542/peds.2005-2633A

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American Academy of Pediatrics

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