

Newborn Screening Strategies and Cost Consequences — Synthesis of Research Findings

Many medical conditions such as hearing loss, sickle cell anemia, and genetic/metabolic disorders affect infants at birth and can be the cause of severe disability or even death. Screening newborns for these diseases can help prevent or limit disability, leading to a healthier and more productive society.

Phenylketonuria

OVERVIEW

In the US, screening newborns for Phenylketonuria (PKU) has the longest history of newborn screening, and serves as a case example illustrating the role MCH programming plays in managing disease. PKU is an inherited disorder where individuals lack the enzyme responsible for breaking down phenylalanine (PHe), leading to mental retardation, delayed speech, seizures, eczema, and behavior abnormalities. Fortunately many of these complications can be avoided with proper management including dietary modifications and monitoring of blood PHe levels.^{1, 2} Managing PKU is a life long endeavor requiring both personal and institutional support. Maintaining cognitive function requires adherence to low PHe diet throughout adulthood. Currently, all states require newborns be screened for PKU at birth, a practice that has resulted in a significant reduction in negative clinical outcomes of the disease. Since the inception of screening and coordinated management, over 95% of children with PKU achieve normal or near-normal intelligence compared to just 1% prior to screening.¹⁷

ECONOMIC IMPACT

These numbers are significant from both a population health and an economic perspective. The Centers for Disease Control and Prevention estimate that the lifetime cost to society for a person with mental retardation in the US is \$1,105,480 (adjusted to 2006 values).³ Screening for PKU and preventing subsequent mental retardation has the potential to save society money in the long term. A recent study examining the cost utility of screening for metabolic disorders found that PKU screening combined with other metabolic disorders both saved money and improved health outcomes.⁴ Studies in the UK have shown similar results.⁵

ROLE OF MCH PROGRAMMING

Managing PKU requires a lifetime of follow up that is especially important as women consider motherhood. During and before pregnancy women with PKU need to maintain low PHe blood levels to ensure proper brain development of their infants. If PKU is not managed adequately, infants have a 93% risk of mental retardation. Women with PKU face multiple barriers to receiving adequate care including lack of insurance coverage for low PHe food supplements, inadequate knowledge by providers, and limited accessibility to regional metabolic clinics.⁶ This is where state MCH programs play a crucial role—many MCH programs ensure that children with PKU are followed beginning in infancy and have access to the resources and information they need.

STATE ACTIVITIES

State MCH program roles related to PKU include a broad range of activities from tracking the number of infants born with PKU to providing long term prenatal follow up for women. Specific examples include:

- Providing active case management, especially as children transition into adulthood (AL, MD, TX)
- Providing financial support enabling women to purchase needed food supplements, which can be expensive (AL)
- Coordinating with external agencies to provide support for families and patients (MD, TX)
- Training physicians on management of PKU and providing referral resources (TX)

Sickle Cell Disease

OVERVIEW

Sickle cell disease (SCD) is a genetic disorder affecting red blood cells and is most common in individuals of African descent, although it is also seen in people of Mediterranean, Indian, and Middle Eastern heritage. With SCD, errors in the cell's DNA change it from a pliable round shape to a brittle sickle form that is easily destroyed, leading to anemia, pain crises, susceptibility to bacterial infection, and stroke.⁷ Strokes, both clinical and sub-clinical, can result in significant reductions in IQ and SCD has also been associated with increases in developmental disorders.^{8,9}

Because SCD is more common in certain populations, it has been argued that screening high risk individuals makes sound economical sense.^{10, 11} One problem with implementing a targeted approach to screening is identifying whom to screen. In our multicultural, multi-ethnic society, identifying at risk patients based on race can be difficult and time consuming.²⁵ In addition to the difficulty of identifying at risk infants, ethical issues of race based discrimination and potential litigation makes a strong case for universal screening. Currently all fifty states require newborn screening for SCD and it has become the standard of care in the U.S.¹²

ECONOMIC IMPACT

The costs of treating an individual with SCD can be significant. Adjusted to 2006 values, the direct costs for hospitalization of sickle cell patients is on average \$9,423, or \$710 million. Among children with SCD in a managed care system, the average annual claim paid by Medicaid was \$14,000, with inpatient services accounting for 72% of these claims. The public sector ultimately pays for a large portion of treating children with sickle cell disease; between 60-70% of SCD hospitalizations are paid for with government funding. Treating patients in comprehensive centers has been shown to decrease the cost of emergency room visits, inpatient visits, and overall healthcare expenditures.²²

STATE ACTIVITIES

State Maternal and Child Health programs provide the links to these cost effective treatment centers by coordinating testing, providing follow up, and referring patients and their families to specialty community programs. Specific state activities include:

- Offering genetic counseling for families of newborns, adults, and couples at risk for having children with sickle cell (MD)
- Developing a regional, comprehensive care system, for Medicaid patients with SCD (MD, SC)
- Providing case management including home visits and yearly health status assessments (MD)
- Referring patients to community groups that provide family support, after school tutoring, and counseling (AL, GA, MD)
- Providing links with outside agencies such as Vocational Rehabilitation to ensure youth with SCD receive adequate care and social support as they transition into adulthood (SC)

Newborn Hearing Screening

OVERVIEW

Congenital hearing loss is one of the most common birth defects known today, with permanent hearing loss occurring in every 1-4 births per 1,000, resulting in approximately 5,000 hearing impaired infants born each year.¹³ Early hearing loss can negatively impact language ability, but children whose hearing loss is identified early and receive comprehensive intervention have significantly improved language outcomes.^{14, 15} In addition to compromised language skills, hearing loss has also been associated with poorer school and work performance.¹⁶ In light of these findings, universal screening for hearing loss of newborns has been recommended since the publication of the 1993 NIH Consensus Development Conference on Early Identification of Hearing Impairment in Infants and Young Children.¹⁷

ECONOMIC IMPACT

The economic impact of hearing loss is significant. When the costs of lost workplace productivity and increased use of medical and social support services are combined, the average lifetime cost per person with hearing loss is \$417,553, adjusted to 2006 values. Nationally this figure is \$2.1 billion.¹⁸ When considering the cost of screening, the expense of the testing and follow up need to be considered, but the long term gains from decreased spending on special education and vocational rehabilitation also need to be taken into account. In a study examining the cost effectiveness of universal screening from a state perspective, the additional lifetime costs to society for a deaf child with delayed language versus normal language was \$486,945 in 2006 values (1,286,909 vs. 799,964). These costs included lost productivity, special education, job training, and medical assistance.^{19*}

* In this analysis, universal screening identified more children and saved more money than a targeted screening approach.

STATE ACTIVITIES

State MCH programs undertake multiple strategies to improve the screening, follow up, and coordination of care of children with hearing impairment. Activities include:

- Conducting media campaigns to educate the public on the importance of hearing tests in infants (CA)
- Providing education and support services that include information on adapting to a disability in the family, advocating for your child, and locating funding sources for items such as hearing aids and speech therapy (AK, CA, NY, VT, WI)
- Establishing regional treatment centers to coordinate care (NY, CA)
- Screening infants at birth for hearing loss and linking families to support services (AK, CA, NY, VT, WI)

¹ National Newborn Screening and Genetic Resource Center. Overview of Newborn Screening. <http://genes-r-us.uthscsa.edu/resources/newborn/overview.htm> Accessed 9/26/06.

² Preventive Services Task Force (US). Guide to clinical preventive services. Baltimore: Williams & Wilkins; c1996. Chapter 44, Screening for phenylketonuria; p. 495-502.

³ Centers for Disease Control and Prevention. Economic Costs Associated with Mental Retardation, Cerebral Palsy, Hearing Loss, and Vision Impairment – United States 2003. *MMWR* January 30; 53(03):57-59.

⁴ Carroll ES, Downs ST. Comprehensive Cost-Utility Analysis of Newborn Screening Strategies. *Pediatrics* 2006; 117;287-295.

⁵ Pandor A, Eastham J, Chilcott J, Paisley S, Beverley C. Economics of tandem mass spectrometry screening of neonatal inherited disorders. *Int J Technol Assess Health Care*. 2006 Summer;22(3):321-6.

⁶ Centers for Disease Control and Prevention. Barriers to Dietary Control Among Pregnant Women with Phenylketonuria --- United States, 1998—2000. February 15, 2002 / 51(06);117-120

⁷ Nietert PJ, Silverstein MD, Abboud MR. Sickle cell anaemia: epidemiology and cost of illness. *Pharmacoeconomics*. 2002;20(6):357-66.

⁸ Ashley-Koch A, Murphy CC, Khoury MJ, Boyle CA. Buchanan GR, DeBaun MR, Quinn CT, Steinberg MH. Sickle cell disease. *Hematology Am Soc Hematol Educ Program*. 2004;:35-47.

⁹ Contribution of sickle cell disease to the occurrence of developmental disabilities: a population-based study. *Genet Med*. 2001 May-Jun;3(3):181-6.

¹⁰ Grosse SD, Olney RS, Baily MA. The Cost Effectiveness of Universal versus Selective Newborn Screening for Sickle Cell Disease in the US and the UK: A Critique. *Appl Health Econ Health Policy*. 2005;4(4):239-47.

¹¹ Panepinto JA, Magid D, Rewers MJ, Lane PA. Universal versus targeted screening of infants for sickle cell disease: a cost-effectiveness analysis. *J Pediatr*. 2000 Feb;136(2):201-8.

¹² NNSGRC. National Newborn Screening Status Report. <http://genes-r-us.uthscsa.edu/nbsdisorders.pdf>. Accessed 9/20/06.

¹³ National Center for Hearing Assessment and Management. Issues & Evidence: Prevalence of Congenital Hearing Loss. <http://www.infanthearing.org/summary/prevalence.html> Accessed September 26th, 2006.

¹⁴ Yoshinaga-Itano C, Sedey AL, Coulter DK, Mehl AL. Language of early- and later-identified children with hearing loss. *Pediatrics*. 1998 Nov;102(5):1161-71.

¹⁵ Moeller MP. Early intervention and language development in children who are deaf and hard of hearing. *Pediatrics*. 2000 Sep;106(3):E43.

¹⁶ Keren R, Helfand M, Homer C, McPhillips H, Lieu TA. Projected cost-effectiveness of statewide universal newborn hearing screening. *Pediatrics*. 2002 Nov;110(5):855-64.

¹⁷ National Institutes of Health. Early Identification of Hearing Impairment in Infants and Young Children. National Institutes of Health Consensus Development Conference Statement. March 1-3, 1993

¹⁸ CDC. Economic Costs Associated with Mental Retardation, Cerebral Palsy, Hearing Loss, and Vision Impairment – United States 2003. *MMWR* January 30; 53(03):57-59.

¹⁹ [Keren R, Helfand M, Homer C, McPhillips H, Lieu TA](#). Projected cost-effectiveness of statewide universal newborn hearing screening. *Pediatrics*. 2002 Nov;110(5):855-64.