ELSEVIER

SEMINARS IN PERINATOLOGY

Newborn Screening Progress in Developing Countries—Overcoming Internal Barriers

Carmencita D. Padilla, MD, MAHPS,*,† Danuta Krotoski, PhD,‡ and Bradford L. Therrell Jr, MS, PhD§,¶

Newborn screening is an important public health measure aimed at early identification and management of affected newborns thereby lowering infant morbidity and mortality. It is a comprehensive system of education, screening, follow-up, diagnosis, treatment/management, and evaluation that must be institutionalized and sustained within public health systems often challenged by economic, political, and cultural considerations. As a result, developing countries face unique challenges in implementing and expanding newborn screening that can be grouped into the following categories: (1) planning, (2) leadership, (3) medical support, (4) technical support, (5) logistical support, (6) education, (7) protocol and policy development, (8) administration, (9) evaluation, and (10) sustainability. We review some of the experiences in overcoming implementation challenges in developing newborn screening programs, and discuss recent efforts to encourage increased newborn screening through support networking and information exchange activities in 2 regions—the Asia Pacific and the Middle East/North Africa.

Semin Perinatol 34:145-155 © 2010 Elsevier Inc. All rights reserved.

KEYWORDS newborn screening, Asia Pacific, Middle East and North Africa, newborn blood-spot screening

Newborn bloodspot screening (NBS), using biochemical markers to detect certain congenital conditions, is a public health measure aimed at the early identification and management of affected newborns in an effort to reduce infant morbidity and mortality. It is a comprehensive system of education, screening, follow-up, diagnosis, treatment/management, and evaluation that must be institutionalized and sustained within public health systems often challenged by economic, political, and cultural considerations.[1] Initiation of NBS in developing countries, such as many in the Asia Pacific (AP) and the Middle East and North African (MENA) regions has been slow because of a variety of factors. While all countries face challenges in implementing NBS, developing countries face additional challenges related to poor economies, unstable governments, unique cultures, geographic extremes, and different public health priorities.[2-5]

The countries in the AP and MENA vary widely in size from very small countries (New Zealand, Singapore, Bahrain, Lebanon, Oman, Qatar, United Arab Emirates) to extremely large countries (China, Mongolia, Algeria, Egypt, Iran, Libya, Saudi Arabia). Some countries are economically developed (Australia, Japan, Korea, New Zealand, Singapore, Taiwan, Israel, Kuwait, Qatar, United Arab Emirates) while others are economically developing (the rest of the AP and MENA Region). Home deliveries continue to be a major challenge in Bangladesh (80%), India (61%), Philippines (62%), Pakistan (80%), Laos (85.7%), Iran (34.4%), Occupied Palestine Territory (38.8%), and Yemen (50%).[4,6] The varied written languages pose unique challenges, particularly in regions where the character sets (ie, Chinese, Arabic, Thai) are not readily understood by outsiders. As a result, experts from developed programs cannot easily communicate their experiences and share materials with some in developing programs. Despite these challenges, NBS is a growing priority in many of the more progressive developing countries.[2,3]

*Department of Pediatrics, College of Medicine, University of the Philippines—Manila, Manila, Philippines.
†Newborn Screening Reference Center, National Institutes of Health (Philippines), Ermita, Manila, Philippines.
‡Eunice Kennedy Shriver National Institute of Child Health and Human Development, National Institutes of Health, Bethesda, MD.
§National newborn Screening and Genetics Resource Center, Austin, TX.
¶Department of Pediatrics, University of Texas Health Center at San Antonio, San Antonio, TX.
Supported in part by HRSA grant U32MC00148.
Address reprint requests to Carmencita D. Padilla, MD, MAHPS, Department of Pediatrics, College of Medicine, University of the Philippines, Pedro Gil Street, Ermita, Manila 1000, Philippines. E-mail: carmencita.padilla@gmail.com

0146-0005/10/$-see front matter
doi:10.1053/j.semperi.2009.12.007

Successful NBS historically has developed from the efforts of an interested individual or group of individuals concerned with improving the life of newborns and their families. Sometimes, these efforts have taken years to develop. While some NBS programs have been initiated as government services, these generally have been confined to small countries or city-states (eg, Hong Kong, Singapore). For sustainability, NBS eventually must intersect with government public health activities. This evolution often has required a delicate balance of economics, politics, government health priorities, personnel, and other resources. Success in developing and institutionalizing NBS typically has resulted from the continued efforts of dedicated leaders willing to gain proficiency in NBS medical and laboratory science to overcome political, cultural, and economic challenges.

Collectively, we have worked with many individuals and groups that are working to initiate and/or improve NBS in developing environments. Together, and with others, we have identified certain characteristics of developed and developing NBS systems that appear to enhance their chances for sustainability: (1) strong leadership in developing pilot studies and working towards national program implementation; (2) strategic advocacy programs targeted at providing policy makers, health professionals and the public with a basic understanding of the operation and value of NBS; (3) strong collaborations between different stakeholder groups (government organizations, nongovernment organizations [NGOs], and individuals) in planning and implementation; and (4) innovative and sustainable financial strategies.

In this report, we will briefly summarize the status of NBS efforts in a large part of the developing world (the AP and MENA), review some of the challenges in implementing and sustaining NBS in an economically developing environment, and discuss some example approaches and experiences in overcoming internal barriers to NBS implementation. Where possible, we will provide examples of successful activities, often drawing from the experiences of the Philippine NBS program and other progressive developing programs.[7] We will focus on the NBS activities that are still developing, acknowledging that there are also many developed programs in these regions that can and have served as models for success. In addition to the well-developed programs in Australia, Hong Kong, Japan, New Zealand, Singapore, Taiwan, and Guam, for purposes of this article we will include South Korea, Thailand, and Israel as developed programs and outside of the scope of this discussion.

Current Status of Screening in the Asia Pacific and the Middle East/North Africa

In assessing the global burden of birth defects and congenital conditions, Christianson et al reported that once infant mortality decreases below 50/10,000 births, the genetic and congenital conditions have important health impact.[8] This is the case in most of the countries in the AP and MENA regions. An overview of the demographics of countries within both regions, including basic information about their NBS coverage, is given in Table 1. In the MENA region, cultural factors have led to larger numbers of consanguineous marriages with a consequent corresponding increased expression of recessive and potentially deleterious conditions in newborns.[11-14] Developed countries have shown the importance of NBS in preventing developmental disabilities and reducing infant morbidity and mortality. Thus, NBS has become a program of increasing importance in developing countries. Table 1 summarizes population totals, annual births, gross national income, fertility rates, and percentages of government budgets allocated for health within the AP and MENA regions to highlight some of the obvious challenges in implementing sustainable NBS.

In recent years, 3 NBS conferences have been conducted in the AP and MENA regions to initiate a dialog concerning experiences and needs. These conferences also provided an opportunity to develop a communications network within the 2 regions as a source of support and information sharing. Summaries of the meetings are available online (http://www.newbornscreening-mena.org/index.html; http://isns.napoleon.ch/upload/dokumente/mena%20nbs%20publication.pdf; and http://www.newbornscreening.ph). Two MENA regional meetings have been held; the first in Marrakech, Morocco (2006), and the second in Cairo, Egypt (2008). The first AP regional meeting was held in Cebu, Philippines (2008), with a second planned for 2010. In these meetings, representatives from screening projects and/or health ministries provided updates concerning NBS implementation activities. Experts from developed programs in various parts of the world also attended and provided assistance in developing national "plans of action" aimed at NBS expansion.

At the first MENA conference, participants from 18 MENA countries developed the so-called "Marrakech Declaration," which states that, "Newborn screening is an important tool in the prevention of disease and disability in our children and thus should be a key part of a comprehensive public health system in all of our countries." Conference participants recommended that "all countries in the region should screen for at least one condition and develop a national model program that takes into account all aspects for post-testing care."[15] At the first AP conference, participants from 11 countries in the AP region developed a "Cebu Declaration"[16] with similar language regarding future planning. Despite the relatively simple goal of screening a single condition in each country, many of the low- and middle-income countries in each region face significant implementation challenges, particularly where health systems are stressed.

Challenges in Implementing Newborn Screening in a Developing Country

Our experiences with both developed and developing NBS systems have identified the following 10 challenges to successfully implementing sustainable NBS[1-4]: