



Screening for Inborn Errors of Metabolism

Philippine Pediatric Society, Inc.
Institute of Human Genetics – National Institutes of Health
Philippine Society for Developmental and Behavioral Pediatrics, Inc.
Philippine Society of Pediatric Metabolism and Endocrinology, Inc.
National Newborn Screening Society of the Philippines

The advancement of newborn screening in developed countries has succeeded in reducing morbidity and mortality associated with certain congenital disorders. Screening within the first few days after birth allows timely medical intervention for diagnosed patients, resulting in more favorable outcomes. Through prompt and appropriate medical management, affected children identified through newborn screening can lead normal lives. There is still a lack of public awareness concerning the benefits of newborn screening. The recently passed Newborn Screening Act of 2004 will ensure that every newborn in the Philippines is given the opportunity to be offered newborn screening. This policy statement describes the roles of the pediatrician, parents, hospital administrators and government. It supports the efforts of the Department of Health and the National Institutes of Health towards the nationwide implementation of newborn screening.

Keywords. newborn screening, phenylketonuria; congenital adrenal hyperplasia; congenital hypothyroidism; galactosemia; homocystinuria, glucose 6 phosphate dehydrogenase deficiency, dried blood spot
URL: http://www.pps.org.ph/policy_statements/screening_inborn_errors.pdf

BACKGROUND

Screening of newborns for congenital disorders began in 1961 when Dr. Robert Guthrie designed an assay for the presymptomatic identification of patients with phenylketonuria (PKU) using dried blood absorbed into special filter paper.¹ Newborn screening has since been a routine component of quality newborn care in the United States (U.S) with newborn coverage of almost 100%. Although newborn screening varies by state in the U.S., screening is done for some 40 different conditions that can result in death or severe disabilities if left undetected and untreated.^{2,3} In the Asia Pacific region, Japan, Hong Kong, Taiwan, Thailand, Singapore, Australia and New Zealand have newborn screening coverage of almost 100%.⁴⁻⁸

Newborn screening was introduced in the Philippines in 1996 through the efforts of the Newborn Screening Study Group with the screening of 5 disorders: phenylketonuria (PKU); congenital adrenal hyperplasia (CAH); congenital hypothyroidism (CH); galactosemia (GAL); and homocystinuria (HCY). In 1998, HCY was dropped from the panel and glucose-6-phosphate dehydrogenase (G6PD) deficiency was added.⁹ Local statistics (NIH Phil, 2004) have shown that at least 33,000 newborns can be saved

annually from mental retardation and death through newborn screening for these 5 disorders: CH - 1:3,296; CAH - 1:5,933; PKU - 1:81,576; GAL - 1:108,768; and G6PD deficiency - 1:58.¹⁰

In 1998, the Department of Health (DOH) explored the inclusion of newborn screening as part of its programs. In 2000, DOH issued Administrative Order No. 1-A s 2000 entitled “Policies on the Nationwide Implementation of Newborn Screening” stating that by 2004, newborn screening “shall be” a part of standard newborn care.¹¹ In December 2003, the Department of Health issued Administrative Order No. 121 s. 2003 establishing a National Newborn Screening System.¹² Cost benefit analysis by Dans et. al. showed that newborn screening for CH had net benefits of US\$5 million annually, when implemented on a nationwide scale.¹³ A subsequent cost benefit study by Padilla et. al. showed that screening for 5 disorders (CH, CAH, GAL, PKU and G6PD deficiency) had net annual benefits of US\$12 million.¹⁴

Despite extensive efforts by both the government and private sector, the coverage of screened newborns has remained low at 3% of the newborn population. There is still a glaring lack of health professional and public awareness on the benefits of newborn screening. Newborn screening

will soon become a more prominent preventive health program with implementation of The Newborn Screening Act of 2004.¹⁵ This Act will require that every baby born in the Philippines be offered an opportunity for newborn screening. Implementation will provide the opportunity to identify and save over 30,000 newborns annually from negative health outcomes including mental retardation and death. Specifically, this law aims to: 1) establish and integrate a sustainable newborn screening system in the public health delivery system; 2) ensure that all health practitioners are aware of the advantages of newborn screening and of their responsibilities in offering newborns the opportunity to undergo newborn screening; and 3) ensure that parents recognize their responsibility in promoting their child's right to health and full development, within the context of responsible parenthood, by protecting their child from preventable causes of disability and death through newborn screening. With the implementation of the Newborn Screening Act of 2004*, it is expected that the general public will be made aware of the benefits of newborn screening and newborns all over the country shall have access to this essential procedure.

This policy statement supports the efforts of the Department of Health and the National Institutes of Health towards the nationwide implementation of newborn screening. The biggest challenge for the Philippines is implementation for home deliveries. The successful implementation of newborn screening will need the commitment of all sectors of society i.e parents, health professionals, non-government and government agencies.

RECOMMENDATIONS

Role of Parents

1. Parents must seek information about newborn screening from the health professional attending to the delivery of their baby.
2. Parents must recognize the importance of newborn screening and that it is part of responsible parenthood to ensure that their baby has newborn screening done at birth.
3. Parents of affected babies must ensure that they follow up regularly with the specialist. Parents must ensure that medication/dietary modification (whichever is necessary for treatment) is strictly followed under the guidance of the specialist.

Role of Health Professionals

1. Health professionals should encourage parents to have their newborns undergo newborn screening for

the congenital disorders included in the screening program.

2. Health professionals should provide adequate information about newborn screening to parents including the screening procedure, the disorders included and the possible consequences of untreated conditions, including costs and benefits.
3. Health professionals are encouraged to organize and participate in continuing medical education activities such as conventions and seminars on newborn screening.
4. Health professionals should serve as model advocates for newborn screening, community and legislative advocates, to encourage state and local governments to enact legislation requiring integration of newborn screening into the existing health care delivery system with mandatory neonatal screening for G6PD, CH, CAH, PKU, and GAL.

Role of Hospital/Lying In Administrators

1. All administrators of hospitals/maternity units/lying-ins must have newborn screening available in their facilities. Information on newborn screening must be available in prenatal clinics, labor rooms, delivery rooms, nurseries and hospital wards.
2. All administrators of hospitals/maternity units/lying-ins must assist in the recall of patients with a positive result in the newborn screening.

Role of Government Agencies

1. Concerned government agencies (Department of Health, Department of Interior Local Government and Philippine Health Insurance Corporation) should ensure the strict implementation of the Implementing Rules and Regulations of the Newborn Screening Act of 2004.

Role of Non-Government Agencies

1. All non-government agencies must be vigilant advocates of newborn screening. Their major contribution will be the education of their members.
2. Coalitions of health professionals, parents, and community leaders should develop and support a widespread campaign and national effort on educating the public about neonatal screening.

* The Newborn Screening Act of 2004 has been passed at both Houses on February 6, 2004 and is currently awaiting ratification by the President.

Document prepared by Committee on Policy Statements
 Chairperson: Carmencita David-Padilla, MD
 Co-chairpersons: Aurora Bauzon, MD; Irma Makalinao, MD
 Members: Cynthia Cuayo-Juico, MD; Nerissa Dando, MD
 Health Policy Consultant: Marilyn Lorenzo, RN, DRPH
 Adviser: Joel Elises, MD
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PANEL OF EXPERT REVIEWERS

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 Joy Lee, MD

Philippine Society for Developmental and Behavioral Pediatrics, Inc.
 Perla Santos Ocampo, MD

Philippine Society of Pediatric Metabolism and Endocrinology, Inc.
 Carmelita Domingo, MD
 Sylvia Estrada, MD
 Leticia Buenaluz, MD
 Lorna Abad, MD

ACKNOWLEDGEMENTS

The Committee on Policy Statements recognizes the contribution of the following:

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 Philippine Society of Pediatric Surgeons, Inc.
 Department of Health – Child Health Program
 Department of Health – Health Policy Development and Planning
 Bureau
 Institute of Human Genetics - National Institutes of Health

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