Inherited Disorders of Metabolism in the Newborn

A Guide for Primary Physicians

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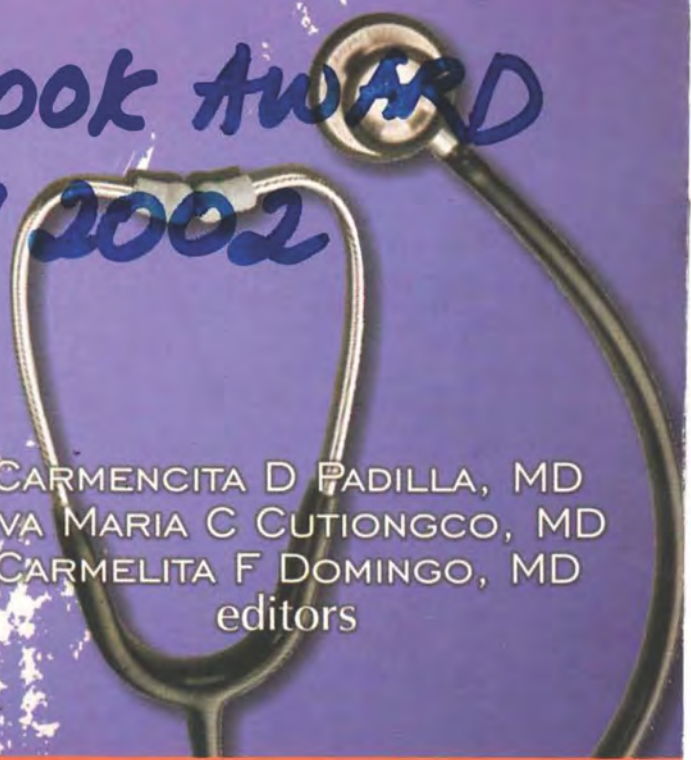

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Foreword

Many countries have adopted newborn screening as an important aspect of preventive medicine because it enables the timely detection and treatment of serious metabolic disorders that can afflict even healthy looking infants.

Though not a new technology, newborn screening was adopted in the Philippines only a few years ago through the efforts of the National Institutes of Health of the Philippines Manila. It was recently made a national program by the Department of Health. As with other health care programs which require the support of the family and the community, it is imperative that we give health practitioners and other health caregivers relevant information to help them more effectively perform their role towards the successful implementation of the program.

We are fortunate that the Institute of Human Genetics saw it fit to produce educational materials that will provide accurate and useful information on how newborn screening benefits both children and their families. This handy guide, which aims to provide health professionals a clearer understanding of inherited metabolic disorders and their management, is just one of these educational materials.

Although intended for general medical practitioners, this handbook contains information that can also be used by other members of the health care team as well as those who may not be directly engaged in health delivery but who decide on health care issues that affect our people. It is a landmark contribution to making newborn screening a reality in all hospitals and health institutions in the country.

ALFREDO T. RAMIREZ, MD
Chancellor, UP Manila

Foreword

The United Nations Convention on the Rights of the Child emphasizes the right of children to survival, protection, development and participation in governance. The health sector plays a crucial role in ensuring that all children will survive and will have the chance to achieve full growth and development.

Guided by the goal of upholding the rights of children to live normal and healthy lives, the Department of Health supports the people behind the Philippine Newborn Screening Program in their efforts to screen newborns for metabolic disorders on a national scale.

At present, the field of metabolic disorders is perceived as the concern only of specialists, such as endocrinologists and geneticists. This handbook is meant to change this perception as it provides general medical practitioners access to relevant information regarding the detection and management of metabolic disorders included in the Philippine newborn screening panel. With the implementation of the Newborn Screening Program in many areas all over the country, we expect an increase in the number of cases detected, thereby increasing the demand for immediate intervention and patient education. Informative and handy, this guide will serve as a tool for medical frontliners in providing more responsive and appropriate services to their clients, especially the newborns who are at the most vulnerable stage in their lives.

The Department of Health extends its appreciation to the Institute of Human Genetics of the University of the Philippines for developing and producing this handbook. This publication represents an important contribution to our collective efforts of reaching and empowering field workers to achieve "Quality Health Services for All Filipinos."

MANUEL M. DAYRIT, MD, MSc
Secretary of Health

Preface

As a result of continuing medical discoveries, technical advances, and a rapidly progressing newborn screening system, more and more newborns are being saved each year through the detection of *silent* (asymptomatic) conditions at birth that, if undetected or detected later, could lead to mental retardation or death. It is inevitable that newborn screening will become an integral part of routine newborn care as its benefits become better understood.

The goal of newborn screening in the Philippines, as in other parts of the world, is to give all newborns a chance to live a normal life. Newborn screening provides the opportunity for early treatment of diseases that are diagnosed before symptoms appear, thus providing affected newborns the chance to grow and develop normally.

This handbook is a sequel to the handbook entitled *Implementing Newborn Screening: A Guide for Coordinators.* It is part of a series of educational materials developed by the Institute of Human Genetics of the National Institutes of Health, UP Manila, as part of its commitment to assisting health professionals in understanding conditions included in the newborn screening panel.

As you go through this handbook, you will learn the reasons for newborn screening and its positive impact not only on children with metabolic disorders but also on families and communities. You will realize that:

- Screening detects silent metabolic conditions as early as the first week of life.

- Screening can prevent death in some instances—e.g., salt wasting CAH.
- Screening facilitates accurate diagnosis when symptoms are ambiguous or nonexistent saving time and money.
- Screening makes effective management for a metabolic disorder possible, preventing associated mental insufficiencies and other health problems.
- Screening blood collection procedures are safe, simple, and relatively inexpensive.
- Screening test procedures are reliable.
- Screening spares families and communities the burden and expense of caring for sick and retarded individuals.
- Screening provides healthy, productive citizens to society.

We believe that if people understand what newborn screening involves, what it prevents, and what it can provide for them and their children, they will want to take advantage of its availability.

Intended for general medical practitioners, this handbook describes each of the five disorders in the current newborn screening panel, namely, congenital hypothyroidism, congenital adrenal hyperplasia, galactosemia, phenylketonuria, and glucose-6-phosphate dehydrogenase deficiency. Primary care physicians, will find in this handbook basic and useful information about each of these inherited disorders. However, this handbook is not intended to replace a textbook. If you wish to have a deeper understanding of the disorders, we suggest that you refer to pediatric or medical textbooks that discuss the disorders in more detail. Our purpose is to provide you with an overview of the disorders, how they are acquired, their clinical manifestations (or the lack of these at birth), the complications that may arise if they are not promptly detected and treated, the role of newborn screening in detecting these disorders, and the need to refer cases to specialists. The handbook also pro-

vides self-assessment questions (SAQs) and their answers (ASAQs) that will help you remember important points when you encounter patients affected with these disorders.

This handbook is about caring for children. Its message is simple: *Children are our most important resource and we must commit to their loving care*. There is no better way of showing this love and ensuring the development of our children than providing for their maximum health beginning at birth.

We hope that you will find the handbook useful as you go about your duties as a primary care physician. This is just one more of the important initial steps in making newborn screening available to all Filipino babies. Let us work together to give all children the gift of a good and healthy life through successful newborn screening.

Happy screening!

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