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NBS02

Newborn Screening Follow-up and Education

This guideline describes the basic principles, scope, and range of follow-up and education activities within the newborn screening program and system.

A guideline for global application developed through the Clinical and Laboratory Standards Institute consensus process.

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Abstract

Clinical and Laboratory Standards Institute guideline NBS02—*Newborn Screening Follow-up and Education* describes the basic principles, scope, and range of follow-up and education activities within a newborn screening (NBS) program and system. NBS systems are responsible for education, screening, follow-up, diagnosis, intervention, and evaluation. Follow-up and education activities are part of the NBS system and play an essential role in facilitating early detection, diagnosis, and intervention for affected babies. This guideline is intended for those involved in any aspect of follow-up and/or education, including health care providers, parents, and others concerned with the health and welfare of newborns.

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Foreword

Newborn screening (NBS) is an essential public health service focused on testing every newborn for certain congenital diseases, groups of diseases, and/or phenotypic differences, including hearing differences, that can result in significant morbidity and/or mortality without early intervention.[1] Screening tests are not diagnostic. Rather, they separate newborns at higher risk of having a disease, group of diseases, or phenotypic difference from newborns at low risk. Therefore, newborns who have screen-positive results, indicating higher risk, must undergo additional diagnostic testing and clinical evaluation to confirm their status as affected or unaffected.

A complete NBS system comprises six parts: education, screening, follow-up, diagnosis, intervention and/or management, and evaluation.[2] Although the NBS system historically focused primarily on the screening tests performed within the public health laboratory, it is now understood that education, follow-up, diagnosis, intervention and/or management, and evaluation are equally important. An effective NBS system provides the infrastructure for universal access, education, and rapid follow-up for at-risk newborns. Parents and/or legal guardians; hospital, birthing facility, and midwifery personnel; health care providers (HCPs); and the NBS program should collaborate to ensure that the NBS system functions effectively and efficiently, providing maximum benefit to the family.

The primary aim of the NBS system is to provide early intervention for affected babies. Pre- and postdiagnostic follow-up helps ensure the accountability of NBS programs and systems. Follow-up, which determines whether NBS systems are achieving and sustaining their primary aims of preventing or minimizing morbidity and mortality, is vital to evaluating the benefits of NBS to an individual throughout his or her life, as well as to the family and society.[3] The quality of follow-up services directly affects the lives of families with at-risk and affected babies. This guideline outlines the role of follow-up services within an NBS system and provides guidance on developing and maintaining effective follow-up services, as well as on educating parents and legal guardians; hospital, birthing facility, and midwifery personnel; and HCPs on their roles in ensuring the success of NBS systems.

Overview of Changes

This guideline replaces the previous edition of the approved guideline, NBS02-A2, published in 2013. Several changes were made in this edition, including:

- Explaining general and NBS-specific terminology, including recent changes:
  - Describing use of the term *special care baby unit and/or neonatal intensive care unit*
  - Clarifying use of the terms *disease, disorder,* and *condition*
  - Clarifying definitions for *short-term follow-up* and *long-term follow-up*
- Expanding discussion of the role of communication and education in the prenatal and postnatal periods and throughout NBS systems
- Discussing timeliness initiatives related to follow-up
- Expanding discussion of postdiagnostic follow-up needs for affected individuals and families
- Outlining considerations for use of advanced screening technologies and their effect on education and follow-up needs
- Describing follow-up considerations for new diseases or groups of diseases added to screening panels

KEY WORDS

congenital heart disease
dried blood spot screening
long-term follow-up
newborn hearing screening
newborn screening
quality assurance
short-term follow-up

Chapter ❶

Introduction

Newborn Screening Follow-up and Education

1 Introduction

1.1 Scope

The primary goal of this guideline is to enhance the overall quality and continuity of newborn screening (NBS) follow-up and education services offered through public health or other NBS programs. The timeliness, quality, and effectiveness of these services directly affect the health and well-being of babies and their families, as well as the effectiveness of the NBS system.

This guideline discusses both the follow-up and education components of the NBS system. Awareness, education, training, and engagement in NBS programs are pivotal to the ongoing success of NBS as a public health initiative. To ensure efficient coordination and informed decision-making, it is important that these efforts span the entire NBS system, including the preanalytical, analytical, and postanalytical phases. As NBS continues to expand and become more complex, NBS programs increasingly need to invest resources in information dissemination and evaluate the success of those efforts in achieving NBS-related communication goals.

Likewise, post-NBS follow-up services have evolved and might now span from the first days after birth to many years after a patient is diagnosed with a disease or trait or found to have hearing differences. Follow-up services include ensuring that all newborns have received a valid screen; establishing vigorous processes to ensure appropriate follow-up for babies with actionable results; and assessing care coordination, family needs, and health outcomes after diagnosis. In general, robust follow-up is an essential part of the screening pathway, contributing to the NBS goals of quickly detecting at-risk newborns and improving health outcomes for affected babies.

The NBS program should assess the resources available in its geographic location for disease diagnosis, treatment and other interventions, and follow-up. A lack of resources can limit the value of screening. Detecting newborns at increased risk for a disease might not be advisable if sufficient resources for care are not available.

This guideline outlines the wide range of follow-up and education activities that should be included in an NBS system. It is intended for global use by public health officials, policy makers, health care providers (HCPs), and anyone involved in any aspect of follow-up or education within NBS systems, including NBS program personnel, confirmatory laboratory personnel, parents, families, and other caregivers. It does not cover other components of the NBS system, such as laboratory methods, disease-specific monitoring, treatment and other intervention protocols, or specific follow-up considerations for point-of-care (POC) screening (eg, newborn hearing screening, critical congenital heart disease [CCHD] screening by pulse oximetry).

Although funding, laws, regulations, and external advisory committees certainly apply to and affect follow-up and education activities within NBS programs, the details of these components are not included in this guideline. However, it is important for NBS programs to ensure that follow-up and education activities are accounted for within funding, regulatory, and advisory structures by including program elements such as health education, short-term follow-up (STFU) and long-term follow-up (LTFU) staffing needs, materials development and dissemination, contracts with specialty centers, and coverage of medical foods and formulas.