International Society of Nurses in Genetics

Newborn Screening: The Role of the Nurse
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Background

Population screening of presumably healthy infants to identify those who might have a rare, serious metabolic and/or genetic condition is a public health initiative (American Academy of Pediatrics, 2000; Baby's First Test, 2011; Health Departments of the United Kingdom, 2000; Holland, Stewart, & Masseria, 2006). These newborn screening (NBS) procedures are not diagnostic. Additional confirmatory testing is required. The goal of NBS is to promptly identify infants with rare conditions and to provide them with appropriate referral and early treatment, an approach that has the potential to significantly reduce morbidity and mortality (American Academy of Pediatrics, 2000; Baby's First Test, 2011). This position statement focuses on NBS procedures that involve obtaining blood samples through heel pricks during infants' first days of life for analysis in laboratories designated for NBS, the ethical issues arising from this process, and the responsibilities of nurses in this process.

In the 1960s NBS was first implemented in the United Kingdom and United States beginning with screening for phenylketonuria (PKU) (American Academy of Pediatrics, 2000; Baby's First Test, 2011; Downing & Pollitt, 2008). New technologies, such as tandem mass spectrometry and DNA analysis, later facilitated inclusion of a wide range of conditions (National Newborn Screening & Genetics Resource Center, 2012) and supported the expansion of NBS programs globally (American Academy of Pediatrics, 2000; Watson, Mann, Lloyd-Puryear, Rinaldo, & Howell, 2006). Currently NBS screening programs exist in all developed countries and in many developing countries (Burgard, et al., 2011). Screening protocols vary by state, region, and country based on the prevalence of particular conditions within the population, the screening and follow-up infrastructure, and the cost of screening and follow-up care (American Academy of Pediatrics, 2000; Baby's First Test, 2011; Holland, Stewart, & Masseria, 2006).

Since 1968, when guidelines were first established, the following criteria have been used to determine which conditions to include on NBS panels (Wilson & Jungner, 1968). The condition must (a) be effectively treatable and (b) its clinical course adequately known. Early detection must (c) enable clinicians to make treatment decisions and/or (d) facilitate supportive

care (American Academy of Pediatrics, 2000), or (e) inform parents' future reproductive decisions. Furthermore, the test must be (f) specific in its ability to identify all or most infants with the condition and sensitive to the particular condition screened, thus minimizing the rate of false-positive results. Finally, (g) experts have long recognized that financial cost and physical as well as psychosocial risks associated with NBS should be matched or outweighed by the benefits (Watson, et al., 2006; Wilson & Jungner, 1968). Although these criteria have been established for more than four decades, technology and NBS practices have evolved to allow testing for an increasing number of diseases which has challenged the limits of the original criteria. For example, some suggest that another valid rationale for screening is to obtain knowledge about the incidence and natural history of rare conditions. (President's Council on Bioethics, 2008; Secretary's Advisory Committee on Heritable Disorders in Newborns and Children, 2011).

Ethical Issues

As the advancement of technology has expanded the scope of NBS, a number of ethical issues associated with its implementation have arisen. Of the many infants screened, only a very small proportion has abnormal results, and a large number of these generally prove to be false-positive results. Furthermore, testing is often carried out without consulting parents. Even when parents are offered options, they seldom receive sufficient explanations of the test to make informed choices (Detmar, et al., 2007; Tluczek, Orland, Nick, & Brown, 2009). Though the risk of harm from the screening procedures is low, it cannot be assumed that the individual and societal benefits justify the assumption that parents should have no role in the screening process unless results are abnormal (Hargreaves, Sinclair & Oliver, 2007). Additionally, large amounts of genomic information about the involved families can be collected, analyzed, and archived with only infrequent attempts at informed consent regarding the process. Long-term follow-up on incidental findings of carrier status is relatively rare and some tests identify genetic variations that have implications for other family members. Nurses are generally involved in all aspects of the NBS process and therefore have opportunities to address these ethical concerns.

Recommendations for Nursing

It is the position of ISONG that a professional nurse has a duty to:

- actively participate in the social contract of a nurse-client relationship through health promotion, disease prevention, and patient advocacy for families affected by NBS remain current in their knowledge of NBS procedures, policies, and implications;

- ensure that parents receive accurate information about NBS (e.g., purpose, meaning, risks, advantages, their rights of enrollment depending on their locale, specimen collection and retention) prior to testing;

- ensure that parents of infants with positive results are offered counseling about the interpretation and implications of their infants' NBS results; and

- if it is known that specimens will be used for research purposes, inform parents that such usage would probably not benefit their own infants but may benefit future generations/populations.

- In addition to the above, it is the position of ISONG that nurse educators have a responsibility to include information about NBS in the curricula of newborn care. Finally, nurses who are prepared at an advanced level are encouraged to engage in policy-making and generate research to advance evidence-based clinical practices and enhance patient outcomes associated with NBS.

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