International Society of Nurses in Genetics
461 Cochran Road, Box #246
Pittsburgh, Pennsylvania USA
Phone +1 (412) 344-1414; Fax: +1 (412) 344-0599
www.isong.org

**Position Statement:**

**Newborn Screening: The Role of the Nurse**

**Introduction**: Population screening of infants to identify those who might have rare, serious disorders including endocrine, metabolic, or genetic conditions, hearing loss, and critical congenital heart defects is a public health initiative in many countries around the world (Baby's First Test, 2011; Centers for Disease Control (CDC), 2019; Holland, Stewart, & Masseria, 2006). The goal of newborn screening (NBS) is to promptly identify infants with possible rare conditions and to provide them with appropriate referral for diagnosis and early treatment, an approach that can significantly reduce morbidity and mortality (Baby's First Test, 2011; CDC, 2019).

NBS procedures are completed during an infant's first days of life. Procedures may include obtaining blood samples for processing and analysis in NBS designated laboratories, pulse oximetry testing for congenital cardiovascular disease, and audiology testing for hearing, among others.

**Issue.** This position statement focuses on NBS procedures, the ethical issues arising from NBS, and the responsibilities of nurses in the NBS process.

**Background.** In the 1960s NBS was first implemented in the United Kingdom and United States beginning with screening for phenylketonuria (PKU) (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, 2014) and supported the expansion of NBS programs globally (Watson, Mann, Lloyd-Puryear, Rinaldo, & Howell, 2006). Currently, NBS programs exist in all developed countries and in many developing countries (Thurell et al., 2015; Burgard, et al., 2011). More recently, routine pulse oximetry screening for congenital heart disease and audiology testing to identify infants who may have hearing loss have been included under the umbrella of NBS (Baby's First Test, 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, technological advances, and the cost of screening and follow-up care (Baby's First Test, 2011; Holland, Stewart, & Masseria, 2006).

**Review.** Guidelines for selecting conditions for the NBS were established in 1968 using the following criteria (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 (Association of Public Health Laboratories, 2015; Watson, et al., 2006; Wilson & Jungner, 1968).

Although these criteria have been established for more than five decades, technology and NBS practices have evolved to allow testing for an increasing number of diseases and conditions. This expansion, or mission shift, 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). Such expansion of the scope and ethical dilemmas associated with NBS are likely to continue. Whole genome sequencing has been proposed as a mechanism for expanding NBS to include thousands of genomic correlates to diseases that include late-onset and adult-onset disorders and disorders with incomplete penetrance (Berg & Powell, 2015). An identification of carrier status for rare Mendelian conditions from infancy has also been proposed (van der Burg, 2017).

**Ethical Issues** As the advancement of technology expanded the scope of NBS, a number of ethical issues associated with its implementation have arisen. Though the overall risk of harm from the screening procedures is low, the ethical principle of autonomy suggests that parents, as surrogate decision-makers for infants, should have a role in screening even in the absence of positive results (Hargreaves, Sinclair & Oliver, 2007). While thousands of infants benefit annually from early diagnosis and treatment of screened conditions, millions of infants are screened each year. Only a very small proportion have abnormal results, many of which prove to be false-positives. The psychosocial consequences and unintentional harms caused by false positives are well documented in the literature. (DeLuca, Kearney, Norton & Arnold, 2011, Holtzman, 2004, Tluczek & DeLuca, 2013). Furthermore, NBS testing is mandatory in some locations and is often carried out without consulting parents. Even when parents are offered options, they seldom receive sufficient information to make informed choices (Tluczek, Orland, Nick, & Brown, 2009; Etchegary et al., 2016). Though the risk of physical harm from the screening procedures is low, the ethical principle of autonomy suggests that parents, as surrogate decision-makers for infants, should have a role in screening even in the absence of positive results (Hargreaves, Sinclair & Oliver, 2007).

**Implications.** Additionally, large amounts of genomic information about the involved families, including incidental and unanticipated findings, can be collected, analyzed, and archived with only infrequent attempts at informed consent regarding the process (van der Burg, 2017). Long-term follow-up on incidental findings of carrier status or identification of genetic variations that have implications for other family members is rare. Furthermore, recent and ongoing calls for the use of residual (or leftover) blood spots as vehicles for research and quality control do not pose significant direct benefits to infants and must therefore be held to a higher ethical standard (Institute of Medicine (US) Roundtable on Translating Genomic-Based Research for Health, 2010). The potential to create a database of genetic information without consent of the newborn, as a vulnerable individual, for use in criminal justice and nonpublic health domains exists. Such ethical dilemmas are likely to increase as the number and range of genomic findings are increased.

**Issue: Recommendations for Nursing:** Nurses are generally involved in all aspects of the NBS process and therefore have opportunities to address these ethical concerns. It is the position of ISONG that all professional nurses involved in NBS have 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 by remaining 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 or opt-out depending on their locale, specimen collection and retention, and anticipated future research) 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

• ensure that parents are provided with understandable, accurate information regarding proposed uses of NBS for research purposes to allow for informed decision-making.

In addition to the above, it is the position of the International Society of Nurses in Genetics (ISONG) that both clinical and academic nurse educators have a responsibility to include information about NBS in the curricula of newborn care and population health. 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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**Revised September 2020 by**:
Lisa M. Blair, PhD, RN, University of Kentucky, USA
Leslie Darmofal, DNP, APRN, ACNS-BC, PHN, Minnesota State University, Mankato, USA
Maryann Campbell, MS, CNS, RNC-NIC, Kaiser Permanente Northern California Region, USA

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