ROLE OF THE NURSE IN NEWBORN SCREENING: INTEGRATING GENETICS IN NURSING EDUCATION AND PRACTICE

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Peter James Bongolan Abad
University of the Philippines Manila
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Aster Lynn Demaisip Sur
Institute of Human Genetics, NIH, UP Manila
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Sibulo Ma. Salve
Philippine General Hospital
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RESEARCH ARTICLE

ROLE OF THE NURSE IN NEWBORN SCREENING: INTEGRATING GENETICS IN NURSING EDUCATION AND PRACTICE

Peter James B. Abad, RN, Msc[1], Ma. Salve K. Sibulo, RN, and Aster Lynn D Sur, RND, RN

Abstract

Nurses are key players in the newborn screening (NBS) program. The inherited nature of the conditions included in the NBS panel means that nurses should be competent in integrating genetics in their practice. However, studies suggest inadequate preparation of nurses in incorporating genetics in their practice. This article aims to discuss how nurses can capitalize on existing population-based genetics programs such as newborn screening to enhance their clinical practice through genetics. An overview of the newborn screening program in the Philippines is provided as well as a discussion on the roles of nurses in genetics in the context of NBS, and a brief discussion of future directions of the Philippine newborn screening program and how this may impact nursing education and research.

Keywords: newborn screening, neonatal screening, nurses, genetics, genomics, nursing practice, nursing education

Introduction

Newborn screening (NBS) is a universally accepted and one of the first population-based genetic screening programs that aims to identify and detect congenital conditions. Early identification of these congenital conditions is crucial as timely interventions could mean saving newborns from severe and irreversible complications, usually intellectual disability, and/or even death (Pourfarzam & Zadhoush, 2013). Newborn screening has evolved through the years from the simple bacterial assay developed by Dr. Robert Guthrie in the 1960s to detect increased phenylalanine levels in newborns with PKU (Guthrie & Susi, 1963) to a comprehensive and complex screening program that now includes detection of over 50 different conditions (Therrell et al., 2015). Typically, newborn screening is done by collecting a heel prick blood sample, but this has also expanded to detecting hearing loss (Wroblewska-Seniuk, Dabrowski, Szyfter, & Mazela, 2017) and critical congenital heart diseases (Kumar, 2016).

Nurses are key players in the newborn screening program and their role is crucial in increasing newborn screening coverage. Considered as front liners, nurses are usually the first contact of parents in a primary care setting or in health facility and this gives them an opportunity to advocate and educate parents on newborn screening. The inherited nature of the conditions detected in newborn screening, mostly inborn errors of metabolism, implies that nurses should be knowledgeable about basic genetics concepts necessary to explain these conditions to parents. Nurses should also be ready to address parents' questions on newborn screening mechanism, discuss what the results could potentially mean for the newborn and the family as well as the various ethical issues that may arise, and provide support to parents. Driven by advances in genetics and genomics, these expectations from nurses are the impetus for the recommendations on integration of genetics in nursing practice (Greco & Salveson, 2009; Jenkins, Grady, & Collins, 2005). The National Coalition for Health Professions Education in Genetics (NCHPEG) and the American Nurses Association published the core competencies in genetics for health professionals and for nurses, respectively, both of which aimed to provide a framework to guide the integration of genetics knowledge, skills, and attitudes into routine health care (Badzek et al., 2008; National Coalition for Health Professional Education in Genetics, 2007). There have been various initiatives to improve competencies of nurses in genetics both in pre-service (e.g., Daack-Hirsch, Dieter, & Griffin, 2011; Garcia, Greco, & Loescher, 2007; St-martin, Bedard, Nelmes, & Bedard, 2011) and graduate nursing programs, and in-service (e.g., Kirk, Tonkin, & Burke, 2008). Despite these initiatives and programs, however, studies suggest that nurses (Aiello, 2017; Coleman et al., 2014; Seven, Pasalak, Guvenc, & Kok, 2017) are inadequately prepared to integrate genetics and genomics in their clinical practice.

[1] Correspondence: Peter James B. Abad, RN, Msc; Address: College of Nursing, University of the Philippines Manila, Pedro Gil, Ermita, 1000 Manila; Email: pbabad@up.edu.ph

In the Philippines, there is also a recognition of the importance of integrating genetics and genomics in nursing education and practice largely due to the increasing use genetics and genomics services (Padilla & Cutiongco dela Paz, 2013). However, a survey on the knowledge, perceptions, beliefs, and practices on genetics and genomics among Filipino nurses working in the US revealed that while 41% had a good understanding of genetics of common diseases, majority of the respondents still reported they need additional continuing education to assist them in integrating genetics and genomics in their practice (Saligan & Rivera, 2014). In this paper, we will provide an example on how nurses can capitalize on existing programs, such as newborn screening, to enhance their clinical practice by integrating genetics. Specifically, this paper aims to provide an overview of the newborn screening program in the Philippines; to explicate the role of nurses in genetics in the context of newborn screening; and finally to discuss the future directions of the Philippine newborn screening program and how this may impact nursing education, and research.

The Philippine Newborn Screening Program

The Philippine NBS program started in 1996 as a pilot research study involving 24 health facilities in Metro Manila to establish incidence of six metabolic conditions namely congenital hypothyroidism (CH), congenital adrenal hyperplasia (CAH), galactosemia (Gal), phenylketonuria (PKU), homocystinuria (HCY), and glucose-6-phosphate dehydrogenase (G6PD) deficiency, to gather adequate data to support for the possible inclusion of newborn screening in the existing health services programs of the Department of Health, and to produce evidence for formulation of national policies and legislation to integrate NBS in the health system (David-Padilla et al., 2009). The Philippines Department of Health formally acknowledged NBS in 2000 as an essential service to improve child health outcomes and adopted the screening of five metabolic disorders (homocystinuria was dropped from the panel because there were no cases detected during the pilot) (David-Padilla et al., 2009; Department of Health, 2000). Because of the observed high number of cases, maple syrup urine disease (MSUD) was included in the panel of disorders in 2012. In 2014, the NBS panel of disorders was expanded from the original six to about 28 disorders to include hemoglobinopathies, amino acid disorders, organic acid disorders, fatty acid oxidation disorders, disorders of carbohydrate metabolism, disorders of biotin metabolism, cystic fibrosis, and endocrine disorders (Department of Health, 2014b, 2014a).

Vital in the NBS program is the ratification of the Newborn Screening Act in 2004 which ensured its institutionalization in the health care delivery system. There were several key provisions of the law that guaranteed universal newborn screening including 1.) designation of the Department of Health as the lead agency in implementing this program; 2.) creation of the Newborn Screening Reference Center that serves as the technical arm of the Department of Health; 3.) establishment of Newborn Screening Centers (NSCs) equipped with NBS laboratory and follow-up program; 4.) instituting NBS as a requirement in licensing and accreditation of health facilities; and 5.) inclusion of NBS cost in insurance benefits of PhilHealth, the National Health Insurance Program (Republic Act No. 9288, 2004). This law also mandated integration of newborn screening in the curriculum of all health professions, and it obliged all health professionals, including nurses, to inform about the availability of newborn screening services and to motivate parents to have their newborn undergo screening (Department of Health, 2004; Republic Act No. 9288, 2004).

Table 1 summarizes the incidence of disorders as of December 2017. The most common are G6PD deficiency (1:58), congenital hypothyroidism (1:2,618), and alpha thalassemia (1:1,481). As of December 2017, newborn screening covered 70.4% of the almost 2.2 million projected newborns, of which 9.5% underwent expanded newborn screening (Newborn Screening Reference Center, 2017). Newborn screening in the Philippines operates under an opt-out model such that parental consent is presumed

Table 1. *Number of confirmed cases and incidence of disorders included in the Philippine NBS panel, 2017*

Disorder	No. Screened	Confirmed Cases	Incidence
Congenital hypothyroidism	10,415,695	3979	1:2,618
Congenital adrenal hyperthyroidism	10,415,695	576	1:18,083
Phenylketonuria	10,415,695	84	1:123,996
Galactosemia	10,415,695	128	1:81,373
Maple syrup urine disease	7,031,276	96	1:73,242
Glucose-6-phosphate dehydrogenase deficiency	10,208,668	176510	1:58
Fatty acid disorders	402,841	15	1:26,856
Organic acid disorders	402,841	10	1:40,284
Hemoglobinopathies			
HbH disease/Alpha thalassemia	402,841	272	1:1,481
Hemoglobin E disease or interacting Hb E/ Beta thalassemia	402,841	20	1:20,142
Beta thalassemia	402,841	3	1:134,280
Hemoglobin D disease	402,841	1	1:402,841

Source: Newborn Screening Reference Center

and screening proceeds routinely unless there is explicit wish from parents for their child to not be screened (Department of Health, 2004; Potter et al., 2015; Republic Act No. 9288, 2004). The usual and acceptable reasons for not screening is religious in nature and this decision by parents is noted down and documented in the newborn's chart (Department of Health, 2004). Collection of a blood spot sample through the heel prick method is done by a trained health professional, usually a trained nurse, a midwife and medical technologists. After which, the sample is transported within 24 hours to the designated Newborn Screening Center (NSC) which runs the laboratory test. Currently, there are six NSCs strategically located in the country to ensure that screening is done in the most timely and efficient manner (Department of Health, 2018). A positive screening result necessitates immediate recall of the newborn within 24 hours. Once a positively screened newborn is located, confirmatory testing and diagnosis is done and once confirmed, treatment and management are instituted (Department of Health, 2004).

The success of this national program relies on the efficiency and effectiveness of mechanisms from pre-screening information dissemination and advocacy, recall of positively screened newborns, timely diagnosis and institution of management, and regular follow-up consultation visits in newborn screening continuity clinics. Throughout this process, nurses have key roles and responsibilities in ensuring that appropriate information are given to parents even during the pre-screening period, facilitating transport of specimen to NSCs, and recall, follow-up, and management of those diagnosed.

Roles of Nurses in Newborn Screening: Exemplar Cases

Philippine General Hospital NBS Nurses Core Group

The Philippine General Hospital (PGH) is the largest tertiary and teaching hospital in the Philippines that serves as a referral center for various conditions. Around 600,000 patients are seen in the hospital every year. In 2018, the hospital attended to 4,828 livebirth deliveries and all newborns underwent NBS. The 100% NBS coverage by the hospital over the years was largely in part due to the efforts of nurses who are part of the Newborn Screening Nurses Core Group who ensure that all parents of newborns are informed about the availability of screening. Established in 2003, this innovative approach is a response to the need to increase screening coverage all over the country considering that newborn screening was not yet fully integrated in the health system then pending the ratification of the law in 2004.

Composed of NBS-trained nurses who are mostly from the maternity and pediatric wards, the aim of the core group is to ensure that pre-screening education are provided, and screening results are communicated appropriately to parents. A referral for newborn screening made anywhere in the hospital will be responded to by an assigned NBS nurse. During the pre-screening education, the parents are given information about the benefits of newborn screening and its risks, as well as the various disorders that are included in the screening panel. They would also orient the parents about the heel prick procedure of collecting blood spot samples. After collecting the blood sample, parents are provided with additional resources, usually a brochure on newborn screening, and contact information should they have any questions later on.

In pre-screening education, nurses are in a position to address not only informational needs of parents, but also the inherent psychosocial and ethical issues that come with the screening process. For example, while NBS in the country operates under an opt-out model and parents have minimal engagement in giving informed consent, providing pre-screening education still ensures that parents know the purpose, benefits, risks, and possible results of the screening which can psychologically prepare them for the results. It has been shown parents who have been given information about the screening beforehand has higher adherence to follow-up in case of positive screening results (Potter et al., 2015). This also allows building of trust and rapport among parents, health care providers, and the NBS program itself and this conveys respect to parents(Manson, 2010).

NBS nurses are also involved in results disclosure. As soon as a positive screening result is received from the Newborn Screening Centers, assigned NBS nurses get in touch with the parents. During the disclosure, the nurse provides information about the specific disorder seen in the screening, gives instructions as to confirmatory testing, and offers psychosocial support to parents. It is during this phase that parents present with various reactions and heightened emotions ranging from being shocked and surprised, to being anxious as to what the disorder is all about. It is in this point that nurses provide psychological support to parents. This can mean assessing for supportive needs of parents, listening to them without compelling them to talk, comforting them and acknowledging their concerns, and helping them to obtain further information by giving them educational resources or directing them to reputable online sources. Nurses also refer them to appropriate specialty depending on the specific disorder, to genetic counseling services for further psychosocial support, and to social services in case of financial needs.

Newborn Screening Continuity Clinics

The Newborn Screening (NBS) Continuity Clinics is a response to the need to establish and strengthen the treatment network of NBS ensuring that timely treatment and continuous management is instituted to diagnosed cases of any of the heritable conditions included in the screening panel (Department of Health, 2014a). Currently, there are 14 operational NBS Continuity Clinics which are administratively under regional medical centers strategically located in the country making the services more accessible(Department of Health, 2018). Each continuity clinic is manned by a part-time medical specialist (usually a pediatrician), and a full-time nurse or genetic counselor.

Long-term follow-up in newborn screening is a crucial period for the parents to understand the natural history of the condition, health and reproductive implications of the diagnosis, to cope and adjust

to the presence of condition, and to sustain recommended management modalities. It is then important to highlight the role of the medical specialist and the nurse to address supportive care needs of the family and to track individuals throughout the life course to ensure continuity of care (Kemper et al., 2008).Supportive care, initially used in the context of cancer, has been studied in rare diseases as well and it encompasses provision of services that meets physical, informational, emotional, practical, and social needs of parents with a child with rare disease (Pelentsov, Laws, & Esterman, 2015). In this sense, providing supportive care means not only addressing immediate information needs about the diagnosis, but also providing additional educational and supportive resources, and attending to the emotional responses of families as well as how this may impact family relationships'(Helm, 2015; Pelentsov et al., 2015). In the context of long-term follow-up in newborn screening, the US Department of Health and Human Services published crucial questions that can serve as guide in addressing supportive care needs of families. One of these crucial questions is whether the family has the necessary competencies to provide home care to their child and whether they are supported to adhere to treatment regimens (Hinton et al., 2011). To illustrate, children diagnosed with disorders of amino acid metabolism (e.g., MSUD) need to strictly adhere to specific low-branched chain amino acid diet closely directed by a metabolic dietician. Nurses ensure that these families understand the importance of observing the strict nutritional requirements and they have the skills to follow nutritional advice. In yet another example, a study on Filipino families with CAH found that the families' understanding of the condition was limited to the point that they thought the condition was 'simple' leading to the underestimation of the importance of medicine and regular follow-up consultation resulting to exacerbation of the condition (Abad et al., 2016). Nurses, equipped with a good knowledge base on the diseases in newborn screening, their treatment, and health implications, provide additional educational resources that can help families understand their child's condition better.

It is also during long-term follow-up that parents, in their attempt to understand more the condition, would ask questions on the genetics of the condition such as the cause, prevention, and reproductive implications. Together with other health professionals such as genetic counselors, nurses can provide basic information on the genetics of these conditions. Most conditions screened in the panel are inherited in an autosomal recessive pattern while G6PD deficiency is inherited through X-linked recessive inheritance. Aside from providing genetics education, however, it is vital for nurses to be aware of potential sociocultural beliefs and issues of families and how these may affect their understanding of the condition. For example in CAH, Filipino families vaguely understood the genetics of the condition pointing to “something in the blood” that caused the disease and all families in the study had limited understating of the reproductive implications of CAH, an autosomal recessive condition (Abad et al., 2016). In a review, Abad et al., (2014) identified seven common Filipino cultural beliefs that may have an impact on how families understand genetic conditions. These cultural beliefs, while not aligned to the medical explanation, should not be disregarded, but rather acknowledged, to ensure that culturally-appropriate care is provided. Awareness of social and cultural issues, as well as understanding clients' knowledge, perceptions, and beliefs, are important competencies of nurses (Badzek et al., 2008). Cultural sensitivity can be addressed by using tools such as the Kleinman's Explanatory Model of Illness, composed of eight questions about the family's perceived cause of the condition, treatment, and management (Kleinman, Eisenberg, & Good, 1978), which can easily be administered in clinic.

Nurses also act as case managers of these children and as such they guarantee coordinated care among the various medical and allied medical specialties required in patient management, and ensure continuity of care including the transition from pediatric to adult care (Hinton et al., 2011). Nurses' role is key in liaising and advocating the needs of patients and families amidst the complexity of medical care required. For example, when nurses identify the need for further discussion of reproductive implications and for psychosocial support, appropriate referral to a genetic counselor can be made. Since there are not enough genetic counselors yet in the Philippines, genetic counseling session can be provided through a telegenetics mechanism. Aside from this, nurses also see to it that non-medical needs such as financial and spiritual concerns are addressed. In such case, nurses work with non-medical professionals such as social workers and the clergy to give appropriate support to patients and families.

Another crucial question that needs to be addressed is whether the family has the skills to self-advocate (Hinton et al., 2011). An example of self-advocacy would be the family's involvement in raising awareness about their child's condition through participation in support groups and other activities that encourage networking with other families affected with similar condition. In a recent survey on supportive care needs of Australian parents with a child with rare disease found that 42% had no access to support groups and 75% had no contact with other parents with a child with similar disease leading to feelings of social isolation and loneliness (Pelentsov, Fielder, Laws, & Esterman, 2016). Nurses should encourage parents to join support groups highlighting the benefits of participating in such groups. In the Philippines, there are a number of disease-specific support groups for MSUD, thalassemia, and CAH (see website of the Professional Society of Genetic Counselors in Asia https://www.psgca.org/programs.html#Groups). The Philippine Society for Orphan Disorders (PSOD), an umbrella organization for rare diseases in the Philippines, was also established to cater and advocate for the needs of families affected with rare diseases (Philippine Society of Orphan Disorders, 2019).

Future Directions and Implications to Nursing Education and Research

Newborn screening in the Philippines will continually expand in the future with the possible addition of critical congenital heart disease (CCHD) screening in the panel. A pilot study on the use of pulse oximetry in the detection of CCHD is underway to provide data and

policy directions to its possible inclusion in the newborn screening program (UP Manila, 2018). These developments does not only provide a platform for nurses to integrate genetics in their clinical practice, but has implications in nursing education and research.

Nurse educators should prepare nurses, both in the baccalaureate and graduate level, to respond to the challenge of increasing use of genetics in clinical practice, and specifically on newborn screening and its impact on families. There have been various strategies used to ensure that genetics is integrated in the curriculum (Daack-Hirsch et al., 2011) but certainly newborn screening offers a good context on how genetics is discussed. For example, the disorders in the panel can be used to explicate the different inheritance patterns and issues like storage of blood sample, presumed informed consent, and communication of results to families can be used to expound on the ethical, legal, and social implications of genetics. There are also a number of good resources available online that nurse educators can use such as the *Online Mendelian Inheritance in Men (OMIM)* and *Genereviews (see De Sevo, 2010)*. Information specific about newborn screening in the Philippines can be freely accessed in the Newborn Screening Reference Center website (https://www.newbornscreening.ph/).

There are also a number of compelling research areas in newborn screening that nurses can be involved in. Much of these topics are along the lines of the ethical, legal, and social implications of newborn screening such as exploration of the degree of engagement of parents in the newborn screening program given the presumed informed consent model in the Philippines program, strategies on how screening results are best communicated to parents and families, a formal survey on the parental supportive care needs, and supporting patients in their transition to adulthood cognizant of issues such as reproductive decision-making and psychosocial adjustments. Nurses can also participate in multidisciplinary studies that would refine the service delivery in newborn screening.

Summary

The expansion of the newborn screening program in the Philippines, with the potential addition later on of pulse oximetry to screen for critical congenital heart disease open opportunities for the evolution of nurses' roles. The genetic nature of most of the conditions included in the screening panel necessitates that nurses are competent to integrate genetics in their clinical practice. In this paper, we provided specific examples on the roles of nurses from pre-newborn screening, disclosure of screening results, and in the long-term management of the disorders. This also serves as an impetus to strengthen nursing education by ensuring that genetics and genomics are taught in both the undergraduate and graduate programs and to ensure that genetics is threaded through in the curriculum. There are also a number of opportunities for nursing research in newborn screening and these are necessary to provide empiric evidence that can guide in improving care for patients and families. Overall, these opportunities provide an avenue for nurses to be key players in the advancement of genetics and genomics.

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ABOUT THE AUTHORS

Peter James B. Abad, RN, MSc is a registered nurse and a genetic counselor. He is currently an Assistant Professor and the College Secretary of the College of Nursing, University of the Philippines Manila. He is also affiliated with the Department of Pediatrics, UP College of Medicine as part of the teaching faculty of the MSc Genetic Counseling program. He is a founding member and currently the Membership Director of the Professional Society of Genetic Counselors in Asia. He is also currently the Director for the Philippines of the Transnational Alliance of Genetic Counseling.

Ma. Salve K. Sibulo, RN is currently a Chief Nurse at the Philippine General Hospital. She has been serving in the national hospital since 1992. She is also the Newborn Screening nurse coordinator of the hospital since 2005. Currently, she is studying Master of Science in Nursing at St. Paul University Manila. She also took units of Master of Science in Genetic Counselling at the University of the Philippines Manila. She obtained her college degree at the Perpetual Help College of Manila.

Aster Lynn D. Sur, RND, RN is currently a Project Development Officer IV and Supervising Nurse of the Clinical Genetics and Research Unit of the Institute of Human Genetics, National Institutes of Health, University of the Philippines Manila. She has been involved in the implementation of the National Comprehensive Newborn Screening (NBS) System in the country since 1999 up to the present. Her previous work included: NBS Follow- up Nurse, NBS Nurse for special projects and Nurse at the Clinical Genetics.

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