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Author manuscript

J Nurs Scholarsh. Author manuscript; available in PMC 2019 May 01.

Published in final edited form as:

J Nurs Scholarsh. 2018 May ; 50(3): 249–256. doi:10.1111/jnu.12380.

The Global Landscape of Nursing and Genomics

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Abstract

Purpose—Nurses have a pivotal role in bringing the benefits of genomics and precision medicine to everyday health care, but a concerted global effort is needed to transform nursing policy and practice to address widely acknowledged deficits in nurses' genomic literacy. The purpose was to conduct a global country and organization review of nursing engagement with genomics, informing a landscape analysis to assess readiness for integration of genomics into nursing.

Design—Global nursing leaders and nursing organizations were recruited using a purposive sampling strategy to complete an online survey that assessed the scope of genomic integration in practice and education, challenges and barriers, and priorities for action.

Methods—The survey was administered online following an orientation webinar. Given the small numbers of nurse leaders globally, results were analyzed and presented descriptively.

Findings—Delegates consisted of 23 nurse leaders from across the world. Genomic services were offered predominantly in specialty centers consisting mostly of newborn screening (15/18) and prenatal screening (11/18). Genomic literacy and infrastructure deficits were identified in both practice and education settings, with only one country reporting a genetic/genomic knowledge and skill requirement to practice as a general nurse.

Conclusions—These data provide insights into the commitment to and capacity for nursing to integrate genomics, revealing common themes and challenges associated with adoption of genomic health services and integration into practice, education, and policy. Such insights offer valuable context and baseline information to guide the activities of a new Global Genomics

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Nursing Alliance (G2NA). The G2NA will use the landscaping exercise as a springboard to explore how to accelerate the integration of genomics into nursing healthcare.

Clinical Relevance—Genomics is relevant to all healthcare providers across the healthcare continuum. It provides an underpinning for understanding health, risks for and manifestations of disease, therapeutic decisions, development of new therapies, and responses to interventions. Harnessing the benefits of genomics to improve health and care outcomes and reduce costs is a global nursing challenge.

Keywords

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As the largest single healthcare professional group worldwide, nurses have a pivotal role in bringing the benefits of genomics to everyday health care; however, a global effort is needed to transform nursing policy, practice, education, and research (World Health Organization [WHO], 2016). Embracing genomic health care requires a prepared workforce. This represents a significant challenge, since deficits in genomic literacy in nursing and other health professions are widely acknowledged (Calzone, Jenkins, Culp, Caskey, & Badzek, 2014; Skirton, O'Connor, & Humphreys, 2012). Therefore, there is a need to assess existing genomic integration, as well as challenges or barriers, and to gauge readiness for a collaborative global effort to increase nursing capacity to integrate genomics into practice.

Background

Developments in genomics and its translation to improve healthcare continue unabated (Davies, 2017). Genomics advances have implications worldwide, across the healthcare continuum, and impact all nurses regardless of academic preparation, role, or clinical specialty (Umberger, Holston, Hutson, & Pierce, 2013). Embracing genomic health care requires a prepared workforce that can inform, educate, and empower people, address existing and novel ethical issues, and anticipate any potential negative impact on vulnerable populations (Badzek, Henaghan, Turner, & Monsen, 2013; Seven, Eroglu, Akyuz, & Ingvoldstad, 2017; Tekola-Ayele & Rotimi, 2015). Nurses have a pivotal role in leading change to advance health, integrating research discoveries into ethical healthcare practice benefiting individuals and societies (Salmon & Maeda, 2016). However, there is substantial evidence that many nurses worldwide lack confidence and competence in genomics, and education provision is inconsistent (Calzone et al., 2014; Skirton et al., 2012). A survey of 10 countries, including the United States, United Kingdom, Israel, Brazil, Pakistan, and South Africa, found that genetics competence is not included within the regulatory standards of six countries and is explicit in only one, defined as "only at the basic level" (Kirk, Calzone, Arimori, & Tonkin, 2011). The conclusion was that concerted and strategic global effort is needed to prepare and enable nurses to drive progress, influence policy, and maximize existing resources to promote nursing literacy in genomics that includes associated ethical, legal, and societal challenges (Kirk et al., 2011). This was echoed by Williams and colleagues, who acknowledge the critical role of nursing in implementation of genomics (Williams, Feero, Leonard, & Coleman, 2017). Nursing policy, education, practice, and research in genomics needs to be strengthened worldwide, and policy

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initiatives in some countries, such as England, may inform how this could be achieved elsewhere (Health Education England [HEE], 2017). Motivated by the need to embrace this challenge, the authors facilitated an interactive event to harness influence and knowledge, with the aim of creating a Global Genomics Nursing Alliance (G2NA; www.g2na.org) to accelerate integration of genomics into everyday professional practice. This article presents the first phase of that wider initiative.

Aims

As a starting point for the establishment of the G2NA, we conducted a country- and organization-specific landscape analysis to assess the factors likely to impact readiness for and scope of genomic integration into nursing policy, practice, and education. We sought to identify the range of genomic services available, the healthcare contexts within which they operate, and the challenges, barriers, and areas of action for nursing. The aim was to provide context and insights into the commitment, capacity, and challenges around the integration of genomics into nursing, and to inform a framework for action for the G2NA. This project was reviewed and approved by the Faculty of Life Sciences and Education Ethics Committee, University of South Wales.

Methods

Recruitment

This project utilized a purposive sampling strategy. Survey participation was limited to delegates attending the inaugural 2017 G2NA meeting. The number of delegates was constrained by available grant funding and meeting space. Country delegates were selected based on their expertise in nursing, health care, services, policy, and leadership within their country. Expertise in genomics was not required. Some delegates were not nurses but represented the nursing community in their country or provided a critical perspective to inform the work of the G2NA. There was an effort to have a broad geographical representation. We also strived to achieve a gender balance similar to the international nursing workforce, which is approximately 16% men (WHO, 2017a). Organizational delegates represented international nursing and genetic organizations: International Council of Nurses (ICN), Sigma Theta Tau International (STTI), International Society of Nurses in Genetics (ISONG), European Society for Human Genetics; two large national genomics and health organizations (U.S. National Human Genome Research Institute and HEE Genomics Education Programme), and a national advocacy group for individuals and families affected by genetic disorders (Genetic Alliance UK). Delegates were identified via international nursing networks and an iterative process by the authors to identify the optimal representative at the most appropriate and highest level (e.g., current president).

Instrument

The WHO describes a landscape analysis as a review of positive and negatives factors that might influence the likelihood of adoption of a new development, initiative, or technology (WHO, 2010). The survey instrument was developed specifically for this project, and questions were designed to ascertain the country- and organization-specific context.

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Questions were adapted with permission from Manolio et al. (2015) and leveled for nursing by the authors, drawing on previous work assessing critical success factors in genetics/genomics integration in nursing (Kirk et al., 2011). The survey was pilot tested by the authors and other genomics and nursing experts, then revised prior to administration. Questions solicited information based on the delegate representation: country versus organization. Country questions sought information on the healthcare system, nursing, and genomics in mainstream and specialist services, as well as challenges and priorities in integrating genomics into nursing. Organization questions requested information on the scope of the organization (national or international), type and size of membership, existing genomic learning resources, and the organization perspective of challenges and priorities in integrating genomics into nursing. All delegates were asked to identify minimal needs to enable and ensure the integration of genomics across nursing practice, education, research, and policy, and to prioritize a list of nine areas for action. The nine areas for action were established through group discussion and anonymous voting at a plenary session at the ISONG Annual 2016 Congress. The survey was administered online in November 2016 following an explanatory webinar. Other demographics and indicators used to assess readiness and inform landscape analysis were obtained through review of routinely available data from the WHO and United Nations (2017).

Analysis

Data were exported into Excel for analysis. Results were tabulated and analyzed using descriptive statistics. There are very few global nurse leaders, so the sample size is necessarily small. Given the small sample we have chosen to not provide percentages.

Results

Population

Nineteen countries were represented at the meeting, of which 18 completed the survey. One country was invited after the survey following a late cancellation and did not complete the survey. Seven organizations were represented, of which four were international, three were not restricted to nursing but had interprofessional membership, and five completed the survey, with two completing the survey from a country perspective. Five men were among the respondents. All delegates were fluent in English.

Country delegates were predominately nursing leaders within their country but did not always have genetics expertise, and most held academic positions. Delegates came from countries with populations (in thousands) ranging from 2,380 (Australia) to 319,929 (United States). The number of nurses and midwives per 1,000 population varied widely from 0.6 (Pakistan) to 17.8 (Switzerland; Table S1; WHO, 2017b).

Half the country delegates (9 of 18) reported that the main source of healthcare services was health insurance systems funded by the government, citizens, employers, or a combination of those entities. Of the remaining, 8 of 18 reported they had a government funded system of which 5 included additional user fees at the time of use. Only one country, Pakistan, reported a decentralized, private system. Country-specific gross domestic product spending on health

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care was as low as 3% (Pakistan) to as high as 17% (United States) (see Table S1) (WHO, 2017c).

Nursing Qualifications to Practice

Most countries reported that the qualification most nurses obtain to practice is a bachelor's degree (9 of 18) or associate degree (4 of 18), with fewer reporting obtaining a diploma (3 of 18) or certificates (2 of 18). Despite this variation in qualification, most countries reported that entry-level training was 3 or 4 years (16 of 18), with 2 of 18 countries reporting 5 years. Most indicated that training occurred in universities or colleges (13 of 18). While hospital-based training was still prominent in three countries, one reported tertiary institutes, and one was transitioning from hospital- to university-based training. Five countries required examination such as a licensure or registration examination to practice. Four countries indicated there was no statutory regulatory body responsible for maintaining a nursing register and setting standards for education and practice. One country reported they do not have a national professional organization to represent nurses' interests.

Required Genomic Training for Nurses

Only one country, Israel, indicated a requirement for all nurses to reach an agreed standard of knowledge and skills in genetics/genomics to practice, via a mandatory 28-hour course. Otherwise, the integration of genomics into nurse training was ad hoc and varied widely based on the country, with some countries reporting no genetic or genomic content included in training. Three countries indicated existence of genetic/genomic competencies applicable to all nurses regardless of clinical role, level of training, or specialty: Japan (Arimori et al., 2007); United Kingdom (Kirk, Tonkin, & Skirton, 2014); United States (Consensus Panel on Genetic/Genomic Nursing Competencies 2009; Greco, Tinley, & Seibert, 2012). Building on U.K. work, interprofessional competencies are available for European countries for primary, secondary, and tertiary (defined as genetic specialist) care (Skirton, Lewis, Kent, & Coviello, 2010). Only 6 of 18 countries reported visible leadership driving developments in nursing to incorporate genomics.

Specialist in Genomics

All 18 countries reported the existence of a specialist genetics service, often in centers of excellence and consisting of genetic testing and counseling provided by trained or accredited individuals whose positions varied by country but included physicians, genetic counselors, and some nurses. Only 5 of 18 indicated there was a recognized specialist genetics nursing role (nurses with specialized training in genetics). Of those countries, four indicated they had agreed standards for specialist genetics nurses. A few countries reported that some genetic counselors are also nurses.

Availability of Genomic Services

The scope of genomic services offered globally varied (Table S2). The most widely available genetic services consist of newborn screening (15 of 18) and prenatal screening (11 of 18), though not prenatal testing (5 of 17). Genomic services were mostly offered in specialized centers only. Some countries reported not having one or more of the following genomic

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services: risk assessment and genetic testing for disease susceptibility; tumor sequencing; targeted therapies; or sequencing of infectious agents. One country reported the only genomic service was newborn screening. Only a few countries reported that genomics of common disease services, when offered, were widely available outside a specialty center: disease screening (2 of 16), disease prognosis (1 of 16), pharmacogenomics (2 of 14), and sequencing for infectious agents (5 of 15).

The nursing roles delivering genomic services were a mixture within and between countries of specialist genetic nurse, specialist nurse, and advanced practice nurse (data not shown). Newborn screening, systematic family history taking, and prenatal screening were interventions where “any nurse” was most likely to be involved (eight, six, and five countries, respectively).

Key Challenges

Several potential challenges to clinical practice and nurse education were considered by both organization- and country-specific delegates. The most significant challenges or barriers to genomic integration into clinical practice consisted of (a) limited access to point of care educational information and clinical decision support; (b) lack of genomic expertise with limited training opportunities; (c) access to critical resources for training; and (d) resources that could link genetic variation to clinical implications (see Table S3a for specific data). High cost or lack of reimbursement and the need for resources to link genetic variation to clinical implications also ranked as significant challenges or barriers. Confusion over consent and privacy issues were considered as only a minimal challenge or barrier.

Delegates identified a need for a cultural shift in the role of nurses in genomics. Comments included the need for development of clear career pathways in genomics for the registered nurse and the wider nursing workforce. Recommendations focused on demonstrating the relevance to nursing leaders such as directors of nursing and those responsible for setting standards.

Education key challenges and barriers identified as the most significant included (a) insufficient curriculum time to cover genomics, (b) insufficient numbers of educators able to teach genomics, and (c) absence of required genomic competency assessments to practice nursing (see Table S3b for specific data). The absence of standards for genomic nursing education was viewed as significant by 12 countries as well as by ISONG, HEE, and ICN. Reluctance to consider different approaches to nurse training that facilitate integration of new knowledge and clinical advances, and the absence of national leadership in driving nursing genomics integration, were also important. Establishing relevance for nursing leaders involved in setting curricula was deemed critical as otherwise there is no incentive to prepare practitioners.

More than three fourths of the countries (14 of 18) reported other significant or major competing priorities, including financial and political uncertainty. The absence of national leadership driving genomics integration into nursing was considered a significant issue.

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Policy Initiatives

Country-specific policy initiatives have largely surrounded investments in large-scale genomic biomedical research. These include National Call for Research into Preparing Australia for the Genomics Revolution in Health Care; Brazil's National Institutes of Science and Technology and its creation of the Family Cancer Network and Institute of Oncogênomica; China Kadoorie Biobank; Japan's Genomic Medical Realization Promotion Council; the U.K. 100,000 Genome Project; and the U.S. Precision Medicine Initiative, now renamed the All of Us Research Program. Initiatives have also focused on specific health issues or ethical and regulatory considerations, such as Pakistan's Punjab Thalassemia Prevention Program; Germany's Genetics Diagnostics Law; Switzerland's National Criteria for Centers of Excellence in Rare Diseases; Taiwan's recommendations for prenatal and newborn screening; the German Ethics Council's position on genetic diagnosis; Turkey's regulation of Genetic Disease Diagnosis Centers; and the U.K. Rare Disease Strategy. However, only 10 of 18 reported these national initiatives acknowledge the implications for nurses. Fewer still include genomic training for nursing and other healthcare professions, although the United Kingdom's Genomics Education Programme is one exemplar (HEE, 2017).

Priority Areas for Action

The top three priority areas for action included raising awareness (22 of 22); education (21 of 22); and resources to support genomics in nursing (20 of 22). The creation of national and international collaborations also ranked highly. High priority areas included efforts to improve the "status and visibility of nurses and nursing, generally and in relation to role in genomics" as well as to facilitate the organization and delivery of genomic healthcare.

Discussion

To the best of our knowledge, this study represents the first landscape analysis of genomic healthcare services and nursing (education and practice) across multiple countries. Our aim was to conduct a landscape analysis that assessed the nursing and genomic trends in genomic literacy, clinical practice integration, and country-specific context inclusive of challenges and barriers. The current landscape revealed many global commonalities. National genomic sequencing initiatives are being implemented across the globe, and yet despite this, the largest single healthcare professional group, nurses, do not yet have genomics fully integrated into their practice, education, and policy. Genomic nursing literacy globally appears to be low given only one country requires genomic training, which is narrow in scope. Improvement may be limited, since the three primary education challenges were insufficient curriculum time for genomics, insufficient educators capable to teach genomics, and the absence of required genomic competency assessments to practice nursing. This contributes to limited genomic translation into practice, with most available services restricted to specialty clinics and not integrated into the general healthcare environment. Understanding this context informs priorities for action and identification of key strategies to influence change, including engagement and education of nursing practice and education leadership, an approach that may be useful for invigorating and sustaining any initiative (Calzone, Jenkins, Culp, & Badzek, in press). All of this can be greatly facilitated by global

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collaboration, the potential to learn from countries further along, and the sharing of expertise and resources to minimize duplication of effort.

Unsurprisingly, the availability and complexity of genomic-based health services varies between countries, with a range of nursing roles involved in their delivery. Most services are located within specialized centers, with more established activities like prenatal and newborn screening, available for many years, being more widely available and being delivered by the nonspecialist nurse. We anticipate that the transition from specialized center to services that are more widely available will occur over time as technology becomes increasingly accessible and genomics becomes embedded within mainstream healthcare practice. However, it is important to be realistic about the scale of the challenge and what this entails. Davies (2017) remarked on the need to reform professional attitudes towards genomics and for a new genomic paradigm to be integrated into all training curricula for all clinicians. According to Davies, adopting genomic technologies requires changes in the design, operation, and workforce of healthcare organizations and raises concerns about the international shortage of skills and expertise. Davies' comments highlight the importance of education, leadership, and willingness to change, the need for appropriate infrastructure, and the value of pooling international expertise.

The education challenges presented here resonate with those identified in the literature. The engagement of nursing leadership is vital to establishing genomics competency as a workforce priority (Calzone et al., in press; Jenkins et al., 2015; Leach, Tonkin, Lancastle, & Kirk, 2016). To achieve engagement, relevancy of genomics must be established, which requires that nursing leaders attain competency in genomics sufficient to inform their decisions on competency standards and infrastructure priorities, such as point of care decision supports (Jenkins et al., 2015). Targeting nursing leadership is challenging as they often view genomics as a niche specialty (Jenkins et al., 2015), but genomics represents a healthcare quality, safety, and cost issue in which larger policy mandates are needed to prioritize training in genomics for the practicing workforce (Calzone et al., in press). This is challenging given that most genomic initiatives globally focus on evidence generation, with limited attention paid to expanding the capacity of the existing healthcare workforce or the infrastructure necessary for effective translation of discoveries into practice. However, with the right government attitudes and investment, such as the U.K. 100,000 Genomes Project, evidence generation can be linked with increasing health professional capacity (HEE, 2017). Hoping that the solution will lie in the future nursing workforce is not realistic. The data from this landscape analysis document common global challenges that are well described in the literature, including lack of faculty capacity to teach genomics, a packed curriculum, and the absence of genomic educational standards (Jenkins & Calzone, 2014; Read & Ward, 2016). Addressing these deficits is hindered by the absence of regulatory bodies globally mandating some form of genomic nursing competency assessment.

The trajectories of genomic translation (outlined above) in both nursing practice (from specialized to widely available) and nurse education lend themselves to measurement. A tool that can capture country-specific and, in turn, global progress in integrating genomics into practice could be of great use to prioritize G2NA ongoing efforts and assess effectiveness.

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Genomics epitomizes a complex competency. The global workforce has little underpinning in the science of genomics, limiting capacity to understand the relevancy and even the literature given some of the terminology used. Many of the health or disease outcomes achieved by using genomics are not readily observable in terms of health or disease outcomes and can also consist of psychosocial outcomes, such as the value of knowing (Garrison, Mestre-Ferrandiz, & Zamora, 2016). For example, identifying an individual with a genetic predisposition to a disease such as cancer provides an opportunity to implement strategies aimed at risk reduction or early detection. Utilizing a pharmacogenomic test to inform treatment options may help alleviate adverse drug events and improve efficacy, superior to the trial and error approach most often utilized (Ciardiello et al., 2014). Rogers' *Diffusion of Innovations* (2003) documents that this complexity and lack of observability all slow adoption. Policy approaches, the involvement of critical healthcare leaders, and the utilization of opinion leaders, such as the G2NA delegates, can help facilitate adoption (Leach et al., 2016). But those G2NA country delegates alone will not be sufficient. The contribution of critical nursing organizations such as the ICN and STTI, which have considerable respect and position in the global healthcare arena, are positioned to influence nursing leaders and policy internationally, and were specifically targeted for participation in this effort. Genomics integration into practice is absolutely an interprofessional issue (Passamani, 2013). Nursing, through global collaboration and interface with critical nursing leaders, can be a catalyst for all health professions to achieve the capacity to integrate genomics into practice and education to realize the healthcare benefits.

The findings from our landscape analysis underpin the future work of the G2NA. This alliance is not targeting the genetic specialist, but is aimed at genomic integration in everyday nursing practice and education though the sharing of resources, expertise, and mobilization of organizations that can help influence nursing leaders and policy directions.

Limitations

These findings have some limitations that need to be considered. The instrument used to collect the data was developed by the project leadership team. Apart from content validity using expert reviewers, the instrument was not otherwise tested for construct validity and reliability as that was not appropriate given the very narrow target audience for this survey.

Given funding constraints, not all countries and international nursing organizations were represented, though the G2NA provides a platform for growth in this arena. Furthermore, the data are based on the knowledge and views of just one individual from each country or organization. This project utilized a purposive sampling strategy. While the authors attempted to identify people optimally positioned to address the state of nursing in their country or organization, a single individual may not have the full details associated with every survey item. There are very few high-level nurse leaders globally who can comment on the state of nursing practice in their country; thus, the numbers we could involve are limited. We do not claim that those involved were representative in any way of the entire nurse leader population.

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Given these limitations, our findings may not accurately reflect the global state of nursing and genomics. Therefore, these findings should be considered as one initial snapshot of a potential state of nursing in genomics that will inform the next steps in establishing the G2NA to facilitate and accelerate the integration of genomics into nursing practice.

Conclusions

The findings from this landscape analysis provide a foundation to inform the development of strategies to address common challenges and prioritize collaborative activities to accelerate the integration of genomics into nursing. The findings also support the concept of global commonalities of pathways to genomic adoption amenable to the development of a tool to guide and track progress. Now more than ever before, nursing exists in a global environment. By working together, we can mobilize information, resources, and strategies to realize the benefits of genomics for the patients that we serve.

Supplementary Material

Refer to Web version on PubMed Central for supplementary material.

Acknowledgments

Thanks are due to the Global Genomics Nursing Alliance delegates who gave their time to respond to the survey. We would also like to acknowledge the contribution of Dr. Juping Yu, University of South Wales, in retrieving and preparing data for Table S1.

This project was funded through a grant from the Wellcome Genome Campus Advanced Courses and Scientific Conferences and supported by Health Education England and the Intramural Research Programs of the National Institutes of Health, National Cancer Institute, and National Human Genome Research Institute, Division of Policy, Communications, and Education.

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Clinical Resources

- Genetic and Genomic Competency Center for Education. http://genomicseducation.net/
- Health Education England. Genomics Education Programme. https://www.genomicseducation.hee.nhs.uk/
- Your Genome. http://www.yourgenome.org/