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Abstract: The knowledge derived from the completion of the Human Genome Project is transforming the health care model, with implications for nursing and repercussions in teaching, practice and research. This article aims to reflect and discuss about genomics-based health care and its implications for nursing from the theoretical and conceptual framework of the “Essential Nursing Competencies and Curricula Guidelines for Genetics and Genomics”.

Keywords: Genetics. Genomics. Nursing education research. Nursing care.

INTRODUCTION

More than 150 years ago, Florence Nightingale demonstrated the importance of epidemiology principles and management of environmental factors in nursing practice.^{1,2} At the same time, Gregor Mendel published the results of his work, through which he explained the possible inheritance of biological traits that are transmitted from generation to generation.^{2,3} Nightingale and Mendel pioneered by introducing, respectively, environmental management fundamentals for the control of infections and genetic inheritance patterns.² These fundamentals and standards continued to evolve and have been applied by nurses worldwide to promote health, prevent disease and manage signs and symptoms of individuals and their families.²

The science of genetics that has expanded recently to genomics⁴ can be defined as the science that studies the genes alone, its operation and the mechanisms by which biological traits are passed from generation to generation and are part of an individual.⁵ The traditional genetic approach focuses on chromosomal abnormalities, genetic diseases and rare syndromes that can be inherited and affect 3% to 5% of the population worldwide.⁵ Genomics, first used in the early 80's⁶, includes the study of all genes in the human genome, as well as their interactions with other genes, and the environment, through the influence of cultural and psychosocial factors.^{7,8}

The completion of the mapping and sequencing of the human genome in 2003 led to the expansion of research technologies, which are now used to identify genetic and genomic factors, determining their influence on people's health.⁴ Information and technologies arising from the Human Genome Project (HGP) have the potential to: a) improve the early identification of individuals and families with or at risk of presenting a genetic condition; b) establish

measures for risk reduction and disease prevention; c) expand the genetic screening of individuals and families; d) support the prognosis and informed decision-making of subjects with or at risk of having genetic diseases; e) develop individualized and personalized care; f) make use of pharmacogenomics, through the selection and use of drugs based on individual genetic variation and metabolism thereof.^{9,10}

In addition, the completion of the HGP has brought a variety of genetic and genomic information, which has become essential to health care.¹¹ Thus, it has been proposed a new care model, which includes the diagnosis, prevention and therapy based on the genetic makeup of each individual, called genomics-based health care.¹² This type of care is concerned with health and disease outcomes as a result of combinations of the human genome and the influences of the external and internal environment of an individual.¹³ To understand the effects of changes in genes in the development of common and complex diseases, health professionals, especially nurses, need to incorporate knowledge of genetics and genomics to their professional practice.¹⁴

Reflecting on the process of nursing care in the different scenarios of nurse's performance is fundamental for the strengthening and the redefinition of their daily practice. Thus, this study aimed to reflect and discuss the implications of genetic and genomic sciences in nursing care, from a theoretical and conceptual framework. This study is justified since that nine of the ten leading causes of morbidity and mortality worldwide have genetic and genomics predisposition.¹⁵ In addition, the speed with which new genomic knowledge, with potential application to care are being discovered has been transforming the health care model that influence the clinical nursing practice.¹³ Thus, the relevance and justification of this study permeate the

contributions of these sciences in the post Human Genome Project era with impacts on health of individuals when considering this issue on nursing care interface.

Historical and conceptual aspects of the genetic and genomic insertion in teaching and nursing practice and the "Essential Nursing Competencies and Curricula Guidelines for Genetics and Genomics":

The issue of insertion of genetic science in nursing curriculum has been discussed for over 50 years.¹⁶ In the 1960s, nurses in North America and Europe began to describe the implications of genetics for their professional practice.^{8,17} In 1962, the first article with recommendations for the inclusion of genetic content in nursing curriculum was published.¹⁶

In 1988 the International Society of Nurses in Genetics (ISONG) was created with the mission of promoting scientific, professional and staff development of nursing in the world, by way of genomic information.⁸ Ten years after its foundation, the document The Scope and Standards of Genetic Clinical Nursing Practice was approved.¹⁷ This document provides international standards and guiding principles of the nursing practice in genetics and genomics.⁸ In addition to establishing the scope of professional practice, ISONG proposed the definition of nursing in genetics and genomics as the protection, promotion and optimization of health and abilities; prevention of diseases and injuries; the alleviation of suffering through the diagnosis of human response, and support the care of genetics and genomics health of individuals, families, communities and populations.⁸

One of the movements to prepare the workforce, faced to the new paradigm of care in the genomic era, is the teaching based on core competencies to this area of practice.^{11,18} Regarding the establishment of competencies for the role of interdisciplinary workforce in health, to incorporate the genetic and genomic care bases, the National Coalition for Health Professional Education in Genetics (NCHPEG), in 2001, was the first organization to establish specific competencies.¹⁹ This initiative served as a guide for creating competencies in genetics and genomics for nurses in many countries, especially the United States and the United Kingdom.^{20,21,22}

After the completion of the HGP, the National Human Genome Research Institute (NHGRI), the National Cancer Institute (NCI) and the National Institutes of Health (NIH), through a collaborative project, devised a proposal for genomics education for nursing.²⁰ The final document containing the proposal was entitled "Essential Nursing Competencies and Curricula Guidelines for Genetics and Genomics".^{20,21,23} The competencies were organized in two areas: professional responsibilities and professional practice. Professional responsibilities include aspects related to the evaluation of knowledge and values, cultural sensitivity, professional development and social support.²⁰ The standards for professional practice follow what is recommended by the nursing process and its steps, including the identification of genomic issues; interventions (the education offering, care and support); and nursing evaluation (genetics/genomics).²⁰

This document, which has been proposed as a standard, internationally recognized, for the integration of genetics and genomics to nursing practice, does not represent only the opinion of some researchers, or a single organization of nursing, or even a government corporation. More than that, the "Essential Nursing Competencies" represent a broad consensus of the community of nurses, approved by 49 nursing organizations and establish knowledge, attitudes and skills needed for these professionals, regardless of practice setting in which they are inserted and their training, whether general or specialist.²⁴ It is notable that the document that sets out the essential competencies does not replace the standards, i.e., the standards and existing guidelines for professional practice in nursing. Instead, it is intended to provide a genetic and genomic perspective on the care.¹¹

Future nurses need to understand not only the foundations of genetics and genomics, but also the implications of these sciences for their customers.²⁵ The American Association of Colleges of Nursing indicated that these are some of the competencies that graduate nursing professionals need to develop: (a) to be educated in the relevant genetic, genomics, pharmacogenetics and pharmacogenomics sciences; (b) to learn about the social impact of genetic and genomic trends in health policies; (c) to be able to access protective and predictive factors, considering those of genetic origin, which influence the health of individuals, families, groups, communities and populations; (d) to raise the health history, including family history, with genetic risks for current and future health problems; (e) to be able to access the evolution of knowledge in genetics and genomics, including specific therapies for customers; (f) to recognize the relationship of genetics and genomics to health, prevention, screening, diagnosis, prognosis, treatment, using the pedigree chart constructed from information collected from family history, using symbols and standardized terminology to such task.²⁶

In order to train nurses, the American Nursing Association (ANA) established that the nursing curriculum needs to prepare them, regardless of their level of expertise, whether generalists or specialists, through the inclusion of genetic and genomic learning experiences, sufficient for them to develop the "Core Competencies in Genetics and Genomics".²⁷

Nursing genomics-based healthcare in the context of nursing practice:

The inclusion of genetic content in nursing practice has its historical roots in community health, prenatal screening and pediatrics.¹¹ With the completion of the HGP, modifications have been incorporated into clinical practice and personalization of care through integration of genomics to health care.²⁸

Recently, leaders in nursing in genetics and genomics in countries like the United States, United Kingdom, Japan, among others, found that the absence of an effective representation of these disciplines in undergraduate curricula can be a major barrier to the integration of these sciences the various areas of professional practice.⁹ It is believed that the integration of these sciences to the daily life of nurses has been restricted for the following reasons: the workforce is not genetically or genomically qualified, not being able to

include this approach in implementing nursing actions; educational resources in the area are limited; the records in the medical documentation/communication are inadequate and lacking sufficient support tools for clinical decisions, such as to interpretation of genetic and pharmacogenomics testing.^{9,12}

In Brazil, both students and teachers of undergraduate courses in nursing still have a very conservative view of genetics and genomics, probably due to the content of current programs are uninformative about the attributions of nurses in this scenario and the absence of a specific direction for professional practice.²⁹ Soon, a political-pedagogical and clinical transformation is needed, to ensure that future nurses have genomic competencies to take care of their customers.^{18,30}

The nursing professional has an extraordinary opportunity to lead the creation of models of care for integration of genetics and genomics, considering each of the above-mentioned gaps.⁹ Nurses play leadership roles, and given this position, they have the chance to expand the health care by integrating it with genetics and genomics, once they have been trained for it.³¹

In Brazil there are several reports of nurses' performance in genetics and genomics. In the 80's nurses started their acting in prenatal counseling³² and later, in the 90's, oncology and geneticist nurses started activity of genetic counseling in oncology.^{33,34}

Nursing practice in genetics and genomics in Brazil is not based only on the recommendations of "Essential Nursing Competencies and Curricula Guidelines for Genetics and Genomics", but also on the regulations of the Law N°. 7498 of June 25, 1986, which regards about the Brazilian nursing practice, and on the Code of Ethics of Nursing Professionals in that country.³⁵ The activities performed by these professionals are related to care, to education, management and research, and include: a) holding nursing consultation (for example, the collection, recording, updating and validation of the family history, to the empowerment for informed decision-making for genetic testing); b) participation on clinical case discussions with the multi professional team; c) planning and implementation of educational actions for health professionals to identify patients and families with or at risk for genetically determined diseases (ex.: early detection of inborn errors of metabolism and hereditary neoplastic syndromes), d) identification of issues and research hypotheses, and development and implementation of research projects; e) implementation and articulation of knowledge related to ethical and legal aspects involved in genetics and genomics health care; f) attention to various subjective experiences of patients regarding genetic disorders and integration of subjective and objective data to understand the context, and prescribe and implement a personalized care plan.^{18,36}

These features show the importance of the nurse, in confidentiality and privacy issues related to genetic and genomic information; and the creation of educational resources. Only then, the ability to integrate these genomic discoveries with the practice will be achieved, in order to

optimize health outcomes in an appropriate and responsible manner.^{9,12}

The need for nurses, in an informed and competent way, to become intimately involved with health care based on genetics and genomics results from the emerging need to optimize health outcomes in the light of powerful tools able to resolve the real health needs of population.³⁶ This need has in view not only the epidemiological transition model already known, but it also has in view the political changes in the health sector, such as the recent approval of the National Comprehensive Care Policy for People with Rare Diseases in the Unified Health System (called SUS) in Brazil.^{37,38}

CONCLUSIONS

Nursing professionals in this century will have to deal with the challenges of explosion of genomic information to provide a personalized health care based on genomics. For this need can be met, it is essential that genetics and genomics are integrated into undergraduate curricula of future professionals. In addition, the teaching of these sciences must be guided by the establishment of curricula that are appropriate to the national context and the educational and health systems in which nursing professionals act.¹⁴

Recently, it has been recognized the need to structure at SUS a network of regionalized and hierarchical services that can offer the possibility of comprehensive care for people with rare diseases with or without genetic predisposition. In order to meet this need, the Brazilian Ministry of Health, through Ordinance N°. 199, of January 30, 2014, established and approved the National Policy on Comprehensive Care for People with Rare Diseases in SUS.³⁸ The organization of this policy is supported by lines of care for users ranging from primary care to the specialized care and is structured on two axes: axis I, composed of rare genetic diseases (80% of rare diseases); and axis II, composed of rare diseases without genetic predisposition. An important contribution of the Policy is the need for training professionals with genetic and genomic knowledge, able to perform genetic counseling, in the SUS.³⁸ However, it still presents some challenges and lacks regulation and implementation.³⁷

Although nursing, as a profession, already recognizes the importance of genetics and genomics in the education of nurses in clinical practice and research, surveys in several countries show that these contents are still limited in undergraduate programs of these professionals.⁹ Therefore, there is urgency to prepare the workforce of health professionals, especially nurses, considering the above aspects, in line with the principles of SUS.³⁷ Also, it is necessary to recognize the accomplishments of Brazilian nurses specialized in genetics and genomics, so that such professionals can, especially considering the new policy above mentioned, serve as a member of the multi professional team of comprehensive care for people with rare diseases under the SUS and have their right to empower themselves assured and to act as a genetic counselor.

REFERENCES

- [1]. Nightingale F. Notes on nursing: What it is and what it is not. London: Harriso. 1859.
- [2]. Williams JK. From Nightingale and Mendel to genomics in health care for children. *J PediatrNurs*. 2009;24(2):171-172.
- [3]. Jorde LB, Carey JC, Bamshad MJ. Medical genetics. 4th ed. Philadelphia: Mosby/Elsevier; 2010.
- [4]. Lea DH, Skirton H, Read CY, Williams JK. Implications for educating the next generation of nurses on genetics and genomics in the 21st century. *J Nurs Scholarsh*. 2011;43(1):3-12.
- [5]. Feetham S, Williams JK. Genetics in nursing. Geneva: International Council of Nurses; 2004.
- [6]. McKusick VA, Ruddle FH. Toward a complete map of the human genome. *Genomics*. 1987;1(2):103-106.
- [7]. Consensus Panel on Genetic/Genomic Nursing Competencies. Essentials of Genetic and Genomic Nursing: Competencies and Curricula Guidelines and Outcome Indicators. 2nd ed. Silver Spring, MD: American Nurses Association; 2009.
- [8]. International Society of Nurses in Genetics (ISONG). American Nurses Association (ANA). Genetics/genomics nursing: scope and standards of practice. Silver Spring, Maryland: American Nurses Association, 2007.
- [9]. Calzone KA, Cashion A, Feetham S, Jenkins J, Prows CA, Williams JK, et al. Nurses transforming health care using genetics and genomics. *Nurs Outlook*. 2010;58(1):26-35.
- [10]. Syuruna EV, et al. Genome based health literacy: a new challenge for public health genomics. *Public Health Genomics*. 2011;14(4-5):201-210.
- [11]. Houwink EJ, Henneman L, Westerneng M, Van Luijk SJ, Cornel MC, Dinant JG, Vleuten CV, et al. Prioritization of future genetics education for general practitioners: a Delphi study. *Genet Med*. 2012;14(3):323-329.
- [12]. Jenkins J, Bednash G, Malone B. Bridging the gap between genomics discoveries and clinical care: nurse faculty are key. *J NursScholarsh*. 2011;43(1):1-2.
- [13]. Jenkins JF, Calzone KA. Are nursing faculty ready to integrate genomic content into curricula? *Nurse Educator*. 2012;37(1):25-29.
- [14]. Skirton H, Lewis C, Kent A, Coviello DA. Genetic education and the challenge of genomic medicine: developing of core competences to support preparation of health professionals in Europe. *Eur J of Hum Genet*. 2010;18(9):972-977.
- [15]. Guttmacher A, Collins FS. Genomic medicine: a primer. *N Engl J Med*. 2002;347(19):1512-1520.
- [16]. Brantl, VM, Esslinger PN. Genetic implications for the nursing curriculum. *Nurs Forum*. 1962;1:90-100.
- [17]. International Society of Nurses in Genetics (ISONG). American Nurses Association. Statement on the Scope and Standards of Genetics Clinical Nursing Practice. Washington, DC: American Nurses Association. 1998.
- [18]. Flória-Santos M, Nascimento LC, Bisson GS, Lopes-Júnior LC, Pinto PS. Desordens genéticas: o papel do enfermeiro. *PROENF Saúde do Adulto SESCAD*. 2011;6(3):71-110.
- [19]. National Coalition for Health Professional Education in Genetics (NCHPEG). Core Competencies in Genetics Essential for all HealthCare Professional (3rded.)2007. Retrieved from <http://www.nchpeg.org/>
- [20]. Jenkins J, Calzone KA. Establishing the essential nursing competencies for genetics and genomics. *J Nurs Scholarsh*. 2007;39:10-16.
- [21]. Kirk M, Tonkin E, Burkes S. Engaging nurses in genetics: The strategic approach of the NHS National Genetics Education and Development Centre. *Journal of Genetic Counseling*. 2008;17(2):180-188.
- [22]. National Health Service National Genetics Education and Development Centre (NHS). Telling stories: understanding real life genetics2010. Retrieved from <http://www.tellingstories.nhs.uk>
- [23]. Jenkins J, et al. Essentials of genetics and genomics nursing: competencies, curricula, guidelines, and outcome indicators. 2nd ed. Silver Spring, Maryland: American Nurses Association. 2009.
- [24]. Calzone KA, et al. establishing the outcome indicators for the Essential Nursing Competencies and Curricula Guidelines for Genetics and Genomics. *Journal Professional Nursing*. 2011;27(3):179-191.
- [25]. De Sevo MR. Genetics and genomics resources for nurses. *J Nurs Educ*. 2010;49(8):470-474.
- [26]. American Association of Colleges of Nursing (AACN). The Essentials of Baccalaureate Education for Professional Nursing Practice. Washington, DC: Author, 2010.
- [27]. Greco KE, Tinley S, Seibert D. Development of the essential genetic and genomic competencies for nurses with graduate degrees. *Annual Review of Nursing Research*. 2012;29. New York: Springer Publishing Co.
- [28]. Blix A. Personalized medicine, genomics, and pharmacogenomics. *Clin J Oncol Nurs*. 2014;18(4):437-441.
- [29]. Novoa MC, FróesBurnham T. Desafios para a universalização da genética clínica: o caso brasileiro. *Revista PanamericanaSalud Publica*. 2011;29(1):61-68.
- [30]. Flória-Santos M. Genetics and nursing: the nurse's role in genetic cancer counseling and molecular study of Breast Cancer and Ovarian Syndrome Hereditary. 2003. Thesis (PhD). Ribeirão Preto Medical School, University of São Paulo, Ribeirão Preto 2003.
- [31]. Andrews V, Tonkin E, Lancaster D, Kirk M. Identifying the characteristics of nurse opinion leaders to aid the integration of genetics in nursing practice. *J AdvNurs*. England. 2014;70(11):2598-2611.
- [32]. Abrahão AR. A integração da genética na prática clínica do enfermeiro. *Acta Paul Enf*. 2000;13(1):203-206.
- [33]. Flória-Santos M, Ramos ES. Genomic-based nursing care for women with Turner Syndrome: genomic-based nursing care. *Rev Latino-Am Enfermagem*. 2006;14(5):645-650.
- [34]. Silva TBC, MacDonald D, Ferraz VEF, Nascimento LC, Santos CB, Lopes-Júnior LC, Flória-Santos,

- M.Perception of cancer causes and risk, family history and preventive behaviors of users in oncogenetic counseling. *Revista da Escola de Enfermagem da USP*.2013;47(2):377-384.
- [35]. Federal Consul of Nursing (COREN). Law N°. 7498 of June 25, 1986, which regards about the nursing practice, and on the Code of Ethics of Nursing Professionals. 1986.
- [36]. Flória-Santos M, Santos EMM, Nascimento LC, Pereira-da-Silva G, Ferreira BR, Miranda DO, Lopes-Júnior LC, Pinto PS. Oncology Nursing practice from the perspective of genetics and genomics. *TextoContexto – Enferm*. 2013; 22(2):526-533.
- [37]. Lopes-Júnior LC, Flória-Santos M, Ferraz VEF, Villa TCS, Palha PF, Bomfim EO, Abrahão CA, Silva S. Feasibility of Integral Attention in Clinical Genetics in National Health System: expanding the discussion. *Texto & Contexto – Enfermagem*. 2014; 23(4):1130-1135.
- [38]. Brazil. Ministério da Saúde. Portaria MS/GM n° 199 of 30 January 2014. Establishing The National Policy on Comprehensive Care for People with Rare Diseases in Unified Health System (SUS). Brasília. 2014. Seção I.