

GENOMICS NURSING: GENETIC COUNSELING IN HEALTHCARE PRACTICES
ENFERMAGEM EM GENÔMICA: O ACONSELHAMENTO GENÉTICO NAS PRÁTICAS ASSISTENCIAIS
ENFERMERÍA GENÓMICA: EL ASESORAMIENTO GENÉTICO EN LAS PRÁCTICAS ASISTENCIALES

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ABSTRACT

This paper aims to propose a reflection on the nurse's activities in genomics area, in the current context of health, with emphasis on the role of genetic counselor. It also analyzes the importance of integrating these professionals in this area of activity, with the growing demand for this service in care practices.

Keywords: Genomics; Nursing; Genetic Counseling.

RESUMO

O presente artigo tem como objetivo refletir sobre a atuação do enfermeiro na área da genômica, no contexto atual da saúde, com ênfase no papel de conselheiro genético. Analisa também a importância da inserção desse profissional na referida área de atuação, frente à crescente demanda desse serviço nas práticas assistenciais.

Palavras-chave: Genômica; Enfermagem; Aconselhamento Genético.

RESUMEN

Este artículo tiene por objetivo reflexionar sobre la actuación del enfermero en el área de genómica, en el contexto actual de la salud. También analiza la importancia de incluir a dichos profesionales en tal área debido a su creciente demanda en las prácticas asistenciales.

Palabras clave: Genómica; Enfermería; Asesoramiento Genético.

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INTRODUCTION

Aiming to perform the mapping of human genes and determine the sequence of its nucleotides, the "Human Genome Project" began in 1990. The initiative arose in the United States with the collaboration of several countries, including Brazil. Its proposal also included the creation of a database with all the genetic information found, facilitating the access to other members of the scientific community. The first stage of this project was completed in 2003, with major discoveries and many practical applications, including the genetic origin of many diseases and the association with its specific genes, treating or preventing many diseases afflicting the humanity.¹

This important research has brought scientific advances related to a new concept in origin, diagnosis, and treatment of diseases, rebounded significantly in health, creating new demand for knowledge to professionals. The incorporation of the concept of genomics in health practices is part of this new reality, exerting great influence on supportive care.²

The genetic information addresses the data obtained from genetic sequences, cytogenetic analysis and the study of hereditary traits. Genomics covers the individual genetic predispositions and his interactions with environmental trigger factors, aiming at a personalized and more effective prophylaxis.³ This area of knowledge is essential in nursing practice because, originally, most of the diseases has a genomic component.⁴

Genomics Nursing is an area expanding and extremely important for care practice that already has documents supporting the work of geneticist nurses in Brazil. One of these publications is the Opinion N° 032, 2011 of the Regional Council of Nursing of São Paulo (COREN-SP), which concludes that the nurse can work in the field of genetics. According to COREN-SP, the nurses are incorporating this knowledge into their practice, both as directors and as educators, acting as determined by institutional protocols.⁵

Another legal support for the work of nurses in the genetics area was the publication of the Resolution of the Federal Council of Nursing (COFEN) N° 468/2014. Its content establishes guidelines for the private work of nurses in genetic counseling in the nursing team.⁶

Thus, the nurse can perform genetic counseling to guide people and their families in all issues to possible diagnosed changes, discussing the ways of prevention and treatment. This professional can also contribute to the development of research projects that aim to improve the population's quality of life.⁴

Given the above, this article aims to reflect on the role of nurses in the genomics area, in the current context of health, with emphasis on the role of genetic counselor.

EVOLVING CHARACTERIZATION OF THE GENETIC COUNSELING

Genetic counseling consists of a monitoring of the individual and/or his family by a skilled professional to provide care related to prevention, diagnosis, prognosis and treatment of diseases related to genetics. This service aims to educate patients on all aspects involved with this problem, using a script that varies as the institutional protocols.⁵

With the use of knowledge in genetics, healthcare is applied in the care to the population about 40 years old.⁷ In some countries, such as USA, Canada, England and Japan, the division of nursing functions in genetic consultations, as a participant of a multidisciplinary team, is already well defined. This professional offers quality services based on protocols that consider ethical, legal and social issues.³

Aiming at the healthcare of the population and devoting to the genetic area, both for the professionals and for the general public, the International Society of Geneticists Nurses was founded in 1988 (ISONG – International Society of Nurses in Genetics) aimed to promote a scientific and professional development of its members, encouraging and disseminating findings related to genomics that will contribute to improving people's quality of life. In 1998, it presented a document that reflects the application of professional skills and responsibilities of nurses working in genetics and genomics around the world.³ This document was subsequently approved by the American Nurses Association (ANA), which is responsible for represent these professionals, providing a positive and realistic view of nursing, emphasizing improvements in health care.⁸

Following the global expansion of this theme in health and given the need to cover care based on genetics and genomics by of its importance, the Ordinance N° 81 was created in Brazil on 20 January 2009, established under the Unified Health System (SUS) and the "National Comprehensive Care Policy in Clinical Genetics" (PNAIGC), to structure a care service specialized to all individuals and families. Among other information, the creation of this Ordinance considered epidemiological data showed that about 5% of pregnancies result in the birth of children with some congenital anomaly, and in developing nations, the predominantly genetic etiology conditions are about 20% of the causes of infant mortality.⁹

Inserted congenital anomalies in Chapter XVII of the International Classification of Diseases (ICD-10), as the last update of the Department of Health System Information - DATASUS, constitute the second cause of infant mortality in Brazil, second only to some conditions originating in the perinatal period, according to Chapter XVI - ICD-1010, demonstrating the persistence of the problem since the creation of the policy.

The special emphasis on genetic counseling was addressed in Ordinance N° 81/2009, and this tool recognized as an essential

factor in health care.⁹ However, even with the creation of this Ordinance, there is a lack of regulation of PNAIGC, still in idealizations plan because of its wide range of assumptions and actions.¹¹

Among the many problems that hinder its implementation at the national level, we can mention the unequal geographical distribution of genetic services in the country, which are mainly concentrated in the South and Southeast region,¹² following a population numerical logic.¹³

Regarding legislation, advances that make this matter remain in Brazil. On January 30, 2014, the “National Policy for Integral Attention to People with Rare Diseases” was established which aims to reduce morbidity and mortality from these diseases. In Article 12 states that one of the care structure axes is composed of rare genetic diseases, which are organized by groups that include congenital or late onset defects, intellectual disability and inborn errors of metabolism. This policy reinforces the contribution of nurses in care teams, defining the professional as essential in the group that will make the care services specialized in rare diseases, including genetic counseling.¹⁴

NURSES AS GENETIC ADVISOR

By acting as a genetic counselor, the nurse must have a knowledge of legal principles and human rights to deal with all the issues involved in this service.¹¹

Often, nurses are faced with situations that require knowledge of the genomic area that will be used in low-complexity procedures, carried out in primary care focused on prevention. These requirements also include identification and appropriate management of people with genetic diseases or even in specialized units and centers of excellence, where the work of an interdisciplinary team is essential for the monitoring of patients and families. This articulation correctly performed allows decreasing the suffering of patients and families, avoiding wrong and unnecessary referrals and correcting introduction of treatments, allowing an individualized approach that directly reflect the quality of life of these people.⁹

Genetics knowledge provides the opportunity to diagnose, prevent and treat diseases, but there is a potential inherent and complex risk, involving ethical implications on the assistance. Questions covering the confidentiality and privacy of genetic information, the control of this information, how it will be used, the preparation of professionals to deal with them, among other topics and questions are part of the complex ethical issues related to genetic technology.¹⁴

The nurse should be prepared to mitigate the psychological consequences arising from the use of genomics in health practices, and to know and respect the privacy protocols and confidentiality of genetic test results.³ In this context, uncertainty about the use of genetic information will emerge, af-

fecting society and individuals, raising debates about how people will react on tests to determine a disease with no available treatment or unfavorable prognosis. Did the parents have the right, in default of the will of the children, trying to identify likely future diseases? Or Will this be just an unnecessary anticipation with psychological effects on the whole family?¹⁵ All these issues should be considered when using genetic information.

Even with the discussions on the ethical and psychological issues involved in the care and genetic counseling, benefits of a specialized monitoring are undeniable. Moreover, the nurse, based on art. 11 of Law N° 7,498/86, can perform nursing consultations¹⁶ and, as a member of a healthcare team, they have support to prescribe care according to the stages of care, such as data collection, diagnosis, planning, implementation and evaluation.⁵

During the data collection stage, as a genetic counselor, the nurse investigating the family history of the patient, allowing to prepare a periodogram, including kinship of first, second and third degrees, affected or not by disease or deceased. From that, nurses trace their plan of care focused on individual characteristics and can be used to pre-genetic counseling and post-tests, working with an interdisciplinary team, following the institutional flows and norms.⁷

Genetic, biological, behavioral, social knowledge and related fields are the bases underlying the practice of nursing in Genomics. Genetic counseling, held from nursing consultations conducted by trained professionals, can ease the anguish and suffering of people with genetic morbidities and also their parents, warning them of the existence or not of the risk of recurrence hereditary diseases. Also, when detected the problem, this service enables knowledge of the prognosis and the performance of a conscious planning of prevention of diseases related to genetic disorders, which can facilitate treatment with a care plan directed to the condition of the patient.⁷

FINAL CONSIDERATIONS

Even under the SUS, there is no definition of the nurse's role as a genetic counselor; nursing is entering this new scenario. In private institutions and some teaching hospitals that have staked out the area, backed by decrees and laws governing its activities in performing nursing consultations, there is satisfactory and qualified work of nurses as interventions provider in the genomics area. The increasing demand of people needing genetic counseling services is a warning for the need for improvement in that area, within the current framework of SUS.¹¹ The improvement of nursing knowledge in genetics and genomics can be an excellent strategy for improvement in prevention, diagnosis, and treatment, contributing to the reduction in morbidity and mortality linked to genetic diseases.

It is essential that nurses improve their genetics and genomics knowledge and encourage its use by other members of their team, not only to adapt to this new demand but also actively participate in treatment decisions, assuming effectively and efficiently their role of genetic counselor to the community and other health professionals.

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