
ONCOLOGY NURSING PRACTICE FROM THE PERSPECTIVE OF GENETICS AND GENOMICS

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ABSTRACT: This study aimed to reflect on oncology nurses' practice from the perspective of genetics and genomics, and their role as a member of the multiprofessional and interdisciplinary cancer genetics counseling team. This reflection is a result of the detailed reading of literature in the area, increased by the authors' experience and research group discussions. In the course of this work, it was verified that the nurse needs to consider genomic-based health care and incorporates essential competencies. These competencies include the ability to mobilize genomic resources in the family history assessment and in the guidelines on genetic testing for families at risk for hereditary neoplastic syndromes. The nursing staff may act as a reference for other members of the health team, with the potential to integrate their knowledge on care, teaching and research in oncology from the viewpoint of genetics and genomics.

DESCRIPTORS: Nursing. Oncology. Genomics.

ATUAÇÃO DO ENFERMEIRO EM ONCOLOGIA NA PERSPECTIVA DA GENÉTICA E GENÔMICA

RESUMO: Este artigo tem como objetivo refletir sobre a atuação do enfermeiro em oncologia, sob a perspectiva da genética e da genômica, e sobre seu papel na condição de membro integrante da equipe multiprofissional e interdisciplinar de aconselhamento genético oncológico. Trata-se de uma reflexão, fruto de leitura minuciosa da literatura da área, acrescida da experiência dos autores e discussões em grupo de pesquisa. No transcorrer desse trabalho, foi possível constatar que o enfermeiro precisa considerar o cuidado de saúde baseado em genômica e apropriar-se de competências essenciais. Essas competências abrangem a habilidade de mobilizar recursos genômicos na coleta da história familiar e nas orientações sobre testes genéticos a famílias em risco para síndromes neoplásicas hereditárias. O profissional de enfermagem pode atuar como referência para os demais membros da equipe de saúde, com potencial para integrar seus conhecimentos na assistência, no ensino e em pesquisas em oncologia, sob a ótica da genética e da genômica.

DESCRIPTORIOS: Enfermagem. Oncologia. Genômica.

PRÁCTICA DE LA ENFERMERÍA EN ONCOLOGÍA EN LA PERSPECTIVA DE LA GENÉTICA Y GENÓMICA

RESUMEN: Este estudio objetivó reflexionar sobre la práctica del enfermero en oncología en la perspectiva de la genética y genómica, y su papel como miembro del equipo multiprofesional e interdisciplinario del asesoramiento genético oncológico. Esta reflexión es resultado de lectura atenta de la literatura, además de la experiencia de los autores y discusiones del grupo de investigación. En el curso de este trabajo, fue posible constatar que el enfermero debe tener en cuenta el cuidado de salud basado en genómica y se apropiar de competencias esenciales. Estas competencias incluyen habilidad de movilizar recursos genómicos en la colecta de la historia familiar y orientaciones sobre testes genéticos para familias en riesgo de síndromes neoplásicas hereditarias. El profesional de enfermería puede actuar como referencia para los demás miembros del equipo de salud, con posibilidad de integrar sus conocimientos en asistencia, enseñanza e investigación en oncología, desde el punto de vista de la genética y genómica.

DESCRIPTORIOS: Enfermería. Oncología. Genómica.

INTRODUCTION

From a physiopathological viewpoint, malign tumors have genetic and molecular bases and can be acknowledge as genetic problems caused by genomic instability.¹⁻² Genomics, a term coined by McKusick and Ruddle,³ is the joint study of all genes in the human genome, including mutual interactions and with the environment, as well as the physical, psychosocial and cultural factors involved, and its implications for health and nursing care.⁴⁻⁵

Genomics applied to health reflects the implications of the Human Genome Project in clinical practice and on the personalization of care.⁶ These implications have changed the health care paradigm, the classification of illnesses, treatment and symptoms management. Through the mapping of the human genome, traditional models for health promotion, disease prevention and the production of new patterns of professional practice are changing, especially in oncology.⁶⁻⁸

Based on these premises, the aim in this paper is to present a reflection of oncology nurses' activities from a genetic and genomic perspective, as well as about their role as a member of the multiprofessional and interdisciplinary cancer genetics counseling team. This reflection derives from careful reading of literature in the area, in addition to the authors' experience and discussions in the GenomicsNursing Study and Research Group.

NURSING ACTIVITIES IN THE AGE OF GENOMICS

For about fifty years, in accordance with scientific discoveries that consequently change care practices for the population, nurses have delivered health care based on genomics, which incorporates gene-based diagnosis, prevention and treatment. In that perspective, health manifestations can be considered as a result of combinations between the human genome and environmental influences.⁹⁻¹¹

One important landmark for nursing professionals' activities was the foundation of the International Society of Nurses in Genetics (ISONG), which currently comprises approximately 400 members in 14 countries, including Brazil.¹² This organization is responsible for defining and establishing the scope of nurses' practice in genetics and genomics on an inter-

national scale. ISONG sets care standards and guidelines, and professional competency levels.⁵ In Brazil, these guidelines direct nurses' actions, which have been similar to other countries, with the first Brazilian publications on the theme dating back to the 1980's.¹⁰ Since then, this number has gradually increased through the conquest of new activity contexts.¹⁰

Essential nursing competences in genetics and genomics

At the same time as publications that informed about the conclusion of the Genome Project, the National Human Genome Research Institute, located in the United States, published its view on the future of genomics, which highlighted the need for workforce training and education for health care delivery in the age of genomics.⁴ In response to this demand, nurses who work at the National Institutes of Health (NIH) planned a nursing education proposal and established the "Essential Genetic and Genomic Nursing Competencies and Curricula Guidelines" (Picture 1).¹³

In 2008, the American Association of Colleges of Nurses, which determines the curriculum contents of North American nursing Bachelor programs, reformulated its guidelines in view of the complexity of nursing practice in the 21st century. This association, in turn, reformulated the document "The Essentials of Baccalaureate Education for Professional Nursing Practice", which started to recommend that nurses: (a) receive education on relevant genetics, genomics, pharmacogenetics and pharmacogenomics sciences; (b) learn about the social impact of genetic and genomic trends in health policies; (c) be capable of discovering protective and predictive factors, considering genetic factors, which influence the health of individuals, families, groups, communities and populations; (d) raise the health history, including the family history, of genetic risks for current and future health problems; (e) be capable of discovering the evolution of genetics and genomics knowledge, including specific client treatments; (f) acknowledge the relation between genetics and genomics and health, prevention, screening, diagnosis, prognosis, treatment selection, treatment effectiveness monitoring, using the pedigree, constructed based on information collected from the family history, using standardized symbols and terminology for that purpose.^{8,14}

Picture 1 - Essential genetic and genomic nursing competencies and curricula guidelines

Area	Competencies
Professional responsibilities	
Recognize when one's own attitudes and values related to genetic and genomic science may affect care provided to clients.	
Advocate for clients' access to desired genetic/genomic services and/or resources including support groups.	
Examine competency of practice on a regular basis, identifying areas of strength, as well as areas in which professional development related to genetics and genomics would be beneficial.	
Incorporate genetic and genomic technologies and information into registered nurse practice.	
Demonstrate in practice the importance of tailoring genetic and genomic information and services to clients based on their culture, religion, knowledge level, literacy, and preferred language.	
Advocate for the rights of all clients for autonomous, informed genetic and genomic-related decision-making and voluntary action.	
Professional practice	
Nursing assessment	Demonstrates an understanding of the relationship of genetics and genomics to health, prevention, screening, diagnostics, prognostics, selection of treatment, and monitoring of treatment effectiveness.
	Demonstrates ability to elicit a minimum of three generation family health history information.
	Constructs a pedigree from collected family history information using standardized symbols and terminology.
	Collects personal, health, and developmental histories that consider genetic, environmental, and genomic influences and risks.
	Conducts comprehensive health and physical assessments which incorporate knowledge about genetic, environmental, and genomic influences and risk factors.
	Critically analyzes the history and physical assessment findings for genetic, environmental, and genomic influences and risk factors.
	Assesses clients' knowledge, perceptions, and responses to genetic and genomic information.
	Develops a plan of care that incorporates genetic and genomic assessment information.
Identification	Identifies clients who may benefit from specific genetic and genomic information and/or services based on assessment data.
	Identifies credible, accurate, appropriate, and current genetic and genomic information, resources, services, and/or technologies specific to given clients.
	Identifies ethical, ethnic/ancestral, cultural, religious, legal, fiscal, and societal issues related to genetic and genomic information and technologies.
	Defines issues that undermine the rights of all clients for autonomous, informed genetic- and genomic-related decision-making and voluntary action.
Referral activities	Facilitates referrals for specialized genetic and genomic services for clients as needed.
Provision of education, care, and support	Provides clients with interpretation of selective genetic and genomic information or services.
	Provides clients with credible, accurate, appropriate, and current genetic and genomic information, resources, services, and/or technologies that facilitate decision-making.
	Consider genetic and genomic influences on personal and environmental risk factors.
	Incorporate knowledge of genetic and/or genomic risk factors.
	Uses genetic- and genomic-based interventions and information to improve clients' outcomes.
	Collaborates with healthcare providers in providing genetic and genomic health care.
	Collaborates with insurance providers/payers to facilitate reimbursement for genetic and genomic healthcare services.
	Performs interventions/treatments appropriate to clients' genetic and genomic healthcare needs. Evaluates impact and effectiveness of genetic and genomic technology, information, interventions, and treatments on clients' outcome.

Resource: Jenkins J, Calzone KA.¹³

Interfaces between academic preparation, professional activity levels and genomics-based health care

Nursing in genetics and genomics is defined as health protection, promotion and optimization; disease and injury prevention; relief of suffering through the diagnosis of human responses, and advocacy, which involves argumentation, search for support and pro-active struggle to defend the rights of individuals, families and communities who receive genomic care.⁵ To take this definition into account in their activities, nurses' role and competences are determined according to their educational level and the complexity of their activities in clinical practice. The two activity levels, proposed according to a generalist or specialized academic background, are the basic and advanced level, respectively, both of which include the application of genetics and genomics in nursing assessment, diagnosis and interventions. These levels, however, differ according to the depth and range of knowledge, skills, professional experience and formal preparation for clinical activities.^{5,9,12} It is highlighted that, in Brazil, the competency levels required for practice are considered in accordance with the law that regulates professional nursing practice and have already been formally acknowledged.¹⁰

Nowadays, despite these new care approaches, many health professionals still consider genetics as a medical specialty only, and not as a part of their daily practice.¹² The increasing importance attributed to knowledge on individuals and their families' family history and genetic and genomic profile, however, will strongly influence preventive medicine in the years to come, not just in oncology, but in all health specialties.^{7,12} Hence, the emphasis needed on genomics-based professional education will turn into an important requisite to the extent that genetics gets integrated into the care routine and stops being solely a specialization area.^{6-7,9}

NURSING INTERVENTIONS IN RISK ASSESSMENT AND IN CANCER GENETIC COUNSELING

In the mid-1990's, changes in rare genes were identified, which could predispose their bearers to an increased risk of malign tumors.^{1,2} On that occasion, for the first time, the American Society of Clinical Oncology proposed recommendations for

the accomplishment of genetic tests, which became part of the oncology care delivered to clients with breast, ovarian, colon, stomach, uterine, thyroid and other primary tumors.² Since then, individuals with a family history of these and other tumors have contacted, or have been referred to specialized genetics services for risk assessment and genetic tests, in case of the probability of finding genetic mutations that predispose to cancer.^{2,12}

As the public gets further access to information about the importance of genetics in cancer development, nurses who work with oncology patients should be prepared to respond to new care demands.^{12,15} Several societies of nursing professionals in oncology, genetics and genomics have published documents that determine nurses' functions in risk assessment and genetic counseling in oncology. These functions can be divided into clinical (care), educational, organizational (consultant, coordinator and administrator) and research activities.¹⁵ To develop these activities, care systemization was proposed through the application of the nursing process in each risk assessment and genetic counseling phase, including: initial assessment, diagnosis, establishment of a care plan, intervention and result assessment, in accordance with ISONG recommendations.⁵

Educative actions

Education is one of the pillars of the cancer genetic counseling process and models for this activity include group and/or individual sessions.^{2,15} Educative actions need to be dynamic and interactive, applying participatory pedagogical strategies that are appropriate for adult education and can attend to their health needs. It is presupposed that those actions are outlined according to the subjects' comprehension abilities, educational level, ways of risk perceptions, personal and family history of tumors, in view of their cultural beliefs and habits.^{11,15} The main topics to be addressed include basic genetics and genomics concepts, cancer as a genetic and hereditary disease, etiological aspects of tumors, environmental risk factors and strategies for prevention and early diagnosis.^{2,15} During these sessions, diagnostic assessment questionnaires can be applied to obtain fundamental data on these individuals' personal and family history, as well as on environmental exposure factors and lifestyle aspects. These questionnaires can be answered at home to stimulate further contact between family members, and to

collect medical documents that can validate the previously reported family history, so that the collected information is as reliable as possible.^{2,11,15} The main aims of these educative practices are to enhance clients' knowledge on the genomic and hereditary aspects of tumors, provide clarifications on the need for future genetic investigation of these individuals and offer conditions for informed decision making about treatment options, monitoring and genetic tests.^{2,11,15}

Cancer Genetic counseling

Genetic counseling emerged in nursing literature at the start of the 1960's, emphasizing psychosocial support and case monitoring as nurses' main responsibility in the multiprofessional team.¹⁶ It became part of systemized nursing language when genetic counseling was included in the Nursing Interventions Classification (NIC), through which it was defined as an interactive process of help, focused on care delivery to individuals, families or groups with or at risk of developing or transmitting a congenital defect or genetic condition.¹⁶ Genetic counseling is also defined as a communication or education process, through which subjects and family members receive information on the nature, benefits, risks and meaning of genetic test results.^{2,11,16} During the counseling process, support is also offered to clients so that they can cope with the implications of possible cancer predisposition test results. Individuals who take these tests need to receive the orientations needed for informed decision making on their health and for voluntary and conscious consent, through the signing of the informed consent term, before blood collection for genetic analyses.^{2,11,16-17}

Nursing consultations

Nurses are professionals, who usually interact quite close and directly with clients, and in general, are the first to have contact with those individuals in health services. During the nursing consultation, they attempt to go beyond the biological by surveying subjects' health needs in their family context, which can be attended to through nursing interventions or forwarding to other multiprofessional genetic counseling team professionals.^{11,16} Interviews and physical investigation are instruments used for nursing assessment purposes. According to ISONG, the data collected during the nursing assessment include client's expectations; their personal and family

health history; data on the family dynamics and structure; information on their health practices; environmental and economic factors that can affect their health; adaptation and disease coping patterns; support systems; values and beliefs, as well as their knowledge about the influence of genetics and genomics on the etiology of their disease.⁵

Family history collection and risk assessment

Among the items in this assessment, cancer family history stands out, as many hereditary neoplastic syndromes are identified through a properly collected and validated history.¹⁻² Some skills are important to collect the family history, including: basic genetics knowledge, communication skills, ability to develop empathetic interpersonal relationships, persistence, and knowledge about ethical and legal aspects that may emerge.¹⁵

When collecting and interpreting family history data, nurses need knowledge about clinical markers that indicate inherited susceptibility to cancer, called "red flags", which include: (a) young age when diagnosed (generally younger than 50 years); (b) bilateral tumors in paired organs; (c) presence of multiple distinct tumors in the same organ; (d) multiple primary tumors in the same individual; (e) presence of tumors in two or more first or second-degree family members; (f) "constellation" of tumors in subjects or families, acknowledged as part of a hereditary neoplastic syndrome previously described in the literature; and (g) associations between cancer and benign injuries.^{1,15}

Health professionals should systematically collect data from the individuals who are going through the consultation, that is, the proband or index case.¹⁵ If (s)he is affected by cancer, it is important to report the primary tumor site, including evidence through anatomical pathology tests with data on the tumor's histologic type and age when the disease was diagnosed.¹ To identify a genetic heritage pattern, the family history should include at least three generations.¹¹ First, questions are asked about first-degree family members health history, always guided by the "red flags". Then, the history of second and third-degree family members is assessed.¹ Both the maternal and paternal branch should be investigated and, in case of death, the age and cause of death should be mentioned. Due to the founder effect observed in different genetic diseases, the ethnic and racial origin of each family member should be part of the collected data. Information about lifestyle and en-

environmental exposure to carcinogenic substances need to be investigated for all family members. In addition, all collected data need to be proven through medical files or reports, anatomical pathology test results and death certificates, as that is the only way to obtain a reliable assessment, leading to a precise diagnosis.^{1,15}

Without available genetic tests to detect susceptibility to certain types of cancer, the family history is an important genomic, effective and low-cost instrument to identify risk categories and individuals that may be more predisposed to tumor development.¹⁵ Based on the family history reported by the client, the nurse can construct the pedigree, which is the representation of this history, using standardized symbols. The pedigree makes it easier to visualize the family history, identification of the genetic heritage patterns and evaluation of individuals at risk, which facilitates access to these data during care delivery by the multiprofessional oncology genetic counseling team.^{1,15}

Like the pedigree, the personal and family history is a very useful resource to estimate the risk of developing cancer. Knowledge about the degree of risk is important for decision making on the accomplishment or not of genetic test, indication of clinical screening conducts and use of chemoprevention measures.² Various statistical methods and models have been proposed to quantify the risk associated with a family history of cancer.² After reaching the diagnosis and determining the risk of developing cancer, in accordance with the family's clinical characteristics, the health team advises the proband about the indication of predictive genetic tests for individual and/or family follow up, and also about the specific prevention and screening program for their case and their family.¹⁷

Genetic tests

When clients decide to take a genetic test, nurses need to broaden their orientations and include topics that need to be taken into account in their consultations, such as their motivations to take the test, how they will process the obtained result and their communication patterns with relatives.¹⁷ Clients' consent to submit to the genetic test needs to be voluntary, but nurses can participate as facilitators in this decision process, offering psychosocial support in the intent to help them understand the risks, benefits and limitations of the tests, their potential results, sensitivity and specificity of the test, without any implications

for their treatment or follow up. These topics need to be part of the informed consent form which, in accordance with ethical and legal standards, all clients need to sign before taking genetic tests.^{2,17}

Most Brazilian genetic counseling services offer tests for the prevalent hereditary neoplastic syndromes in the population, like hereditary breast cancer syndromes, when the most frequently tested genes are *BRCA1* and *BRCA2*; and colorectal and gynecological cancer syndromes, in which the repair genes *MLH1*, *MSH2*, *MSH6*, *PMS1* and *PMS2* can be investigated.^{1,2,17} Those tests are accomplished to detect germline mutations inherited in high-penetrance genes, which are associated with the tumor type observed in individuals and families who seek counseling.^{2,17} Other intermediary penetrance mutations have also been reported in families with hereditary breast and colon cancer, but their impact in clinical practice has not been well-established yet. Those high and intermediary-penetrance mutations are considered rare and, more commonly, inherited susceptibility to cancer is attributed to a number of varying sequences in low-penetrance genetic material. The genomic location of many of these low-penetrance variations has been established through association studies across the genome, which are not part of routine clinical recommendations yet. Private institutions already offer those tests though, through genetic tests offered directly to the consumer, whose potential impact in the population is completely unknown so far.^{2,15}

Follow up and returns

Clients follow up is an essential component of the genetic counseling process. The information clients receive during consultations is very complex, so that nurses need to follow the evolution of their clients across this process and evaluate their understanding about the counseling.¹⁵ The returns and follow up are important to update the family history, as well as to reinforce healthy behaviors with regard to cancer prevention and check clients' adherence to screening and early detection programs of future injuries. Ideally, nurses should contact the families once per year to check for changes in the family history.^{12,15}

POSSIBILITIES AND CHALLENGES

In view of the above, nursing professionals who work in oncology can apply their knowledge to assess clients' risk of tumor development, with

a focus on genetic and genomic aspects.¹⁵ This assessment should be a fundamental part of their routine and particularly include the collection and registration of the family history, facilitating forwarding to specialists. Nurses are the professionals who need to reinforce care recommendations and the information that genetics experts provide. They need to be competent to provide orientations and support, and constantly assess the individuals' reactions to the care received.^{12,17}

ISONG recommends that nursing professionals who want to act at a more advanced professional practice level in genetics and genomics take a graduate program that addresses those themes or oncology.^{5,15} Through that training, nurses will be apt for a detailed and personalized risk assessment of individuals and their relatives, using statistical model that include available algorithms to calculate probabilities of cancer and to identify possible bearers of genetic mutations.^{2,15} These professionals can discuss the implications of the genetic test and provide counseling; acting as references for other nursing team colleagues, and can also participate in and/or conduct research in the area.^{12,15,17}

Oncology personalized medical techniques are gradually permitting the detailed characterization of the tumors individual biology, and of their bearers.² The therapeutic decisions that are based on this personalization have included the integration of complex clinical-pathological, technical and socioeconomic considerations.^{2,6-7} Personalized medicine involves all health professionals and appoints the need to educate these subjects about the meaning and implications of genomic information applied to their daily professional reality.^{2,6} The increasing number of gene-based approaches and the technological advances deriving from the Human Genome Project are leading to redefinitions in the pathophysiology, prevention and treatment of different conditions. Although nursing represents the main workforce, which delivers care all over the world, most nurses have not taken the initiative yet to confront these advances, often due to knowledge deficits or difficulties to access updated information sources in the context of their daily practice.¹⁸ Nursing professionals need training and continuing education programs, as they will increasingly play a central role in the planning and execution of genomics-based health care.

CONCLUSION

Now more than ever, it is fundamental for nurses to gain essential competences in genetics

and genomics, which is no longer an option but a need in nursing practice in the 21st century. They need to advance and assume leadership roles on initiatives aimed at the integration of these sciences into oncology care, focused on individuals and their families. Thus, nursing professionals can serve as references for other health team members and, within a genetics and genomics perspective, apply their knowledge in clinical care, teaching and oncology research.

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