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Practices and ethical implications in genetic counseling: a scientific review from a nursing perspective

Práticas e implicações éticas no aconselhamento genético: uma revisão científica sob a perspectiva da enfermagem

Prácticas e implicaciones éticas en el asesoramiento genético: una revisión científica desde la perspectiva de la enfermería

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ABSTRACT

Objective: To investigate, through current literature, the role of nurses in genetic counseling, in addition to highlighting the contribution of this professional, highlighting its implications in clinical practice and health promotion. **Literature review:** The analysis of bibliographic sources revealed that genetic counseling plays a central role in the early identification of hereditary diseases, promoting the understanding of heredity, prevention strategies and the sharing of information between patients and their families. In addition, the role of nurses as a fundamental agent in facilitating this process was highlighted, contributing significantly to the quality of life of patients. **Final considerations:** It can be considered that genetic counseling is an essential tool in the approach to genetic conditions, with a direct impact on the prevention, diagnosis and management of diseases. In this context, nurses emerge as essential mediators, integrating genetic knowledge into care practice, strengthening the bond with patients and families and enhancing health outcomes.

Keywords: Genetic counseling, Genetics, Heredity, Nursing.

RESUMO

Objetivo: Investigar, por meio da literatura atual, a atuação do enfermeiro no aconselhamento genético, além de pontuar a contribuição desse profissional, destacando suas implicações na prática clínica e na promoção da saúde. **Revisão bibliográfica:** A análise das fontes bibliográficas revelou que o aconselhamento genético desempenha um papel central na identificação precoce de doenças hereditárias, promovendo o entendimento da hereditariedade, estratégias de prevenção e o compartilhamento de informações entre pacientes e suas famílias. Além disso, destacou-se a atuação do enfermeiro como agente fundamental na facilitação desse processo, contribuindo significativamente para a qualidade de vida dos pacientes. **Considerações finais:** Pode se considerar que o aconselhamento genético é uma ferramenta essencial na abordagem de condições genéticas, com impacto direto na prevenção, diagnóstico e manejo das doenças. Nesse contexto, o enfermeiro emerge como mediador essencial, integrando o conhecimento genético à prática do cuidado, fortalecendo o vínculo com pacientes e famílias e potencializando os resultados em saúde.

Palavras-chave: Aconselhamento genético, Genética, Hereditariedade, Enfermagem.

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RESUMEN

Objetivo: Investigar, a través de la literatura actual, el papel del enfermero en el consejo genético, además de resaltar el aporte de este profesional, destacando sus implicaciones para la práctica clínica y la promoción de la salud. **Revisión bibliográfica:** El análisis de las fuentes bibliográficas reveló que el asesoramiento genético juega un papel central en la identificación temprana de enfermedades hereditarias, promoviendo la comprensión de la herencia, las estrategias de prevención y el intercambio de información entre los pacientes y sus familias. Además, se destacó el papel del enfermero como agente fundamental para facilitar este proceso, contribuyendo significativamente a la calidad de vida de los pacientes. **Consideraciones finales:** Puedes considerar que el asesoramiento genético es una herramienta esencial en el abordaje de las condiciones genéticas, con un impacto directo en la prevención, diagnóstico y manejo de las enfermedades. En este contexto, el enfermero emerge como un mediador esencial, integrando el conocimiento genético en la práctica del cuidado, fortaleciendo el vínculo con los pacientes y familiares y potenciando los resultados de salud.

Palabras clave: Consejo genético, Genética, Herencia, Enfermería.

INTRODUCTION

Genetics is the area of science that investigates genes and their heredity. In a DNA molecule, the four nitrogenous bases can be organized in different sequences, each representing a specific set of genetic instructions. The information contained in DNA is encoded based on the order of the bases, generating a unique set of instructions that guides the synthesis of specific cellular products. According to the same authors, the sequences of bases or nucleotides contain information that, when encoded, results in characteristics in the genes (LUNA ME, et al., 2021).

During reproduction, the chromatin presents within the cell compacts into a chromosome, making this process of cell multiplication safer. However, failures occasionally occur, which result in harmful changes to the gene. “Some monogenic or multifactorial pathologies, such as diseases, are associated with this: autoimmune, oncological and metabolic diseases, among others” (LUNA ME, et al., 2021). In this sense, several genetic disorders can affect human health, such as Polycystic Ovary Syndrome (MEDEIROS AJG, et al., 2023), cancer, for example, breast cancer (MARTIN CA, et al., 2022), Turner syndrome (ARAÚJO ACP, et al., 2022), Marfan syndrome (GONÇALVES IM, et al., 2024), Dementia and Parkinson's (RECH RS, et al., 2022). According to the authors (COUTINHO HDM, et al., 2022) chromosomal syndromes are genetic disorders that affect intrauterine development, due to errors in haploid cells.

Medical Genetics applied to Nursing helps in the tracking and identification of probable genetic pathologies. In this context, as reported by the authors, nursing plays a fundamental role in the genetic counseling process. The nurse, when aiding the patient and their family, uses tools such as the pedigree to assess family history, contributes to reproductive planning and acts in the transmission and guidance of genetic information essential for the understanding of patients (AHMED MOHAMED H, et al., 2023).

Furthermore, during prenatal care, the nursing professional monitors the patient continuously, but their role extends beyond pregnancy, beginning with family planning, especially in the presence of a genetic disease, such as sickle cell anaemia. In these cases, the nurse plays an essential role in family planning, prenatal care and postnatal follow-up of the individual (ARAÚJO CM, et al., 2023). Composed of several aspects, it is an interdisciplinary process that aims to help individuals understand and deal with the medical and family implications of genetic information (AHMED MOHAMED H, et al., 2023). This process, according to the same authors, is extremely relevant for hereditary diseases, such as cystic fibrosis (ALI EA, et al., 2024), Huntington's disease (RIBEIRO PSC, et al., 2021), and several forms of hereditary cancer, such as breast and ovarian cancer associated with mutations in the BRCA1 and BRCA2 genes (MURAD A, 2023). In these circumstances, Genetic Counseling (GC) allows patients to make informed decisions based on data about genetic testing, prevention strategies, and therapeutic options. In this context, nursing assumes a fundamental role, especially in the interface between the patient and the health team (WAINSTEIN T, et al., 2023).

Nurses are supported to act as mediators between technical information and the patient, who may be experiencing high levels of emotional stress (BRASIL COFEN, 2024). For example, in cases of women with a family history of breast cancer, the nurse can help in understanding the risks and benefits associated with carrying out genetic testing to detect mutations in the BRCA genes. In addition, the nurse offers emotional support, helping the patient to deal with the impact of a diagnosis of genetic predisposition to cancer and with other issues involved in the process, such as the adoption of preventive measures (SERNA CDRL, et al., 2024).

The management of neurodegenerative diseases is another example of the role of the nurse, situations in which the health professional helps patients and their families to understand the implications of a positive predictive testand physical activity interventions appear to be beneficial for older adults with neurodegenerative diseases. In these cases, the nurse's role involves providing information about the progression of the disease, family planning options and helping the patient understand the available alternatives, respecting their autonomy and individual preferences (ALANAZI MA, 2024).

This study conducted a literature review with the aim of analysing and discussing the role of nurses in genetic counselling, emphasizing their practices, ethical implications, challenges and associated benefits. Thus, it sought to highlight the nurse as a central figure in the GC process, acting as a guide and mediator of complex information, in addition to providing emotional and ethical support to patients and their families.

LITERATURE REVIEW

Changes during cell reproduction can result in genetic mutations, which are often associated with the development of hereditary diseases. Although the human genome is largely uniform, specific modifications in the genetic material of a cell can trigger genetic disorders. When these changes occur in somatic cells, they are classified as mutations; in germ cells, they can give rise to genetic syndromes (MOORE L, et al., 2021).

This study explored these pathologies, as well as their relationship with nursing care practices. It was demonstrated that nurses play a crucial role in genetic counselling, guiding patients on genetic characteristics, inheritance patterns, and preventive strategies through genetic testing and diagnosis. In addition, their role in reproductive counseling for families with a history of genetic diseases stands out, providing detailed information on heredity and using genetic tests to predict and prevent diseases, such as breast cancer (AHMED MOHAMED H, et al., 2023).

GENETIC COUNSELING (GC): CONCEPTS AND RELEVANCE

Genetic counseling (GC) is a tool that deals with human problems and aspects related to the occurrence or risk of a genetic pathology in a family. Nursing professionals, for example, mediate this process and aim to assist patients and families in understanding the disease, diagnosis, preventive measures and management of the pathology. In addition, it also analyzes heredity, thus aiming to study risks and preventive measures (FELDMAN J, et al., 2023).

In this context, the use of GC in families with a history of hereditary diseases allows a detailed understanding of the specific heredity of that family and how this contributes to the transmission of pathogenic genes that can lead to the development of genetic pathologies. Since genetics studies the transmission of genes to future generations, GC assesses hereditary aspects and the incidence of certain conditions in a family, identifying risks and possible diseases that may manifest in the future. The use of genetic counseling in this context is highly beneficial, since, by knowing the risks, it is possible to adopt prophylactic measures or make an early diagnosis (PAULA LCC, et al., 2023).

During genetic counseling, the role of nursing is to serve as a mediator throughout the process. In this sense, the nurse acts as an intermediary between scientific and healthcare knowledge and patients and their families. Nursing, based on ethical and scientific principles, can use genetic counseling to evaluate the genetic aspects of a family, thus assisting in reproductive planning, preventive measures and the diagnosis and understanding of genetic diseases (NASCIMENTO DM e SILVA VA, 2023).

NURSE COMPETENCIES AND SKILLS IN GENETIC COUNSELING

Genetic counseling is defined, according to Pires (Pires, 2024), as a communication process that encompasses human problems associated with the occurrence or likelihood of a genetic pathology occurring in a family. This way, the nursing professional acts as a mediator in this process, studying the family history and outlining probable future occurrences of genetic disorders. In this way, enabling patients to know their family history, assisting in reproductive planning, in addition to transmitting knowledge to the patient about heredity (NASCIMENTO DM e SILVA VA, 2023).

The professional must have knowledge in Genetics, including hereditary aspects, nursing care for patients and families carrying a pathogenic gene or even the pathology itself, prophylactic measures, in addition to having knowledge on how to use tools such as the Heredogram for the study of families (REIS ASN, et al., 2021). According to the same authors, the mediator of the genetic counseling process must be grounded in essential ethical skills, such as respect for the subjectivity and autonomy of the patient, in addition to ensuring confidentiality, responsibility and professional competence. This role must promote the principles of beneficence and offer emotional support. Since genetic counseling involves communicating various genetic issues present in a family, it is essential to address scientific, interpersonal and ethical aspects (REIS ASN, et al., 2021).

Nurses seeking to work in GC must have in-depth knowledge of genetics. In this context, nurses' specialization in this area allows them to work as geneticists, ensuring that the professional has solid and specific training in genetics (BRASIL COFEN, 2024).

During the genetic counseling process, tools such as the pedigree can be used, which maps genetic aspects of a family, including marriages, affected individuals, recessive and dominant genes, consanguinity, among others. The pedigree provides a comprehensive view of family generations, allowing a detailed analysis of hereditary occurrences and risks, as well as the propagation of genes across generations (FELDMAN J, et al., 2023).

APPLICATIONS OF GENETIC COUNSELING IN NURSING ROUTINE

Genetic counseling applied to Nursing practice enables the monitoring of families, the analysis of the probability of genetic diseases, the guidance of patients when diagnosed with a genetic pathology, the prenatal monitoring of patients with genetic diseases, such as patients with sickle cell disease, the screening of predisposition, among others (AHMED MOHAMED H, et al., 2023).

A very frequent example in nursing practice, according to authors (BRITO BG e OLIVEIRA SB, 2023; COUTINHO HDM, et al., 2022) is prenatal monitoring and family planning. Thus, genetic counseling enables the study of risks of having a child with a genetic syndrome or disorder, in addition to enabling the monitoring of heredity and family guidance.

GC has a beneficial informative, preventive and diagnostic role. Some examples illustrate this positive effect of its application, such as reproductive planning based on family knowledge, the reduction of future generations affected by genetic pathologies, the identification of predisposition, early diagnoses and reduction of mortality (SILVA MPS e LOPES GS, 2023). In addition, another positive impact is the provision of knowledge to families at a theoretical and assistance level.

Although it is an extremely important area, there are limitations in specific knowledge about Genetics, tools for its implementation and a lack of investment in this area in nursing (ALMEIDA JF, et al., 2021).

THE ROLE OF THE NURSE IN THE APPLICATION OF GENETIC COUNSELING

Nursing professionals work directly in patient care, playing a fundamental role in guiding families and individuals, developing preventive strategies and predicting possible occurrences in future generations. Nurses are also responsible for transmitting knowledge and care to patients, positively impacting their quality of life, understanding pathologies and heredity, and promoting essential knowledge for the management and care of diseases (NASCIMENTO DM e SILVA VA, 2023).

One of the benefits of the nurse's role in genetic counseling is support for reproductive planning, which allows for the assessment of risks and occurrences, helping to identify possible diseases and adopt preventive measures. In the case of sickle cell anemia, a genetic disease, genetic counseling allows for the analysis of risks and identification of the probability that a child of carrier parents will be affected by the disease due to heredity. In this way, reproductive planning mediated by the nurse contributes to the parents' informed decision-making and to the in-depth understanding of the disease and associated hereditary patterns (COSTA PN,et al., 2024).

According to Martin et al.(MARTIN CA, et al., 2022)among the benefits of genetic counseling, the positive impact on reducing mortality and morbidity in hereditary diseases stands out. In the case of cancer, for example, the identification of pathogenic genes makes it possible to track possible future pathologies, allowing the adoption of prophylactic measures, knowledge of the respective risks and early diagnosis. In this way, mortality and morbidity rates can be significantly reduced.

Nursing plays a crucial role in promoting health and preventing genetic diseases. In care practices, nurses maintain direct contact with patients and their families, and in the context of genetic counseling, they act as a mediator in the process, facilitating the transmission of information and assuming the role of health educator. This professional analyzes the genetic and hereditary profile of a family, contributing to the prevention of genetic diseases through preventive guidance, diagnosis and counseling, thus promoting health in a comprehensive manner (AHMED MOHAMED H, et al., 2023).

Ethical considerations for genetic counseling

Ethics is a principle related to patient autonomy, ensuring their freedom to make informed decisions. In this context, the professional responsible for genetic counseling must provide the patient with detailed information about all stages of the process. This approach aims to ensure active patient participation, promoting understanding of the implications, phases, potential risks and associated results. In addition, it is essential to address the scientific and ethical issues involved, ensuring informed and conscious decision-making (NORA CRD, et al., 2022).

Confidentiality and privacy are fundamental pillars in the genetic counseling process, considering that patient and family data constitute sensitive information. In this context, professionals must strictly ensure the protection of this information, avoiding its disclosure or undue exposure. It is essential that the patient has their privacy protected and feels safe to actively participate in the genetic counseling process. Confidentiality, in addition to being a fundamental ethical principle, is an inherent requirement of professional practice and must be strictly observed (LUGO NT, 2023).

Ethics in genetic counseling, according to Nora et al. (2022), covers topics such as autonomy, confidentiality, justice and beneficence. Patients must be properly informed so that they can make decisions autonomously and consciously, preserving the privacy of their genetic information, which, in turn, may have implications for family members. In addition, it is essential to ensure equitable access to services, allowing everyone to benefit from genetic advances, regardless of their socioeconomic conditions. Genetic counselors must act with empathy and sensitivity, seeking to minimize patients' suffering and offer support at decisive moments, especially in delicate situations, such as prenatal diagnoses.

Future perspectives and research needs in nursing and genetics

Although genetic counseling is highly relevant, it has limitations that highlight the need for advances in nursing practices. One of the main barriers is related to specific scientific knowledge in genetics. Many professionals lack in-depth training, constant updating and specialization in genetic aspects, including scientific foundations, clinical management and interpretation of genetic data. This gap limits immersion in essential information for professional development, impacting the quality of care and reinforcing the need for investment in education and research in the area (BRITO BG e OLIVEIRA SB, 2023).

The literature highlights the importance of developing health policies that promote the integration of genetics into nursing practice. These policies aim to train professionals to act more effectively in genetic counseling, incorporating knowledge about genetics into care, prevention and educational practices, thus expanding the scope and quality of care provided to patients (CUNHA CMSLM, et al., 2020).

Genetic counseling represents a tool with great potential in nursing practice. Through this approach, genetics is applied to the study of a family's hereditary characteristics, to the analysis of the risks of occurrence of genetic pathologies, to the guidance of family planning and to the management of care. In addition, the process includes detailed guidelines related to the diagnosis and monitoring of carrier individuals, promoting more personalized care based on scientific evidence (NUNES MR e CANABARRO ST, 2021).

Additionally, technologies such as genetic and predictive testing play a crucial role in predisposition analysis, enabling more accurate diagnoses and personalized treatments. In this context, genomic technologies, such as the CRISPR gene editing technique, and the use of artificial intelligence, which can be applied to the study, management and analysis of genetic data, also stand out, expanding the possibilities for intervention and improving the understanding of the molecular bases of diseases (LOPES JÚNIOR LC, 2021).

Furthermore, biomarkers can assist in neonatal screening and early diagnosis, and preventive care that includes health guidance and education and risk tracking (SANTOS RA e MARTINS W, 2024). In summary, through the application of these technologies, beneficial impacts such as early diagnosis allow for greater treatment success and more precision in the study of the disease and the effectiveness of treatment measures. For example, in the treatment of breast cancer, according to the authors, new diagnostic technologies have significantly contributed to reducing negative rates, while increasing beneficial impacts in the health sector. These benefits include increased success rates of treatments, early diagnosis of genetic conditions, the effectiveness of preventive measures and the accuracy of personalized treatments, resulting in substantial improvements in clinical outcomes (SILVA GO, et al., 2024).

FINAL CONSIDERATIONS

Genetics is the science that studies genes and heredity, investigating the processes by which genes are transmitted to future generations. During reproduction, genetic material is compacted and propagated, enabling the transmission of pathogenic genes. Consequently, some families are more predisposed to genetic diseases. Furthermore, genetic errors in haploid cells can result in disorders classified as syndromes. This study concluded that these pathologies impact individuals and their families both emotionally and physically. In this context, early diagnosis, understanding of hereditary patterns and reproductive planning, mediated by genetic counseling, demonstrate significant benefits, especially in the identification and prevention of diseases such as cancer. Nurses, as trained professionals, play an essential role in the management, diagnosis and prevention of genetic disorders, using genetic counseling as an indispensable tool to optimize health care and promote quality of life.

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