GENETIC COUNSELING IN NURSES' CARE PRACTICES

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Genetic Counseling In Nurse Care Practices
O aconselhamento genético nas práticas assistenciais do enfermeiro

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Abstract

DNA is an organic compound that contains genetic information in order to coordinate the development, functioning and transmission of the hereditary characteristics of every living being. By studying it, major discoveries are being made, such as the association of specific genes with the origin of many diseases that afflict humanity, thus making it possible to treat or prevent these pathologies, thus having a significant impact on the health area, creating new demands for knowledge from the professionals who work in it. Among them are nurses, who are including this knowledge in their care practices all over the world, both in academic teaching and in research and patient education. This research, which is a quantitative/qualitative, descriptive and exploratory cross-sectional study, aimed to analyze the knowledge of Primary Health Care nurses regarding genetic counseling. To this end, a questionnaire was created by the researcher containing open and closed questions. Descriptive statistics were used to analyze the objective questions, using graphs generated by the *Google Forms* platform. The analysis of the discursive questions was organized according to Bardin, following the three stages of discourse analysis. Through the study, it was possible to observe that nurses have a basic knowledge of the subject, that they have difficulties in approaching genetics in their nursing consultations and do not carry out genetic counseling during their care practices, and the main factor would be the lack of an instrument that facilitates the consultation focused on genetics.

Keywords: Genetic Counseling; Nursing; Genetics.

Resumo

O DNA é um composto orgânico que contém informações genéticas no intuito de coordenar o desenvolvimento, funcionamento e transmissão das características hereditárias de cada ser vivo. Ao estudá-lo, grandes descobertas estão sendo realizadas, tais como a associação de genes específicos com a origem de muitas doenças que afligem a humanidade, permitindo assim, tratar ou prevenir essas patologias, repercutindo então, de forma significativa na área da saúde, criando novas demandas de conhecimento aos profissionais que nela atuam. Dentre eles, destacam-se os enfermeiros, que estão incluindo esses conhecimentos em suas práticas assistenciais em todo o mundo, tanto no ensino acadêmico, quanto na pesquisa e na educação dos pacientes. Esta pesquisa, que se trata de um estudo quantitativo/qualitativo, descritivo e exploratório de caráter transversal, teve como objetivo analisar o conhecimento dos enfermeiros da Atenção Primária à Saúde referente ao aconselhamento genético. Para tal, foi aplicado um questionário criado pelo pesquisador contendo questões abertas e fechadas. Para a análise das questões objetivas foi realizada uma estatística descritiva, utilizando-se os gráficos gerados pela plataforma *Google Forms*. Já a análise das questões discursivas foi organizada segundo Bardin, seguindo as três etapas de análise dos discursos. Através do estudo foi possível observar que os enfermeiros possuem um

conhecimento básico a respeito do tema, que apresentam dificuldades para abordar a genética em suas consultas de enfermagem e não realizam o aconselhamento genético durante as suas práticas assistenciais e, o principal fator seria a falta de um instrumento que facilite a consulta voltada a genética.
Palavras-chaves: Aconselhamento Genético; Enfermagem; Genética.

INTRODUCTION

DNA (*Deoxyribonucleic Acid*), also known as the genome, is an organic compound that contains genetic information in order to coordinate the development, functioning and transmission of the hereditary characteristics of each living being. It is made up of 46 chromosomes (23 from the father and 23 from the mother), together they store and pass on all the genetic information of the human being (VIEIRA, et al., 2006).

By studying DNA, great discoveries are being made, such as the association of specific genes with the origin of many diseases that afflict humanity, thus making it possible to treat or prevent these pathologies. A pioneering study began in 1990 with the aim of mapping human genes and determining the sequence of nucleotides that make them up - the "Human Genome Project". The initiative originated in the United States and involved the collaboration of several countries, including Brazil. Its proposal also included the creation of a database with all the genetic information found, facilitating access for other members of the scientific community (OFFICE OF SCIENCE, 2016).

Research has brought major scientific advances related to a new concept in the origin, diagnosis and treatment of diseases, which has had significant repercussions in the health area, creating new demands for knowledge from professionals in the field (JENKINS and LEA, 2005).

These genetic and technological advances have created a new demand for health professionals, considering the relationship between genetic aspects and the increased risk of certain pathologies. Among these professionals, nurses stand out, as they are including this knowledge in their care practices all over the world, both in academic teaching and in research and patient education (FLORIDA-SANTOS and NASCIMENTO, 2006).

Although little has been said about nursing in genetics, the brilliant and important work of these professionals in this area is not new and is only expanding. Genetic counseling has been present in nursing literature since the early 1960s, when psychosocial support and case follow-up were emphasized as nurses' responsibilities (FLORIA-SANTOS and NASCIMENTO, 2006).

The term genetic counseling (GC) was coined by the American physician Sheldon Reed in 1947, outlining a new and broad field of service for geneticists. GA can be understood as the communication process that deals with the human problems associated with genetic diseases in a family, involving an interdisciplinary team in an attempt to help individuals and/or families understand the medical facts, including diagnosis, risk of recurrence, prognosis and available treatments (BERTOLLO, 2013).

Nurses have been working for over forty years as counselors and educators in the field of genetics, helping to broaden professionals' knowledge of the health-disease process in relation to biological and psychosocial aspects, generating a transformation in the way the population is cared for. Genetic counseling became part of the systematized language of nursing when it was added to the *Nursing* Interventions Classification (NIC). According to the general attributions of nurses, their education and training, it can be concluded that they can also work in the field of genetics. For this, it is recommended that there is an institutional protocol describing the activities that fall within the competence of this professional (COREN-SP, 2011; FLORIA-SANTOS and NASCIMENTO, 2006).

Nursing, as the science of care, is an area of paramount importance for the practice of genetic counseling and seeks to offer a quality service taking into account ethical, social and cultural issues, free from illegal conduct such as malpractice, recklessness and negligence. The nurse will be able to contribute to all issues pertaining to possible genetic alterations, as well as drawing up research projects that support the improvement of the population's quality of life. It is worth emphasizing that nurses will also assist patients and their families at all stages of the health-disease process (COREN-SP, 2011).

METHODS

This is a quantitative/qualitative, descriptive and exploratory cross-sectional study.

A questionnaire created by the researcher was used, developed using the *Google Forms* platform, containing open and close-ended questions. Participants were only given access to the questionnaire if they accepted the informed consent form (ICF), which was shown immediately after the study was presented (https://forms.gle/5XUka3ejSPXteeS29).

Participants were instructed on the correct way to fill in the questionnaire, highlighting that it could be completed wherever the respondent felt most comfortable, requiring only a device with internet access, which could be a computer, laptop, tablet or even a cell phone.

An instrument was developed which, after validation, will be made available to the Primary Health Care units in the municipality where the research was concentrated, in order to encourage and facilitate nursing consultations focused on genetics/genetic counseling (Appendix A).

Inclusion and exclusion criteria

The study population consisted of nurses working in Primary Health Care.

The inclusion criteria were: being a nurse for more than six (6) months, working in Primary Health Care and accepting the terms presented in the ICF. Exclusion criteria: Those on leave or on vacation were excluded.

Research tool

There are various instruments to help to carry out a research. The questionnaire was the option chosen to carry out this investigation. To clarify the choice, we defined the instrument "questionnaire" within the perimeters presented by Amaro et al (2005): "A questionnaire is a research instrument that aims to collect information based, generally, on the inquisition of a representative group of the population under study. To this end, a series of questions are asked covering a topic of interest to the researchers, with no direct interaction between them and the respondents" (AMARO et al., 2005, p.5).

The importance of questionnaires is also due to the ease with which a large number of people can be questioned in a relatively short space of time.

The questionnaire developed by the researcher contains ten questions, both discursive and multiple-choice, covering knowledge, performance and the importance of genetic counseling in the view of the respondents.

Data collection

Data were collected using a questionnaire created by the researcher, developed using the *Google Forms* platform. Participants were given instructions on how to fill it in and could take as long as they felt necessary to answer it, within the time limit.

The questionnaire is made up of questions related to the knowledge or lack thereof regarding genetic counseling in nurses' care practices. The contact details of the researcher and his supervisor were made available through the ICF, so that the participants would be able to answer any questions that might arise during the course of the research.

Data analysis

Descriptive statistics were used to analyze the closed-ended questions, using graphs generated by the *Google Forms* platform. The data was entered into Microsoft Excel 2016 spreadsheets to calculate the nurses' knowledge of genetic counseling in their care practice. The analysis of the open-ended questions was organized according to Bardin, following the three stages of discourse analysis: 1st pre-analysis; 2nd analytical description; 3rd inferential interpretation.

It is worth noting that the open questions were transcribed in order to keep the answers reliable.

Ethics committee

In accordance with the precepts established by Resolution 466/2012, which regulates research with human beings, this research was approved by the Research Ethics Committee of the Faculdade Dinâmica do Vale do Piranga (FADIP), CAAE 45299821.5.0000.8063.

RESULTS AND DISCUSSION

According to Wanda Horta (1979), nursing is the science and art of assisting human beings (individuals, families and communities) in meeting their needs. In other words, nurses are taking on an increasingly decisive and proactive role in identifying the care needs of the population, as well as in promoting and protecting the health of individuals in their different dimensions, and it is important to act with humanity, cordiality and a clinical eye in order to reach this patient in all aspects.

The incorporation of the concept of genetics into health practices is part of a new reality, exerting a major influence on care. Genetic information covers data obtained from gene sequences and cytogenetic analysis, as well as the study of hereditary characteristics. Genomics, in turn, covers individual genetic predispositions and their interactions with triggering environmental factors, with a view to personalized and more effective prophylaxis. This area of knowledge is fundamental in nursing practice, since most diseases originally have a genomic component (JENKINS, 2008).

It is very common for nurses to come across situations that require knowledge in the field of genetics, especially in Primary Health Care. The nurse, as a genetic counselor, will investigate the patient's family history, drawing up a heredogram that includes first, second and third degree relatives, whether or not they are affected by a disease or have already died. From this point on, they will draw up a care plan geared towards the individual's characteristics, and may use pre- and post-genetic test counseling, working together with a multidisciplinary team and complying with institutional flows and standards. These nursing consultations can also ease the suffering of those with genetic pathologies, as well as their parents (GUIMARÃES E COELHO, 2010).

It is essential that nurses train themselves to offer a quality service when dealing with genetic counseling, providing increasingly holistic and personalized care for each patient.

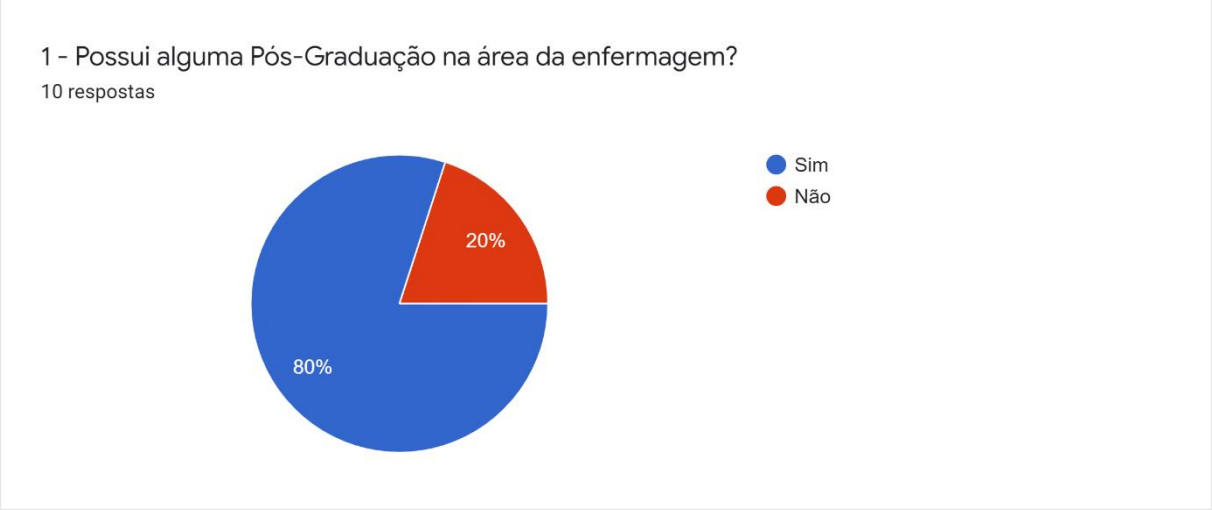
The training of trained nurses and specialists in the field of genetics and genomics is becoming increasingly important in the world, since the ten leading causes of mortality have a genetic component, the integration of this knowledge is of fundamental importance for the era of precision medicine, in order to provide the best health care to patients (CHAIR et al., 2019).

The research was carried out in a town in the Piranga Valley region, in the Zona da Mata region of Minas Gerais, which has 13 units in its Primary Health Care network, where the study was concentrated. The questionnaire was presented and sent to the 13 working nurses, 10 of whom answered it.

The results of the survey will be described in full below, according to the data obtained after organization and descriptive statistical analysis of the responses of the 10 participants.

Figure 1 illustrates the results of the organization of responses in relation to the variable "Do you have a postgraduate degree in nursing?", where it can be seen that the majority of respondents (80%) have a postgraduate degree.

Figure 1. Distribution of responses regarding whether or not they have a postgraduate degree in nursing.



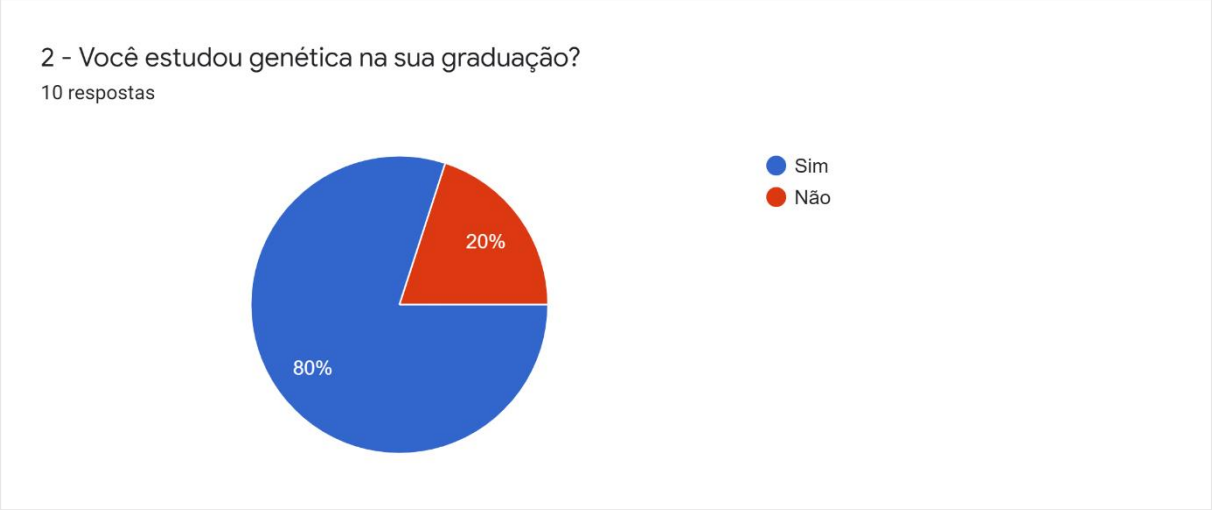
Source: Survey data (2022)

Faced with an increasingly competitive job market, health services are looking for nurses with the highest level of technical and scientific knowledge to work with new technologies and innovations in diagnostic and therapeutic methods. Currently, undergraduate courses offer a generalized view of nursing care, while postgraduate courses provide a vision of specialist nursing in order to meet the need for in-depth study and updating, both in terms of achieving professional autonomy and better integration into the highly sophisticated and technological job market (BURGUESE, 2019).

According to the Faculty of Medical Sciences of Santa Casa de São Paulo (2021), the main objective of postgraduate studies is to deepen knowledge, providing nurses with specific learning, i.e. making them specialists in a particular area, as well as helping them to provide better patient care through training, bringing new knowledge about the health area, promoting updates on new technologies, training students to promote more humanized care and preparing postgraduates to deal with new diseases and protocols.

Figure 2 shows the percentage of respondents who had or had not studied genetics during their undergraduate studies. It can be seen that 80% studied the precepts in their subjects.

Figure 2. Distribution of responses regarding having or not having studied genetics during undergraduate studies.



Source: Survey data (2022)

In 1962, the first article citing the importance of including genetics content in the nursing curriculum was published and, during the 1960s and 1970s, genetics was conceptualized as an important component of nursing practice, especially in the areas of community health and maternal-child nursing (FEETHAM et al., 2004).

In the 20th century, curricula for undergraduate nurses had to be adjusted as emerging technologies, such as the electrocardiogram, antibiotic therapy and dialysis, changed the daily life of nursing (LEA et al., 2011).

Currently, one of the main challenges for the training of nursing professionals is the integration of genetics and genomics into health care. Given the advances made by these sciences, nurses need to update their knowledge in these areas and incorporate it into their care (CALZONE et al., 2010; HSIAO et al., 2012; THOMPSON; BROOKS, 2011).

In Brazil, both students and teachers in nursing courses still have a very conservative view of genetics, probably due to the content of the programs, which are not very informative about the duties of nurses, without specific guidance for nursing practice in genetics and based on the biomedical model. Therefore, there is an urgent need for pedagogical and clinical transformation (FLORIDA-SANTOS, 2004).

Teachers working with undergraduate nurses need to start a debate to include, in the training of these professionals, in addition to the new genomic technologies, aspects related to their implications for individual and collective care (LEA et al., 2011).

Question 3 - If not, were the basic principles covered in the subjects?

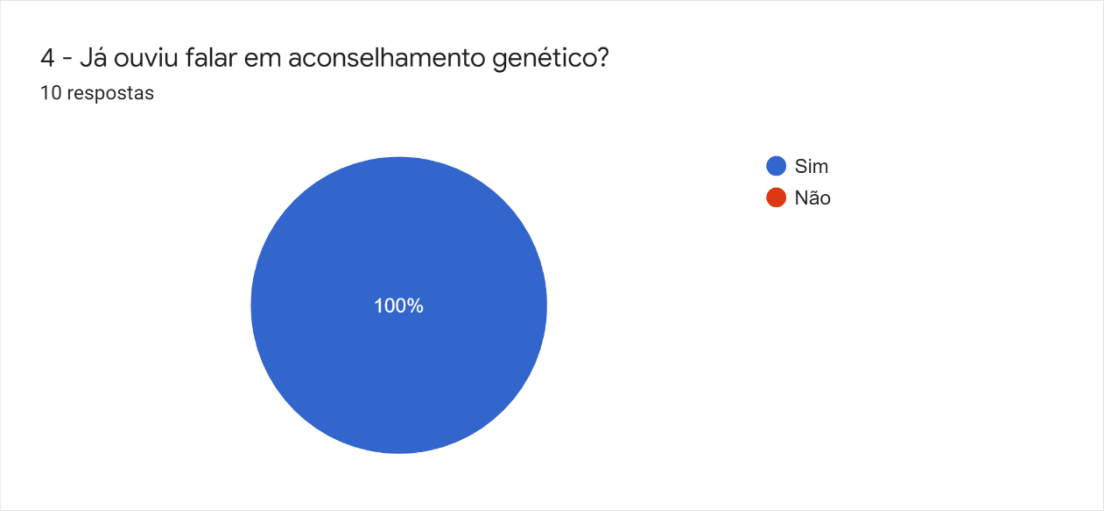
Question 3 asked whether the basic precepts of genetics had been covered by the nurses interviewed during their undergraduate studies. Although the majority (85.7%) had some prior knowledge, 14.3% had not even had contact with genetics during their academic life.

Although nursing as a profession already recognizes the importance of genetics and genomics in clinical practice, surveys carried out in various countries show that this content is still limited in undergraduate nursing courses (JENKINS and LEA, 2005).

The teaching of genetics and genomics needs to be consolidated by articulating the principles of basic genetics with the contents of human genetics and genomics, integrated with clinical practice. To this end, it is necessary to reformulate the teaching-learning process, which is currently deficient, as it is notoriously difficult for most students to retain knowledge of subjects taught at the beginning of the undergraduate course, which remains necessary in the professionalizing cycle and in practice (SCHANAIDER, 2002).

Figure 3 shows whether the nurses had any prior knowledge of what genetic counseling is, where 100% said they knew about it.

Figure 3: Distribution of answers to the question "Have you ever heard of genetic counseling?



Source: Survey data (2022)

Genetic counseling consists of monitoring the individual and/or their family by a trained professional, with the aim of providing care related to the prevention, diagnosis, prognosis and treatment of genetic-related diseases. This service aims to guide patients through all the aspects involved with the problem in question, using a script that varies according to institutional protocols (COREN-SP, 2011).

It is clear that the topic is not new to the respondents, and that health care, using knowledge of genetics, has been applied to the population for around 60 years and since then, nursing has been a part of it. In some countries, such as the United States, Canada, England and Japan, the role of nurses in genetic consultations, as part of a multidisciplinary team, is already well defined. This professional offers quality services based on protocols that take ethical, legal and social issues into account (AMERICAN NURSES ASSOCIATION, 2007).

Question 5 - In your opinion, genetic counseling is:

In question 5, an open-ended question, nurses were asked for their opinion on what genetic counseling is about. The following are the words of the nursing professionals, collected through the survey instrument.

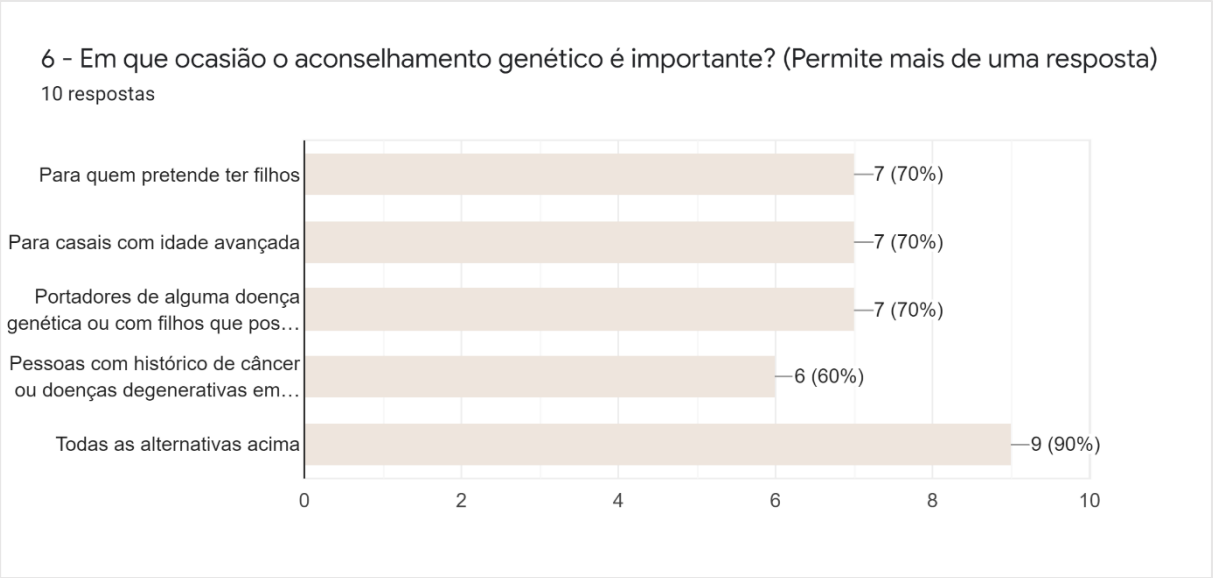
> ENF1: *"Very important to avoid various diseases and malformations";*
> ENF2: *"Genetic counseling consists of checking the probability of a genetic disease occurring in a family. With this probability, it is possible to guide couples who are thinking of having children."*
> ENF3: *"It's a health education process that aims to guide people about the likelihood of events associated with heredity";*
> ENF4: *"It is work focused on the human problems associated with the occurrence or risk of a person having a hereditary disease in a family."*
> ENF5: *"Offer guidance on genetic diseases, hereditary probabilities and changes related to the genetic inheritance brought by children."*
> ENF6: *"Advising the patient of the possibility of a child being born with a certain genetic disease, when there is a case in the family."*
> ENF7: *"Study to detect possibilities of genetic diseases that may run in the family";*
> ENF8: *"Important to carry out in primary care";*

From the statements highlighted above, it can be seen that the nurses taking part in the research are aware of the topic and know the importance of carrying out health education focused on genetics, carrying out genetic counseling in their nursing consultations and promoting actions aimed at the health of the population.

ENF9: *"Assessment of the possibility of the person having a child with a certain disease. And after this assessment... give the person the opportunity to have the child or not".* It's important to note that genetic counselors act as educators and provide all the data regarding certain pathologies, available treatments, expected traits and conditions, how the pathology will progress, but they can never decide for future parents whether or not they will go ahead with the pregnancy, or wish to become pregnant.

Question 6 measured whether respondents were aware of the importance of genetic counseling and in which cases it is indicated.

Figure 4 - Distribution of answers to the question "When is genetic counseling important?



Source: Survey data (2022)

Genetic counseling is an important tool in the field of hereditary diseases, as it addresses educational and reproductive aspects that are essential for improving the quality of life of patients with certain genetic pathologies. It is indicated for different clinical scenarios, such as: consanguineous couples, family history of genetic diseases, habitual or repeated pregnancy loss, infertility, suspected hereditary cancer (GUIMARÃES and COELHO, 2010).

Question 7 - In your opinion, which professionals can provide genetic counseling?

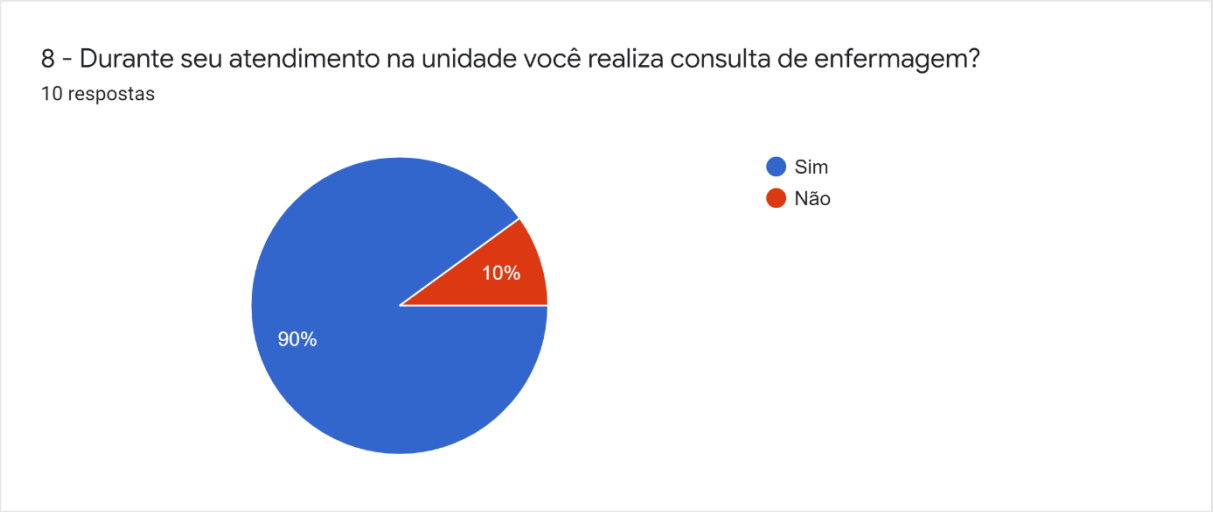
In question 7, an open-ended question, nurses were asked which professionals can carry out genetic counseling, where 90% of respondents included nurses as counselors, in addition to other professionals.

Oliveira and others (2017) point out that nurses can carry out genetic counseling through their patient's family history, through data collection, diagnosis, planning and evaluation, but

many nurses are unaware that they are supported by their council and that they should provide care and health education focused on genetics during their consultations, even if they don't have a specialization. This lack of knowledge can be seen in the following statement: *"I was unaware that a professional other than a doctor could provide genetic counseling"* (ENF1).

After discussing which professionals can carry out genetic counseling, the nurses were asked, in question 8, if they carry out nursing consultations in the unit where they work. It was noted that there are still professionals who don't carry out nursing consultations.

Figure 5 - Distribution of responses regarding the care provided in the unit, and whether the nurse provides nursing consultations?"



Source: Survey data (2022)

According to the Ministry of Health's Ordinance No. 1.625 of July 10, 2007, it is the specific responsibility of nurses in Family Health Teams (NFHs) to carry out nursing consultations. Nursing consultations are a private activity provided by nurses, as can be seen in Resolution 606/2019, in which health problems are identified and nursing measures are prescribed and implemented with the aim of promoting, protecting, recovering or rehabilitating the patient (CAIXETA, 2009).

Through the nursing consultation, the nurse can identify and solve the individual's health problems, as well as indicate measures and care in the face of the interaction between the professional and the patient, in addition to being a liberal activity, of a scientific nature and exclusive to nurses. This professional must bear in mind that they cannot impose their

own reality on others. Dialogue must encourage respect for the thoughts and attitudes of the person being assisted, since the active participation of the patient during the consultation is fundamental (GOMES et al., 2012).

In order to improve care for the population and the autonomy of the professional when carrying out the nursing consultation, since, according to resolutions, only the nurse can carry out such care, it is essential that all Primary Health Care nurses carry out this action.

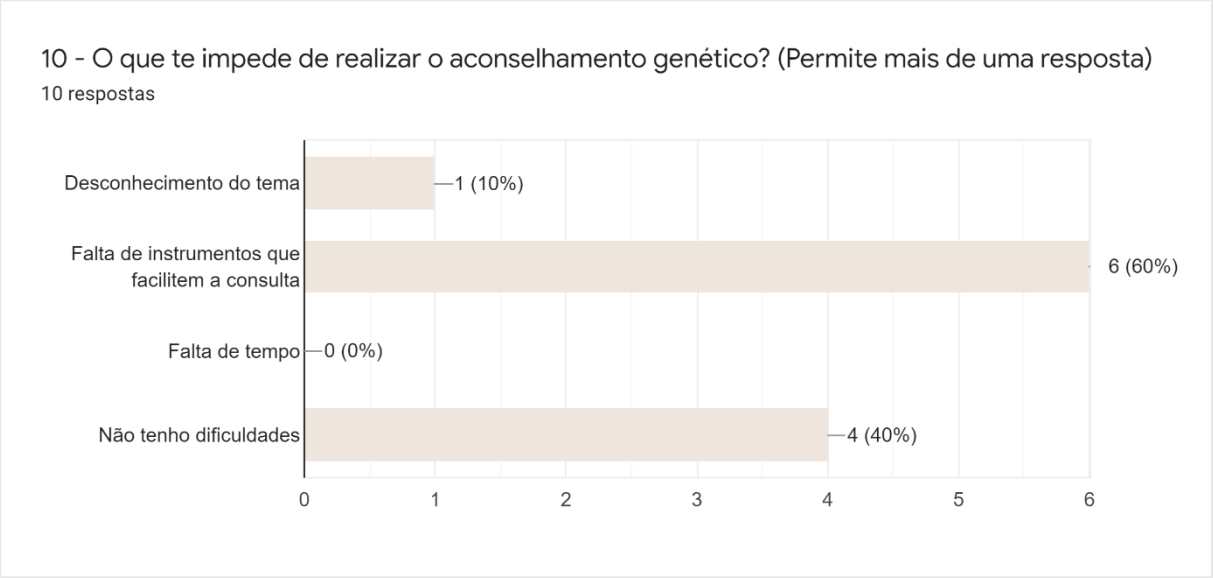
Although nursing consultations are considered important, nurses still face difficulties in carrying them out. The fact that they don't always have a suitable place, or even an office just for consultations, is one of the most common obstacles.

Question 9 - During the anamnesis, you ask questions related to: (allows more than one answer)

Question 9, a close-ended question, asked whether during the anamnesis the nurse asks about: the presence of hereditary diseases (100%), problems that arise in the prenatal or postnatal period (100%), a history of mental retardation (70%), a history of abortion and consanguineous relationships (90%), or doesn't ask any questions about the subject (10%).

Question 10 illustrates the results of the organization of answers in relation to the variable "What prevents you from carrying out genetic counselling?", where more than half (60%) said that it was due to the lack of a tool to facilitate nursing consultations focused on genetics.

Figure 6. Organization of the answers to the question "What prevents you from undergoing genetic counseling?"



Source: Survey data (2022)

The nursing consultation is an activity of great importance and resoluteness when carried out properly by the nurse. In addition to knowledge and constant training, it requires adequate and complete instruments (COSTA, BUSTAMANTEL and SILVA, 2013).

In 2003, nurses who attended the ANA's annual convention were interviewed and stated that they do not feel qualified to provide care that involves knowledge of genetics, but all agreed that genomics is profoundly transforming nursing practice (JENKINS and LEA, 2005).

FINAL CONSIDERATIONS

Genetic Counselling is an essential practice to help patients and counsellors understand the deleterious genetic conditions in which they are involved, humanizing the process that the person and their family go through from the suspicion and diagnostic confirmation of a genetic anomaly to the prognosis.

The offer of GA allows not only the nature of the risks to be determined, but also the information available, treatments, consequences of each option, psychological costs, among

others, as it addresses educational and reproductive aspects that are indispensable for improving the quality of life of patients with certain genetic pathologies.

Through the study, it was possible to observe that nurses have a basic knowledge of the subject, that they have difficulties in approaching genetics in their nursing consultations and do not carry out genetic counseling during their care practices, and the main factor would be the lack of an instrument that facilitates the consultation focused on genetics, becoming a very important loss for Primary Health Care, since the growing demand of people who lack genetic counseling services serves as a warning of the need for improvements in this area, within the current scope of the SUS.

As the professional who has the most direct and indirect contact with the patient, improving nurses' knowledge of genetics and genomics can be an excellent strategy for improving prophylaxis, diagnosis and treatment, helping to reduce morbidity and mortality rates linked to genetic diseases.

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