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Genetic counselling in preconception and prenatal care provided by midwives, an international review

Summary

Promote the change from genetic counselling carried out in preconception and prenatal check-ups, to genetic counselling provided by midwives themselves and at the same level of care with an open perspective to multiculturalism and plurality. This would mean improving primary care for women and their families, with a cost-effective activity practised by midwives. However, considering the experience of other countries where midwives undertake this activity, we believe it is essential that within the midwife's training in sexual and reproductive health, there is a high component of training in genetics in order to be able to carry out this task with the safety and effectiveness required by this change in care. Key words: prevention, preconception, counselling, genetic counselling, midwives.

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Introduction

The Genetic Counselling Society of the USA has defined genetic counselling as the process of helping individuals understand and adjust to the family, psychological and medical implications of a genetic diagnosis of a specific disease.[1] In 1997, Abramski and Chapple in the United Kingdom mentioned that the involvement of midwives in genetic counselling plays a fundamental role in terms of the information given to patients in a preconceptional or prenatal state.[2] In Chile, the Ministry of Health (MINSAL) defines preconception and prenatal screening as basic actions for gestational control, including the detection of congenital malformations and/or chromosomal alterations.[3]

Carrol et al. in 2009 stated that it is essential to provide genetic education in primary care, respecting the beliefs, expectations and culture of the population. However, in our country this task is not yet a reality, since genetics education programmes for health careers are far from reaching the proposed standards.[4]

The overall objective of this review is to analyse the aspects of genetic counselling in preconception and prenatal care that should be taught by midwives internationally during the period 2010-2020.

Methodology

A literature review was carried out using primary research sources to collect, analyse and synthesise evidence on the topic of genetic counselling in preconception and antenatal care provided by midwives internationally between 2010 and 2020, using six electronic databases: EBSCO, ELSEVIER, Google Scholar, PUBMED, Taylor & Francis and Wiley. The selection was limited to documents dealing with genetic counselling in preconception and prenatal consultations provided by midwives, also included were articles defining genetic counselling and the work of midwives in genetics, in addition to other sources of information such as guides, regulations and clinical care manuals pertaining to Chile. The search was carried out in English, French, Italian and Spanish, and all those that presented a different language were discarded.

The thesaurus descriptors used for the bibliographic search are: "Genetic counseling", "Midwives of midwifes", "Genetic counselor", "Genetic", "Perinatal", "Prenatal", "Preconception" and its translation in Spanish: "Asesoramiento genético", "Matronas o parteras", "Consejero genético", "Genética", "Perinatal", "Prenatal", "Preconcepcional", which for the purposes of this research are consistent with the keywords. Boolean operators used were "AND/OR", time and language filters were applied. The documents were then selected by title and summary of interest for this research. To conclude, a reading and analysis of the usefulness of the different documents selected was made.

Results

Genetic counselling in preconception and prenatal check-UPS.

The study concluded that genetic counsellors are more likely to perform invasive genetic testing during their own pregnancies without medical indication. The reasons for this could be on the one hand that they are affected by the frequency with which they see such genetic disorders, which makes them more susceptible to believe that something bad will happen with their pregnancies. On the other hand, the information that they themselves know and give about it, as well as the greater accessibility they have to these diagnoses because they work in this area.[5]

At the Neurosciences Centre, Department of Reproductive Sciences and Dentistry located in Naples, Italy, a retrospective study of genetic counselling in couples was conducted from 1993 to 2013, from a total of 1269 prenatal diagnoses performed in that period of time. The results showed that the prenatal diagnostic tests used were amniocentesis in 14.9% and chorionic villus sampling in 85.1% of cases. The genetic diagnostic diseases found were: 41.6% Betatalasemia, 15.1% cystic fibrosis and in 43.3% of cases other rare genetic disorders.[6]

At the S. Orsola-Malpighi Hospital, Bologna, Italy, a study was conducted in 2015 to assess the attitude of pregnant women of

advanced maternal age in relation to the detection of chromosomal abnormalities by invasive prenatal diagnosis and the impact of genetic counselling. It was concluded that 52.6% of the women preferred individual genetic counselling, however, this was also associated with a perceived higher risk of chromosomal abnormalities. On the other hand, group genetic counselling sessions had a better impact in terms of clarification of doubts and better understanding of the topic.[7]

Farrel's 2015 study assessed how healthcare professionals counselled women about genetic pathology testing during the first trimester in antenatal consultations; the results of this research highlighted the following themes: 1. It is important to identify levels of knowledge and practice in the application of genetic pathology testing. 2. There is a need to know the educational needs of pregnant women. 3. Women's preferences about decision making.[8]

A London study evaluating the components of genetic counsellor education concluded that genetic counsellors are not fully prepared to provide genetic counselling due to a lack of clinical experience based on master's programmes in 8 countries, including Australia, France, the Philippines, Cuba, Taiwan, Israel, Saudi Arabia and South Africa. Although some of the programmes combined formal education with more clinical capacity, the form and duration of each of the clinical placements in the different programmes is unknown.[9]

In France, since 2006, it has been established by decree that multidisciplinary prenatal diagnosis teams must have a genetic counsellor, who plays an important role in the care provided during preconception and prenatal care. The role of the genetic counsellor is to collect gestational data such as: maternal age, pregnancy, parity, gestational age in weeks, date when the foetal anomaly was discovered, additional tests and previous obstetric consultations.[10]

A descriptive study conducted at Yamaguchi University Hospital, Japan, interviewed women who had an older gestation and asked them about their experience with the genetic information provided during their pregnancies. The researchers concluded that a misperception of risk can lead women to seek more invasive interventions than required, as they have an incorrect perception of their own risks. The patient has the right to receive clear information in order to be aware of these risks. Doctors, midwives and nurses should provide more appropriate and effective information in a sensitive way.[11]

A study carried out in London by the European Association of Human Genetics and the EUROGENTEST network during the year 2020 reached the consensus that the key to improving healthcare provision related to the application of genetic testing in Europe is directly related to the education provided to healthcare professionals, which must be adequate and up to date.[12]

In the UK, specifically at the Fetal Medicine and NHS Foundation Health Centre for Women and Children in Birmingham, the evolution of prenatal genetics was studied. The results show that there has been a change in the trends of the most commonly used genetic tests for prenatal diagnosis, which are QF-PCR, CMA and Mendelian testing. In the years 2008-2011 the predominant test for diagnosis was QF-PCR/karyotyping (53%), CMA (8%), Mendelian (38%), on the other hand in the period of 2011-2014 began to predominate the use of Mendelian tests (43%) but still the other two tests were almost in the same percentage QF-PCR (25%) and CMA (31%) and finally in the period of 2014- 2019 Mendelian tests continue to predominate (53%) and the other two tests are used in lower proportion QF-PCR (22%) and CMA (24%).[13]

Contributions of genetic counselling to public health

Strategic role of nurses in genetic literacy, providing educational tools in the context of public health. In Japan, genetic information requires the development of a code of ethics to generate public awareness and thus ethical use. Genetic literacy stems from the term scientific literacy and is defined as "the ability to use scientific thinking for personal and social purposes in relation to genomics and genetic diseases", with the aim of providing genetic education through the school system. Nursing professionals have the potential to help patients gain knowledge about genetics, but in practice they often do not do so. Practitioners have noted that genetic aspects of health care counselling should be covered in addition to environmental and socio-economic factors, providing a clearer and more reasonable explanation of disease risks.[14]

In Chile, genetic counselling is not recognised as an independent clinical discipline and is only provided by physicians specialising in genetics. Of the 28 clinical geneticists in Chile, 86.7% specialise in dysmorphology and 67.7% in prenatal diagnosis, among other specialties. As in most Latin American countries, Chile also lacks formal postgraduate programmes in genetic counselling as an independent clinical discipline. There is only one fellowship programme in clinical genetics for physicians since 2004, which remains in place to date. According to the Royal College of Physicians of the United Kingdom, it is recommended that the ratio of clinical geneticists to population should be 0.75 per 250,000 people. In Chile this ratio is not met, as there should be 49.4 specialists and so far there are only 32% of what is recommended. There should be a multidisciplinary team for the delivery of genetic counselling, but doctors consider this to be a barrier rather than an opportunity, since, by involving other professionals, patients may be confused by the information given by different people, contradicting what is said by the medical geneticist. There are a variety of barriers to access to genetic counselling and education, from the small number of professionals specialising in this area, to the financial cost of having counselling for the public health population. There is a need to increase genetics education and thus motivate more professionals to specialise in order to provide better health care from the beginning of development.[15]

Discussion

In some countries such as France, the genetic counsellor plays an important role in supporting the informed consent process, which involves a thorough discussion of the benefits and risks of both fetal and maternal genetic testing.[10] In Italy, on the other hand, the genetic counsellor provides timely advice to families before and after genetic testing, informing them of the possible pathologies that may be found, clinical features, genetic test results, possible treatments and therapies for the genetic alteration found, and finally informing and educating them about the prevalence of concomitant conditions.6

In the USA, the midwife has a strategic role in the delivery of genetic counselling due to her full involvement in the reproductive process and delivery care, however, the discussion on the delivery of genetic counselling focuses on midwives' knowledge and confidence about genetic issues. Results from a 2012 survey of US midwives indicate that while midwives consider genetic counselling to be fundamental to prenatal care, they perceive that they do not have the necessary skills to provide comprehensive genetic counselling.[16] Following Crane's theoretical line, Farrel in 2015 illustrates the need for practitioner-led strategies in counselling for informed decisions about genetic testing that can impact on prenatal care.[8]

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In this way we can clearly see how North Americans and Europeans are including the role of genetic counselling within preconception and prenatal care according to their needs, in turn, envisioning the importance of midwives, nurse-midwives and midwives in performing this as a fundamental component of professional care.

In the UK in mid-2013, the idea was put forward that the provision of genetic information should be based on bioethical principles and that those who fulfil the role of genetic information provider, in this case the counsellor, should be registered on the national register of genetic counsellors and have certification of their knowledge to ensure safe, effective and correct access to the basic principles of genetics.[17]

Recently in Japan, Kawasaki et al.[14] proposed the need to spread "genetic literacy" as an effective measure to help resolve the ethical use of genetic information. In general, the public is finding more and more genetic information available, but it is often not put to proper use, so instead of being a benefit, it becomes a disadvantage as it increases public concerns about not having the necessary tools to discriminate between what is useful and what is not.[14]

Both measures described by researchers in Japan and the UK focus on the use of genetic information in an ethical and welfare-enhancing way, either through education of the general population or through a licensed counsellor.

In 2010, Skirton from the UK mentions in his article the competencies of nurses and midwives in basic genetics. They discuss the importance of confidence as well as the knowledge and skills that these professionals must have under the legal framework required in their country. However, at the end of the research, it was discovered that midwives still do not fully achieve the competencies in genetics according to UK standards, suggesting a lack in the performance of activities that require genetic knowledge for their development, such as prenatal and preconception checks.[18]

The various proposals in improving the foci of genetic knowledge, confidence and skills of midwives with genetics may help with the premise that health professionals should empower pregnant women to be aware of their risks and encourage them to make informed decisions according to their potential genetic risk, a premise proposed by Murakami in 2016.[19]

Conclusion

Genetic counselling improves the expectations of clients of advanced maternal age in possible pregnancies, whether, genetically, biologically, psychologically and socially; Communication is the key to reach clients and their families by providing appropriate genetic counselling.

a. Improving the perception of risk in possible or actual pregnancies in cases of risk factors or abnormal prenatal outcomes.

b. Significant improvement is sought in the genetics education provided to students during undergraduate studies and implementation of postgraduate programmes for those seeking to specialise.

c. The midwife has a strategic role in the delivery of genetic counselling, as they are the professional who provides the gateway to the health system for the vast majority of women.

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Conflicts of interest

The authors declare that they have no conflicts of interest.

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