JOURNAL OF HEALTHCARE SCIENCES
Volume 2 Issue 12 2022, Article ID: JOHS2022000593
http://dx.doi.org/10.52533/JOHS.2022.21216
e-ISSN: 1658-8967

Review

Psychosocial Effect and Role of Genetic Counselling in Primary Health Care

Shada Baoum [1*], Asmahan Balobaid [1], Ayman Aljedany [1], Maha Awlia [2], Abdulaziz Alqahtani [3], Zahra AlGhanim [4], Saad ALJandan [5], Mohammed Alkhidhr [6], Ali Alfehid [7], Abdullah Al jabal [8], Walaa Alabadi [9]

[1] *Primary Healthcare, King Fahad General Hospital, Jeddah, Saudi Arabia*
[1] *Primary Health Care, King Abdullah Medical Complex, Jeddah, Saudi Arabia*
[2] *Primary Health Care, Makkah Health Cluster, Makkah, Saudi Arabia*
[3] *Home Care Department, King Fahad Medical City, Riyadh, Saudi Arabia*
[4] *Primary Health Care, First Health Cluster, Qatif, Saudi Arabia*
[5] *NICU, King Faisal General Hospital, Al-Ahsa, Saudi Arabia*
[6] *College of Medicine, King Saud University, Riyadh, Saudi Arabia*
[7] *College of Medicine, Medical University of Warsaw, Warsaw, Poland*
[8] *Department of Emergency Medicine, Asir Central Hospital, Abha, Saudi Arabia*
[9] *College of Medicine, Vision Colleges, Riyadh, Saudi Arabia*

Correspondence should be addressed to **Shada Baoum**, Primary Health Care, King Fahad General Hospital, Jeddah, Saudi Arabia. Email: baoum.shadaa@gmail.com

Received: 15 December 2022, Accepted: 27 December 2022, Published: 31 December 2022

Abstract

Medical knowledge of both prevalent and rare diseases is changing as a result of recent advanced innovations. In the team that provides genetic services, such as genetic risk assessment, counseling, and continuous medical care for patients with inherited predisposition to disease, primary healthcare plays a crucial role. Primary care physicians can integrate genetic counseling within the larger context of a patient's overall health state since they are accustomed to prioritizing among competing clinical needs. The ongoing relationship that develops between physicians and patients over time is another aspect of primary care that is well-suited for genetic counseling. As patients' needs and emotional states change over time, primary care may provide continual patient education and counseling. When patients ask their primary care physicians about genetic tests and capabilities, they are frequently ill-equipped to respond for which further research along with the training of physicians is needed. Additionally, the majority of patients are also unprepared for the plethora of psychosocial problems that result from genetic testing. The primary care physician is more equipped to incorporate psychosocial factors into any discussion of genetic information since they are more familiar with the patient's broader perspective. When a patient is identified with genetic or familial predisposition to an illness, a number of psychosocial problems may arise. If their risk is revealed, people can be concerned about stigmatization, insurance eligibility, and employment discrimination. The purpose of this research is to review the available information about psychosocial effect and role of genetic counselling in primary health care.

Keywords: *genetic, counseling, primary, care, psychosocial*

Introduction

Although Sheldon Reed first used the term genetic counseling in 1947, the field of genetic counseling is still relatively new compared to medicine and allied health sciences. Although skilled professionals from a variety of fields can engage in genetic counseling, genetic counsellors have received specialized training. The process of assisting people in comprehending and adjusting to the physical, psychological, and familial ramifications of the hereditary contributions to disease is defined as genetic counseling. Interpretation, risk assessment, education, and counseling are all part of the process (1). The mapping of the human genome captured the public's imagination who saw the project as a great expedition; a revolution that would change society forever. The human genome sequencing pledged emerging trends in medicine, innovative treatments for disease, and improved understanding of man's genetic and biological composition. Genetic testing has the capacity to forecast an individual's risk of getting an illness, even while the individual is asymptomatic, for a number of specialized but continuously expanding disorders. Genetic testing has the ability to significantly lower the associated morbidities and mortalities for certain diseases, and in some cases, completely prevent the condition, if adequate measures are adopted to prevent or slow the beginning of the disease. Additional advantages include decreasing hospitalizations and health care costs in addition to easing pain and suffering (2).

Clinicians in primary care play a crucial part in genetic counseling. This function is different from what genetic counsellors typically do. To begin with, primary care physicians frequently participate in the early steps of selecting who may benefit from genetic assessment, assisting patients in determining when it is acceptable to explore genetic information, and preparing them for consultation. Second, the nondirective approach promoted by genetic counseling differs from the counseling and guiding philosophies prevalent in generalist practice. Third, primary care physicians frequently understand the patient's family, which is the setting in which genetic information affects the patient. They may be in a good position to take a family-based strategy for controlling genetic risk because they are likely to care for family members in addition to the identified patient. As diseases progress, family members are newly diagnosed, and patients enter new stages of their life cycles, genetic information such as the family history takes on additional significance and occasionally more urgency in primary care. In the treatment and long-term management of those who are identified to be at risk for genetic illness, patient continuity will be crucial (3).

Individuals display a limited sensitivity to genetic risk information, and it appears that testing perceptions and behavioural reactions are influenced by mindsets of illness risk. Testing motivations are complicated, and efforts are being made to create decision aids. Genetic testing's findings on psychological and behavioural effects show a wide range of results, from minimal or positive effects to negative effects that may be difficult to detect using standard measures of general well-being. Information about genetic risk may affect health behaviour, as per the recent research studies. Genetic testing of children is causing growing concern since the research depicts that it has wide-ranging effects on family dynamics (4). In tertiary care settings, genetics services have traditionally been primarily compartmentalized. Majority of the population face geographic, financial, and psychological access hurdles as a result. Theoretically, allowing patients to obtain genetic services in the context of primary care, which is associated with decreased patient expenses and travel time, would be one of the greatest methods to more effectively and fairly incorporate genetics into healthcare. Family physicians, however, lack faith in their capability to practice genetic medicine. In addition, although while the majority of primary care physicians think that adopting genomic medicine is not their job, they do believe that developments in genetic medicine will enhance patients' health outcomes (5). The purpose of this research is to review the available information about psychosocial effect and role of genetic counselling in primary health care.

Methodology

This study is based on a comprehensive literature search conducted on November 11, 2022, in the Medline and Cochrane databases, utilizing the medical topic headings (MeSH) and a combination of all available related terms, according to the database. To prevent missing any possible research, a manual search for publications was conducted through Google Scholar, using the reference lists of the previously listed papers as a starting point. We looked for valuable information in papers that discussed the information about psychosocial effect and role of genetic counselling in primary health care. There were no restrictions on date, language, participant age, or type of publication.

http://dx.doi.org/10.52533/JOHS.2022.21216

Discussion

Genetic counseling is a communication process that gives patients and families the ability to make independent decisions and utilize new genetic knowledge. The ability to provide genetic counseling and the knowledge of genetic counsellors are essential for the efficient application of genomic medicine (6). Over the past ten years, the revelation of genes linked to increased risk of colorectal, ovarian, and breast cancer has expanded the understanding of cancer susceptibility. The chance of developing cancer can be predicted through genetic testing for abnormalities in a number of genes. The demand for genetic counseling and predictive genetic testing for cancers has significantly increased over the past decade and is expected to continue to rise as the genetics of other common diseases are unrevealed. In genetic counseling, an effort is made to help a person understand how likely it is that they may inherit a disorder as well as how to deal with that possibility without becoming overly anxious. Therefore, for genetic counseling to be regarded beneficial, there must be proof that it enhances a person's perception of their probability of developing the disease and their understanding of the genetics of the condition without having a negative emotional impact (7).

Frequently, the earliest indications of genetic diseases are noticed during routine healthcare examinations. It may thus be difficult for the physician and the patient to decide what to do next. Human genetics is a fast-developing discipline; therefore, many physicians are unsure of which aspects of a patient's medical history call for a trip to the genetics clinic and which ones just need to be observed. Primary care physicians frequently ask clinical geneticists and genetic counsellors questions about a range of genetic conditions. These include discussing test results and other genetic concerns with a patient or family member, aiding with interpreting specialized testing, and the appropriateness of patient and laboratory referrals (8).

Evidence from literature

Primary care physicians are readily available to all patients for any complaint, request, or inquiry, thus having a special position in the healthcare system. To fulfil the demands of their patients that are generated by the swift progress in genomics, primary care professionals must get education. Improved genetics literacy among primary care physicians is necessary to enable their involvement in the discussion of the promises and hypes of genomic medicine and to help them distinguish between practical applications in healthcare that are beneficial and those that are not. If developments in basic and clinical science in the genomics of prevalent chronic diseases in practice and midwifery care are successfully translated, genetics may have an impact on routine primary care practices. However, changes will only be successful if they seamlessly integrate into practice routines (9). Rose et al. described in their study that the patients thought the informational level was appropriate and were happy to get genetic counseling service in primary care. Every session included a discussion of the genetic basis of cancer, and a subsequent multiple-choice question revealed that the genetic information had been retained well. Many individuals who had concerns regarding a family history said they were relieved after the consultation, indicating that the counselling provided was suitable. One benefit of primary care involvement in pedigree evaluation is that the recommendations made can be reiterated at later consultations (10).

It is becoming increasingly understood that an individual's responsiveness is significantly influenced by their emotional responses to risk information. Distinct emotions have different effects on how individuals react to risk information, with fear leading to risk-averse decisions and anger leading to risk-seeking. Therefore, emotional responses to risk information may cause people to decline screening. Fear of learning their risk level and actual possibility of having the disease, as well as animosity toward physicians, may lead people to forego screening and take risks instead. If a person has had prior unfavourable encounters with a certain event or object, negative reactions to risk information are more likely to manifest (11). The primary care physician should be aware of the risks to the ideal pregnancy posed by hereditary and non-genetic hazards. Pregnancy, or the anticipation of pregnancy, serves a variety of psychological purposes. Couples could struggle with the conflict between their feeling of duty and their desire to have, raise, and care for a child. The couple may have emotions upon learning about risks to a desired pregnancy, necessitating careful counseling. To understand the psychological effects of providing genetic counseling and genetic screening in preconception primary care, further research is required. The many counseling techniques that can be used when concentrating on non-genetic and genetic risk factors in primary care should be known to primary care professionals (12).

http://dx.doi.org/10.52533/JOHS.2022.21216

The outcomes of genetic research have a great impact on the development of medicine. The importance of prevention, personal attitudes, and behaviour has increased at the same time, not only in relation to the disease but also in regard to one's own health. This field of study and intervention should be taken into account for these reasons, not only from a medical standpoint but also from an ethical and psychological one. It is important to take a comprehensive approach to this issue, psychological support often thought of as a parallel intervention becomes a part of genetic counseling: the focus of the intervention shifts to the person, rather than just the issue (13). Families with down syndrome babies are recommended to receive genetic counseling, which may help them adapt to the difficulties brought on by this diagnosis. There are numerous methods of genetic counseling used globally, despite the fact that the fundamental counseling objectives are the same. Results of a pilot study showed that following two sessions of genetic counseling in a public healthcare facility over the course of 30 days, 30 mothers of Down syndrome babies younger than 6 months old were questioned. High levels of satisfaction were expressed. In 44% of cases, respondents thought the information they had learned about Down syndrome after just two sessions was technically ambiguous. The majority of mothers (96.7%) said genetic counseling was helpful and offered psychological support. The model was deemed adequate, but more research is required to determine how to increase this population's capacity for knowledge retention (14).

Primary care physicians lack the expertise needed to perform genetic services in an efficient manner. The lack of knowledge is not the only problem since family physicians and other professionals will only recommend genetic testing if it will benefit their patients. How to impart genetic knowledge to primary care physicians in a way that respects their cultural values, expectations, and beliefs is a difficult problem. For health care workers, fundamental competences in genetics have been established by numerous organizations as criteria for genetic education. Although limited, published assessments of genetic education programs have demonstrated an improvement in patient referral decisions for patients with a family history of breast and ovarian cancer as well as an increase in cancer genetic knowledge. It is generally understood that educational interventions, particularly interactive ones, have the power to alter physicians' behaviour (15). Survey assessing the knowledge of primary care physicians regarding the genetic counseling practice revealed that in terms of basic genetic knowledge, physicians said that chromosome abnormalities, xeroderma pigmentosum, and genetic disorders were their most knowledgeable subjects. In terms of genetic counseling, physicians said that they direct the parents of and couples with a risk of having a child affected by a genetic disease to an expert or a genetic counseling center. Only 20.7% of respondents were aware of the ethical principles and practices involved in genetic counseling. Physicians believed they lacked sufficient knowledge of genetics and genetic counseling, and 83.9% said they would like to take a course (16).

Harding et al. stated in their study that primary care physicians agreed that it was their responsibility to collect family histories, evaluate risks, choose the best management plans, and arrange referrals. Primary care physicians considered it part of their job to address patients' concerns as well as inform and counsel patients about the moral, psychological, and medical implications of genetics. Primary care physicians reported a small number of genetic contacts in their practices, although they did not categorically label some procedures, like family history evaluation, as practicing genetic medicine. Rare genetic disorders were less frequently observed; the majority of clinical exposures mentioned by primary care physician were related to pregnancy or inherited malignancies. Primary care physician employed genetic information as a screening tool since they were aware that many health disorders have a hereditary component. Although understanding the genetic basis of a problem was thought to be advantageous, primary care physician felt that more information was needed about the interpretation of genetic test results, clinical value, cost-effectiveness, and communication techniques. Primary care physician in rural and urban areas shared comparable views on the importance of genetics in primary care (17). Primary care being immediate contact has vital role in the genetic counseling. The best approaches to inform patients about the importance of genetic medicine to their health and the role of primary care in providing genetic services both require further study as the available literature is limited. Furthermore, academic informative sessions for genetic counseling among primary care physicians can be helpful in increasing and improving their skills.

Conclusion

Genetic counseling provided by primary care physicians may be more directive since it is the first line of contact with the community although training of primary care physicians for advanced genetic counselling is need of

http://dx.doi.org/10.52533/JOHS.2022.21216

hour to ensure effective practice and prevention. Further research can be beneficial in signifying the importance of role of primary care.

Disclosure

Conflict of interest

There is no conflict of interest

Funding

No funding

Ethical consideration

Non applicable

Data availability

Data that support the findings of this study are embedded within the manuscript.

Author contribution

All authors contributed to conceptualizing, data drafting, collection, and final writing of the manuscript.

References

1. Skirton H, Cordier C, Ingvoldstad C, Taris N, Benjamin C. The role of the genetic counsellor: a systematic review of research evidence. European journal of human genetics : EJHG. 2015;23(4):452-8.

2. Cooke-Hubley S, Maddalena V. Access to genetic testing and genetic counseling in vulnerable populations: the d/Deaf and hard of hearing population. Journal of community genetics. 2011;2(3):117-25.

3. Martin JR, Wilikofsky AS. Genetic counseling in primary care: longitudinal, psychosocial issues in genetic diagnosis and counseling. Primary care. 2004;31(3):509-24, viii-ix.

4. Cameron LD, Muller C. Psychosocial aspects of genetic testing. Current opinion in psychiatry. 2009;22(2):218-23.

5. Slomp C, Morris E, Price M, Elliott AM, Austin J. The stepwise process of integrating a genetic counsellor into primary care. European journal of human genetics : EJHG. 2022;30(7):772-81.

6. Patch C, Middleton A. Genetic counselling in the era of genomic medicine. British medical bulletin. 2018;126(1):27-36.

7. Braithwaite D, Emery J, Walter F, Prevost AT, Sutton S. Psychological impact of genetic counseling for familial cancer: a systematic review and meta-analysis. Familial cancer. 2006;5(1):61-75.

8. Facher JJ, Robin NH. Genetic counseling in primary care. What questions are patients likely to ask, and how should they be answered? Postgraduate medicine. 2000;107(3):59-60, 3-6.

9. Houwink EJF, van Luijk SJ, Henneman L, van der Vleuten C, Jan Dinant G, Cornel MC. Genetic educational needs and the role of genetics in primary care: a focus group study with multiple perspectives. BMC family practice. 2011;12(1):5.

10. Rose P, Humm E, Hey K, Jones L, Huson SM. Family history taking and genetic counselling in primary care. Family practice. 1999;16(1):78-83.

11. Metcalfe A, Werrett J, Burgess L, Clifford C. Psychosocial impact of the lack of information given at referral about familial risk for cancer. Psycho-oncology. 2007;16(5):458-65.

12. Riedijk S, Oudesluijs G, Tibben A. Psychosocial aspects of preconception consultation in primary care: lessons from our experience in clinical genetics. Journal of community genetics. 2012;3(3):213-9.

13. Borreani C, Gangeri L. Genetic counselling: communication and psychosocial aspects. Tumori. 1996;82(2):147-50.

14. Micheletto MR, Valerio NI, Fett-Conte AC. Effects of a genetic counseling model on mothers of children with down syndrome: a Brazilian pilot study. Journal of genetic counseling. 2013;22(6):784-94.

15. Carroll JC, Rideout AL, Wilson BJ, Allanson JM, Blaine SM, Esplen MJ, et al. Genetic education for primary care providers: improving attitudes, knowledge, and confidence. Canadian family physician Medecin de famille canadien. 2009;55(12):e92-9.

16. Tomatir AG, Sorkun HC, Demirhan H, Akdağ B. Genetics and genetic counseling: practices and opinions of primary care physicians in Turkey. Genetics in medicine : official journal of the American College of Medical Genetics. 2007;9(2):130-5.

17. Harding B, Webber C, Ruhland L, Dalgarno N, Armour CM, Birtwhistle R, et al. Primary care providers' lived experiences of genetics in practice. Journal of community genetics. 2019;10(1):85-93.

http://dx.doi.org/10.52533/JOHS.2022.21216