Original Article

Impact of the Absent Genetic Counseling on Muscular Dystrophy in Taif City

Ghaliah Obaid Alnefaie[1], Naif Edah Alomairi[2], Turk Obaid Al Nefaie[3], Sahar Mohammed Alnefaie[4], Nisreen Khalid Aref Albezrah[5], Rahaf Mansi Alalawi[6], Ghadah Homood Al-swat[6], Fai Nasser Alsukhayri[6], Hatun Abdullah Althagafi[6], Shouq Mohammed Alsalmi[6], Wejdan Dhafer Alamri[6], Rawabi Abdullah Althubaiti[6], Maryam Saud Aljaid[7*]

[1]Department of Pathology, College of Medicine, Taif University, Taif, 21944, Saudi Arabia. [2]Department of Internal Medicine, College of Medicine, Taif University, Taif 21944, Saudi Arabia. [3]Department of Family Medicine, Ministry of National Guard Health Affairs (MNGHA), Jeddah, Saudi Arabia. [4]Department of Surgery, College of Medicine, Taif University, Taif 21944, Saudi Arabia. [5]Department of Obstetric and Gynecology, College of Medicine, Taif University, Taif 21944, Saudi Arabia. [6]Department of Medicine, Taif University, Taif 21944, Saudi Arabia. [7]Department of Pediatrics, College of Medicine, Taif University, Taif 21944, Saudi Arabia.

Abstract

Muscular dystrophies are neuromuscular diseases and heterogeneous neuromuscular disorders that share similar clinical features and dystrophic changes on muscle biopsy. Genetic counsellors assess clients' patients for a variety of inherited conditions, such as birth defects. They review genetic test results with individuals and families and support them in making decisions based on those results. In this study, we examine the impact of the absence of genetic counselling on patients with muscular dystrophies. 168 patients with MDs aged 18 to 58 were asked to complete the Genomics Outcome Scale, which consists of six items. Several cognitive, decisional, behavioural, and emotional control items are included, as well as items assessing their ability to plan for the future. There have been two groups of participants (muscular dystrophies patients), one group has consulted with a genetic counsellor (GC), and the other group has not consulted with GC. The results determine that lack of knowledge about family risk in the group not visited GC 81% with a p-value of 0.000. In addition, visiting a genetic counsellor had a significant impact on different aspects of managing disease, making decisions, and planning for the future. 17% of MDs who had visited GC and were single feel neutral, neither agreeing nor disagreeing that they can make decisions regarding their health. It is important to consider the impact of genetic counselling on MDs. Genetic counselling involves helping individuals understand and adapt to the psychological, psychological, and familial implications of a gene's influence on disease.

Keywords: Genetic counselling, MDs, Genomics outcome scale, Impact, Muscular dystrophies

Introduction

Among muscular dystrophy MDs, there is a clinically, genetically, and biochemically heterogeneous group of disorders that share a clinical manifestation of progressive muscle weakness as well as pathological appearances on muscle biopsy that are indicative of muscle dystrophy. These diseases are characterized by progressive muscle weakness affecting the limbs, axial muscles, and facial muscles to varying degrees. According to the type of disorder, severity, age at onset, rate of progression, complications, and outcome vary greatly [1].

As a result of muscle biopsy, the severity of the results can vary, depending on the pathological findings. These findings may include variations in the size of muscle fibres, degeneration and regeneration of the muscles, and an increase in fibrosis [2]. Many genes are responsible for MD phenotypes. Considering this, it is not surprising that there is a wide range of phenotypes associated with MDs, which may involve cardiac or respiratory muscles, the central nervous system, or the ocular structure [3]. Several new genes have been identified in the past few years, which has contributed to a more comprehensive understanding of both the clinical and molecular aspects of MDs. About 60% of the cases can be identified with the help of comprehensive gene panels, suggesting that both new genes and unusual mutations of the currently known genes are responsible for the remainder of the cases [3].

Identifying the genetic basis for the most common forms of muscular dystrophy has led to an unexpected expansion of the clinical range of variants, including allelic disorders that

Address for correspondence: Maryam Saud Aljaid, Department of Pediatrics, College of Medicine, Taif University, Taif 21944, Saudi Arabia.
Maryam@tu.edu.sa

How to cite this article: Alnefaie GO, Alomairi NE, Al Nefaie TO, Alnefaie SM, Albezrah NKA, Alalawi RM, et al. Impact of the Absent Genetic Counseling on Muscular Dystrophy in Taif City. Arch Pharm Pract. 2023;14(S):A06231448.

differ from the original muscular dystrophy described [4, 5]. The most common muscular dystrophies can be categorized according to their clinical features and age of onset (for example, limb-girdle muscular dystrophies, Emery-Dreifuss muscular dystrophies, and congenital muscular dystrophies) [6]. It has been found that the prevalence of muscular dystrophy as a group ranges from 19.8 to 25.1 per 100,000 persons [7].

There has been a shift in the objectives of genetic counseling over the last three decades. Reviewing past literature reveals two main schools of thought [8]. The first emphasizes the prevention of birth defects and genetic disorders while the second emphasizes the improvement of psychological well-being for patients who are adjusting to a genetic condition or risk. Even though both types of goals advocate for the patient to make their own reproductive decisions, the former focuses on the patient making choices that will mitigate the impact of genetic disorders on the patient's life [8]. Health care providers may have different types of goals based upon their training and orientation in genetics, their sociocultural values, or their healthcare settings' priorities. As a matter of fact, there are several reasons to dismiss the prevention of birth defects as a worthwhile objective. Using a genetic counselling sub-specialty as a framework may assist counsellors in establishing appropriate counselling goals and specific aims for populations in the reproductive, paediatric/adult, and common disease settings [8]. A genetic health care provider should work toward reaching a consensus on the objectives of genetic counselling, considering the needs of the patients, given the current level of genetic information, technologies, and the need to evaluate genetic counselling practices [8].

In recent years, genetic counselling has become increasingly common in virtually all medical specialties to assist patients in understanding and adjusting to the physical, psychological, and familial ramifications of genetic contributions to disease [9]. These include obstetrics, paediatrics, cancer, cardiology, and neurology.

In 1969, Sarah Lawrence College in New York established the first master's level genetic counselling program in the United States [10, 11]. It is estimated that there are currently approximately 7,000 genetic counsellors (GCs) working in more than 28 countries, according to a recent study [9]. It is estimated that more than 60% of genetic counsellors in the world are in North America, where there are more than 4000 genetic counsellors and 39 master's degree programs [9]. While there are approximately 100 GCs in the Middle East, most of which are in Israel and Saudi Arabia [9].

The field of Genetic Counselling is relatively new in Saudi Arabia. According to the Saudi Commission for Health Specialties, the first-degree program in genetic counselling was approved for higher education in 2015 [12]. Although many Saudi families are seeking genetic counselling in order to gain more information regarding their health status and future plans, genetic counsellors in Saudi Arabia face many challenges. Saudi Arabia also has distinctive issues related to the prevalence of consanguineous marriages (over 50%). As compared to other regions, Saudi Arabia has a high prevalence of inherited disorders [13]. As a consequence of a number of factors, this significant increase has been observed, among them high rates of consanguinity, rapid population growth, and older parental ages at childbearing ages [14-16].

A genetic counselling program can make a significant difference in the treatment of MDs and may serve as an early intervention process to prevent and improve health outcomes.

In this study, we aim to investigate the possible encouraging effect of genetic counselling as it pertains to patients who have been diagnosed with MDs.

Materials and Methods

A total of 168 muscular dystrophies patients were surveyed using the Genomics Outcome Scale (GOS) six-item questionnaires, which included seven-point Likert rating scale questions (1: strongly disagree, 2: disagree, 3: slightly disagree, 4: neutral, 5: slightly agree, 6: agree, and 7: strongly agree). A study was conducted at Al Hada Armed Forces Hospital, Taif, Saudi Arabia, which is part of the General Medical and Surgical Hospitals Industry, exclusively for Saudi military personnel and their families. A study was carried out between November 2022 and March 2023. Patients with MDs are included in the study, whereas patients without MDs are excluded. Based on six items, the Genomics Outcome Scale (GOS) survey was designed as a shorter version of the GCOS-24 [17]. In order to provide an assessment tool that can be used both within and outside of clinical genetics services, as well as to reduce respondent burden [18]. As part of the questionnaire, socio-demographic information was collected, including age and marital status. Participants were asked to self-report their history of muscular dystrophy and previous genetic counselling visits. The GOS survey included six elements to measure the impact that genetic counsellor GC leaves on patients with MDs. It includes items demonstrating cognitive control, decisional control, behavioural control as well as emotional regulation and assessing their ability to plan for the future.

Informed consent was obtained from all participants prior to completing the questionnaire. The research and ethics committee of the general directorate of medical services has approved this project (Reg. No. H-02-T-078 and Reference: REC-2022-673).

Numbers and percentages will be used to present categorical data. In order to compare the average 7-point Likert scale responses among respondents. A Mann-Whitney U-test with two tails is used. A p-value of 0.05 will be used to determine statistical significance for all responses. All data were

analysed using Statistical Package for Social Sciences version 22.0 for Windows (SPSS, Inc., Chicago, IL).

Results and Discussion

Patient Characteristics

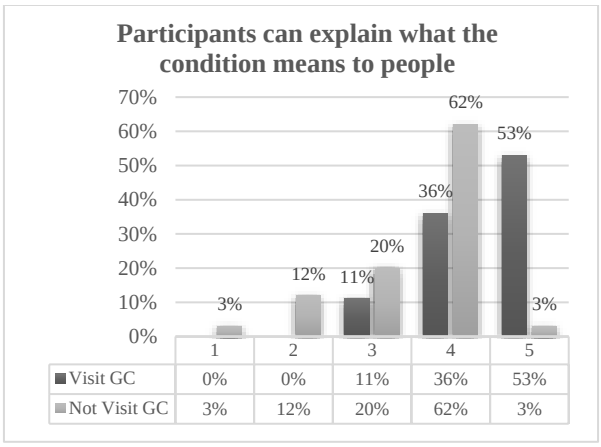
A total of 168 muscular dystrophy patients participants were divided into two groups for the study. 56% of the control group participants have been diagnosed with muscular dystrophy and have visited a genetic counsellor. In the case group, 44% of the participants complained of muscular dystrophy and did not seek genetic counselling. There was no statistical significance between the two groups in terms of gender, marital status, and family history **(Table 1)**.

Table 1. Study muscular dystrophy patients' characteristics.

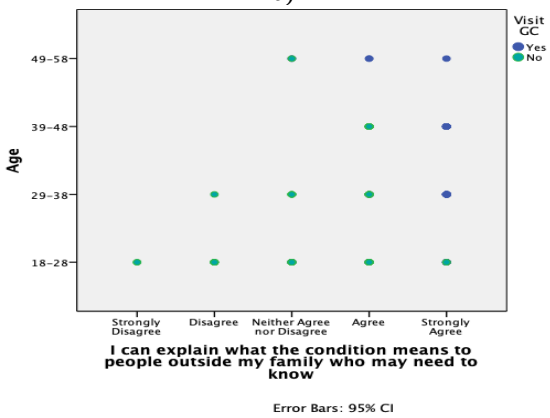
	Visited Genetic Counselling Group	Not Visited Genetic Counselling Group
Total number	94	74
Age, median (IQR)	33 (18-58)	30 (18-58)
Gender		
Males	67	51
Females	27	23
Marital Status		
Married	38	24
Single	56	50
Do you have any personal history of congenital muscular dystrophy?		
Yes	27	26
No	67	48

Explaining the Condition to Others

Regarding the ability of patients s to explain what the condition means to people outside their family who may need to know (e.g., teachers, social workers), participants in the control group agreed and strongly agreed with 36% and 53% respectively (p=0.05, p=0.001) that they are able to explain. In contrast, 62% of the case group agreed that they could **(Figure 1a)**. Only 3% of the respondents strongly disagreed that they could explain their condition, and their age group ranged from 18 to 28 with p=0.000. Among the participants in both groups who are between the ages of 39 and 48, the case group agreed, and the control group strongly agreed that they could explain the condition **(Figure 1b)**.



a)

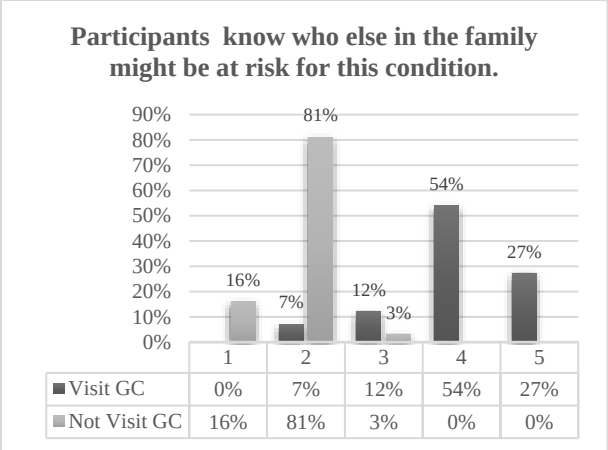


b)

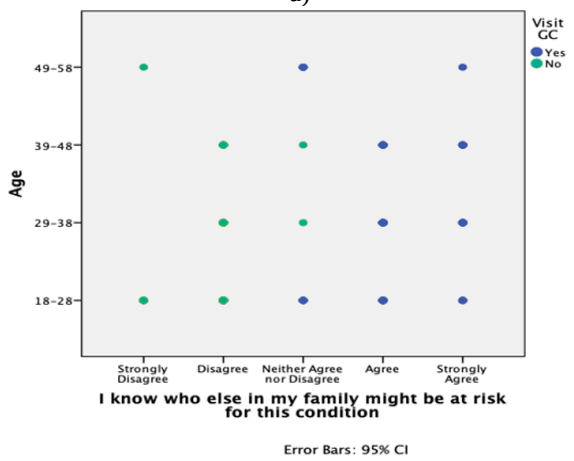
Figure 1. Explaining muscular dystrophy.
a) There are two groups of participants in this study: muscular dystrophy patients who visited the Genetic Counsellor GC (control group) and those who did not visit the Genetic Counsellor GC (case group). The five-point linker is as follows: 5 strongly agree, 4 agree, 3 neither agree nor disagree, 2 disagree, 1 strongly agree. In the control group, responses ranged from neutral to strongly agreed, whereas in the case group, responses varied widely. b) the chart shows responses based on age group. In the case group of participants between 18 and 28 years of age, there was a strong disagreement regarding how the condition could be explained. Error bars are 95% confidence intervals.

Family Risk

It was found that in the control group, 54% and 27% of participants agreed and strongly agreed that they are aware of who else in their family may be at risk of this condition (p=0.000). In contrast, 81% of participants in the case group disagreed (p=0.001) **(Figure 2a)**. In the case group, 16% of participants aged between (18 - 28) and (49 - 58) strongly disagreed that they have knowledge about family risk (p=0.000) **(Figure 2b)**.



a)

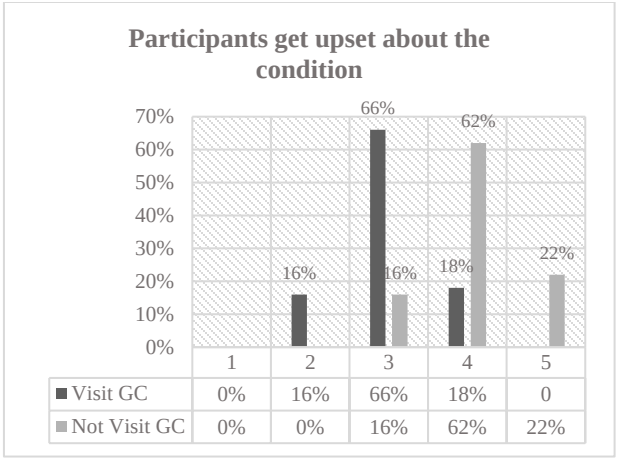


b)

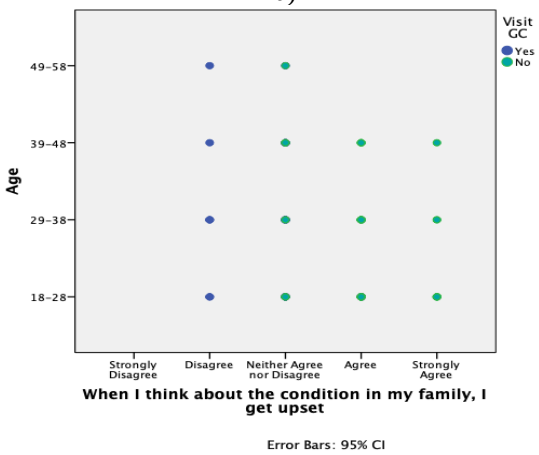
Figure 2. The family is at risk for muscular dystrophy. a) An analysis of the bar chart displays the participants' responses to two different groups: muscular dystrophy patients who visiting a GC (control group) and not visiting a GC (case group). According to the five-point linker, 5 strongly agree, 4 agree, 3 neither agree nor disagree, 2 disagree, 1 strongly agree. Responses from the case group ranged from neutral to strongly disagreed, while responses from the control group varied. b) According to the chart, the responses vary based on the age group. Control group members in all age groups strongly agreed that they were aware of the family risk. Error bars are 95% confidence intervals.

Feeling About the Condition

In the control group, the majority of muscular dystrophy patients who had visited GC when they thought about their condition felt neutral rather than upset or unsatisfied with 66%, p=0.0341. A significant proportion of patients with muscular dystrophy who had not visited GC reported feeling upset about their condition, with a p-value of 0.0450 **(Figure 3a)**. There is no sense of upset among most control group participants aged 49 to 58. In the case group, however, the participants from the same age group reported feeling neutral with a p-value of 0.000 **(Figure 3b)**.



a)



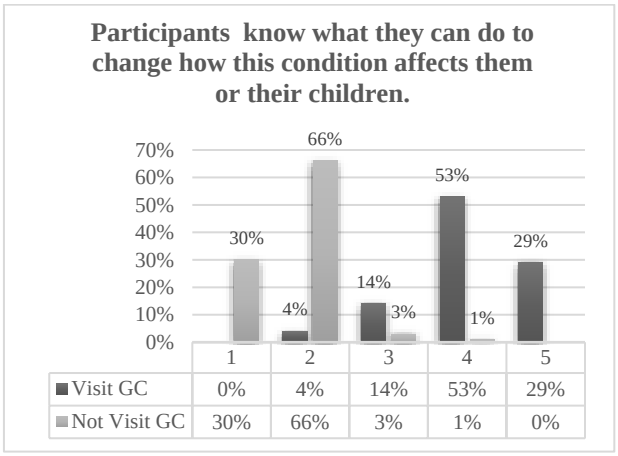
b)

Figure 3. A feeling of upset about the condition. a) A bar chart illustrates the results of the survey participants' responses: muscular dystrophy patients who visited GC (control group) or did not visit GC (case group). A five-point linker indicates that: 5 strongly agree, 4 agree, 3 neither agree nor disagree, 2 disagree, and 1 strongly agree. In both groups, there is no strong disagreement about the fact that they feel upset about their condition. Participants in the case group are the only ones who strongly agree with the statement that they feel upset. In contrast, participants from the control group disagree with the statement. b) The chart shows the results according to the age group. Neither group, in any age group, strongly disagreed that they are upset. Error bars are 95% confidence intervals.

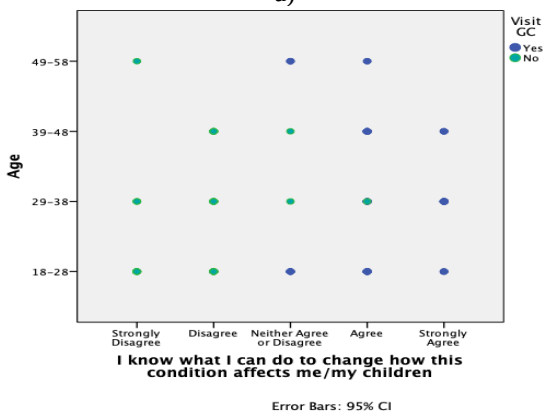
Change the Effect of the Condition

In the control group, 53% of participants agreed and 29% strongly agreed that they know what they can do to change how this condition affects them or their children (p=0.0450 and p=0.050, respectively). Conversely, in the case group, 66% and 30% of respondents disagreed and strongly disagreed, respectively (p=0.010 and p=0.0345) **(Figure 4a)**. With a p-value of 0.001, the majority of the control group in the age range 49-58 years strongly disagreed that they knew what they could do to change how this condition affected

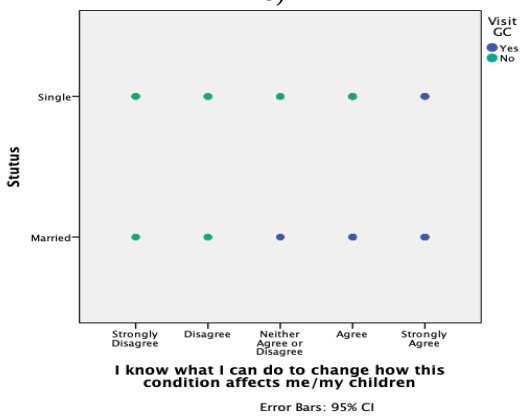
them or their children. Furthermore, most participants in the control group who have visited GC and are single strongly agree that they know what they can do to change the way this condition affects them or their children p= 0.002 **(Figure 4c)**.



a)



b)

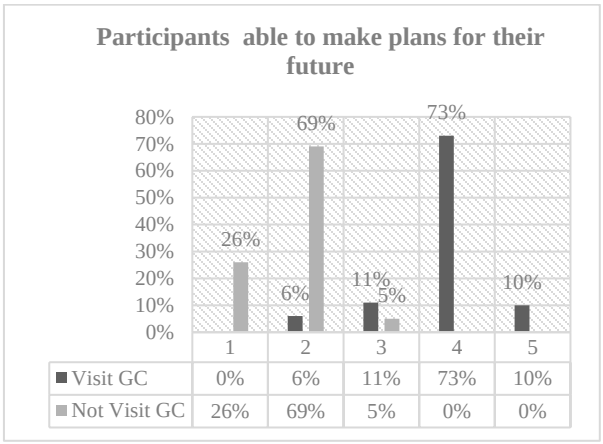


c)

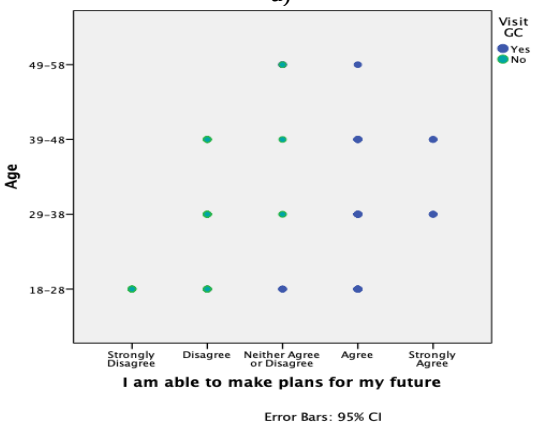
Figure 4. Being aware of what can be done to change the effects of muscular dystrophy.
a) A bar chart shows the participants' responses with respect to their visit to GC (control group) or not visiting GC (case group). Five-point linker: 5 strongly agree, 4 agree, 3 neither agree nor disagree, 2 disagree, 1 strongly agree. The control group agreed that they are conscious, while the case group disagreed.
b) The chart illustrates responses based on age group. Among the participants in the case group aged 49-58 years, all strongly disagreed that they are aware of what needs to be done in order to improve their condition.
c) The chart illustrates responses based on the statutes. In the control group, all single participants strongly agreed that they knew how to change how this condition affected them or their children. Error bars are 95% confidence intervals.

Future Plan

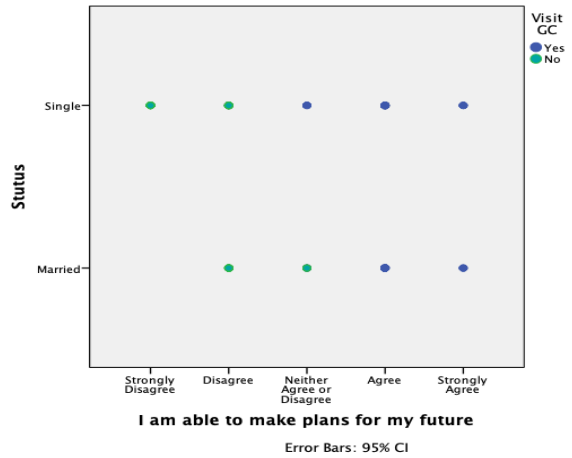
In the control group, 73% agreed that they were able to make plans for their future with a p-value of 0.020. However, 69% of participants in the case group disagreed that they were capable with p=0.0345 **(Figure 5a)**. Only participants from the case group ages 18-28 strongly disagreed with the statement that they are able to make plans for their future with a p-value of 0.050. With a p-value of 0.0001, participants in the age group 49-58 years who are in the case group considered the future plans neutral. None of the married participants in either group strongly disagreed that they are capable of making future plans.



a)



b)

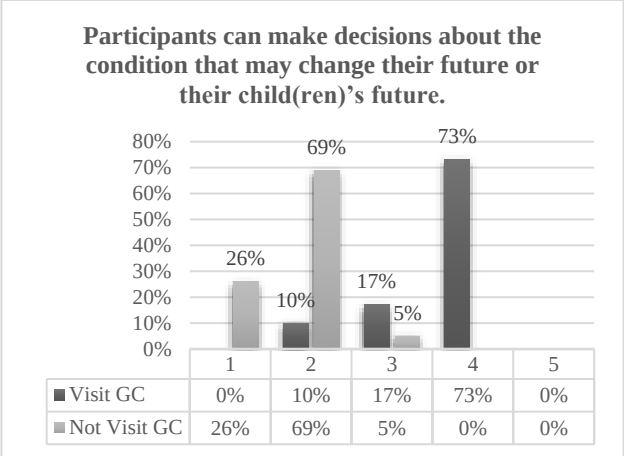


c)

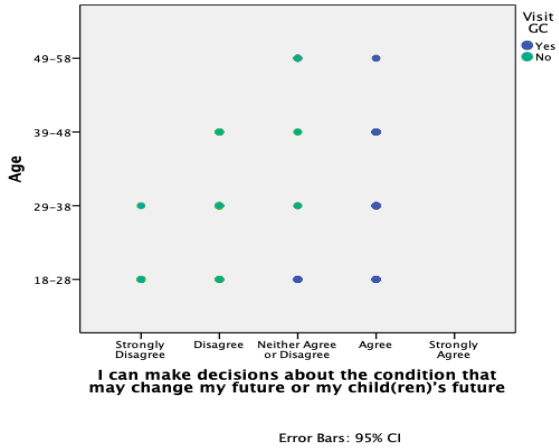
Figure 5. Future plans.
a) There are two groups of participants represented in the bar chart: muscular dystrophy patients who visited GC (control group) and those who did not visit GC (case group). Five-point linker: Five strongly agree, four agree, three neither agree nor disagree, two disagree, and one strongly agrees. It was agreed by the control group that they are capable of making plans for the future, while the case group disagreed.
b) Based on the age group, the chart illustrates the responses. In the case group, all participants aged 49-58 years did not agree or disagree that they could make plans, while in the control group, all participants agreed.
c) The chart illustrates responses based on the statutes. Singles who are not married in the case group are the only ones who strongly disagree that they are capable of making future plans. Error bars are 95% confidence intervals.

Decisions About the Condition

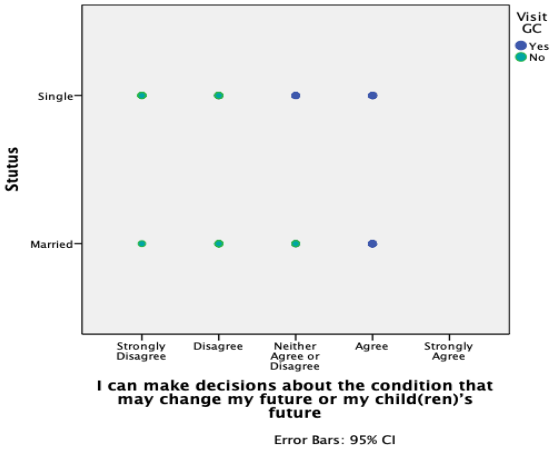
A majority of 73% of those in the control group who had visited GC agreed that they can make decisions about the condition that may impact their future or their children's future with p=0.020. While 69% of participants in the case group who had not visited GC disagreed that they could make decisions regarding conditions that could change their future or the future of their children with p=0.0345 **(Figure 6a)**. A majority of the participants in the 49–58year age group in the case group did not agree or disagree that they could make decisions about the condition that could change their future or the future of their children, p=0.0001 **(Figure 6b)**. A control group of single, non-married participants was divided into neutral and agreed participants. Although most of the married individuals in the control group chose that option, they acknowledged that they could make the decision **(Figure 6c)**.



a)



b)



c)

Figure 6. Decision-making.
a) The chart in Figure A displays the participants' responses in two groups: muscular dystrophy patients who visited GC (control group) and those who did not visit GC (case group). A five-point linker is presented below 5 strongly agree, 4 agree, 3 neither agree nor disagree, 2 disagree, and 1 strongly agree. No one from either group strongly agrees that they can make decisions about the condition that may affect their future or the future of their children. b) The chart illustrates the responses based on the

age group. Among participants aged 49 to 58, those in the case group neither agreed nor disagreed that they were able to make decisions, while those in the control group agreed that it was possible. c) The chart illustrates the responses based on the statutes. In the control group, all married participants agreed that they are capable of making decisions for themselves. Error bars are 95% confidence intervals.

There are a variety of muscular dystrophies MDs that are clinically and genetically heterogeneous neuromuscular disorders, and where muscle biopsy results are consistent with dystrophic muscle disease [2]. Providing genetic counselling entails assisting individuals in understanding and adapting to the psychological, psychological, and familial implications of a gene's contribution to a disease. The following steps are integrated into this process: Interpretation of the family and medical histories to determine the likelihood that the disease will occur or recur. An introduction to inheritance, testing, management, prevention, resources, and research all that to assist in promoting informed decisions and adjusting to risk or condition [19].

In this study, we examine the impact of the absence of a genetic counsellor on patients with muscular dystrophies. As muscular dystrophy patients demonstrate their ability to explain their condition, patients strive to gain a comprehensive understanding of their disease. For that helping patients cope with and adapt to genetic information is an important aspect of genetic counselling [20]. Researchers found that whether a patient consults GC does not affect his knowledge of his condition. Pulse neurologist physicians usually provide patients with a detailed explanation of their condition. Our study, however, showed that young patients between the ages of 18 and 28 do not usually have the ability to discuss their condition. According to the new concept analysis, young people with chronic illness have similar views about themselves and their lives compared to their healthy peers [21].

Muscular dystrophies are inherited diseases. It is therefore necessary to know the family risk associated with muscular dystrophies for patients with MDs. Study results indicate that MDs patients who do not receive GC are significantly less aware of who else in their family is at risk of the disease than those who do receive GC. In seeking GC, women's perception of genetic risks and their level of concern appear to be associated with their perception of genetic risk, which may be in accordance with actual risk figures [22].

It is understandable that having any kind of disease makes the patient feel sad and upset. As demonstrated in this study, the majority of MDs patients who did not consult with GC felt upset about their condition. In contrast, MDs patients who have visited GC have exhibited a significantly neutral attitude towards the illness. Therefore, seeing GC can have an impact on a patient's emotional response to the illness. A number of patients have reported an improvement in communication between them and their spouses and other family members as a result of GC. As an expert in his field, the counsellor is viewed by patients as a valuable source of both information and support [23].

In presenting genetic information, it has been suggested that pitting intellect against emotion may lead to psychological harm 8. In order to aid the client in interpreting the situation as it is, rather than as he or she wishes it were, GC is essential to addressing the patient's fears, hopes, defences, and rationalizations [24]. Genetic counselling should include a mental health component, according to Hecht and Holmes in 1972 [25].

In this study, a significant percentage of patients who consulted GC knew what they could do to control how this condition affected them or their children, while a significant percentage of patients who did not consult GC did not know what to do. The process of genetic counselling is an educational and psychotherapeutic process that emphasizes genetic information in a dynamic, interactive way. Patients are assisted in personalizing technical and probabilistic genetic information through a therapeutic relationship established between providers and patients, promoting self-determination, and enhancing their abilities to cope with changes as time goes on. Using genetic information should be facilitated in such a way that minimizes psychological distress and enhances personal control for patients [26].

Furthermore, patients exhibited similar responses with regard to the ability to make future plans, genetic counselling made a significant impact on the ability of patients to plan their futures. GC aims to assist clients in understanding the scientific causes of genetic conditions and to provide them with feelings of mastery through the discussion of resources and the presentation of future options. By providing these resources, genetic counselling can assist in facilitating the adaptation process of patients. Information about future reproductive decisions and their meaning for families may also influence those decisions, which makes long-term decision-making in this setting less urgent than in the prenatal setting [8].

A genetic counsellor also found that patients who visited him or her were more likely to make decisions about their future or the future of their children than those who did not see GC. Helping individuals/couples understand their options and current medical information in order to make informed decisions, assisting individuals/couples in coping with their genetic disorders, and removing patient guilt or anxiety [8]. In addition to improving the general health of the population, the goal of preventing disease or abnormality was still strongly supported. This model of genetic counselling emphasizes the importance of empowering patients to make their own decisions, but also apparently trusts clients to make preventative choices [8].

As part of these goals, genetics health providers were acknowledged that these issues could be emotionally difficult and that they needed support in dealing with them. The fine line between past eugenic goals and those of preventative genetic counselling was discussed by Kessler in 1979. It was highlighted that there is a discrepancy in whether the prevention goals are societal or individual in nature. Genetic counselling consists of communicating genetic information to patients, assisting them in making appropriate decisions, and assisting them in coping with the information and consequences of genetic disorders [27].

Considering the impact of genetic counselling on MDs should be emphasized. There is a common goal in genetic counselling for common diseases, which is the enhancement of health-promoting behaviours and an understanding of personalized disease risk [28-30]. The goal reflects a public health objective of preventing disease. Providing genetic counselling for common diseases may also include discussions of reproductive options and risks to children; however, the primary goal is to promote health.

Conclusion

It is important to consider the impact of genetic counselling on MDs. Genetic counselling involves helping individuals understand and adapt to the psychological, psychological, and familial implications of a gene's influence on disease.

ACKNOWLEDGMENTS: The authors would like to acknowledge the Deanship of Scientific Research, Taif University for funding this work. The authors would like to extend their acknowledgment to Al-Hada Armed Forces Hospital (Taif, Saudi Arabia), for approving the conducting of the study.

CONFLICT OF INTEREST: None

FINANCIAL SUPPORT: None

ETHICS STATEMENT: Approval for this study was sought and obtained by the research ethics committee of the medical services' general directorate at the Armed Forces Hospital, Taif region, Saudi Arabia (Reg. No. H-02-T-078 and Reference: REC-2022-673). All research was performed in accordance with relevant guidelines/regulations. Written informed consent was obtained from all participants before their recruitment into the study.

References

1. Emery AE. The muscular dystrophies. Lancet. 2002;359(9307):687-95.
2. Bertini E, D'Amico A, Gualandi F, Petrini S, editors. Congenital muscular dystrophies: a brief review. Seminars in pediatric neurology; 2011: Elsevier.
3. Zambon AA, Muntoni F. Congenital muscular dystrophies: What is new? Neuromuscul Dis. 2021;31(10):931-42.
4. Mercuri E, Muntoni F. The ever-expanding spectrum of congenital muscular dystrophies. Ann Neurol. 2012;72(1):9-17.
5. Guglieri M, Straub V, Bushby K, Lochmüller H. Limb–girdle muscular dystrophies. Curr Opin Neurol. 2008;21(5):576-84.
6. Mercuri E, Muntoni F. Muscular dystrophies. Lancet. 2013;381(9869):845-60.
7. Theadom A, Rodrigues M, Roxburgh R, Balalla S, Higgins C, Bhattacharjee R, et al. Prevalence of muscular dystrophies: a systematic literature review. Neuroepidemiology. 2015;43(3-4):259-68.
8. Biesecker BB. Goals of genetic counseling. Clin Genet. 2001;60(5):323-30.
9. Abacan M, Alsubaie L, Barlow-Stewart K, Caanen B, Cordier C, Courtney E, et al. The global state of the genetic counseling profession. Eur J Hum Genet. 2019;27(2):183-97.
10. Heimler A. An oral history of the National Society of Genetic Counselors. J Genet Couns. 1997;6(3):315-36.
11. Stern AM. A quiet revolution: the birth of the genetic counselor at Sarah Lawrence College, 1969. J Genet Couns. 2009;18:1-11.
12. Balobaid A, Qari A, Al-Zaidan H. Genetic counselors' scope of practice and challenges in genetic counseling services in Saudi Arabia. Int J Pediatr Adolesc Med. 2016;3(1):1-6.
13. Al-Owain M, Al-Zaidan H, Al-Hassnan Z. Map of autosomal recessive genetic disorders in Saudi Arabia: concepts and future directions. Am J Med Genet Part A. 2012;158(10):2629-40.
14. Middle I, Al-Salloum A, Al-Herbish A, Qurachi M, Al-Omar A. Regional variations in the prevalence of consanguinity in Saudi Arabia. Saudi Med J. 2007;28(12):1881-4.
15. Al-Gazali L, Hamamy H, Al-Arrayad S. Genetic disorders in the Arab world. Bmj. 2006;333(7573):831-4.
16. El-Hazmi M, Al-Swailem A, Warsy A, Al-Swailem A, Sulaimani R, Al-Meshari A. Consanguinity among the Saudi Arabian population. J Med Genet. 1995;32(8):623-6.
17. McAllister M, Wood AM, Dunn G, Shiloh S, Todd C. The Genetic Counseling Outcome Scale: a new patient-reported outcome measure for clinical genetics services. Clin Genet. 2011;79(5):413-24.
18. Grant PE, Pampaka M, Payne K, Clarke A, McAllister M. Developing a short-form of the genetic counselling outcome scale: The genomics outcome scale. Eur J Med Genet. 2019;62(5):324-34.
19. Resta R, Biesecker BB, Bennett RL, Blum S, Estabrooks Hahn S, Strecker MN, et al. A new definition of genetic counseling: National Society of Genetic Counselors' task force report. J Genet Couns. 2006;15:77-83.
20. Kessler S. Genetic counselling: Psychological dimensions: Academic Press; 2013.
21. Taylor RM, Gibson F, Franck LS. A concept analysis of health-related quality of life in young people with chronic illness. J Clin Nurs. 2008;17(14):1823-33.
22. Scarupa HJ. Genetic Counseling: Helping People Cope with Inherited Ills. New Dir. 1987;14(1):3.
23. Bernhardt BA, Biesecker BB, Mastromarino CL. Goals, benefits, and outcomes of genetic counseling: client and genetic counselor assessment. Am J Med Genet. 2000;94(3):189-97.
24. Kallmann FJ. Psychiatric aspects of genetic counseling. Am J Hum Genet. 1956;8(2):97.
25. Hecht F, Holmes LB. What we don't know about genetic counseling. Mass Medical Soc; 1972. p. 464-5.
26. Biesecker BB, Peters KF. Process studies in genetic counseling: peering into the black box. Am J Med Genet. 2001;106(3):191-8.
27. Kessler S. The processes of communication, decision making and coping in genetic counseling. Genetic Counselling: Elsevier; 1979. p. 35-51.
28. Karpov VY, Mal GS, Bakulina ED, Rysakova OG, Fedorova TY, Svetlichkina AA. Functional Capabilities of the Heart of Young Men Who Regularly Experience Muscle Loads. J Biochem Technol. 2021;12(4):90-3.
29. Moga TD, Moga I, Venter A, Cavalu S. Sarcopenia of Iliopsoas and Abdominal Muscles and Coexistence with Osteoporosis—Case Presentation. Pharmacophore. 2021;12(6):61.
30. Bodnar P, Bodnar Y, Bodnar T, Soroka Y, Liudmyla B. Histological Changes in Muscles During the Lower Extremities Thrombosis in Individuals with Gastrointestinal Tract Cancer. Int J Pharm Res Allied Sci. 2021;10(3).