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Evaluation of quality of life in patients with muscular dystrophy by socio-demographic characteristics

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Abstract

Aim: Genetic myopathies are diseases that progress and impair quality of life. It was aimed to evaluate the relationship between socio-demographic characteristics and the quality of life of 146 patients.

Materials and Methods: The WHOQOL-BREF scale was applied, and the scores in the questionnaire were converted to WHOQOL 4-20 and WHOQOL 0-100 score ranges and the relationships between the WHOQOL-BREF questionnaire results were evaluated.

Results: There was no significant difference in the scale results according to gender and age ($p>0.05$ for all). There were significant findings regarding education level, marital status, and disease severity on the WHOQOL-BREF scale.

Conclusion: It is thought that it can be a reference for researchers by revealing the findings that will increase the patient's quality of life.

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Introduction

Genetically Caused Muscular Dystrophies (GCMD) are primary muscle diseases that cause dystrophic changes in the histochemical structure of the muscle with gene mutation. The main clinical feature of these disorders is that they are a progressive disease group [1]. It is common in consanguineous marriages, clinical symptoms and course may differ between siblings with the same diagnosis; those with mild symptoms are not detected, they occur at different ages, the incidence of GCMDs cannot be determined precisely, and prevalence detection is used more frequently according to the case level [2, 3]. In GCMD, muscle weakness in the extremities can lead to joint contractures, accelerating the dystrophic process.

GCMD includes signs of eye, partial hearing, metabolic, hormonal, and cognitive disorders, endocrine disorders, and central nervous system involvement of varying severity. Behavioral disorders such as obsessive-compulsive disorder, schizotypal personality disorder, and lack of empathy have been observed. Although laboratory values, electromyography, and muscle biopsy are used for diagnosis, a definitive diagnosis can be made by gene analysis with technological development [4].

Mild ones may lead a life close to a regular Activity of Daily Living (ADL) value, while increased dependency may develop in their moderate to severe life. Information showing which parameters are related to the patient's ADL and functional level is limited. In addition, information on the adult group's quality of life, vital needs, and mental health is scarce, and most studies have focused on pediatric muscle groups [5-6].

There is no basic cure for GCMDs; physical and occupational rehabilitation and proper nutrition are often the main supports. Quality of life (QOL) is a concept that mainly affects an individual's satisfaction in adapting to living conditions [7].

Materials and Methods

Genetic myopathies are diseases that progress and impair quality of life. The aim of this study is evaluating the relationship between socio-demographic characteristics and

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the quality of life of 146 patients who applied to the neuromuscular disease unit.

Study design and sample size

According to the genetic results, a cross-sectional study included 146 eligible patients from 236 patients with primary muscle disease. If the number of individuals in the population is known, the formula for the sample size to be reached is as follows. n=(Nt2 pq)/(d2 (N-1)+t2 pq).

In the formula;

N: Number of individuals in the population

n: Number of individuals to be sampled

p: Frequency of occurrence (probability of occurrence) of the investigated event

q: Frequency of non-occurrence of the investigated event (probability of not occurring)

t: The theoretical value found in the t table at a certain degree of freedom and detected error level.

d: It represents the desired ± deviation according to the incidence of the event.

The sample size was calculated using the above formula. After the calculation, it was found that the sample size should be n=146 in order to obtain the findings with a 5% margin of error in the 95% confidence interval.

Conditions were to be over the age of 18, to have enough education to fill out the scale, and to be a voluntary participant in the study. The purposeful sampling method was used, which is one of the probabilistic sampling methods. Sampling methods were used in the study. Although it was desired to reach the whole population for the study, inclusion criteria were determined due to reasons such as reluctance to participate, inability to reach some patients, and illiteracy of some patients. These inclusion criteria are: being 18 years of age or older, being able to read and understand the given questionnaire, having muscular dystrophy, and participating in the study voluntarily.

Socio-demographic data such as age, gender, marital status, education level and disease severity were recorded and analyzed. The patients were called for control at intervals of 3-6 months, depending on the condition of the disease, and a home rehabilitation program and necessary medical treatment were planned.

The WHOQOL-BREF survey was performed to determine the QOL of the participants. The patients themselves filled the survey forms, and filling out a form took approximately 30 minutes on average. The scores obtained from the survey were transformed into WHOQOL 4-20 and WHOQOL 0-100 score ranges, and relationships between the socio-demographic data of the patients and WHOQOL-BRIEF survey results were evaluated.

WHOQOL-BREF survey

WHOQOL-BREF survey is a shortened form of WHOQOL-100; this is a scale approved by the World Health Organization [8]. It consists of 26 questions in four domains, including physical, psychological, social relationships, and environment sections. The physical domain consists of daily activities, energy, fatigue, mobility, pain and discomfort, dependence on medicinal substances and medical aids, work capacity, sleep, and rest. The psychological domain includes items questioning negative and positive feelings, self-esteem, body appearance, personal belief/spirituality/religion, learning, thinking memory, and concentration. The social relationships domain involves questions related to social support, intimate relationship, and sexual activity. Lastly, the environmental domain questions financial resources, freedom, health and social care, physical safety and personal security, home environment, opportunities for acquiring new skills and knowledge, participation in recreation and leisure activities, and physical environment (noise, traffic, climate, etc.) and transport. Questions in WHOQOL-BREF are answered with a 5-Likert scale. WHOQOL-BREF scores were then transformed into two score ranges, WHOQOL-BREF 4-20 and WHOQOL-BREF 0-100, based on an algorithm considering the number of answered questions in each domain analyzed. A Zero-point indicates the worst possible health condition, while 100 points represent the best possible QOL [8].

Ethical considerations

The ethics committee approval for this cross-sectional study was obtained from University of Health Sciences Gazi Yasargil Training and Research Hospital (date: 05.03.2021 number: 688). This study has been conducted in accordance with the principals set forth in the Helsinki Declaration. The data of the study were collected between 06.03.2021 and 20.11.2021.

Data analysis

Frequency and percentage values from descriptive statistics for categorical variables were used to reveal and explain the socio-demographic characteristics of the patients. Mean, standard deviation, and standard error values were used for continuous variables. Independent Sample t-Test was used to determine the difference in the patient's quality of life between the two groups. One-Way Analysis of Varina's (ANOVA) was used to determine the difference in patients' quality of life between more than two groups [9]. For both t-test and ANOVA analysis, continuous variables are expected to be normally distributed. For this reason, according to the results of the normality test, it was seen that the continuous variables were normally distributed. Because kurtosis and skewness values were between -1.5 and +1.5. The $p<0.05$ value was taken as a reference for statistical significance. Results were analyzed at 95% confidence intervals. All analyzes were performed with the Statistical Package for the Social Sciences (SPSS) 25 version [10].

Results

The majority of the participants with Muscular Dystrophies are male (58.2%, n=85), the lowest rate according to education level is 8.9% (n=13) secondary graduate, the highest rate is 28.1% (n=41) primary graduate, major

Table 1. The quality of life percentile scores of patients with muscular dystrophies (n=146).

Sub Parameters	x̄	SD
General health status	27.65	21.73
Physical	35.91	17.34
Psychological	52.85	21.50
Social relations	49.54	27.08
Environment	44.04	17.37

Table 2. The quality of life scores by gender.

	Sex					
Sub Parameters	Female		Male		t	p
	x̄	se	x̄	se		
General health status	26.64	2.67	28.38	2.43	.477	.634
Physical	35.42	2.41	36.26	1.76	.287	.774
Psychological	51.09	2.76	54.11	2.33	.837	.404
Social relations	49.31	3.71	49.70	2.79	.085	.932
Environment	42.62	2.48	45.7	1.71	.839	.403

*p<0.05.

most of them were single (64.4%, n=94), and according to disease severity status, 35.6% (n=52) of the patients were found to have mild disease severity. Also, the average age is 32 (min: 18; max: 64).

There are two types of scoring according to the quality of life scale. The first of these is the raw scores. Raw scores consist only of the sum of the items in the relevant subparameter. It was determined that the highest quality of life subparameter score according to the raw score was the environmental quality of life score ($\bar{x}$ =22.10±5.56), and the lowest quality of life subparameter was general health status ($\bar{x}$ =4.21±1.74).

There are two types of scoring according to the quality of life scale. The second of these is the percentage points given in Table 1. Percentile scores consist of the sum of the items in the relevant subparameter and the scores that are weighted according to the items. It was determined that the highest quality of life subparameter score according to the percentage score was the psychological quality of life score ($\bar{x}$ =52.85±21.50), and the lowest quality of life subparameter was general health status ($\bar{x}$ =27.65±21.73) (Table 1).

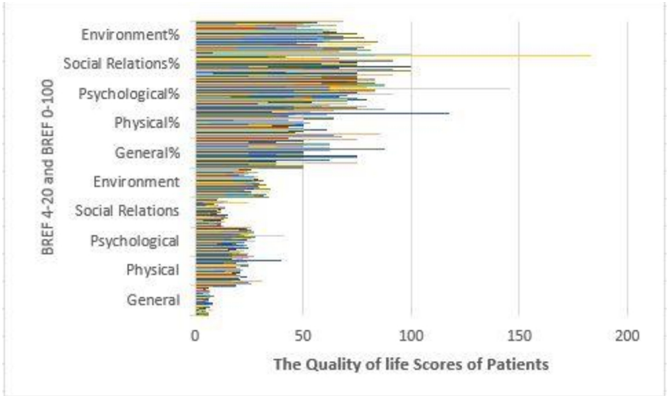

Figure 1. BREF 4-20 Versus BREF 0-100.

The comparison of the participant patients' raw quality of life scores with the percentile quality of life scores is given in figure 1. Accordingly, It was determined that the highest quality of life sub-parameter score according to the raw score was the environmental quality of life score, and the lowest quality of life sub-parameter was general health status. It can be seen in figure 1 the highest quality of life sub-parameter score according to the percentage score was the psychological quality of life score, and the lowest quality of life sub-parameter was general health status.

There is no statistically significant difference in the patient's quality of life according to gender and age, as seen in Table 2.

Social relations and environmental sub-parameter scores, which are among the quality of life scores of the patients, show a statistically significant difference according to the education level, one of the socio-demographic characteristics (p<0.05). According to the Tukey Post Hoc test results to find out which groups the difference originated from, a difference was found between the literate and high school groups. According to this result, the quality of life score of high school graduate patients was higher in social relations and environmental sub-parameters than in the literate group. No statistically significant difference between the other groups, as seen in Table 3.

Among the patients' quality of life scores, general health status and physical sub-parameter scores show a statistically significant difference according to marital status, one of the socio-demographic characteristics (p<0.05). According to the Tukey Post Hoc test results, which was conducted to find out which groups the difference originated from, it was determined that there was a difference between the divorced and married groups. Accordingly, married patients' life scores were higher than divorced patients' general health status and physical quality. According to Table 3, It can be seen that no statistically significant difference was found between the other groups.

The general health status and physical sub-parameter scores of the patient's quality of life show a statistically significant difference according to the severity of the disease, which is one of the socio-demographic characteristics (p<0.05). According to the results of the Tukey Post Hoc test performed to find out which groups the difference originated from, it was determined that there was a statistically significant difference between the groups with mild and very severe disease. Accordingly, the general health status and physical quality of life scores of patients with mild disease were higher than those with severe disease. According to results that can be seen in Table 3, there is no statistically significant difference found between the other groups.

Discussion

The majority of the patients included in the study were male, the median age was 32, the lowest rate was secondary school graduates, the highest rate was primary school graduates, most of the patients were single, and according to the severity of the illness, it was found that the most patients had mild disease severity.

Table 3. The quality in life scores by socio-demographic features (n=146).

		Sub Parameters				
Socio-demographic features		General health status	Physical	Psychological	Social relations	Environment
		x̄ ± se	x̄ ± se	x̄ ± se	x̄ ± se	x̄ ± se
Education status	Literate	24.24±3.72	34.95±3.75	48.23±4.00	42.92±3.95	38.73±3.45
	Primary school	24.08±3.24	32.05±2.40	48.98±3.91	43.69±5.38	40.85±2.91
	Secondary school	30.76±6.58	33.79±4.49	51.92±4.82	39.10±6.95	39.42±4.07
	High school	32.56±3.75	40.22±2.86	57.12±3.01	59.86±3.49	51.31±0.01
	College/university	30.92±4.52	38.72±2.98	62.06±3.91	59.64±4.51	47.03±3.63
F		1.110	1.035	1.650	3.001	3.109
P		.358	.399	.151	.013*	.011*
Marital status	Single	25.53±2.26	34.04±1.75	50.48±2.07	49.82±3.01	43.08±1.91
	Married	33.51±3.04	40.80±2.50	56.29±2.72	49.64±3.24	46.34±2.17
	Divorced	12.50±5.59	25.00±7.23	65.00±24.17	43.33±14.04	40.62±7.84
F		3.485	3.525	1.995	.135	.648
P		.033*	.032*	.140	.874	.525
Disease severity status	So mild	35.41±3.84	42.85±4.96	56.94±7.88	62.50±5.59	53.64±6.42
	Mild	35.81±2.83	41.41±2.60	54.72±2.44	54.16±4.16	46.03±2.45
	Moderate	26.30±3.29	35.63±2.24	54.68±3.02	51.38±3.63	43.42±2.30
	Severe	19.23±4.82	30.49±4.84	49.67±7.22	40.38±6.98	43.75±5.24
	Very severe	16.66±3.89	26.85±2.77	46.60±5.19	38.88±4.70	39.35±3.55
F		4.725	4.020	.878	2.271	1.141
P		.001*	.004*	.479	.065	.340

*p<0.05.

QOL is the evaluation or perception of the general functioning of the patients in their daily lives. When the QoL status of these patients is examined, the quality of life (QoL) is an essential issue in the management of the disease today [11]. QoL measurements provide helpful information about the results of interventions in patients. In GCMD diseases, they are suitable conditions for the measurement of QoL. In this way, it is possible to observe the contribution of the interventions and treatment practices to the quality of life and the progress of their functionality in GCMD patients.

Several studies in the literature investigate QoL in MD patients using various surveys and scales. The most commonly used QOL surveys/scales in the literature include WHOQOL and WHOQOL-BREF, 36-Item Short-Form Health Survey, Pediatric Quality of Life Inventory (PedsQL), Duchenne MD Module, Quality of Life in Neurological Disorders (NeuroQOL), Individualized Neuromuscular Quality of Life (QoL), Quality of Life Profile, Psychosocial Well-Being Questionnaire, TNO-AZL Questionnaire for Adult's Quality of Life (TAAQoL) and EuroQol-5D (EQ-5D) [6,12-20]. In the present study. There is still a need for widely recognized measurement tools with proven validity and reliability to assess the effects of GCMDs on QOL and to accurately quantify changes in MD's QOL resulting from interventions, rehabilitation, and treatment programs. Current scales do not measure all areas of the QOL. In 2011, the Centers for Disease Control and Prevention (CDC) held a meeting on the priorities of MD research. A significant gap was found in the quality of life scales, as it was concluded that these scales did not adequately measure the emotional aspect of the illness, sense of personal significance, participation in society, and access to care [21]. In addition, the diversity of QoL scales used in evaluating GCMD patients makes it difficult to compare studies and obtain guidance.

This study investigated factors affecting the quality of life in patients with muscular dystrophy. In this context, the relationships between gender, marital status and education level, and disease severity of GCMD patients were analyzed with the widely used WHOQOL-BREF questionnaire. The studies mentioned above mainly investigated the quality of life in GCMD patients. Since they usually die early, they were excluded from the study, and the WHOQOL-BREF questionnaire was applied to myopathies with slow progression. It was seen that the result of this scale was significant in GCDMs. This study determined that the highest quality of life subparameter score according to the raw score was the environmental score, and the lowest quality of life subparameter was general health status. On the other hand, it was determined that the highest quality of life sub-parameter score according to the percentage score was the psychological score, and the lowest quality of life sub-parameter was general health status.

According to our results, social relations and environmental sub-parameter scores show a statistically significant difference according to the education level. The difference was found between the literate and high school groups. Ac-

cording to this result, the quality of life score of high school graduate patients was higher in social relations and environmental sub-parameters than in the literate group. No statistically significant difference was found between the other groups. We attributed this result to those GCMD patients with a higher educational level showing a higher adherence to treatment plans and higher participation in social relations and the environment.

It was found that there was no statistically significant difference in the patient's quality of life according to gender and age.

Marital status can mainly be classified as single, marriage problems (separation, divorcing, etc.), and married. In our study, the marriage status of GCMD patients was classified as single, married, or divorced. In this study, general health status and physical sub-parameter scores show a statistically significant difference according to marital status. It was determined that there was a difference between the divorced and married groups. Accordingly, married patients' life scores were higher than divorced patients' general health status and physical quality. No statistically significant difference was found between the other groups. Studies evaluating QOL according to marital status have reported lower QOL levels in married people due to marriage problems. In a study by Han et al., evaluating QOL according to the marital status of male and female participants using the EQ-5D scale, QOL was reported as higher in single compared to married participants, both in women and men. In the same study, QOL was investigated according to marital status among cancer patients, and it was found that QOL was lower in single cancer patients [22]. In a study from Indonesia, gender, marital status, and education level were evaluated as the predictors of QOL in older people using the WHOQOL survey. Different results among studies might have resulted from the differences between the scales used and the participants included. Han et al. investigated QOL in healthy and cancer patients, while Gondodiputro et al. evaluated QOL in older people [23-24]. In addition, economic, cultural, and educational differences among countries may contribute to differences between studies on this issue.

The relationship between the severity of the disease and the quality of life has been tried to be revealed. The physician scored the disease severity for each patient. According to the results, general health status and physical subparameter scores show statistically significant differences according to the severity of the disease. It was determined that there was a statistically significant difference between the groups with mild and very severe diseases. Accordingly, the general health status and physical quality of life scores of those with mild conditions were higher than those with very severe diseases. No statistically significant difference was found between the other groups.

When we look at the previous studies in the literature, the relationship between the WHOQOL scale and the functionality of GCMD patients was examined in general. In this respect, our study is a first in the literature.

Conclusion

The results of this study show that quality of life increases as education level, marital status, and disease severity increase in patients with DCMD. On the other hand, the quality of life in these patients does not differ according to gender. They should be followed up periodically. Further multicenter and comprehensive studies with a more significant number of parameters to be examined are needed. In addition, there is an urgent need in the literature for studies comparing the quality of life before and after rehabilitation and treatment programs in patients with muscular dystrophies. Besides, to increase the quality of life of individuals, physical rehabilitation (healthcare professionals intervene in the physical problem that will worsen by interfering with the disease),social rehabilitation (not keeping them away from society, including them in clubs, and revealing their unique abilities, if any), environmental rehabilitation (land construction, architectural sidewalks, and building entrances, landscaping, designing public vehicles), psychiatric rehabilitation (treating symptoms related to mental illnesses or possible mental losses, ensuring individual responsibility, gaining a sense of recovery, reducing family and social pressure, increasing motivation) measures should be applied. Institutions have some duties to develop a rehabilitation service understanding of rare diseases and to raise awareness.

Study limitations

This study has some limitations. First, the study was conducted in a single center with limited socio-demographic parameters. WHOQOL scores could be compared between more data groups (income level, comorbidities, etc.). QoL measurements could be performed before and after interventions. Finally, WHOQOL scores could be compared between patients with different forms of MD. Nevertheless, being the first study in the literature to investigate QOL and socio-demographic features of MD patients indicates the strength of our research. In this respect, we think that our research will be guiding for further comprehensive studies.

Ethics approval

The ethics committee approval for this cross-sectional study was obtained from University of Health Sciences Gazi Yasargil Training and Research Hospital (date: 05.03.2021 number: 688).

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