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ORIGINAL ARTICLE

Functional level and its relationship to upper extremity function, pain, and muscle stiffness in children with Duchenne muscular dystrophy

Dilan Savaş[1] · **Tülay Tarsuslu Şimşek**[2]

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Abstract

Background The progressive symptoms of Duchenne muscular dystrophy (DMD) negatively affect upper extremity skills, and this may have an effect that reduces the independence of daily life.

Aims The purpose of this study is to investigate the relationship between functional level and upper extremity function, pain, and stiffness in children with DMD.

Methods A total of 38 children with DMD were participated. The functional level of the upper and lower extremities was assessed using Brooke scale and Vignos scale. Upper extremity function, pain and stiffness were assessed using Upper Limb Short Questionnaire (ULSQ). The correlation between ULSQ and Brooke and Vignos scales was calculated.

Results A moderate positive correlation was calculated between ULSQ total scores and Vignos scale ($r=0.52$, $p<0.001$) and Brooke scale ($r=0.65$, $p<0.001$). There was a moderate positive correlation between Vignos scale scores and ULSQ subscores of function ($r=0.42$, $p<0.05$) and stiffness ($r=0.56$, $p<0.001$); no significant correlation was found between pain scores and Vignos scale ($p=0.053$). There was a moderate positive correlation between the function ($r=0.54$, $p<0.001$), pain ($r=0.40$, $p<0.05$), and stiffness ($r=0.62$, $p<0.001$) subscores of the ULSQ with the Brooke scale.

Conclusion In our study, there was a significant relationship between the functional level of patients with DMD and upper extremity function, pain, and muscle stiffness.

Keywords Duchenne muscular dystrophy · Function · Functional level · Pain · Stiffness · Upper extremity

Introduction

Muscular dystrophies are a heterogeneous group of diseases defined by dystrophic pathological changes in muscle biopsy. Clinically, they are characterized by progressive muscle weakness affecting skeletal muscles, but due to significant differences in genetic and biochemical characteristics, the distribution of the affected muscles, the degree of respiratory and cardiac involvement, and the involvement of other organ systems differ [1].

Duchenne muscular dystrophy (DMD) is a disease associated with muscle degeneration and necrosis [2]. It is the most common hereditary muscle disease in childhood and occurs in about 8.3 out of 100,000 males [3]. DMD is caused by mutations in the gene encoding the dystrophin protein in Xp21, which is responsible for stabilizing the sarcolemma during muscle contraction [4, 5]. DMD is characterized by complete or partial deficiency of the dystrophin cell membrane protein [6, 7]. The absence of dystrophin causes instability and decreased protein levels, leading to progressive fibril and membrane damage [8]. Patients with DMD become wheelchair dependent around the age of 12 on average and usually die in their 20 s due to respiratory failure [9]. Currently, there is no definitive treatment for DMD. However, an increase in the life expectancy of patients can be achieved thanks to treatments such as corticosteroids that slow down the progression of the disease and supportive treatments [10]. Treatment in DMD is a multidisciplinary (medical, surgical, and rehabilitative) approach according to patients' symptoms. With the progression of the disease, patients with DMD continue with impaired extremity functions for the rest of their lives [11]. In a study examining

✉ Dilan Savaş
savas.dilan1@gmail.com

[1] Institute of Health Sciences, Dokuz Eylul University, Mithatpasa Street No:1606, TR-35340 Balcova, Izmir, Turkey

[2] Faculty of Physical Therapy and Rehabilitation, Dokuz Eylul University, Izmir, Turkey

the existing literature on scales assessing upper extremity function in DMD, it was seen that each of the scales used provided useful information, but none of them were completely ideal. The difficulty of defining "perfectly fit" across scales suggests that they were not specifically designed for DMD. It is therefore thought that the spectrum of activities assessed, and the scoring system may not reflect the abilities or difficulties associated with DMD-specific differences, weakness, and progression of contractures [12]. Compensation for limitations in the upper extremities may be more difficult in patients with DMD. This may result in a restriction in the functional independence of patients [13], activity limitation, and a decrease in quality of life [14]. Loss of upper extremity functional skills has been reported due to muscle pain and muscle stiffness symptoms in the vast majority of patients [11].

In the literature, it is stated that patients with DMD have weakness experiences in their upper extremities starting from their ambulatory period, and this muscle weakness progresses from proximal to distal parts of the extremities. A few years after the loss of ambulation, upper extremity movements that can be performed against gravity are generally limited to forearm and hand functions, and severe losses are experienced in shoulder abduction and extension. With the progression of muscle weakness, the movements that can be performed in the upper extremity become limited to the hand, wrist, and finally the fingers [12]. In non-ambulatory patients with DMD, assessing the upper extremity becomes more difficult due to muscle weakness, contractures, and compensation strategies [15]. Progressive weakness in upper extremity functions causes compensations. In such cases, when the patients want to touch their face, they use some compensation strategies by flexing their head and trunk or by making climbing movements over the other arm with their fingers. Compensations performed by the patients to do any movement or activity may not provide information about the existing muscle weakness, but the important problem is the difficulties that the patients have in using the upper extremity [16].

Upper extremity skills in patients with DMD are essential for self-care and many other daily activities in daily life, and also have a major role in mobility and ambulation skills. In the future, when muscle weakness progresses and affects ambulation, good upper extremity function becomes important for maintaining ambulation and mobility of the patient. The patient, who can maintain mobility and ambulation, can participate in daily life. Thus, with a good upper extremity function, important contributions can be made to the quality of life and life satisfaction of the patient who remains functional in daily life. However, the progressive symptoms of the disease may also negatively affect upper extremity skills at certain periods of life, and this may have an effect that reduces the independence of daily life. The aim of our study, which was planned based on all this information, is to examine the relationship between functional level and upper extremity function, pain and muscle stiffness in children with DMD.

Material and methods

Study design and participants

This study was an observational study. A total of 38 children with DMD between the ages of 5–18 were participated. The sample size was calculated as a minimum of 34 participants by G* Power by taking 90% power, $\alpha = 0.05$, correlation pH1 = 0.68, and correlation pH0 = 0.3 [17]. Inclusion criteria were being diagnosed with DMD, being between the ages of 5–18, not having any additional neurological disorder, volunteering to participate in the research. Exclusion criteria were the children diagnosed after 10 years of age, children who had never taken corticosteroids, and children aged 14 years or older but were still ambulatory. The questionnaires were filled in by patients themselves or by their parents/legal guardians. All participants and their families were informed, and their given informed consent was taken before the participation. The Non-Invasive Research Ethics Board of Dokuz Eylul University approved the study in conformity with the Declaration of Helsinki principles (ref no: 2019/07–65).

Questionnaires and scales

Socio-demographic information registration form

It was created by the researchers after a literature review. In the form, the patient's age, height, body weight, and age of diagnosis were questioned.

Brooke upper extremity functional classification

The Brooke Upper Extremity Functional Classification, developed by Brooke et al. is used to evaluate the upper extremity functions in patients with DMD. Depending on what activities the affected person can perform with the upper extremities, a rating between 1 and 6 is given. Level 1 means the patient can start movement with their arms at their sides and bring their hands fully together above their head, level 6 means they cannot bring their hands to their mouth and cannot use their hands functionally [18].

The upper limb short questionnaire

The ULSQ is a 14-item questionnaire developed to evaluate upper extremity function, muscle pain, and muscle stiffness

in patients diagnosed with DMD. This questionnaire contains 5 items to assess upper extremity function, 6 items to assess pain, and 4 items to assess muscle stiffness. The questions are evaluated with 0 or 1 points according to the answer given. The questionnaire can be answered by the patients or the caregiver. Lower scores mean fewer problems in the upper extremities [13]. The ULSQ takes approximately 10–15 min to complete.

Vignos scale

It is a scale used to evaluate lower extremity functions in patients with neuromuscular disease. It has 10 functional levels according to the ambulation ability of patients. At the first level, the patient can walk and climb the steps without assistance, but at the last level, patient is confined to bed [19].

Data analysis

Statistical analysis was performed using the Statistical Package for Social Sciences (SPSS®) Windows 22.0 program. The Shapiro–Wilk test was applied to test the normality assumption. Correlation between the ULSQ with the Brooke scale and Vignos scale were calculated to determine relationship with children's functional level and upper extremity function, pain, and muscle stiffness**.** The median, interquartile range, mean, and standard deviation values of the ULSQ, Brooke, and Vignos scale levels and scores were calculated in the evaluation of the upper extremity and functional levels of patients with DMD. Pearson's correlation test was used for correlation, *p*-values < 0.05 were considered statistically significant. The Pearson's *r* correlation coefficients for all data were interpreted as 0.00–0.10 to be a negligible correlation, 0.10–0.39 to be a weak correlation, 0.40–0.69 to be a moderate correlation, 0.70–0.89 to be a strong correlation, and 0.90–1.00 to be a very strong correlation [20].

Results

The socio-demographic and functional level findings

A total of 38 children with a mean age of 10.58 ± 3.99 years were included in the study. The mean age of diagnosis was calculated as 4.06 ± 2.33 years (Table 1). According to the findings from the Vignos scale, 28.9% of children were at grade 9 which means using a wheelchair, while 5.3% can walk and climb stairs without assistance (Table 1). According to upper extremity functions findings, 47.4% of the children were at grade 1 in Brooke scale and could perform a full circle abduction movement while placing their hands on the head. On the other hand, 5.3% of the children were at level 5 and could use their hands to hold a pen or pick up coins from the table, even if they could not bring their hands to their mouths (Table 1). The mean ULSQ score of the children was calculated as 5.97 ± 3.06 out of a maximum of 14 points (Table 1). According pain scores of the ULSQ, 63.4% of the children had shoulder pain. The socio-demographic and functional level findings of children are shown in Table 1.

Factors associated with upper extremity function

There was a moderate positive correlation between the age of children and function ($r = 0.45$, $p < 0.05$) and stiffness scores of the ULSQ ($r = 0.46$, $p < 0.05$). No significant relationship was found between pain scores and age ($p = 0.08$) (Table 2). There was a moderate positive correlation between the duration of wheelchair usage and stiffness scores ($r = 0.54$, $p < 0.001$) and the total scores of the ULSQ ($r = 0.42$, $p < 0.05$). There was no significant correlation between the age of diagnosis and the subscores and total score of the ULSQ ($p > 0.05$) (Table 2).

The relationship between functional level and upper extremity function, pain and muscle stiffness

In the correlation analysis that was used to calculate the relationship between upper extremity function, pain, muscle stiffness, and functional level of children, there was a moderate positive correlation between the total score of ULSQ and the Brooke scale levels ($r = 0.65$, $p < 0.001$). A moderate positive correlation was calculated between the sum scores of ULSQ, function ($r = 0.53$, $p < 0.001$), stiffness ($r = 0.62$, $p < 0.001$) and pain ($r = 0.40$, $p < 0.05$), and Brooke scale levels (Table 3). There was a moderate positive correlation between children's ULSQ total score and Vignos scale levels ($r = 0.53$, $p < 0.001$). While there was a moderate positive correlation between Vignos scale levels and the function ($r = 0.41$, $p < 0.05$) and stiffness ($r = 0.58$, $p < 0.001$) scores of the ULSQ, no significant correlation was found between pain and Vignos scale levels ($p > 0.05$) (Table 3).

Discussion

The results of our study support our hypothesis and showed that there is a relationship between functional level and upper extremity function, pain, and muscle stiffness in children with DMD.

Good upper extremity function requires good proximal muscle control. When the shoulder joint is stabilized, an increase in upper extremity performance and hand functions is observed [21]. As a result of the studies of Janssen et al. they showed that patients with DMD continue with impaired

Table 1 Socio-demographic and functional level findings

	Median (IQR 25/75)	Min–max	Mean ± std. deviation	Percent/number
Age (y)	9.5 (8.00/13.00)	5–18	10.58 ± 3.99	*n* = 38
Length (cm)	125.5 (114.00/152.50)	90–180	133.07 ± 25.14	*n* = 38
Weigth (kg)	30 (22.00/49.75)	16–80	36.57 ± 18.63	*n* = 38
Age of diagnosed (y)	4 (1.75/6.50)	1–8	3.06 ± 2.33	*n* = 38
Brooke scale				
Grade 1	-	-	-	47.4 (*n* = 18)
Grade 2	-	-	-	18.4 (*n* = 7)
Grade 3	-	-	-	23.7 (*n* = 9)
Grade 4	-	-	-	5.3 (*n* = 2)
Grade 5	-	-	-	5.3 (*n* = 2)
Grade 6	-	-	-	-
Vignos scale				
Grade 1	-	-	-	5.3 (*n* = 2)
Grade 2	-	-	-	13.2 (*n* = 5)
Grade 3	-	-	-	31.6 (*n* = 12)
Grade 4	-	-	-	7.9 (*n* = 3)
Grade 5	-	-	-	7.9 (*n* = 3)
Grade 6	-	-	-	5.3 (*n* = 2)
Grade 7	-	-	-	-
Grade 8	-	-	-	-
Grade 9	-	-	-	28.9 (*n* = 11)
Grade 10	-	-	-	-
Subscores of ULSQ				
Function	3.00 (3.00/2.00)	0–5	2.50 ± 1.19	*n* = 38
Pain	3.00 (1.00/3.00)	0–6	2.32 ± 1.61	*n* = 38
Stifness	.00 (.00/3.00)	0–3	1.18 ± 1.33	*n* = 38
ULSQ total score	6.00 (3.00/9.00)	0–12	5.97 ± 3.06	*n* = 38

y year, *cm* centimeter, *kg* kilogram, *ULSQ* Upper Limb Short Questionnaire, *IQR* the interquartile range, *Min* minimum, *Max* maximum

extremity functions for the rest of their lives [11]. In DMD, which is a progressive disease, the increase in symptoms with age causes limitations in functionality [22]. As patients with DMD get older, they become more dependent on activities of daily living. This is seen as an important factor showing that aging and loss of muscle strength are associated with restrictions in activities of daily living [23]. Following the literature, the results of our study show that the increase in the age of the patients affects the upper extremity functions. It is thought that the increase in functional

Table 2 Relationship between descriptive and health status results with ULSQ

Subscores of ULSQ	Variables					
	Age (y)		Use of wheelchair (y)		Age of diagnosed (y)	
Function	*r*	0.45	*r*	0.33	*r*	0.18
	p	< 0.05*	*p*	< 0.05*	*p*	0.26
Pain	*r*	0.28	*r*	0.18	*r*	−0.75
	p	0.08	*p*	0.26	*p*	0.65
Stifness	*r*	0.46	*r*	0.54	*r*	−0.09
	p	< 0.05*	*p*	< 0.001*	*p*	0.95
ULSQ total score	*r*	0.49	*r*	0.42	*r*	−0.006
	p	< 0.05*	*p*	< 0.05*	*p*	0.97

ULSQ Upper Limb Short Questionnaire, *r* Pearson correlation coefficient

*$p < 0.05$

Table 3 The relationship between functional levels with ULSQ

Subscores of ULSQ	Brooke scale		Vignos scale	
Function	*r*	0.53	*r*	0.41
	p	< 0.001*	*p*	< 0.05*
Pain	*r*	0.40	*r*	0.30
	p	< 0.05*	*p*	0.06
Stifness	*r*	0.62	*r*	0.58
	p	< 0.001*	*p*	< 0.001*
ULSQ total score	*r*	0.65	*r*	0.53
	p	< 0.001*	*p*	< 0.05*

ULSQ Upper Limb Short Questionnaire, *r* Pearson correlation coefficient

*$p < 0.05$

problems that occur with age is due to disease-specific symptoms, especially muscle weakness that starts in the proximal muscles and progresses to the distal due to the progression of the disease, and problems in motor control as a result of the loss of stabilization.

A moderate positive correlation was found between Brooke and Vignos scale and extremity functions. This shows that the effects on the upper extremity functions in relation to the functional levels of both the lower and upper extremities. The results of our study showed that the change in ambulation abilities of children have a relationship to their upper extremity functions. Studies have shown that patients with non-ambulatory DMD have a significantly need for more support in their educational life compared to those with ambulatory [24]. The problems experienced by patients with DMD in using their extremities functionally cause functional limitations in daily living activities or accessing problems in school life [25]. According to the results of our study, the decrease in upper extremity functionality with the increase in Vignos levels show that patients may experience limitations in their upper extremity functions during daily living activities with the progression of the disease. The increase in both the Vignos levels and the Brooke levels indicates that patients experience more loss of function in their upper extremities in the later stages of the disease. In the literature, it has been stated that there is a strong relationship between postural control and fine motor skills in children aged 3–11, and it has been reported that the increase in postural control increases fine motor skills [26]. So that, a significant relationship was found between wheelchair use and upper extremity functions in our study. It is thought that this result is due to the loss of ambulation that occurs with the progression of the disease, causing weakness in postural control and consequently impaired upper extremity functions.

Pangalila et al. reported the similar rate of pain in the shoulder and arm in a study they conducted with patients with DMD [27]. Several studies showed that shoulder pain is more common in patients with DMD than pain in other parts of the upper extremity [28, 29]. Janssen et al. reported that pain is most common in the shoulder, followed by the elbow, proximal of the arm (upper and lower part of the arm), and distal of the arm (wrist and fingers) [13]. The results of our study were found to be consistent with the literature, as pain is commonly seen in the shoulder. The frequency of pain continues in the form of pain in the lower/upper parts of the arms and pain in the extremity, respectively. It has been thought that the prevalence of pain in the proximal and larger muscle groups may be due to the earlier weakening of the muscle groups that are used more and occupy a large volume due to the course of the disease, the inability to fulfill their stabilization task, and the more common fatigue symptoms. Pain and muscle weakness are important symptoms that affect each other [11]. In the literature, it has been stated that many problems in the musculoskeletal system cause pain in neurological diseases. It was found that long-term spasticity in CP was associated with the development of contractures. In the same study, it was stated that problems such as focal spasticity or dystonia may cause focal muscle spasms [30]. This information obtained from the literature shows that the problems affecting the musculoskeletal system, characterized by long-term fixed posture or muscle weakness, are matched with the signs of pain and muscle spasms in the later stages. In our study, a moderate positive correlation was observed between the upper extremity functional level and the symptom of pain. The results obtained from our study showed that as the functional level of the upper extremity worsened in children with DMD, symptoms of pain may be more common. With the progression of the disease, it is thought that the increase in DMD-specific symptoms such as muscle weakness, muscle stiffness, and fatigue will cause patients to avoid activity, and with the effect of this situation, contractures that may occur in the later stages of the disease will reduce functional skills and increase pain.

It has been stated in the literature that the increase in pain, which will limit functions, affects activities of daily living [30]. The findings of our study showed the importance of physiotherapy and rehabilitation practices aimed at protecting and increasing upper extremity skills in patients with DMD as a result of the decrease in their upper extremity skills in the later stages of the disease. In our study, there was no significant relationship between Vignos scale and pain. Since the Vignos scale determines the ambulation problems, it was thought that the children included in the study did not have serious ambulation problems yet may have affected this result. Studies have frequently mentioned the importance of the upper extremity in terms of maintaining ambulation and participation in daily life in patients with DMD. However, it was observed that the number of studies investigating the factors affecting the upper extremity skills was insufficient, and the findings and effects of pain were not

adequately studied. It is thought that there is a need for more detailed studies on the stages of the disease, upper extremity skills, and the findings of pain in patients with DMD.

In the literature, it has been reported that muscle stiffness increases with the stage of the disease in patients with DMD [31]. In the results of our study, muscle stiffness increased with age. Considering that DMD is a progressive disease, this increase in muscle stiffness with age can be associated with the stage of the disease in line with the literature. Jannsen et al. reported that limitations and complaints caused by muscle stiffness negatively affect upper extremity function [32]. It was observed that the children included in our study had not severe signs of muscle stiffness. It was interpreted that the low problems experienced by the children in the finding of muscle stiffness did not cause a significant regression or limitation in their hand functions. There is a need for studies that evaluate the functions of the upper extremity in patients with signs of muscle stiffness, and studies that will examine the relationship between the functional level of the upper extremity and stiffness in detail. Another important finding of our study is the good positive correlation between Brooke scale level and muscle stiffness. Our results showed that patients with good upper extremity function also have less muscle stiffness; stiffness signs may be associated with disease progression. With the progression of the disease, functional level regression and limitations occur, and in advanced stages, muscle stiffness occurs with the increase of soft tissue contractures and muscle weakness. Along with muscle weakness and soft tissue contractures, upper extremity skill limitations of patients can also be seen. When the literature is examined, it is seen that complaints of pain and stiffness have a negative effect on general functional skills [33]. In the results of our study, in parallel with the study findings of Janssen et al. [11, 32], it was interpreted that the stronger relationship between stiffness and upper extremity functional level than pain might be due to the fact that the finding of muscle stiffness in DMD patients was seen before the finding of muscle weakness and fiber destruction, and pain. A moderate correlation was observed between the Vignos scale and the sign of stiffness. This situation can be explained by the increase in stiffness in the upper extremity due to progression, along with the change in the ambulation levels of the patients in the advanced stages of the disease, which is consistent with the literature.

According to the results of our study, a good correlation was observed between the ULSQ total score and Brooke scale. Similarly, there was a moderate relationship between Vignos scale values and the ULSQ. Although upper extremity limitation is experienced as muscle weakness progresses from proximal to distal in patients with DMD, ambulation continues and the patient prefers to continue ambulation until the muscle weakness worsens. As a result of our study, these limitations determined at functional levels show that patients may encounter problems in performing self-care activities, in activities that require participation at school, and in any situation that requires functional use of their upper extremities. Our results also remind us of the importance of proximal region rehabilitation in the upper extremity in terms of continuation of upper extremity use in patients with DMD, preventing muscle weakness from the early period and reducing pain.

There are some limitations of our study. When the relationship between the age at diagnosis of the patients included in our study and upper extremity function, pain, and muscle stiffness was examined, it was seen that there was no significant relationship between the subscores of the ULSQ and the total score. It was thought that this situation was caused by the fact that most of the patients were diagnosed at an early stage and started treatment and thus were not at a good level in terms of functionality.

Conclusion

In this study, a significant correlation was found between functional levels of patients with DMD and upper extremity function, muscle pain, and muscle stiffness. Identifying the problems experienced by children and deciding on interventions will have a positive impact on children's quality of life. In the literature, it is stated that it is very important to evaluate upper extremity functions in patients with DMD, to observe functional changes in different stages of the disease, to determine the progression of the disease, to develop the content of the treatment program that can be applied, and to support the patient in realizing functional independence in their daily life [18, 23, 34, 35]. For these reasons, we think that facilitating the clinical evaluation of pain, muscle stiffness, and dysfunction, which are common symptoms of DMD, is important because it is a progressive disease.

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Declarations

Conflict of interest The authors declare no competing interests.

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