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THE EFFECT OF ANKLE LIMITATION ON DYNAMIC BALANCE AND FUNCTIONAL CAPACITY IN CHILDREN WITH DUCHENNE MUSCULAR DYSTROPHY

Duchenne Musküler Distrofili Çocuklarda Ayak Bileği Limitasyonunun Dinamik Denge ve Fonksiyonel Kapasite Üzerine Etkisi

Halil ALKAN[1] *Mahsum EREN*[2]
[1]*Muş Alparslan University, Faculty of Health Sciences, Muş*
[2]*Beykent University, Vocational School, İstanbul*

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ABSTRACT

The aim of this study is to investigate the effect of ankle limitation on dynamic balance and functional capacity in children with Duchenne Muscular Dystrophy (DMD). This cross-sectional study included thirty-six 6- to 11-year-old children with DMD who were followed up in the special education rehabilitation centers. In addition to recording of the demographic data of the children, ankle limitations were recorded in degrees using a goniometer. Dynamic balance and functional capacity of the patients were evaluated using the Timed Up and Go (TUG) test and 6-Minute Walk Test (6MWT), respectively. Ankle limitation had a statistically significant effect on dynamic balance and functional capacity of children with DMD ($p = 0.008$ and $p = 0.006$, respectively). In children with DMD, dynamic balance and functional capacity deteriorate as their ankle limitation increases. Treatment methods applied to increase/maintain joint range of motion in children with DMD should be added to the rehabilitation programs as early as possible.

Keywords: Ankle limitation, Duchenne muscular dystrophy, Dynamic balance, Functional capacity.

ÖZ

Bu çalışmanın amacı DMD’li çocuklarda ayak bileği limitasyonunun dinamik denge ve fonksiyonel kapasite üzerine etkisini araştırmaktır. Bu kesitsel çalışma, özel eğitim rehabilitasyon merkezlerinde izlenen 6-11 yaşlarında DMD'li otuz altı çocuğu içermektedir. Çocukların demografik verilerinin kaydedilmesine ek olarak ayak bileği limitasyonları gonyometre kullanılarak derece cinsinden kaydedildi. Hastaların dinamik dengesi ve fonksiyonel kapasitesi sırasıyla Süreli Kalk ve Yürü (SKYT) testi ve 6 Dakika Yürüme Testi (6DYT) ile değerlendirildi. Ayak bileği kısıtlılığının DMD'li çocukların dinamik dengesi ve fonksiyonel kapasitesi üzerinde istatistiksel olarak anlamlı bir etkisi vardı (sırasıyla $p= 0.008$ ve $p= 0.006$). DMD’li çocukların ayak bileği limitasyonu arttıkça dinamik dengeleri ve fonksiyonel kapasiteleri azalmaktadır. DMD'li çocuklarda eklem hareket açıklığını artırmak/sürdürmek için uygulanan tedavi yöntemleri biran önce rehabilitasyon programlarına eklenmelidir.

Anahtar kelimeler: Ayak bileği limitasyonu, Dinamik denge, Duchenne musküler distrofi, Fonksiyonel kapasite.

Halil ALKAN 🖂*, mailto:fzthalilalkan@hotmail.com*
Muş Alparslan University, Faculty of Health Sciences, Muş

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INTRODUCTION

Duchenne muscular dystrophy (DMD) is the most common type of progressive muscular dystrophies seen in 1/3500 live male births. Progressive and irreversible muscle weakness occurs as a result of deletion, duplication, or point mutation in the Xp21 gene, which encodes dystrophin in the short arm of the X chromosome (Akima et al., 2012; Bushby et al., 2010a; Goemans et al., 2013). The absence of dystrophin makes muscle fibers more susceptible to damage, necrosis and degeneration, causing them to lose function and be replaced by non-contractile connective and adipose tissue (Akima et al., 2012). Together with functional muscle tissue loss; joint deformities, cardiopulmonary problems, increased body mass (obesity and osteoporosis in long term), decreased locomotion, decreased physical activity and exercise capacity, anxiety and depression are observed in the patients (McDonald, 2002; Olle, Pivarnik, Klish, & Morrow Jr, 1993). Due to these multiple problems, the main goals in DMD include protecting the locomotion system to slow the progression of the disease and keeping vital functions at the best possible level. Hence, spinal deformities, muscle contractures, joint limitations, respiratory and cardiac problems are accelerated in the child with DMD who loses ambulation in the early period (Karaduman, Yılmaz, & Alemdaroglu, 2014).

In muscular dystrophies, after muscle weakness, joint deformity is the second most important clinical feature that adversely affects the locomotor system (McDonald et al., 1995). In particular, deformities in lower limb joints disrupt standing balance and accelerate the loss of ability to walk (Rose, Burns, Wheeler, & North, 2010). This is also shown to restrict mobility and balance in Charcot-Marie-Tooth, a neuromuscular disease (Burns, Ryan, & Ouvrier, 2009). Due to a number of active and passive neurophysiological mechanisms, equinus occurs in the ankle joint together with limitation in dorsiflexion (Rose et al., 2010). Early loss of ambulation is the most important complication that develops secondary to ankle limitation in DMD (Farmer, Pearce, Whittall, Quinlivan, & Patrick, 2006; Kaya, Alemdaroğlu, Yılmaz, Karaduman, & Topaloğlu, 2014).

Similar to other neuromuscular diseases, in DMD, ankle joint limitation together with muscle weakness disturbs balance and leads to stumbling and falling (Kaya et al., 2014). Furthermore, decreased safety of ambulation leads to a decrease in the activity levels of the patients causing social isolation; it also brings along undesirable effects on bone mineral density, physical fitness, and cardiorespiratory functions (Kaya et al., 2014).

To the best of our knowledge, there is no study in the literature investigating the effect of ankle limitation on dynamic balance in children with DMD; and there is only a limited number of studies investigating its' effect on functional capacity. Therefore, the purpose of the study was to investigate whether:

i. Ankle limitation had an impact on dynamic balance in children with DMD.
ii. Ankle limitation had an impact on functional capacity in children with DMD.

MATERIAL AND METHOD

The cross-sectional study was carried out in special education and rehabilitation centers located in Istanbul city center. To determine the sample size of the study, regression analysis was based. According to this analysis, it is suggested that the number of samples should be at least 10-15 times the number of independent variables to be included in the regression model (Field, 2005).

Participants

The study group consisted of 36 children with DMD between the ages of 6-11 who were followed up in the various special education rehabilitation centers. The participants were the children with DMD who were; already prescribed a home-based program, regularly followed up, on steroid therapy, and using night orthosis. The inclusion criteria were set as: being diagnosed with DMD, being in the age range of 6-11 years, having Level 1 or 2 on the Vignos Lower Extremity Functional Scale (Vignos, Spencer, & Archibald, 1963), and the absence of additional medical conditions (such as cardiac risk, congenital heart defect, intake of medication affecting heart rate or metabolism). Children who did not meet the inclusion criteria were excluded from the study.

Outcome Measures

Name and surname, demographic data and physical characteristics such as height, body weight and body mass index (BMI) of the children with DMD were recorded. Objective performance tests were carried out in one session for each child by an expert (Physiotherapist M.E.), who had 7 years of experience in pediatric rehabilitation. Each child was evaluated respectively within a period of approximately 30 minutes. In order to prevent fatigue during the assessments, the children with DMD were allowed to rest for 2-3 minutes after each assessment test. Then, when the child felt well, the next assessment was carried out.

Functional Level

Lower extremity functional levels of the participants were evaluated using the Vignos Lower Extremity Functional Scale (Vignos et al., 1963). Developed by Vignos, this scale has a score range of 1-10, in which level 1 represents the best functional state and level 10 stands for the worst level of functionality. The Vignos Lower Extremity Functional Scale is a standard method for determining the functional level of the lower extremity in the clinical evaluation of children with DMD (Vignos et al., 1963). The children with DMD in Level 1 and 2 of the Vignos scale were included in the study (Level 1: walks and climbs stairs without assistance; Level 2: walks and climbs stairs with aid of handrail in less than 12 seconds).

Ankle Limitation Assessment

Using a universal goniometer, ankle dorsiflexion limitations of the participants were measured bilaterally and recorded in degrees. The universal goniometer was reported excellent for intratester intersessional intraclass correlation coefficient values for all measurements obtained among subjects with DMD (Pandya et al., 1985). With the child in the supine position, the lateral malleol was used as the pivot point, and the stable arm of the goniometer was parallel to the midline of fibula. The mobile arm was aligned to the lateral midline of the 5^{th} metatarsal bone. Since muscle weakness hinders patients to actively perform the movement, passive range of motion was recorded for the children with muscle weakness. The ankle limitations of the children with DMD were calculated by subtracting from the passively measured ankle ROM degree from the optimal ROM that should be. Ankle limitations in children were recorded for both feet, right and left (Bach & Lieberman, 1993; Kaya et al., 2014). For each participant, the mean of left and right ankle limitation was calculated and used in the statistical analysis.

Functional Capacity

Functional capacity of the participants was evaluated with 6-Minute Walk Test (6MWT). 6MWT is a measure of quality of life as it reflects submaximal walking performance and functional capacity (McDonald et al., 2013). This valid and reliable test is recommended as a primary outcome measure in DMD for therapeutic treatments and for evaluating the course of the disorder (McDonald et al., 2010).

The test was performed indoors, along a flat, straight, enclosed corridor with a hard surface. The walking course was 25m in length and a cone was placed at the turnaround point. To signal the direction of the turn, the floor was marked with counterclockwise arrows using a brightly colored tape. A stopwatch was used to measure walking time. The children were asked to walk at their best speed but not to run, hop, or jump. For each participant, the walking distance was recorded in meters. During the test, the children were given verbal encouragements to motivate them complete the test (Laboratories, 2002; McDonald et al., 2010).

Balance Assessment

To evaluate the balance of the children, Timed Up and Go (TUG) test was used. TUG is a valid, practical, and objective measure to evaluate functional mobility and dynamic balance in the pediatric population (Williams, Carroll, Reddihough, Phillips, & Galea, 2005). In our country, the mean TUG values of children with DMD and healthy peers according to the Vignos Lower Extremity Functional Scale have been previously reported (Alkan et al., 2017).

With the subject seated in a standard chair, he was asked to rise from the chair (without holding the armrests), walk for 3 meters, turn around, walk back to the chair, and sit down. For each child, the time was recorded in seconds. The test was performed three times with short rest intervals (one-two minutes) and the mean of the three values was used for the statistical analysis (B. Aras, O. Aras, & Karaduman, 2011).

Statistical Analysis

Statistical analyzes were made using “IBM® SPSS © 24 software”. Compliance of numerical variables to normal distribution was performed using visual (histogram and probability graphs) and analytical methods (Shapiro Wilk tests). Descriptive statistics of the numerical variables were expressed as means and standard deviations. Descriptive statistics of the categorical variables were expressed as numbers and percentages. Pearson correlation test was used for the relationships between numerical variables. According to the correlation coefficient, the degree of correlation was interpreted as low correlation between 0.05-0.4, moderate correlation between 0.4-0.7 and high correlation between 0.7-1.0 (Murat Hayran & Mutlu Hayran, 2011). Univariate regression analysis was used to study the effect of independent variable (ankle limitation) on the dependent variable (TUGT and MWT). For the regression analysis, assumptions such as linear relationship between variables, extreme values, multicollinearity, and normal distribution of estimation errors were taken into account. Statistical significance level was $p<0.05$.

Ethical Considerations

The ethics committee approval was obtained from Muş Alparslan University, Non-Interventional Clinical Research Ethics Committee with the decision number E.14279 and number 15-27 on 29.12.2020 to conduct the study. Informed consent was obtained from the families prior to prospective follow-up of the children.

RESULTS

Demographic data and clinical characteristics of the participants are presented in Table 1.

Table 1. Clinical Characteristics of Children with DMD

		DMD (n=36)	Min-Max
		Mean ± SD	
Age (months)		95.14 ± 13.87	72-121
Height (cm)		120.14 ± 9.8	106-136
Weight (kg)		24.83 ± 4.91	17-38
BMI (kg/m^2)		17.14 ± 2.10	11-20.9
Ankle limitation (°)		19.46 ± 5.26	9.50-32.50
TUGT (sec)		7.07 ± 1.87	3.12-12.13
6-MWT (m)		370.15 ± 66.78	260-510
		n (%)	
Vignos Lower Extremity Functional Classification	Level 1	24 (66.7)	
	Level 2	12 (33.3)	

DMD; Duchenne Muscular Dystrophy, TUGT; Time up and go test, BMI; Body mass index, MWT; Minute walk test, SD; Standard deviation

When the relationship between the ankle limitation of the children included in the study and TUGT and 6MWT was examined, it was observed that there was a positive, moderate statistical relationship between the ankle limitation of the children with DMD and TUGT (TUGT, r = 0.435, p = 0.006). A moderate, negative correlation was found between the ankle limitation and 6MWT (6MWT, r = -0.451, p = 0.008).

The correlations between ankle limitation, TUGT and 6-MWT in children with DMD are given in Figure 1 and 2.

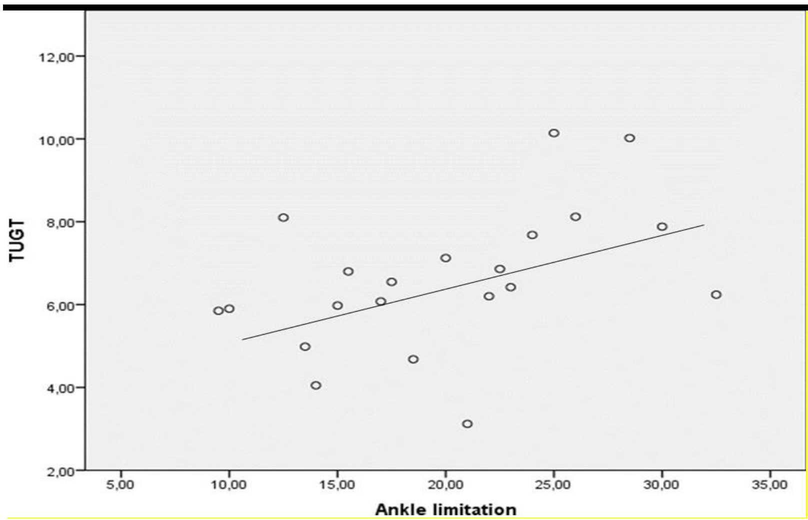

Figure 1. Correlations Between Ankle Limitation and TUGT in Children with DMD (N = 36), DMD; Duchenne Muscular Dystrophy, TUGT; Time Up and Go Test, ($y = 0.435x + 3.905$, $R^2 = 18.9$, $p < 0.01$).

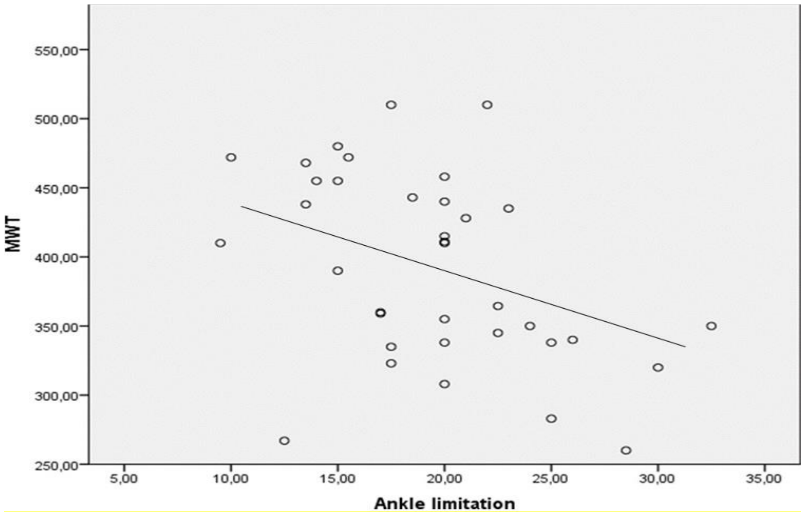

Figure 2. Correlations Between Ankle Limitation and 6-MWT in Children with DMD (N = 36), DMD; Duchenne Muscular Dystrophy, MWT; Minute Walk Test, ($y = - 0.451x + 505.056$, $R^2 = 20.3$, $p < 0.01$).

According to the results of the regression analysis, the explanatory effect of ankle limitation on dynamic balance and functional capacity in children with DMD was found to be statistically significant ($R^2 = 18.9$, $p = 0.008$ and $R^2 = 20.3$, $p = 0.006$, respectively). To put it more clearly, as the ankle limitation of children with DMD increases, their dynamic balance periods increase. That is, their dynamic balance decreases. Similarly, it was found that the functional capacity of the children with DMD decreased as the ankle limitation degree increased. The variances rate of the ankle limitation on the dynamic balance and functional capacity of children with DMD were also found to be 18.9% and 20.3%, respectively (Table 2).

Table 2. The Variable that Affect the Dynamic Balances (TUGT) and Functional Capacities (6MWT) of Children with DMD

	R^2	F	β	t	*p* value
Ankle limitation (°)	0.189	7.914			**<0.001**
			0.435	2.813	**0.008**
	0.203	8.685			**<0.001**
			-0.451	-2.947	**0.006**

DMD; Duchenne Muscular Dystrophy, TUGT; Time up and go test, MWT; Minute walk test

DISCUSSION

According to our study results, in children with DMD dynamic balance and functional capacity deteriorate as their ankle limitation degree increases.

In muscular dystrophies, evaluating balance and identifying the factors affecting it are important in shaping preventive and therapeutic approaches in rehabilitation practices. The importance of balance in children with DMD becomes more prominent over time due to disorder progression and children’s growth (Allsop & Tecklin, 1989). In children with DMD, ankle limitation together with muscle weakness causes the balance to deviate from normal reference points relative to the gravity line. As a result of these deviations, children develop postural adaptations to achieve/maintain their static balance (Baptista, Costa, Pizzato, Souza, & Mattiello-Sverzut, 2014). In a study conducted by Romano and his colleagues using 3D gait analysis in 20 DMD children, they showed that there is a relationship between ankle plantar/dorsiflexion angles and functional mobility (Romano et al., 2019). Similarly, Burns et al. reported that ankle limitation affected mobility and balance in children with Charcot-Marie-Tooth disease (Burns et al., 2009). Regarding other factors that affect balance in children with DMD, some studies reported a significant negative relationship between muscle weakness and balance (Kaya, Alemdaroğlu, Yılmaz, Karaduman, & Topaloğlu, 2015) and others stated that dynamic balance worsens as the functional level decreases (Alkan et al., 2017). Our study results also indicate that ankle limitation has a moderate impact on dynamic balance in children with DMD. In other words, in line with the findings of our current study, it can be said that as the ankle limitation of children with DMD increases, their dynamic balance decrease. At this point, it can be predicted that exercise, orthotics and surgical applications applied for ankle limitations (Ainslie, 2012; Bushby et al., 2010b) in terms of protecting/increasing the mobility and balance of children with DMD are also important in terms of treatment methods. Hence, in many studies in children with DMD, it is emphasized that the applications for the ankle (especially orthotics and surgery) are of great importance in terms of maintaining ambulation and delaying the onset of contracture in these children (Ainslie, 2012; Bushby et al., 2010b).

Since 6MWT perfectly reflects functional capacity in children with DMD (McDonald et al., 2013), it is recommended to be used as a primary outcome measure in therapeutic treatments and follow-up of the disease course (McDonald et al., 2010). Studies examining the relationship between ankle limitation and functional capacity in children with DMD are limited. Kaya (Kaya et al., 2014) and Akkurt (Akkurt, Gürbüz, Karaduman, & Yilmaz, 2019) used 6MWT to study the relationship between ankle limitation and walking function in children with DMD. Both studies reported significant relationships between ankle limitation and 6MWT. In another study comparing children with DMD with their healthy peers and using the biomechanical method, the researchers showed that there is a relationship between the sagittal ankle angles in the sagittal plane and the 6MWT (Romano et al., 2019). Similarly, Scott et al. (Scott, Hyde, Goddard, & Dubowitz, 1981) reported a significant relationship between ankle limitation and functional level in 59 children with DMD aged 4-12 years. Unlike the afore-mentioned studies, the present study reveals the cause-effect relationship, rather than the general relationship between ankle limitation and functional capacity in children with DMD. Thus, it clarifies this relationship and highlights the adverse effect of ankle limitation on functional capacity and gait. This emphasizes the necessity of surgical, orthotic and rehabilitation approaches to increase/maintain ankle range of motion in children with DMD in order to preserve their ambulation ability.

CONCLUSION

In children with DMD, dynamic balance and functional capacity deteriorate as their ankle limitation degree increases. This indicates that the treatment applications applied to increase/maintain joint range of motion in children with DMD should be added to the physiotherapy and rehabilitation programs in the earliest possible period.

Limitations

This study has some limitations that need to be addressed. Firstly, the use of goniometer, which is a subjective method due to impossibilities, to measure the ankle limitations of children with DMD instead of objective biomechanical methods. Secondly, since only the passive (but not active) ankle limitation of the children was measured, the effects of active and passive limitation could not be studied separately. The reason for this is that active range of motion cannot always be completed in children with DMD due to muscle weakness (Bach & Lieberman, 1993; Kaya et al., 2014). Third; Age (McDonald et al., 2010), functional level (Alkan et al., 2017), muscle strength (Kaya et al., 2015) known to be associated with functional tests in children with DMD could not be included in the regression model due to the small number of samples. Thus, together with ankle limitation of these factors, the inability to reveal the effect sizes separately by including multiple regression analysis is also a limitation. Therefore, there is a need for studies with sufficient samples to include these independent variables in the model. Fourth, the inability to reach all children with DMD registered in rehabilitation centers due to the pandemic process can be shown among the limitations of this study.

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