Research Article

Association Between Muscle Fascicle Length Changes And Functional Performance in Children With Duchenne Muscular Dystrophy A Cross-Sectional Observational Study

Dimiana Samir Fied [1]*, Amira Mohamed El-Tohamy[2], Marwa Waly Eldin Ali[3], Ahmed S. Awad[2]

[1] Department of Growth and Development Disorders in Children and Its Surgery, Faculty of Physical Therapy, Deraya University, Minya, Egypt.
[2] Department of Physical Therapy for Pediatrics, Faculty of Physical Therapy, Cairo University, Cairo, Egypt.
[3] Department of Pediatrics, Faculty of Medicine, Minya University, Minya, Egypt.
Corresponding author:
Dr. Dimiana Samir Fied Yanny
Department of Growth and Development Disorders in Children and Its Surgery Faculty of Physical Therapy, Deraya University, Minya, Egypt. , Email: demyana.samer@deraya.edu.eg

Abstract

Background: Muscle tissue gradually deteriorates in Duchenne Muscular Dystrophy (DMD), a hereditary genetic disorder that causes functional impairment and early mortality. Children with DMD typically develop pseudohypertrophy of the calf muscle and struggle to run, jump, or climb stairs. Their functional performance abilities are impacted by their frequent falls. The study objective was to look at the relations between functional performance and alterations in the gastrocnemius muscle's fascicle length in DMD children.

Methods: Twenty-six ambulant DMD children who were between the ages of six and twelve and did not have pulmonary or cardiac conditions participated in cross-sectional observational research. Fascicle length was measured using ultrasound imaging and motor function (MF) was assessed utilizing MFM-32 scale to gage these kids' functional performance.

Results: The study's findings demonstrated that fascicle length and MF ability had no significant relationship (P>0.05) (r=0.039; P=0.850). This insignificant relationship indicates that in children with DMD, changes in fascicle length are inconsistent with changes in MF measurements.

Conclusion: this study demonstrated that in ambulant children with DMD, changes in muscle fascicle length don't directly correspond to changes in functional performance.

Keywords: Duchenne Muscular Dystrophy; Fascicle Length and Functional performance.

Introduction:

Muscle tissue gradually deteriorates in Duchenne muscular dystrophy, a hereditary genetic condition that causes functional impairment and early mortality.[1] Increasing the extensor muscle weakness rather than the flexor weakness is common characteristic in DMD patients. This weakness is further proximal rather than distal and influences the lower limbs rather than the upper limbs. This muscle weakness reduces function performance [2]. Children with DMD typically have frequent falls as a result of calf muscle weakness, which makes it difficult for them to run, jump, or climb stairs [3]. Imaging evaluations and motor function (MF) tests are the main methods used in clinical evaluation of illness progression [4]. On ultrasonographic (US) examination, children with DMD showed decreased bone echogenicity and increased muscle echogenicity; in many cases, the usual fascial demarcation was also disrupted [5]. US can be used to diagnose patients with DMD. Additionally, the results of the US scan may indirectly represent the functional capacity of children with DMD [6].

With the benefits of ease of use and strong repeatability in clinical practice, US is regarded as a very dependable method for evaluating skeletal muscle. The use of this modality for muscle examination has been made easier by developments in portable US technology. Nevertheless, there are still a number of difficulties with using muscle US [7]. Muscle atrophy, intramuscular fibrosis, and fatty infiltration are all sonographically detectable neuromuscular disorders, and muscle US is a good method for evaluating these conditions [8]. Muscle US provides a strong specificity and sensitivity for neuromuscular disorders identification as reported in prospective studies. Current research suggests that several neuromuscular disorders may show with distinctive US characteristics, possibly assisting differential diagnosis, despite the lack of evidence from extensive studies [9]. During contraction, the architectural features of muscles can change significantly. Fascicle length (FL) reduced during static contraction of the knee extensors at all angles of the knee joint, with a more noticeable loss of around 30% when the knee was almost fully extended and only about 5% when it was flexed [10]. Gastrocnemius tightness can restrict ankle

dorsiflexion, particularly when the knee is maintained in full extension. Because peak dorsiflexion typically occurs just before the heeling-off, at a point when patient's knee is nearly fully extended, decreased gastrocnemius flexibility may hinder the normal forward progression of the tibia over the foot during mid-stance, thereby affecting the mechanics of gait [11].

The majority of the study's DMD children showed clinically noticeable plantar-flexion contractures, which are typified by a limited range of motion for ankle dorsiflexion. This restriction might be linked to shortening of non-contractile periarticular tissues and/or tightness of the gastrocnemius muscle. Furthermore, the occurrence of plantar-flexion contractures was further corroborated by the greater stiffness coefficient seen in children with DMD [12]. A validated assessment instrument designed to evaluate MF in people with neuromuscular illnesses at varying degrees of disease severity is the MFM. It has shown strong repeatability, construct validity, and concurrent validity and is appropriate for ambulatory people and those who are not [13]. The **MFM-20/32** acts as the main outcome measure because it evaluates both fine and gross motor skills and is generally accepted as a valid and trustworthy method for assessing the severity of the disease and tracking its course in people with neuromuscular diseases [14]. The relationship between FL and overall MF may not be straightforward. Investigating this association may help clarify whether FL obtained by US has a meaningful relationship with functional performance and whether it can provide clinically relevant information during the assessment and follow-up of ambulant DMD children.

Timeline of the Study:

The study was conducted between September 2024 and October 2025. FL was measured using US [15], DMD Children have their functional performance evaluated using the MFM-32. [16], The functional level was measured using the Vignos scale (VS) [17].

Research Question:

Does functional performance in DMD children correlate with changes in gastrocnemius fascicle length?

Hypotheses:

Function performance and FL in DMD children would not be related.

Methods:

Participants:

A cross-sectional study was conducted among boys diagnosed with DMD. The study population comprised 26 boys between 6 and 12 years of age who were receiving follow-up care at the Pediatrics Clinic, Faculty of Physical Therapy, Deraya University, Minya, Egypt. Relevant clinical and medical information was obtained from diagnostic and laboratory investigations, together with data documented in the patients' electronic medical records.

Ethical approval was granted in 2024 by the Ethics Committee of the Faculty of Physical Therapy, Cairo University (Approval No. P.T.REC/012/005360). At enrollment, all participants demonstrated calf muscle pseudohypertrophy and retained the ability to ambulate independently. DMD diagnosis was confirmed based on established clinical and diagnostic evidence, including serum creatine kinase levels, characteristic clinical manifestations, genetic testing, and muscle biopsy findings. Children were not eligible for participation if they had severe scoliosis, significant cardiac or respiratory disorders, or pronounced contractures that could interfere with the assessment of functional capacity.

Data collection:

Sample Size estimation:

With a significance level of $\alpha = 0.05$ (two-sided), a power studying with 80% ($1-\beta = 0.80$), and an expected effect size of $|\rho| = 0.50$, this estimation indicated that **26 patients** were required to detect a significant association between FL and functional capacity in DMD boys.(https://g-power.apponic.com/)

Procedures

Fascicle length measuring:
The gastrocnemius FL variations in DMD children were measured using a 2D US equipment (mindray dp 10) with a frequency of 7.5 MHz. The gastrocnemius FL is measured when the cases are lying prone on the plinth with their knees extended and their feet outside the plinth. The distance between the fascicle's junctions with the superficial and deep aponeuroses was determined to be the FL.[18].

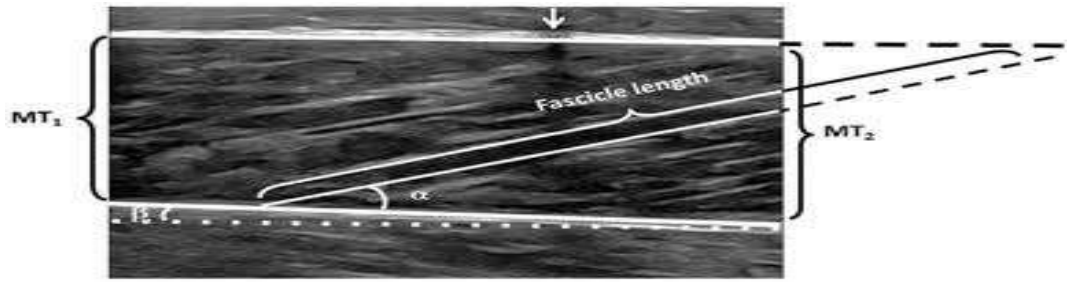

Fig.(1) Measurement of fascicle length

Functional level selection of children.
The VS, which has a strong intraclass correlation, was used to evaluate the functional level of DMD children. There are ten grades on the scale, with Grade 1 being the highest functioning level and Grade 10 the lowest. More precisely, children are classified as Grade 1 if they can walk and ascend stairs without assistance and as Grade 2 if they can do so with the use of a railing [17].

Motor function assessment MFM-32
The MFM is one quantitative instrument used to assess functional performance in neuromuscular disorders cases. It is used to assess the severity of an illness and track its development over time. The measure was created especially for use by rehabilitation physicians and physiotherapists in standard clinical settings [13]. The 32 items on the MFM are divided into three domains: D3 (distal MF), D2 (axial and proximal MF), and D1 (standing and transfers). Each item is scored on a four-point Likert scale ranging from 0 to 3. The scoring system was based on four levels of performance. A score of 0 was assigned when the participant was unable to begin the movement or sustain the required initial posture. A score of 1 denoted an attempt in which the movement was completed only partially. A score of 2 was given when the task could be performed but was accompanied by compensatory strategies, including slower execution, impaired motor control, or lack of coordination. A score of 3 indicated normal execution of the task, characterized by complete performance with purposeful and well-controlled movements performed at a steady pace [19].
The User's Manual, accessible in English, Spanish, and French contains the scoring criteria for each MFM item. Lower scores indicate more functional impairment. The overall score, which is given as a percentage of the highest possible score, is calculated by adding the scores from the 32 individual items. Additionally, the MFM offers three domain-specific subscores (D1, D2, and D3) that offer a more thorough evaluation of the severity and pattern of motor impairment. Physiotherapists with specialized expertise in MFM administration performed the evaluations. It took around 36 minutes to perform the entire evaluation [20].

Data Analysis:
All statistical procedures were conducted using SPSS software, version 25 (SPSS Inc., Chicago, IL, USA). Continuous variables, including age, VS, FL, and MF scores, were summarized using the mean and SD. The affected side was treated as a categorical variable and reported as frequencies and corresponding percentages. The relationship between FL and MF among DMD boys was examined using Pearson's correlation coefficient to determine both the magnitude and direction of the association.

RESULTS
This study comprised 26 boys with DMD, whose ages varied from 6.00 to 12.00 years (Table 1), with a mean age of

7.69 ±1.43 years. With a minimum of 2.00 and a high of 7.00, the VS mean value (Table 1) was 3.23 ± 1.68. For children with DMD, the afflicted side distribution (Table 1) were 12 (46.20%) left side and 14 (53.80%) right side.

A. Demographic findings for DMD boys

Table (1) displays the demographic information for DMD boysin the population study group. The study included twenty-six boys who had been diagnosed with DMD; their ages varied from 6.00 to 11.00 years, and the mean was 7.69 ±1.43 years (Table 1 and Figure 1). VS values ranged from 2.00 to 7.00, with a mean of 3.23 ± 1.68 (Table 1 and Figure 2). For children with DMD, the afflicted side distribution categories was 12 (46.20%) on the left side and 14 (53.80%) on the right side (Table 1 and Figure 4).

Table (1): Demographic findings for children in study group

Quantitative variables		Mean ±SD	Minimum	Maximum
Age (year)		7.69 ±1.43	6.00	11.00
Vignos scale		3.23 ± 1.68	2.00	7.00
Qualitative variables		Number	Percentage	
Gender	Boys	26	100%	
	Girls	0	0.00%	
	Left	12	46.20%	

Data are expressed as mean ±SD, or frequency (%)

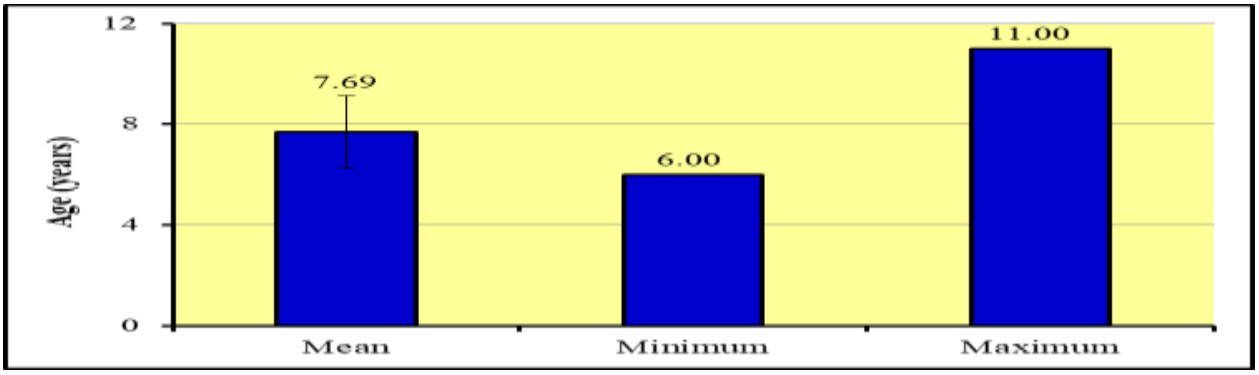

Figure (2): Mean, minimum, and maximum age values of the children included in the study group.

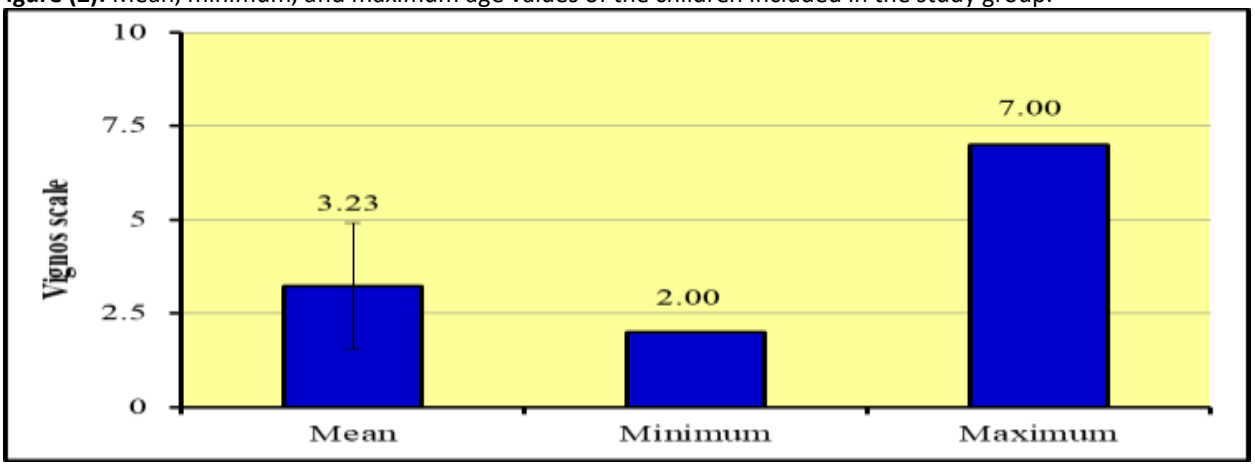

Figure (3): The mean, minimum, and maximum Vignos scale in the study group.

B. Descriptive statistics for main variable outcomes in study group

Table (2) displays the descriptive statistics of the primary variable outcomes for children with DMD in the population study group. Children's MF measurements had a mean of 80.13 ±11.04 (Table 2 and Figure 4), with a minimum of 54.16 and a maximum of 94.70. Children's FL had a mean of 2.34 ±0.64 (Table 2 and Figure 5), with a minimum of 0.94 and a maximum of 3.74. Children's muscle thickness ranged from 0.90 to 1.73, with a mean of 1.33 ±0.24 (Table 2 and Figure 6).Children's knee flexion degree was 19.12 ±8.08 on average (Table 2 and Figure 7), with a minimum of 5.60 and a high of 32.70. Children's ankle planter flexion had a mean of 90.74 ±8.59 (Table 2 and Figure 8), with a minimum of 80.10 and a maximum of 108.50.

Table (2): Descriptive statistics for main variable outcomes in study group

Items	Mean ±SD	Minimum	Maximum
Motor function measurement	80.13 ±11.04	54.16	94.70
Fascicle length	2.34 ±0.64	0.94	3.74

Data are expressed as mean ±SD

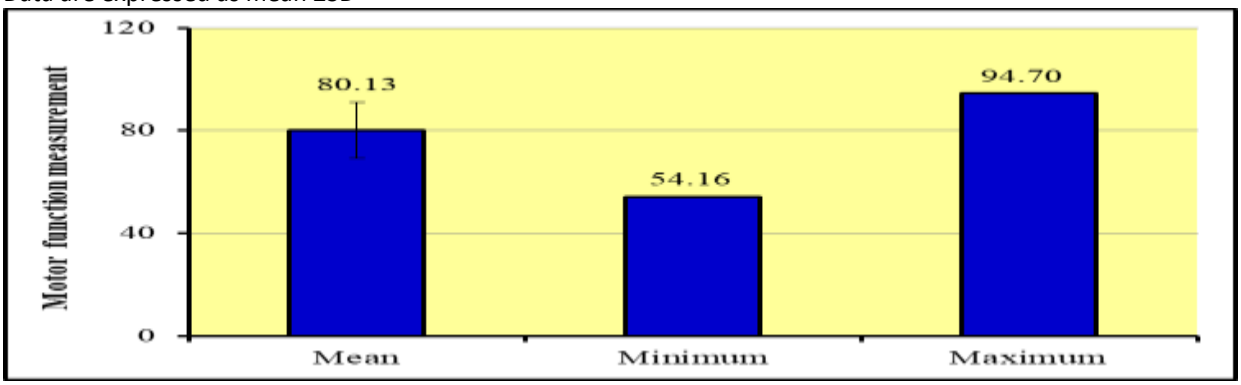

Figure (4): The mean, minimum, and maximum values of children motor function measurement in study group.

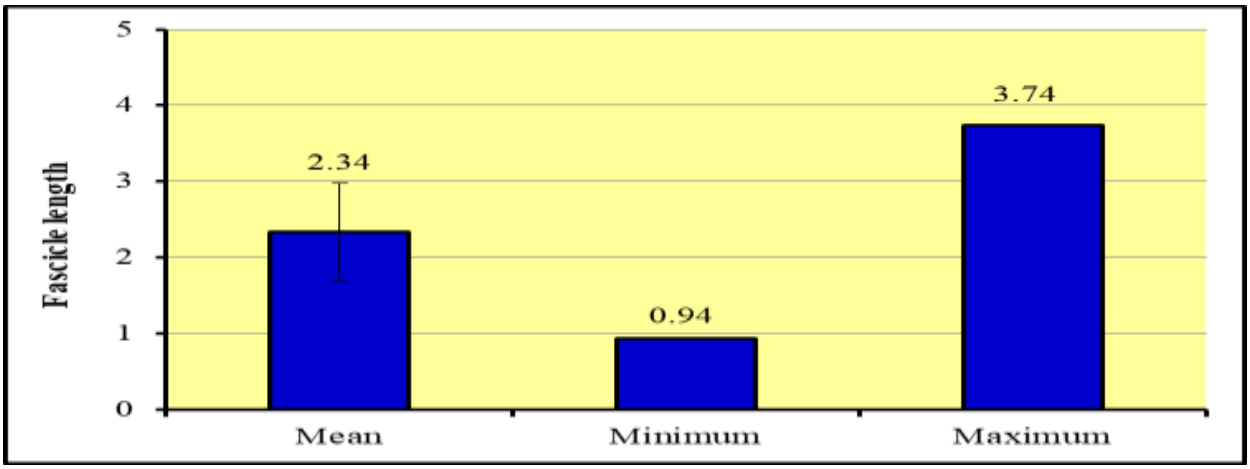

Figure (5): The mean, minimum, and maximum values of children fascicle length in study group.

The two variables' Pearson correlation coefficients were computed. (assessments of muscle architecture and MF) in DMD children are shown in Table (3). The correlation analyses revealed no statistically significant associations (P > 0.05) between MF assessment and FL (r = 0.039, P = 0.850) or muscle thickness (r = 0.140, P = 0.494). These findings suggest that variations in muscle thickness and FL were not consistently associated with changes in MF among children with DMD.

A negative correlation was observed between ankle plantar-flexion and MF assessment (Figure 1), indicating an inverse relationship between the two variables. Specifically, as ankle plantar-flexion increased, MF scores decreased among children with DMD.

C. Relation between fascicle length and motor function measurement in children with DMD

The bi-variate Pearson correlation coefficients between independent (FL) and dependent variable (MF). (Table 4)

This correlational analysis's findings showed that FL and MF assessment had no significant relationship (P>0.05) (r=0.039; P=0.850). This insignificant relationship indicates that in children with DMD, changes in FL are inconsistent with changes in MF.

Table 4. Correlation between fascicle length and motor function measurement in children with DMD.

Items	Fascicle length	Motor function measurement
Mean ±SD	2.34 ±0.64	80.13 ±11.04
95% confidence interval	2.07 – 2.59	75.66 – 84.59
r-value	0.039	
P-value	0.850	
Significance	NS	

**Data are presented as mean ± SD. P-value indicates the probability value, and NS denotes a non-significant result.

Discussion

This study was designed to examine whether gastrocnemius muscle architectural characteristics, with particular emphasis on muscle thickness, are associated with functional capacity in children with DMD. The sample consisted of 26 ambulatory boys aged 6–12 years with genetically confirmed DMD, whose functional status varied from grades 1 to 7 according to the VS.

The findings revealed insignificant correlation between MFM-32 and gastrocnemius muscle thickness. This lack of association suggests that muscle architectural measurements alone may not adequately indicate the muscle functional capacity. Although muscle thickness is generally associated with force-generating capacity in healthy muscle, its reliability as a functional indicator in DMD may be limited by pathological changes such as pseudohypertrophy, fibrosis, and fatty infiltration, which can increase muscle size without preserving its contractile function.

The lack of a strong association between fascicle length and functional performance may be due to the complex pathological changes occurring within dystrophic muscle. DMD is characterized by progressive degeneration of muscle fibers, defective regeneration, inflammation and replacement of contractile tissue by adipose and connective tissue. These pathological processes progressively reduce muscle force-generating capacity and functional performance [21]; [22]. Consequently, a structural measurement such as fascicle length may not necessarily reflect the functional quality of the muscle.

Altered muscle architecture has been demonstrated in children with DMD including changes in gastrocnemius fascicle length and muscle thickness. In particular, ultrasound studies have identified alterations in the gastrocnemius architecture in the early stages of DMD indicating that architectural anomalies may precede manifest functional decline [23]; [24]). However, an architectural change does not necessarily imply a direct relation with functional performance. This interpretation is supported by the present findings, which do not reveal any significant correlation between fascicle length and the MFM-32 score, although measurable architectural changes are present. Previous ultrasonographic studies have indicated that the apparent increase in muscle size in children with DMD is primarily attributable to the replacement of muscle tissue by connective tissue and fat rather than true muscular hypertrophy [5];[25] This finding is consistent with the results of the present study, which showed no significant association between measured muscle thickness and functional performance or mobility. Similarly, although FL is

often considered a predictor of muscle shortening velocity, it was not significantly correlated with MFM scores in the current study [26], This may be due to the loss of elastic characteristics and fibrotic shortening of muscle fibers, which alter fascicle form without improving function.

One possible explanation is that fascicle length is only one component of muscle architecture and does not account for muscle quality. In DMD, the development of fatty infiltration and fibrosis may significantly decrease the amount of effective contractile tissue, even when the overall size or fascicle arrangement of the muscle appear relatively intact. In particular, muscle echogenicity measurements have been increasingly studied as a clinically feasible method to assess disease-related changes in DMD. Increased echogenicity relates to pathological alterations in muscle composition, and may give information regarding disease severity not attainable from the measurement of muscle dimensions only [27];[28]. Therefore, the lack of correlation observed in the present study may indicate that muscle composition and tissue quality are more closely related to functional ability than resting fascicle length.

Another possible reason for the lack of strong correlation might be the multi-dimensional aspect of functional performance. The MFM-32 evaluates different components of motor function relative to axial, proximal, and distal movements. Thus, functional tasks are dependent on the combined contribution of multiple muscle groups and not the isolated function of the gastrocnemius. In ambulatory DMD children, performance is influenced by strength and integrity of the hip, knee and ankle musculature, trunk control, balance, joint mobility and compensatory movement strategies [21]. Consequently, a measurement obtained from a single muscle may have limited ability to explain variability in a global functional measure such as the MFM-32.

The gastrocnemius however is a major contributor to locomotor function as it contributes to ankle plantarflexion and propulsion during gait. But the connection between its static architecture and its dynamic function is complicated. The fascicles interact mechanically with the tendon and aponeurosis and thus do not necessarily change length in the same way as the whole muscle-tendon unit during locomotion [29]. demonstrated that gastrocnemius fascicles can operate with relatively limited length changes during locomotion while substantial changes occur in the length of the muscle-tendon unit. Therefore, a resting ultrasound measurement of fascicle length may provide limited information regarding how the gastrocnemius functions dynamically during walking.

Gait abnormalities, including reduced ankle dorsiflexion and excessive plantarflexion during stance, can develop in association with gastrocnemius–soleus muscle weakness and the progressive onset of ankle joint contractures [30]. These biomechanical findings are supported by the current research, which shows that compensatory posture and joint mechanics are more strongly linked to functional decline than static muscle measurements [31].

The lack of a relationship between FL and functional performance in this study is consistent with the findings of [32] and [33], who stated that adipose and connective tissue infiltration, rather than an increase in functional muscle mass, is the primary cause of the noticeable hypertrophy in DMD calves. However, some previous research [34].

This distinction is particularly relevant to DMD, in which progressive muscle weakness, altered muscle-tendon properties, and joint contractures can modify gait mechanics. Children with DMD frequently develop compensatory gait strategies to maintain forward progression despite reduced muscle strength. These strategies may include increased ankle plantarflexion, alterations in knee position, and modifications of proximal joint motion. Such compensatory mechanisms may allow children to maintain ambulation despite substantial muscle pathology. Therefore, functional performance may be more strongly influenced by the interaction between muscle weakness, joint mobility, and gait adaptations than by resting fascicle length alone.

The current findings should also be interpreted in relation to the age and functional characteristics of the sample. The participants in the present study were ambulatory children between 6 and 12 years of age. During this stage of DMD, functional decline is progressive but may vary considerably between individuals. Children of a similar age may demonstrate substantial differences in muscle quality, strength, contracture development, and gait pattern. This biological variability may weaken the correlation between a single architectural parameter and functional performance. Furthermore, because the study included only ambulatory children, the range of functional impairment may have been relatively restricted, potentially reducing the magnitude of detectable correlations.

The absence of a significant association between fascicle length and MFM-32 performance does not necessarily indicate that fascicle length is clinically irrelevant. Rather, fascicle length may represent a structural marker that is better interpreted in conjunction with other architectural and functional parameters. Previous research suggests that muscle architecture can provide useful information regarding disease-related changes, but its interpretation should consider muscle strength, tissue composition, joint mobility, and functional status simultaneously [35]. Thus,

ultrasound assessment of fascicle length may have greater clinical value when incorporated into a multimodal assessment rather than used as an isolated predictor of function.

The findings further suggest that future investigations should move beyond static measurements of muscle architecture. Longitudinal studies are required to determine whether changes in fascicle length over time are associated with subsequent functional decline. Such studies would be particularly valuable because the present cross-sectional design cannot establish temporal or causal relationships. In addition, dynamic ultrasound assessment during gait or controlled ankle movement could provide information regarding fascicle excursion and fascicle strain that cannot be obtained from resting measurements. Combining dynamic ultrasound with quantitative MRI, particularly MRI-derived muscle fat fraction, could also help distinguish architectural alterations from changes in the amount of viable contractile tissue.

The findings highlight the importance of preventing contractures and preserving joint flexibility to maintain functional mobility in children with DMD. Although muscle ultrasonography remains a valuable tool for diagnosis and disease monitoring, clinicians should interpret increased muscle thickness with caution, as it may reflect pathological pseudohypertrophy rather than preserved muscle strength [36],[5]. From a rehabilitation perspective, physical therapists should incorporate regular stretching and range-of-motion exercises, particularly for the hamstrings and gastrocnemius muscles, to minimize the development of contractures [37]. Functional training and gait re-education should also be included to optimize compensatory strategies and improve energy efficiency during mobility. Finally, longitudinal monitoring should incorporate functional outcome measures such as the M -32 rather than relying on muscle fascicle length measurements to assess changes in functional performance[38].

Conclusion

The present study demonstrated that functional performance in ambulant children with DMD was not significantly associated with variations in gastrocnemius muscle thickness. Instead, functional limitations may be more closely related to joint restrictions encountered during daily activities than to changes in muscle thickness alone. Therefore, gastrocnemius muscle thickness may have limited value as a predictor of functional performance in DMD, as pathological changes such as pseudohypertrophy, muscular fibrosis and fatty infiltration can obscure the muscle's actual contractile properties. These findings emphasize the importance of early physiotherapy interventions aimed at maintaining proper joint alignment, preserving muscle and joint flexibility, and promoting functional mobility.

Limitations:

The interpretation of the study findings should take into account several methodological limitations. First, incorrect lower-limb placement may have been caused by fear or anxiety during the US evaluation, which might have affected the precision of muscle architecture measures. Second, direct comparison with current results was hampered by the paucity of prior research on the connection between muscle architecture and functional performance in DMD. Third, the research only examined the gastrocnemius muscle; it did not evaluate the quadriceps or tibialis anterior, two additional muscles that are crucial to gait. Furthermore, the very small sample size may restrict the findings' generalizability and reflects the rarity of DMD. Lastly, the cross-sectional design did not allow for the study of long-term changes in muscle structure or functional performance, but it did allow for the assessment of relationships at a single time point.

Recommendations for Further Research:

Longitudinal study designs should be used in future studies to track changes in muscle morphology over the course of DMD development. To distinguish real muscle tissue from fatty infiltration, quantitative imaging methods like MRI-based fat fraction analysis should be used. The potential of physiotherapy therapies to retain functional skills, reduce the development of contractures, and preserve FL should be further investigated. A more thorough knowledge of the degree and course of muscular involvement in DMD may also be obtained by evaluating many lower-limb muscles.

Implications for Physiotherapeutic Management

In ambulatory DMD children, differences in muscle thickness alone may not be a reliable indicator of functional performance.

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Statement of Data Availability
The data supporting the findings of this study are available from the corresponding author upon reasonable request.

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Written informed consent was obtained from the parents or legal guardians of all participating boys before their enrollment in the study.

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Author Contributions:
Dimiana Samir Fied was responsible for study conceptualization, formal analysis, methodology, validation, investigation, resource provision, data curation, and project administration. She also contributed to manuscript development by preparing the initial draft and subsequently reviewing and editing the final version. Amira Mohamed El-Tohamy, Marwa Waly Eldin Ali, and Ahmed S. Awad contributed to the conceptualization, methodology, formal analysis, validation, investigation, resource provision, data curation, supervision, and project administration of the study. They also participated in drafting, reviewing, and editing the manuscript. All authors critically reviewed and approved the final manuscript for submission.