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Original Article

Clinical Characterization of 53 Egyptian children diagnosed as Duchenne Muscular Dystrophy in a Tertiary Unit In Upper Egypt

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Abstract

Background: Muscular dystrophies, which are characterized by progressive muscle atrophy, are a heterogeneous group of inherited muscular diseases.

Aim of work: Clinical aspects in children with Duchenne muscular dystrophy at Sohag University Hospital. **Subjects and methods:** This was cross-sectional research performed at the pediatric department and pediatric neurology unit, Sohag University Hospital over two years, from 2022 to 2023.

Results: In our research, 53 children had a confirmed Duchenne diagnosis, divided into two groups according to ambulation: Group A: 33 (62.3%) ambulant children. Group B: 20 (37.7%) non-ambulant children. The verbal and total IQ tests were significantly higher in group A than group B. The EF (ejection fraction) and FS (fraction shortening) were significantly higher in group A than group B. All patients had hyporeflexia. Regarding muscle state, there was calf hypertrophy in 33 (100%) of group A and 3 (15%) in group B, and Muscle wasting in 17 (85%) of group B compared to no cases in group A, which was significantly different. As regard skeletal abnormalities, 27 (81.8%) patients in group A had lordosis compared to 4 (20%) patients in group B, while 4 (20%) had scoliosis, 5 (25% had scoliosis and joint stiffness), and 5 (25% had joint stiffness) in group B, with no cases in group A with a statistically significant difference. Functional ability (North Star score) was positively correlated with young age.

Conclusion: Younger age and ambulant status were associated with better cardiac, muscular, skeletal, cognitive, and functional outcomes and abilities in Duchenne muscular dystrophy patients.

Key Words: Clinical Characterization, Duchenne Muscular Dystrophy.

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Introduction

Duchenne muscular dystrophy [DMD] is an uncommon neuromuscular illness that is X-linked in nature. Estimates place its occurrence at approximately 1 in 3,500 to 1 in 5,000 live male births. Mutations in the dystrophin gene [DMD] cause DMD, leading to a near-absence or complete lack of dystrophin, an essential protein responsible for maintaining the integrity of muscle cells. DMD is characterized by a severe dystrophin shortage that leads to progressive muscle degeneration & function loss, ultimately resulting in premature mortality, which typically occurs by age 30. [(1)]

DMD typically manifests during early childhood and is characterized by motor challenges such as impaired achievement of developmental milestones, recurrent accidents, high fatigability, and hypertrophy of the calf muscles. Individuals experience a progressive decline in mobility throughout the illness, culminating in the inability to walk by early adolescence & subsequent functional impairment of the upper extremities, which hinders the execution of even the most fundamental self-care activities. Scoliosis may arise as a consequence of trunk muscle atrophy & frequently necessitates surgical intervention for the preservation of respiratory function.[(2)] Insufficiency of dystrophin within the cardiac muscle tissue gives rise to cardiomyopathy, which subsequently culminates in the need for mechanical ventilation. [(3)]

Due to limited reports from low-resource settings, particularly from Upper Egypt; possible differrences in clinical features due to differences in access to healthcare facilities and novel therapeutic options. The objective of this research was to examine clinical aspects of Duchenne muscular dystrophy in children at Sohag University Hospital.

Patients and Methods

Study type & region: This cross-sectional research was conducted at the pediatric department and pediatric neurology unit at Sohag University Hospital. The duration of the study was two years. We separated the individuals into two cohorts based on the still ambulance, and then studied and compared the different predictive factors in both groups.

Inclusion criteria:

- The age of onset varies from three to eighteen years.
- Clinical presentation characteristic of Duchenne muscular dystrophy
- During the research period, patients whose clinical manifestation was confirmed by specific biochemical analysis or genetic testing presented to the pediatric department & neurology outpatient clinic.

Exclusion criteria:

- Offspring with additional congenital muscular dystrophy
- Children who suffer from additional myopathies
- CNS disorders, including brain injury and spinal muscular atrophy, should be noted.
- Feminine gender

Methods: All individuals were exposed to:

- **Complete history-taking:**
- **Personal history**: age, name, duration, age of onset, course, & complication of the disease.
- **The presenting symptoms focus on:**

A. Neurological manifestation such as frequent falls, difficulty rising from a sitting position, waddling gait, toe walker, large calf muscle, muscle stiffness, loss of ambulation, learning disabilities, and psychiatric abnormalities.

B. Extra neurological manifestations such as cardiac arrhythmia, dilated cardiomyopathy, and respiratory failure

- **Physical examinations**
  Data from patient examinations focusing on neurological examination as a regression of motor power, calf muscle hypertrophy, and increasing muscle tone.
- **Regression of Motor Power** it indicates a decline in muscle strength. This was assessed through a systematic evaluation of muscle groups using the Medical Research Council (MRC) scale. The scale typically ranges from 0 to 5, with 0 indicating no muscle movement & 5 indicating normal strength.

- **Investigational Studies:**
- **Routine laboratory investigations:**

- Complete blood count (CBC).
-Erythrocyte sedimentation rate and C- reactive protein
-Liver and kidney functions.
- Serum muscle enzyme: serum CK LDH level

- Thyroid function tests
- **Radiological investigation**
- **Echocardiography**

Outcome Measurements and Follow-Up
- IQ test: the Stanford Bient intelligence scale [4].
- North Star Ambulant Assessment (NSAA) [5].

We divided cases into ambulant and non-ambulant cases, and we performed North Star for ambulant cases.

Ethical Consideration: The approval of the Sohag Faculty of Medicine research ethics committee was obtained. Written consent was obtained from the guardian. The data that was obtained from participants was confidential. No report or publication concerning this study identified the study participants by name. Before the participants were admitted to this study, the purpose and nature of the study, as well as the risk-benefit assessment were explained to them. Informed consent was obtained.

Data management and statistical analysis: data collected throughout history, basic clinical examinations, laboratory investigations, and outcome measures were coded, entered, and analyzed using SPSS (statistical package for social science) version 25 (Armonk, NY: IBM Corp.). Two types of statistics were done**:**

Descriptive statistics: According to the type of data, qualitative data were represented as number and percentage, and quantitative data were represented by the mean ± SD.

Chi square test was used for analysis of qualitative data and unpaired t test was used to compare quantitative data. A value of 0.05 was used as a significant level.

Results

Molecular studies were done for all of them to confirm the diagnosis of DMD. At first MLPA DMD gene mutation technique was applied and showed that 44(62%) patients were positive while 27(38%) patients were negative. Those patients with negative results were further evaluated by whole exome sequence (WES) technique which revealed that 9(33%) patients were positive for DMD, so the total number of patients confirmed to have DMD were 53 and the remaining 18 were still negative for DMD diagnosis. We divided the 53 children with a confirmed Duchenne diagnosis into two groups based on their ability to walk.

Group A included 33 (62.3%) ambulant children.
Group B included 20 (37.7%) non-ambulant children the age and BMI were significantly higher in group B than group A, where age ranged from 3.00 to 18.00 years in group A, with a mean of 7.65±3.08, and in group B it ranged from 10.00 to 16.00, and the mean was 12.90±1.62 years, while BMI ranged from 12.5 to 23.4 kg/m2 in group A, with a mean of 17.22±2.57, and in group B it ranged from 13.9 to 34.8, and the mean was 23.48±4.72 **(Table 1).**

Table 2 showed that there was no significant difference between groups as regards residence, family history, or consanguinity.

The neurological examination of the included patients revealed hypotonia in 33 (100%) of group A and 13 (65%) of group B, as well as hypotonia with joint contracture in 7 (35%) of group B, which was significantly different (P = 0.001). All of the patients had weak deep tendon reflexes. In terms of muscle state, 33 (100%) of patients in group A had calf muscle hypertrophy, compared to only 3 (15%) of patients in group B. Also, 17 (85%) of the patients in group B had muscle wasting, while none of the patients in group A did.

This was a statistically significant difference (P = 0.001). As regards the Gower sign, it was present in 31 (93.9%) of patients in group A and absent in 2 (6.1%), while it was absent in all patients in group B, which was significantly different (P = 0.001). LDH and CPK were markedly high in both groups, with no significant difference between the two groups as regards LDH and CPK **(Table 3).** The IQ test verbal and IQ test total were significantly higher in group A than group B (p <.001) **(Table 4).**

The EF (ejection fraction) and FS (fraction shortening) were significantly higher in group A than group B, where EF ranged from 50 to 92 in group A, with a mean of 69.79±11.07, and in group B, it ranged from 34.00 to 72.00, and the mean was 51.80±11.80, while FS ranged from 25 to 48 in group A, with a mean of 36.12±5.61, and in group B, it ranged from 17 to 37, and the mean was 26.45±6.79. As regard other echo findings, in group B, there were 9 (45%) patients with dilated left ventricles in comparison to only 3 (9.1%) patients with the same finding in group A, while dilated left ventricles with MR (mitral regurgitation) and TR (tricuspid regurgitation) were

higher in group A than in group B. These findings had a significant difference of p = 0.026 **(Table 5).**

Table 6 shows the functional ability, which was measured by the North Star score in group A; it ranged from 5 to 34, with a mean of 18.12±6.99.

As regard skeletal abnormalities, the included patients showed that 27 (81.8%) patients in group A had lordosis compared to 4 (20%) patients in group B, while 4 (20%) had scoliosis, 5 (25%) had scoliosis & joint stiffness, and 5(25%) had joint stiffness in group B, with no cases in group A with a statistically significant difference (p = 0.011) (**Table 7).**

The distribution of the studied patients regarding medication showed that in group A, 27 (81.8%) of patients had current steroid therapy, which was continuous in 13 (48.1%) of them, while only 6 (30%) patients in group B received steroid therapy, which was mainly interrupted, which was statistically significant (p = 0.00) as regards dietary supplement (L-carnitine). 30 patients in group A, in contrast to 11 patients in group B, had a dietary supplement (L-carnitine), which was statistically significantly higher in group A than group B (p = 0.002) (**Table 8).** Currently receiving steroid therapy was significantly associated with lower BMI, higher IQ verbal, IQ total, and a North Star score in the studied patients **(Table 9).**

Functional ability (North Star score) was positively correlated with higher IQ verbal and IQ total (**Table 10).**

Table (1): Age and BMI of the confirmed DMD patients (n=53)

		Group A (n=33)	**Group B (n=20)**	**Total**	P-value
Age (years)	Range	3.00 - 18.00	10.00 - 16.00	3 - 18	0.001**
	Mean± S. D	7.65 ± 3.08	12.90 ± 1.62	9.63 ± 3.66	
BMI (kg/m2)	Range	12.5 - 23.4	13.9 - 34.8	12.5 - 34.8	0.001**
	Mean± S. D	17.22 ± 2.57	23.48 ± 4.72	19.58 ± 4.75	

Table (2): Residence, family history, and consanguinity of the confirmed DMD patients (n=53)

	Group A (n=33)		**Group B (n=20)**		**Total**			P-value
	N	**%**	**N**	**%**	**N**	**%**	χ^2	
Residence								
Urban	14	42.4%	7	35.0%	21	39.6%	0.287	0.592
Rural	19	57.6%	13	65.0%	32	60.4%		
Family history								
Negative	19	57.6%	15	75.0%	34	64.2%	1.644	0.200
Positive	14	42.4%	5	25.0%	19	35.8%		
Consanguinity								
No	14	42.4%	7	35.0%	21	39.6%	0.287	0.592
Yes	19	57.6%	13	65.0%	32	60.4%		

Table (3): Neurological examination and CPK and LDH level of confirmed DMD patients, n:53

	Groups				Total		χ^2	P-value
	Group A (n=33)		Group B (n=20)					
	N	%	N	%	N	%		
Muscle tone								
Hypotonia	33	100.0%	13	65.0%	46	86.8%	13.31	0.001**
Hypotonia with contracture of joints	0	0.0%	7	35.0%	7	13.2%		
Deep Tendon Reflexes								
Hyporeflexia	33	100.0%	20	100.0%	53	100.0%	0.00	1.00
Muscle state								
Calf hypertrophy	33	100.0%	3	15.0%	36	67.9%	41.29	0.001**
Muscle wasting	0	0.0%	17	85.0%	17	32.1%		
Gower sign								
Negative	2	6.1%	20	100.0%	22	41.5%	45.26	0.001**
Positive	31	93.9%	0	0.0%	31	58.5%		

CPK and LDH enzymes									
CPK	Range	515 - 35930	328 - 59000	328 - 59000	0.626
	Mean± S. D	10057 ± 7936	11591 ± 14856	10636 ± 10953	
LDH	Range	206 - 12269	214.00 - 8734	206 - 12269	0.912
	Mean± S. D	1120 ± 2051	1182 ± 1847	1144 ± 1959	
	Range	11.69 ± 1.35	10.35 ± 1.49	328 - 59000	

Table (4): IQ findings in confirmed DMD patients n:53

IQ test		Group A (n=33)	Group B (n=20)	Total	P-value
IQ test verbal	Range	50 - 98	60 - 83	50 - 98	0.001**
	Mean± S. D	80.79 ± 9.91	71.10 ± 7.81	77.13 ± 10.25	
IQ test total	Range	52 - 105	64 - 86	52 - 105	0.001**
	Mean± S. D	83.97 ± 10.60	74.30 ± 8.13	80.32 ± 10.75	

Table (5): ECHO findings in confirmed DMD patients n:53

Cardiac function		Group A (n=33)	Group B (n=20)	Total	P-value
EF	Range	50.00 - 92.00	34.00 - 72.00	34 - 92	0.001**
	Mean± S. D	69.79 ± 11.07	51.80 ± 11.80	63 ± 14.27	
FS	Range	25.00 - 48.00	17.00 - 37.00	17 - 48	0.001**
	Mean± S. D	36.12 ± 5.61	26.45 ± 6.79	32.47 ± 7.66	

Echo findings	N	%	N	%	P-value
Normal	25	75.8%	9	45.0%	0.026*
Dilated left ventricle	3	9.1%	9	45.0%	
Dilated left ventricle, MR	2	6.1%	1	5.0%	
Dilated left ventricle, TR	3	9.1%	1	5.0%	

Table (6): The North Star score among ambulant DMD patients (n=33)

	Unable		Modified Method		Normal	
	No.	%	No.	%	No.	%
Stand	4	12%	9	27%	20	61%
Walk	3	9%	25	76%	5	15%
Stand up from chair	3	9%	25	76%	5	15%
Stand on one leg-right	5	15%	12	36%	16	48%
Stand on one leg-left	5	15%	12	36%	16	48%
Climb box step-right	7	21%	21	64%	5	15%
Climb box step-left	7	21%	21	64%	5	15%
Descend box step-right	7	21%	21	64%	5	15%
Descend box step-left	7	21%	21	64%	5	15%
Gets to sitting	3	9%	25	76%	5	15%
Rise from floor	3	9%	25	76%	5	15%
Lifts hand	0	0%	10	30%	13	39%
Stand on heel	4	12%	12	36%	17	52%
Jump	9	27%	17	52%	7	21%
Hop right leg	10	30%	18	55%	5	15%
Hop left leg	10	30%	18	55%	5	15%
Run (10m)	10	30%	18	55%	5	15%
Range Mean± S. D	5.00-34.00 18.12±6.99					

Table (7): Skeletal abnormalities of the confirmed DMD patients

Skeletal examination	**Groups**				**Total**		χ^2	**P-value**
	Group A (n=33)		**Group B (n=20)**					
	N	**%**	**N**	**%**	**N**	**%**		
Normal	6	18.2%	2	10.0%	8	15.1%	31.78	0.001**
Lordosis	27	81.8%	4	20.0%	31	58.5%		
Scoliosis	0	0.0%	4	20.0%	4	7.5%		
Scoliosis & joints stiffness	0	0.0%	5	25.0%	5	9.4%		
Joints stiffness	0	0.0%	5	25.0%	5	9.4%		
Total	33	100.0%	20	100.0%	53	100.0%		

Table (8): Medications received by the studied patients, n=53

Received medication	**Groups**				**Total**		χ^2	**P-value**
	Group A (n=33)		**Group B (n=20)**					
	N	**%**	**N**	**%**	**N**	**%**		
Steroid Therapy								
Current	27	81.8%	6	30.0%	33	62.3%	14.23	0.001**
Previous	6	18.2%	14	70.0%	20	37.7%		
Regimen of current steroid use								
Interrupted	14	51.9%	6	100.0%	20	60.6%	4.77	0.029*
Continuous	13	48.1%	0	0.0%	13	39.4%		
Dietary supplement (L-Carnitine)								
Yes	30	90.9%	11	55.0%	41	77.4%	9.17	0.002**
No	3	9.1%	9	45.0%	12	22.6%		

Table (9): Relation between steroid therapy and BMI, IQ verbal, IQ total and North star score in the studied confirmed patients, n=53

Steroid therapy	Current (n=33)			Previous (n=20)			P-value
	Min	**-**	**Max**	**Min**	**-**	**Max**	
	Mean	**±**	**S. D**	**Mean**	**±**	**S. D**	
BMI (Kg/m2)	12.50	-	34.80	14.80	-	27.00	0.002*
	18.26	±	4.79	21.77	±	3.90	
IQ Verbal test	50.00	-	98.00	60.00	-	98.00	0.001*
	80.18	±	9.83	72.10	±	9.07	
IQ test	52.00	-	105.00	64.00	-	101.00	0.002*
	83.39	±	10.45	75.25	±	9.44	
North star score	**Current (n=27)**			**Previous (n=6)**			0.024*
	8.00	-	34.00	5.00	-	22.00	
	19.48	±	7.17	11.83	±	6.94	

Table (10): Correlation between Functional ability (North star score) and Age, BMI (Kg/m2), IQ Verbal test, and IQ test, n=53

	Functional ability (North star score)	
	r	**P**
Age	-0.017	0.924
BMI (Kg/m2)	-0.115	0.523
IQ Verbal test	0.977	0.001*
IQ test	0.963	0.001*

Discussion

In our study, the 53 children who had a confirmed Duchenne diagnosis were divided into two collections concerning ambulation: **Group A** included 33 [62.3%] ambulant children. **Group B** included 20 [37.7%] non-ambulant children.

A study conducted in Saudi Arabia by AlSaman et al.**(6)** reported that 28.9% of individuals were wheelchair-dependent full-time at the time of enrollment, 50% of ambulatory individuals could run, and 63.9% could ascend stairs.

In our study, the age was significantly higher in group B than group A, this agrees with Ricotti et al., (1) who reported that the ambulant had an average age of 7.9 years. In contrast, the non-ambulant group had an average age of 14.2 years. This age distribution between the groups underscores the progressive deterioration of motor functions in DMD, with ambulatory capacity declining as patients age.

AlSaman et al.**(6)** reported that, in this investigation, the average age at which ambulatory individuals & full-time wheelchair users were enrolled was 7.0 years & 10.1 years, respectively. This suggests a phase of substantial progression of DMD illness occurring among these ages. This result is additionally corroborated by the fact that the mean ages of the individuals who demonstrated the ability to run or ascend stairs were approximately one year lower [6.6 & 6.9 years, respectively] than those who were unable to do so [8.0 and 7.6 years, respectively].

In our research, we noted no significant variance amongst collections as regards residence, family history, and consanguinity.

In a comprehensive database of DMD patients [0–18 years old] in East China, Li et al. (7) reported that 23.1% of the probands had a family medical history of the disease.

In our study, the IQ test verbal and IQ test total were significantly higher in group A than group B ($p < .001$).

Banihani et al.**(8)** demonstrated that individuals with DMD suffer from intellectual disabilities. Behavioral issues & learning difficulties are included.

An intellectual impairment is present in thirty percent of infants who have been diagnosed with Down syndrome, as indicated by an IQ of eighty-five percent or less, which is one standard devi-

ation below the generally accepted population mean of one hundred. [9]

In our study, as regards heart, there was a significant increase in cardiomyopathy and heart failure in group B compared to group A. Also, the EF and FS were significantly higher in collection A than group B, where EF ranged from 50 to 92 in group A.

In our study, as regards the echo finding in group B, 9 [45%] patients had a dilated left ventricle in comparison to only 3 [9.1%] patients with the same finding in group A, while DLV with MR [mitral regurgitation] and TR [tricuspid regurgitation] was higher in collection A than in collection B. These findings had a significant difference ($p = 0.026$).

The research revealed that approximately one-third of these individuals developed cardiomyopathy by the age of 14, one-half by the age of 18, & all individuals were older than eighteen years. Despite the considerable prevalence of dilated cardiomyopathy among individuals with DMD, the majority remain asymptomatic until the later stages of their illness. This has been attributed to the patients' increasing incapacity to engage in physical activity, which obscures the functional impairment that is typically among the initial indicators of heart failure. **(10)**

Cardiomyopathy progressively becomes more prevalent among individuals with DMD as they enter their second decade of life. **(10,11)**

Since cardiac myocytes also contain dystrophin, cardiac involvement is unavoidable.**(12)** The ventricular myocardium exhibited a diverse range of pathological changes, which were attributed to the combined effects of myocardial wasting & remodeling caused by progressive cardiac muscle degradation leading to decreased systolic function. Following the demise of myocytes, the latter process takes place alongside fibrosis & secondary lipid infiltration. **(13)**

Li et al.**(7)** reported that the cardiac functions of 46.4 percent of DMD males were evaluated. We prescribed cardiac medication for the management of four young males who presented with cardiac function abnormalities.

In our study, as regards reflexes, all patients had hyporeflexia, and as regards muscle state, there was significant increase in calf hypertrophy in group A than group B, and muscle wasting was higher in group B compared to group A [$P = 0.001$]. Also, the skeletal abnormalities of the included patients showed that patients in group A had higher percentage of lordosis compared to patients in group B, who had scoliosis & joint stiffness with no cases in group A with a statistically significant difference [$p = 0.011$].

Ricotti et al. **(1)** found that upper limb function, especially at the shoulder level, gets worse more quickly in DMD patients who can't walk. The total PUL score drops by 4.13 points each year, and the shoulder level drops by 0.97 points. In contrast, ambulant patients displayed a milder, statistically insignificant decline in shoulder function over a year, aligning with DMD's characteristic progression from proximal to distal muscle groups.

When utilizing the MyoGrip & MyoPinch to assess distal upper limb strength, a consistent decrease in the predicted percentage grip & pinch force was noted. In comparison to grasp force, grip force decreased more rapidly annually in ambulant boys [$p < 0.001$] & at a slower rate in non-ambulant boys [$p < 0.001$] [$p < 0.001$ in ambulant boys and $p < 0.001$ in non-ambulant boys]. **(14)**

Pane et al. **(15)** found a strong connection between how functionally stable a person was and how much their Performance of Upper Limb (PUL) scores changed over 12 and 24 months in people with Duchenne Muscular Dystrophy. Specifically, ambulant boys with better general function [6MWT >350 meters] showed more stability in PUL scores, with minimal deterioration. Conversely, those with reduced functional ability [6MWT <250 meters] exhibited a greater PUL decline, particularly in the shoulder domain, losing an average of 3.8 points over two years. Notably, 86% of these patients lost their ability to walk within the same period. Interestingly, the residual 14% who preserved ambulation experienced a lesser decline in PUL scores, indicating a more stable course in both upper & lower limb functions.

In our research, we observed no significant variance between the current & previous use of steroid drugs as regards CPK and LDH levels, although their levels were higher in individuals with previous therapy.

In individuals who have Duchenne muscular dystrophy, serum creatine kinase [CK] levels are almost always substantially elevated, & they peak around the age of two. In certain cases, the value may exceed the upper limit of normal by a factor of twenty. Nevertheless, this value diminishes ov-

er time due to the progressive replacement of muscle cells with adipose cells & fibrous tissue. [16]

One study done in Pakistan by Hashim et al. [17]demonstrated that exhibited creatine kinase to have a specificity of 91 percent & a positive predictive value of 88.8%, respectively, in instances of DMD, with a sensitivity & negative predictive value of one hundred percent.

In our study, the distribution of the studied patients regarding medication showed that in group A, current steroid therapy, which was continuous was statistically significantly higher than group B where patients received steroid therapy, which was mainly interrupted [p = 0.00] as regards dietary supplement [L-Carnitine] was statistically significantly higher in collection A than collection B [p = 0.002].

Ricotti et al. [1] revealed a slightly lower proportion of daily steroid use in the non-ambulant collection compared to the ambulant collection, which could be related to the progression of the disease or differences in treatment strategies as DMD advances.

AlSaman et al.[6] reported that corticosteroids were prescribed to slightly over forty percent of individuals as an initial component of their treatment management strategy. At the time of the research enrollment, however, the percentage of individuals receiving corticosteroid treatment had increased to nearly sixty percent. Although the increase in corticosteroid use among diagnosis & study enrollment is a positive development, it is worrisome that the proportion is not significantly higher in a population whose motor function is deteriorating swiftly. Uncertainty surrounds the causes of the low use of corticosteroids in Saudi Arabia [SA], but they may be related to Middle Eastern concerns regarding the adverse effects of corticosteroids**.**

The Duchenne Natural History Investigation, conducted by the Cooperative International Neuromuscular Research Group among 2006 & 2016, documented that 87 percent of individuals underwent corticosteroid treatment. [18] Similarly, the STRIDE Registry reported that 89.2% of individuals underwent corticosteroid treatment [89.2%]**.**[5]

In our study, functional ability [North Star score] was positively correlated with young age.

Mazzone et al. [19] It was reported that a 24-month follow-up of an Italian cohort yielded an equivalent level of assurance, as demonstrated by the 330-meter 6MWD. two percent of the boys who walked a minimum of 330 meters at baseline ceased walking within the next two years. Similarly, after a period of two years, approximately 98% of boys in their cohort who obtained an NSAA score of 17/34 [approximately fifty-two units on the linearized NSAA] were ambulant.

Ricotti et al. [20] described the progressional slope after age seven, when motor function begins to deteriorate. Based on their findings and the average linearized NSAA score of 73 [=27/34 in the raw scale] at seven years of age, the transformed NSAA scale indicates an overall decline rate of eight linearized units per year.

Study limitations of this study included its cross-sectional design, limited number of cases, and lack of advanced studies, such as genetic testing**.**

Conclusion

In this research of children with confirmed Duchenne muscular dystrophy, there were significant differences between ambulant and non-ambulant groups in mean age (younger in the ambulant group), thyroid hormone levels, IQ scores, cardiac abnormalities, muscle status, skeletal deformities, and treatment history. Both EF and FS were significantly higher in the ambulant collection. The ambulant collection had more calf hypertrophy and less muscle wasting, while the non-ambulant group had more left ventricular dilation, mitral/tricuspid regurgitation, scoliosis, and joint stiffness. Ambulant children received more continuous steroid therapy and L-carnitine. Overall, younger age and ambulant status were associated with better cardiac, muscular, skeletal, cognitive, and functional outcomes and abilities in Duchenne muscular dystrophy patients.

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