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DISABILITIES IN CHILDREN WITH DUCHENNE MUSCULAR DYSTROPHY: A PROFILE

K. P. Sivaraman Nair,[1] Anisya Vasanth,[2] M Gourie-Devi,[2] Arun B. Taly,[2] S Rao,[3] N Gayathri[4] and T Murali[1]

From the Departments of [1]Psychiatric and Neurological Rehabilitation, [2]Neurology, [3]Bio-statistics and [4]Neuropathology, National Institute of Mental Health and Neurosciences, Bangalore, India

Proper assessment of disabilities is essential for rehabilitation of patients with Duchenne muscular dystrophy. The aim of this study was to identify and quantify the disabilities in children with Duchenne muscular dystrophy and correlate them with impairment. Thirty-one patients with Duchenne muscular dystrophy of age four years and above were studied. The motor functions were evaluated using total motor score, upper and lower extremity function grades and timed function tests. Disability was quantified with Barthel index. The mean scores of motor scales were: total motor score -52 ± 7.8, total functional grade -4.4 ± 1.9 and timed function score -12.5 ± 5.8. Barthel index scores ranged from 45–95 with a mean of 70.8 ± 12.7. Motor scales correlated with each other and with Barthel index. Thirty children had disabilities in multiple spheres of life, which were significantly influenced by the motor power. Barthel index was useful in identifying and quantifying specific areas of disabilities in these children. Evaluation of disabilities using specific measures may be crucial for planning comprehensive management.

Key words: Duchenne muscular dystrophy, disability, Barthel index, rehabilitation.

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Correspondence address: A. B. Taly, Department of Neurology, National Institute of Mental Health and Neurosciences, Hosur Road, Bangalore 560 029, India. E-mail: taly@nimhans.kar.nic.in

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INTRODUCTION

Duchenne muscular dystrophy (DMD) is the most common inherited progressive disorder of skeletal muscles. Despite rapid advances in knowledge about molecular genetics of DMD, there is yet no cure. The mainstay of treatment is to reduce the disabilities, prevent complications, prolong mobility and improve quality of life (1, 2). Conventional protocols for evaluation of children with DMD focus on motor strength testing and do not address the issue of disabilities (3–5). Barthel index (BI) is valid, reliable, simple and easy to use (6). This scale has been widely used to assess the outcome of stroke, traumatic brain injury and spinal cord injury (7). The objective of the current study was to obtain the profile of the disabilities in children with DMD using BI and to correlate them with motor impairment.

PATIENTS AND METHODS

Thirty-one boys of age 4–13 years (8.1 ± 2.1) with DMD, attending the Neuromuscular clinic for comprehensive rehabilitation participated in this study. Informed consent was obtained from the parents. The diagnosis of DMD was based on clinical, biochemical, electromyographic and histopathological features. Immunohistochemistry for dystrophin was done in muscle biopsy specimens from three children and all showed absence of dystrophin. Children below four years of age and those not co-operative for evaluation were excluded from the study. Disability was defined as any restriction or lack of ability to perform an activity in the manner or within the range considered normal for a human being (8).

The children were evaluated at the time of the monthly follow-up visit at the neuromuscular clinic. All assessments were made in the presence of at least one parent or guardian. The strength of deltoids, pectoralis major, biceps, triceps, glutei, iliopsoas, hamstrings and quadriceps muscles of both right and left sides were graded according to Medical Research Council (MRC) grades: Grade 0—no movement, Grade 1—palpable contraction, Grade 2—active movement with gravity eliminated, Grade 3—active movement against gravity, Grade 4—active movement against resistance, but weaker than other side and Grade 5—active movement against full resistance (6). The total motor score (TMS) was derived by adding the motor grades of all 16 muscles tested and TMS ranged from 0–80. The children were assigned a functional grade based on arm and leg function tests according to the protocol for DMD suggested by Brooke et al. (3, 9). The arm and leg function grades were added to get the Total Functional grade (TFG), which varied from 2 (normal) to 16 (bed-bound with no useful hand functions). The time taken by the children to stand from supine, climb four standard steps and run or walk 30 feet as fast as possible was noted. Based on the time taken, each task was given a score: 0–>120 seconds, 1–100 to 120 seconds, 2–80 to 100 seconds, 3–60 to 80 seconds, 4–40 to 60 seconds, 5–20 to 40 seconds and 6–<20 seconds (9). The sum of all three time scores was designated as timed function score (TFS) from 0 to 18. Their disabilities were quantified using BI by interviewing the parents. It is a scale of 10 items with a score range of 0 to 100. The score zero indicates total dependence and 100 maximum independence (7). The correlation among TMS, TFG, TFS and BI was assessed using Pearson's co-efficient of correlation and the influence of motor power on BI was determined by linear regression analysis.

RESULTS

The TMS for the 31 boys ranged from 30 to 67 (52.0 ± 7.8). The mean TMS for upper limbs was 27.6 ± 4.3 (18–36) and for lower limbs was 24.4 ± 4.7 (12–36). There was good correlation between the TMS of upper and lower limb muscles (Pearson's co-efficient of correlation ($r = 0.5131$, $p < 0.01$). The TFG varied from 2 to 10 (4.4 ± 1.9) and had good correlation with TMS ($r = -0.5009$, $p < 0.01$) and TFS ($r = -0.7706$, $p = 0.01$).

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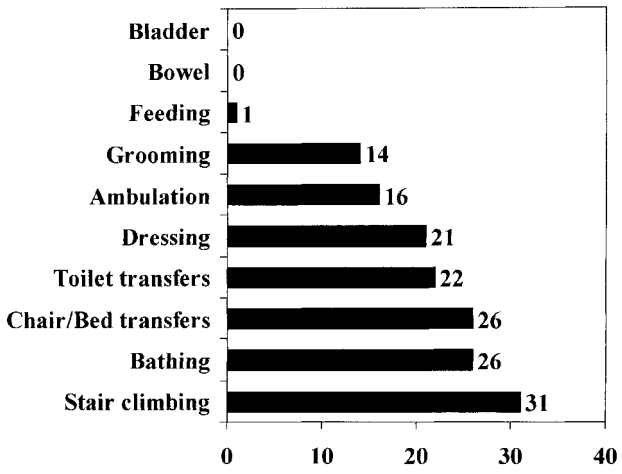

Fig. 1. Prevalence of disabilities among items included in Barthel index in children with Duchenne muscular dystrophy. The items are arranged according to the frequency.

The mean TFS was 12.5 ± 5.8 (0–18). This correlated well with TMS ($r = 0.4221$, $p = 0.05$) and TFG ($r = -0.7706$, $p = 0.01$).

The BI ranged from 45–95(70.8 ± 12.7). Table I shows the correlation between BI and TMS, TFG and TFS. All children were dependent for at least one activity. Fig. 1 shows prevalence of partial or total dependence in various functional activities included in BI. The number of areas in which these children were dependent varied from one to eight: one—1, two—1, three—1, four—10, five—6, six—7, eight—1 and the mean number of disabilities per child was 5.1 ± 1.6. All except three children were variably dependent for four or more activities. The linear regression of BI on TMS had a slope of 0.239 ($p = 0.031$).

DISCUSSION

In DMD the motor weakness becomes apparent when the child starts walking, but proper measurement of strength and disabilities may be difficult in very young children. Brooke et al. observed that by three years of age children with DMD co-operate with test procedures (9). Hence, in the present study, only children four years of age and above were included. Selectivity of the muscle involvement is a typical feature of all muscular dystrophies and therefore the pattern of muscle involvement may determine the disabilities. Brooke et al. noted that hip muscles, knee extensors and rotators of the shoulders were severely affected while other muscles were relatively preserved (3).

Table I. *The correlations between different scales and Barthel index in children with Duchenne muscular dystrophy*

	Barthel index	
	r	*p*
Total motor score	0.3886	<0.05
Total function grade	−0.5991	<0.01
Timed function score	0.6761	<0.01

r = Pearson's correlation co-efficient.
P-level of significance = <0.05.

Scott et al. observed that weakness was always symmetrical and extensor groups were weaker than flexors and proximal groups weaker than the distal (5). While there is a significant relationship between upper and lower extremity strength and functional grades, they may not be entirely equivalent (10). In the current study a good correlation was recorded between the TMS of upper and lower limb muscles.

The functional grading system indicating involvement of arms and legs was applied in the present study (3). Using the same scale, Brooke et al. have reported wide differences in functional ability of patients at a given age (11). McDonald et al. also noted a "non-linear" relationship between functional level grades and motor strength (12). In contrast, Scott et al. found good correlation of motor ability with muscle strength (5). In the current study also a good correlation between muscle strength and functional grades was noted (Table I).

The performance on timed function tests depends on the patients' mood, ability to co-operate, functional state and training (3, 11). The timed walking tests predicted the loss of independent ambulation (12). Scott et al. noted a good correlation between walking times and total muscle strength (5). A similar linear relation was noted between TMS and TFS in the current series. The TFG and TFS denote the maximal functional ability in a clinic setting. They do not denote the habitual functioning of the child (9). A good correlation between TFG and TFS was seen in the present study. While these scales may give information about arm and leg functions in general they do not give details of disabilities.

The protocols designed for the evaluation of DMD focus on motor strength, range of motion and specific motor tests done in a laboratory setting (3, 5). Fowler et al. assessed disability by measures of mobility, upper extremity function, cardiopulmonary adaptations, cardiac and pulmonary complications and psychosocial adjustment (4). Firth et al. reported that 62% of parents of children with DMD had problems in bathing, lifting, cleaning, dressing and feeding the disabled child (13). These children also had significant learning disabilities (14). However, the conventional protocols of DMD do not give an accurate profile of these problems. BI is the best known scale for assessing the activities of daily living. It covers most of the common daily activities, mobility and bowel and bladder functions. Lue et al. reported that BI is acceptable in evaluating disability in children with DMD (15). They noted that sitting balance and hip contractures significantly influenced performance of activities of daily living tasks. In the current study the BI correlated well with TMS, TFG and TFS (Table I) and was significantly influenced by the motor power.

Dependency for self care activities and locomotion is common in DMD (12). In the present study all children except one were dependent for multiple activities. Every one of them needed assistance for climbing stairs (Fig. 1). Modification of

environment like provision of ramps or lifts in schools or relocation of classrooms to ground floor will help these children to be more independent. The majority of these boys needed assistance for transfers. Hence, teaching proper transfer techniques to patients and caregivers should be an integral part of the rehabilitation program for them. About half of the children studied were dependent for locomotion. The independent locomotion can be prolonged by proper physiotherapy, prevention of contractures and suitable orthoses (2). Other activities for which these children were dependent included bathing, dressing, and grooming. Appropriate training using simple adaptive devices like long handled brushes and combs, latch openers, and hooks to pull up trousers can make these children more independent. None of the boys had any problems with control of bowel and bladder functions but 26 of them needed assistance for toilet transfers. This may be overcome by providing western sitting instead of traditional Indian toilets that require squatting. Firth et al. and Lue et al. noted that most of these children are independent in feeding (13, 15). In the present series also only one child had problems with eating (Fig. 1).

There was a good correlation between conventional scales consisting of motor tests and BI in children with DMD. Disabilities in activities of daily living, transfers and locomotion are common among these children. Motor power had significant influence on the disability. The available protocols do not address these issues. Qualitative and quantitative evaluation of the functional skills and disabilities is essential for program planning and meaningful interventions and therefore separate assessment of disabilities using specific scales is essential.

REFERENCES

1. Kakulas BA. Problems and solutions in rehabilitation of patients with progressive muscular dystrophy. Scand J Rehab Med 1999; 39: 23–37.
2. Vasanth A, Gourie-Devi M. Rehabilitation of patients with neuromuscular disorders. In: Taly AB, Nair KPS, Murali T (eds.). Neurorehabilitation: principles and practice. Bangalore: Department of Psychiatric and Neurological rehabilitation, National Institute of Mental Health and Neurosciences 1998: 149–164.
3. Brooke MH, Griggs RC, Mendell JR, Fenichel GM, Schumate JB, Pellegrino RJ. Clinical trial in Duchenne dystrophy 1. The design of the protocol. Muscle Nerve 1981; 4: 186–197.
4. Fowler Jr WM, Abresch RT, Aitkens S, Carter GT, Johnson ER, Kilmer DMD, et al. Profiles of neuromuscular diseases: Design of the protocol. Am J Phys Med Rehabil 1995; 74: S62–S69.
5. Scott DM, Goddard I, Dubowitz M. Quantification of muscle function in children: A prospective study in Duchenne muscular dystrophy. Muscle Nerve 1982; 5: 291–301.
6. Wade D. Measurement in Neurological rehabilitation: Oxford: Oxford University press, 1992.
7. Dobkin BH. Neurological rehabilitation: Philadelphia: FA Davis, 1996.
8. World Health Organization. WHO international classification of impairments, disabilities and handicaps: Geneva: World Health Organization, 1980.
9. Brooke MH, Fenichel GM, Griggs RC, Mendell JR, Moxley R, Miller JP, Province MA, CIDD Group. Clinical investigation in Duchenne dystrophy: 2. Determination of the power of therapeutic trials based on the natural history. Muscle Nerve 1983; 6: 91–103.
10. Lord JP, Portwood MM, Fowler WM, Lieberman JS, Carson R. Upper vs lower extremity functional loss in neuromuscular disease. Arch Phys Med Rehabil 1987; 68: 8–9.
11. Brooke MH, Fenichel GM, Griggs RC, Mendell JR, Moxley R, Florence J, et al. Duchenne muscular dystrophy: patterns of clinical progression and effects of supportive therapy. Neurology 1989; 39: 475–481.
12. McDonald CM, Abresch RT, Carter GT, Carter MD, Fowler WM, Johnson ER, et al. Profiles of neuromuscular diseases: Duchenne muscular dystrophy. Am J Phys Med Rehabil 1995; 74: S70–S92.
13. Firth MA, Gardner-Medwin D, Hosking G, Wilkinson E. Interviews with parents of boys suffering from Duchenne muscular dystrophy. Dev Med Child Neurol 1983; 25: 466–471.
14. Billard C, Cillet P, Barthez M, Hommet C, Bertrand P. Reading ability and processing in Duchenne muscular dystrophy and spinal muscular atrophy. Dev Med Child Neurol 1998; 40: 12–20.
15. Lue YJ, Chen SS, Jong YJ, Lin YT. Investigation of activities of daily living performance in-patients with Duchenne muscular dystrophy. Kao Hsiung I Hsueh Ko Hsueh Tsa Chih 1993; 9: 351–360. (In Chinese).