Journal of Experimental and Clinical Medicine
https://dergipark.org.tr/omujecm

Research Article

J Exp Clin Med
2023; 40(3): 522-529
doi: 10.52142/omujecm.40.3.17

Clinical profiles of neuromuscular disorders: A tertiary hospital experience

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Received: 18.03.2023 • **Accepted/Published Online:** 21.08.2023 • **Final Version:** 30.09.2023

Abstract

This study aimed to identify the symptoms and disease-related features of the most common neuromuscular disorders (NMD) to establish an appropriate multidisciplinary approach in a specific field.A total of 46 patients, 26 (56.5%) women and 20 (43.5%) men with a median age of 46.0 (17.0-72.0) years were divided into three groups according to the affected part of the motor unit: Group I (neuronopathy group, n=11), motor neuron diseases; Group II (neuropathy group, n=13), peripheral neuropathies; and Group III (myopathy group, n=22), myopathies. Demographic and clinical features, functional level, muscular strength, balance, dexterity, activities of daily living, functional performance, global cognitive status test scores, respiratory symptoms, and pulmonary test results were recorded from patient files.There was a significant difference between the mean ages of Groups II and III (p =0.046). A significant difference was observed in muscle weakness between Groups II and III (p = 0.044). The prevalence of dysarthria and speech disorders was higher in Group III (33.3%) compared to Groups II (10%) and I (12.2%). The summarized mini-mental test scores were significantly lower in Group I than in Groups II and III (p =0.047 and p = 0.034, respectively). The nine-hole peg test scores were significantly longer in Group I than in the other groups (p = 0.034 and 0.038, respectively). In this study, NMD were divided into three main groups, and the most common symptoms and clinical findings were evaluated. Fatigue and muscle weakness were the most common symptoms shared by all groups. The mean age of the disease was lowest in the myopathy group. Dysarthria was most common in the neuropathy group. Cognitive impairment was most common in the neuronopathy group.

Keywords: neuromuscular disorders, neuronopathy, peripheral neuropathy, myopathy, clinical features, experience

1. Introduction

Neuromuscular disorders (NMD) are a complex group of heterogeneous, acquired, or hereditary disorders that affect different motor unit parts (1). Injury can occur in motor neurons, sensory neurons, axons, Schwann cells, neuromuscular junctions, muscles, or any combination of these regions (2). Therefore, the term NMD includes a wide variety of different syndromes (3). A systematic neurological evaluation is crucial in diagnosing these complex diseases (4).

The prevalence of NMD is not precisely known and differs across countries. In Turkey, there are approximately 100,000 (Social Security Institutions [SSI]) with NMD in different groups. It is known that there are 70,000 neuromuscular patients in Western Europe, 40,000 in the USA, and over 35,000 in the UK (1, 5). These disorders are rare, genetic, progressive, and untreatable. Some of these cases are autoimmune and can be treated. Symptoms can appear at any age, from childhood to adulthood, and range from mild to severe sensory and/or motor impairment and cardiac or respiratory involvement, which may require life support and/or result in death (6). Muscle weakness is the most common symptom shared by all these disorders (3). The onset and progression of muscle weakness provide some clues for the diagnosis of the disease. Proximal muscle weakness is a presenting symptom of myopathies such as Duchenne and Becker muscular dystrophies, fascioscapulohumeral, limb-girdle, juvenile spinal muscular atrophy, polymyositis, mitochondrial, glycolytic, or lipid storage myopathy, and also Lambert-Eaton myasthenic syndrome. Proximal muscle weakness may be described by patients as a complaint of difficulty combing hair, getting up from a chair, or climbing stairs. Distal muscle weakness can manifest as difficulty in performing finely coordinated movements, such as grasping, jumping off pavement, or descending stairs. Myotonic muscular dystrophy, distal myopathy, scapuloperoneal dystrophy, amyotrophic lateral sclerosis, and distal spinal muscular atrophy are predominantly associated with distal weakness. Distal weakness indicates neuropathy when there is an early loss of deep tendon reflexes and accompanying sensory loss. The trade of muscle weakness is also a guide for the diagnosis. Muscle weakness with remissions and flare-ups

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that fluctuate throughout the day, often worse in the evening than in the morning, is usually caused by myasthenia gravis (7). In addition to muscle weakness, patients should be questioned about common symptoms such as cramps, spasms, and stiffness (8). A comprehensive neuromuscular examination of each patient with NMD is required to determine an effective treatment plan and to initiate rehabilitation. A neuromuscular examination often includes cranial nerve function testing, manual muscle testing, an inspection of muscle atrophy, hypertrophy, and fasciculation, observation of gait, rising from a chair, evaluation of the activity of deep tendon reflexes and the presence of pathologic reflexes, as well as assessing sensory function (3, 7). A long-term multidisciplinary approach is often required (9). This study aimed to review the demographic and clinical features, functional status, current findings, and complications of patients with NMD, and to compare these findings of three main groups: neuronopathies, neuropathies, and myopathies.

2. Matherials and Method

2.1. Study design and patients

This was a retrospective study of all eligible patients admitted to the Physical Medicine and Rehabilitation Department of a tertiary hospital between September 2020 and October 2021. The data of the patients were analyzed from the patient files. The study was performed in accordance with the Declaration of Helsinki, and approval from the local ethics committee was obtained (No: 91/05, 06.07.2020). Medical files of all hospitalized neuromuscular patients with motor neuron involvement, peripheral neuropathies, or myopathies were reviewed. Patients with (i) involvement of the neuromuscular junction and (ii) missing information in their files were excluded. A total of 46 patients, 26 (56.5%) women and 20 (43.5%) men were included in the study. These patients were divided into three groups based on the affected component of the motor unit: Group I (neuronopathy group, n=11), motor neuron diseases; Group II (neuropathy group, n=13), peripheral neuropathies; and Group III (myopathy group, n=22), myopathies.

2.2. Demographic variables and clinical features

Demographic characteristics and clinical features of the patients, including the following variables: age (year), sex (male/female), job (white collar, blue collar, housewife, retired), weight (kilogram), height (centimeter), body mass index (BMI) (kg/m²), comorbidities (yes/no), age of disease onset (year), firt presenting symptom, disease duration (year), muscle weakness (yes/no), fatigue (yes/no), spasticity (yes/no), muscle twitching (yes/no), cramps (yes/no), pain (yes/no), pain level last week by a score from 0 to 10 on the Visual Analog Scale (VAS), dysphagia (yes/no), dysarthria (yes/no), speech disorders (yes/no), independent walking (yes/no), age at which independent walking ends (year), use of an assistive or adaptive device (yes/no), use of a wheelchair (yes/no), respiratory support (yes/no), enteral nutrition (yes/no), osteoporosis (yes/no), number of falls in the last six month, and history of bone fracture (yes/no), were recorded from the patient files.

2.3. Examination findings and clinical assessment tests

Muscle strength was evaluated using the Medical Research Council (MRC) test, bilateral upper and lower extremity range of motion (ROM) measurements with a goniometer, posture analysis according to the New York Postural Rating Scale, hand grip strength evaluation with Jamar hand dynamometer and pinch meter, and dexterity with nine-hole peg test (NHPT) (10). The 6-minute walking test (6 MWT) for functional capacity (11), 5-time sit and stand test (12), one-leg standing test (13), and stair and climb test for lower extremity strength and balance (14), Brooke and Vignos scale for functional status (15), Functional Independence Measure (FIM) for daily living activities (16), Functional Ambulation Scale (FAS) for ambulation levels (17), and standardized mini-mental test for global cognitive status (18) were obtained from patient files. Pulmonary function test (PFT) measurements were also performed.

2.4. Statistical analysis

SPSS version 22.0 software (IBM Corporation, Chicago, IL, USA) was used for the statistical analysis. Categorical variables and other discrete and continuous variables were represented as percentages, numbers, and medians (min-max), respectively. Variables with a normal distribution were represented as mean±standard deviation (SD). The Kolmogorov-Smirnov test was used for data distribution analysis. Continuous and non-parametric variables were compared using the Mann-Whitney U test. Fisher's exact and chi-square tests were used to compare the categorical variables. Comparisons among groups were performed using the Kruskal-Wallis test or ANOVA. Post-hoc analysis with Bonferroni correction was used for pairwise comparisons. A p-value of less than 0.05 was found to be statistically significant.

3. Results

Forty-six neuromuscular patients who met the inclusion criteria and were followed up in the neuromuscular disease unit of our hospital were enrolled in this study. The main pathologies were presented according to the group in which patients were included: Group I: Motor neuron involvement (11 patients), 5 Amyotrophic Lateral Sclerosis (ALS), 3 poliomyelitis, and 3 Spinal Muscular Atrophy (SMA). Group II: Peripheral neuropathies (13 patients): 6 diabetic polyneuropathies, 2 Charcot-Marie Tooth (CMT) diseases, 3 chronic inflammatory demyelinating neuropathies (CIDP), and 2 Guillain-Barré syndrome (GBS). Group III: Myopathies (22 patients): 8 myotonic dystrophies, 2 polymyositis, 5 muscular dystrophies, 3 core myopathies, and 3 mitochondrial myopathies.

The study population consisted of 26 (56.5) % women and 20 (43.5%) men. The median age of the population was 46.0 years (17.0-72.0). Age of patients in Group I: 54.0(17.0-72.0) years; Group II: 52.0 (21.0-67.0) years; Group III: 38.0 (17.0-

69.0) years. There was a significant difference between the mean ages of Groups II and III (p =0.046). The median age of onset of the first symptoms was 25.5 (1.50-68.0) years, while the median age of diagnosis was 27.5 (1.5-71.0) years. When comparing the median age of disease onset in the three study groups, a significant difference was observed between Group II and III (p = 0.028). The other demographic characteristics of the patients are presented in Table 1. When comparing the disease-related data of the three study groups, as shown in Table 2, a significant difference was found in muscle weakness between Group II and III (p = 0.044). The prevalence of dysarthria and speech disorders was higher in Group III (33.3%) when compared to Group II (10%) and I (12.2%) (Table 2).

Table 1. Demographic characteristics and clinical features of patients

Variables	All patients (n=46)	Group 1 (n=11)	Group 2 (n=13)	Group 3 (n=22)	P1	P2	P3
Age (year), median (min-max)	46.0 (17.0-72.0)	54.0 (17.0-72.0)	52.0 (21.0-67.0)	38.0 (17.0-69.0)	0.593	0.183	**0.046***
Gender, n (%) Female Male	 26 (56.5) 20 (43.5)	 6 (54.5) 5 (45.5)	 7 (53.8) 6 (46.2)	 13 (59.1) 9 (40.9)	 0.974	 0.811	 0.771
Occupation,n(%) White collar Blue collar Housewife Retired	 15 (32.6) 6 (13.0) 16 (34.8) 9 (19.6)	 3 (27.2) 4 (36.4) 2 (18.2) 2 (18.2)	 3 (23.0) 2 (15.4) 4 (30.8) 4 (30.8)	 9 (40.9) 0 10 (45.5) 3 (13.6)	 0.473	 0.384	 0.065
Age of disease onset (year), median (min-max)	27.5 (1.5-71.0)	42.5 (1.5-71.0)	44.5 (8.0-61.0)	23.0 (7.0-52.0)	0.857	0.057	**0.028***
Height (cm), mean±SD	164.28±9.02	161.44±9.47	167.60±8.60	163.78±8.95	0.145	0.523	0.145
Weight (kg), median (min-max)	66.0 (42.0-100.0)	63.5 (48.0-80.0)	67.0 (42.0-100.0)	67.0 (45.0-95.0)	0.556	0.502	0.992
BMI (kg/m²), mean±SD	24.41±5.38	23.66±3.59	23.41±6.68	25.11±5.43	0.995	0.534	0.507
Comorbidities, n (%)	24 (52.2)	11 (100)	3 (23.1)	10 (45.5)	0.111	0.116	0.924

Values are mean±SD (standard deviation), median (min-max) or percentage (n,%) *p values are statistically significant (p < 0.05) and are shown in bold. Group 1: Neuronopathy group, Group 2: Neuropathy group, Group 3: Myopathy group, P1: P value between group 1 and group 2, P2: P value between group 1 and group 3, P3: P value between group 2 and group 3.

Table 2. Disease-related symptoms and clinical features of patients

Variables	All patients (n=46)	Group 1 (n=11)	Group 2 (n=13)	Group 3 (n=22)	P1	P2	P3
Muscle weakness, n (%)	42 (91.3)	10 (90.9)	10 (76.9)	22 (100)	0.229	1.000	**0.044***
Muscle atrophy, n (%)	27 (58.7)	8 (72.7)	5 (38.5)	14 (63.6)	0.090	0.440	0.179
Fatigue, n (%)	37 (80.4)	9 (81.8)	10 (76.9)	18 (81.8)	0.604	0.998	0.653
Spasticity, n (%)	4 (8.7)	2 (18.2)	1 (7.7)	1 (4.5)	0.560	0.237	0.724
Muscle twitching, n (%)	21 (45.7)	7 (63.6)	4 (30.8)	10 (45.5)	0.100	0.280	0.427
Cramp, n (%)	20 (43.5)	4 (36.4)	6 (46.2)	10 (45.5)	0.691	0.690	0.895
Pain, n (%)	28 (60.9)	8 (72.7)	8 (61.5)	12 (54.5)	0.646	0.262	0.719
Disphagia, n (%)	9 (19.6)	2 (18.2)	4 (30.8)	3 (13.6)	0.646	0.637	0.211
Dysarthria, n (%)	10 (21.7)	2 (18.2)	6 (46.2)	2 (9.1)	0.204	0.572	**0.013***
Speed disorders, n (%)	13 (28.3)	3 (27.3)	7 (53.8)	3 (13.6)	0.231	0.346	**0.015***
VAS (0-100), median (min-max)	60.0 (0.0-100.0)	80.0 (0.0-80.0)	65.0 (0.0-80.0)	30.0 (0.0-100.0)	0.548	0.095	0.232
İndependent walking (year), median (min-max)	33 (71.7)	9 (81.8)	9 (69.2)	15 (68.2)	0.594	0.380	0.677
Age of ending independent walking (year), median (min-max)	41.0 (2.0-64.0)	42.0 (42.0-42.0)	52.0 (51.0-60.0)	25.0 (2.0-64.0)	0.163	0.517	0.060
Assistive technology, n (%)	8 (17.4)	2 (18.2)	1 (7.7)	5 (22.7)	0.931	0.863	0.637
Adaptive device, n	3 (6.5)	1 (9.1)	0	2 (9.1)	0.905	0.935	0.998

(%)							
Wheelchair, n (%)	7 (15.2)	10 (90.9)	1 (7.7)	6 (27.3)	0.998	0.142	0.387
Respiratory insufficiency, n (%)	1 (2.2)	10 (90.9)	0	1 (4.5)	---	0.493	0.473
Enteral nutrition, n (%)	2 (4.3)	2 (18.2)	0	0	0.214	0.091	1.000
Falls, n (%)	21 (45.7)	5 (45.5)	4 (30.8)	12 (54.5)	0.653	0.959	0.465
Bone fracture, n (%)	8 (17.4)	4 (36.4)	2 (15.4)	2 (9.1)	0.361	0.060	0.586

Values are median (min-max) or percentage (n,%) *p values are statistically significant ($p < 0.05$) and are shown in bold. Group 1: Neuronopathy group, Group 2: Neuropathy group, Group 3: Myopathy group, P1: P value between group 1 and group 2, P2: P value between group 1 and group 3, P3: P value between group 2 and group 3. VAS: Visual Analog Scale.

The SMMT scores were significantly lower in Group I than in Group II and III (p =0.047 and 0.034, respectively). NHPT scores for the left hand were significantly longer in Group I than in Group II and III (p = 0.034 and 0.038, respectively). No significant difference was observed in other clinical findings and physical examination tests, including the Jamar hand dynamometer and pinch meter, NHPT scores for the right hand, 6 MWT, five times sit and stand test, one-leg standing test, stair and climb test, Brooke and Vignos scale, FIM scores, FAS scores, and PFT measurements (p>0.05), as shown in detail Table 3 and 4.

Table 3. Clinical assessment tests and functional tests

Variables	All patients (n=46)	Group 1 (n=11)	Group 2 (n=13)	Group 3 (n=22)	P1	P2	P3
FAS level	5.0 (0.0-5.0)	5.0 (0.0-5.0)	4.5 (1.0-5.0)	4.0 (1.0-5.0)	0.731	0.511	0.771
Brooke score	1.0 (1.10-6.0)	1.0 (1.0-5.0)	1.0 (1.0-2.0)	1.0 (1.0-6.0)	0.567	0.570	0.217
Vignos score	3.0 (1.0-10.0)	2.0 (1.0-10.0)	2.0 8(1.0-9.0)	3.0 (1.0-10.0)	0.616	0.452	0.845
FIM-Motor	88.0 (21.0-91.0)	85.0 (33.0-91.0)	86.0 (42.0-91.0)	90.0 (21.0-91.0)	0.723	0.929	0.706
FIM-Cognitive	35.0 (18.0-35.0)	35.0 (35.0-35.0)	35.0 (30.0-35.0)	35.0 (18.0-35.0)	0.715	0.273	0.514
FIM-Total	121.0 (56.0-126)	120.0 (68.0-126.0)	121.0 (72.0-126.0)	122.0 (56.0-126.0)	0.681	0.853	0.773
SMMT score	28.0 (26.0-30.0)	27.0 (24.0-30.0)	29.5 (29.0-30.0)	30.0 (27.0-30.0)	0.047	**0.034***	0.737
Jamar-right hand (kg)	16.63 (2.83-36.3)	21.43 (3.2-36.3)	17.0 (11.6-32.3)	13.06(2.83-22.66)	0.882	0.054	0.117
Jamar-left hand (kg)	14.58(1.0-34.0)	24.15 (1.0-34.0)	18.0 (9.40-31.6)	10.36(2.40-24.33)	0.834	0.088	0.193
Pinchmeter-right hand (kg)	7.33 (2.6-23.0)	7.75 (3.0-23.0)	7.5 (2.60-14.83)	6.41 (2.66-17.33)	0.472	0.175	0.675
Pinchmeter-left hand (kg)	7.33 (2.5-22.6)	7.50 (2.5-22.6)	7.5 (3.0-12.0)	7.16 (2.66-14.33)	0.250	0.087	0.791
Nin-hole peg test-right (sec)	26.0 (19.28-43.70)	27.30 (20.0-43.7)	24.9 (21.0-39.0)	25.0 (19.28-43.0)	0.532	0.400	0.693
Nine-hole peg test-left (sec)	26.0 (19.0-65.0)	29.3 (20.0-65.0)	24.37 (21.0-27.0)	26.5 (19.0-35.0)	0.034	**0.038***	0.557
5-time sit-to-stand test (sec)	17.0 (0.0-60.0)	19.5 (10.0-54.0)	17.26 (11.0-60.0)	14.0 (0.0-43.0)	0.784	0.269	0.419
Stair-climb test-up (sec)	5.0 (2.52-24.0)	4.31 (3.0-10.0)	8.0 (3.0-20.0)	5.0 (2.52-24.0)	0.362	0.689	0.551
Stair-climb test-down (sec)	4.0 (2.0-57.0)	3.60 (2.80-9.60)	3.0 (2.0-14.0)	4.08 (2.0-57.0)	0.900	0.621	0.485
One-leg standing test-right (sec)	6.0 (0.0-77.0)	6.0 (0.0-69.0)	3.5 (0.0-77.0)	12.0 (0.0-52.0)	0.996	0.983	0.980
One-leg standing test-left (sec)	4.5 (0.0-180.0)	4.75 (0.0-14.0)	2.5 (0.0-180.0)	8.0 (0.0-57.0)	0.246	0.687	0.343
6MWT (m)	330.0 (9.0-630.0)	225.0 (9.0-560.0)	280.0 (30.0-510.0)	349.0 (90.0-630.0)	0.859	0.237	0.325

Values are median (min-max). *p values are statistically significant ($p < 0.05$) and are shown in bold. Group 1: Neuronopathy group, Group 2: Neuropathy group, Group 3: Myopathy group, P1: P value between group 1 and group 2, P2: P value between group 1 and group 3, P3: P value between group 2 and group 3. FAS: Functional ambulation scale, FIM: Functional Independent Measure, SMMT: Summarized Mini-Mental Test, 6MWT: 6 Minutes Walking Test. Kg: kilogram, sec: second, m: meter.

Table 4. Respiratory symptoms and pulmonary functional tests

Variables	All patients (n=46)	Group 1 (n=11)	Group 2 (n=13)	Group 3 (n=22)	P1	P2	P3
Cough, n (%)	27 (58.7)	5 (45.5)	7 (53.8)	15 (68.2)	0.462	0.209	0.117
Respiratory tract infection, n (%)	3 (6.5)	0	1 (7.7)	2 (9.1)	0.998	0.328	0.907
FEV1, median (min-max)	78.9 (49.5-120.0)	120 (120.0-120.0)	90.95 (78.9-103.0)	76.05 (49.5-98.0)	0.397	0.046	0.267

FVC, median (min-max)	78.0 (53.4-116.0)	116.0 (116.0-116.0)	88.7 (78.0-99.4)	75.45 (53.4-106.0)	0.380	0.130	0.550
FEV1/FVC, median (min-max)	86.0 (73.0-92.0)	86.5 (86.5-86.5)	86.35 (85.7-87.0)	82.5 (73.0-92.0)	0.133	0.704	0.599

Values are median (min-max) or percentage (n,%). P values are statistically significant ($p < 0.05$). Group 1: Neuronopathy group, Group 2: Neuropathy group, Group 3: Myopathy group, P1: P value between group 1 and group 2, P2: P value between group 1 and group 3, P3: P value between group 2 and group 3.

4. Discussion

We aimed to review the clinical features of patients diagnosed with NMD and compare the symptom findings of three main groups: neuropathies, neuronopathies, and myopathies. In this study, fatigue and muscle weakness were the most common symptoms shared by all groups. The mean age of the disease onset was lowest in the myopathy group. Dysarthria was most common in the neuropathy group. Cognitive impairment was most common in the neuronopathy group. Although there was no statistically significant difference between the three groups, the use of assistive technology was most common in the myopathy group, dysphagia was most frequent in the neuropathy group, pain was most common in the neuronopathy group. While the frequency of falls was highest in the myopathy group, the highest fracture rate was observed in the neuronopathy group.

In the present study of the three main groups of NMD, it was observed that the median age and diagnosis age of patients with myopathies (Group III) were significantly lower than in the neuropathy group ($p=0.044$ and 0.028, respectively). Disease onset and progression may differ in each group of NMD. Myopathies can be described according to their clinical manifestations, onset, histopathological features, or eponyms (19). In this study, except for two patients with polymyositis, all other patients had congenital types of myopathies in Group III. Therefore, the median age and age at diagnosis were lower in the myopathy group than in the other groups.

There are approximately 600 different NMDs that can be inherited or acquired despite differences in etiology and severity, and all NMDs share (progressive) muscle weakness as a clinical feature (20). In the present study, muscle weakness was the most common symptom in patients with myopathy, neuronopathy, and neuropathy (100%, 90.9%, and 76.9%, respectively). Muscle weakness was significantly higher in the myopathy group than in the neuropathy group ($p = 0.044$). The distribution of muscle weakness usually indicates the primary affected area within the neuromuscular system (21). Weakness of the proximal muscle groups, such as the hip or shoulder girdle, is generally due to myopathy or neuromuscular junction (NMJ) disorders, with few exceptions. Conversely, weakness of the distal muscle groups is usually, but not always, due to neurogenic causes, such as peripheral polyneuropathy or amyotrophic lateral sclerosis (4). Proximal weakness often first appears in the muscles of the lower extremities. Patients usually present with complaints of difficulty walking and climbing stairs. Proximal weakness in the upper extremities manifests as an inability to raise the arms above the shoulders. Limb weakness is the most common symptom, but there are several other symptoms, including weakness in the speaking, swallowing, and breathing muscles (7, 22). Muscle pain, cramps, extreme fatigue, atrophy or pseudohypertrophy, fasciculation, and stiffness are other significant symptoms that can be seen in NMD (8).

Fatigue was the most frequently reported symptom in patients with NMD (80.4%) in this study. Fatigue can be acute, chronic, central, or peripheral. According to some studies, as many as 80% of patients with NMD can report fatigue, similar to our study (23-25). Several studies have reported fatigue as an essential symptom, especially in NMD such as ALS, post-poliomyelitis syndrome, CIDP and GBS, CMT hereditary sensitive-motor neuropathies, and NMJ diseases (26-30). Fatigue may also be seen in NMD as the first symptom before any motor deficit develops. For example, fatigue is evaluated at onset because it is a crucial factor in diagnosing metabolic myopathies (31).

In this study, the incidence of dysarthria and dysphagia was highest in the neuropathy group (30.8% and 42.6%, respectively). A few studies have examined the prevalence of dysphagia and dysarthria in patients with NMD (32-34). In a study by Knuijt et al. (33) a prevalence of dysphagia of 36–58% and dysarthria of 46–62% was found in adult patients with NMDs. In the same study, there was a moderate but notable association between dysphagia and dysarthria ($rs = 0.40$; $p < 0.01$). While dysphagia is usually mild, dysarthria is moderate to severe in 15% of dysarthric patients. In a study by Audag et al. (32) dysphagia was observed in 45% of all NMD patients screened for dysphagia with the Sydney Swallow Questionnaire (SSQ). The median SSQ scores were higher than the cutoff value in patients with myotonic syndromes, ALS, and facioscapulohumeral dystrophy. In the literature, dysphagia in patients with polyneuropathy has been reported frequently in critical illness polyneuropathy and Guillain-Barre syndrome, especially in patients with prolonged requirements for orotracheal intubation and tracheostomy (35-37).

In this study, pain was most frequently observed in the neuronopathy group (72.1%). There are few studies investigating the prevalence of pain in motor neuron diseases, and they reported that the frequency of pain varies between 15% and 84% (38, 39). Pain can occur at all stages of the disease and is reported to be mild to moderate in severity (38). There are some differences between pain levels in the gradually progressing forms of NMD. It is seen that the intensity of pain is high in neuropathic diseases such as CMT, especially in demyelinating forms, and diabetic neuropathy

(40, 41). In addition, the most frequent forms of muscular dystrophy, myotonic type 1, and facioscapulohumeral are also present in painful NMD (42).

NMD significantly affects the affected person's independence in daily changes. Their gradually progressive nature reduces their functional capacity, and they need more support over time; this may be due to the caregiver and/or assistive devices. Therefore, this assistance is essential for maintaining a minimum level of personal autonomy (43, 44). In our study, the use of assistive technology was highest in the myopathy group (22.7%). With the progression of muscle weakness, more caregiver assistance, assistive devices, and environmental regulations are required for people with NMD to perform their professional activities and maintain their level of daily activities (43). Studies on the use of assistive technology in the literature have been most frequently performed in patients with Duchenne muscular dystrophy (45, 46). Falling is one of the most common clinical problems in patients with NMD. In our study, the highest rate of falls was observed in the myopathy group, whereas the highest rate of bone fracture was observed in the neuronopathy group. Data from case-control studies involving patients with axonal polyneuropathy and polio have shown an increased incidence of falls in specific patient populations (47, 48). In a study investigating fracture risk in NMD, 47% of the patients were shown to have a moderate to high fracture risk. In the same study, two-thirds of the patients were not included in the osteoporosis screening and treatment program. The authors stated that patients with NMD should be screened routinely for osteoporosis, and in this way, early treatment can be achieved to reduce the risk of fragility fracture (49).

The effects of NMD on the spectrum of cognitive function are not yet understood. While NMD is known to affect motor functions the most in patients, the cognitive effects of these conditions may be significant (50). In this study, the patients' SMMT scores were significantly lower in the neuronopathy group than in the neuropathy and myopathy groups ($p = 0.047$ and 0.034, respectively). In a study by Phukan et al. (51) comorbid dementia was observed in approximately 14% of newly diagnosed ALS patients. In the same study, cognitive impairments were observed in more than 40% of ALS patients without evidence of dementia. In a review by Consonnia et al (52), older age, rapidly progressing ALS, bulbar-onset, advanced disease stages were reported as related factors mainly associated with cognitive involvement. In the management of patients with motor neuron disease, it should be kept in mind that cognitive disorders are common, and patients should be evaluated in this respect.

NMD is a broad group of diseases separated into three main groups, and the most common symptoms and clinical findings were evaluated. In this study, fatigue and muscle weakness were the most common symptoms shared by all groups. The mean age of the disease onset was lowest in the myopathy group. Dysarthria was most common in the neuropathy group. Cognitive impairment was most common in the neuronopathy group. Although there was no statistically significant difference between the three groups, the use of assistive technology was most common in myopathy group, dysphagia was most frequent in the neuropathy group, pain was most common in the neuronopathy group. The frequency of falls was highest in the myopathy group, and the fracture rate was highest in the neuronopathy group. We believe that such demographic and clinical studies will be beneficial in promoting effective healthcare services for these patients and appropriate rehabilitation programs at each stage of the disease.

Conflict of interest

The authors declared no conflict of interest.

Funding

No funding was used for the study.

Acknowledgments

We acknowledge Mrs Fatma Ballı and Zeynep Aykın for their assistance in data collection.

Authors' contributions

Concept: Ö.Z.K., Design: Z.T.B., Data Collection or Processing: Y.T.Y., Analysis or Interpretation: D.C., Literature Search: E.U., Writing: Z.T.B.

Ethical Statement

The ethical committee of Dışkapı Yıldırım Beyazıt Training and Research Hospital approved the study protocol (Ethics Code; 91/05, 06.07.2020). All patients gave verbal consent as this was a retrospective study with an interview of the patient and a review of his medical file. No interventions were applied to the participants, and the institutional review board approved the study based on this verbal consent.

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