Chapter 11

Neuromuscular Diseases and Rehabilitation

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Additional information is available at the end of the chapter

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Abstract

Neuromuscular diseases (NMDs) are a heterogeneous group of diseases that are inherited or acquired, resulting from an abnormality in the anterior horn motor cells, peripheral nerves, neuromuscular junctions, or muscles.

In NMDs, evaluation is performed to monitor the progression of the disease, to determine the appropriate treatment methods, to investigate the efficacy of treatment methods, and to predict and prevent possible complications. Evaluation methods should include evaluation of respiratory functions, muscle testing, normal joint movement, evaluation of flexibility, evaluation of motor functions and functional capacities, functional posture, and gait analysis.

The aim of physiotherapy approaches is to improve the quality of life of patients and their families. The rehabilitation program should include the protection of the functional level of the patient and the physical and psychological functions, increasing the physical and mental capacity of the patient, and slowing the progress of the disease symptoms. The rehabilitation program should include the preservation of muscle strength, an exercise program for the prevention of contractures, increasing respiratory function and aerobic capacity, gait and balance training, fall prevention, walking aid training, psychosocial approach, vocational counseling, ergotherapy processes and nutrition expert support.

Keywords: neuromuscular diseases, physiotherapy, rehabilitation, ICF, assessment

1. Introduction

1.1. Identification and classification of NMD

Neuromuscular diseases (NMDs) are a heterogeneous group of diseases that are inherited or acquired, resulting from an abnormality in the anterior horn motor cells, peripheral nerves, neuromuscular junctions, or muscles [1]. The most common neuromuscular diseases are

motor neuron diseases, neuropathies, neuromuscular junction diseases, and muscular diseases based on anatomic localization [2].

Motor neuron diseases are a group of diseases that progress with lower and/or upper motor neuron involvement in motor neurons in the anterior horn of medulla spinalis. They are characterized by muscle weakness associated with fasciculation and atrophy that differs with respect to the location and function of the affected motor neurons. Hereditary spinal muscular atrophy (SMA) is the most common motor neuron disease. SMA is caused by the degeneration of the anterior horn cells in the spinal cord and brain stem and by the activation of the corticospinal tract. Progressive symmetrical weakness, hypotonia, hyporeflexia or areflexia, muscle atrophy, fasciculation typically affecting the tongue, and postural tremor in the fingers are typical signs and symptoms of SMA. Spasticity may also be seen as a disease-specific finding in patients with upper motor neuron involvement such as amyotrophic lateral sclerosis (ALS) [3].

Neuropathies are peripheral nerve diseases with sensory and motor symptoms. The observation of sensory changes in addition to muscle weakness distinguishes peripheral nerve diseases from diseases of other components of the motor unit. It causes demyelination in the nerve and axonal degeneration in the nerve by affecting the nerve myelin sheath and/or axon [4]. The most common neuropathies are hereditary, but there are also different syndromes such as inflammatory, toxic, and infectious neuropathies [2]. The loss of muscular strength and muscular atrophy is observed starting from feet and legs in the lower extremity and hands in the upper extremity, while paresthesia/dysesthesia is observed in a stocking-glove distribution [5]. The most common Charcot-Marie-Tooth (CMT) disease or hereditary motor and sensory neuropathy constitutes a genetically heterogeneous group of diseases affecting the peripheral nervous system. CMT, which is characterized by the abnormal development or degeneration of the peripheral nerve, has different genetic pattern transitions. The disease begins during infancy in many cases. Symptoms include inadequate gait, muscular atrophy and weakness progressing from the distal to proximal extremities, foot deformities such as cavus deformity, deep tendon reflex loss, and sensory loss in the distal extremities. Inappropriate gait may be evident as walking with jumping and there may be a prone to falling [6–8]. Hereditary neuropathies are diseases affecting peripheral nerves and classified as hereditary motor and sensory neuropathies (HMSNs), hereditary motor neuropathies, hereditary sensory neuropathies, and hereditary sensory and autonomic neuropathies. Autonomic neuropathies include loss of sweating, bladder dysfunction, constipation, and impotence in males [5, 9].

Neuromuscular junction diseases are autoimmune diseases that result from the destruction, impairment, or absence of one or more proteins during neuromuscular transmission. Most neuromuscular junction diseases are acquired and occur associated with presynaptic, synaptic, and postsynaptic disorders. Myasthenia gravis (MG), the result of a postsynaptic disorder, is the most common neuromuscular transmission disorder [10, 11]. MG has two clinical forms: ocular and generalized. Weakness in the ocular form is limited to the

eyelids and extraocular muscles. In generalized myasthenic patients, however, there is also a weakness in the bulbar, extremity, and respiratory muscles in varying degrees due to the cranial involvement. As a result, ptosis, diplopia, dysphagia, and dysarthria are observed. Myasthenic weakness typically fluctuates during the day, usually the least in the morning, and worsens as the day progresses with prolonged use of muscles, especially those that are stiff [12–15].

The main problem in muscle diseases involves the degeneration in muscle rather than in the nerve. It is a group of genetic diseases proceeding with progressive muscle weakness that causes subsequent limitation of joint movements, shortness of muscles, a decrease in respiratory capacity, and posture disorders. The loss of function in the body and in the upper and lower limb, impairment of organization of postural reactions, fatigue, loss of cardiopulmonary adjustment, and disturbed psychosocial condition are among the clinical and functional problems encountered [16, 17]. Different functional levels and clinical characteristics may be observed ranging from minimal influence to being confined to bed depending on the type of disease, the age of onset, and the location of the affected muscle group (proximal and/or distal). Bed confinement is observed in the early period in Duchenne musculoskeletal dystrophy (DMD) and Becker muscular dystrophy (BMD). However, facioscapulohumeral musculoskeletal dystrophy (FSHMD) and limb-girdle muscular dystrophy (LGMD) progress slowly and decrease the patient's functional ability and quality of life by causing scoliosis, wing scapula, difficulty in going up or down the stairs, toe walking and lordotic posture, difficulty in standing up from a sitting or squatting position, and Gower's sign. Gower's sign indicates the weakness of the proximal muscles, namely those of the lower limb. The sign describes a patient that has to use their hands and arms to "walk" up their own body from a squatting position due to lack of hip and thigh muscle strength, and contractures are observed [18]. In many types of diseases, the decreased pulmonary function is due to the respiratory muscle weakness and spinal deformities (kyphoscoliosis), which leads to respiratory tract infections and respiratory disorders. Cardiomyopathy and arrhythmia lead to cardiac failure and this cardiorespiratory complications cause death [19]. In the literature, various classifications were done according to the rate of progression of the disease, the affected area, and the body part involved, but the most recent classification was done according to the Belgian Neuromuscular Disease Registry [20]. The classification of neuromuscular diseases is given in **Table 1**.

According to the international classification of health, function, and disability (ICF) by the World Health Organization (WHO), neuromuscular disorders are associated with disability in body structure and function, resulting in problems with activity and participation [21]. These problems in muscle diseases can be addressed in two parts: primary and secondary. Primary disorders are muscular pain, atrophy, pseudohypertrophy, myotonia, and the loss of postural control, while the secondary ones are fatigue, difficulty in transfer activities and mobility problems, exercise intolerance, contractures, respiration, and psychological problems. The problems observed in the patient are related to the type of disease, pathogenesis, and progression of illness [22].

1. Muscular dystrophies	2. Myotonic and relaxation disorders	3. Myopathies	4. Disorder of the neuromuscular transmission	5. Disorder of the motor neurons	6. Neuropathies
1. Congenital muscular dystrophy	1. Thomsen-type myotonia congenita	**a. Congenital myopathies**	1. Myasthenia gravis	1. Amyotrophic lateral sclerosis	**a. Hereditary**
2. Duchenne muscular dystrophy	2. Becker-type myotonia congenita	1. Central core disease	2. Congenital myasthenia	2. Primary muscular atrophy	1. Hereditary motor and sensory neuropathy
3. Becker muscular dystrophy	3. Paramyotonia congenita	2. Multiminicore disease	3. Lambert-Eaton syndrome	3. Postpolio syndrome	2. Hereditary neuropathy with liability to pressure palsies
4. Dystrophinopathy	4. Familial periodic paralysis	3. Nemaline myopathy	4. Other disorders of neuromuscular transmission	4. Primary lateral sclerosis	3. Hereditary sensory & autonomous neuropathy
5. Facioscapulohumeral dystrophy	5. Other myotonic disorders	4. Myotubular		5. Werdnig-Hoffman spinal muscular atrophy	**b. Inflammatory**
6. Limb girdle muscular dystrophy		**b. Myopathy**		6. Intermediate spinal muscular atrophy	4. Guillain-Barré syndrome
7. Emery-Dreifuss muscular dystrophy		5. Centronuclear myopathy		7. Kugelberg-Welander spinal muscular atrophy	5. Chronic inflammatory demyelinating polyneuropathy
8. Distal myopathy		6. Fiber-type disproportion myopathy		8. Adult spinal muscular atrophy	6. Multifocal motor neuropathy
9. Oculopharyngeal muscular dystrophy		7. Metabolic myopathies		9. X-linked bulbo-spinal muscular atrophy or Kennedy's disease	7. Vasculitis
10. Myotonic dystrophy type 1		8. Muscle glycogenoses		10. Distal spinal muscular atrophy	8. Neuropathy associated with paraproteinemia
11. Myotonic dystrophy type 2		9. Disorders of fatty acid metabolism mitochondrial myopathy		11. Hereditary spastic paraplegia	9. Neuropathy associated with plasma cell dyscrasia
12. Other muscular dystrophies		**c. Inflammatory myopathies**		12. Other disorders of motor neurons	10. Amyloisdosis
		10. Polymyositis			11. Neuropathy in systemic disease
		11. Dermatomyositis			12. Other neuropathies
		12. Inclusion body myositis			7. Hereditary ataxias and others
		13. Other myopathies			

Table 1. Classification of neuromuscular diseases [20].

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2. Body structure and function disorders in NMD according to the international classification of function

2.1. Loss of strength

The progressive loss of strength, severity of which changes depending on the type of the disease, is one of the leading problems that constitute the deficiencies seen in neuromuscular diseases. The functional deficiencies seen in neuromuscular diseases vary depending on the localization of affected muscle groups and secondary outcomes caused by muscle weakness vary depending on the type and progression of the disease but it should be remembered that the severity of the disease may vary due to the individual differences among the patients [23]. The loss of strength can be seen in the distal and/or proximal region. Also, the loss of strength can also be seen in the neck and mimic muscles. The reason for the progressive loss of muscle strength in neuromuscular diseases varies according to the nature of the disease. It is due to the reduction in the number and size of intrinsic contractile fibers in muscle diseases that are followed by replacement of these fibers by fat infiltration and connective tissue. These changes are caused by the disturbance in the nerve stimulation and transmission pathways necessary for muscle contraction [7, 18]. Also, the decrease in the optimal length of the muscle and deterioration of the sarcomere structure resulting from decreased physical activity secondary to the disease is among the causes of the development of immobilization. It has been shown in the literature that the muscle ceasing to the contraction has lost half of its strength after 3–5 weeks [24]. For example, patients with FSMD typically show weakness around the shoulder and facial muscles, thereby weakening the activities involving the upper extremity for function, while DMD typically shows weakness around the hips of the patients. Thus, patients have difficulty in activities involving the lower extremities [23, 25]. Approximately 75% of muscle patients have muscle strength loss in the proximal limb of the extremity, 20% in the facioscapulohumeral, and 4% in the scapuloperoneal part of the body [26, 27]. However, in acquired diseases (such as inflammatory myopathy), a loss of strength occurs subacutely. Some neuromuscular diseases affect specific muscles. FSHMD should be considered if there is asymmetric loss of strength in the muscles around the scapula, humerus, facial, and mimic muscles. Myotonic dystrophy (MD) should primarily be considered if there is involvement of the frontal and facial muscles, as well as the sternocleidomastoid muscle and the distal (especially tibialis anterior) muscles [26].

In CMT disease, in which foot and ankle problems and especially muscle weakness are commonly seen, weakness typically begins in the intrinsic muscles of the foot and follows the peroneus brevis and longus, tibialis anterior, extensor digitorum longus, and extensor hallucis longus muscles. This weakness pattern causes the muscle imbalance; while the plantar flexors remain relatively strong, the dorsiflexors weaken and consequently leads to the contraction of the Achilles tendon. As a secondary to this muscle imbalance, together with calcaneus inversion, forefoot adduction, and claw toe, pes cavus deformity develops typically in CMT disease [28, 29]. Also, symptoms such as drop head, ptosis, dysphagia, and dysarthria can be seen in oculopharyngeal muscular dystrophy [18]. In patients with peripheral neuropathy, however, the loss of muscle strength has been shown to be more prominent in the foot dorsiflexors, knee extensors, and hip flexor muscles [30].

2.2. Postural control and balance problems

The strength necessary for the movement of the extremities is generated, collected, and transferred to the upper extremity from the lower extremity by the postural control. Thus, the segments from the proximal to distal that are independent of each other operate in a specific interaction and concordance during functional activities [31].

The efferent system is composed of the structures necessary for postural control including vestibular, visual, and somatosensory inputs. The somatosensory input disorders can disrupt the postural control and lead to falls in neuromuscular diseases, which causes various conditions such as the weakness of the proximal or distal extremity (like patients with myopathy), axial weakness (myositis or amyotrophic lateral sclerosis), stiffness (myotonia), slow muscle contraction (nemaline myopathy), intermittently varying weakness (myasthenia gravis), sensory polyneuropathic end-effector proprioceptive (myasthenia gravis), and sensory polyneuropathy resulting in proprioceptive insufficiency [32–34]. While the efferent system is also necessary to maintain a similar upright stabilization, it provides effective postural correction after perturbations during posture. The inadequacy of the efferent system can disrupt balance control and cause falls [35].

One of the problems that threaten postural control in neuromuscular diseases is the progressive nature of muscle weakness; the other is the inactivity due to the loss of ambulation following the progression. In both cases, spinal stabilization is affected due to the motor and postural reasons, leading to spinal problems. In cases where the ambulation continues despite the decrease in muscular strength, many postural problems such as lordosis, kyphosis, scoliosis, and wing scapula are observed due to the increase in the compensatory responses in the body [32, 36].

Scoliosis develops before the loss of ability to walk in 30% of muscle patients. The restriction of the paraspinal muscles causing the lordotic sitting posture in extension emerges as a result of locking of the posterior facets of the vertebral bone and the vertebral bones remaining flattened [37]. In addition, kyphotic sitting position is also preferred by patients as a result of the weakened paraspinal muscles. This kyphotic position prevents the locking of the posterior facets of the vertebral bones and causes the opening of the joint faces, vertebral rotation, and lateral curve formation. Finally, the functional scoliosis develops as a result of the inability of the vertebrae to resist to the gravity, the multifaceted adverse influencing of the posture, and difficulty in controlling this effect [38–40].

One of the major problems in the clinical postural control is pelvic obliquity. It affects the sitting balance and causes increased pressure on the lower ischial tuberosity, making the sitting position uncomfortable [41]. Also, the hip joint left on the higher side in the pelvic obliquity tends to be subluxated. Subluxation of the hips may also be observed secondary to progressive muscle weakness [42]. The increasing pelvic obliquity affects spinal stabilization. This condition affects the sitting balance of the patients and causes complaints of pain [43].

Hip movements are an important factor in correcting lateral balance; hence, proximal muscle weakness can disrupt the ability to balance during external shocks. Distal muscle weakness can lead to falls through different ways; the obstacles that do not look dangerous in normal

barrier-free environments can cause the patients with dropped foot to stumble and lose balance, increasing the risk of falls. In studies, it is emphasized that the rotations and movements of the ankles during standing is essential for maintaining and correcting the balance [44–47].

The wing scapula is observed in patients due to power loss in the shoulder girdle. Wing scapula adversely affects upper extremity functions [48]. Kyphosis and dropped head syndrome have been reported in some myopathies, particularly due to loss of strength in the neck extensor muscles. Patients with dropped head syndrome experience severe walking difficulty with loss of spinal smoothness [49, 50].

Proprioception plays an important role in stabilizing the body during both comfortable posture and unexpected postural perturbations. Therefore, patients with peripheral neuropathy are unstable when their eyes are closed during standing. In addition, reflex responses to postural perturbations are either delayed or decreased in amplitude or both [51–53].

In addition to the above-mentioned findings that are directly related to postural control, postural control plays an important role in many activities such as stair climbing, wheelchair activities, writing, bathing, makeup, shaving, eating, toilet needs, and in-bed mobility [54, 55].

2.3. Atrophy

In this disease group that proceeds with progressive muscle weakness, the patients become more inactive and sedentary in time because the muscles are the active structures responsible for movement in the body. It is stated in the literature that atrophy develops between 14 and 17% of muscle fiber after 72 h of immobilization [56]. Atrophy in muscular diseases develops later compared to peripheral nerve diseases. Selective atrophy of certain muscles may be associated with disease-specific disorders [27].

2.4. Pseudohypertrophy

Pseudohypertrophy is a false hypertrophy seen in the muscle. The hypertrophic appearance of fibrils in the affected muscles results from the replacement of the fibrils by the fat and connective tissue. It does not cause a real increase in the strength of the muscle even though the muscle volume increases [57]. The hypertrophy in the muscle fibers that have not yet been lost is accompanied by the increase in the fat and connective tissue, and thus muscle mass increases. It is most commonly seen in the gastrocnemius-soleus muscle group, occasionally in quadriceps, biceps brachii, and deltoid muscles. In addition, the presence of atrophic muscles around the muscles of the pseudohypertrophy may exaggerate this enlarged image [26].

2.5. Myotonia

Clinically, myotonia refers to any condition that prevents the relaxation of the muscles after contraction. The relaxation difficulty is evident in the first movement of the muscle after its resting position. Relaxation difficulty is reduced when the same movement is repeated after which the movements become easier. Although this is the only symptom in some diseases, in myotonic dystrophy it is associated with permanent muscle strength loss [26]. Myotonia seen

in MD and ALS is a factor affecting patients' lives negatively in activities such as handshaking and jar opening.

2.6. Pain

Pain is an important problem in most of NMDs, but it is not typically a direct consequence of disease, and now researchers agree that chronic pain is a common symptom that can be seen in all forms of NMDs [58]. In a study on 511 NMPs involving DMD, BMD (myotonic musculoskeletal dystrophy (MMD), metabolic myopathy, FSHMD, and MG), Guy-Coichard et al. evaluated the frequency, characteristics, and effects of pain; they found that the pain was moderate to severe in NMDs and emphasized that pain should be regularly assessed in this patient group [59]. Pain observed in neuromuscular diseases is the result of progressive muscle weakness, fatigue, ligament laxity or stretching, and abnormality of walking and posture [60]. In neuropathic conditions, however, pain-causing mechanisms are neurogenic inflammation, abnormal involvement of the sympathetic nervous system, and the neuroplasticity changes in the central nervous system. In a study aiming to determine the structure and location of pain in muscle patients, 73% of the patients complained of pain, and 27% of them were found to have severe pain [58]. In another study involving the evaluation of pain in 125 patients with neuromuscular disease from different groups, 73% complained of pain, 62% had chronic pain, and 15% had severe pain. The localization of the pain was reported to be spinal column in 81%, shoulder girdle in 54%, hip in 47%, and knee in 47%. In addition, 67% of these patients reported an increase in pain with walking and 68% with standing [60].

2.7. Contracture

Joint contractures, subluxations, and dislocations are common problems in NMDs [61, 62]. When the strength of the muscles around the same joint is different from each other, the joint tends to remain at a certain position, the corresponding muscle becomes shortened, and this position of the joint is fixed in time, resulting in the formation of contracture. The development of contracture occurs in patients with NMDs in time. The stiffness, which develops in the tendon and increases until contracture occurs, can be prevented or reduced by exercise or splints holding the joint in the opposite position [22, 26].

Flexion contracture is seen in hip joint with loss of strength of the hip extensor. The lumbar lordosis is increased so that the upright posture can be sustained and the loss of strength can be compensated. However, the progression of the weakness in the muscles of the pelvic girdle causes the knee flexion contracture to develop [62]. The increasingly apparent loss of strength in the gluteus maximus and quadriceps muscle leads to an excessively increased lordosis. As the patient tries to stabilize hips in the extension, lordosis becomes more pronounced. As the muscles weaken, the patient tries to increase stabilization by pulling the arms back and increasing the lordosis to pull the center of gravity behind the hip joint. Due to the loss of dorsiflexor muscle strength, plantar flexion contracture develops in the ankle joint, resulting in toe walking [48].

2.8. Cardiopulmonary problems

Neuromuscular diseases cause respiratory problems mainly due to the inadequacy of the respiratory muscles, namely upper airway muscles (mouth and tongue muscles), external intercostal muscles, diaphragm, and abdominal muscles. The restrictive type of respiratory problem is observed in particular. Along with the involvement of the primary respiratory muscles in Becker MD and FSHMD, respiratory problems are encountered. The compliance of the chest wall is reduced, and then, there becomes a decrease in the total lung capacity. The disease may also cause kyphoscoliosis that increases the respiratory problem. Muscle fatigue in MG bears respiratory problems, whereas bulbar involvement in ALS causes increased secretion, inability to close glottis, and decreased respiratory control. In MM dystrophy and congenital myotonic dystrophy, a direct cardiac involvement is in the form of cardiomyopathies observed in addition to respiratory problems [63, 64]. Respiratory disturbances and cardiomyopathy may follow the development of scoliosis [65]. Respiratory muscle strength is insufficient in muscle patients. In particular, the maximal inspiratory (MIP) and expiratory (MEP) pressures are below normal values [66, 67].

The respiratory muscle weakness that emerges in the late stages of any neuromuscular disease results in hypoventilation and hypercapnia. This causes restrictive pulmonary impairment and reduced exercise capacity [68, 69]. Moreover, during terminal stages of all diseases with muscle involvement, the weakness in the respiratory muscles restricts coughing. This problem may result in difficulty in swallowing and aspiration pneumonia. This in turn may cause the mechanical ventilator to be connected [69]. While cardiac involvement is mostly a result of respiratory involvement, it can be primarily seen as cardiac muscle involvement in some NMDs. In patients whose heart muscle is affected, the heart will have difficulty adapting to changes in the body in the event of any increase in physical activity [70, 71].

The presence of cardiomyopathy resulting from a lack of dystrophin in the myocardium and cardiac Purkinje fibers affects the cardiopulmonary response to exercise. Myocardial dysfunction remains silent due to decreased physical activity until the end of the disease process. The effect of cardiomyopathy and restrictive pulmonary disease on physical activity is much greater in NMDs with slower progression [1].

3. Activity problems in NMD

Although most neuromuscular disorders have progressive and clinically distinct features, the most important common feature is that they lead to functional problems and activity limitations at various levels [72]. The activity limitations due to the functional deficits vary depending on the localization of the affected region, on the type of the disease with secondary outcomes caused by loss of strength, and even on the patient [22]. The patients with muscle diseases usually go to the doctor with complaints such as difficulty in climbing stairs or a slope, getting up from the sitting position, walking, raising arms and reaching up, and washing the head. The patients may also complain about numbness in feet and hands and not

being able to open eyes, and difficulty in swallowing. At the end of the natural progression of the disease, there may be a loss of strength in both the proximal and distal muscles and the inability to perform many activities such as wheelchair activities, writing, bathing, applying makeup, shaving, eating, toilet needs, and mobility within the bed [73].

Fatigue, cardiopulmonary effect, and exercise intolerance cause an increase in fat mass and contractures. It also causes a decrease in the efficient use of locomotion (such as reduced walking space and more energy expenditure), a decreased motivation of the patient, a reduction in support of the social environment for activity, an increase in depression, and social barriers. These restrictions all result in a decline in physical activity performance in NMDs [1]. The progressive muscle weakness present in NMD directly affects the daily activities of the patient. Depending on the degree of muscle weakness in the proximal part of the body, patients may have difficulty in fine hand skills such as self-care, dressing, and hygiene. The patients may also have difficulty in transfers requiring upper extremity support, ambulation activities, reaching to mouth due to the weakness of the distal muscle groups, bilateral use of cutlery, and nail clipping [74]. In a study involving 208 neuromuscular patients, Pieterse et al. have grouped the activities that the patients had difficulty under the following headings: (1) communication, (2) eating and drinking, (3) transfer, (4) walking and moving around, (5) transportation, (6) lifting and carrying, (7) fine hand skill, and (8) use of arms higher than shoulder level [75].

3.1. Fatigue

More than 60% of patients with NMD complain about severe fatigue along with muscle weakness and many other problems. Fatigue has a significant effect on exercise limitation and includes the physiological fatigue and the experienced fatigue [76]. The physiological fatigue is defined as an exercise-dependent reduction in maximal voluntary muscle strength while the experienced fatigue is defined as difficulty in initiation and maintenance of voluntary activities [77, 78]. The physiological fatigue is the one with both peripheral and central components, either muscular or related to limitation initiated in the central nervous system (CNS). Many mechanisms may cause this fatigue and exercise limitation [79]. In neuromuscular diseases, there is the fatigue (peripheral fatigue) stemming from the local effects on the muscle function on one side, and there is the fatigue (central fatigue) arising from the level of CNS due to the feedback of the pathological condition in the peripheral nervous system on the other side. This type of fatigue protects the muscle from being damaged in the long term during the improperly set physical activities [77]. This symptom is perceived by patients as a different, abnormal, and feeling of being more tired than before the onset of disease [80]. In a study, which included the neuromuscular disease groups of HMSN, FSHMD, and MG, is indicated that severe fatigue is associated with functional impairment in daily life. The level of fatigue has been found to be significantly related to the muscular strength, the level of physical activity stated by the patient, sleep disorders, and pain. The fatigue level and the physical activity level are mostly associated with functional impairment in daily life [81].

3.2. Exercise intolerance and the impact on functional capacity

Exercise intolerance is a type of abnormally severe tiredness that develops with a certain movement in the muscle involved in that movement. In individuals with NMHD, several

factors such as loss of functional muscle tissue, unused muscles, injuries due to excessive use, cardiopulmonary involvement, contractures, decreased locomotion adequacy (decreased walking speed and increased energy expenditure), decreased patient motivation, less social participation for activity, increased depression, and increased social barriers lead to a decline in physical activity [1]. Three problems affecting functional capacity and determining the level of physical activity in NMD are often striking. These are muscle weakness, difficulty in exercising, and fatigue. These problems result in a decrease in physical activity and a sedentary lifestyle [1]. Unfortunately, sedentary behavior increases exercise intolerance and causes a decrease in functional capacity in the subsequent periods, resulting in a low quality of life. The occurrence of fatigue is dependent on which of the energy-producing metabolic pathways were used during the activity. For example, if a patient develops exercise intolerance while walking but this patient is not uncomfortable while running fast, this should primarily suggest the impairment of lipid metabolism, which enables slow movements with type-1 muscle fibers. On the other hand, if the same complaint occurs in the arm muscles, for example, in an arm movement such as serial and rapid wiping, it should be primarily considered that the glycogen metabolism used by the type-2 muscle fibers is defective in this case [82]. Individuals who have been diagnosed with NMD are more likely to live a sedentary life when compared to physically healthy people. A person with a lower level of physical activity will later have an increased body weight even with a normal diet. Increased body weight makes the patient more inactive, and this continues in a vicious cycle [83–85]. Therefore, when it is considered that all of the components required for exercise are relatively affected in NMDs, a reduction in exercise capacity in this disease group is inevitable.

3.3. Walking and mobility problems

Mobility is defined as the movement of the person around self and transition from one position to another in a safe manner; it is a function that should be carefully observed throughout the course of the disease in NMD [86]. NMDs progress with disorders limiting the patients' mobility and their independence in daily life activities. The reason for this is that the progressive weakness of the muscles affects the functional levels of the patients negatively after a while, leading to the defects in mobility activities such as standing up, walking, running, and stair climbing [87]. In the majority of NMDs, there is a chronic clinical course emerging slowly or progressing with a rapid decline in muscle strength that leads to impaired motor function. Neuromuscular patients suffer from difficulty in standing up from the ground as clinical signs of weakness of hip extensors, quadriceps, and trunk muscles. The increasingly apparent loss of power in the gluteus maximus and quadriceps muscles leads to an excessively increased lordosis. As the patient tries to stabilize hips in extension, lordosis becomes more pronounced. As the muscles weaken, the patient tries to increase stabilization by pulling the arms back and increasing the lordosis to pull the center of gravity behind the hip joint. To expand the area of support while walking, the patient walks like a duck (Trendelenburg gait) [88]. Fracture contracture is seen in hip joint with the loss of strength in hip extensor and the lumbar lordosis is increased so that the upright posture can be sustained and the loss of strength can be compensated. However, the progression of the loss of strength in the muscles of the pelvic girth causes the knee flexion contracture to develop [39]. The weakness of the hip flexor and the eccentric muscle reduces the length of stride. The weakness of the knee

extensor reduces the knee flexion moment in the midstance phase. Due to the weakness of dorsiflexion, plantar flexion contracture obstructs the stabilization of the foot during standing as well as the toe-off during the swing phase [22].

Contractures may lead to postural disorders such as kyphoscoliosis and scoliosis, which can interfere with the continuation of sitting and lying activities, and as a result, pain. Furthermore, ortheses that help maintain the force, which reduce the development of contractures and deformities, and surgical medical procedures lead to additional disorders [89]. In the advanced stages of NMDs, various problems are faced such as wheelchair dependence, being confined to bed, difficulty in in-bed transfers, inability to maintain mobility during climbing the stairs without holding to something like railing, not being able to stand up from the sitting position, and falling in and out of home [1]. This makes the patients dependent during their daily life activities.

3.4. Psychological and other problems

Even in normal individuals, being ill affects a person psychologically. In a condition such as that of neuromuscular patients where the disease is chronic and has many symptoms, this can make one feel more vulnerable. While some of the patients may cope with the problems associated with the illness and adapt to the social life, some of them respond to these problems with limitations in school, social life, and daily activities. These restrictions manifest themselves as a decrease in independence and self-confidence, limited participation, and social isolation, particularly in youngsters. For example, of two patients with muscular diseases with the same mobility level and the same severity of pain, one may continue to work while the other may quit or even show severe depression. In patients with NMDs, patients with increasing problems are severely affected, and with symptoms such as pain and fatigue, patients may not maintain their psychological well-being [90].

4. Participation problems in NMD

It has been shown in the literature that mobility (transfers and walking), which is one of the parameters of social participation, housekeeping, community life, education, work, and leisure activities were affected in NMD [91]. Biomechanical problems present in NMDs lead to participation problems as a result of the increased energy expenditure levels and fatigue [92].

4.1. Falling and fear of falling

According to ICF, falling affects participation by deterioration of the affected body structure and body functions [21]. Falling often has a complex form; it is defined as lying on the ground or at a lower level by accident, except conscious positional change for resting on seats, walls, or other objects, and is affected by multiple factors [93–96]. There are different descriptions, such as intrinsic and extrinsic factors, associated with the risk factors of falling [94, 95]. These descriptions include age, duration of illness, presence of prior falls, fear of falling, the number of used medicines, use of antihypertensive medicine, reduction in mobility level,

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and in-home and out-of-home dangers [97, 98]. Also, risk factors include muscle weakness, the presence of falling history, gait abnormality, balance disorder, the use of assistive device, mobility limitation, visual disturbances, arthritis, depression, cognitive impairment/mental status changes, postural hypotension, vertigo/dizziness, incontinence, and chronic diseases [99, 100]. Prevention and reduction of falling have a positive effect on the patient's activity and participation level [101].

4.2. Environmental factors

Participation problems are encountered as a result of the interaction of environmental factors experienced. Slippery floors, bed-chair heights, poor lighting, unsuitable ancillary equipment, improper building designs, and broken or uneven pavements may be listed among the factors [93, 102].

4.3. Psychological factors

As the disabilities and difficulties start to be permanent in a patient's life, the patient may lose hope completely. Such people remove themselves from activities and social life, and face many problems including depression as a consequence [103, 104]. Hopelessness, the thought that no one can help them, is a common feeling for neuromuscular patients [105]. In a study of 88 male patients with progressive musculoskeletal dystrophy, it was found that patients had problems in participating in private life and professional life and that more than half of the cases considered themselves to be socially isolated [104]. In his work to assess participation in myotonic dystrophies, Gagnon noted that the participation problems of these patients were about communication, personal care, and interpersonal relationships [106].

In NMD, the restriction of participation occurs in professional life, leisure activities, home, family, community, and social life [104]. Muscle weakness and reduced aerobic capacity may negatively affect participating in the professional life that requires long-term physical activity. Deterioration of interpersonal interaction and relationships is another reason for the restriction of participation. Sometimes, this restriction can lead to extreme consequences such as no communication with people or not being able to eat or drink in public. Participation in the community and social life can also decrease due to depression and fatigue. The psychological burden of having a degenerative and terminal illness affects the participation of the individual. The presence of each of these factors has significant adverse effects on the quality of life [107].

5. Assessment methods in rehabilitation of NMD

Nowadays, the key to planning a good rehabilitation program is to know the characteristics of the disease and the problems it will cause in the patient, and to evaluate the patient in detail in light of these features. In NMDs, evaluation is performed to monitor the progression of the disease, to determine the appropriate treatment methods, to investigate the efficacy of treatment methods, and to predict and prevent possible complications. Some evaluation

methods are used, in which the disorders of the body structure and function, and the limitations of activity and participation are evaluated. These methods include evaluation of respiratory functions, muscle testing, normal joint movement, evaluation of flexibility, evaluation of motor functions and functional capacities, timed performance tests, functional posture, and gait analysis.

5.1. Assessment of muscle strength

Due to the diversity of neuromuscular diseases, it is crucial to determine the pattern or spatial distribution of weakness in the evaluation of muscular weakness to distinguish the etiology. It should be determined whether the loss of strength is general (e.g., bilateral, proximal, distal, or all) or localized [71, 108, 109]. The presence of the weakness predominantly in the proximal muscle groups or the dominance in the distal muscles, its presence on one or both sides of the body, and impairment in a single nerve or a group of nerves reflect the pattern of weakness [70]. In the case of huge weakness, it is determined whether the loss of function is proximal or distal, and which functional activities are limited by muscle weakness [108]. If the patient is having difficulty in standing up (around the hips) or combing the hair (around the shoulders), it indicates proximal muscle weakness, commonly observed among the weaknesses in myopathic diseases. If the weakness is pronounced around the hip, there are difficulties in getting up and down the stairs, standing up from the chair or toilet, or standing up from the ground or squatting position. If there is a weakness in the shoulder, there are many functional difficulties such as lifting heavy objects, reaching to and taking the objects on high shelves, brushing teeth, bathing, dressing, and combing the hair [108–110]. Generally speaking, proximal muscle weakness seen in the form of a limb-girdle pattern in the arms and legs and the muscle group of the shoulder and the hip circumference suggests a myopathic process (process affecting the muscles directly); the presence of distal weakness, however, primarily suggests prevalent polyneuropathy.

However, some myopathies cause distal weakness. This pattern usually cannot be diagnosed and diagnostic errors are possible in relation to the neurogenic feature. The types of myopathic diseases with this atypical distal phenotype are known as distal myopathies and include MD and inclusion body myositis [108–110]. In less frequent cases of distal myopathies and neuropathies, patients complain of distal upper extremity weaknesses that cause difficulty in activities such as opening jars, buttoning, turning the key, and turning the door handle. Patients with distal lower-limb muscle weakness complain about tripping over the pavement edges, having difficulty walking on uneven surfaces, or dragging feet during walking. Patients experience difficulties standing on toes (m. gastrosoleus) or in activities involving hands (intrinsic muscles) [108, 109].

Methods such as manual muscle testing, dynamometric evaluation, isokinetic (eccentric and concentric) evaluation, and surface electromyography (EMG) are used in the clinical evaluation of muscle strength, but manual muscle test (MMT) is the most commonly used method because of its easy application in clinical practice. Static and isometric contractions of the muscles are also measured with cable tensiometers or dynamometers. These measurements are superior methods because they provide numerical data in NMD, are objective, and reflect

changes in muscular strength [111]. However, manual muscle strength measurements remain valid today because of the low reliability of dynamometric measures in muscle groups that cannot complete their movements against the gravity or distal muscle groups [112, 113]. Nevertheless, the physiotherapist who will perform the muscle testing in NMD should be able to analyze whether the factors that limit muscle strength stem from weakness, loss of motor control, pain and/or fatigue, or whether the movements completed during manual muscle testing through compensations. Also, the physiotherapist should consider the time and quality of contraction, the range of motion (ROM) of the joint, and patient's ability to maintain contractions. For this reason, the evaluating physiotherapist should have performed a significant number of MMT-related applications in neuromuscular diseases [22]. Another method is to find the maximum weight that a person can lift at once. For this purpose, isokinetic instruments have been developed that measure the maximum force at the specified speed in NMDs. These tools give information about both concentric and eccentric contraction. Studies have shown that it is reliable in good unipolar joints, especially elbows and knees. The use of these instruments is somewhat reliable because it is difficult to isolate the shoulder, wrist, and ankle muscles while measuring their strengths during particular movements. It is stated that it is more suitable to perform the force measurement using hand dynamometers providing better stabilization in these joints [114].

5.2. Assessment of range of motion and flexibility

Since there is the loss of muscle fibers in the intrinsic muscle tissue, contractures due to necrosis and fibrosis of the muscle fibers in NMDs, biomechanical analysis of the movement, and normal joint range of the motion and flexibility should be assessed for tracking the disease progression. The range of motion of the joint is monitored objectively by goniometric measurements at regular intervals [115]. The active and passive normal range of the motion should certainly be assessed. The physiotherapist should maintain a record of the presence of limitation and whether the limitation is due to the muscle, the joint capsule, the tendon, or the pain and make a comparison of the agonist-antagonist flexibility. The physiotherapist should consider this situation in the treatment program [22]. In muscle-induced limitations, shortness of muscles should be particularly assessed, particularly since the shortness of hip and knee flexors, plantar flexors, lumbar extensors, latissimus dorsi, pectoral muscles, tensor fascia latae, and quadratus lumborum muscles are considered functional [116].

5.3. Assessment of motor function

There are some tests evaluating motor function in NMDs.”Motor Function Measurement (MFM)” is a scale with demonstrated validity and reliability developed to evaluate motor function in all NMDs. It was indicated that this is a scale, which evaluates the severity of motor impairment in the NMD group with good psychometric properties. It was indicated that, other scales, MFM is adapted to the severity of deficits at every level in patients who can or cannot walk and that it evaluates all of the head, trunk, lower, and upper extremities [117]. Some of these tests are specific to the disease, while others focus only on one area of the body. The “Spinal Muscle Atrophy Functional Motor Scale” was prepared for SMA patients

[118], the "Amyotrophic Lateral Sclerosis Functional Classification Scale" and the ALS score for patients with ALS [82], and the "Hammersmith Motor Skill Score" for patients with DMD [119]. Among the scales focusing only on one region/function of the body, Trunk Control Test and Trunk Impairment Scale are for the trunk [120, 121], the "Brooke Upper Extremity Scale" is for upper extremity [122], the "Vignos Lower Extremity Scale" is for lower extremity [123], the "North Star Ambulatory Assessment" is for ambulation [124] and the "Jebsen-Taylor Hand Function Test" and "Activlim" are for the hand functions [125, 126].

5.4. Performance tests

The timed and controlled tests of the subsequent tests applied to the patient, evaluating the patient's ability to perform a specific activity in a specific time interval. These include some activities such as rolling from the supine to the prone position, rolling from the prone to the supine position, rising to the sitting position from the lying position, standing up without sitting, walking 10 m, climbing 10 steps up and down, and putting on a t-shirt and taking it off [127]. The Minnesota, Purdue pegboard, nine-hole peg test, and Jebsen hand skill tests were developed and are the most frequently used timed tests to assess hand functions [55]. There are no average values for the timed performance tests. The results are interpreted by comparing the clinical findings with the subsequent tests administered to the patient.

5.5. Fatigue

Two different methods are used in fatigue evaluations: electrophysiological tests and scales. Since electrophysiological tests are expensive, scales are more commonly used in clinics. The Multidimensional Fatigue Inventory (MFI) [128], the Fatigue Severity Scale [129], the Piper Fatigue Scale (PFS) [129, 130], the Short Fatigue Questionnaire (SFQ) [131], the Chalder Fatigue Scale (CSF) [132], Fatigue Impact Scale (FIS) [133], and Visual Analog Scale (VAS) are among the scales used for assessment of fatigue [134, 135]. While each of these tests has its advantages and disadvantages and is used in a large population of patients, none of the tests have been specifically developed for neuromuscular patients.

5.6. Respiratory function

The most commonly used evaluation methods in the clinic are the pulmonary function tests including spirometric measurements of "forced vital capacity (FVC)" and maximum inspiratory and expiratory pressures (MIP, MEP) [136]. Cardiopulmonary exercise tests are the evaluation of respiratory muscle strength, thoracic environment measurements, and assessment of respiratory frequency. In the literature, the recommendations on which respiratory evaluation should frequently be done in neuromuscular disease are given in **Table 2** [137].

5.7. Assessment of cardiac functions

Symptoms of heart disease seen in NMDs depend on the severity of skeletal muscle insufficiency and the severity and type of effect. The degree of neuromuscular insufficiency may modulate the symptoms of heart involvement and over time and may sometimes suppress

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Test	Frequency
History, physical examination	Six monthly and in acute conditions
Lung function test (FVC, FEV1, VC—upright and supine)	Six monthly and after acute conditions
MIP, MEP	Six monthly and after acute conditions
Cough peak expiratory flow	Six monthly and after acute conditions
Polysomnography	At least yearly, symptom oriented and after acute conditions

Abbreviations: FVC, forced vital capacity; FEV1, forced expiratory volume; VC, vital capacity; MIP, maximum inspiratory pressure; MEP, maximum expiratory pressure.

Table 2. Respiratory follow-up of patients with NMD [137].

these symptoms [138]. The most commonly used screening test in clinic has two types: resting electrocardiography (ECG) and ambulatory ECG (Holter). Cardiac rhythm, intraventricular state, and ectopic beats can be evaluated noninvasively with ECG [139, 140]. In particular, cardiopulmonary exercise tests in individuals with severe neuromuscular disease generally show a decrease in maximal oxygen consumption, a decrease in pulmonary ventilation, a reduction in work capacity, and an elevation of resting heart rate. These findings reflect respiratory muscle involvement, cardiac decay, and poor physical fitness [69].

5.8. Pain

The characteristics of the pain should be recorded, such as localization, type, frequency, day-night difference, factors that increase or decrease the pain, change in pain with movement, and the presence of a response to a pain reliever. It should be established which of the causing factors of the pain are involved such as muscle imbalance, trigger points, joint tension, and muscle spasm. One-dimensional and multidimensional scales are used to assess pain severity. One-dimensional scales are intended to measure pain intensity directly, and the patients make the assessment themselves. They are used especially in the evaluation of acute pain and in monitoring the efficacy of applied pain reliever. Among the one-dimensional scales are the verbal category, numerical and visual comparison scale, and Burford Pain Thermometer [141–147]. Multidimensional scales are thought to be useful in certain cases to assess all aspects of pain in chronic pain [143]. These include the McGill Melzack Pain Questionnaire, the Dartmouth Pain Questionnaire, the West Haven-Yale Multidimensional Pain Inventory, the Memorial Pain Assessment Card, the Wisconsin Brief Pain Inventory, the Pain Perception Profile, and the Behavioral Models [141–147].

5.9. Assessment of aerobic (functional) capacity

The purpose of evaluating functional capacity is to assess whether or not the maximal or submaximal activities can be performed in nonclinical settings. The ultimate standard to evaluate the person's aerobic exercise response is maximal increasing cardiopulmonary exercise test [148]. A suitable cardiopulmonary exercise test allows determining the underlying

pathophysiological mechanisms. These mechanisms include broad assessment of the exercise response, the objective determination of the functional capacity and impairment, the measurement of the appropriate intensity required for long-term exercise, the amount of factors limiting exercise, and the contribution of various organ systems involved in exercise [149]. Functional capacity is assessed by maximal cardiopulmonary exercise tests, motorized treadmill, and stationary bicycle ergometer. However, submaximal tests are recommended in situations where maximal testing increases the patient's risk status and hinders his/her potential abilities, especially in individuals with significant risk for cardiovascular problems, and in cases where multiple cases are to be tested [148, 150]. The 6- or 12-min walking test is a form of submaximal exercise assessment, finds extensive use in the field, is employed in pulmonary diseases and heart disorders, and evaluates the response to various treatment interventions especially pharmacological treatments and exercise training. The ability to walk at a specified distance is a quick, simple, and inexpensive way to evaluate physical function. Walking is also a critical important component of the quality of life because it is highly necessary to accomplish the daily life activities and reflects patient's capacity [151, 152]. The validity study of the 2-min walk test has been conducted in neuromuscular diseases, and it was put to use in recent years since its period of implementation is short and it does not cause fatigue [153, 154].

5.10. Assessment of functional mobility and falling

In clinical practice, the activity and participation problems are also encountered as a result of disorders seen in body structure and function in neuromuscular diseases according to ICF. However, walking emerges as a function related to all of ICF subparameters (body structure and functions, activity, participation, personal and environmental factors). In particular, two important conditions must be considered for mobility evaluation according to ICF: the *capacity* is the ability of an individual to perform a function or a task and what the individual is capable of doing in his/her environment is the *performance* [21].

Functional mobility, bed mobility, transfer, transfer grounds, gait, and wheelchair should be evaluated. The physiotherapist should assess the level of effort to initiate movement, weight transfer, postural alignment, motion timing and motion completion, patient balance, support surface, walking assistance, and energy expenditure level [22]. The Rivearmed Mobility index is also used in mobility analysis [155]. Walking analysis is done by observational and three-dimensional analysis methods. Some parameters and assessments may be used to analyze the gait. These parameters include step-length asymmetry, position of the ankle during heel strike, knee angle in heel strike, knee flexion angle in stance phase, single extremity support, state of foot and ankle in push phase, knee flexion in swing phase, body position, presence of Trendelenburg sign in frontal plane, knee cap in transverse plane, and foot angle and arm posture. The evaluation should be advanced to investigate possible causes in case of detection of a possible impairment in these standards [156]. For three-dimensional analysis, computerized video cameras, passively reflected signal indicators, multicomponent power platforms, dynamic electromyographic analysis, and temporospatial gait analysis systems are used [154]. The timed-up-and-go test, 30-s chair-stand test, the 4-Stage Balance test [157–160], and risk analysis tools such as the Fall Risk Assessment Tool and STRATIFY (St. Thomas

Risk Assessment Tool) may be used for the evaluation of falling in neuromuscular diseases [161–163]. However, there is no specific cutoff value for NMDs in these tests.

5.11. Evaluation of activities of daily living (ADL)

Functional level deteriorates due to progressive muscle weakness in NMD, and dependence in ADL increases, while the tests such as Barthel, Katz, and Lawton are used for clinical evaluation of ADL in neurological patients [164]. FIM is the most preferred method for determining the levels of ADL in NMD. The advantage of FIM is that it has proven validity and reliability in evaluating many diseases and that it was found highly reliable by practitioners even when implemented by specialists with varying education and experience [165].

5.12. Sensory evaluation

In neuromuscular diseases with sensory involvement, especially in peripheral neuropathy, there is a need for frequent sensory evaluation. Surface sensations such as light touch (cotton), pain (sharp-blunt test), and sense of temperature (with hot-cold) and deep sensations such as vibration sensation, pressure recognition, touch localization, joint position, and motion sense should be evaluated [166].

5.13. General health measurements

These measurement methods give a general profile of health such as well-being, functions, social, and emotional health. The most commonly used assessments in NMD in this group are "Nottingham Health Profile (NSP)" and the Short Form SF-36 Quality of Life Survey [167].

6. The aim and content of the rehabilitation program

Although there have been some promising studies recently, there is no known curative approach to NMD. Physiotherapy and rehabilitation programs are gaining importance to maintain muscle strength, functional capacity, and quality of life as long as possible and to keep the patient in social life [168]. The lack of therapeutic approaches that can curb the progression of the disease in a large proportion of neuromuscular diseases increases the importance of preventive, supportive, compensatory, and rehabilitative approaches. The aim of physiotherapy rehabilitation approaches is to improve the quality of life of patients and their families. Applications in that direction are to delay muscle weakness or loss of strength, to prevent muscle shortness and distortion in joints, to prevent respiratory problems, to maintain the walking activity for as long as possible, to educate the family, to support and keep the function at different stages of the disease, and to increase functional capacity [169]. The rehabilitation program should include the protection of the functional level of the patient and the physical and psychological functions, increasing the physical and mental capacity of the patient, and slowing the progress of the disease symptoms. Patients have a significant number of clinical problems, so rehabilitation should be done with a multidisciplinary team.

6.1. The characteristics of the physiotherapist present in the rehabilitation team of neuromuscular patients

The effectiveness of the rehabilitation program to be administered in NMDs depends on the ability of physiotherapists to assess and analyze the main causes of the patient's problems [22]. As a prominent member of the rehabilitation team, the physiotherapists who will evaluate and implement the rehabilitation in NMD should be specialized for this group of patients and should be able to individualize the rehabilitation program. The physiotherapist should be able to identify the needs of NMD and be able to individualize the treatment program based on needs. The physiotherapist should have knowledge about the pathophysiology of the disorders in the patient and about the progression of the disease. The physiotherapist should be able to serve in different settings (inpatient and outpatient clinics, at home, at home-care facilities, and in workplace arrangements) and various age groups. The physiotherapist should be able to follow up the patient with the short/long-span controls during the planning of research on NMD, the development and implementation of outcome measurements, the setting of treatment interventions, and the natural progression of the disease and should be able to document the process well [22].

The rehabilitation program should include the muscle strength preservation, an exercise program for the prevention of contractures, increasing in respiratory function, increasing the functional (aerobic) capacity, walking and balance training, fall prevention and the stages of deciding walking aids, nutrition expert support, psychosocial approach, vocational counseling, and ergotherapy processes.

6.2. Preservation of muscle strength and prevention of contractures

Many international researchers agree on the use of exercise therapy in neuromuscular diseases. There are many physical and psychological benefits of exercise such as muscle strength preservation, prevention of contractures, increased flexibility, reduced energy expenditure, relieving fatigue, reducing pain, depression, social isolation and loneliness, ensuring the participation of the individual active life, sustaining mobility, and increasing the quality of life. However, the number of studies with a high level of evidence about type, intensity, frequency, and speed of the exercise is limited. When deciding on a possible exercise program, the pathophysiology, onset, severity and the progression of the disease, the age and sex of the patient, and the intensity and frequency of the exercise to be given should be considered [170]. In general, there is a consensus on the positive effects of mild to moderate exercise programs on muscular strength without causing significant muscle damage, particularly in the early stages of neuromuscular disease with moderate progress when the muscle strength was not severely affected [171]. Combined use of upper/lower extremity exercises with neck and body exercises is preferred in clinical practice due to the ability to spread force from strong muscles to weak muscles and relieve fatigue [172].

6.2.1. Stretching and normal range-of-motion exercises

Stretching and normal range-of-motion exercises can prevent the limitation of joint mobility that develops as secondary to the muscle weakness [170]. The static stretching, which is usually used

for treatment in NMDs, is performed by proper alignment of the joint and bringing the muscle to its maximum length along the joint during stabilization of the unmovement joints. When the movement reaches the end, the position is held for at least 10 s and repeated. Although healthy individuals are recommended to make one to two repetitions a day or three to seven repetitions per week regarding the frequency of stretching, there is no definite information for neuromuscular patients. Daily stretching exercises are recommended. It has been reported that ROM increases with stretching frequency and the improvement was maintained for 4 weeks after the exercise ends [173]. Surveys show that a little 5-min static stretching causes a change in the muscle-tendon unit [174]. Resting splints can be used during sleep to prevent contractures. When bed confinement has developed, lower extremities can be stretched using the body weight with standing table [175].

6.2.2. Strength training

Strength training can be done with resistance exercises. Resistance exercise training is one of the most effective ways to improve the functional capacity of the neuromuscular system. However, the potential benefits and risks of strength training in neuromuscular diseases are still a controversial subject in the literature. Progressive strengthening exercises are also commonly used to increase muscle strength in neuromuscular diseases [170]. Progressive strengthening exercises improve lean body mass, muscle protein mass, contractile strength, strength, and physical function. This improvement varies according to the rate of progression of the disease. There is a consensus in recent years about the benefits of mild to moderate intensity strength training (25–40% of maximum weight) on muscle strength without any deleterious effects, especially for slow-progressing neuromuscular diseases [176]. Considering that the high-intensity (50–70% of maximum weight) eccentric- or concentric-type exercise programs would cause mechanical stress on muscle fibers and increase muscle weakness, they are not recommended for use on dystrophic types with rapid progression and membrane instability (such as Duchenne muscular dystrophy) [170]. However, studies with opposite point of view also exist suggesting that high-intensity training is beneficial with appropriate selection of patients [175]. However, no additional contribution to muscle strength and endurance was shown in a comparison of the maximum intensity weight training to the medium- and low-intensity weight training [171].

Strengthening principles applied especially in healthy muscles should be used carefully here. The level of intensity and resistance, which is above the patient's muscle strength, strain the patient, and cause muscle fatigue, should be avoided. The patient should be recommended to do the exercises in parts during the day so as not to cause fatigue. It should be started with little or no resistance and few repetitions, and the frequency, duration, and resistance of the exercise should be monitored with monthly or tri-monthly evaluations. The intensity and resistance of exercise should be revised if the patient experiences pain, muscle spasms, fasciculations, and excessive fatigue after exercises [177].

Electrical stimulation commonly used in the clinic for strengthening should also be used with caution in NMDs. Since all muscle fibers contract at the same time with electrical stimulation, it can increase degeneration in patients with low muscle fiber counts. For this reason, patients with muscle strength below three should use current types that will not cause fatigue [177].

6.2.3. In-water exercises (aqua therapy)

In-water exercises are the most appropriate exercise method for this group of patients. The water lift supports weakened muscles, permits functional movement, and in some cases can also be used as a resistance exercise. Pool exercises treat all muscle groups and maximize the aerobic capacity of the patient. It is particularly effective in a group of patients with limited energy levels. It is recommended in the literature to apply 45 min twice a week. Limitations of pool therapy are lack of accessibility and insurance payment [178].

6.3. Aerobic endurance training

Aerobic endurance training generates physiological responses that are different from strength training. Sufficient intensity and duration for aerobic training that involves the use of large muscle groups and will not cause fatigue is 50–85% of VO2max and 30 min. Aerobic training causes stimulation in the heart, the peripheral circulation, and the musculoskeletal system. As a result, circulation of more oxygen in the body leads to an increase in cardiac output, capillary density, and vascular transmission. For this benefit, aerobic exercises such as swimming, walking, and cycling can be performed, which put fewer burdens on the musculoskeletal system [179].

According to the American College of Sports Medicine guidelines, it is adequate to improve cardiorespiratory fitness for most aerobic training when an optimal frequency of 70–85% of maximum heart rate and 60–80% of maximum oxygen consumption are combined with an optimal frequency of 3–5 days per week [180]. A systematic review by Cup et al. suggests that an aerobic training at this intensity can be recommended to the NMD patients with a good functional level. In most of the studies included in the review, the cycling or treadmill exercises done at least three times a week and the use of approximately 70% of the heart rate reserve or the use of an estimated maximum heart rate are recommended. It is stated that the whole program lasting at least 10 weeks for both muscle strengthening and aerobic exercises and regular physical therapist supervision increases the effectiveness and improves the safety and suitability of the exercise [181]. However, in only 30% of all studies involving muscle strengthening and aerobic exercises, training lasted for less than 10 weeks, with an average of 5 weeks. In the literature, it is indicated that the aerobic exercise training in conjunction with muscle strengthening exercises is effective at evidence level of 2 or 3, especially in muscle diseases and neuromuscular patients with heterogeneous features [182].

6.4. Development of postural control

The activity of coming to sitting position from the supine position is an important activity of the body against the gravity and is one of the first stages of mobility. During this activity, which is very important for muscle patients also, the anterior trunk muscles contract concentrically while the posterior trunk muscles contract eccentrically. The difficulty of the patient in

coming to the sitting position should bring into mind the possibility of encountering mobility problems and that the treatment should involve the precautions related to the trunk [121].

The equilibrium reactions have been tested in patients with isolated muscle weakness, and it was concluded that the muscle weakness is important. Although patients with distal leg weakness are particularly prone to stumble-like stability disorders, the stability has been observed to decrease following the external perturbations of balance in proximal muscle weakness [32]. In previous studies, it has been shown that balance correction strategies assessed by dynamic posturography can vary depending on body parts where muscle weakness is present. Some muscle responses are sensitive to balance perturbations, especially in the sagittal (anterior-posterior) plane, while others are found to be sensitive to the frontal plane or a combination of these two planes. This reveals that the proximal and axial muscles (such as paraspinal or gluteus medius) are more frontal-focused, the lower-limb muscles are more sagittal-focused sensitivity, and the knee muscles have the sensitive role in both directions in muscle response sensitivities following proximal to distal disturbances [183, 184]. Therefore, the patient maintains the balance based on these sided sensitivities, and the question whether the patients with distal muscle weakness are more distressed in the sagittal plane and those with proximal lower extremity weakness are more distressed in the frontal plane should be answered. It is thought that this information can also help in the planning of therapeutic interventions. For example, patients with complete proximal weakness are unstable in balance correction strategies associated with the frontal plane; these patients will need a different intervention than those with complete distal weakness and possibly unstable in strategies associated with the sagittal plane [185].

6.5. Increasing respiratory capacity

If there is a coughing weakness in the patient, airway cleaning techniques such as air stacking (glossopharyngeal breathing), mechanical, and manual coughing should be applied as soon as possible [186]. Increasing the pulmonary capacity, breathing exercises, diaphragm breathing exercises, and thoracic expansion exercises aim to maximize the expansion of the patients' lungs and should be taught to the patient.

Pulmonary expansion therapy and maximal insufflation therapy (mask or mechanically assisted hyperinsufflation) increase the forced inspiratory vital capacity. It is reported in the literature that maximum insufflation therapy is important in increasing peak cough flow for neuromuscular patients with vital capacities less than 1500 mL [187]. Manually supported coughing techniques should be continued for maximum expansion. Secretion mobilization can also be provided by a positive expiratory pressure device. With a positive expiratory pressure device, patients breathe freely and breathe against a moderate resistance; air pressure activates secretions, preventing atelectasis. Traditional chest physiotherapy techniques used for airway cleaning should be taught to this patient population. This involves taking the patient to different positions, then clapping on the chest wall, vibrating, and coughing. However, it should be taken into account that Trendelenburg, lateral recumbent, and prone positions are difficult to tolerate in NMDs [186].

6.5.1. Mechanical insufflation-exsufflation (assisted coughing device)

The assisted coughing device operates according to the principle of vacuum cleaning. Cleansing of strong expiratory flow and secretions is achieved without tracheostomy by applying negative pressure after maximal insufflation with a positive pressure of the oronasal mask. It is also known to be more effective than aspiration catheters in tracheostomized patients. The use of a peak cough flow below 160 L/min in NMDs is found appropriate. However, in recent publications, a cough flow of at least 300 L/min was used to initiate maximum assisted mechanical cough assistance [187–189]. The assisted coughing device produces an airflow of approximately 10 L/s with pressures between -40 cm HO_2 and 40 cm HO_2. It is a very vital and efficient device to use in patients using a mechanical ventilator and has reduced coughing ability [190]. Noninvasive mechanical ventilation devices are employed in later stages of NMDs. Indications for noninvasive mechanical ventilation are shown in **Table 3**.

6.6. Reduction of pain

The mechanism of pain has not been identified in detail in neuromuscular patients. For this reason, the physiotherapist should choose the appropriate treatment modality based on the pain source he/she has identified. From among the physiotherapy techniques, ultrasound, TENS, hot/cold application, and massage may be used but there are very few studies on the effectiveness of these techniques. There are even contradictory results regarding the increased muscle destruction of the ultrasound and hot application. Physiotherapists should be so cautious in their use. It is thought that TENS is preferred because it uses different ways of inhibiting pain.

In the loss of muscle strength, joint pain can often be associated with improper alignment or excessive stretching of the joint capsule. Pain relief is possible with proper alignment of the joint and removal of excessive tension. However, since the weakness of each NMD patient is seen in different forms, it should be analyzed well before choosing the appropriate treatment. External support such as splints can be used when muscle weakness makes self-stabilization impossible. An external shoulder splint may be utilized for the shoulder pain resulting from shoulder subluxation while an abdominal mattress may be preferred for back pain due to the excessive weakening of the abdominal muscles [22].

Shoulder pain may arise due to the single-point cane, which NMD patients often prefer for cosmetic reasons. In this case, a four-point walker may be used if the patient's energy

- Chronic daytime hypercapnia with $PaCO_2 \geq 45$ mmHg
- Nocturnal hypercapnia with $PaCO_2 \geq 50$ mmHg
- Daytime normocapnia with a rise in $PTcCO_2$ of ≥ 10 mmHg during the night
- A rapid, significant reduction in VC
- MIP <50 H_2O cm or 60% and FVC <40%

Abbreviations: $PaCO_2$, partial pressure of carbon dioxide (CO_2) in the blood; $PTcCO_2$, transcutaneous carbon dioxide; VC, vital capacity; MIP, maximum inspiratory pressure; FVC, forced vital capacity.

Table 3. Indications for noninvasive ventilation in neuromuscular diseases [137].

expenditure level is not increased [154]. If the size of the four-point walker is not suitable for the patient and the patient bends forward, this may be a possible reason for back pain. If the patient has a lack of postural control and the arms carry the whole load when the walker is used, this may be considered as a possible cause of pain in the upper extremity. If the patient's upper extremity strength is unable to carry the weight of the walker and the patient has upper extremity and back pain, then the arm-assisted wheeled walker can be used to reduce pain.

The shoulder pain that occurs in patients using wheelchairs may be due to the improperly positioned inadequate arm support, which prevents the alignment of the humerus in the glenohumeral joint, while the hip and back pain may be caused by improper knee-hip level and foot support. Wheelchair cushions, unsuitable pillows, and the bed should be considered as possible reasons for pain. The preference of a pressure distributing bed is also an important factor in reducing pain in bed dependence [22].

6.7. Walking and balance training

Mobility target should be determined according to the evaluation and the progression of the disease. Walking training should include endurance training, teaching the use of walking aids, orthotic approach and fall prevention, learning safe-fall techniques, teaching how to stand up after falling, and teaching energy conservation techniques. In NMDs, the level of energy expenditure during walking increases [191]. For this reason, physiotherapists should choose the most appropriate aerobic exercise for the patient; the disease and exercise tolerance should be closely monitored. Physiotherapists should evaluate and recommend ambulatory assistive devices, transfer supports, and orthoses for improved walking, energy conservation, and safety if the patient develops weakness. However, the use of wrong assistive devices may alter the patient's optimal gait pattern and may prevent gait function or cause new problems in the patient. It has been shown that walking aids affect stability in a negative way in a group of studies in the literature. The reason for stability to be affected is that the upper- and lower-extremity balance reactions, normally used to protect against falls and protective, are restricted by the walking aid [192]. There is also the problem of lower-extremity tripping over the mobility aid. It is stated that the reciprocal movements necessary to use the walking aid in these situations are difficult to achieve. Choosing the right walking aid, using the right size, and training with the walking aid can reduce the risk of falling [193]. Walking aids prevent falls when used safely; it should be kept in mind that they may be the most important reason for falling if misused [154]. When the falling story is evaluated, the frequency of falls, during which activity the fall occurred, the balance, sense, proprioception, the characteristics of the fall area, and the home environment should be evaluated. Balance and proprioception training should be given as a result of these evaluations. Recommendations for orthosis can be made by a physiotherapist, an orthotist, or a doctor. When orthosis advice is given, the desired function, weight, and device tolerance should be considered, and a lightweight material should be used. The articulated orthoses may be granted if the patient has muscular strength to control the dynamic orthosis, fixed orthoses may be given if not. Walking training should be provided with the orthosis given [22].

6.8. Training for the activities of daily living and adaptive approach

It has been shown that there is a negative correlation between manual muscle test results and FIM results in studies performed [194]. The manual muscle test score of 3 is a critical threshold value. The average muscle strength value being 3 is an important indicator that the patient is dependent/independent in the activities of daily living or a candidate for dependence [195]. Weakness in the upper extremities causes patients to lose their independence in basic activities of daily living such as dressing, nutrition, and personal hygiene. In addition, muscle weakness around the shoulder in diseases with more pronounced proximal muscle weakness causes difficulty in performing activities such as shaving, makeup, and weight lifting; distal muscle weakness causes difficulty in gripping and increases the functional deficiencies of patients in activities such as writing, turning the faucet on/off, and unlocking the door with the key [127]. It has been found that the activities indicated by the patients as the most challenging are climbing stairs (72%), taking along walk (40%), and getting on or off the bus (18%) [195].

To be able to perform the activities of daily living independently, the functional capacity of the individual and the environmental conditions necessary to carry out the activity should be able to match fully with each other. When there is a discrepancy between these two parameters, problems arise which affect the quality of life of the person negatively. These problems can be solved by increasing the functional capacity of the individual, reducing environmental demands, and adapting the environment to the individual [196]. For example, if a person needs to pick up any item at the top shelf, he/she should be able to lift his/her arms over his/her shoulder. If he/she cannot lift his/her arms, he/she cannot be considered completely independent of this activity. This problem can be solved by strengthening the shoulder muscles to allow the arms to be raised above the head, or by lowering the height of the shelf with a manually adjustable cabinet to the level at which the patient can lift his/her arms [195]. Neuromuscular patients need different aids at various periods of the disease to be able to perform their daily life activities independently as the disease progresses. Assistive devices include tools designed or modified to increase the functional capacities of persons with disabilities that can be considered in a broad spectrum. This equipment can range from simple tools such as jar openers, pen holders, electronic environmental control systems, toilet lifts to prevent fatigue, handlebars, forks, and spoons with thickened stalk, and adjustable beds to complex technological tools. Parallel to the developments in technology, assistive devices are also renewed every day. As the complexity and technology increase, the costs of the assistive devices also increase, making it difficult for patients to obtain these devices. Appropriate devices prescribed to a neuromuscular patient increase the patient's quality of life [195, 197].

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