Approaches and directions for the physiotherapeutic management of patients with Duchenne muscular dystrophy

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Abstract: Duchenne muscular dystrophy (DMD) affects cardiac and skeletal muscles, but represents a condition that causes numerous secondary pathologies. Although there is no curative treatment available, multidisciplinary approaches, centered on the needs of the patient, improve the quality and duration of life. Therapies such as physiotherapy, occupational therapy, physical activities, respiratory physiotherapy, but also the management of cardiac, osteo-muscular and neuro-psychiatric manifestations, are continuously developing, (as new data becomes available) are the key of DMD management today. These multidisciplinary approaches can lead to good long-term outcomes by helping patients with muscular dystrophy reduce muscle damage, early detect and treat heart failure, and manage cognitive impairment as best they can.

Key words: Duchenne muscular dystrophy, physiotherapy, recovery

1. Introduction

In the last three decades, medicine has evolved a lot, so that numerous discoveries have been registered in all medical specialties and new therapies have appeared that have come to the aid of patients, increasing their life expectancy and implicitly the quality of life.

Currently, we cannot say that there is an actual cure for Duchenne Muscular Dystrophy (DMD), however, medical, surgical, occupational therapy, speech therapy and other specialties can improve the clinical picture of DMD.

This disease produces changes in the cardiac and skeletal muscles, but also non-muscular effects with consequences due to muscle weakness.

Motor disorders are highlighted in the following forms: the presence of the Gowers sign (manifests when the child tries to get up), the walk is rocking, on the tips, with loss of balance that leads to frequent falls, difficulties in running or climbing, inability to jump, difficulty going up and down stairs.

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Global motor disability is accompanied by pseudohypertrophy of the calves, muscle hypotonia, collapse of the plantar arch, muscle pain accompanied by cramps. The loss of motor skills also causes low resistance to exertion and the inability to control head movements in the elevated position.

The non-motor impairment leads to the installation of behavioral disorders, cognitive disability, delays in height and weight growth, attention deficit, learning difficulties, language disability. These losses are accompanied by the advancement of the disease and a series of cardiological, orthopedic and respiratory complications that end up putting the patient's life at risk.

In this context, the care of the patient with DMD must be individualized, specific to each stage of the disease and must be provided by a multidisciplinary team consisting of a neurologist, rehabilitation doctors, physiotherapists, psychologists and geneticists.

Along with them, nurses, dieticians, occupational therapists and, sometimes even social workers must be involved. Thus, this team can manage the psychosocial needs of patients diagnosed with DMD in order to help them integrate more easily into social life.

Monitoring patients is continuously done from the moment of diagnosis throughout life and aims to personalize the care of the patient according to the stage of the disease and his age [4].

For optimal care in DMD, guidelines have been created for effective disease management, based on models of approach and treatment of the condition. The existing guidelines lack evidence to support them, due to the lack of large-scale randomized controlled trials for DMD.

The indications are based on the experience of specialists in the field, and are created in accordance to selected and analyzed evidence, following recognized methodologies, with the aim of achieving a common point of view. The given indications refer to the directions of approach and management of DMD in the general way, but it is necessary that they fold on the characteristics and personal needs of the patient taking into account the new discoveries that appear over time in the specific therapies of DMD [13].

Treatment of patients diagnosed with DMD must be multidisciplinary, judicious, and always focused on the patient's well-being.

2. The multidisciplinary management

2.1. Physiotherapy, occupational therapy and physical activity

These forms of therapy are part of standard management. Physiotherapy is considered crucial for the success of treatment, as it has achieved good short-term results, such as maintaining the autonomy of these individuals [6], [11], [10].

Physical activity is another method that has been used to treat DMD [1],[7]. Exercise is used in people with DMD to reduce muscle damage, muscle contractures and bone fractures and to increase the amount of time the patient is functionally independent [9], [12].

The balance for a good treatment is directly related to the process of muscle
fatigue, because during a strenuous activity, metabolic and physiological changes occur that can lead to sensations of asthenia and fatigue.

Therefore, although there are studies describing the entire process of fatigue and degradation of skeletal muscle fibers and the nervous system during physical exercise, few studies have evaluated these parameters in patients with associated diseases [5].

The recovery treatment addressed to the patient will be personalized, depending on his needs, the response to the chosen therapeutic methods and his tolerance to the applied physical-kinetic program. Prevention of sprains and strains is achieved by stretching exercises for the ankles, knees, hips, hand joints and neck.

Physical activities or exercises, carried out constantly, have the role of maintaining mobility and reducing the risk of contractures and deformations caused by the disease. Therapeutic and reflexogenic massage can beneficially complement a patient’s recovery program. Current research is intended to improve current physiotherapy practice and treatment itself, contributing to an improved quality of life for these patients.

2.2. Respiratory physiotherapy

Assessment of respiratory function is done annually from the moment of diagnosis and every six months after loss of ambulation.

The guidelines for respiratory care contain specific techniques for respiratory re-education with the aim of obtaining respiratory patterns adapted to the stages of the disease, the application of postural bronchial drainage, cough re-education, respiratory gymnastics and their role in the early stages of the disease [4].

An increase in the quality of respiratory care in patients with DMD was achieved, and the ability to provide respiratory support, led to a change in their main causes of death.

Thus, the main cause was no longer respiratory failure, but acute heart damage caused by dilated cardiomyopathy, fibrosis, conduction disturbance.

2.3. Management of cardiac disfunctions

Assessment of respiratory function is done annually from the moment of diagnosis and every six months after loss of ambulation.

The management of cardiac Prevention of sprains and strains is achieved by stretching exercises for the ankles, knees, early detection of heart failure symptoms hips, hand joints and neck. Physical activities or exercises, carried out constantly, have the role of maintaining time of diagnosis and annually thereafter. mobility and reducing the risk of Evaluation should include clinical contractures and deformations caused by examination, echocardiography, cardiac the disease. Therapeutic and reflexogenic magnetic resonance imaging (MRI) and massage can beneficially complement a electrocardiography. patient’s recovery program. Current Periodic evaluation of patients can research is intended to improve current establish their physical condition and physiotherapy practice and treatment itself, cardiorespiratory adaptation to exercise. contributing to an improved quality of life. The results obtained in the evaluations will be used to dose the intensity of the physical effort applied within the kinetotherapeutic recovery programs.

2.4. Management of osteo-muscular manifestations

Specific to DMD is the appearance of muscle contractions (a shortening of the muscles that causes deformation) of the limbs and at the level of the spine the appearance of scoliosis accompanied by lumbar hyperlordosis.

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With the loss of locomotion comes a rapid progression of muscle and tendon contractures and scoliosis. Altered bone metabolism, with reduced bone mineralization, is affected by corticosteroid treatment and therefore compression fractures may occur.

The main objective to control the evolution of osteo-muscular disorders is to improve or preserve muscle function and to prevent secondary bone deformations (primarily at the level of the spine).

If orthopedic interventions and surgical procedures are needed, they must be analyzed by the care team, which will include the orthopedist and surgeon, the physiotherapist, the orthotist and the occupational therapist.

For patients who have preserved their mobility and ambulation, the goals of kinetic recovery aim at preventing retraction of the Achilles tendons, knee and hip flexors, and iliotibial bands.

Physiotherapy and wearing special orthoses aim to improve and maintain orthostatic posture and gait. From the moment of losing the ability to ambulate, it is necessary to focus on adopting appropriate sitting positions, either in bed or on the wheelchair, with the aim of preventing the worsening of scoliosis and musculotendinous contractures of the lower limbs and to increase the patient's comfort.

Also, a kinetotherapeutic program will be applied that will aim to prevent contractures at the level of the upper limbs, with the main goal of preserving fine motor skills [13].

2.5. Management of neuropsychiatric disorders

In patients affected by DMD, neurobehavioral disorders such as ADHD (hyperactivity/hypoactivity), manifestations of the autistic spectrum, obsessive-compulsive disorders, but also emotional disorders such as depression and anxiety are common. In this sense, the physiotherapist can use muscle relaxation techniques, sedative and muscle relaxant massage, hydro-kinetotherapy, magnetic field therapy, etc.

The multidisciplinary team that monitors each patient must also include a psychologist or a psychiatrist, as appropriate, with training and experience in the diagnosis, evaluation and treatment of mental disorders in the context of Duchenne disease.

Complex neuropsychiatric and psychological assessment is necessary for the correct diagnosis, especially when there are deficiencies and delays in the dynamics of the patient's development.

Neuropsychological evaluations should be done when cognitive disability and emotional and behavioral adjustment difficulties are evident.

In order to monitor the neuropsychological evolution of the patient, assessments should be done periodically, from the moment of diagnosis of mental health problems.

Both patients and their families need psychosocial support to be able to overcome the effects of the disease's impact on them [3].

Further research carried out with the aim of enriching and improving the knowledge related to the manifestations of DMD, provides specific
recommendations that aim to identify, monitor and address them, and is extremely important for the goal of increasing the life quality of patients and their caregivers [2].

3. Conclusions

Although the recovery methods and guides of the pathology are not fully defined, the complex approach, and the continuous research to discover the most effective ways to recovery, promises a better care of the patients, and an increase in the quality of life of both those affected and their families, which are part of the recovery process, most of the time.

The goals of surveillance and care protocols for patients with DMD should include increased attention to early detection of cases among children. The sooner the detection is done, the greater is the patient's chance of increasing his life span and decrease the rate of complications. Improving the quality of medical care and the methods of clinical and paraclinical examination of children with DMD, will allow controlling the evolution of the disease and prevent the occurrence of complications.

In conclusion, research to define effective methods of rehabilitation for patients with DMD is an important aspect that must be supported, and for which it is necessary a permanent concern on the part of all medical specialties.

References

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