Chapter 1

Functional Evaluation for Duchenne Muscular Dystrophy

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Introduction

Duchenne Muscular Dystrophy (DMD) is a genetic, progressive and irreversible disorder linked to the X chromosome, which affects one in every 3,500 male children born alive. Induces muscle weakness as result of a deficiency or absence of the protein dystrophin, resulting in functional motor abilities loss, which starts when the child is between three and seven years old. Falls and difficulty running, rising from the floor, and climbing steps are common at this phase. Gait loss occurs between nine and twelve years of age [1,2].

Functional assessment usually includes the evaluation of muscle strength and joint mobility. The development of evaluation protocols has become increasingly important for clinical evaluation or as biomarker in research projects. The most used functional scales to assess muscular dystrophy are the Vignos Scale [3], the Brooke scale [4], the Hammersmith Functional Motor Scale [5], the Egen Klassifikation [6], the Motor Function Measure [7] and the North Star Ambulatory Assessment [8].

Vignos et al. [3] elected functional activities such as walking, going up and down stairs, sitting and standing from the chair and from the floor as essential in classifying the stages of patients with DMD. Vignos scale ranges from 1 (less severe) to 10 (more severe cases).

The Brooke Scale assesses subjectively the upper limb function in Duchenne and Becker muscular dystrophies [4]. It involves measuring the timed performance of six tasks, which require more proximal than distal control [9].
Hammersmith Functional Motor Scale aims to assess motor skills and to monitor the clinical progression of patients with muscular dystrophy. It has 20 items and ranges from 0 to 40. Higher scores indicate higher functional independence [5]. It evaluates upper and lower limbs, trunk and pelvis. Score zero means that the patient is incapable of performing the task, one is given when assistance is needed and two when the patient can perform the task without any assistance [10].

The Egen Klassifikation (EK) aims to quantify the functional limitations in patients in advanced phases of DMD and other neuromuscular diseases. It is divided in 10 categories and scores the tasks from 0 to 3, ranging from 0 to 30. Higher scores denote more severe functional impairments [11,6,12]. This scale is specific for wheelchair-dependent patients and it is strongly correlated to muscle strength, contractures, number of years of wheelchair dependence and forced vital capacity. The score on EK may be considered to consider the prescription of non-invasive ventilation [13,11].

The Motor Function Measure is indicated for neuromuscular diseases in general. It provides detailed assessment of both proximal and distal muscles [14] and evaluates patients from 7 to 60 years of age [7]. It has good reliability and responsiveness [7,15]. MFM-32 has 32 items, divided in three dimensions: D1–standing and transferring; D2–axial and proximal control; D3–distal control [7]. MFM-20 contains 20 of the 32 items of MFM-32. It can be used with children between 2 and 7 years of age [16].

North Star Ambulatory Assessment (NSAA) is used for ambulatory patients with DMD. It derives from Hammersmith Functional Motor Scale, and includes items as running and jumping, possible for pre-clinical patients. It has 17 activities and allows time measurement on 10 meters walking and rising from the floor as additional information. It has good reliability and good to moderate responsiveness [8,17,18].

Although they were not originally developed for patients with muscular dystrophies, two scales have been frequently used with this population. The Medical Research Council Scale [19] and the six-minute walking test [20] are common in clinical and research protocols for muscular dystrophy assessment.

The Medical Research Council Scale (MRC) assess muscle strength. It ranges from 0 to 5 and it is reliable when applied by the same professional [19] or by members of a well-trained and experienced group of professionals [21]. In muscular dystrophy muscle weakness results in the progressive loss of functional abilities.

The six-minutes walking test is an outcome measure to assess functional capacity in DMD. It was firstly developed for patients with cardiopulmonary diseases and it evaluates the distance covered in six minutes. Studies have also described the step length, cadence, velocity, falls,
cardiac frequency and arterial blood pressure obtained during the performance of this task. It has excellent reproducibility and sensitivity to detect DMD progression [20]. Mazzone et al. [17] reports that the association of the six minute walking test and the North Star Assessment is useful to assess the motor function in ambulatory patients with DMD.

More recently, the compensatory movements developed by children to maintain the performance of several activities have also been described and quantified. With the progression of DMD, there is a need to employ compensatory movements to perform functional tasks [22]. Significant variation is observed, due to genetic and environmental heterogeneity [23].

**Functional Evaluation Scale for DMD**

The Functional Evaluation Scale for DMD (FES-DMD) aimed to quantify and score compensatory movements in specific activities [24,22,25,26]. It has four domains: D1–sitting and standing from the chair [25]; D2–10 meters walking [26]; D3–climbing up and down steps [24]; D4–sitting and standing from the floor [22].

FES-DMD is used to evaluate and follow important changes in muscular synergies [27]. Its development and testing focused on demonstrating that the compensatory movements performed during everyday activities are observable, analyzable and measurable, and thus able to reflect the evolution of Duchenne muscular dystrophy (DMD).

The assessment with FES-DMD is performed by therapists and involves recording and watching the films of the functional activities, collecting data, generating partial and total scores, timing each activity and generating reports. To facilitate all these tasks, we developed a software that uses Brazilian Portuguese and was called FES-DMD-DATA. Its usability was demonstrated in a recent study [28].

The use of FES-DMD is simple, inexpensive, practical, and also provides permanent and standardized records. FES-DMD is sensitive to functional changes over time, with consistent responsiveness. FES-DMD makes physical therapy clinical evaluation faster and can less stressful for the patient. As the tasks are recorded, activities do not have to be repeated by patients for complete assessment. The risk of omitting relevant details is lower.

Domain 1 (FES-DMD-D1) evaluates sitting and standing from the chair. The movements of sitting and standing chair are highly relevant to physical therapy evaluation of patients not only with muscular dystrophies but with neurological diseases or dysfunctions of the lower limbs or trunk in general [29,30].
FES-DMD-D1 aims to detect compensatory movements on different phases of this task, as knee hyperextension, hip rotation, and lateral trunk flexion with or without rotation, usually associated with hyperextension or rotation, and head lateral flexion. We found a moderate correlation between the scores obtained by FES-DMD-D1 and the time of performance of sitting and standing from the chair and between the age and the activity of sitting [25,31].

Changes in speed of sitting and rising from a chair may be clinically significant indicator of therapeutic intervention [32]. Different studies have used this task to evaluate the response to treatment with corticosteroids [33,34].

Jeng et al. [29] described the presence of stages during the lifting movement of the chair: flexion phase, transfer, extension and stabilization. Millington et al. [35] cite that the problems that can difficult sitting and rising from a chair are muscle weakness, especially quadriceps and spine erector, decreased perception or sensory impairment, balance and coordination dysfunctions. Galli et al. [36] noted that obesity, back pain and muscle weakness can disrupt sitting and rising from a chair.

In DMD, progressive muscle weakness, leads to balance and coordination dysfunction, which difficults the performance of sitting and rising from a chair, getting up and sitting on the floor and walking [37]. In FES-DMD-D1, Hukuda et al. [25] found moderate correlation between age and timed performance (r=0.69 and r=0.66, respectively, for sitting and rising from the chair), but no correlations with the Vignos Scale.

Domain 2 (FES-DMD-D2) was elaborated to provide detailed kinesiological analysis of gait, describing compensatory movements during 10 meters walking. The total score on the domain correlated to the timed performance and to age. Higher age and time were associated to higher number of compensatory movements.

Biomechanical and muscular adaptations explain how children with DMD walk for some period with limited muscle strength [38, 39]. The weakness of the muscles that stabilize the pelvis induces anterior tilt of the trunk, also associated with the shortening of the plantar flexor muscles [38]. Therefore, children walk on their toes and perform trunk lateral movements.

The weakness of extensor muscles results in higher use of hips flexor muscles and ankles plantar flexions to maintain body support and keep performing gait. The disease progresses gradually and gait speed and cadence decrease. Children are usually unable to walk around the age of twelve [39, 37].

The gait of children with DMD has been investigated with the kinematic analysis of specific joints associated with kinanthropometry, force platforms and electromyo-
graphy (Graudrealt et al., 2007). These devices are essential in experimental research. However, they are not feasible in clinical practice, when the assessment is carried out with the direct observation and descriptive records.

In this domain, moderate positive correlations were found between the total scores and timed performance (r=0.50). Patients with lower times showed less compensatory movements. The increase in the number of compensatory movements may generate extra biomechanical difficulties. Compensatory movements must be incorporated to the gait pattern and may cause cardiac and respiratory overload.

Domain 3 - FES-DMD-D3 - evaluates climbing up and down steps. It showed excellent reliability (repeatability and reproducibility) [24]. The phases of going up and down stairs are described in the literature. Studies discussed the risk of falls during stairs descent [40], the influence of age [41] and of orthoses [42]. The description of kinesiological characteristics during going up and down stairs was published by Mc Fadyen [43] and became an important reference on this topic.

In the study of Vignos et al. [3], the assessment of going up and down stairs was essential to determine the clinical staging of patients with DMD. Scott and Mawson [44] agreed that the evaluation of this activity could describe and detect changes in functional independence. Functional evaluation can be used as an additional parameter to measure disease progression [45] and develop treatment strategies.

In DMD, lower limbs muscle weakness begins with changes in hip and knee angles associated to deficits of ankle dorsal flexors and quadriceps [37]. When going down stairs, patients tend to accelerate the displacement of the center of mass forward [46].

Muscle strength is needed on the way up the stairs, to support the body weight against gravity. However, the demand is higher when descending stairs, because the boys must control movement acceleration with railing support. As DMD progresses, patients cannot perform eccentric contractions with the lower limbs antigravity muscles. In going up and down stairs, the time of going up stairs had highest correlation with FES-DMD that the time of going down (r=0.83 vs. r=0.40).

Stair climbing showed a strong correlation with the Vignos Scale (r=0.92, p<0.05) and, while going down stairs showed moderate correlation (r=0.70, p<0.05) [24]. This can be justified because the Vignos Scale considers climbing stairs as a milestone in the progression of patients with DMD [3]. Climbing up and down stairs were strongly related involving the timed motor performance (r=0.79, p<0.05) and FES-DMD score (r=0.83, p<0.05) 18 months prior to gait loss [27].
For the scarcity of studies in literature about the description of the movement sitting and standing from the ground in healthy children, Escorcio et al. [47] carried out a pilot study in order to observe how the healthy children would perform these activities. Based on this information they developed the FES-DMD-D4, which is composed of two parts, the evaluation of rising from the ground and sitting on the ground. The domain showed good repeatability and very good reliability.

Correlations were found between standing and sitting on the floor, age and Vignos scale. Escorcio et al. [22] found a weak correlation between sitting on the ground and Vignos Scale (r=0.21). However, rising from the floor was moderately correlated to Vignos (r=0.56).

The correlations between the scores and the timed performance were strong for standing (r=0.79) and poor for sitting on the ground [22]. With the evolution of DMD, progressive loss of eccentric control is observed, disrupting the performance of muscles that act against gravity. During the descent phase, boys accelerate the projection of hands to the ground and bend the trunk, increasing the risk of falling. During sitting on the ground, the compensatory movements are a typical way of dealing with the weakness of lower limbs muscles.

During standing from the floor, the most common compensatory movements occurred during the passage quadrupedalism to bipedalism, with frequent need for external support. This can be explained by factors such as difficulty contracting hip and trunk extensor muscles, difficulty during semi-kneeling because of the weakness of hip flexors, and difficulty performing hip knee and ankle extension in closed kinetic chain [27].

In the study of the activities of FES-DMD, the relationship between the number of compensatory movements and timed performance became evident. These findings indicate that timing these tasks can bring additional information on the progression of DMD. An increase in the number of compensatory movements indicates a poorer functional performance [37,27,25,26,24,22].

Respiratory Assessment in DMD

Another essential evaluation, due to the power loss of the trunk support muscles and breathing is the respiratory system clinical and functional assessment. Respiratory complications are the primary cause of morbidity and mortality in Duchenne muscular dystrophy (DMD) as progressive respiratory muscle weakness leads to hypoventilation [48,49] and/or recurrent atelectasis and pneumonia, secondary to decreased cough effectiveness [50,51]. Furthermore, it has been postulated that due to the decreased range of motion of the chest wall and lungs, stiffening of the ribcage may occur, reducing chest wall and lung compliance. Most deaths not due to respiratory complications are due to cardiomyopathy. Life has been described as being statistically prolonged by the use of part-time Noninvasive mechanical ventilation and by the
use of cardioprotective medications [52].

Garcia et al [53] evaluated the chest expansion by measuring the chest circumference in a group of healthy children and children with DMD. They found significant differences in all measures between groups, with the DMD group characterized by lower measures. When the values of chest expansibility were analyzed, greater values of free thoracic mobility (allowing use of compensatory movements) were found when compared to the guided evaluation (which restricts the use of compensatory movements), only in the DMD group.

To achieve greater thoracic expansibility, participants performed compensatory movements mainly in the head, associated with the shoulder and thoracic and lumbar spine. Therefore, these children created compensatory strategies, on the same manner that they compensated for other motor functions, with the aim of improving respiratory function. These compensatory movements seem to be beneficial as they allow an increase in thoracic expansibility.

The studies performed by Nicot et al [54] and Caromano et al [55] demonstrated a correlation between age and inspiratory and expiratory muscle strength in DMD patients. Caromano et al [55] also observed the presence of correlation between the percentage of fat with maximal expiratory pressure in patients with DMD. Despite generalized muscle dysfunction, diaphragmatic function is relatively well preserved. Several researchers have also studied the correlation between expiratory muscle strength and cough [56,57,58]. However, the relationship between cough capacity and respiratory muscle weakness has not been discussed.

Although expiratory muscle function is crucial for an effective cough, the lung volume attained before expiratory muscle contraction also plays an important role. If insufficient precough volume is obtained due to inspiratory muscle weakness, cough capacity decreases in spite of functional expiratory muscles [59]. As a result, a lack of deep breathing leads to atelectasis, where the lung and chest walls lose their elasticity and compliance in the chronic stage [60]. In addition, the loss of lung compliance may exacerbate cough weakness by restricting dynamic airway compression [59].

Park et al. [61] observed that expiratory muscle strength was more strongly correlated than inspiratory muscle strength with cough capacity. Risk of respiratory failure in neuromuscular diseases is directly related to lung capacity. Untreated patients with a vital capacity of one liter in DMD show 8% survival in 5 years [62]. Therefore, that maintenance of lung capacity may have a significantly positive effect on mortality.

McKim et al. [63] assessed the long-term effect on measures of forced vital capacity (FVC) before and after the introduction of regular lung volume recruitment
(LVR) maneuvers (breathstacking) in DMD. Cough peak flow (CPF) at initial introduction of LVR improved from 144.8 to 232.8 L/min. The mechanism of improvement in the long-term decline of FVC, however, is most likely related to changes in compliance of the lung and chest wall, particularly because LVR is a passive therapy, not associated with muscle strengthening. Currently, improvement has been observed even in the presence of progressively weaker respiratory muscles. Therefore, the rate of FVC decline improves dramatically with initiation of regular LVR in DMD patients.

Patients with severe pulmonary restriction without adequate tracheobronchial clearance of secretions often progress to respiratory failure requiring hospitalization, intubation, suction and invasive mechanical ventilation. Still, the presence of secretion in bronchi quickly leads to tracheobronchitis and/or bacterial pneumonia [64].

The higher survival in patients with DMD occurs not only with the combined therapy of non-invasive mechanical ventilation, but also with measures for cleaning tracheobronchial secretions. These measures include assisted cough by chest compression during coughing, and/or the lung inflation, performed with the aid of a ventilation bag [65]. Controlled trials demonstrated significant increases in lung volume recruitment by air stacking. Lung volume recruitment, whether by glottis air stacking or by passive lung insufflations, limits chest wall contractures and lung restriction, increases CPF and voice volume, and promotes lung growth and impedes chest deformity for children with DMD [66].

Assistance to cough is also effective with the use of mechanical assistance, that promote insufflations and exhalations [67]. The CPF, when less than 4.5-5 liters/sec, indicates the need for manually assisted coughing during periods of airway congestion. Cough flows were increased from 2.5 +/- 2.0 liters/sec to 4.3 +/- 1.7 liters/sec by manually assisted coughing [66].

Considering all the possibilities for clinical complications arising from muscular dystrophy, we reinforce that detailed and targeted evaluation should take place to better clinical decision making for meeting the specific needs of each patient.

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