AVALIAÇÃO FUNCIONAL EM DISTROFIA MUSCULAR DE DUCHENNE

FUNCTIONAL ASSESSMENT IN DUCHENNE MUSCULAR DYSTROPHY

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RESUMO

O desenvolvimento de protocolos de avaliação torna-se cada dia mais importante. A busca por avaliações padronizadas e fidedignas visa possibilitar um efetivo monitoramento da evolução dos pacientes. Na distrofia muscular de Duchenne (DMD) ocorre fraqueza muscular o que resulta na perda progressiva das habilidades funcionais. No tratamento da DMD cálculo de medidas de força como a Medical Research Council e algumas escalas como funcionais como a Egen Klassifikation, a Motor Function Measure, a Hammersmith Functional Motor, a Escala Funcional de Brooke, a Escala Vignos, o 6-Minute Walk Test e a North Star Ambulatory Assessment e FES-MD são utilizadas visando aumentar a acuidade das avaliações. Na maioria das escalas, os dados e informações gerados tendem para o classificatório e não para o descritivo, sendo que todas possuem vantagens e desvantagens em relação às outras. Mais recentemente foi elaborada e analisada a confiabilidade da Escala de Avaliação Funcional para DMD, composta por quatro domínios. Nos últimos anos, o número crescente de abordagens terapêuticas eficazes em DMD refletiu no aumento da necessidade de medidas de resultados validadas, confiáveis e responsivas para serem utilizadas também em ensaios clínicos. As escalas de avaliações devem ser sensíveis o suficiente para detectar as alterações clinicamente significativas que ocorrem ao longo do tempo. Conhecendo a evolução da DMD, faz-se necessário, escolher o método de avaliação mais adequado para cada paciente a fim de traçar um plano terapêutico que deve ser iniciado o mais precocemente possível dentro de uma abordagem interdisciplinar.

Palavras chave: distrofia muscular de Duchenne, avaliação, escalas

ABSTRACT

The development of evaluation protocols becomes increasingly important. The search for standardized and reliable assessments aims to enable effective monitoring of patient outcomes. In Duchenne muscular dystrophy (DMD) muscle weakness occurs which results in the progressive loss of functional abilities. In the treatment of DMD calculation force measures such as the Medical Research Council and some scales as functional as Egen Klassifikation, the Motor Function Measure, the Hammersmith Functional Motor Scale Functional Brooke, the Vignos scale, the 6-Minute Walk Test and the North Star Ambulatory Assessment and FES-MD are used to increase the accuracy of the assessments. Most of the scales, the data and information generated tend to qualifying and not descriptive, and all have advantages and disadvantages over the other. More recently, it was drafted and analyzed the reliability of Functional Rating Scale for DMD, consisting of four domains. In recent years, the increasing number of therapeutic approaches effective in DMD reflected in an increased need for outcome measures validated, reliable, and responsive to being used also in clinical trials. Ratings scales should be sensitive enough to detect clinically significant changes that occur over time. Knowing the evolution of DMD, it is necessary to choose the most appropriate valuation method for each patient in order to map out a treatment plan that should be started as early as possible within an interdisciplinary approach.

Keywords: Duchenne muscular dystrophy, evaluation, scales.

1 - INTRODUCTION

Protocols with clinical assessment scales, laboratory tests and functional independence measures are necessary to define the clinical characteristics and the progression of neuromuscular diseases. Functional evaluation tools must be reliable to monitor the patients and to describe their outcomes properly.

Recently, the number of possible therapeutic approaches for patients with Duchenne muscular dystrophy (DMD) has increased. This reflected on the need of validated outcome measures

(MAZZONE et al., 2011; MAZZONE et al., 2013; KANG, 2013). The assessment of the functional independence is fundamental to show the response to treatment, to determine if the intervention was successful (VUILLEROT et al., 2012).

Many functional scales have been developed to assess the motor function of patients with neuromuscular diseases, but only some have had their reliability, responsivity and validity tested (MAZZONE et al., 2010). Functional assessment scales must be sensitive enough to detect clinical changes over time and are usually useful to complement the muscle strength measure (SCOTT; MAWSON, 2006; VUILLEROT et al., 2010).

In Duchenne muscular dystrophy, the patient gradually loses muscle strength. This results in progressive reduction of functional independence (KANAGAWA; TODA, 2006; BUSHBY et al., 2010). In the evaluation of patients with DMD, many scales are used. The Vignos classification (VIGNOS et al., 1963), the Brooke scale (BROOKE et al., 1981), the Hammersmith Functional Motor Scale (MAIN et al., 2003) and the Egen Klassifikation (MARTINEZ et al., 2006) provide classificatory information. The Medical Research Council (FLORENCE et al., 1992), the Motor Function Measure (BÉRARD et al., 2005), the North Star Ambulatory Assessment (MAZZONE et al., 2009), the six-minute walking test (MCDONALD et al., 2010) and the Functional Evaluation Scale for DMD (FERNANDES et al., 2010; ESCORCIO et al., 2011; HUKUDA et al., 2013; CARVALHO et al., 2014) provide more specific information for clinical decision-making, particularly in physical and occupational therapy practice.

The Motor Function Measure (BÉRARD et al., 2005), the North Star Ambulatory Assessment (MAZZONE et al., 2009) and mainly the Functional Evaluation Scale for DMD (FERNANDES et al., 2010; ESCORCIO et al., 2011; HUKUDA et al., 2013; CARVALHO et al., 2014) describe the compensatory movements in functional tasks. As DMD progresses, the number of compensatory movements increase, to minimize functional losses (HUKUDA et al., 2013; MARTINI et al., 2014).

Vignos Classification

The Vignos classification ranks the functional independence from zero to ten. Zero means pre-clinical stage and 10 is the score given for patients restricted to bed (VIGNOS et al., 1963). This scale shown the general functional state, but is not sensitive to assess DMD progression (BÉRARD et al., 2005; MAIN et al., 2003), mainly in wheelchair dependent patients (MAIN et al., 2003).

Egen Klassifikation

The Egen Klassifikation (EK) aimes 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 functional impairments (STEFFENSEN et al., 2001; MARTINEZ et al., 2006; FAGOAGA et al., 2013).

It is specific for wheelchair-dependent patients. EK is strongly correlated to muscle strength, contractures, number of years of wheelchair dependence and forced vital capacity. It provides clinical information to help decision-making involving the prescription of non-invasive ventilation (LYAGER et al., 1995; STEFFENSEN et al., 2001).

Hammersmith Functional Motor Scale

Hammersmith Functional Motor Scale aims to assess motor skills and to monitor clinical progression. It has 20 items and ranges from 0 to 40. Higher scores, indicate higher functional independence (MAIN et al., 2003). It evaluates upper and lower limbs, trunk and pelvis and scores as 0 when the patient is incable of performing the task, 1 when assistance is needed and 2 when the patient can perform the task without any assistance. It has been shown to be

reliable to evaluate children with spinal amiotrophy (MERCURI et al., 2006).

Brooke Scale

The Brooke Scale assesses subjectively the UL function in Duchenne and Becker muscular dystrophies (BROOKE et al., 1981). It involves measuring the time of performance of six tasks, which require more proximal than distal control (JUNG et al., 2012). It has been shown to be valid and reliable and it is easy and fast to use. However, is not sensitive to detect the disease progression.

Medical Research Council

The Medical Research Council scale (MRC) assess muscle strength with the patient positioned on dorsal, ventral and lateral decubitus. It ranges from 0 to 5. 0 - no contraction, 1 – observable contraction, 2 - active movement, 3 - movement against the gravity force, 4 - move against gravity and resistance forces (given by the therapist´s hand) and 5 - normal strength.

It is reliable when applied by the same professional (FLORENCE et al., 1992) or by members of a well-trained and experienced group of professionals (SOUZA et al., 2014). It can be represented as the Total Muscle Strength (SCOTT et al., 1982).

Total Muscle Strength = Sum of bilateral scores x 100

Number of muscles tested x 5

The relationship between muscle strength and functional performance is controversial in literature. Some studies showed poor relationship between these variables (KANAGAWA; TODA, 2006; MAZZONE et al., 2010), while others showed strong relationships between muscle strength and motor function (DINIZ et al., 2012; SOUZA et al., 2014).

Six minute walking test

The six-minute walking test is an outcome measure to assess functional capacity in DMD. It was firstly developed for patients with cardiopulmonary diseases and evaluates the distance covered in six minutes. Studies have also described the step length, cadence, velocity, falls, cardiac frequency and arterial blood pressure on this task. It has excellent reprodutibility and sensibility to detect DMD progression (MCDONALD et al., 2010). Mazzone et al. (2010) reports that the association of the six minute walking test and the NSAA is useful to assess the motor function in ambulatory patients with DMD.

Motor Function Measure

The Motor Function Measure (MFM) is viable for patients with neuromuscular diseases, aged from 6 to 60 years (BÉRARD et al., 2005). It has good reliability and responsivity (BÉRARD et al., 2005; VUILLEROT et al., 2012). MFM-32 has 32 items, divided in three dimensions: D1 – standing and transferring; D2 – axial and proximal control; D3 – distal control. Each item is scored 0 – cannot initiate or maintain the initial position, 1 – initiates performance, 2 – the movement is imperfect or incomplete 3 – the movement is complete and controlled (BÉRARD et al., 2005). MFM-20 contains 20 of the 32 items of MFM-32. It can be used with children between 2 and 7 years of age (DE LATTRE et al., 2013). MFM is indicated for all kind of neuromuscular disease, because it provides detailed assessment of both proximal and distal muscles (IWABE et al., 2008).

North Star Ambulatory Assessment

North Star Ambulatory Assessment (NSAA) assesses ambulatory patients with DMD. It derives from Hammersmith Functional Motor, and includes items as running and jumping, possible for pre-clinical patients treated with steroids. It has 17 activities, scored as 2 – normal; 1 – modified but performed independently; 0 – assistance needed. It allows the measurement of time on 10-m walking and rising from the floor as additional information, without applying to the total score. It has good reliability and good to moderate responsivity (MAZZONE et al., 2009;

MAZZONE et al., 2010; MAYHEW et al., 2013).

Functional Evaluation Scale for DMD

The Functional Evaluation Scale for DMD aimed to quantify and score compensatory movements in specific activities. It has four domains: D1 – sitting and standing from the chair (HUKUDA et al., 2013); D2 – walking (CARVALHO et al., 2014); D3 – climbing up and down steps (FERNANDES et al., 2010); D4 – sitting and standing from the floor (ESCORCIO et al., 2011).

D1 has two phases: sitting and standing from the chair. The sitting evaluation is divided into three phases, namely: phase I or flexion phase, composed of five items; phase II or contact of the hip with the chair phase, composed of four items; and phase III or extension phase, composed of three items. The standing evaluation is divided into three phases: phase I or flexion phase, composed of four items; phase II or transference phase, composed of six items; and phase III or extension phase, composed of four items (HUKUDA et al., 2013).

D2 is divided on three phases: phase I or stance, composed of five items; phase II or swing, composed of three items; phase III or stance and swing, composed of six items (CARVALHO et al., 2014).

D3 is divided in climbing up and climbing down steps. The assessment of climbing up steps is divided in five phases. Phase I or preparation - standing, composed of three subitems; phase II or propulsion, composed of eight subitems; phase III or swing - pelvis, composed of seven items; phase IV or swing – lower limbs, composed of three items; phase V or support, composed of four items. The assessment of climbing down is divided in four phases, phase I or preparation – standing, composed of three items; phase II or propulsion, composed of five items, phase III or swing, composed of ten items, and, phase IV or support, composed of nine items (FERNANDES et al., 2010).

D4 is composed by two parts: standing from the floor and sitting on the floor. Standing from the floor is divided in five phases. Phase I or dorsal decubitus to flexion and/or dorsal decubitus to lateral decubitus, composed of five items; phase II or sitting, composed of three items; phase III or quadrupedalism, composed of two items; phase IV or kneeling, composed of four items and, phase V or standing, composed of five items. Sitting on the floor is divided in three phases: phase I or trunk flexion, composed of four items; phase II or ankles and knees flexion, composed of two items and phase III or sitting, composed of six items (ESCORCIO et al., 2011).

To get the final score, all scores on the subitems of each phase and all the phases must me added. Higher scores denote a higher number of compensatory movements. FES-DMD also proposes the measurement of the time of each activity. All activities of FES-DMD have shown good reliability intra and inter-raters and good responsiveness (FERNANDES et al., 2010; ESCORCIO et al., 2011; HUKUDA et al., 2013; CARVALHO et al., 2014).

FES-DMD proposes documenting all tasks in videos to allow repeating the domains and phases as many as needed without causing fatigue to the patient (MERCURI et al., 2008). This allows more careful and detailed observation and analysis and generates a permanent databank. As DMD progresses, the muscle strength diminishes and the number of compensatory movements increase, to minimize functional losses (VUILEROTT et al., 2010; HUKUDA et al., 2013; MARTINI et al., 2014).

2 – CONCLUSION

The scales used in neuromuscular diseases, more specifically in DMD, reviewed in the present chapter, show some or all the psychometric properties, such as reliability, validation and responsiveness. DMD shows genetical and

clinical heterogeneity (BROOKE et al., 1989), therefore, the evaluation scales are fundamental to allow a more specific assessment and follow-up during the multiprofessional rehabilitation program.

3 – REFERENCES

BÉRARD, C.; PAYAN, C.; HODGKINSON, I.; FIAN, J. A motor function measure scale for neuromuscular diseases. Construction and validation study. Neuromuscular Disorders, v. 15, p. 463-470, 2005.

BROOKE, M. H.; GRIGGS, R. C.; MENDELL, J. R.; FENICHEL, G. M.; SCHUMATE, J. B.; PELLEGRINO, R. J. Clinical Trial in Duchenne dystrophy. I. The design of the protocol. Muscle Nerve, v. 4, p. 186-197, 1981.

BROOKE, M. H.; FENICHEL, G. M.; GRIGGS, R. C.; MENDELL, J. R.; MOXLEY, R.; FLORENCE, J. et al. Duchenne muscular dystrophy: patterns of clinical progression and effects of supportive therapy. Neurology, v. 39, p. 475-481, 1989.

BUSHBY, K.; FINKEL, R.; BIRNKRANT, D. J.; CASE, L. E.; CLEMENS, P. R.; CRIPE, L. et al. Diagnosis and management of Duchenne muscular dystrophy, part 2: implementation of multidisciplinary care. Lancet Neurology, v. 9, p. 177-189, 2010.

CARVALHO, E. V.; HUKUDA, M. E.; ESCORCIO, R.; VOOS, M. C.; CAROMANO, F. A. Development and reliability of the functional evaluation scale for Duchenne muscular dystrophy, Gait domain: a pilot study. Physiotherapy Research International. In press.

DE LATTRE, C.; PAYAN, C.; VUILLEROT, C.; RIPPERT, P.; DE CASTRO, D.; BÉRARD, C.; POIROT, I.; MFM-20 STUDY GROUP. Motor function measure: validation of a short form for young children with neuromuscular diseases. Archives of Physical Medicine and Rehabilitation, v. 94, p. 2218-2226, 2013.

DINIZ, G.; LASMAR, L.; GIANNETTI, J. Motor assessment in patients with Duchenne muscular dystrophy. Arquivos de Neuro-psiquiatria, v. 70, p. 416-421, 2012.

ESCORCIO, R.; CAROMANO, F. A.; HUKUDA, M. E.; FERNANDES, L. A. Y. Development of an evaluation scale for sitting and standing from the ground for children with Duchenne muscular dystrophy. Journal of Motor Behavior, v. 43, p. 31-36, 2011.

FAGOAGA, J.; GIRABENT-FARRÉS, M.; BAGUR-CALAFAT, C.; FEBRER, A.; STEFFENSEN, B. F. Translation and validation of the Egen Klassifikation scale for the Spanish population: functional assessment for non-ambulatory individuals with Duchenne's muscular dystrophy and spinal muscular atrophy. Revista de Neurologia, v. 56, p. 555-561, 2013.

FERNANDES, L. A. Y.; CAROMANO, F. A.; HUKUDA, M. E.; ESCORCIO, R.; CARVALHO, E. V. Elaboration and reliability of functional evaluation on going up and down stairs scale for Duchenne muscular dystrophy (DMD). Revista Brasileira de Fisioterapia, v. 14, p. 518-524, 2010.

FLORENCE, J. M.; PANDYA, S.; KING, W. M.; ROBISON, J. D.; BATY, J.; MILLER, J. P.; SCHIERBECKER, J.; SIGNORE, L. C. Intrarater reliability of manual muscle test (Medical Research Council scale) grades in

Duchenne's muscular dystrophy. Physical Therapy, v. 72, p. 115-122, 1992.

HUKUDA, M. E.; ESCORCIO, R.; FERNANDES, L.; CARVALHO, E.; CAROMANO, F. Evaluation scale development, reliability for sitting and standing from the chair for Duchenne muscular dystrophy. Journal of Motor Behavior, v. 45, p. 117-126, 2013.

IWABE, C.; MIRANDA-PFEILSTICKER, B. H.; NUCCI, A. Medida da função motora: versão da escala para o português e estudo de confiabilidade. Revista Brasileira de Fisioterapia, v. 12, p. 417-424, 2008.

JUNG, I. Y.; CHAE, J. H.; PARK, S. K.; KIM, J. H.; KIM, J. Y.; KIM, S. J.; BANG, M. S. The correlation analysis of functional factors and age with duchenne muscular dystrophy. Annals of Rehabilitation Medicine, v. 36, p. 22-32, 2012.

KANAGAWA, M.; TODA, T. The genetic and molecular basis of muscular dystrophy: roles of cell-matrix linkage in the pathogenesis. Journal of Human Genetics, v. 51, p. 915-926, 2006.

KANG, P. B. Beyond the gowers sign: measuring outcomes in Duchenne muscular dystrophy. Muscle Nerve, v. 48, p. 315-317, 2013.

LYAGER, S.; STEFFENSEN, B.; JUHL, B. Indicators of need for mechanical ventilation in Duchenne muscular dystrophy and spinal muscular atrophy. Chest, v. 108, p. 779-785, 1995.

MAIN, M.; KAIRON, H.; MERCURI, E.; MUNTONI, F. The Hammersmith functional motor scale for children with spinal muscular atrophy: a scale to test ability and monitor progress in children with limited ambulation. European Journal of Paediatric Neurology, v. 7, p. 155-159, 2003.

MARTINEZ, J. A. B.; BRUNHEROTTI, M. A.; ASSIS, M. R.; SOBREIRA, C. F. R. Validação da escala motora funcional EK para a língua portuguesa. Revista da Associação Médica Brasileira, v. 52, p. 347-351, 2006.

MARTINI, J.; VOOS, M.; HUKUDA, M. E.; RESENDE, M.; CAROMANO, F. Compensatory movements during functional activities in ambulatory children with Duchenne Muscular Dystrophy. Arquivos de Neuro-psiquiatria, v. 72, p. 5-11, 2014.

MAZZONE, E. S.; MESSINA, S.; VASCO, G.; MAIN, M.; EAGLE, M.; D'AMICO, A. et al. Reliability of the North Star Ambulatory Assessment in a multicentric setting. Neuromuscular Disorders, v. 19, p. 458–461, 2009.

MAZZONE, E.; MARTINELLI, D.; BERARDINELLI, A.; MESSINA, S.; D'AMICO, A.; VASCO, G. et al. North Star Ambulatory Assessment, 6-minute walk test and timed items in ambulant boys with Duchenne muscular dystrophy. Neuromuscular Disorders, v. 20, p. 712-716, 2010.

MAZZONE, E.; VASCO, G.; SORMANI, M. P.; TORRENTE, Y.; BERARDINELLI, A.; MESSINA, S. et al. Functional changes in Duchenne muscular dystrophy: a 12-month longitudinal cohort study. Neurology, v. 77, p. 250-256, 2011.

MAZZONE, E. S.; PANE, M.; SORMANI, M. P.; SCALISE, R.; BERARDINELLI, A.; MESSINA, S. et al. 24 month longitudinal data in ambulant boys with Duchenne muscular dystrophy. PLoS One, v. 8, e52512, 2013.

MAYHEW, A.; CANO, S.; SCOTT, E.; EAGLE, M.; BUSHBY, K.; MANZUR, A.; MUNTONI, F.; NORTH STAR CLINICAL NETWORK FOR NEUROMUSCULAR DISEASE. Detecting meaningful change using the North Star Ambulatory Assessment in Duchenne muscular dystrophy. Developmental Medicine; Child Neurology, v. 55, p. 1046-1052, 2013.

MERCURI, E.; MESSINA, S.; BATTINI, R.; BERARDINELLI, A.; BOFFI, P.; BONO, R. et al. Reliability of the Hammersmith functional motor scale for spinal muscular atrophy in a multicentric study. Neuromuscular Disorders, v. 16, p. 93-98, 2006.

MERCURI, E.; MAYHEW, A.; MUNTONI, F.; MESSINA, S.; STRAUB, V.; VAN OMMEN, G. J. et al. Towards harmonisation of outcome measures for DMD and SMA within TREAT-NMD; Report of three expert workshops: TREAT-NMD/ENMC Workshop on outcome measures, 12th–13th May 2007, Naarden, The Netherlands; TREAT-NMD Workshop on outcome measures in experimental trials for DMD, 30th June–1st July 2007, Naarden, The Netherlands; Conjoint Institute of Myology TREAT-NMD Meeting on physical activity monitoring in neuromuscular disorders, 11th July 2007, Paris, France. Neuromuscular Disorders, v. 18, p. 894–903, 2008.

MCDONALD, C. M.; HENRICSON, E. K.; HAN, J. J.; ABRESCH, R. T.; NICORICI, A.; ELFRING, G. L. et al. The 6-minute walk test as a new outcome measure in Duchenne musculardystrophy. Muscle Nerve, v. 41, p. 500-510, 2010.

SCOTT, O. M.; HYDE, S. A.; GODDARD, C. ; DUBOWITZ, V. Quantitation of muscle function in children: a prospective study in Duchenne muscular dystrophy. Muscle Nerve, v. 5, p. 291-301, 1982.

SCOTT, O. M.; MAWSON, S. J. Measurement in Duchenne muscular dystrophy: considerantions in the development of a neuromuscular assessment tool. Developmental Medicine; Child Neurology, v. 48, p. 540-544, 2006.

SOUZA, M.; HUKUDA, M. E.; FÁVERO, F.; VOOS, M.; CAROMANO, F.; OLIVEIRA A. Relationship between muscle strength and motor function in Duchenne muscular dystrophy. Journal of Musculoskeletal and Neuronal Interactions. In press.

STEFFENSEN, B.; HYDE, S.; LYAGER, S.; MATTSSON, E. Validity of the EK scale: a functional assessment of non-ambulatory individuals with Duchenne muscular dystrophy or spinal muscular atrophy. Physiotherapy Research International, v. 6, p. 119-134, 2001.

VIGNOS, P. J. JR.; SPENCER, G. E. JR.; ARCHIBALD, K. C. Management progessive muscular distrophy of childhood. JAMA, v. 184, p. 89-96, 1963.

VUILLEROT, C.; GIRARDOT, F.; PAYAN, C.; FERMANIAN, J.; IWAZ, J.; DE LATTRE, C. et al. Monitoring changes and predicting loss of ambulation in Duchenne muscular dystrophy with Motor Function Measure. Developmental

Medicine; Child Neurology, v. 52, p. 60-65, 2010.

VUILLEROT, C.; PAYAN, C.; GIRARDOT, F.; FERMANIAN, J.; IWAZ, J.; BÉRARD, C. et al. Responsiveness of the motor function measure in neuromuscular diseases. Archives of Physical Medicine and Rehabilitation, v. 93, p. 2251-2256, 2012.

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