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Motor Function Measure Scale (MFM): New Instrument for Follow-Up Brazilian Patients with Neuromuscular Disease

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1. Introduction

Neuromuscular disorders include a variety of conditions that affect motor neurons (spinal muscular atrophy), peripheral nerves (neuropathy), neuromuscular junction (myasthenia gravis) or muscle fibers (myopathy) (1).

Myopathy is characterized by primary and generally irreversible skeletal muscle tissue degeneration, including genetic, inflammatory, metabolic or endocrine disorders and the different are charaterized by the muscle fibre type affected, mode of inheritance, age of onset and course of evolution (2,3,4).

The term muscular dystrophy has been used in cases of rapidly progressive myopathy as well as of slow progressive degeneration of muscle, such as myotonic dystrophy (2).

Myotonic Dystrophy (MD) is defined as the most common inherited myopathy in adults, with multisystemic involvement (cardiovascular, respiratory, nervous, visual, endocrine), autossomal dominant pattern and distinct clinical manifestations (2,5,6). Depending on the genetic trait, MD is classified as type 1 (MD-1), type 2 (MD-2) (7), or type 3 (8); being the type 1 the most common and the type 3, very rare.

The MD-1, described by Steinert in 1909, is caused by expansion of CTG nucleotides repeat in the region of the gene for dystrophy myotonic protein kinase (DMPK) on chromosome 19; over 35 and ranging from 80 to more than 4,000 repetitions in the affected individuals. This abnormal protein is responsible for the disability of muscle, cardiac and nervous cells, involving several systems (9).

This disease can be classified in: congenital, infantile, classic (age of onset between 10 to 50 years) and with minimal involvement (10). Usually the earlier the onset of symptoms, the greater the number of repetitions of the nucleotides (7,11).

The muscle involvement is the main clinical feature with variations in the degree of weakness (facial, neck and distal muscles of limbs), as well as myotonia. In the congenital form, the deficit is prominent at birth without myotonia. In the infantile form, weakness is relatively mild, and in adults form there is slowly progression of the symptoms (11).

In contrast to muscular dystrophy, the congenital myopathy is described as non-progressive or slowly progressive and rarely fatal disease. Among them is included the Congenital Fibre Type Disproportion (CFTD) (12). The CFTD is a congenital myopathy described by Brooke (13) as having generalized weakness, hypotonia at birth and slow progression of symptoms associated with abnormal histological predominance of type 1 muscle fibres, and smaller size of at least 12% than type 2. The patients have abnormalities such as congenital hip dislocation, foot deformities, kyphoscoliosis, ligament laxity, high palate and underweight (14,15,16,17).

The CFTD pattern suggests a autossomal dominant trait although sporadic recessive cases has been described (18,19,20,21). Distinct mutations in the gene that codes the α-skeletal muscle actin (ACTA1), selenoprotein N (SEPN1) and α-tropomyosin (TPM3) proteins were identified in some cases, but the molecular mechanisms that cause the disparity of the fibres are still unknown (22,23).

Individuals with CFTD show different degrees of weakness, more severe in the early stages of development, especially in the lower limbs (20). Generally they have a good prognosis, but in some cases they may be associated with respiratory (14,20) or cardiac (16,21) failures.

Considering the CFTD and MD-1, both have weakness as the main physical limitation (17,24,25) and the individuals become more and more dependent to achieve their routine activities. Rehabilitation programs must measure and maximize patients motor skills and optimizing their functionality.

Now a days, several therapeutic techniques and other health professionals assessment tools can be used for patient selection, therapeutic monitoring and to establish prognosis for recovery (26).

Generally, the evaluations are qualitative test, not allowing for the individual assessment of the recovery of the better patients (27). Currently, Medical Research Council scale (MRC) for measuring muscle strength is being used for clinical examination and patient follow-up however, it does not reflect the real abilities of each individual (28)There are several scales to measure function in neuromuscular diseases, including the Barthel Index (BI), Vignos scale and Motor Function Measure (MFM) (28).

The MFM have been developed and validated for neuromuscular diseases by the research group of the Department of Pediatric Reeducation L'Escale, Lyon, France.This is a more comprehensive, specific and functional scale, analyzing the function of the head, trunk, proximal and distal segments in several neuromuscular diseases (28).

The scale comprises 32 items, including static and dynamic evaluations, divided into three dimensions:

- Dimension 1 (D1): a standing position and transfers, with 13 items
- Dimension 2 (D2): axial and proximal motor function, with 12 items.

- Dimension 3 (D3): distal motor function, with seven items, six of which are related to the upper limbs.

Each item is graduated on a 4-point scale (scores 0 to 3), with the instructions detailed in the scoring manual, specific to each item. Score 0 - can not start the requested task or can not keep the starting position. Score 1 – initializes the item. Score 2 - partially performs the requested movement or fully realized, but imperfectly. Score 3 - completes the item, with controlled movement (normal).

In cases of tendon retraction or joint limitation, the individual is graduated as not presenting adequate strength to perform the movement, preventing them from receiving the maximum degree. The total score and each dimension are expressed in percentages relative to maximum score (96 points).

In 2008, Iwabe et al. (29) demonstrated the reliability of the Portuguese version's MFM (P-MFM), showing a high correlation intra and inter examiner results.

The aim of this chapter is to describe the validation of the P-MFM and its applicability to evaluate the motor function in muscular myopathy and dystrophy individuals.

2. P-MFM Validation

The population comprised a total of 65 patients, 37 male and 28 female, average 33.09 years (8-60 years), with laboratory findings confirming clinical diagnosis of congenital muscular dystrophy (n = 7), Duchenne (n = 5), Becker (n = 4), limb girdle (n = 4), facioscapulohumeral (n = 8), distal myopathy (n = 4), mitochondrial (n = 3), centrocore (n = 6), congenital fibre types disproportion (n = 1), myotonic dystrophy (n = 21) and spinal muscular atrophy (n = 2); outcome in the Neuromuscular Diseases Clinic of the Faculty of Medical Sciences, Campinas State University (UNICAMP).

Patients were evaluated according to the P-MFM, BI and Vignos scales. All evaluations were performed by the same examiner and BI questions were answered by the patient, or in some cases with the help of their parents.

Statistical analysis - the total scores and each of the three dimensions of P-MFM were correlated with Vignos scale and BI by the Spearman correlation coefficient, with significance level of 5% ($p < 0.05$).

It was observed that in P-MFM scale, both its three dimensions and total score, were correlated negatively and significantly with the Vignos scale, and correlated positively and significantly with the BI (Table 1).

P-MFM	Vignos scale	BI
Dimension 1	-0,858*	0,946*
Dimension 2	-0,852*	0,871*
Dimension 3	-0,671*	0,736*
Total	-0,894*	0,980*

* - $p < 0,001$

Table 1. Correlation between P-MFM, Vignos scale and BI.

Validation is defined as the ability of an instrument to measure a particular aspect, and for this it is necessary the correlation with other validate scales, and similar characteristics (30). Miller et al. (31) defined some factors for the scale's development and validation. The instrument must represent the function at the moment, following the patient's evolution over the time, and each individual serving as his own control.

The MFM was developed for this purpose, containing items easily implementable and understandable by patients from different age groups (6-60 years). This scale has the capacity of analyze the most important motor functions and deficiency in several neuromuscular diseases. It can measure the activity of the proximal and distal segments members; as well as the standing position and transfering at the moment and over time (28).

The capacity of MFM scale to analyze the various body segments and their mobility in all neuromuscular diseases, empathizes its use in research and clinical. To be applied in Brazil, it was necessary perform the validation process of the Portuguese version, considering that the scale item was approved in reproducibility and reliability (29).

In Brazil, there are only two assessment instruments validated for patients with dystrophy (32).

The P-MFM validation study used two functional scales, the BI and Vignos scale (33,34,35,36). BI and Vignos scale are clinical instruments often used to assess the level of functionality in neuromuscular diseases (35). These two scales were used in the study by Nair et al. (37), with Duchenne muscular dystrophy, showing to be a valid instrument for assessing the functional limitations in this patients.

Previous validation studies about functional neuromuscular diseases using the BI and Vignos scale (28,38,39) demonstrated the correlation between them. Studies analyzing the functionality of individuals with neuromuscular diseases after clinical or surgical treatment, or correlating it to other parameters, such as muscle strength using these same scales (40,41,42,43,44) established them as easy to use instruments.

In this study, we found a high significant correlation among the P-MFM, Vignos scale and BI, allowing for validation of the Portuguese version of MFM.

3. Applicability of P-MFM in family with CFTD, associated muscle magnetic ressonance

We studied members of a family with clinical and laboratory CFTD. They were evaluated for muscle strength (MRC scale) and motor function by the scale P-MFM (29) and were previously examined in the Neuromuscular Diseases Clinic through physical examination, serological and neurophysiological tests and muscle biopsy. One sample was taken from the biceps muscle from the father in a family. The obtained sample was fixed in isopentane and frozen in liquid nitrogen. Sections were stained with hematoxylin-eosin (H & E), Gomori trichrome modified (TRI) or oil red O and analyzed by histochemical techniques for nicotinamide adenine dinucleotide phosphatase, nicotinamide dehydrogenase tetrazolium reductase (NADH-TR), succinate dehydrogenase (SDH) and immunohistochemistry for slow and fast myosin, desmin and alpha B crystalline.

Muscle magnetic resonance imaging (mMRI) was performed in magnetic field of 2.0 Tesla, T1-weighted images in axial plane for the leg muscles from each patient, according to De Cauwer et al. (45).

Image data were analyzed quantitatively according to the degree proposed by Mercuri et al. (46) and modified by Nucci (47).

0 = normal appearance
1 = slight appearance of "moth-food", with sporadic areas of hyperintensity.
2 = moderate appearance of "moth-food" with related areas of hyperintense spaced, comprising less than 30% of muscle volume.
= 2.5 appearance of "moth-food" with moderate hyper spaced areas, comprising 30 to 60% of muscle volume.
3 = severe appearance of "moth-food", with numerous areas of confluence of hyperintense with muscle still present in the periphery.
4 = complete fatty degeneration, with replacement of muscle by connective tissue and fat.

The family pedigree is shown in Figure 1. It was not possible to examine patient´s mother (I-1) and patient´s uncle (I-2). Throughout the family´s history, the patient´s mother was indicated as asymptomatic, and the uncle as having the phenotype very similar with myopathy aspects.. Thus, I-2 was marked as affected in the pedigree.

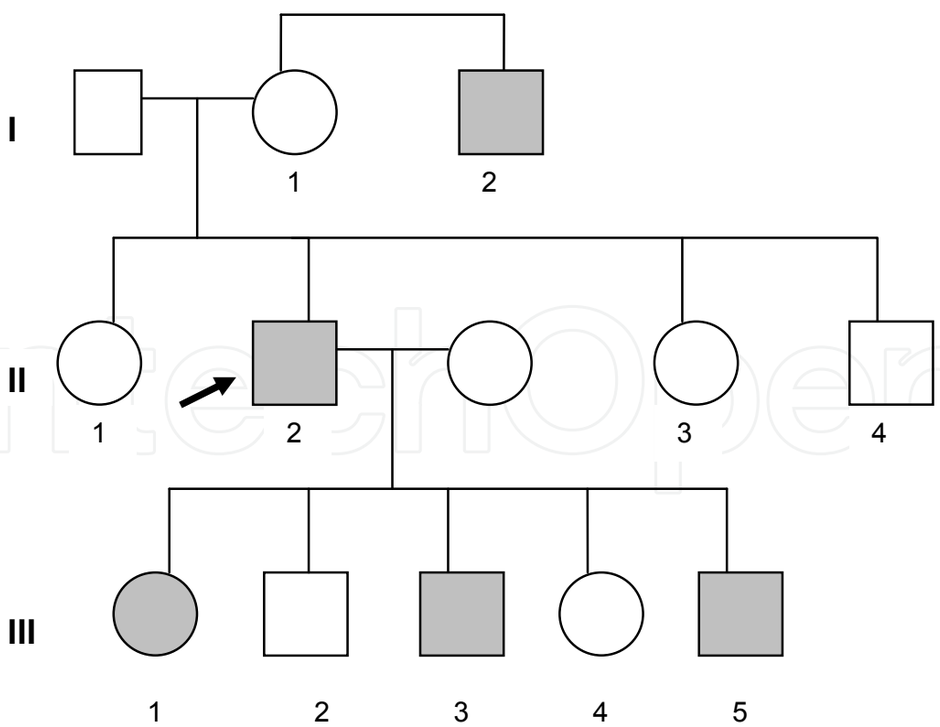

Fig. 1. Family pedigree. The grey figures are the affects cases. The white figures are the normal cases

CASE II -2 Male, 48-year-old with consanguineous parents, complaining about muscle weakness since childhood, considered a "sick child" due to limitations in physical activities and difficulty in gaining weight. The acquisition of motor milestones was delayed, just being able to walk around 5 years old. The initial clinical examination showed a collaborative and lanky patient (1.78 meters, 48 kg), with severe scoliosis dextro-convex compensate cervical, and high palate. A complex gait was observed due to the misalignment of the spinal cord and the tendency to walk with his feet fallen. The muscles stretch reflexes were hypoactive, but the cranial nerves, the cognition and sensibility were normal. The laboratory findings for creatine kinase (CK) showed 181 U / L (normal values below 170), and the study for motor and sensory nerves conduction were within normal limits. Electromyography (EMG) of the deltoid and biceps brachii showed the most potential of motor units with respect polyphasic, low amplitude and short duration, myopathy indicative,, despite the present right quadriceps were relatively normal. The electrocardiogram (ECG) and routine laboratory tests showed no abnormalities. The main morphologic abnormality in biopsy was a disproportion of the fibre types with small type 1

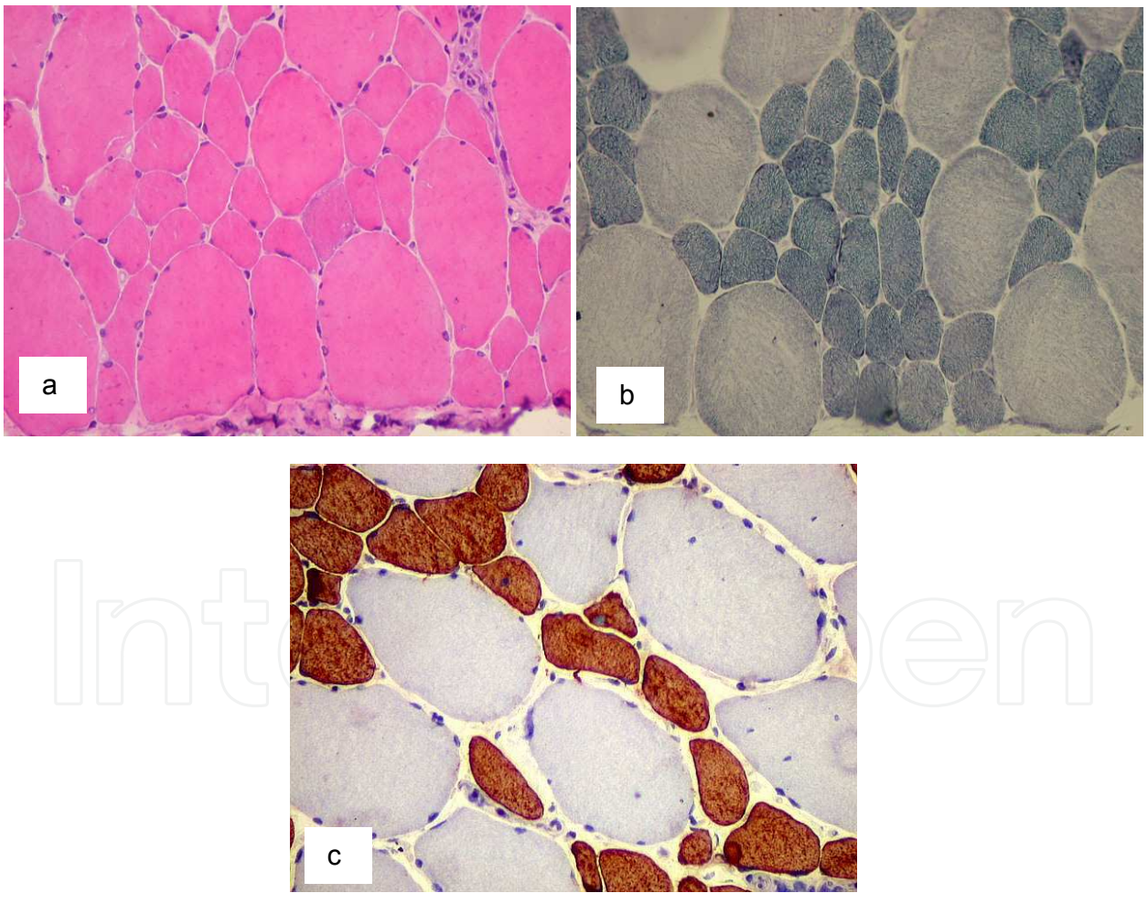

Fig. 2. a - H & E staining showing two populations of muscle fibres with different diameters average. b - NADH-TR staining where small diameter fibres show a higher oxidative activity (type 1) than the large fibres (type 2). c - Immunohistochemistry for myosin slow - small diameter fibres are positively stained for slow myosin fibres (type 1), in contrast to the larger fibres that are unmarked (type 2).

fibres (Figures 10 a-c). The electron microscopy showed neither central focus, minifocus, nemaline bodies nor mitochondrial alterations, also no protein deposits were observed.

The most significant data from the biceps muscle biopsy from case II-2 is illustrated in figure 2 c.

CASE III - 1 Female, 13 years old, daughter of the Case II-2, non-consanguineous parents. She was born at 40 weeks gestational age, 50 cm height, 2.670g weight, cesarean delivery for fetal distress, and a history of reduced fetal movements. The child presented a congenital hypotonia and delayed motor development, acquiring the standing posture approximately at 19 months of age with a clumsy posture. She did not gain weight like a normal child and physical activities were restricted. Like her father, she showed progressive deviation of the spine, and recently complained with pain in the dorsal region, especially during physical activity. On examination, the patient was a tall and thin child, with marked kyphoscoliosis, long face, high palate, atrophy muscle and global hypoactive muscle stretch reflexes. There were no motor deficits in the face or external ophthalmoparesis. CK values were between 65 to 73 U / L (normal below 145 U / L). Glucose, IgA, IgG and IgM, transaminases, and electrolytes were normal, but with a TSH value of 7.23 IU / ml (normal 4.5) and FT4 of 1.68 ng / dl (normal range). Sensory nerve conduction velocity (median, ulnar, radial and sural) and motor nerves (median, ulnar, peroneal and tibial) were within normal limits. EMG of deltoid, biceps, rectus femoris, tibialis anterior and gastrocnemius showed myopathic changes. The patient was doing physical therapy in 50-minute sessions per week in pediatric neurology ambulatory UNICAMP. At 15 years old she had an episode of severe pneumonia which complicated by fatal septicemia.

CASE III - 3. Male, 10 years old, third son of the case II -2. He was born at 38 weeks gestational age with cesarean delivery, and had a history of reduced fetal movements. The child presented a congenital hypotonia and delayed motor development to roll, sit and crawl, acquiring the standing posture around 11 months age, and unsteady gait around 2 years old. According to his parents, he used to present recurrent episodes of urinary tract infection, pneumonia and ear infections during childhood. In the first consultation, there was a decreased in motor performance, diffused muscle hypotrophy, scoliosis, high palate and hypoactivity of muscle stretch reflexes. There was no facial weakness or external ophthalmoparesis. Sensibility and cognition were preserved. Values of CK, glucose, IgA, IgG and IgM, transaminases, electrolytes, vitamin B12 and TSH were normal. The EMG test was not performed at the request of his father.

CASE III - 5. Male, 9 months of age, born after 38 weeks of pregnancy, cesarean delivery, and with a history of reduced fetal movements. He had a slight delay in motor development, and at clinical examination an evident hypotonia, with preserved muscle stretch reflexes. Laboratory findings revealed mild microcytic hypochromic anemia, CK of 89 U / L (normal 170 U / L) and TSH and T4 L-us normal. Clinical monitoring was suggested by over time. He was re-evaluated at 2 years and 6 months of age. At this time he was walking without support, with mild myopathic patterns. The muscle stretch reflexes were hypoactive, hypotonia clearly present and dextro-convex scoliosis and lumbar cord. For treatment physiotherapy was indicated. We did not evaluate him using the scale P-MFM due to this method is recommended for patients over 6 years.

Figure 3 a-c illustrate the data from the mMRI for cases II-2, III-1 and III3.

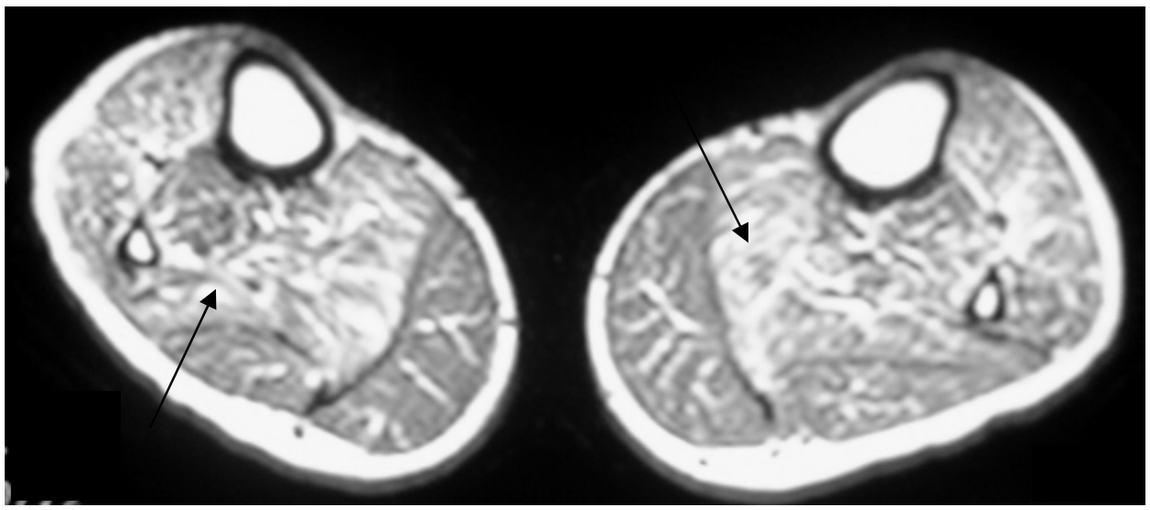

Fig. 3.a - Case II-2. mMRI, T1-weighted images and axial sections showing fatty infiltration of muscle compartments of the anterior, posterior deep and superficial leg, with greater involvement of the anterior and deep posterior compartment.

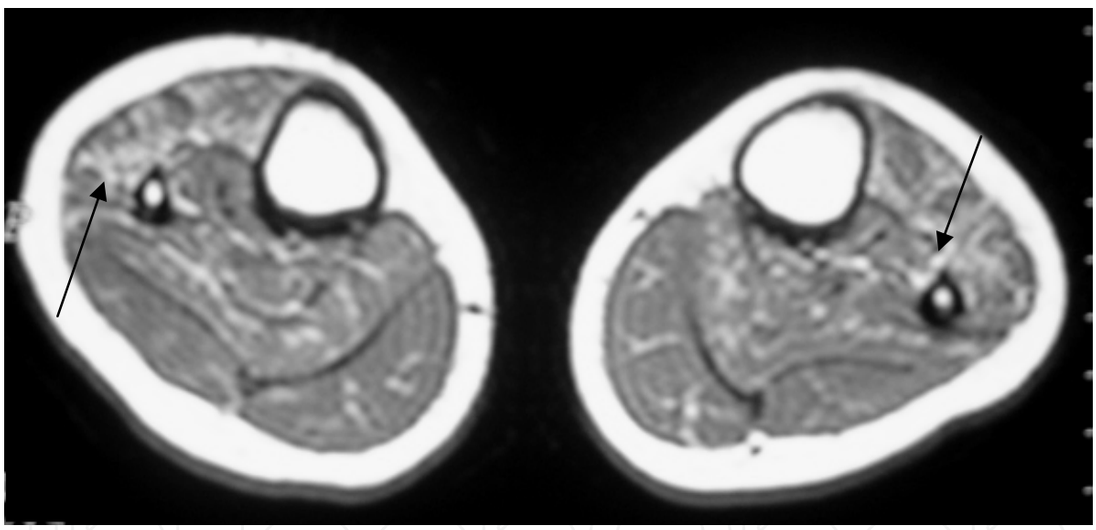

Fig. 3.b - Case III-1. mMRI, T1-weighted images and axial sections showing fatty infiltration of muscle compartments of the anterior, posterior deep and superficial leg with less alteration of the superficial posterior compartment. Note the symmetry condition.

Figure 4 represents the scores of each dimension and the total score of P-MFM in cases II - 2, III - 1 and III - 3, at different ages.

Different types of muscle fibres are present in the muscles of normal adults in a typical mosaic pattern, with ratio of approximately 1 / 3 of fibre type 1 (Figures 2a and 2b). The differentiation of the fibres occurs between the 22° week of gestation and the first year of life. At birth, the child had about 40% of type 1 fibre. The percentage of these fibres increased up to 60% in the first year of life and remained unchanged until adulthood. The sizes of type 1 and 2 fibres are almost equal in childhood, with little variability in relation to adults (75).

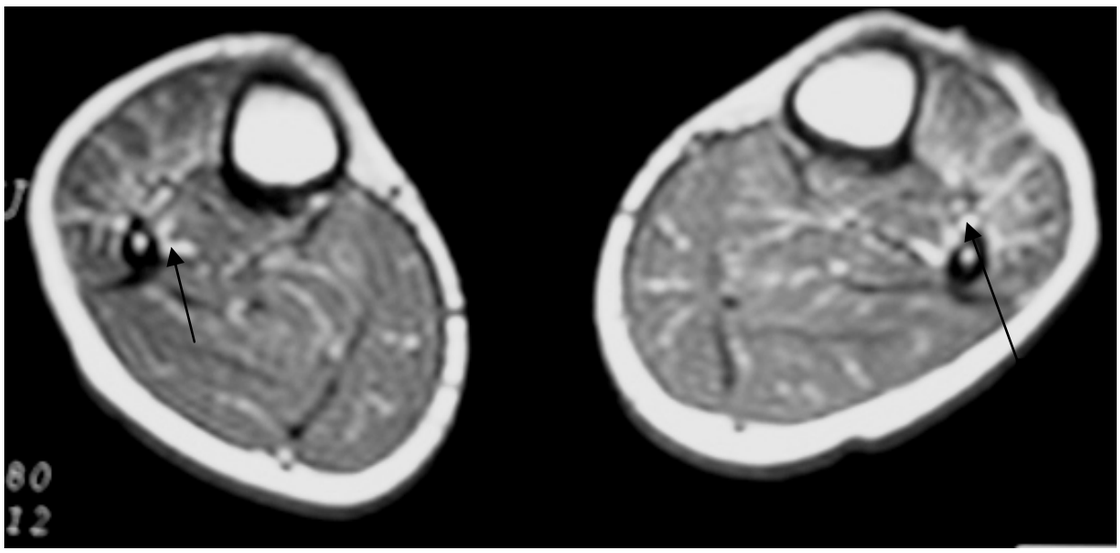

Fig. 3.c - Case III-3. mMRI, T1-weighted images and axial sections showing fatty infiltration of muscle compartments of the anterior, posterior deep and superficial leg with less alteration of the superficial posterior compartment. Note the relative preservation of muscle compared with the previous cases.

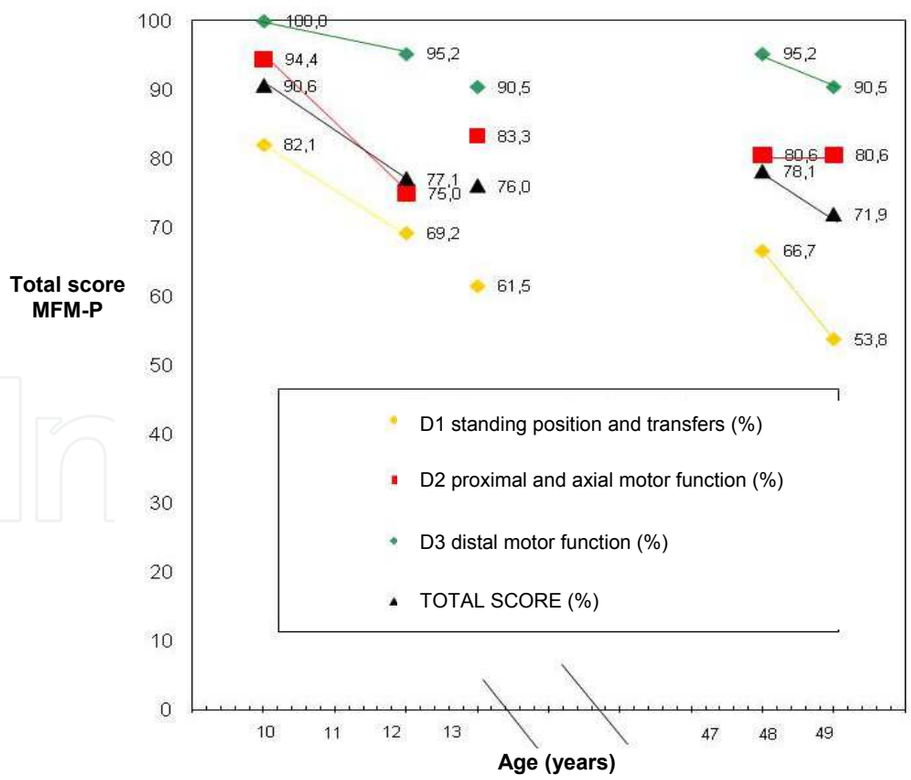

Fig. 4. Distribution of scores for each dimension and total score of the P-MFM from each patient. The evaluation at 10 and 12 years refers to case III-3, the evaluation at 13 years refers to the case III-1, and evaluation at 48 and 49 years to case II-2.

The mutation in the genes ACTA1 (22), SEPN1 (48) or TPM3 (23) express the morphological and histochemical alteration in CFTD (49). Since the first description of this disease, approximately 67 cases have been described (16), but just a few cases originally in Brazil (50).

Clinical characteristics of patients in the study were similar to those described in previously works, such as congenital hypotonia, delayed motor skills, kyphoscoliosis and high palate. Also data for additional tests as EMG myopathic pattern and CK levels are normal or slightly altered according to the literature (14,17,51,52,53,54).

The natural course of CFTD is in most cases characterized by slow progression of weakness, affecting mainly the lower limbs beginning at the proximal muscles and progressing to distal. However, in some patients weakness is widespread (16,17). The deterioration of muscle strength was observed in our patients, with difficulties to standing, walking and running. During assessment of motor function, all patients showed greater difficulties in activities related to standing position and transfers (Dimension 1) due to loss of muscle strength.

All the seven cases described by Sobrado et al. (17) had difficulty in activities like those described in our study (raising themselves from a chair or from the floor, walking or running on heels), in addition to muscle weakness in lower limbs (grade 3 to 4). These data agree with descriptions of Linssen et al. (55) in which cases presenting degree of muscle strength equal to 4, have functional limitations.

Some patients with CFTD have severe respiratory complications, as with the patient in the case III - 1. This condition could happen due to hypotrophy of type 1 fibres found in the respiratory muscles, including the diaphragm (14,16,20,22,48,49) or as a result of severe kyphoscoliosis, which progressively diminished lung capacity. Thus, it is important that patients be monitored for maintenance of a postural alignment and breathing function.

The mMRI is proposed as a useful method to study congenital and metabolic myopathy (56), although there are few publications using this technique in CFTD (17). In the current study, it was possible to qualitatively observe intense changes in distal segments of the lower limbs in all images of affected patients. These changes correspond to an increased leg muscle weakness with difficulties in performing activities according to the P-MFM scale.

The evaluation of muscle function by P-MFM associated with the examination of mMRI led to a full characterization and motor phenotype evaluation in these patients. The first evaluation using the P-MFM in family's members showed a co-occurrence of more intense abnormalities in the mMRI with the worst scores in standing position and transfer. There was also a correlation between the age and severity of the mMRI and P-MFM score.

4. Correlation between muscle strength and P-MFM in myotonic dystrophy

The study included a total of 21 patients, 10 males and 11 females, from 20 to 60 years old, with an average of 38.14 years, and with clinical-laboratory diagnosis of MD-1, outcoming the Neuromuscular Diseases Clinic Clinical Hospital of UNICAMP.

Patients were evaluated by MFM-P (29) and submitted to examination of muscle strength by MRC scale (1976) that includes 14 muscle groups of upper limbs (UL), 14 groups in the

cervical muscles and lower limbs, trunk flexors and extensors. Muscles were grouped according to the segment's function.

Statistical analysis - The correlation of each of the three dimensions and the total score P-MFM, with the degrees obtained by the MRC scale in the muscle groups studied was made using Pearson's correlation coefficient.

Patients showed a variation in the degree of muscle strength from 2 to 4. In the distal muscle groups, such as flexors, extensors of the fingers and wrist extensors, the degrees of force were grade 2-4. And the proximal muscles strength varied from 3 to 4.

In the lower limbs, muscle strength ranged from grade 2 to 5. In distal muscles the strength varied from 2 to 4 and the proximal muscles strength ranged from grade 3 to 5. Also the evaluation of axial muscles showed strength ranged varying from grade 2 to 4.

The deficits found in patients were symmetrical in both the axial region and in the upper and lower segments.

To analyze the distribution of the total score and each dimension of the P-MFM scale values were arbitrarily classified as: mild (100 to 70%, independent patient), moderate (69.9 to 50%, partially dependent) and severe (<50%, dependent).

A higher number of patients presented lower scores (<50%) in activities related to P-MFM Dimension 1 (standing position and transfers) (Table 2).

Score	Dimension (score)			
	D1	D2	D3	Total score
Mild (100– 70%)	4 (89,74 – 76,92)	18 (100 – 80,56)	20 (100 – 71,43)	12 (95,83 – 72,92)
Moderate (69,9– 50%)	7 (69,23 – 53,85)	3 (63,89 – 58,33)	01 (57,14)	09 (66,67 – 54,17)
Severe (< 50%)	10 (48,72 – 25-64)	----	---	---

Table 2. Number of patients according to scores obtained in each dimension and total score of P-MFM

Analyzing the correlations between the degrees of upper limb strength in each dimension and total score P-MFM, we observed a significantly positive correlation between the proximal muscles and Dimension1 and Dimension 2, and correlation between the distal muscles and Dimension 3.

Similarly for lower limbs, we observed a significantly positive correlation between the plantar flexors and extensors of the hips strength with the scores of D1; and also between the finger extensors and dorsiflexors of the ankles strength with the scores of D2. The strength values of the finger extensors, dorsiflexors, ankle inverters and eversion were significantly positive correlated with the score of D3; and the plantar flexors, dorsiflexors, eversion ankles, and hip extensors strength were significantly positive correlated with the total score of P-MFM.

Analyzing the correlations between the neck and trunk strength, with each dimension and the total score of P-MFM, we observed a significantly correlation between the neck and abdominal flexor with D1; between neck flexors with the D3; and between neck flexors and abdominal flexor with total score.

The assessment of functional capacity and degree of muscle strength in patients with neuromuscular disease are essential aspects for their diagnosis and follow-up. Assistance in clinical decisions, treatment, prevention of any complications (like respiratory failure or retractions), indication of the type and intensity of exercise are important aspects to be considered (57,58).

The MD-1 shows a pattern of muscle weakness primarily affecting the facial muscles, neck flexors, and dorsiflexors of fingers. The proximal muscles may not show deficits or mild clinical signs (2,59,60). Lindeman et al. (59,61) showed that in MD-1 there is a great variability in degrees of strength in the affected muscle groups (62.63). The symmetric and progressive muscular weakness involves proximal muscles (64). The deficit predominantly distal and axial deficits, as well as the later weakness proximal segments was also demonstrated in studies by Whittaker et al. (60), Lindeman and Drukker (65) and Lindeman et al. (66). Similar topography of motor impairment was observed in this study, in patients with MD-1.

The strength graduated in 4 on the MRC scale, defined as the ability of active muscle contraction against moderate resistance, was correlated with a negative impact on the ability to perform functions like running, climbing stairs and walking (55,59,67,68,69,70,71,72).

In this study, we found a great variability in the degree of muscle strength with greater involvement in distal segments of the lower limbs, and motor skills assessed by the P-MFM and a greater activity limitation in the standing position and transfers. The positive correlations obtained between muscle groups and the dimensions of P-MFM were restricted, noting that the most significant one occurred between the groups with lower degree of strength of the distal segment (hand and foot) and D3.

According to Whittaker et al. (60) the weakness of flexors and extensors of fingers and wrist is the major cause of disability in DM-1. This early involvement of distal muscles allowed the correlation of these groups, especially with the D3. The proximal and axial body segments act as posture stabilizer factors to provide a stable basis for the distal movement (73). Considering that during the measurement of the scale items the subjects had trunk and limbs supported, they did not need an effective action from the muscles of the trunk and proximal limbs. So, it was possible to correlate specifically the strength of distal muscles with their respective activities.

Poor limitations in proximal muscle groups (extensors of the hip and shoulder) were observed in few activities (Dimensions 1 and 2) limiting other activities (60,65,66) in late stage of this disease.

The proximal muscular deficit can influence the performance function (64, 74, 75, 76.77, 78). According to Galassi et al. (79) and Linssen et al. (55) even when the weakness is predominantly in the distal limb segments, there is a bilateral proximal atrophy area in the computer tomography scans from patients with muscular dystrophy, also demonstrating the involvement of bilateral and proximal muscles, even when there is a clear manifestation of symptoms.

Lindeman et al. (59,61,76)and Aldehag et al. (80) correlated a specific muscle with a particular activity (like the quadriceps strength and standing up from a chair) in patients with DM-1, and found a positive correlation between strength and function. However Dawes et al. (81) found no correlation between muscle strength and function, when these activities were more complex.

Besides strength, other variables may be influencing the individual's functional ability, such as age (82), gender and weight (83). In our cases these variables were not considered because they were not included in the objectives.

So, it is important to analyze a large number of myotonic dystrophy patients to observe any correlation between strength and motor function.

5. Final considerations

The use of MFM proved to be an interesting tool in the study of the congenital myopathies as CFTD; although we were limited by time and sample size. The correlation of severe myopathy and mMRI data for smaller scale scores P-MFM, enables the use of a non-invasive diagnostic tool to study the course of myopathy diseases. They also indicate that both methods could be used in to analyse the results of pharmacological interventions and rehabilitation in these diseases.

The P-MFM has a high reliability and validity to be used as a tool for clinical diagnosis and monitoring of neuromuscular diseases, allowing the inclusion of Brazilian patients in international clinical trials. Thus, we can mention that:

1. The positive correlations between the BI, Vignos scale and P-MFM allowed the validation of P-MFM in Brazil.
2. The application of P-MFM could demonstrate the progressive dysfunction in two of the three members of a family with CFTD. The muscle magnetic resonance showed that the more severe the motor function, the worse were the changes in the image worst severe functional motor, the worst change images.
3. The application of P-MFM in MD-1 showed the predominance of distal myopathy, expressed by the lower scores of muscle strength correlating positively with the scores mainly in D3.

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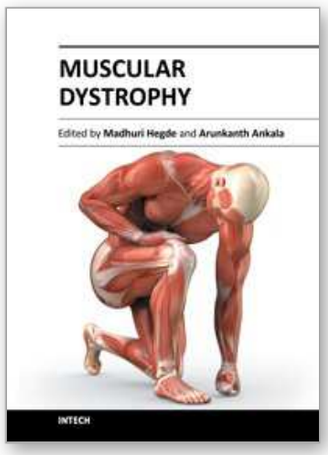

Muscular Dystrophy
Edited by Dr. Madhuri Hegde

ISBN 978-953-51-0603-6
Hard cover, 544 pages
Publisher InTech
Published online 09, May, 2012
Published in print edition May, 2012

With more than 30 different types and subtypes known and many more yet to be classified and characterized, muscular dystrophy is a highly heterogeneous group of inherited neuromuscular disorders. This book provides a comprehensive overview of the various types of muscular dystrophies, genes associated with each subtype, disease diagnosis, management as well as available treatment options. Though each different type and subtype of muscular dystrophy is associated with a different causative gene, the majority of them have overlapping clinical presentations, making molecular diagnosis inevitable for both disease diagnosis as well as patient management. This book discusses the currently available diagnostic approaches that have revolutionized clinical research. Pathophysiology of the different muscular dystrophies, multifaceted functions of the involved genes as well as efforts towards diagnosis and effective patient management, are also discussed. Adding value to the book are the included reports on ongoing studies that show a promise for future therapeutic strategies.

How to reference
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Cristina Iwabe, Anamarli Nucci, Beatriz Helena Miranda Pfeilsticker and Luis Alberto Magna (2012). Motor Function Measure Scale (MFM): New Instrument for Follow-Up Brazilian Patients with Neuromuscular Disease, Muscular Dystrophy, Dr. Madhuri Hegde (Ed.), ISBN: 978-953-51-0603-6, InTech, Available from: http://www.intechopen.com/books/muscular-dystrophy/motor-function-measure-scale-mfm-new-instrument-for-evaluation-diagnosis-and-monitoring-in-brazilian

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