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RELATO DE CASO

Limb-girdle muscular dystrophy and physical therapy: update and case report

Distrofia muscular de cinturas e fisioterapia: atualização e relato de caso

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Abstract

The term limb girdle muscular dystrophy (LGMD) has been introduced to delineate a distinct form of muscular dystrophy with predominantly proximal upper and lower extremity weakness. PAS, 71 years old, male, retired, diabetic and hypertensive. He reports that the clinical picture started in mid-2010 with complaints related to atrophy of the shoulder girdle and thigh region. "When looking at myself in the mirror I started to notice that I was losing muscle in my shoulders". The beginning of the clinical picture dragged on, still with muscle shape close to normal to perform daily activities. He points out that the cervical region bothers him a lot, mainly due to the inclined position in which

he is. In this clinical case, we present neurological and functional evaluations, as well as complementary exams that help in the detection of LGMD. New studies, including a larger number of patients with myopathy of the waist and limbs, are needed. We believe that the best form of rehabilitation for this group of myopathies is not to look for overuse damage and, undoubtedly, work in search of function, not primarily strength.
Keywords: myopathy, rehabilitation; limb girdle muscular dystrophy

Resumo

O termo distrofia muscular de cinturas (DMC) foi introduzido para delinear uma forma distinta de distrofia muscular com fraqueza predominantemente proximal nas extremidades superiores e inferiores. PAS, 71 anos, sexo masculino, aposentado, diabéticos e hipertenso. Ele relata que o quadro clínico iniciou em meados de 2010 com queixas relacionadas à atrofia da cintura escapular e região da coxa. “Ao me olhar no espelho comecei a perceber que estava perdendo músculos nos ombros”. O início do quadro clínico se arrastou, ainda com formato muscular próximo ao normal para realização das atividades diárias. Ele ressalta que a região cervical o incomoda bastante, principalmente pela posição inclinada em que se encontra. Neste caso clínico apresentamos avaliações neurológicas e funcionais, bem como exames complementares que auxiliam na detecção de DMC. Novos estudos, abrangendo um número maior de pacientes com miopatia de cintura e membros, são necessários. Acreditamos que a melhor forma de reabilitação para esse grupo de miopatias é não buscar danos por uso excessivo e, sem dúvida, trabalhar em busca de função e não principalmente de força.
Palavras-chave: miopatia, reabilitação; distrofia muscular de cinturas.

Introduction

The term limb girdle muscular dystrophy (LGMD) has been introduced to delineate a distinct form of muscular dystrophy with predominantly proximal upper and lower extremity weakness. Families with evidence of both autosomal recessive and autosomal dominant modes of inheritance have been described. The recognition of other disorders presenting with weakness in a limb girdle distribution, such as the spinal muscular atrophies, dystrophinopathies, inflammatory and metabolic myopathies, casted doubt on the existence of LGMD as a separate entity [1-2].

We present a case of muscular dystrophy of the waist and limbs and, based on the impairments and functional disabilities resulting from muscle weakness, we point out some proposals for rehabilitation.

Case Report

PAS, 71 years old, male,retired. Diabetic and Hypertensive. He reports that the clinical picture started in mid-2010 with complaints related to atrophy of the shoulder girdle and thigh region. "When looking at myself in the mirror I started to notice that I was losing muscle in my shoulders". The beginning of the clinical picture dragged on, still with muscle shape close to normal to perform daily activities. He points out that the cervical region bothers him a lot, mainly due to the inclined position in which he is. Neurological examination: On inspection: muscle atrophy is noted in the shoulder and pelvic girdles. Muscle Strength: In the main muscle groups evaluated in the upper and lower limbs, the predilection for weakness occurred in the proximal third of the brachial and bilateral crural muscles (figure 1). The most intermediate and distal segments had muscle strength grade 45 (MRC). Normal Deep Reflexes. Superficial and deep sensitivity: normal. Cranial Nerve Nuclei: Normal. Physical function: Height 1.74 (m), weight 77.2 kg, body mass index (BMI) 25.5 kg/m^2, muscle mass 55.1 kg, % fat 23.9, FFM/m^2 18.2. Despite presenting functional independence when evaluated by the Barthel index (scoring 95/100), when performing the Short Physical Performance Battery hort Physical Performance Battery (SPBB) it presented three points (3/12) characterizing a “low capacity”. After evaluating the peripheral muscle strength by means of digital dynamometry, it was found that the handgrip strength was preserved, for the dominant limb reaching 104.2% in relation to the predicted value (figure 2A), and 119.4% of the predicted value in the non-dominant limb (figure 2B). After the assessment of inspiratory muscle strength (MIP) was performed using digital manovacuometry, it was observed that 91.8% were obtained in relation to the predicted values (figure 2C). There were no changes in static and/or dynamic balance compatible with the risk of falling, assessed using the Timed Up and Go (TUG), the patient completed the course in 9 seconds. Electroneuromyography: myopathic pattern with axial predominance (Figure 1). MRI of the brachial plexus: denervation with liposubstitution in the proximal brachial and crural third. Muscle biopsy: Immunohistochemical changes for dystrophy and sarcoglycans, which were positive, suggesting myopathy of proximal predominance.It is noteworthy that the patient has lateral neck tilt due to several inadequate synergies of movement that he performed for years. Initially, a picture of dystonia was thought, but the electroneuromyography ruled it out (figure 3). As a result of the thoracolumbar anchorage, we noticed filiform hydromyelia throughout the entire length of the medulla (figure 4). The diagnosis of recessive bulbospinal amyotrophy was excluded.

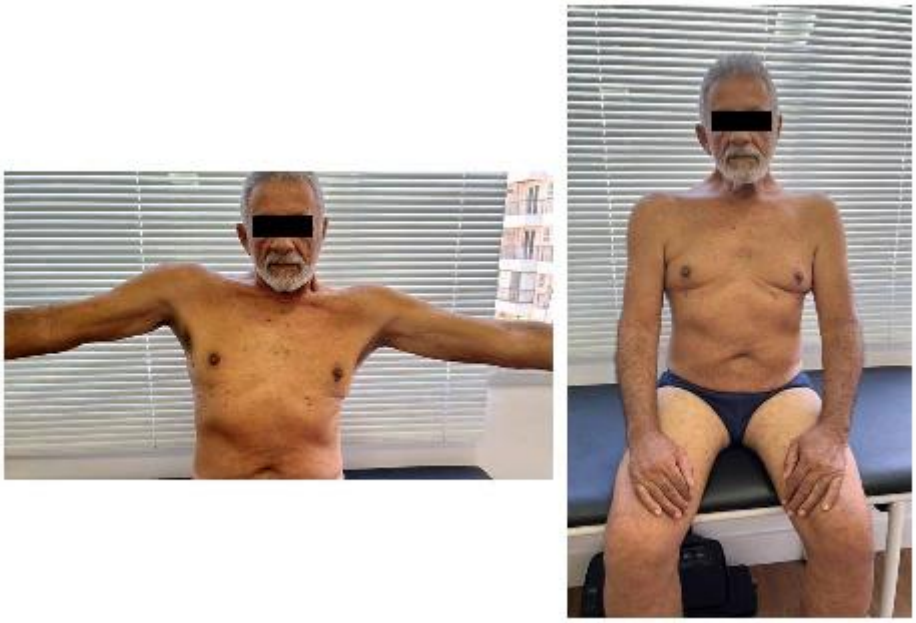


Figure 1 - *Note brachial proximal muscle atrophy and difficulties in raising the arms. Note proximal brachial and crural muscle atrophy*

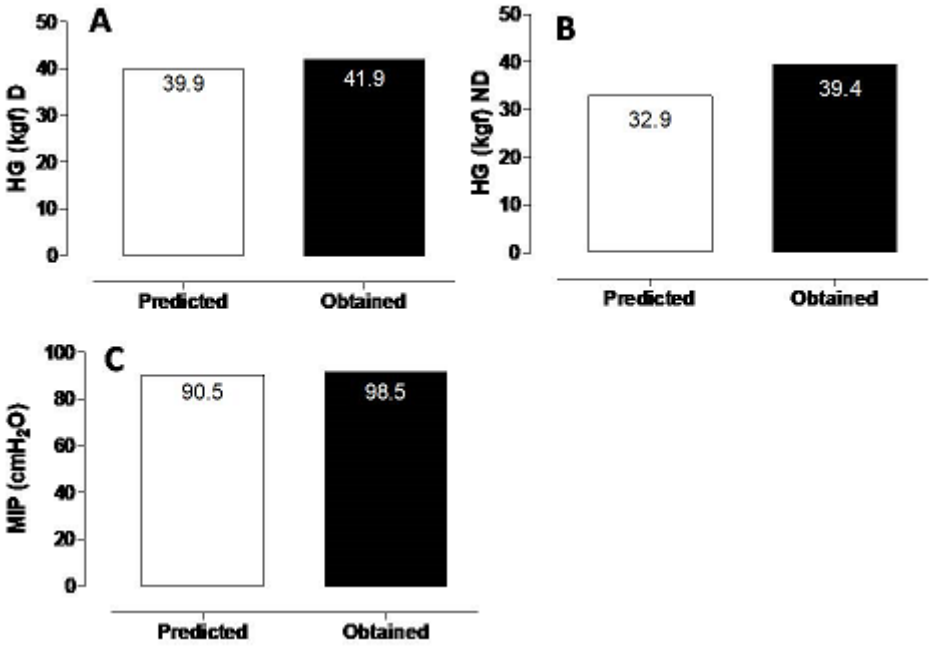
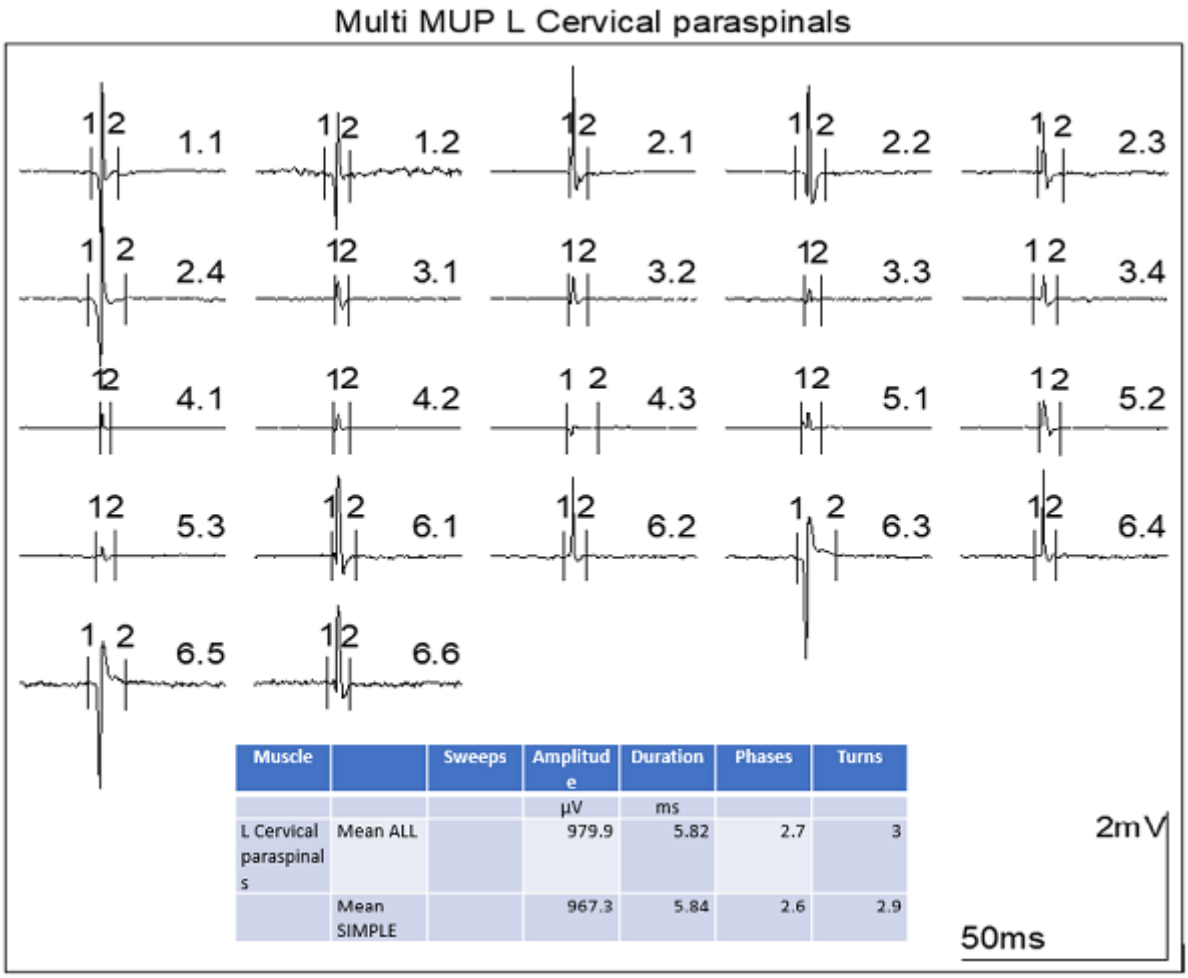


Figure 2 - *Handgrip strength (HG) for the dominant limb (A) non-dominant limb (B), maximal inspiratory pressure – MIP (C) predicted value in the non-dominant limb (figure 3B). After the assessment of inspiratory muscle strength (MIP) was predicted vs. obtained*



Muscle		Sweeps	Amplitude	Duration	Phases	Turns
			µV	ms		
L Cervical paraspinals	Mean ALL		979.9	5.82	2.7	3
	Mean SIMPLE		967.3	5.84	2.6	2.9

Figure 3 - *Quantitative Multi Motor Unit Potential Analysis of the cervicalparavertebral musculature showing motor units of reduced duration and amplitude. Indicative of myopathic disease*

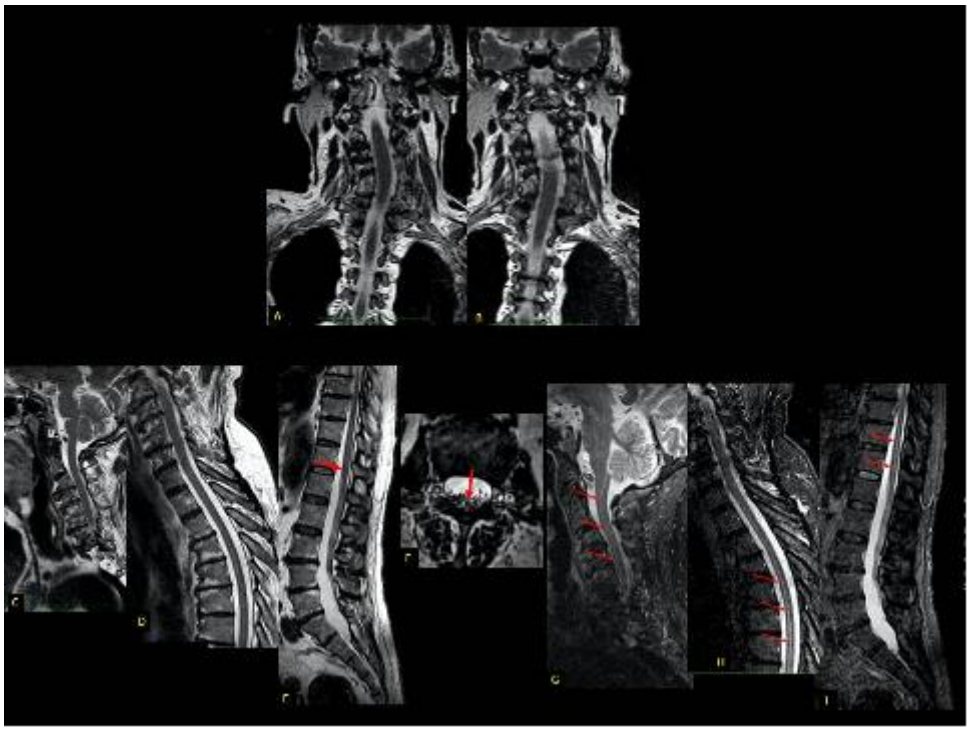


Figure 4 - *Neck pain 1 year ago. Paresthesia in the third and fourth fingers of the right hand. Coronal T2 (A and B) shows left cervical scoliosis. There are extensive degenerative changes in the cervical (C), dorsal (D) and lumbosacral (E) spine, with osteophytes in the vertebrae, degenerative changes in the vertebral plateaus, dehydration and reduced disc height. Note the stretched and retracted medullary cone posteriorly inside the dural sac (arrow on E) due to the presence of a thick terminal filum (arrow on F). There are extensive degenerative changes in the cervical (C), dorsal (D) and lumbosacral (E) spine, with osteophytes in the vertebrae, degenerative changes in the vertebral plateaus, dehydration and reduced disc height: As a consequence of anchoring, we noticed filiform hydromyelia throughout the entire length of the medulla (arrows in G-I-sagittal STIR)*

Discussion

The spectrum of conditions encapsulated by this subset ranges from severe and fatal congenital muscular dystrophies with onset in infancy to mild forms of limb and girdle weakness with onset in adulthood and minimal respiratory compromise. The list and classification of muscular dystrophies are undergoing near-constant revision, based largely on new insights from genetics and molecular medicine [3].These advances are reflected in the development of new therapeutic approaches, some of which have already led to clinical trials in the dystrophinopathies and limb-girdle dystrophies4.We present a case of muscular dystrophy of the waist and limbs and, based on the physical and neurological examination, we suggest possibilities for rehabilitation treatment [4].

Cup *et al.* [5], aimed through a systematic review to summarize and measure evidence on rehabilitation for patients with neuromuscular diseases, including myopathies. The search sources were: Cochrane Central Register of Controlled Trials

and Cochrane Database of Systematic Reviews, Medline, CINAHL, EMBASE (Rehabilitation and Physical Medicine). Study Selection comprised: Randomized Clinical Trials (RCTs) and Controlled Clinical Trials (CCTs). The patients in the study had to suffer from the NMDs listed below: diseases of the motor neuron, motor nerve roots or peripheral nerves, neuromuscular transmission or muscular diseases (myopathies). For data extraction, two reviewers decided on the inclusion or exclusion of articles to evaluating the methodology employed. A level of evidence was assigned to each subgroup of NMD and each type of intervention. Initially (data synthesis) 58 studies were included: 12 RCTs, 5 CCTs and 41 other research projects. After the first evaluation pairing, 19 other projects were excluded. There is level II evidence ("likely to be effective") for strengthening exercises in combination with aerobic exercises for patients with muscle disorders. Level III evidence ("indications of efficacy") was found for aerobic exercise in patients with muscle disorders and for the combination of muscle strengthening and aerobic exercise in a heterogeneous group of muscle disorders. Finally, there is level III evidence for breathing exercises for patients with myasthenia gravis and for patients with myotonic muscular dystrophy. Adverse effects of exercise therapy were negligible. The available evidence is limited, but relevant for physicians and professionals who have little access to therapeutic practices. Multicenter studies, using the International Classification of Functioning (ICF), will probably improve the comparability of results5.

Recent studies are in line with earlier ones further supporting safety and efficacy of exercise in patients with polymyositis or dermatomyositis. There is an urgent need for larger randomized controlled trials also including patients with inclusion body myositis to further increase knowledge of disease mechanisms causing disability, exercise effects, and what exercise program is most efficient in patients with different entities of idiopathic inflammatory myopathies [6].

Orsini M *et al.* [7], described the case of a patient with mitochondrial myopathy. In addition, they sought numerous studies involving short-term, moderate-intensity aerobic training in this myopathy group. The results were satisfactory in the improvement of respiratory capacity, resistance to fatigue and tolerance in the execution of basic and instrumental activities of daily living.in patients with mitochondrial myopathy [7].

Other authors point out that training programs rehabilitative treatment composed of high-intensity exercises intensity, although well tolerated by patients with neuromuscular diseases with weakness (medium to moderate), can cause deleterious effects on the musculature.skeletal striated latature. high resistance exercises do not seem to offer greater advantages when compared to light to moderate exercises.

Exercise-based training protocols for patients with myopathies of various etiologies should measure the intensity, duration, frequency and type of activities proposed [8-10].

The basic facilitation procedures provide tools for the therapist to help the patient gain efficient motor function and increased motor control. The basic procedures can use to treat patients with any diagnosis and or condition, although a patient condition may rule out the use of some of them. Evidence based physiotherapy treatment is based upon external support of the therapeutic care/intervention combined with the expertise and experience of the therapist, adapted to the needs and objectives of the patient. The results presented reinforce that the techniques of PNF, when employed after a correct kinetic-functional diagnosis, promote satisfactory results in the management of the muscular weakness and training of the functional abilities [11-13].

Despite the reports described in the literature regarding the possible involvement of the respiratory muscles in patients with limb-girdle muscular dystrophy [14-16], the patient described has preserved ventilatory musculature. It is recommended that every patient with muscular dystrophy undergo an adequate respiratory assessment so that any changes can be identified as soon as possible. The reduction in respiratory function and respiratory muscle strength and resistance is directly related to the individual's functional condition.

Another issue that should always be present in the evaluation of patients with Limb-girdle is the verification of the presence of sarcopenia [17], a condition that may be superimposed on the primary clinical condition and lead to functional decline, morbidity and mortality [18], but the patient presented here does not fulfill sarcopenia criteria (muscle strength – hand grip, muscle quantity or quality – bioimpedance, physical performance – TUG) despite having a “low capacity” SPPB.

A fact that deserves to be highlighted is that although the patient presented a reduction in peripheral muscle strength assessed by the MRC, this was not demonstrated in the assessment using the hand grip. For clinical conditions such as the Limb-girdle, using at least two ways to measure strength can provide important information regarding the staging of both the clinical and functional condition [19].

New studies, comprising a larger number of patients with myopathy of the waist and limbs, are needed. We believe that the best form of rehabilitation for this group of myopathies is not to look for overuse damage and, undoubtedly, work in search of function, not primarily strength.

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