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BENIGN MONOMELIC AMYOTROPHY WITH PROXIMAL UPPER LIMB INVOLVEMENT

Case report

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ABSTRACT - Monomelic amyotrophy (MA) is a rare condition in which neurogenic amyotrophy is restricted to an upper or lower limb. Usually sporadic, it usually has an insidious onset with a mean evolution of 2 to 4 years following first clinical manifestations, which is, in turned, followed by stabilization. We report a case of 20-years-old man who presented slowly progressive amyotrophy associated with proximal paresis of the right upper limb, which was followed by clinical stabilization 4 years later. Eletroneuromyography revealed denervation along with myofasciculations in various muscle groups of the right upper limb. We call atention to this rare location of MA, as well as describe some theories concerning its pathophysiology .

KEY WORDS: lower motor neuron, monomelic amyotrophy, spinal amyotrophy, eletroneuromyograpy.

Amiotrofia monomélica benigna com comprometimento proximal do membro superior: relato de caso

RESUMO - A amiotrofia monomélica é condição rara em que a amiotrofia neurogênica é restrita somente a um membro superior ou inferior. Usualmente esporádica, possui um início insidioso e evolução lenta de 2-4 anos após as primeiras manifestações, seguida por estabilização. Relatamos o caso de um homem que aos 20 anos apresentou lentamente amiotrofia e paresia proximal no membro superior direito, estabilizando-se em quatro anos. A eletroneuromiografia identificou a presença de desnervação e fasciculações em diversos músculos proximais do membro superior direito. Chamamos a atenção para esta rara localização desta entidade, assim como descrevemos algumas das hipóteses da fisiopatologia.

PALAVRAS-CHAVE: neurônio motor inferior, amiotrofia monomélica, amiotrofia espinhal, eletroneuromiografia.

Monomelic amyotrophy (MA) is a rare disorder in which neurogenic amyotrophy is restricted to a superior or inferior limb, generally sporadic, and characterized by gradual, often insidious onset[1-2]. It differs from other lower motor neuron disorders in that it involves only one limb. Upper motor neuron signs are usually absent, and progression slowly evolves over months or even years, followed by clinical stabilization of symptoms[3]. MA affects younger individuals (second to third decade of life) and predominantly males, also characterized by a typical geographic distribution[4]. The majority of cases are encountered in Asia, mainly in Japan and India. There are few reports of MA in western countries[5-8].

Although the cause still remains unknown, neuropathologic studies reveal a focal lesion in the anterior horn motor cells of the spinal cord. Its relation to other lower motor neuron diseases, like amyotrophic lateral sclerosis (ALS) for instance is uncertain[1,5]. Distal muscles groups are usually affected. MA is also known as Hirayama´s disease[9].

We report a case of a male patient, who presented with signs and symptoms of this disorder, though in the proximal instead of distal muscle groups of his upper limb, a very rarely described location according to the literature.

CASE

A 23-years-old man, white, medical student, was consulted at the Neuromuscular Disorders Inpatient Division of

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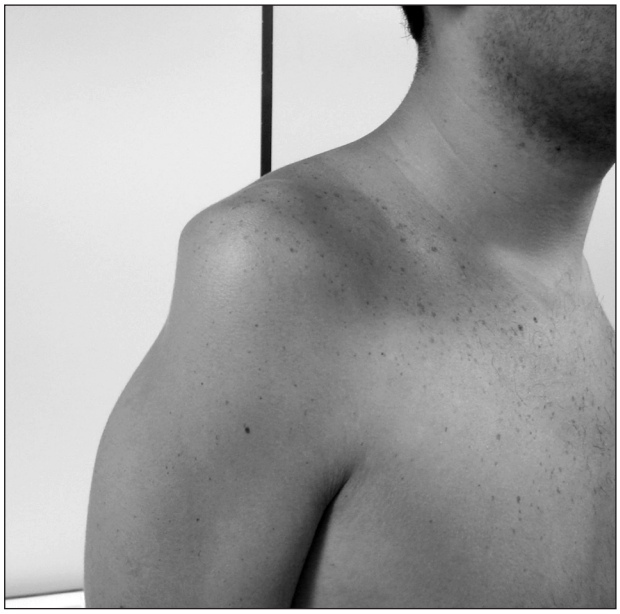

Fig 1. Marked atrophy of right deltoid muscle.

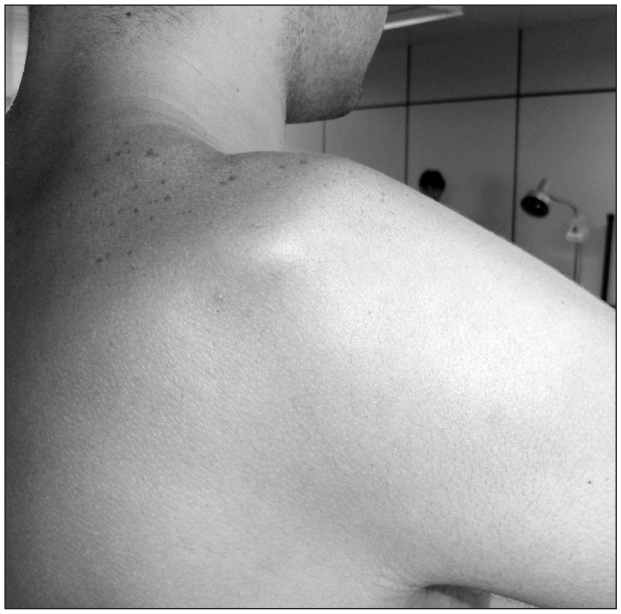

Fig 2. Difficulty in abduction movement of upper right limb.

Table. Paresis in proximal muscles of the upper right limb (Medical Research Council).

Muscles	Right upper limb	Left upper limb
Teres minor	3	5
Teres major	3	5
Subescapular	3	5
Infraspinatus	3	5
Supraspinatus	3	5
Deltoid	3	5
Biceps	3	5
Extensor carpi radialis longus and brevis	5	5
Triceps	5	5
Flexor digitorium superficialis and profundus	5	5
Interosseous	5	5

Antonio Pedro Teaching Hospital in May 2004. During this year, the patient reported that two years prior he began weakness in certain parts of his right upper limb, especially during physical activity. His clinical exam revealed marked atrophy and weakness in the upper right limb (scapular region and proximal third) associated a difficulty in abduction movement of upper right limb, (Figs 1 and 2, respectively). Table shows variable degrees of paresis in proximal muscles of the upper right limb of this patient. The bicipital and estilorradial (pronator) reflexes were abolished on that side and sensibility was normal. Cranial nerve evaluation was also normal. Electromyography (EMG) showed reduced motor action potentials in the right deltoid muscles. The infraspinatus, biceps and deltoid muscles showed signs of denervation, with the infraspinatus group also presenting fasciculations. There was mild fibrillation in the first interosseous muscle on the left and an increase in the amplitude of the motor action potential of the right lateral vastum muscle. Both motor and sensitive nerve conduction studies as well as a cervical spinal cord magnetic resonance image (MRI) were normal. The motor symptoms progressed over the next four years, having been clinically stable for the last year.

DISCUSSION

Monomelic amyotrophy was originally described in Japan in 1959 by Hirayama et al., who had observed young male patients with insidious onset, slowly progressive one-sided hand and forearm atrophy. The first terms used to describe such a condition were "juvenile muscular atrophy of unilateral the upper extremity"[9]. The first report of a unilateral nonprogressive lower limb atrophy dates from 1981[10].

The term "monomelic amyotrophy" was first coined in 1984 by Gourie-Devi, who had observed both upper and lower limb atrophy, stressing the fact that the atrophy was always restricted to a single limb[11]. However, such a term has since been contested by some authors due to reports of bilateral involvement[5].

The benign nature of MA, which helps distinguish it from other lower motor neuron disorders, explained by the fact that involvement of other limbs is absent in the majority of patients and in its restriction to only the lower motor neuron[12,17]. Its occurrence is essentially sporadic, there being no relation to any genetic predisposition such as that seen in, for example, the spinal muscular atrophy[13]. Reports of familial cases of MA are extremely rare, though there is the possibility of an autossomic recessive or dominant trait[4,14-15]. Although MA is a rarely seen in the western world, it comprises approximately 12,8% of lower motor neuron diseases in India[16].

The main clinical findings seen in patients with MA include weakness and atrophy restricted to a single upper or lower limb. The degree of muscular atrophy may not necessarily correlated with the degree of weakness[1,12]. In a few exceptional cases, bilateral involvement is observed. An irregular distal tremor (minipolymyoclonia) also seen in spinal muscular atrophy is present in the upper limb in the majority of cases. This type of tremor is present during rest and is worsened by voluntary activity as well as stress[5,12]. Less common findings include cramps and autonomic disturbances such as hyperhydrosis, aggravation of symptoms during cold and distal cyanosis[11,18]. Myofasciculations are rarely seen. Tendon reflexes may be normal, increased or decreased in the affected segment[5,13]. Neither the pyramidal tracts, nor the cranial nerves, extrapyramidal, somatosensory or cerebellar systems are involved[12].

MA of the upper limb is most commonly reported type in the literature of western countries, while the lower limb is most commonly involved in India[19]. In the upper limb, weakness and atrophy are known to affect the hand and forearm muscles to a greater extent, with sparing of the braquiorradialis, thus giving the impression of an oblique amyotrohpy. Chief complain among patients are lack of forceful hand and finger flexion along with difficulty of fine, discriminative hand movements. The isolated involvement of the arm and shoulder muscles, especially the braquial biceps and deltoid, is rare. The most commonly involved lower limb muscles are the femoral quadriceps and leg muscles, and, less commonly, pelvic and feet muscles[5,18].

The majority of patients present an insidious, slowly progressive course anywhere from 2 to five years, followed by spontaneous stabilization of the disorder. The mean age of onset in over 90% of cases is between 18 and 22 years of age, sometimes extending to 15 to 35 years of age. There is a marked predilection for males of 10:1[1,12]. Although MA is clinically evident in only one limb, EMG and MRI may reveal subclinical involvement in other muscle groups, as in owner case.

The cause of MA is unknown though diverse hypotheses such as viral infection, vascular failure/insufficiency of the spinal cord, excessive physical activity, contact sports, and focal spinal atrophy as a result of spinal cord injury due to forced flexion of the cervical vertebral column have been considered. However, these theories are not sufficient to account for its unique geographic distribution and its predilection for asians[12].

The concept that anterior horn motor cells are essentially involved is based upon eletromyographic and clinical evidence and also upon histological evidence of neurogenic atrophy. The changes seen at EMG are characterized by polyphasic action potentials of increased amplitude, representing mechanisms of denervation and reinnervation. Myofasciculations may be seen. In some instances, subclinical changes may be detected in clinically healthy unaffected muscle groups. Nerve conduction studies are normal[1,5]. The absence of difuse, disseminated denervation is an important factor for diagnostic exclusion of lower motor neuron diseases such as amyotrophic lateral sclerosis, the progressive spinal amyotrophies as well as other hereditary muscular disorders[5].

The diagnosis of MA should always be considered in a patient presenting with slowly progressive unilateral amyotrophy in a single limb associated with weakness. Other possible causes for these symptoms should be ruled out, as well as close follow-up be started as son as possible for these patients should any additional signs of lower motor neuron disease occur. The differential diagnosis of MA include all disorders that present painless unilateral weakness and atrophy restricted to a single limb. Distal spinal muscular atrophy is characterized by bilateral, symmetric involvement, progressively smoldering clinical course and positive family history in many patients. The absence of involvement of bulbar nerves and pyramidal tracts as well as of other muscles separates MA from the initially localized form of amyotrophic lateral sclerosis. Structural lesions such as herniated disk, syringomielia, neoplasms and arteriovenous

malformations present with symptoms of altered sensitivity as well as characteristically defined images on MRI of the vertebral column[5,20].

This patient presented with a very rare form of MA that affected the proximal muscles of the upper limb. The weakness was essentially of the right shoulder, contrary to most commonly seen locations of MA such as the midlle and distal thirds of the upper limb[5,18]. The absence during four years of corticospinal tract signs, of somatossensory involvement as well as involvement in other muscle groups are fundamental for the diagnosis[5,12,20]. The pattern of denervation on EMG was confined to the key muscles related to the anterior horn cells of the first cervical spinal cord segments, thus justifying the focal nature of the disease. A subtle alteration in the EMG of the contralateral limb muscle and of another one in the lower limbs in our patient may be compared to that described in other case reports, always being subclinical or clinically silent (assymptomatic). The importance of our case report lies in the singular location of one proximal muscle of the upper limb, up until know rarely described in the medical literature.

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