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Pseudopolyneuritic form of amyotrophic lateral sclerosis: Marie-Patrikios type

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ABSTRACT

Introduction: Amyotrophic lateral sclerosis (ALS), also called motor neuron disease (MND), is a progressive, neurodegenerative, and inexorable disease that affects the neurons of the anterior horn of the spinal cord, as well as the lateral funiculus. A rare variant of ALS was first described in 1918 by Patrikios and Marie, called the pseudopolyneuritic form or Marie-Patrikios disease. It is characterized by an initial manifestation with melting of the feet, distal weakness of the muscles of the anterior compartment of the leg, and absence of the Achilles tendon reflex. We present an atypical case of ALS, marked by polyneuropathy and involvement of upper and lower motor neurons.

Case Report: A 70-year-old man reported that approximately four years ago he started having pain in the thoracic region with subsequent paresis in the lower limbs. Initially, compressive myelopathy, transverse myelitis, and spastic paraparesis of various causes were thought to be the cause. However, the non-impairment of the superficial and deep sensibility, obviously, with absence of sensorial level, associated to the absence of specific imaging findings in the thoracic and lumbar spine, a normal complete laboratory, ruled out such hypotheses.

Conclusion: We highlight that the pseudopolyneuritic form presented in this study has a better prognosis and survival rate when compared to other subtypes of ALS. Thus, a detailed investigation including physical, neurological, and electrophysiological examination is essential to establish the diagnosis and increase the scarce knowledge about this condition.

Keywords: Achilles tendon reflex, Amyotrophic lateral sclerosis, Heterogeneity, Pseudopolyneuritic form

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INTRODUCTION

Amyotrophic lateral sclerosis (ALS), also called motor neuron disease (MND), is a progressive, neurodegenerative, and relapsing disease which affects the neurons of the anterior end of the spinal cord, as well as the lateral funiculus [1]. The incidence in the population is heterogeneous and ranges from 0.73 to 1.89 cases per 100,000 persons per year in South Asia and Northern Europe, respectively [2, 3]. It is a progressive condition

comprising degeneration of the motor system at various levels: bulbar, cervical, thoracic, and lumbar [4].

A rare variant of ALS was first described in 1918 by Patrikios and Marie, called the pseudopolyneuritic form or Marie-Patrikios disease. It is characterized by an initial manifestation with melting of the feet, distal weakness of the muscles of the anterior compartment of the leg, and absence of the Achilles tendon reflex, mimicking a peripheral neuropathy. Patellar and upper limb tendon reflexes may show hyperreflexia [5].

This form may be confused with some clinical conditions, such as lumbar plexopathy, fibular neuropathy, and distal myopathy, due to the presence of foot melting. Therefore, the patient's clinical history, neurological examination, and electrophysiological evaluation should be carefully analyzed for a differential diagnosis [6].

Other syndromes related to this spectrum of ALS disorders include progressive bulbar palsy, progressive spinal amyotrophy, primary lateral sclerosis, unstable arm syndrome, and unstable leg syndrome—pseudopolyneuritic form of ALS [7].

Symptoms related to upper limb and bulbar involvement are minimal or even nonexistent, although they invariably occur in advanced stages of the disease. Furthermore, it is difficult to distinguish in early stages from other disorders such as distal spinal atrophy, lumbosacral radiculopathy, and multifocal motor neuropathy [8].

Histopathologically, a cellular degeneration had been described preferentially of the lumbar spinal cord. Later, severe loss of lower motor neurons throughout the spinal cord and depletion of the minimal neurons in the middle zone of the anterior horn of the lumbar spinal cord were described. However, to date, knowledge about the clinical and histopathological findings of the variant is limited [9].

Therefore, the aim of the present study is to demonstrate, through a case report, a rare form of ALS, the pseudopolyneuritic or Marie-Patrikios form, and thus, to discuss the current knowledge about the clinical and histopathological findings of the variant.

CASE REPORT

A 70-year-old man reported that approximately four years ago he started having pain in the thoracic region with subsequent paresis in the lower limbs. Initially, compressive myelopathy, transverse myelitis, and spastic paraparesis of various causes were thought to be the cause. However, the non-impairment of the superficial and deep sensibility, obviously, with absence of sensorial level, associated to the absence of specific imaging findings in the thoracic and lumbar spine, a normal complete laboratory (Vitamin B12, HTLV, genetics for spastic paraparesis, among marked inflammatory, infectious, and immunological) and normal cerebrospinal fluid, ruled out such hypotheses. It is worth mentioning that the electroneuromyography identified a sensorimotor polyneuropathy with axonal predominance. Neurological examination revealed amyotrophy and paresis [Medical Research Council (MRC): grade 3/4] in the lower limbs, with Babinski's sign bilaterally present, and mild spasticity (Ashworth 1) in the evaluated groupings (quadriceps, gastrocnemius, soleus, and posterior tibial), as well as in the finger flexors. No myofasciculations were observed. In the upper limbs only hyperreflexia with Hoffman's sign present on the right. Muscle strength and sensitivity were normal. After six months, the patient returns for a medical consultation with complaints related to the progression of muscle weakness, confining him to a wheelchair; (MRC: 1/2) in the muscles of the lower limbs. At this point muscle weakness had already started to affect the upper limbs (MRC: 3/4) (Figure 1); being associated with myofasciculations. His cranial nerves were normal with normal cognition and higher functions. Upon further electroneuromyography examination, a neurogenic pattern with fibrillation potentials, positive waves, and myofasciculations was demonstrated. Added to this was a predominance of neurogenic (long duration, polyphasic, and large amplitude) motor unit action potentials (MUAPs). Findings of compatibility to sensorimotor polyneuroradiculopathy and plexopathy, more pronounced in the lower limbs, with acute and chronic denervation were also described. With disease progression and new findings of first and second motor neuron involvement, the patient was diagnosed with ALS pseudopolyneuritic form of Marie-Patrikios; an association between polyneuroradiculopathy and motor neuron disease.

Unfortunately, the patient has been showing rapid evolution, already with damage in the four limbs and breathing. Dysphagia is already present on some occasions, mainly with liquids.

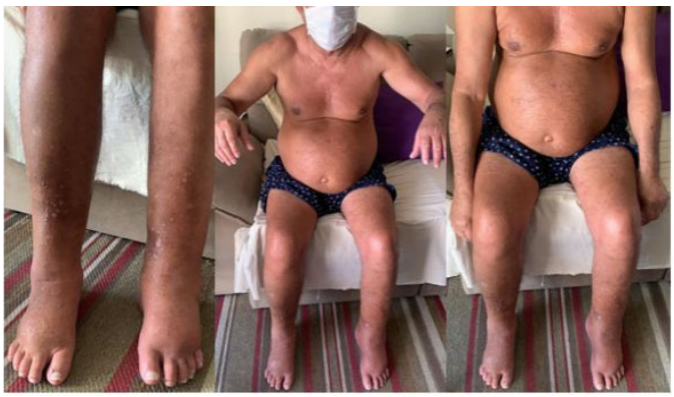

Figure 1: Lower limbs edema and distal muscle weakness at upper limbs.

DISCUSSION

Amyotrophic lateral sclerosis is a prototype of motor neuron disease and numerous theories have been

proposed for its etiopathogenesis, but none so far have been unifying, among them: genetic factors, excitotoxicity, oxidative stress, mutations in the copper-zinc SOD1 gene, mitochondrial dysfunction, neurofilament, protein aggregation, etc. The average age of disease onset varies between 55 and 65 years, and men are more affected compared to women, with a ratio of 1.5:1, respectively [10].

The pseudopolyneuritic form of ALS (Table 1) is a subtype characterized by distal weakness of the lower limbs and absence of the Achilles tendon reflex. The etiopathogenesis of the Patrikios form is defined by the loss of myelinated fibers in the corticospinal tract of the thoracic and lumbar spinal cord segments [11].

This variant was discovered by Pierre Marie and first described by Patrikios, thus named the Marie-Patrikios form. It was described as a syndrome which includes weakness of distal onset in the lower limbs, clinically asymmetric, with absence of tendon reflexes in the lower limbs, slow progression, and late manifestations of the upper motor neuron. Moreover, this subtype occurs preferentially in women when compared to other forms of ALS [12].

Studies indicate that this form has a significantly better prognosis compared to the bulbar form of ALS. The survival time with this form is the onset of symptoms ranges from 30 to 69 months, and the frequency is 1–17.5% [13].

The recognition of this form of ALS is essential for clinicians, since the combination of distal lower limb weakness, in addition to the absence of the Achilles reflex, often suggests a peripheral neuropathy. In clinical practice, the identification of hyperreflexia in the patellar and/or upper limb reflexes may be the key to the diagnosis of ALS, even if in exceptional cases no signs of upper motor neuron involvement are observed [14].

Table 1: Characteristics of ALS pseudopolyneuritic or Marie-Patrikios form

Pathogenesis	Loss of myelinated fibers in the corticospinal tract of the thoracic and lumbar spinal cord segments
Weakness pattern	Distal weakness of the lower limbs and absence of the Achilles tendon reflex, upper motor neuron late manifestations
Progression	Slow progression
Gender	Preferentially in women
Prognosis	Better prognosis compared to the bulbar form of amyotrophic lateral sclerosis. The survival time ranges from 30 to 69 months

CONCLUSION

We highlight that the pseudopolyneuritic form presented in this study has a better prognosis and survival rate when compared to other subtypes of ALS. In this regard, due to its symptomatological particularities, there may be a misunderstanding regarding the differential diagnosis of polyneuropathy. Thus, a detailed investigation including physical, neurological, and electrophysiological examination is essential to establish the diagnosis and to increase the scarce knowledge about this condition. The polyneuritic form of Patrikios is in the hall of variants of the classic ALS. Differential diagnosis is based on physical and electrophysiological examination. The prognosis, although considered by some authors as less aggressive, does not seem to us to be different from the classic cases of ALS.

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Author Contributions

Marco Orsini – Conception of the work, Design of the work, Acquisition of data, Drafting the work, Final approval of the version to be published, Agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved

Antônio Marcos da Silva Catharino – Analysis of data, Interpretation of data, Revising the work critically for important intellectual content, Final approval of the version to be published, Agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved

Valéria Camargo Silveira – Design of the work, Analysis of data, Revising the work critically for important intellectual content, Final approval of the version to be published, Agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved

Carlos Henrique Melo Reis – Analysis of data, Interpretation of data, Revising the work critically for important intellectual content, Final approval of the version to be published, Agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved

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Guarantor of Submission

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Consent Statement

Written informed consent was obtained from the patient for publication of this article.

Conflict of Interest

Authors declare no conflict of interest.

Data Availability

All relevant data are within the paper and its Supporting Information files.