CASO CLÍNICO

Progressive solitary sclerosis: isolated lesion, progressive deficits
Esclerose solitária progressiva: lesão única, défices progressivos

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Abstract

Introduction: A single demyelinating lesion is a possible cause of progressive disability, an entity known as Progressive solitary sclerosis. These cases do not fulfil the diagnostic criteria for Multiple sclerosis, although some authors suggest a continuum between Progressive solitary sclerosis and Primary Progressive Multiple sclerosis.

Clinical case: We report the case of a woman that presented with an isolated intrinsic cord lesion which led to a progressive left lower limb motor deficit. The extensive investigation undergone excluded other causes of myelopathy and serial MRIs did not show progression of the spinal cord lesion.

Conclusions: This case illustrates a clinic-radiological paradox, and its etiology is still unknown. Since there seems to be an association between these two conditions, a revision of Multiple sclerosis diagnostic criteria was proposed to include this presentation.

Resumo

Introdução: A presença de uma lesão desmielinizante única tem vindo a ser apontada como uma causa possível de incapacidade progressiva, entidade conhecida como Esclerose solitária progressiva. Nestes casos os critérios para esclerose múltipla não são cumpridos, embora alguns autores tenham sugerido um continuum entre Esclerose solitária progressiva e a forma primária progressiva da Esclerose múltipla.

Caso clínico: Os autores apresentam o caso de uma doente com uma lesão intramedular única que acarretou défice motor progressivo no membro inferior esquerdo. A extensa investigação realizada excluiu causas de mielopatia e a vigilância imagiológica não mostrou progressão da lesão.

Conclusões: Este caso descreve um paradoxo clinico-radiológico, cuja causa permanece ainda desconhecida. Foi sugerido que esta apresentação seja integrada nas revisões dos critérios de diagnóstico da Esclerose múltipla, dado existirem alterações que apontam para uma associação entre estas duas patologias.

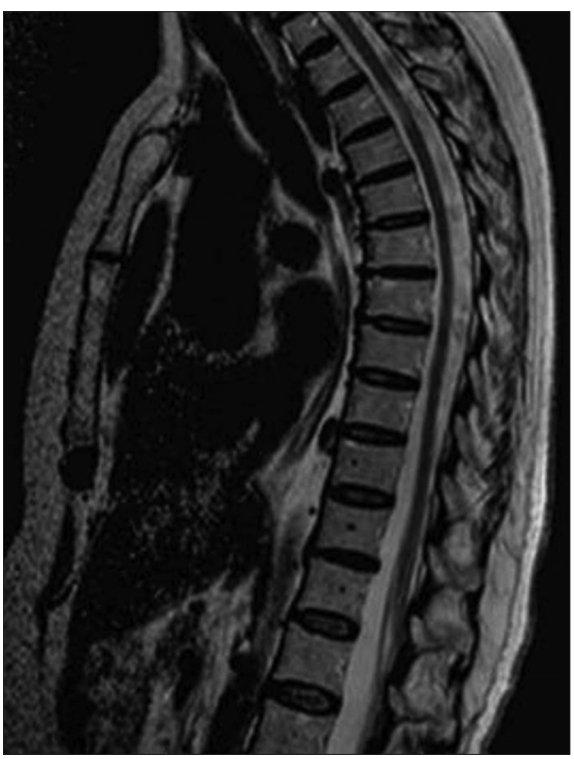
Introduction

When a patient presents with a progressive motor deficit a multitude of diagnoses must be pondered. Among these the inflammatory demyelinating diseases (IDD) are an important group. In some cases, a single demyelinating lesion of the central nervous system (CNS) may cause a progressive motor deficit, a condition known as Progressive solitary sclerosis (PSS). It was first described in 1990. Since then few cases have been described and the largest series was published in 2016.[1] To our knowledge this is the first report of a case of PSS in Portugal.

Case report

A 67-year-old woman with a past medical history of an anxiety disorder, presented to our clinic in 2014 with monoparesis and sphincter dysfunction. The first symptom the patient reported was a feeling of "lump in the throat" in 2010, although she denied dysphagia for liquids and solids, even if it was never formally tested. In 2011, for this reason, she was observed by a Neurologist. Because of an asymmetric evaluation of algic sensation in her legs, a spinal cord magnetic resonance imaging (MRI) was performed and showed an anterior left sided intrinsic cord lesion at D3-D4 with slight edema and gadolinium enhancement. At this stage brain MRI disclosed one nonspecific T2 hyperintense subcortical parietal lesion. About one year later she repeated the spinal cord MRI and there was a decrease in size of the isolated lesion, without gadolinium enhancement, making a neoplastic lesion unlikely. In the following two years there was a progression of the left lower limb motor deficit and the patient underwent the following investigation: CSF analysis with no oligoclonal bands (OCBs) or malignant cells; blood tests (including immunological study and aquaporin-4 antibodies) were negative; visual evoked potential studies, somatosensory evoked potentials and electromyography were normal. In 2014, she was admitted to the Neurology ward of our clinic. On examination, she had a pure motor monoparesis grade 3+/5 of the left lower extremity with brisk reflexes and indifferent cutaneoplantar reflex on the left side. The previous studies were repeated and we confirmed the preceding findings: blood tests (including vitamin B12, folate and angiotensin converting enzyme), viral serologies and immunological study were normal; CSF analysis showed 8 cells, normal protein level (0,49 g/L), borrelia antibodies were negative, as well as syphilis testing. OCBs were absent. MRI angiography excluded a dural arteriovenous fistula. The patient received IV methylprednisolone for 5 days without any benefit. After discharge, her motor function continued to decline, resulting in an EDSS of 6, since unilateral assistance is required for her to walk. No immunomodulatory or immunosuppressive treatment was instituted. Spinal cord and brain MRI was repeated last April showing mild atrophy of the thoracic cord lesion and there is no evidence of dissemination in space (Fig. 1).

Fig. 1. Spinal cord MRI (2017)



Discussion

The patient presented seems to fulfil the criteria proposed by Schmalstieg el al[2] for PSS: (1) MRI evidence of a focal, T2 hyperintense lesion involving the corticospinal tracts in the brainstem or upper cervical spinal cord; (2) progressive motor deficit (duration ≥ 1 year) attributable to the lesion on MRI; (3) MRI of the brain and spinal cord did not fulfil the requirements for dissemination in space; (4) no clinical history suggesting relapses affecting other portions of the central nervous system. Importantly, no alternative diagnosis was identified after extensive testing and follow up, which is a key first step

when diagnosing an IDD. For instance, Neuromyelitis optica spectrum disorders, with MOG-Abs or AQP4-Abs, can also present with spinal cord lesions, that although different among each other, typically exhibit a longitudinally extensive myelitis and follow a relapsing course with no progressive phase.

From all the reported cases, some other characteristics have been defined. Age of onset ranges from 23 to 71 years[1], and the majority are women. There is no history of relapses and motor disability accumulates, with a mean EDSS of 6[1,3] after 7,5 years.[2] The lesions found on MRI, resemble demyelinating plaques,[2] and are usually located in the cervico-medullary junction or the cervical spinal cord.[2-6] Nevertheless, the thoracic spinal cord and subcortical white matter may also be the lesion site.[1] As in our patient, nonspecific brain MRI white matter T2 hyperintensities may be found in some cases.[1]

CSF analysis may show OCBs in 50% of patient[1] and electrodiagnostic studies do not demonstrate abnormalities establishing spatial dissemination.[6]

One curious aspect of this condition is the extent of disability that comes from a single lesion. No explanation has been found, but it has been suggested that it is due to the strategic location of the lesions in places where there is concentration of functionally critical tracts.[2]

Immunomodulatory and immunosuppressive treatments have been used:[1,3,6] methylprednisolone, cyclophosphamide, azathioprine, plasma exchange, mitoxantrone, natalizumab, glatiramer acetate, methotrexate and there were isolated reports of disease stabilization with immunosuppression,[6] but no treatment showed definite benefits in this group of patients . Also, symptomatic treatments, such as dalfampridine, could be tried.[1]

The presence of a progressive course, well circumscribed lesions, involving short segments of the spinal cord, and of OCBs has led to the suggestion that there is a continuum between PSS and primary progressive multiple sclerosis (PPMS).[4] However, this group of patients does not fulfil the criteria for dissemination in space,[7] and may represent an MS variant. According to some groups,[1] such patients might be considered in future revisions of MS diagnostic criteria. ■

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