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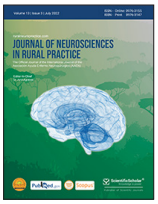

Case Report

Brachial monomelic amyotrophy as an initial manifestation of stiff person syndrome

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ABSTRACT

Stiff person syndrome (SPS) is characterized by rigidity of truncal and proximal muscles. The presence of abdominal and paraspinal rigidity is a defining clinical feature of SPS. It is rarely associated with the lower motor neuron (LMN) features. We report a patient with SPS whose initial clinical presentation was that of brachial monomelic amyotrophy (BMA). A 24-year-old gentleman presented with a history of the left upper limb wasting and weakness. In addition, he reported stiffness of the lower limbs and abdomen while walking. On examination, patient had left upper limb monomelic amyotrophy and hypertonia, exaggerated deep tendon reflexes in all four limbs. He also had abdominal and paraspinal rigidity. Serum was strongly positive for GAD 65 antibodies suggestive of SPS. Patient showed dramatic improvement to immunomodulation. Patient presented with features of BMA. Symptoms related to SPS were mild. Abdominal rigidity was the clue to the diagnosis. LMN features have been reported previously in stiff person plus syndrome with an atypical course and progressive encephalomyelitis with myoclonus and rigidity, but not in classical SPS.

Keywords: Anti-nerve antibodies, Auto-antibodies in neurological disorders, GAD antibodies, Hirayama disease, Lower motor neuron syndromes

INTRODUCTION

Stiff person syndrome (SPS) is characterized by rigidity of truncal and proximal limb muscles, associated with painful spasms.[1] SPS can be associated with additional clinical features such as encephalopathy, myoclonus, and rarely lower motor neuron (LMN) features.[2] Brachial monomelic amyotrophy (BMA) is characterized by atrophy and weakness of the upper limb in C7, C8, and T1 distribution.[3] We report a patient with SPS who presented with features of BMA.

CASE REPORT

A previously healthy 24-year-old gentleman presented with history of progressive thinning and weakness of the left upper limb for 1 year. For 8 months, patient reported difficulty in walking due to limb and abdominal stiffness. There were no symptoms in the right upper limb and no bowel, bladder, or speech or swallowing disturbances. Patient did not give any history of spasms, poliomyelitis, or neck trauma.

On examination, palpation revealed rigidity over abdominal muscles and paraspinal muscles. Minipolymyoclonus was observed over the left distal upper limb. Wasting was seen over the left thenar and hypothenar muscles, dorsal interossei, and long flexor muscles of the left forearm with sparing of brachioradialis [Figure 1]. Occasional fasciculations were seen over triceps. Rigidity was noted over both upper limbs and lower limbs (LL>UL). Motor examination revealed weakness of small muscles of the left hand, exaggerated deep tendon reflexes (DTR), and extensor plantar response. Gait examination revealed exaggerated lumbar lordosis.

Investigations revealed normal nerve conduction study while electromyography (EMG) of upper and lower limb muscles showed fasciculations and polyphasic motor unit action potential in the left abductor digitiminimi only. EMG testing of other limb muscles, abdominal and spinal muscles was normal. MRI spine showed lower cervical cord atrophy (C8, T1) while imaging of the brain was normal. Flexion extension studies of MRI spinal cord showed no evidence of dynamic compression. Serum was strongly positive for glutamic acid decarboxylase (GAD) 65 antibodies, tested by immunoblot assay as well as enzyme immunoassay (1125 IU/ml), hence, confirming a diagnosis of SPS. Blood investigations did not reveal evidence of associated immune disorders or other paraneoplastic antibodies.

Patient was treated with 90 g of intravenous immunoglobulin, clonazepam 2 mg/day, and gabapentin 600 mg/day with which he reported complete improvement in symptoms. At 9 months of follow-up, patient reported complete resolution

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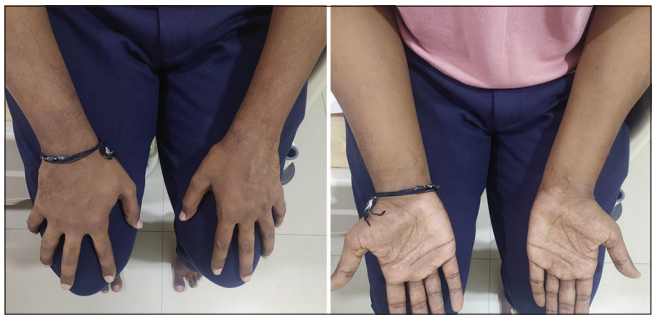

Figure 1: Atrophy of first dorsal interossei, long flexors of forearm and thenar, and hypothenar muscles of the left upper limb.

of stiffness of the lower limb and non-progression of the upper limb wasting and weakness.

DISCUSSION

The presentation of our patient led to the initial diagnosis of BMA. According to defining criteria by Tashiro *et al*., diagnosis of Hirayama disease requires the presence of normal reflexes.[3] However, exaggerated reflexes in involved and uninvolved limbs have been reported in BMA.[4] Differential diagnosis of BMA includes multifocal motor neuropathy with conduction block, distal spinal muscular atrophy, amyotrophic lateral sclerosis (ALS), and cervical myelopathy.[5] The presence of LMN involvement of the upper limbs with the upper motor neuron (UMN) signs in the lower limbs as seen in our patient would clinically localize the lesion to an intramedullary spinal cord pathology or to motor neuron disease. MRI of the spinal cord showed atrophy and no evidence of dynamic cervical cord compression that might be encountered in patients with Hirayama disease.[6] Exaggerated lower limb reflexes and rigidity of the lower limbs in the background of UMN and LMN features in the upper limb might lead to a strong clinical suspicion of motor neuron disease and warrantee routine EMG evaluation at subsequent follow-up visits to look for ALS. The benign and self-limiting course as well as lack of progression to ALS in patients with BMA with exaggerated reflexes in uninvolved limbs has been previously documented.[7]

The presence of abdominal wall rigidity was an important clinical clue that suggested the possibility of SPS despite the presence of LMN features. Symptoms pertaining to SPS were mild and without features of progressive encephalomyelitis with rigidity and myoclonus (PERM). The presence of LMN involvement in a patient with SPS classifies our patient as having stiff person plus syndrome.[2] Our patient did not have painful spasms or EMG findings of continuous motor unit firing (CMUF) at rest, hence, partially satisfying the criteria of SPS proposed by Dalakas.[1] However, one study showed the presence of CMUF in only 61.9% of patients with SPS.[8]

Whether BMA and SPS coexist in our patient or he has stiff person plus syndrome is a matter of debate. To the best of our knowledge, there has been only one case report of clinical motor neuron involvement in SPS with an atypical course. His autopsy showed spinal cord infiltration with cytotoxic T cells and microglial activation. Clinical evidence of LMN involvement has been observed more frequently in PERM but not in SPS.[9] Involvement of motor neurons beyond those involved in GABAergic transmission have been reported in SPS,[10] further reiterating the expanded clinical spectrum of SPS.

CONCLUSION

BMA can be rarely seen in association with SPS. In our patient, the clinical presentation was dominated by BMA with subtle features of SPS. SPS can be readily misdiagnosed as motor neuron disease (ALS or monomelic amyotrophy) if the clinical presentation of rigidity and exaggerated DTR is associated with LMN features. A high index of suspicion and mandatory examination of abdominal muscles will enable prompt diagnosis.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent.

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Nil.

Conflicts of interest

There are no conflicts of interest.

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