STIFF LIMB SYNDROME: AN UNUSUAL PRESENTATION.

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INTRODUCTION

Stiff-person syndrome (SPS) is a rare autoimmune neurological disorder characterized by continuous muscle activity causing severe rigidity and episodic spasms in axial and limb muscles leading to sever pain.
Different phenotypes have been described: stiff trunk syndrome (classic), stiff limb syndrome, progressive encephalomyelitis with rigidity and myoclonus (PERM).
SPS is associated with a large number of antibodies.
In this paper we illustrate a focal anti-glutamic acid decarboxylase (anti-GAD) antibody mediated variant of the disease.

CASE REPORT

A 56-years-old woman presented an acute onset of spasms in left shoulder, spreading to left leg in a month with rapid increase in frequency and duration; because of the rigidity, the patient began showing a progressive difficulty in walking with frequent falls.
She underwent an orthopedic surgery after a femur fracture due to a fall and then she was admitted to our hospital.
The neurological examination disclosed intermittent cramps and stiffness in left shoulder and leg with a decreased movement range; no other pathological findings were discovered.
During hospitalization, she developed a localized itch over the left infrascapular region, probably due to rigidity.
We used a panel of antibodies to intracellular antigens by which we detected a high titre-of anti-GAD antibodies (540 IU/ml).
Results from other laboratory tests, including viral exams, anti-ANA, ENA, ANCA and the neuronal antibodies to extracellular antigens (anti VGCC, anti VGKC, anti-GLUR3, anti-GABA B, anti NMDAR, anti CASPR2, anti LGI1, anti AMPA antibodies), were negative.
Needle EMG (EMG testing infraspinatus, deltoideus, pectoralis major, biceps brachii, flexor carpi radialis, abductor pollicis brevis, opponens pollicis, infraspinatus, rectus abdominis, rectus femoris, vastus medialis, paraspinales D5-D6, sx) showed a focal continuous motor unit activity and a co-contraction of agonist and antagonist muscles; brain and cervical spine MRI scans were normal.
In order to rule out a paraneoplastic aetiology, a CT scan of thorax and abdomen and a mammography were taken without significant results.
The patient was diagnosed with SPS associated with the presence of GAD antibodies; treatment with delorazepam (3,5 mg/die) improved symptoms, including itch (notalgia paresthetica?).

CONCLUSIONS

Several diseases can mimic SPS (for example Isaac syndrome and tetanus); differentiating the phenotypes of SPS may be a diagnostic challenge: clinical features, EMG patterns and immunological tests allow a prompt diagnosis in order to avoid a progression of disease with an important disability.

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