STIFF PERSON SYNDROME IN A PATIENT WITH THYROIDITIS AND MISDIAGNOSED LADA. CASE REPORT

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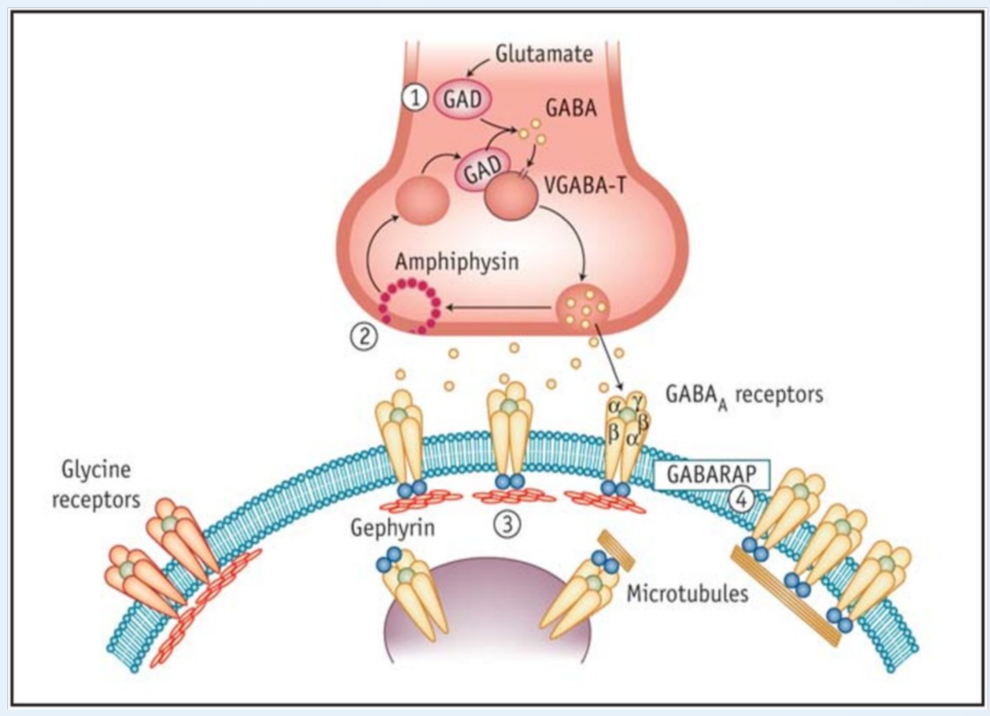
Background: Stiff person syndrome (SPS) is a rare neurological disorder characterized by fluctuating stiffness and superimposed episodic spasms in axial and proximal limb muscles. SPS is mostly reported in women and associated with the presence of autoantibodies against glutamic acid decarboxylase (GAD). The autoimmune pathogenesis of SPS is supported by the strong association with some autoimmune-mediated disorders such as type 1 diabetes mellitus (DM1), pernicious anemia, and thyroiditis. However, it can also present as paraneoplastic or cryptogenic variant.

Case report: A 49-year-old Caucasian woman, with a recent history of misdiagnosed latent autoimmune diabetes of adulthood (LADA), was admitted to our neurologic unit because of difficulty ambulating associated with fluctuating stiffness in the left leg, painful episodic spams in trunk and limb muscles and frequent falls. In the six years previously, were referred two episodes characterized by subjective dizziness, nausea, vomiting and vertical diplopia resolved after intravenous steroid therapy. Moreover, in November 2009, she presented rigidity and superimposed spasms of paraspinal and limb muscles associated with fear of crossing a street without assistant. These symptoms lasted several months and resolved spontaneously. In the month before our observation, she performed brain and spinal MRI that showed gliotic aspecific brain lesions, lumbar disk herniation with no spinal cord lesions and cauda neurinoma. After psychiatric evaluation, she received diagnosis of conversion disorder and assumed diazepam with mild benefit. At the time of admission, the neurological exam revealed precautionary gait, increased tone in the thoracic and abdominal paraspinal muscles with hyperlordosis, nystagmus in horizontal gaze and upward and tendon reflex increased.
Electromyography showed subcontinued activation of paraspinal muscles and co-contraction of agonist and antagonist muscles in the left wrist flexion. Onconeural antibodies and autoimmune encephalitis antibodies was all normal, whereas high *serum* titer of *anti-GAD antibodies* was found *(455 UA/mL)*. Moreover, anti-nuclear antibodies (ANA) and anti-thyroid antibodies were positive with normal hormones levels. The *CSF* analysis showed intrathecal IgG synthesis and the positivity of *anti-GAD antibodies (134 UI/mL)*. Total body PET-TC did not show pathological FDG-glucose uptake. A diagnosis of SPS was made and she was treated with diazepam, baclofen, and Intravenous Immunoglobulin (IVIG) with benefit particularly on painful spasms.

Conclusions: Our case supports the need of close follow-up in LADA patients. Truncal and proximal limb stiffness associated with painful spasms are often underestimated symptoms from both neurologist and endocrinologist leading to psychiatric diagnosis. A timely recognition of SPS the disease and prompt treatment can improve the quality of life of SPS patients.

Figure 1. Pre- and post-synaptic target antigens in SPS.



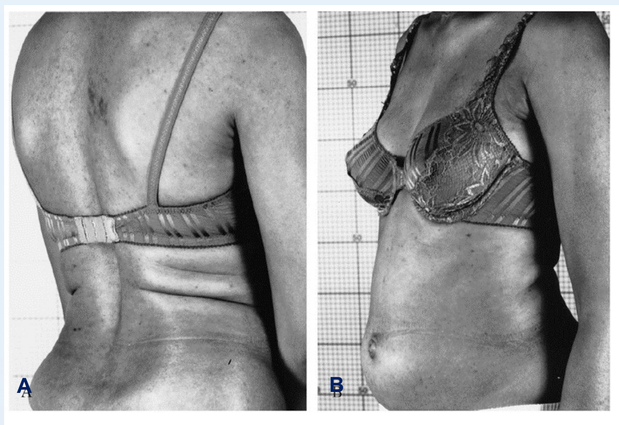
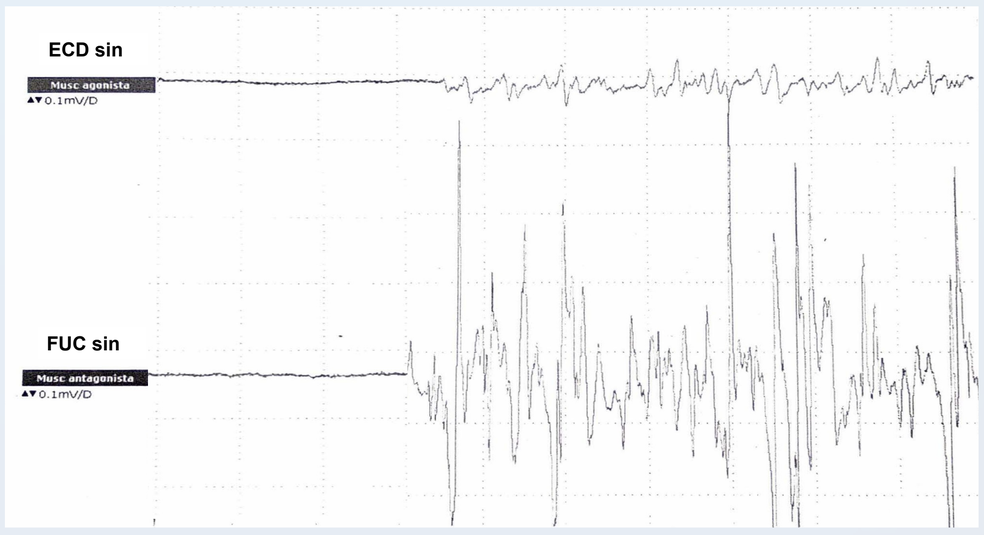


Figure 2. Hyperlordosis with co-contracture of the thoracolumbar paraspinals (A) and abdominal muscles (B)

Figure 3. Electromyographic evidence of co-contraction of agonist and antagonist muscles in the left wrist flexion



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