



WEB OF SCIENCE



STIFF-PERSON SYNDROME - CASE PRESENTATION



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Abstract

Introduction: Stiff-person syndrome (SPS), formerly known as “stiff-man syndrome”, is a rare neuro-immunological disease, the understanding and diagnosis of which have greatly advanced with the identification of disease-associated autoantibodies. It is associated with high levels of antibodies against glutamic acid decarboxylase (GAD Ab), a protein in inhibitory nerve cells that is involved in the synthesis of the main inhibitory neurotransmitter called gamma-aminobutyric acid (GABA). GABA helps control muscle movement and prevent hyper-excitability within the brain and spine. The symptoms of SPS may develop when the immune system mistakenly attacks certain nerve cells (neurons) that produce GAD, leading to a deficiency of GABA in the body.

SPS may be associated with other autoimmune disorders, more frequently diabetes. Less commonly, affected individuals may also develop inflammation of the thyroid (thyroiditis), pernicious anemia and vitiligo.

Material and methods: We present the case of 35 years old woman, diagnosed with stiff-person syndrome from 2017. The symptomatology had an insidious onset with walking difficulties (gait abnormalities), muscle pain and rigidity in the inferior limb, and dysarthria. In addition to muscular stiffness, she also developed muscle spasms, which occurred spontaneously or in response to various triggering events. In 2014 she was diagnosed with Latent Autoimmune Diabetes in Adults. The patient was anamnestic (history of the illness), clinically and para-clinically evaluated.

Results and discussions: Taking into account the anamnestic and clinical information, also the presence of GAD antibodies and the associated autoimmunity (diabetes and recently diagnosed thyroiditis), we can say that we are dealing with one of the rare cases of stiff-man syndrome.

Conclusions: SPS is a rare disorder and it is very difficult to diagnose. With an early recognition of the disease and prompt treatment, the quality of life of SPS patients can be improved. Stiffness and spasms interfere with the ability of these patients to fully mobilize affected joints, and they are at risk of developing further complications. Physical therapy may have a role in the management of this disease, as these patients need to be taught how to properly stretch and maintain joint mobility as a lifelong commitment.

Although currently there is no cure for SPS, symptoms can be treated by augmenting spinal cord γ -aminobutyric acid-mediated activity with benzodiazepines and baclofen. Plasmapheresis and intravenous immunoglobulin have been used to diminish the underlying autoimmune response. Though the first line of drugs for SPS is benzodiazepines and baclofen, their dose-related adverse effects are of major concern.

KEY-WORDS: *stiff-person syndrome, GAD antibodies, muscle spasms, neuro-immunological disease*