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Abstract

Stiff-Person Syndrome: Seeing Past Comorbidities to Reach the Correct Diagnosis

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ABSTRACT

Stiff-person syndrome (SPS) is a rare disorder seen in approximately one in one-million people. Although it is rare, the symptoms and findings of a typical case should paint a clear clinical picture for those who are familiar with the disease. The primary findings in SPS include progressive axial muscle rigidity as well as muscle spasms. These symptoms most commonly occur in the setting of antibodies against Glutamic Acid Decarboxylase (GAD), the rate-limiting enzyme in the production of Gamma-Aminobutyric Acid (GABA), which is the primary inhibitory enzyme in the central nervous system. Here, we report the case of a 65-year-old African American female with a past medical history of hypothyroidism, anxiety and depression with psychotic features who presented with axial muscle rigidity and lactic acidosis. She had been symptomatic for several months, and reported extensive workups performed at two previous hospitals without a definitive diagnosis. A complete neurological and musculoskeletal investigation yielded no positive findings except for the presence of GAD antibodies. The patient was treated with diazepam, tizanidine, and Intravenous Immunoglobulin (IVIG) with significant improvement, thus solidifying the diagnosis of SPS, a rare autoimmune and/or paraneoplastic syndrome. This case served as an incredible learning opportunity for the author and will likewise help others to gain an understanding of the disease process that is sufficient to reach the correct diagnosis when seen in a clinical setting.

Keywords: Stiff-person syndrome, GAD antibodies, Neurologic, Musculoskeletal, Rigidity

Abbreviations

SPS: Stiff Person Syndrome; GAD: Glutamic Acid Decarboxylase; GABA: Gamma Amino Butyric Acid; IVIG: Intra Venous Immuno Globulin

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