STIFF PERSON SYNDROME: A CASE REPORT

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BACKGROUND

Stiff Person Syndrome (SPS) is a rare central nervous system disorder characterized by progressive muscle stiffness, rigidity, and painful spasm involving the axial muscles, which can lead to impaired ambulation and overall decrease in function. Spasm includes both agonist and antagonist muscles. It is caused by anti-GAD65 antibodies, leading to a decrease in GABA levels. SPS is commonly seen in conjunction with other auto-immune disorders. The PERM variant of SPS is a more severe and rapidly progressive variant. These patients may also have brainstem dysfunction and alterations in consciousness. Treatment of SPS usually begins with high doses of benzodiazepines or baclofen, and can progress to IVIG in resistant cases.

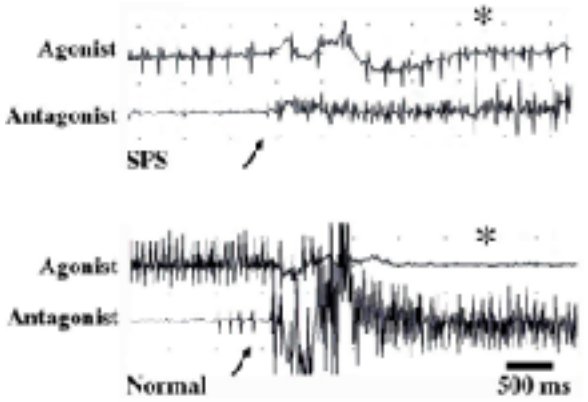
CASE PRESENTATION

38 year old male who developed vertigo associated with nausea and vomiting in March of 2017. He went on to develop coordination deficits and difficulty ambulating, requiring walker assistance, followed by speech and swallowing difficulties, eventually requiring PEG placement. Ambulation progressively worsened and he was wheelchair bound October of 2018, bed-bound thereafter. He also became mute at this time. He was hospitalized for an episode of unconsciousness after he was found unresponsive on the ground. Medical workup revealed anti-GAD65 antibodies and a diagnosis was made of cerebellar ataxia and stiff person syndrome.

PMH: Hepatitis B, GERD, hypothyroidism
PSH: Gastrointestinal stromal tumor resection
Allergies: No known drug allergies

PHYSICAL EXAM

Deficits included mutism, left sided visual gaze preference, stiff and rigid muscle movements with increased muscle tone, and right greater than left sided weakness. He exhibited bradykinesia and poor trunk control. His gait was ataxic with a left to right lean. Moderate impairment of coordination was noted in the bilateral upper extremities.



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HOSPITAL COURSE

There were no significant medical complications during his hospital course. From a rehabilitation perspective, he progressed slowly with minimal improvement in functional gains. Speech therapy allowed for small portion pureed feedings with nectar thick liquids for pleasure feeds in conjunction with bolus tube feeds to meet his nutritional requirements.

DISCUSSION

Stiff Person Syndrome is a rare neurological disorder, most commonly presenting in the third to sixth decade of life. The exact incidence and prevalence of SPS is unknown, but it is estimated to be one in one million, with a 2:1 ratio of females:males. It is a suspected there is a genetic component, however this has not been proven.

This patient's presentation of rapidly progressive SPS, along with alteration in consciousness, is suggestive of the PERM variant. Although not formally diagnosed with the PERM variant of SPS, a review of this case leads the review(s) to believe that the patient presentation is suggestive of a PERM variant. Physical Therapy plays a critical role in treatment of SPS and will be most beneficial when used with anti-spasm medications. Stretching, with or without ultrasound, can be used on spastic muscles to help alleviate pain. Thoracolumbar flexion exercises are indicated in patients with a lumbar hyperlordosis. Isometric and isotonic core exercises are useful in patients with poor trunk control. Gait training is indicated in patients able to or preparing to stand and assistive devices should be considered when necessary. Exercises used to improve coordination are recommended, as this can increase independence, functional mobility and decrease the assistance required to perform ADLs.

This patient follows with a neurologist, who manages the IVIG treatments at a university setting hospital system. Having a multidisciplinary team, as well as a strong support system, is crucial for SPS patients to help manage the potentially devastating consequences of the disease. Physical and Occupational Therapy should be directed to decrease pain, improve ADLs, increase functional mobility and minimize contractures.