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Case report

A case report of patient with cerebellar variant of stiff person syndrome

Ewa Maludzińska [a,*], Monika Rudzińska [b], Artur Stępień [c], Andrzej Szczudlik [d]

[a] Department of Neurology, The John Paul II Specialist Hospital, Krakow, Poland
[b] Department of Neurology, Medical University of Silesia, Faculty of Medicine, Katowice, Poland
[c] Nuclear Medicine Department, 5-th Clinical Military Hospital, Krakow, Poland
[d] Department of Neurology, University Hospital, Jagiellonian University Medical College, Krakow, Poland

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ABSTRACT

Stiff person syndrome (SPS) is a rare autoimmune neurological disorder with antibodies against antigens involved in neurotransmission of gamma-aminobutyric acid (GABA). About 10% of patients with SPS may develop ataxia. This cerebellar variant is a distinct subset of SPS with more severe and complex clinical phenotype.

We report the clinical, neuropsychological and neuroradiological findings in a 39-year-old female with cerebellar variant of SPS.

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1. Introduction

Stiff person syndrome (SPS) is a rare and underdiagnosed neurological disorder characterized by the muscle rigidity and superimposed spasms. The rigidity begins insidiously at the truncal muscles and spreads to the legs. Painful spasms are precipitated by movement, emotional distress and auditory startle. Some patients do not show the classic axial distribution of stiffness. It may start focally from one lower limb, giving rise to the diagnosis of stiff limb syndrome considered as a focal form of SPS in which the symptoms are confined to a limb, although sometimes this progresses to involve the axial musculature as well. Antibodies against glutamic acid decarboxylase (anti-GAD) are diagnostic marker of the SPS, but they are also described in patients with insulin-dependent diabetes mellitus and in patients with cerebellar ataxia. Some patients with SPS may develop additional neurologic abnormalities, including ataxia (10% of patients), epilepsy (5–10% of patients), abnormal eye movements and mental disorders (phobias, anxiety, talkativeness, obsessions). Patients with ataxia (the cerebellar variant of SPS) have more severe and complex clinical phenotype of SPS with more prominent stiffness and spasms in the leg and trunk, cerebellar ataxia; dysarthria; ataxic gait, abnormal eye

* *Corresponding author at*: Department of Neurology, John Paul II Specialist Hospital, 80 Prądnicka Street, 31-202 Krakow, Poland. Tel.: +48 126142730; fax: +48 126142729.
E-mail address: neurologia@onet.eu (E. Maludzińska).
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movements with impaired saccades, deficient smooth pursuit and gaze-holding nystagmus [1,11].

We report the clinical, neuropsychological and neuroradiological findings in a 39-year-old female with cerebellar variant of SPS.

2. Case report

A 39-year-old woman was referred to the department of neurology with a three-year history of cerebellar ataxia and progressive muscle rigidity of axial and limb musculature of unknown origin. In 2008, she began to complain of unsteady gait, vertigo and diplopia. Neurological examination revealed nystagmus and broad-based gait.

Six months later, she reported low back pain and stiffness in the thoracic and lumbar spine with an exaggerated upright posture. She developed cramps of thoracic and abdominal muscles and exaggerated lumbar lordosis. The stiffness has spread to the proximal limb muscles. The spasms of both legs and the low back were usually precipitated by passive and voluntary movement, as well as unexpected noise, but at times occurred spontaneously. Rigidity increased over months and patient was virtually unable to walk outdoors without a cane. Over the last several months, her back spasms had become progressively more painful with exacerbations during stressful situations resulting in frequent falls and difficulty with standing up. She was afraid to walk even with an aid. Past medical history did not reveal any neurological or psychiatric disorder or autoimmune disease. Her family history was unremarkable. She did not smoke or use illegal drugs.

On admission, neurological examination revealed symmetric paraspinal and lower extremities muscle rigidity, an exaggerated lumbar lordosis and a prominent thoracic scoliosis with vertical nystagmus, cerebellar dysarthria, mild dysmetria and dysdiadochokinesia of the upper limbs. A slightly increased tone was noted in her left upper limb, but muscle strength and range of motion were both normal. Strength of the lower limbs could not be assessed because of rigidity and spasms. Sensory examination was normal. Deep tendon reflexes were normal in upper limbs. Knee and ankle jerks were very brisk bilaterally. Pathological reflexes were absent. The gait was slow and stiff because of the rigidity in her both legs. She had problems to initiate gait and could not walk unassisted because she was afraid of falling.

In 2008, neuropsychological study including Rey Auditory Verbal Learning Test, Clock Drawing Test, Verbal Fluency Test, Trail Making Test, Stroop Test, Wisconsin Card Sorting Test, serial number subtractions (7 from 100) revealed impaired selectivity of attention and executive dysfunction. When compared with the neuropsychological assessment completed in 2011 including the same tools, the examination showed continued impairment of higher-order cognitive functions and was suggestive of involvement of frontal-subcortical regions. The assessment revealed increased executive dysfunction and language problems, impairment in short-term memory, learning disturbance, decreased verbal fluency and mental speed, reduced self-criticism, deficits of attention.

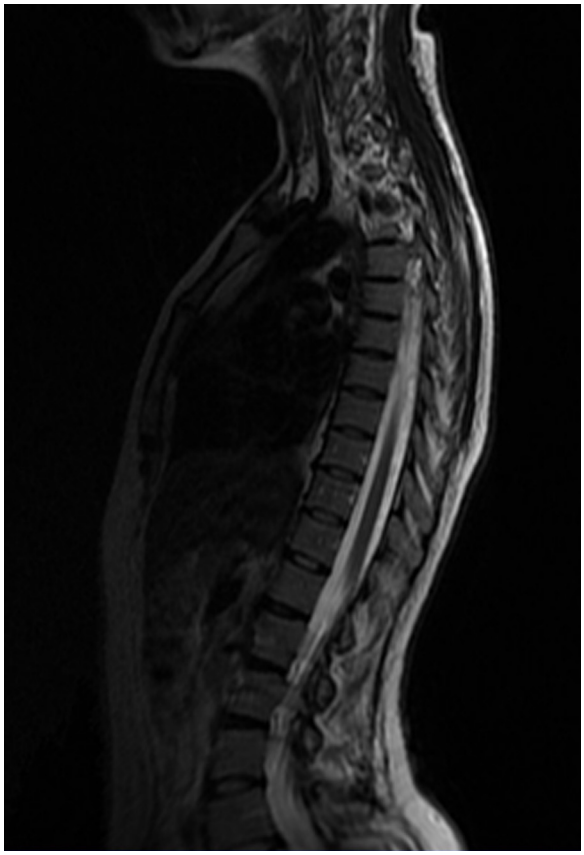

Fig. 1 – MRI of the spine shows an exaggerated lumbar lordosis.

Electromyography revealed continuous motor unit activity in agonist and antagonist muscles at rest. Magnetic resonance imaging (MRI) of the spine before and after gadolinium injection showed thoracic scoliosis and hyperlordosis (Fig. 1). MRI of the brain before and after gadolinium injection showed no evidence of atrophy of frontal lobes or medial temporal lobes (Fig. 2).

Single photon emission computed tomography (SPECT) studies showed bilateral hypoperfusion in frontal lobes, especially on the right side (Fig. 3). Visual and auditory evoked potentials were normal. Results of the routine blood biochemical analyses (including complete blood count, serum electrolytes, blood urea nitrogen, creatinine, glucose, liver enzymes, thyroid-stimulating hormone, ceruloplasmin) and urinalysis were all normal. Anti-GAD autoantibodies were found and their level was above 20000 IU/mL (normal value $<$10 IU/mL). Anti-amphiphysin and paraneoplastic antibodies were not detected. Gene analysis for spinocerebellar ataxia type 1 (SCA1) found no abnormalities. Paraneoplastic antibodies were not detected. Analysis of the cerebrospinal fluid (CSF) wasn't carried out, because the patient refused lumbar puncture.

The patient was diagnosed with SPS according to currently accepted clinical criteria [1]. Oral diazepam was administered at the dose of 30 mg daily and marked reduction of the stiffness was observed. The patient was able to walk with a walking stick. After the increase of diazepam to dose 30 mg/day, levetiracetam at 1000 mg/day was introduced. With this