Stiff man syndrome (SMS), also known as stiff person syndrome, was first described by Moersch and Woltman in 1956 for stiffness and spasms of the extremities and axial muscles. Over time, variant forms of this syndrome such as paraneoplastic SMS, rigid limb syndrome, and progressive encephalomyelitis with myoclonus and rigidity have been identified. Solimena et al. indicated a relationship between the disease and glutamic acid decarboxylase antibody (anti-GAD) positivity and type 1 diabetes mellitus (T1DM) (McKeon, 2012) (Clardy et al., 2013) (Solimena, Folli, Aparisi, Pozza, & De Camilli, 1990). SMS is very rare in children (Clardy et al., 2013). In this article, a case of pediatric SMS with symptoms of walking difficulty, lower extremity stiffness, and involuntary movements is presented.

1. Case

A 9-year-old male patient was admitted to hospital with the symptoms of contraction in both legs, involuntary movements, and difficulty in walking. He had a history of contraction in his left leg two days previously and he fell from a ladder while he was conscious, and then had difficulty in walking due to pain in his waist and leg. He was admitted to another hospital's emergency department, radiographs were obtained and the results were normal. He was discharged with analgesic treatment only. He experiences contraction in both legs and inability to walk when he wakes up the next morning. He described tenderness in the lumbar region. Both legs occasionally formed a dystonic posture and he described severe pain at these times. In his medical history, he had been followed up for a year with the diagnosis of T1DM and is receiving insulin treatment. The family history was unremarkable. Physical examination revealed spasm and tension in both leg muscles that increased with palpation. A neurologic examination revealed that deep tendon reflexes were normactive. Plantar response was bilateral flexor, and clonus was not
observed. The strength of proximal and distal muscles was normal in the extremities, but with stiffness in the thoracolumbar muscles. Cranial nerve examination, cerebellar tests, and sensory examination were normal. Anal sphincter reflex was normal. Laboratory tests revealed normal hemogram, blood glucose, liver, and kidney function tests, and electrolytes (sodium, calcium, potassium and magnesium). Creatinine phosphokinase level was found to be 1447 IU/L (upper limit of 210 IU/L). The thyroid function tests were normal, antinuclear antibody and anti-helix DNA antibody were positive, and the anti-GAD level was 1000 IU/L (<1 IU/L). Chest X-ray, abdominal and thorax ultrasonography, peripheral smear, and tumor markers were normal. On the first day, the patient underwent spinal and cranial magnetic resonance imaging (MRI) in case of any possible spinal trauma and MRI resulted as normal. Focal seizures were considered because of the intermittent stiffness and spasms. Electroencephalography (EEG) was performed and found to be normal. The contractions in the legs increased when he got excited when healthcare staff entered the room or when he wanted to move. Baclofen (body weight 22 kg) 30 mg/day and sertraline 25 mg/day were started as treatment. However, the spasms in the lower extremity muscles increased and began to progress to the hip and axial muscles. On the third day, needle electromyography (EMG) was applied to the lower extremity muscles, which showed continuous spontaneous discharges with neurogenic motor unit potentials (MUP) in bilateral lower extremity muscles. Findings were considered as neurogenic MUP changes and spontaneous continuous muscle contraction in lower extremity muscles. The patient was diagnosed as having SMS based on the clinical and EMG findings. On the fifth day, although baclofen (100 mg/day) was maximized, clonazepam was added to the treatment due to the lack of response. An initial dosage of clonazepam 0.5 mg/day was given and then increased by 0.5 mg every day. The maximum 2 mg/day clonazepam was given on the eighth day. The patient benefited from clonazepam. The spasms regressed and he started to walk again within a week and discharged from the hospital.

2. Discussion

SMS is not well recognized, even in the adult age group, and can be missed in the pediatric age group (Clardy et al., 2013). SMS is characterized by increased spasms and lumbar lordosis, especially in the axial muscles and lower extremity muscles. The spasms increase with tactile, acoustic or emotional stimuli (H. M. Meinck et al., 1994). Classic SMS (adult and child) occurs with stiffness and spasms in the lumbar region and lower extremities, but there are other forms of SMS such as isolated extremity or body rigidity, and progressive forms of encephalomyelitis have been described. Diagnosis is made through clinical findings (Clardy et al., 2013)(H.-M. Meinck, 2001). In our case, spasms
were increased when the patient was excited by healthcare staff entering the room, but this suggested psychical causes too. In our case, contractions first started in the lower extremity and progressed to axial muscles over time.

Although the etiology of SMS is not known, its association with autoimmune diseases shows that autoimmune mechanisms play a role. Gamma-aminobutyric acid (GABA) is a major inhibitory transmitter and GAD plays role in the synthesis of this transmitter. The role of autoimmunity against GAD is not yet understood because the same antibody is found in different autoimmune diseases (H.-M. Meinck, 2001). Anti-GAD positivity is frequently reported, especially in patients with T1DM, Hashimoto thyroiditis, Graves' disease, and myasthenia gravis (Clardy et al., 2013; Solimena et al., 1990; Baizabal-Carvallo, 2019; Dayalu & Teener, 2013). Anti-GAD positivity in patients with SMS is found in 80% of cases (Clardy et al., 2013). The anti-GAD level was found to be high in our patient who had T1DM. It is unclear why some autoimmune diseases with the same autoantibodies accompany SMS. However, different antibodies such as amphiphysin or glycine receptor, especially those described in adults with SMS, are also known (Clardy et al., 2013). It is reported that paraneoplastic SMS may occur in some adult malignancies such as lymphoma and lung cancer (Jun et al., 2015). Therefore, our patient was examined for malignancies and no sign of any malignancy was detected. It has been reported that baclofen, benzodiazepines (diazepam, clonazepam), steroids, plasmapheresis, intravenous immune globulin, and rituximab can be used in the treatment of SMS (Clardy et al., 2013; Rineer & Fretwell, 2017; Sarva, Deik, Ullah, & Severt, 2016). Our patient did not respond to baclofen, but significant improvement was observed after benzodiazepine use. Although autoantibody positivity was high, immune regulators were not necessary in our case.

In conclusion, SMS should be considered in the differential diagnosis in patients with muscle stiffness and increased spasms induced with stimulus. Care should be taken in terms of accompanying autoimmune diseases and malignancies in these patients.

Learning Points:

• Symptoms of SMS such as stiffness and spasms, especially in the lower extremities, cause gait difficulties.

• Autoimmune disorders such as type 1 diabetes mellitus can coexist with this syndrome. The relationship is not well understood.
• Treatment include benzodiazepines (e.g. diazepam, clonazepam) and immunotherapies (e.g. immunoglobulin, plasmapheresis, steroids. We obtained a good response to clonazepam.

Written informed consent was obtained from the parents for publication.
References
