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Facial onset sensory and motor neuronopathy syndrome — a rare variant of motor neurone disease

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To the Editors

A 65-year-old right-handed woman reported a four-year history of numbness of the left side of her face as well as in her left fingers and forearm. Then, slowly progressive weakness of the muscles of the left hand along with the left wrist flexors appeared, accompanied by global atrophy of the left upper limb muscles. In 2016, an ambulatory nerve conduction study (NCS) revealed selective axonal sensory neuropathy in the left upper limb, although a needle electromyography was not done. In 2017, magnetic resonance imaging (MRI) of the cervical spine showed discopathy at C5-C6 and C6-C7 levels. However, no improvement was noted after surgery, and the patient complained of stiffness and a pulling sensation of the neck, left upper limb and trunk. Due to progressive and mainly distal weakness of the left upper limb, the patient started dropping objects that she was holding, and was unable to lift her arm above the level of her head. In addition, she reported fatigue of the masseter muscles while eating and periodically less clear, slurred speech, without swallowing impairment. Paresthesia of the face, neck and left limb were unresponsive to pregabalin, gabapentin and antidepressants treatment.

In 2018, the patient was for the first time admitted to the Department of Neurology at the Medical University of Warsaw. Neurological examination revealed peripheral left facial nerve paresis, reduced pain sensation, temperature and touch in all divisions of the left trigeminal nerve, atrophy of the shoulder girdle muscles on the left and slightly of the left upper extremity, fasciculations in the left triceps muscle, slight weakness of the wrist flexors and extensors, hand and left hand fingers, and disturbed sense of pain, temperature and touch on the left upper limb. Anti-ganglioside, anti-myelin associated glycoprotein, anti-sulfatide, and anti-acetylcholine receptor antibodies were negative. Results of cerebrospinal fluid analysis, Lyme serology, lung functional tests, motor-evoked potentials for right thumb abductor and right short toe flexor muscles, brain and cervical spine MRI were not relevant. Brachial plexus MRI with contrast revealed asymmetrical atrophy of the rotator cuff, rhomboid, deltoid, serratus anterior, latissimus dorsi and biceps brachii muscles. There were no changes of the brachial plexus cords bilaterally (Fig. 1 A–B). NCS confirmed axonal sensory neuropathy in the left upper limb (Tab. 1). A needle electromyography recording from the muscles of the left upper limb showed significant denervation in the muscles supplied by C5-Th1 segments, with features of chronic reinnervation and marked reorganisation of motor units. Chronic reinnervation was also present in the left vastus lateralis muscle. The blink reflex indicated abnormal conduction at the level of the brainstem, as both sides R2 components had prolonged latency after left stimulation. Treatment with steroids (5 g methylprednisolone i.v. in October 2018) followed by intravenous immunoglobulins (105 g, 2.0 g/kg bw in July 2019 and two maintenance doses: 50 g in September 2019 and 60 g in October 2019) was introduced without any improvement. Finally, a diagnosis of facial onset sensory and motor neuronopathy syndrome (FOSMN) — a rare variant of motor neurone disease (MND) — was established, and riluzole 100mg/d was started.

In June 2020, the patient was readmitted to the Department of Neurology with severe progression of dysphagia. She was underweight, with a body mass index of 18 kg/m^2 (an unintentional loss of 22 kg since April 2019). Neurological

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Table 1. Nerve conduction studies

Motor nerves	Right		Left	
	Amp [mV]	CV [m/s]	Amp [mV]	CV [m/s]
Median	7.0	58.3	4.4	58.3
Ulnar	9.1	62.1	6.9	54.3
Radial	not done		4.5	70.0
Peroneal	1.4	41.3	0,3	54.8
Tibial	not done		13.9	52.7
Sensory nerves	**Right**		**Left**	
	Amp [uV]	CV [m/s]	Amp [uV]	CV [m/s]
Median	32	55.0	2.6	68.8
Ulnar	13	65.1	1.4	73.3
Radial	40	68.6	2.6	67.8
Peroneal superficial	not done		11	57.1
Sural	not done		8.8	59.1
Lateral antebrachial cutaneous	12	74.3	not done	

examination revealed absent corneal reflex on the left and weak on the right, no palatal and pharyngeal reflexes, dysarthria, peripheral left facial nerve paresis and impaired sense of touch on the left side of the face, weakness of the masseter muscles, weakness of axial muscles and head droop, left hemiparesis with global atrophy of the upper limb (the patient was able to abduct the arm to 10 degrees), and left side hypoaesthesia. She was able to walk alone, and could perform the squat manoeuvre with support. NCS results were comparable with the previous set, but needle electromyography showed progression of neurogenic changes in right biceps brachii, with reinnervation of muscles that previously were not affected (right first interosseus dorsalis and vastus lateralis). Percutaneous endoscopic gastrostomy (PEG) was performed.

Facial onset sensory and motor neuronopathy was first described by Vucic et al. in 2006 as a 'syringomyelia-like' condition [1]. It is characterised by facial onset sensory abnormalities which can spread to the scalp, neck, upper trunk and extremities, followed by lower motor neurone involvement. Bulbar symptoms, such as dysarthria and dysphagia, muscle weakness, cramps and fasciculations, can present later. It affects both genders, with a male-to-female ratio of 1.92:1, the mean age at onset is 54.0 years, and the mean disease duration is 8.9 years [2].

The diagnosis of FOSMN is based on medical history and clinical characteristics. Distinctive electrophysiological findings include reduced amplitude of sensory nerve action potentials. The blink reflex is usually abnormal, with either a delayed or absent R2 response. The pathogenesis of FOSMN syndrome is unclear. The possibility of an immunological mechanism was raised after the confirmation of auto-antibodies in a few patients. While some patients have partially responded to various immunotherapies, an immunological mechanism has been considered. There is the possibility of a potential pathophysiological link with amyotrophic lateral sclerosis (ALS), although there have been no familial cases of FOSMN to date [3]. Post-mortem evidence of sensory and motor neuronal degeneration within the trigeminal sensory nuclei, dorsal root ganglion, brainstem and spinal cord motor nuclei has been described [1]. FOSMN should be differentiated neurophysiologically from brachial plexus injury, as well as syringomyelia and other brainstem pathologies, trigeminal sensory neuropathy, motor neurone disease and Kennedy's Disease [4].

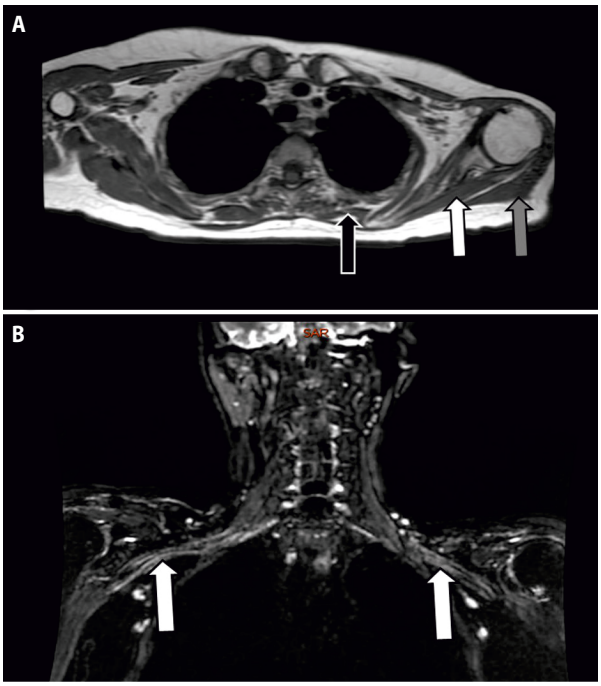

Figure 1. Magnetic resonance imaging. Atrophy of rotator cuff (white arrow), rhomboid (black arrow) and deltoid muscles (grey arrow) on left side on axial T1-weighted image (**A**), and similar thickness of brachial plexus cords bilaterally on STIR image (white arrows) (**B**)

It is worth discussing in more detail damage to the brachial plexus, which poses a serious diagnostic and therapeutic medical challenge. The basis for the diagnosis of brachial plexus function is a clinical examination and neurophysiological studies. It is characteristic that the amplitudes of sensory potentials decrease from five days post-damage, reaching their lowest values after 11 days. In the case of compound muscle action potentials (CMAP) amplitudes, abnormalities can occur 3-7 days after injury. Pathological changes in muscle function appear approximately three weeks after injury. The first NCS examinations should be carried out up to 3–4 weeks after the injury, as the Wallerian degeneration process will end. Incorrect parameters are shown first in motor, rather than sensory, potentials [5]. Maintaining the correct CMAP amplitude with accompanying muscle weakness at least seven days after injury suggests neuropraxia. If the difference in amplitude between the symptomatic and asymptomatic side is 50–75%, this indicates a moderate axons loss; more than 75% unregistered indicates axonotmesis or neurotmesis. F wave study should be performed only with reference to the long nerves. If the sensory nerve action potential (SNAP) is recorded, this suggests proximal damage to the sensory neuron. If the SNAP amplitude is reduced, or the sensory potential has not been recorded, this suggests a postganglionic plexus damage. In needle electromyography studies, denervation activity can be recorded 10–14 days after injury [5].

In conclusion, patients with FOSMN syndrome typically present with insidious, slowly evolving unilateral or bilateral numbness of the face. This is followed by bulbar and proximal (neck and arms) weakness. Despite the fact that FOSMN is mainly a lower motor neurone disease, some upper motor neurone signs make this condition an ALS mimic. Where appropriate, clinicians should consider percutaneous gastrostomy.

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