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CASE REPORT Open Access

Atypical motor neuron disease variant: facial-onset sensory and motor neuronopathy syndrome (FOSMN)

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Abstract

Background: Facial-onset sensory and motor neuronopathy (FOSMN) is a rare disease whose cardinal features are initial asymmetrical facial sensory deficits slowly evolving and followed by bulbar symptoms and spreading of sensory and motor deficits from face to scalp, neck, and extremities.

Case presentation: We described a 70-year-old man who presented with 6-month history of facial numbness on the left side, and gradual worsening of symptoms. Over several months, facial muscle weakness, dysarthria, and fasciculation had progressed. NCS, needle EMG and blink reflex responses suggested the diagnosis of FOSMN. The ganglioside panel (anti-GM1 and Gd1b) was positive. Considering the FOSMN autoimmune pathology hypothesis, IVIG treatment was given.

Conclusion: In this case, we aimed to highlight the key clinical aspects of FOSMN and how to differentiate it from motor neuron disease and bring FOSMN to the attention of neurologists as a recently recognized and distinctive disorder.

Keywords: FOSMN, Neuronopathy, Dysphagia, Motor neuron disease (MND)

Background

Motor neuron disease (MND) is a broad, heterogeneous group of neurological disorders that result from dysfunction of upper motor neurons (UMN), lower motor neurons (LMN), or both. The most common clinical phenotype is sporadic amyotrophic lateral sclerosis (ALS). However, a growing number of new, atypical, primary neurodegenerative forms of MND and ALS have now been described [1]. Little is known about the etiology and pathophysiological mechanisms of facial-onset sensory and motor neuronopathy (FOSMN) syndrome, a rare primary neurodegenerative disorder. FOSMN syndrome is a rare, slowly progressive disease of the lower motor neurons with sensory impairment that mainly affects the face, bulbar region, and upper limbs.

In this report, we present this case to emphasize that knowledge of a rare disease such as FOSMN syndrome is essential for differential diagnosis.

Case presentation

A 70-year-old man reported a 2-year history of numbness on the left side of his face. Over time, complaints of speech disorder and difficulty in chewing were added. Then within 6 months, slowly progressive weakness of the left-hand distal muscle appeared to attend global atrophy of the left upper limb muscles. Due to progressive distal weakness of the left upper limb, the patient began to drop objects and could not raise his arm above head level. Soon after, he had weakness in his left leg and difficulty walking. Then the patient was admitted to the neurology department of our hospital. Neurologic examination revealed weakness in bilateral masseter muscles, more

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prominent on the left, and decreased sensation of touch, temperature, pain in all three branches of the trigeminal nerve and absent corneal reflexes on the left side. Speech was dysarthric, palatal and pharyngeal reflexes were absent. Atrophy and fasciculations were observed on the tongue. Light touch on the limbs, vibration, and proprioception were intact. Strength testing revealed weakness in the neck flexors, left arm, and proximal leg muscles. Muscle stretch reflexes and coordination were normal.

In nerve conduction study (NCS), by Natus Medical Incorporated, Pleasanton, USA, sensory nerve action potential (SNAP) amplitude could not be obtained in the left median, ulnar, and sural nerves. SNAP amplitude is low in the right median, ulnar, and sural nerves. Compound muscle action potential (CMAP) amplitudes of the left ulnar, peroneal, and bilateral tibial nerves are small, conduction velocity is normal, and conduction block is not observed. The bilateral median nerve, right ulnar nerve, and peroneal nerve are normal. The latency of the bilateral tibial F response is prolonged. Fibrillation, PKD and fasciculations were observed in the tongue, bilateral lower and upper limb muscles. Motor unit potential (MUP) loss, prolonged duration, and high amplitude of MUPs were noted. It has been observed that the interference pattern decreases.

The blink reflex abnormalities were characterized by prolonged R1 and R2 responses. Importantly, abnormal blink reflexes were observed in the context of normal facial nerve CMAP responses, suggesting dysfunction in the afferent limb of the blink reflex pathway.

The patient was diagnosed with facial-onset sensory and motor neuronopathy syndrome (FOSMN), a rare variant of motor neuron disease (MND).

The patient was planned for differential diagnosis, but the patient left because he lived in France. When the patient was called for control 7 months later, we learned that the patient died. The investigations of the patient made in France were revised, Anti-acetylcholine receptor antibodies were negative. The results of the cerebrospinal fluid analysis were normal. Lyme, HIV, syphilis serology were negative. Vasculitis and tumor markers were negative. The ganglioside panel (anti-GM1 and Gd1b) was positive. Magnetic resonance imaging (MRI) of the cervical spine was normal. There was mild diffuse cortical atrophy on cranial MRI. There was no significant finding in the left deltoid muscle biopsy. In the left sural nerve biopsy, there were findings consistent with the Wallerian degeneration of the nerve. Repetitive nerve stimulation (RNS) from the nasal muscle was normal.

The patient, whose ganglioside panel was also positive, was given intravenous immunoglobulins at a dose of 2 g/kg in the autoimmune pathology hypothesis and riluzole 100 mg/day was started. The patient partially benefited from the treatment. But 7 months later, his condition worsened, dysphagia required percutaneous endoscopic gastrostomy (PEG) tube placement, and the patient died after pneumonia.

Discussion

Facial-onset sensory and motor neuronopathy was first described by Vucic and colleagues in 2006 as a 'syringomyelia-like' condition [2]. FOSMN syndrome is characterized by usually unilateral facial sensory deficits and paresthesia, then slowly spreads to the scalp, neck, upper trunk, and upper extremities. Later, lower motor neuron signs such as fasciculation, cramps, atrophy, and weakness arise in the affected areas. Patients develop dysphagia and weakness in the distal muscle. It is a rare and slowly progressive neurological disorder. Although neurodegenerative and autoimmune mechanisms have been suggested in the pathophysiological process underlying FOSMN syndrome, they have not been fully elucidated [3]. Autopsy cases of FOSMN syndrome, finding loss of neurons in trigeminal sensory nuclei, facial nerve nuclei, hypoglossal nerve nuclei, anterior horns, and dorsal root ganglia (DRG), suggest that the disease is a neurodegenerative process. However, positive antiganglioside antibodies and partial response to immunotherapies with intravenous immunoglobulin (IVIg) infusion and plasma exchange suggest the autoimmune pathology hypothesis.

At present, there are no formal diagnostic criteria for FOSMN. FOSMN is based on the medical history and clinical features and diagnosis is made by excluding other diseases. Extensive genetic testing has been performed in various cases, which included ALS genes (C9orf72, FUS, SOD1, TARDBP), oculopharyngeal muscular dystrophy (PABPN1), Kennedy disease (AR), panels for spinocerebellar ataxia as well as whole-exome sequencing. The genetic testing has not given any insight so far [4]. Low head syndrome and early bulbar involvement may have more aggressive clinical progression and worse prognosis [2].

In electrophysiological findings, include reduced the amplitude of sensory nerve action potentials. The blink reflex is usually abnormal and the R2 response is prolonged or absent.

FOSMN must be differentiated from brachial plexus injury, syringomyelia, and other brainstem pathologies, trigeminal sensory neuropathy, motor neuron disease, and Kennedy's disease [5]. Idiopathic trigeminal neuropathy or central lesion should be considered in patients with isolated sensory findings. Cranial MRI was normal in our patient. ALS should be considered when bulbar weakness is a progressive or dominant feature, especially since FOSMN has been suggested to be an atypical form of ALS. Kennedy's disease was also ruled out because

of its symmetrical onset. Brachial plexopathy and syringomyelia were excluded because cervical MRI did not explain current findings.

Conclusion

FOSMN is an extremely rare neurological disease that begins with sensory impairment spreading along with a craniocaudal distribution, followed by motor manifestations. It should be differentiated from other diseases with similar complaints. More cases and research should be conducted to establish diagnostic criteria for FOSMN and to gain more information.

Abbreviations

FOSMN: Facial-onset sensory and motor neuronopathy syndrome; MND: Motor neuron disease; UMN: Upper motor neurons; LMN: Lower motor neurons; ALS: Amyotrophic lateral sclerosis; NCS: Nerve conduction study; SNAP: Sensory nerve action potential; CMAP: Compound muscle action potential; MUP: Motor unit potential; MRI: Magnetic resonance imaging; RNS: Repetitive nerve stimulation; PEG: Percutaneous endoscopic gastrostomy; DRG: Dorsal root ganglia; IVIg: Intravenous immunoglobulin.

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Author contributions

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Competing interests

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