Facial onset sensory and motor neuronopathy syndrome (FOSMN): first description of the neuropsychological profile

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Background and Purpose

Facial onset sensory and motor neuronopathy (FOSMN) syndrome has been described in 2006 [1], and few cases are reported in literature. The aetiology is still unknown, but some authors tend to consider the pathology as neurodegenerative disease, on the basis of the TDP-43-positive neuronal inclusions [2-3]. FOSMN shares many common clinical features with amyotrophic lateral sclerosis (ALS), nevertheless some differences are present. In particular, FOSMN is characterized by sensory symptoms and slow progression. Furthermore, subclinical or clinically evident frontal dysfunction is often part of the ALS spectrum, whilst no neuropsychological data have been collected in FOSMN patients. We aim to describe the clinical and instrumental characteristics of our patient, in order to expand the spectrum of FOSMN syndrome.

Case Report

In May 2016, a 70-year-old Caucasian man presented with a 10-year history of perioral numbness. Over the next years, he developed paraesthesiae initially involving all the trigeminal divisions bilaterally, and gradually progressing to upper and lower limbs. Meanwhile, he manifested dysphagia with weight loss of 8 Kg, bilateral global upper limb weakness, and muscle twitching.

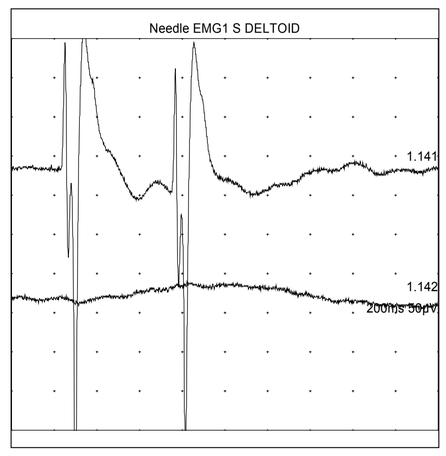
The neurological examination showed a reduced pinprick sensation in all divisions of the trigeminal nerve bilaterally, and absence of corneal reflexes. There was global wasting of limb muscles bilaterally, with diffuse fasciculations. His neck flexion was weak, as were proximal arm muscles bilaterally. Reflexes were globally and symmetrically reduced, with bilateral flexor plantar responses. There was a reduced vibratory sensation at the lower limbs.

Negative investigations included brain and cervical imaging exams, serum electrophoresis, autoantibody screen, anti-GM1 IgM antibodies, neoplastic markers, onconeural antibodies, CSF analysis. EMG/NCS showed widespread neurogenic changes and fasciculations in spinal (A, B) and bulbar muscles examined, and reduced sensory and motor nerve action potentials in the limbs. The blink reflex was abnormal, with delayed R2 response bilaterally (C, D). Motor evoked magnetic potential examination was normal. The jaw reflex was absent bilaterally (E, F). Neuropsychological profile was normal, except for reduced verbal short term memory (G). Genetic analysis (SOD1, C9orf72, FUS, TDP-43) is ongoing. No treatment was started.

Conclusions

Neuropsychological functions have been never studied in FOSMN patients. Our patient underwent the neuropsychological assessment in use at our Institute for ALS patients. The cognitive profile was substantially normal, despite the long time elapsed since the onset of FOSMN symptoms. This represents the first report of cognitive screening in FOSMN patients. While a cognitive follow-up of our patient is advisable, and further data must be collected from a wider cohort, the relative sparing of cognitive function in our case may be interesting in view of the possible inclusion of FOSMN in the group of TDP-43 proteinopathies.

A



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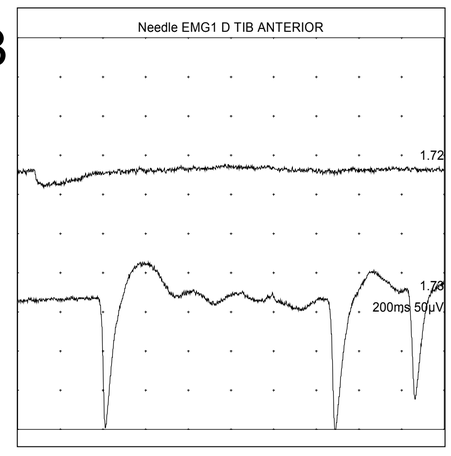

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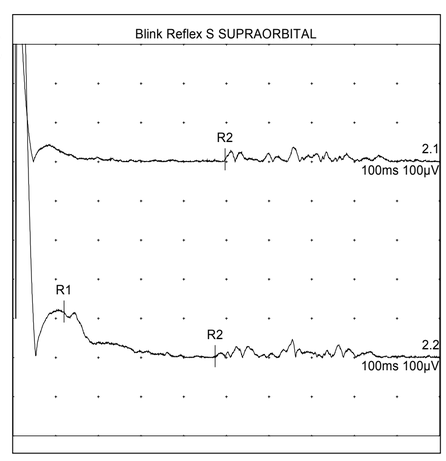

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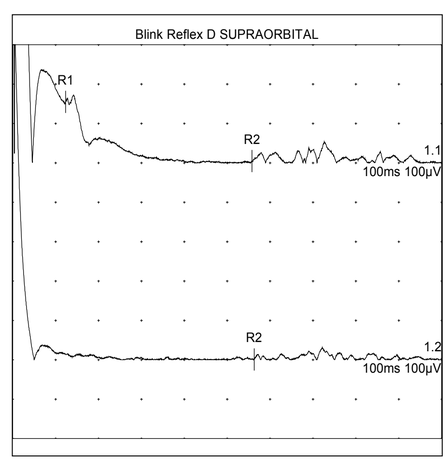

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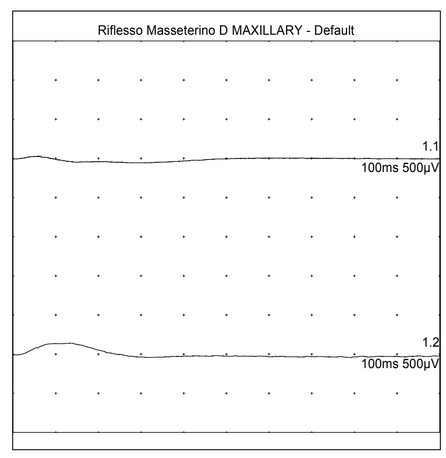

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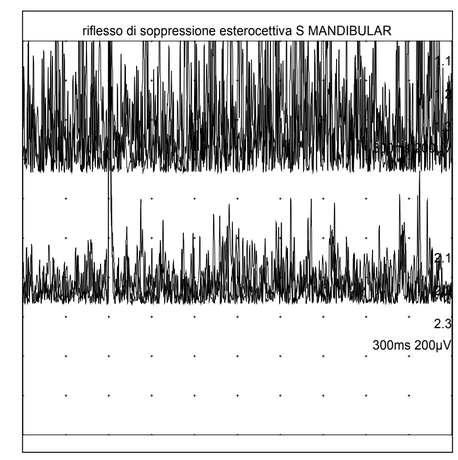

G

	Punt. Grezzo	Punt. Corretto	Punt. Equivalente	Cut-off
Mini Mental State Examination	29	26.7	*Nella norma*	< 24
Funzioni Mnestiche				
Span Verbale	3	2.75	**0**	< 3
Digit Span	5	5	4	< 3.75
Test di Corsi	5	5	4	< 3.75
15 parole di Rey R.I.	34	37.1	3	< 28.53
15 parole di Rey R.D.	6	7.2	3	< 4.69
Breve Racconto				
- Rievocazione immediata	7.7	7.9	4	< 3.09
- Rievocazione differita	8	8	4	< 2.38
Figura di Rey (recall)	18	18.5	4	≤ 9.46
Funzioni Logico Esecutive				
Matrici Colorate di Raven 1947	30	30.1	4	<18.96
Frontal Assessment Battery (FAB)	15	14.9	2	≤ 13.4
Funzioni Attentive				
Trail Making Test A	76	60	2	> 93
Trail Making Test B	128	81	4	> 282
Matrici Attentive	54	51.75	4	< 31
Stroop test				
Interferenza Tempo	15.5	8.25	4	≥ 36.92
Interferenza Errori	0	0	4	≥ 4.24
Linguaggio				
Fluenza per categorie fonologiche (FAS)	28	25.6	3	< 17.35
Fluenza per categorie semantiche	42	42	4	< 25
Denominazione Visiva	64		*Norma*	< 56
Abilità visuospaziali e prassiche				
Figura di Rey copia	36	36	4	≤ 28.87
Cognizione sociale				
Test di Teoria della Mente	12		*Norma*	< 11
Test delle Situazioni Sociali:				
- comportamenti normativi	14		*Norma*	< 12
- violazioni	22		*Norma*	< 21
- livello di violazione	53		*Norma*	< 24

References

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