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Moebius syndrome in association with hypogonadotrophic hypogonadism and focal motor demyelinating peripheral neuropathy with persistent conduction blocks

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Introduction

Congenital, non-progressive facial paralysis and a restricted abduction of one or both eyes are symptoms of Moebius Syndrome (MBS) [1]. Peripheral neuropathy, Kallmann syndrome, intellectual disability, and social impairment are also evident in a subgroup of people with inherited congenital facial nerve atrophy and oculomotor nerve palsy [2]. Moebius syndrome with hypogonadotropic hypogonadism is associated with progressive peripheral neuropathy and is often axonal and occasionally demyelinating [3-6]. Electromyography in this uncommon clinical type shows high amplitude motor unit potentials with reduced recruitment in the leg muscles when an axonopathy is present. Electroneurography has shown that motor and sensory nerves exhibit lower amplitudes, different axonal properties, and slightly slower conduction velocities [2-7]. We describe, for the first time, a case of a patient with MBS along with chronic multifocal demyelinating motor neuropathy and persistent conduction blocks due to hypogonadotropic hypogonadism.

Case history

A seventeen-year-old male subject who had previously been diagnosed with MBS for congenital paresis of both VII cranial nerves and limited eye abduction displayed growing ankle dorsiflexion weakness during childhood, which was more pronounced on the left side. There were no hints that the neuropathy was inherited. Additionally, the patient had anosmia and hypogonadotropic hypogonadism (Kallmann syndrome). An equinovarus deformity of the foot and ankle as well as bilateral

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Table 1: Neurophysiological parameters in lower limbs motor conduction studies.

Nerve	Muscle	Latency	Amplitude	Rel Amp	Segment	Distance	Lat Diff	Velocity	Rel Vel	Area 1-5
		ms	mV	%		mm	ms	m/s	%	mVms
Left Peroneal - EDB										
Ankle	EDB	5.23	0.9	100	Ankle - EDB					5.8
Fibular Head	EDB	12.46	1.3	135	Fibular Head - Ankle	300	7.23	41	100	6.8
Popliteal fossa	EDB	17.29	0.3	20.5	Popliteal fossa - Fibular Head	120	4.83	25	59.8	
Right Peroneal - EDB										
Ankle	EDB	4.63	3.3	100	Ankle - EDB					13.5
Fibular Head	EDB	10.98	3.8	115	Fibular Head - Ankle	330	6.35	52	100	18.5
Popliteal fossa	EDB	12.90	3.5	92.1	Popliteal fossa - Fibular Head	80	1.92	42	80.4	15.4
Left Tibialis - AH										
Ankle	AH	3.21	20.6	100	Ankle - AH	80				67.5
Popliteal fossa	AH	11.92	17.9	87	Popliteal fossa - Ankle	400	8.71	46	100	66.5
Right Tibialis - AH										
Ankle	AH	4.71	20.1	100	Ankle - AH	80				70.5
Popliteal fossa	AH	12.85	18.6	92.7	Popliteal fossa - Ankle	350	8.15	43	100	64.6

drop feet with steppage gait were discovered during a physical examination before admission to the neurorehabilitation ambulatory unit. In the anterior-lateral compartments of the legs, there were signs of muscle hypotony and partial atrophy, which were more obvious on the left side. Before the neurorehabilitation examination, untrained doctors working in a general medical facility treated stiffness in the left calf muscles with an ineffective injection of botulinum toxin A (left Gastrocnemius caput medialis and Tibialis Posterior muscles). After this therapy, the patient reported worsening muscular trophism throughout the entire left leg and no improvement in their motor impairments. The subject displayed no abnormalities in breathing or swallowing and showed no evidence of upper motor neuron involvement.

Electrodiagnostic studies

The following common approaches are part of the neurophysiological protocol used in the electrodiagnostic examinations carried out in the Neurorehabilitation Unit EMG Lab of General Hospital - University of Padua. Blink reflex, motor conduction studies of the seventh cranial nerve, medial, ulnar, peroneal, and tibialis posterior motor nerves with the registration of F waves, medial, ulnar, and sural sensory nerve conduction studies, and needle electromyography of the face (Orbicularis oculi and oris muscles), limbs, and heart are all examples of such studies (Deltoid, Abductor pollicis brevis, first dorsal Interosseus, Tibialis anterior, Peroneus longus, Gastrocnemius caput medialis, Tibialis Posterior, Extensor digitorum brevis). Exams verified earlier findings that there was no motor unit activation in the face muscles' EMG or absence of blink and VII cranial nerve motor nerve responses. Chronic neurogenic EMG findings of the bilateral Tibialis anterior and Peroneus longus were found by limb needle electromyography. Additionally, the left Gastrocnemius caput medialis and the left Tibialis Posterior displayed mild neurogenics (previously injected with botulinum toxin A). Electroneurography showed a small drop in motor conduction velocity in the right Tibialis posterior nerve and a bilateral partial conduction block in the Peroneal nerves at the fibular head (cMAP amplitude and area decline) (See Table 1 and Figures 1, 2).

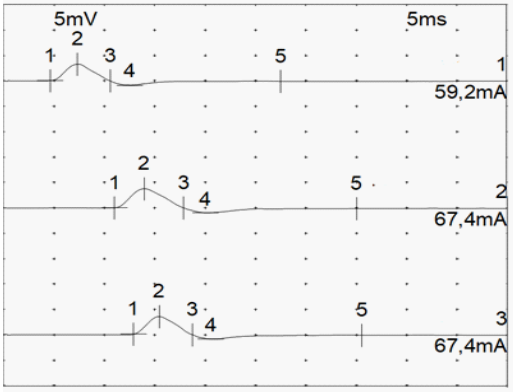

Figure 1: Partial conduction block in right peroneal nerve at fibular head (cMAP amplitude and area decay).

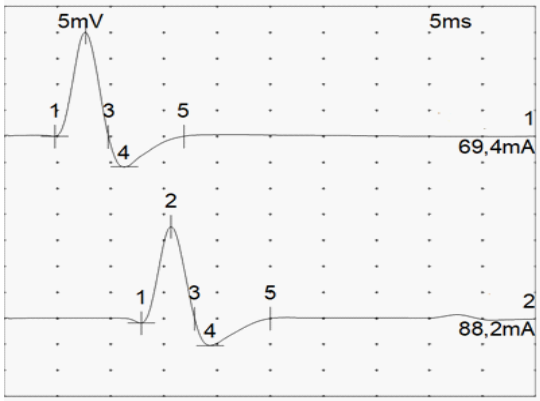

Figure 2: Slight reduction of motor conduction velocity in right tibialis posterior nerve.

Except for a slightly decreased number of F-Waves induced by stimulation of lower limb motor neurons, all other neurophysiologic parameters in needle EMG, Motor, and Sensory conduction investigations were within normal ranges. After that, during a follow-up appointment, a dorso-lumbar spine MRI ruled out disc herniation or foraminal stenosis at that level crushing the L4 or L5 nerve roots, and a knee ultrasound exam ruled out a common source of peroneal nerve compression at the fibular head.

Discussion

Along with facial and oculomotor congenital paresis [1], the MBS also had bilateral drop feet and a steppage gait. After receiving an untimely injection of botulinum toxin in the left calf muscles during childhood, the patient's motor skills for walking were delayed and slowly got worse [8]. Electroneurography of the lower limbs revealed focal motor peripheral neuropathy with persisting conduction blockages of the peroneal nerve at the fibular head in this clinical version of Moebius syndrome. In muscles of the anterior-lateral compartments of the legs, needle EMG revealed high amplitude motor unit potentials with reduced recruitment, which is consistent with the existence of dorsiflexor muscle abnormalities. Other myotomes not innervated by the Peroneal nerve exhibit demyelinating aspects of the neuropathy with intact sensitive responses and normal needle EMG findings, according to electrodiagnostic research [2-7]. Only one research has described the co-existence of MBS and focal motor peripheral neuropathy with chronic conduction blocks [3-6]. The findings of this study also aid in the planning of rehabilitation therapy for the syndrome, which includes functional electrical stimulation of the leg muscles in addition to traditional physical therapy and AFO bracing to increase foot dorsiflexion and avoid leg muscle atrophy [8].

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