

## CASE REPORT

### **A Rare Genetic Disorder Characterized by Facial Paralysis: Mobius Syndrome**

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We report an extremely rare disorder characterized by facial paralysis, Mobius Syndrome. Also clinical, radiological and electrophysiological findings are described, and management aspects are discussed.

A 5-year-old boy with Mobius syndrome is presented. He applied to our center because of lacking facial expressions; inability to smile; eye sensitivity; hearing problems, speech difficulties and pes plano valgus deformity. Magnetic resonance imaging (MRI) of the brain showed a slight dilatation of the left ventricle more than expected. In electromyographic (EMG) evaluation, the amplitude of the muscle potentials evoked by facial nerve stimulation was reduced on left side. Auditory brainstem evoked responses demonstrated waveforms I-V were abnormal. The amplitude wave V was less than wave I on left. A normal 46 XY karyotype was present. The other clinical - metabolic screening tests were normal.

Although it appears to be genetic, its precise cause remains unknown and the medical literature presents conflicting theories. Whether the nerve, brainstem, or muscle aplasia is the primary event has not been established. Abnormal blink reflexes and loss of motor activity with EMG showed a defect at the facial nuclear level. Masking of supranuclear lesions by nuclear defects should also be kept on mind. Abnormal ABR responses may suggest inclusion of eighth cranial nerve.

Mobius Syndrome is a rare disorder characterized by permanent facial paralysis. Only about 300 cases have been described in the English-language literature. The sixth and seventh cranial nerves are not fully developed. Other cranial nerves can also be affected. Although it appears to be genetic, its precise cause remains unknown and the medical literature presents conflicting theories.<sup>[1,2]</sup>

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### CASE REPORT

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A five-year-old boy admitted to ENT Department having asymmetry and facial function loss; eye sensitivity; hearing problems; and speech difficulties. Generalized hypotonia associated with facial diplegia, weak sucking and swallowing reflexes were noted during his delivery at birth. He was able to hold his head in five months and able to sit when he was ten months old.

He was unable to blink his left eye, smile, or frown during the examination. He had facial diplegia and convergent strabismus secondary to paralysis of the facial and abducens nerves on the left side. Pupillary reflexes were normal, but corneal reflexes were impaired on left side. (Figure 1a,b)



**Figure-1a:** Face at rest    **Figure-1b:** Face during crying

He was able to speak but experienced difficulties especially with closed mouth sounds.

Additionally there was a weakness present in his lower extremity and pes plano valgus deformity (Fig 2). A normal 46 XY karyotype was present. The other clinical - metabolic screening tests were normal.



**Figure 2:** Pes Plano Valgus deformity

Temporal bone computed tomography was unremarkable. Cranial magnetic resonance imaging (MRI) showed the slight dilatation of the left ventricle. (Fig3).



**Figure-3:** Cranial MRI

The electrophysiological tests were resumed as: Facial nerve conduction velocity was calculated by evoked EMGs of the mentalis muscle. It was reduced on the left side. The facial nerve evoked potential amplitudes were reduced on left side. Blink reflex responses were prolonged in terms of latency. The amplitude ratio of waves I and V was reversed on the left side.

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### DISCUSSION

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Von Graefe described Congenital facial and abducens palsy in 1880. Paul Julius Möebius drew attention to patients with congenital nonprogressive bilateral facial and abducens palsy in 1888. Because of these

contributions, Möbius is now the eponym used to describe the syndrome. In 1939, Henderson broadened the definition and included cases with congenital unilateral facial palsy<sup>[1,2]</sup>

Möbius syndrome is a rare disorder and the prevalence is reported as 0.002-0.0002% of births. Most cases are sporadic, but autosomal-dominant, recessive and X-linked patterns of inheritance have been described. Although it appears to be genetic, its precise cause remains unknown and the medical literature presents conflicting theories. Whether the nerve, brainstem, or muscle aplasia is the primarily affected, has not been established. Möbius believed that the condition was degenerative or toxic in origin and that it involved the nuclei of the affected nerves. Some authors suggest that the underlying problem is an inherited congenital hypoplasia or agenesis of the cranial nerve nuclei<sup>[2,3]</sup>. Approximately 2% of cases appear to have a genetic basis. In addition, theories of vascular etiologies of the syndrome have many proponents. One such theory involves disruption of flow in the basilar artery or premature regression of the primitive trigeminal arteries. A second vascular theory is a disruption of the subclavian artery supply that involves interruption of the embryonic blood supply. Cocaine and prostaglandin abuse is occasionally associated with the occurrence of this syndrome<sup>[4]</sup>.

Lower cranial nerves may be involved including 6th to 12th. Oculomotor and trochlear nerves are rarely affected. The facial nerve is involved in all cases and usually bilateral. Abducens nerve functions are abnormal 75% of cases. Hypoglossal nerve lesions are found in only a minority of cases. The ocular muscles are always involved when the tongue is affected<sup>[2,3]</sup>. The patient in this case had unilateral facial and abducens nerve paralysis.

In cases with Möbius Syndrome, conductive (due to serous otitis media) or sensorineural hearing loss may be noted. Speech problems are reported in 76-90% of the patients. Their speech is usually difficult to understand because of the patient's inability to close his or her lips and make labial articulations. In some cases, speech impairment may be severe. Musculoskeletal abnormalities occur in one third of the patients. These abnormalities may include equinovarus, brachydactyly, syndactyly, congenital amputations, arthrogryposis, smallness of

limbs, and occasionally hypoplasia or absence of the pectoralis major muscles (Poland anomaly). Bilateral club foot formation occurs in almost one third of the patients<sup>[1,5]</sup>. Both hearing-speech problems and pes plano valgus deformity existed in our patient.

There are no specific laboratory study findings for Möbius syndrome. Reports of radiological and electrophysiological findings are limited. Some reports describe nonspecific calcifications of the basal ganglia on the brain computerized tomography and the brainstem may appear hypoplastic with straightening of the fourth ventricular floor on the MRI studies<sup>[6]</sup>.

Timing of the injury to the facial nerve may be important, and electromyography (EMG) can assist in this regard. Jaradeh et al. suggested the primary cause of neural involvement was prenatal brainstem damage involving the motor nuclei and their internuclear connections<sup>[7,8]</sup>. Cattaneo et al reported two different defined phenotypes based on their EMG data. The authors postulated that the first group had rhombencephalic maldevelopment, while the second group seemed to have acquired a nervous system injury during intrauterine life<sup>[9]</sup>. The patient's abnormal blink reflexes, loss of facial nerve responses and absent motor activity on needle electromyography indicated a defect at the facial nuclear level. However, the nuclear defect might mask an additional supranuclear defect. Abnormal ABR responses suggested the cochlear nerve could also be involved.

Physical and speech therapy may improve motor skills and coordination, and help to better control speaking and eating abilities. If necessary hearing aid and ventilation tube insertions may be performed. Both speech and physiotherapy were designed for our patient. Ophthalmologists recommended delaying surgery for strabismus as the condition frequently improves with age. Secondary to incomplete eyelid closure, treatment for corneal ulcerations or abrasions may be required.

Möbius Syndrome is a rare disorder characterized by permanent facial paralysis. The sixth and seventh cranial nerves are not fully developed. The syndrome can also be associated with abnormalities in other parts of the body. In this case, a 5-year-old boy with Möbius syndrome is presented, had unilateral facial and abducens nerve paralysis and Pes Plano Valgus deformity.

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