Case report

Moebius Syndrome: ACase Report on an Uncommon Congenital Syndrome

ABSTRACT

Moebius syndrome (MS) is a rare congenital disorder characterized by facial paralysis and limitations in eye movement due to cranial nerve involvement, with potential additional craniofacial and limb abnormalities. We present the case of a two-year-old girl with MS who presented with watering in both eyes. Clinical examination revealed left-sided facial paralysis, convergent strabismus, and limb malformation. The etiology of MS remains uncertain, with both genetic and environmental factors implicated. Diagnosis relies on clinical criteria, with supportive care and multidisciplinary management essential for optimizing outcomes. Early rehabilitation is crucial, and interventions may include surgical correction and therapies addressing associated issues. MS poses significant challenges, necessitating careful evaluation and management strategies to improve patient outcomes and quality of life.

Keywords: Moebius syndrome (MS); Congenital; Facial paralysis; Cranial nerve palsies

1. INTRODUCTION

Moebius syndrome (MS) is a rare congenital disorder charecterized by either unilateral or bilateral nonprogressive congenital facial paralysis (involving the VII cranial nerve) accompanied by limitations in eye movement (involving the VI cranial nerve). Additionally, it may present with other cranial nerve impairments, abnormalities in the orofacial region, and limb defects [1].It was initially recognized by A. Von Graefe, who documented cases involving congenital VI and VII nerve palsies. However, it was in 1888 that Paul Julius Moebius, a German neurologist, received recognition for his insightful reports on the syndrome's clinical features [2].

While the involvement of the VI (abducens) and VII (facial) cranial nerves is central to the diagnosis of MS, the clinical spectrum can be variable. In some instances, additional cranial nerve palsies may be present, including the XII (hypoglossal) nerve responsible for tongue movement and the III (oculomotor) nerve critical for eye movements. Involvement of other cranial nerves, such as the trigeminal (V), vagus (X), and spinal accessory (XI), has also been documented [3]. Furthermore, MS can be associated with various malformations, most notably limb abnormalities like Poland syndrome (underdeveloped pectoral muscles and a specific palmar crease) or other hand and finger anomalies [4].We present the case of an infant with MS who presentedto our hospital with watering in both eye difficulties.

2. PRESENTATION OF CASE

We report a two-year-old girl, the youngest of three siblings, from a nonconsanguineous marriage. The pregnancy was uneventful, and regular checkups confirmed healthy development. The pregnancy was carried to term without any incidents, and there was no history of exposure to toxins or medications, including abortive products. The delivery was medically assisted by vaginal delivery with no history of trauma or forceps use, and the child adapted well to extrauterine life with an imprecise birth weight.

The patient was brought to our hospital for watering in both eyes at the age of 20 months. The mask-like face was, however, noticed by the mother when she was six months old. The child had, up to this point, delayed psychomotor development, with walking acquired at 12 months and speech at 18 months. On clinical examination, the patient showed left-sided facial paralysis with deviation of the eye and lip, and effacement of the nasolabial fold was noted, which was more accentuated during crying. Mild convergent strabismus of the same was also noted. Furthermore, the child had limb malformation, but after receiving treatment, she became fine.

Comment [ГК1]: Author does not mention what treatment protocol was applied to the patient ?

Fig. 1. Patient with left sided facial palsy

3. DISCUSSION

Moebius Syndrome is a rare and complex disorder characterized by the involvement of typically two or more cranial nerves, commonly the 7th and 6th, potentially accompanied by additional limb or craniofacial anomalies. The incidence of MS is roughly 1 per 250,000 live births, with no significant gender disparity. While sporadic cases are predominant, familial occurrences, constituting around 2% of all affected individuals, have also been observed [1].

The etiology and pathogenesis of Moebius syndrome remain uncertain and subject to debate. Since its initial description by von Graefe and Moebius, there has been ongoing discussion regarding whether the syndrome is primarily genetic or influenced by environmental factors, although it is believed to involve both. While the specific gene

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associated with Moebius syndrome has yet to be identified, it is speculated to be linked to alterations in certain regions of chromosomes 3, 10, or 13. Certain medications are considered potential risk factors, including exposure to toxic substances, impaired development of the rhombencephalon's vascular system, and ischemic or hypoxic events after the fifth week of gestation.1 Misoprostol, a synthetic prostaglandin E1 analogue, is frequently cited as a substance implicated in Moebius syndrome [5].

Physiopathological findings have led to the proposal of four categories elucidating MS manifestations and imaging results. The first category involves underdevelopment of cranial nerve nuclei due to congenital abnormalities. Our case aligns with the second category, characterized by neuronal degeneration and loss resulting from peripheral facial nerve defects. The third category exhibits reduced neurons, along with degeneration, gliosis, focal necrosis, and brainstem nuclei calcifications due to inadequate blood supply or infection. The last group entails primary myopathic changes without CNS or cranial nerve involvement [6].

Clinically, facial nerve paralysis, whether unilateral or bilateral, presents with a mask-like appearance, accompanied by drooling, and a lack of smile, which may appear unattractive or even grimacing due to involvement of specific subcutaneous muscle bundles [7]. Involvement of the sixth cranial nerve leads to restricted eye movement horizontally, an inability to perform saccadic movements, and may result in optic nystagmus. Although vertical gaze paralysis is uncommon in Moebius syndrome, it can occur in up to 25% of cases involving the third pair of cranial nerves [8]. A study conducted in Mexico focusing on ophthalmological involvement in Moebius syndrome revealed a variety of manifestations, including limited ocular sequestration and facial paralysis in all cases, along with esotropia, epicanthus, and entropion. Additionally, a history of abortion-inducing agents used by the mother during the first trimester of pregnancy was noted in a significant percentage of cases [9]. Moebius syndrome can also affect other cranial nerves, such as the hypoglossal nerve, resulting in language and speech disorders, as well as drooling; the trigeminal nerve, the oculomotor nerve, and less commonly, the glossopharyngeal nerve. It may also be associated with various malformation syndromes, including micrognathia, cleft palate, heart malformations, urinary and orthopedic anomalies, and digital malformations of the upper limbs [3].

Moebius syndrome can be detected during infancy, characterized by symptoms such as difficulty in sucking due to inadequate lip closure, lack of facial expressions, incomplete eyelid closure during sleep, excessive salivation, and inward deviation of the eyes (esotropia). Newborns with Moebius syndrome commonly face challenges with swallowing and breathing, with approximately 90% exhibiting craniofacial malformations that increase the risk of airway blockage [3].

The diagnosis of Moebius syndrome relies primarily on clinical criteria, as there are no specific laboratory tests associated with the condition. Radiological imaging, while less useful for diagnosis, may occasionally reveal calcification at the site of dorsal ischemic necrosis of the pons on head computed tomography (CT) scans or brainstem hypoplasia on magnetic resonance imaging (MRI) [10]. Abramson et al. introduced the CLUFT scoring system (Cranial Nerves, Lower extremity, Upper extremity, Facial structural anomalies, Thorax) for classification and assessment of dysmorphic features [11]. This scoring system serves multiple purposes, including categorizing anomalies by location and severity, facilitating comparison between patient groups, analyzing surgical management outcomes, and providing insights into pathogenesis.

Management of Moebius syndrome requires a multidisciplinary approach, including respiratory support, possible tracheotomy during the neonatal period, and multiple surgeries such as facial reconstruction, correction of strabismus, or orthopedic interventions [12]. Early rehabilitation is crucial for optimizing recovery and improving outcomes and quality of life. At an advanced stage, Moebius syndrome can have significant psychological and social impacts, with individuals expressing challenges in social interactions and being frequently misunderstood due to limited facial expressions.

4. CONCLUSION

Diagnosing MS requires consideration of both ophthalmological and general signs, and it can be challenging in atypical cases. Management of these patients necessitates a multidisciplinary team approach. Management focuses on supportive care while parental education is crucial. Ocular protection is essential to prevent complications. Additional interventions may include physiotherapy, occupational therapy, and speech therapy to address associated issues.

CONSENT

As per international standard, parental written consent has been collected and preserved by the author(s).

ETHICAL APPROVAL

As per international standard or university standard written ethical approval has been collected and preserved by the author(s).

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