Moebius Syndrome: A Rare Case Report

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ABSTRACT

Moebius syndrome is an extremely rare disorder. Only approximately few hundred cases have been reported in literature. In a nationwide survey reported in 2003, the prevalence of this syndrome was at least 0.002% of births for the years 1996 to 1998. The definition and diagnostic criteria for Moebius syndrome vary among authors. In most studies, it is defined as congenital facial weakness combined with abnormal ocular abduction. The list of signs and symptoms mentioned in various sources for Moebius syndrome includes more than 20 peculiar features. Here, we report an interesting case of Moebius syndrome in a 12-year-old male patient with all the important peculiar signs and symptoms.

Keywords: Moebius syndrome, Congenital facial diplegia, Congenital oculofacial paralysis, Loss of function of motor cranial nerves.

INTRODUCTION

Moebius syndrome is a rare congenital disturbance which is described in most studies as congenital facial weakness combined with abnormal ocular abduction. In 1888, Moebius reported patients with congenital, nonprogressive facial and abducent nerve palsy.1,2 This relatively uncommon congenital anomaly has been given various names: Moebius syndrome, congenital facial diplegia, congenital oculofacial paralysis, nuclear agenesis and congenital nuclear aplasia.2 Varying definitions have hampered a clear delineation of the Moebius syndrome. Isolated congenital facial palsy and the extended phenotype of congenital facial palsy with ocular muscle weakness, with or without craniofacial dysmorphisms were criteria for diagnosis of Moebius syndrome.2 Recently, it has been suggested that facial palsy with impairment of ocular system is the primary criterion for Moebius syndrome. Dysfunctions of other cranial nerves and orofacial malformations are commonly associated features but they are not necessary for the diagnosis, making the syndrome extremely variable in its clinical manifestations.2,3

Nerves commonly affected in this syndrome are facial (in all cases), abducent (75% cases), hypoglossal (20% cases) and oculomotor (5% cases). Most cases are diagnosed during infancy. Facial and ocular symptoms are usually the presenting problems. Facial diplegia is the most noticeable feature along with inability to close the mouth.2 Skin appears devoid of wrinkles. In adults, lower lip is usually everted and prominent. Speech problems are reported in 75 to 90% of these patients.1,2

The list of signs and symptoms mentioned in various sources for Moebius syndrome includes more than 20 peculiar features. Some of them are facial paralysis, lack of facial expressions, inability to smile, inability to suck, feeding difficulties, swallowing difficulties, choking problems during infancy, eye defects, motor delays, high palate, speech difficulty and hearing problems. Other associated congenital defects include jaw deformities, tongue deformities, limb deformities, club foot, missing fingers, webbed fingers and low muscle tone.1,3

The aim of this article is to illustrate the clinical features, radiological features and dental abnormalities in a rare case of Moebius syndrome.

CASE REPORT

A 12-year-old male patient came to our hospital with a chief complaint of inability to close his mouth since birth (Fig. 1). His history revealed that he was also unable to close his left eye since the same duration. He had undergone several neurological and ophthalmological investigations but no satisfactory diagnosis and treatment were given to him. He was born to normal parents with no history of consanguineous marriage. His other two siblings were normal.

On general examination, the patient appeared thin built and undernourished. His height was 143 cm and weight was 25 kg. All vital signs were under normal limits. Patient had normal IQ levels. No abnormality was detected in either of his upper limbs or lower limbs. His sensory functions were normal.
Four cranial nerves (facial, abducent, oculomotor and hypoglossal) were affected. Signs of facial nerve involvement were loss of motor functions on left side of face; hence, he had an expressionless face (Fig. 1). He was unable to close his left eye (Figs 1 and 2) and raise his left eyebrow (Fig. 2). There was absence of wrinkles on his left forehead (Fig. 2). Patient was neither able to whistle or blow air between his cheeks nor was able to smile. Sign of hypoglossal nerve involvement was deviation of his tongue to the paralysed side (Fig. 3).

Sign of abducent nerve involvement was his inability to completely abduct his left eye (Figs 1 and 2). Signs of oculomotor nerve involvement were patient’s inability to rotate his left eyeballs upwards or downwards and can only partially move it in medial and lateral directions (Figs 1 and 2). Other ocular manifestations were increased lacrimation from left eye along with presence of ptosis and strabismus (Figs 1 and 2).

Other facial and intraoral features were wide nasal apertures, incompetent lips, hypertrophic and everted lower lip (Fig. 1), macroglossia and fissured tongue (Fig. 4). Premature molar contacts bilaterally (Fig. 5) leading to anterior open bite and high vault palate.

Based on history given by the patient and features shown on clinical examination, case was diagnosed as Moebius syndrome for which the differential diagnosis was Melkersson Rosenthal syndrome, congenital facial muscular atrophy, muscular dystrophy, cerebral palsy and congenital myopathies.

Investigations performed for the case were routine hematological investigations which were under normal limits and radiological investigations which included orthopantomogram, lateral cephalogram, computed tomography and magnetic resonance imaging of the skull. Radiographs supported the clinical findings. Orthopantomogram and lateral cephalogram (Fig. 6) showed occlusal disturbances leading to anterior open bite. Computed tomography and magnetic resonance imaging of skull showed no significant abnormalities.

The patient is under treatment of a team comprising of oral physicians, pedodontists, orthodontists, and oral surgeons who are planning surgical correction for occlusal abnormalities. He is also advised surgical correction for strabismus and ptosis. He is under regular follow-up and care.

DISCUSSION

Moebius syndrome is a rare disorder. Only approximately 300 cases have been reported in literature. The prevalence in the United States is reported as 0.002 to 0.0002% of births, or 1 case per 50,000 newborns. Because of variety of clinical findings, etiology of Moebius syndrome is unknown. Etiologies that have been suggested includes: Dysplastic or degenerative developmental mishap, myopathies, peripheral neuropathies, vascular etiology, exposure to drugs, trauma during gestation period etc. Few familial cases with autosomal dominant transmission have also been reported.1-2,4 There are
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Journal of Indian Academy of Oral Medicine and Radiology, July-September 2011;23(3):267-270

Physical findings entirely depend on the case definition of Moebius syndrome. By using the most commonly accepted definition, the typical phenotypic appearance is an immobile facial feature with various glaze palsies as seen in our case report. The flattened facial expression causes patients to have difficulties in relating to others because of their inability to convey emotions.2,5 External ocular palsies, including ptosis accompany facial diplegia in 80% of patients.5 The intelligence is usually normal but may be underestimated because of absence of facial expressions. An interesting syndrome is seen in association with Moebius syndrome called as Kallmann syndrome, in which anosmia and hypogonadism are also present.1-3

Numerous orofacial abnormalities are present, like hypoplastic upper lip, microstomia, mouth-angle drooping, hypoplasia of mandible, gothic palate, tongue weakness, fissured tongue, tongue atrophy and open bite.6 Most of these abnormalities are present in our case. Other rare anomalies that can be seen in patients with Moebius syndrome include talipes equinovarus, brachydactyly, syndactyly, congenital amputations, arthrogryposis, smallness of limbs and occasional hypoplasia or absence of pectoralis major muscles (Poland anomaly).7 Skin abnormalities have also been reported including café au lait pigmentation and webbing of axilla.2

No diagnostic laboratory studies yield findings specific to Moebius syndrome. Very few cases of this syndrome have been described in the radiological literature. CT and MR imaging findings include hypoplasia of the pons or medulla, depression of the 4th ventricle, absence of the medial colliculus at the level of the pons, absence of the hypoglossal prominence, calcification in the pons in the region of the abducens nuclei and cerebellar hypoplasia.1,8 The course of facial nerve in the temporal bone can be depicted in polytomography or coronal CT scans. Electromyography may be used to differentiate this condition from muscular dystrophies.9

No definite treatment for this syndrome is available. The surgical goal in Moebius syndrome is far more modest and differs in patients with unilateral developmental facial paralysis. It is impossible to restore a true smile in these masks-like expressionless faces. Ocular surgeries, orthognathic surgeries and surgical corrections for other associated abnormalities may be tried. Medical care is only supportive and symptomatic. Complications depend on the severity of the patient’s deficits. They may include aspiration pneumonia, corneal ulceration/abrasion, dysphagia and poor nutrition. Consultations may be required from pediatricians, pedodontists, oral physicians, orthopedic surgeons, ophthalmologists, psychologists and speech therapists.1,2,6,10

CONCLUSION

Moebius syndrome is a rare disorder which not only causes physical abnormalities but also social and psychological problems. Children with this syndrome may grow up to feel alone and ‘defective’. Parental education is required early in the child’s life. Although neither the etiology nor the pathogenesis of this syndrome has yet been elucidated, several
theories regarding them have been postulated but none has been satisfactorily accepted. There is no single course of medical treatment. Treatment is supportive and in accordance with symptoms.

REFERENCES