

Case Report

Moebius syndrome: A Rare case report

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Abstract: Moebius syndrome is a rare syndrome with congenital non progressive bilateral or unilateral, complete or partial paralysis of the 6th and 7th cranial nerves. Orofacial anomalies and limb malformations may be associated with the disorder. The 3rd, 4th, 5th, 9th, 10th and 12th cranial nerves may also be affected. It is of unknown etiology with sporadic occurrence. This case report illustrates clinical features and dental abnormalities in a rare case of moebius syndrome.

Keywords: Congenital, Cranial nerve palsies, Facial paralysis, Moebius syndrome, Smileless syndrome

INTRODUCTION

Moebius syndrome was first described by German neurologist “Paul Julius Mobius” in 1888 [1]. The condition is one of the rare disorders amongst the disorders of oromandibular limb hypogenesis. Only approximately 300 cases have been reported in literature. The incidence is 1 in 50,000 of live births. Moebius syndrome is a rare non-progressive congenital disorder with the primary diagnostic criteria of congenital facial diplegia and abducent nerve palsy. Orofacial anomalies and limb malformations may be associated with the disorder. We report a case of moebius syndrome in a 8 year old female patient.

CASE REPORT

An 8 year old female patient reported to the department with a chief complaint of decayed teeth since 4 – 5 years, she was unable to co-operate with oral hygiene methods and because of difficulty in swallowing since childhood, she was fed on an exclusively soft consistency diet. She also gave history of sucking and feeding problems in infancy and was bottle fed till 2 years of age. She gave history of delayed developmental milestones.

She had visited the audiologist and speech pathologist with complaint of unintelligible speech at 5 years of age and it was found that she had predominant omission errors of bilabial, labiodental and palatal phonemes and distortion errors of fricatives and rhotics. She had history of nasal regurgitation during the early

years and currently for liquids occasionally. The orofacial mechanism revealed inadequate sucking and blowing functions with restricted movement of the tongue. She had been referred for nasoendoscopy to assess the velopharyngeal port which revealed grooved soft palate with moderate movement of posterior pharyngeal wall and lateral wall. There were no significant otological symptoms and hearing screening revealed hearing sensitivity within normal limits.

She had visited the psychologist at 6 years of age where the Wechsler's Pre Primary and Primary Scale of Intelligence and Vineland Social Maturity Scale (VSMS) tests were administered. She obtained an IQ of 62 indicating ‘mild’ mental retardation. On the VSMS, her social age indicate ‘dull normal’ level of functioning. She got her tooth extracted 1 year back and it was an unpleasant experience. There was no history of consanguineous marriage. Her mother's pregnancy was uneventful and ended at term. There was no history of birth trauma. Neither her brother nor anyone else in the family had any such problem.

No abnormality was detected in either of her upper and lower limbs. On extra-oral examination, patient had convex profile (fig 1), moebius like expressionless face (fig 2) and an underdeveloped hypoplastic mandible. She had impaired facial expressions i.e. forehead wrinkling and smiling. She was unable to close and abduct her eyes and had incompetent lips (fig 3). The mother reported that these

features were present since her birth. On intraoral examination multiple carious teeth, crowding of teeth (fig 4), deep bite and generalized inflammation of gingiva (fig 5) was present. She was unable to protrude and move her tongue (fig 4). She had high arched palate and cleft palate (fig 6). Panoramic radiograph was taken. The panoramic radiograph showed mixed dentition and developing permanent teeth in the jaw (fig 7).



Fig 1: convex profile of patient.



Fig 2: typical Moebius like expressionless face.

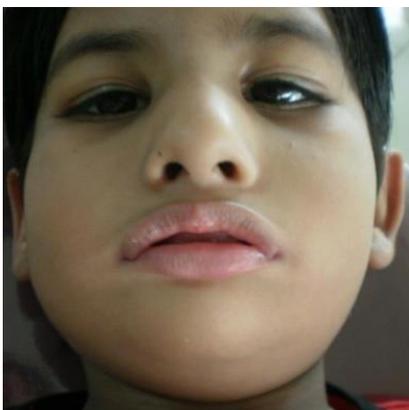


Fig 3: inability to close the eyelids and incompetent lips.



Fig 4: multiple carious teeth, crowding of teeth and inability to protrude her tongue.



Fig 5: inflammation of gingiva and deep bite.



Fig 6: high arch palate and cleft palate.



Fig 7: Panoramic radiograph showing mixed dentition and developing permanent teeth in jaws.

Based on the history given by the patient and in view of these findings of congenital facial diplegia, bilateral ocular impairment of abduction the patient was diagnosed to have Moebius Syndrome. The child and her parents were instructed about the importance of maintaining at-home oral hygiene. The extraction of grossly decayed teeth and pulpectomy of carious teeth was done. A multidisciplinary approach has been

established with pediatricians, ophthalmologists, psychologists, physical and speech therapists. She is under regular follow up and care.

DISCUSSION

Moebius syndrome is one of the rare disorders amongst the oromandibular limb hypogenesis which is described in most studies as congenital facial weakness combined with abnormal ocular abduction. Moebius described it as a rare syndrome with congenital non progressive bilateral facial and abducent nerve palsy. It may be associated with other craniofacial dysmorphisms and congenital abnormalities of the extremities. It is usually detected in infancy. This relatively uncommon congenital anomaly has been given various names such as Congenital facial diplegia, Congenital nuclear agenesis, Congenital oculo-facial paralysis, Smileless syndrome [2].

Nerves commonly affected in this syndrome are facial (in all cases), abducent (in 75% cases), hypoglossal (in 20%) cases and oculomotor (in 5% cases). Facial diplegia is most common noticeable feature along with inability to open the mouth. 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 [3].

Abramson et al actually classified and graded the syndrome on the basis of the clinical findings of cranial nerve palsies and musculoskeletal anomalies by using the acronym CLUFT (cranial nerve, lower limb, upper limb, face, and thorax). This grading system included cranial nerve features of either partial or complete 6th or 7th nerve palsies or both; lower extremity findings of talipes equinovarus, ankylosis, longitudinal, or transverse deficits; upper extremity involvement with digital hypoplasia or failure of formation; structural facial findings of cleft palate, micrognathia; and thoracic findings of scoliosis, pectoral hypoplasia, or other chest wall deformity[4].

Because of variety of clinical findings, etiology of Moebius syndrome is unknown. Despite the unclear pathogenicity of this syndrome, it is mostly believed that in the embryonic development, during the sixth week of pregnancy, a momentary stoppage or reduction in the circulation of the thoracic artery or one of its peripheral ramifications primes the pathogenic mechanism of the syndrome and results in different degrees of syndrome severity, depending on the length and intensity of the vascular interruption[5].

There is evidence that genetic and environmental factors, however, are related to this syndrome. Among the generic factors are: descriptions of dominant autosomal inheritance, recessive autosomal inheritance, translocation between the short arm of chromosome 1 and the long arm of chromosome 13 ; and involvement of the long arm of chromosome 3,

with the critical region being delimited by markers at the chromosome 3q21-22[6,7].

The most frequently associated environmental factors are: hyperthermia; generalized hypoxia; gestational diabetes; exposure of pregnant women to infection (rubella); and the use of certain drugs (benzodiazepines, misoprostol, alcohol, cocaine, and thalidomide) during the first trimester of pregnancy. These factors have also been related to vascular defects, such as transient fetal ischemia, which is one of the most widely accepted theories that explain the origin of this syndrome[7-8].

Moebius syndrome's clinical characteristics include loss of facial expression ("mask-like facies"), impaired stomatognathic system functions, and inability to close the eyelids. In addition, a number of congenital anomalies of the extremities and brachial musculature have been observed radiographically, including: talipes equinovarus (clubfoot); digital anomalies; unilateral absence of pectoral musculature; congenital hip dislocation; and arthrogyrosis (permanent or persistent joint flexure). Other ocular abnormalities include: restricted lateral eye movements; ptosis; nystagmus; strabismus; and conjunctivitis[7].

Numerous orofacial abnormalities are present like micrognathia; microstomia; hypoplastic upper lip; gothic or cleft palate; tongue deformities; bifid uvula; hypoplastic teeth; congenitally missing teeth; open bite; gingivitis; hypoplasia of the mandible; drooling; angular cheilitis nursing bottle caries; lack of lip seal; drooping mouth angle; and inability to perform mandibular lateral movements and protrusion[9]. The increased risk of caries and periodontal diseases in Moebius syndrome patients has been widely reported and is frequently associated with a soft-consistency diet and ineffective oral hygiene, due to impaired tongue movements, masticatory function, and motor ability as found in the reported case.

The criteria for diagnosis are difficult to define; however, the following guidelines should assist in making a diagnosis of Moebius syndrome.

Criteria for diagnosis

- (1) Complete or partial facial nerve paralysis is essential for the diagnosis of Moebius syndrome.
- (2) Limb malformations (syndactyly, brachydactyly or absent digits, and talipes) are often present.
- (3) The following additional clinical features in association with complete or partial facial nerve paralysis may also be present and should be helpful in making a clinical diagnosis of Moebius syndrome: bilateral or unilateral ocular nerve palsies (commonly of the abducens and less commonly of the oculomotor and trochlear nerves); hypoplasia of the tongue owing to hypoglossal nerve paralysis; swallowing and speech difficulties owing to trigeminal, glossopharyngeal, and

vagus nerve palsies; malformations of the orofacial structures (bifid uvula, micrognathia, and ear deformities); other anomalies of the musculoskeletal system, for example, Klippel-Feil anomaly, absence of the sternal head of the pectoralis major, rib defects, and brachial muscle defects are also present [10-11].

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 [12-13]. 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[14].

No definite treatment for this syndrome is available. It is directed toward the restoration of motion secondary to the facial nerve palsy, which results in mask-like facies and inability to smile. Facial reanimation using free pectoralis minor transfer is an invaluable aid to such patients and allows for greater social interaction by means of the ability to smile. 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. Consultations may be required from pediatricians, pedodontists, oral physicians, orthopedic surgeons, ophthalmologists, psychologists and speech therapists.

In conclusion, because of the variable findings in Moebius syndrome, it is important to use a meticulous evaluation process for each child, which may involve a variety of professional disciplines. Recognizing the problems the child presents and establishing an adequate treatment plan is a major responsibility for those professionals involved. The dentists' knowledge of Moebius syndromes etiology and clinical features is important to make these professionals familiar with this condition and improve their participation in the multidisciplinary teams usually involved in the treatment of these patients.

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