Case Report

Moebius Syndrome: A Case Report

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Abstract

Moebius syndrome consists of congenital, complete or partial facial nerve palsy with or without paralysis of other cranial nerves (most commonly an abducens paralysis) and often associated with other malformations of the limbs and orofacial structures.

In this case, a ten days old male child with moebius syndrome is presented, clinical findings are described and management aspects are discussed.

Key words: Congenital syndrome, facial palsy in neonate, moebius syndrome

Introduction

Paul Julius Moebius, a German neurologist, in 1892 described a clinical entity of bilateral combined palsies of the 6th and the 7th cranial nerves that subsequently carried his name [1]. Though the first description of congenital facial diplegia was given by Von Graefe in 1880[2]. Moebius Syndrome is congenital non-progressive complete or partial, unilateral or bilateral facial nerve palsy with or without paralysis of other cranial nerves (III, V, VI, IX, X) and physical abnormalities such as multiple limb malformations (syndactyly , brachydactyly , or absent digits and talipes) [3].

The most obvious symptoms of this syndrome include a loss of facial expression and jeopardized functions of the stomatognathic complex [3]. Ocular presentations include restricted lateral eye movements, ptosis, nystagmus, strabismus, conjunctivitis and inability to close the eyelids [3].

Mental retardation is probably over diagnosed owing to mask-like facies, drooling of saliva, strabismus and speech difficulties. In one study mild to moderated mental retardation is known to occur in 10% of the cases [4] while in other moderate to severe mental deficiency was said to be present in at least half of the children with clinical features of moebius syndrome [5].

The purpose of this report is to describe a rare clinical case of moebius syndrome.

Case Report

A ten days old male patient who was born full term at 38 weeks with intrauterine growth retardation (IUGR) came to hospital with abnormal facial expression. The parents were unrelated and young and healthy at the time of conception. There was no history of maternal drug intake.

The infant was brought to the pediatricians with the complaints of a restricted mouth opening and an expressionless face. There was incomplete closure of the left eyelids during sleep and drooling of saliva.

Generalised hypotonia was seen, muscle tone was being more decreased around the shoulders than in the lower extremities. No clonus was elicited. Limb malformations were not obvious although minor syndactyly was present. His right upper limb appeared to be hypoplastic with fourth partial syndactyly.

Manuscript received: 04th Sep 2014
Reviewed: 15th Sep 2014
Author Corrected: 20th Sep 2014
Accepted for Publication: 29th Sep 2014

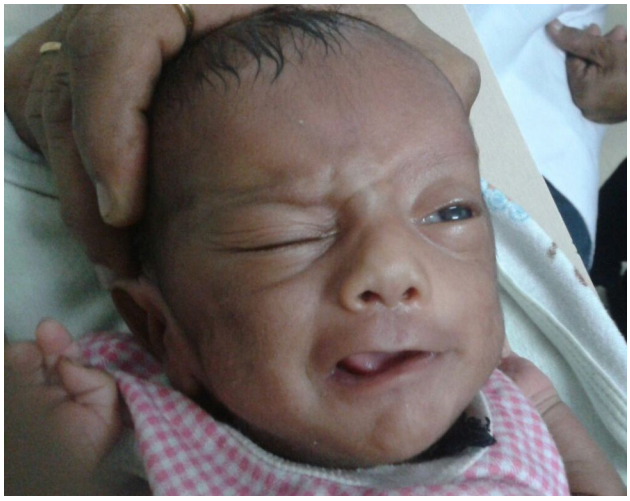

Fig 1 and 2: Photographs of the infant showing some of the dysmorphic features (Inability to close left eye, expression less face)

Discussion

The etiology of this syndrome was considered to be a congenital absence of the motor nuclei of abducens and facial nerves, but genetic and environmental factors can also be involved. Moebius in his original description of the condition postulated that the anomaly resulted from degeneration of the nuclei of the sixth and seventh cranial nerves [6,7].

Heubner [8] suggested nuclear agenesis or hypoplasia as the chief underlying cause of moebius syndrome, which is supported by others [9]. Exposure to infections, alcohol, cocaine, thalidomide or misoprostol was also related in association with moebius syndrome [10]. It was found in most cases that this syndrome is because of the infarction of brain stem nuclei during fetal life [10].

The pathogenesis of cranial nerve palsies associated with limb anomalies is difficult to explain. An ischemic process resulting from an interruption in the vascular supply during early fetal development, around four to six weeks of gestation, may result in facial and limb anomalies characteristic of moebius syndrome [2].

The hypothesis is supported by the report of moebius – like facies resulting from damage to the tegmentum of the midbrain secondary to basilar artery thrombosis, possibly related to birth trauma [2]. Another explanation for cranial nerve dysfunction and limb anomalies may be a primary metameric defect in the brain stem nuclei and somite mesoderm of the limb buds. The occurrence of Poland's anomaly in moebius syndrome is probably related to a common pathogenetic mechanism. Poland's anomaly is believed to result from a restricted defect in the metamerisation process of the somite mesoderm [2].

As far as genetics is concerned, most of the reported cases have been sporadic and both sexes are affected with equal frequency. There are no prevalence figures and the exact population incidence is not known. Pedigrees with an autosomal dominant, autosomal recessive and X-linked recessive inheritance pattern have been described [11].

Criteria for Diagnosis [2]

1. Complete or partial 7th nerve paralysis – essential for diagnosis.
2. Limb malformations (syndactyly, brachydactyly or absent digits, and talipes) are often present.
3. Some additional features may also be present:

    a) Bilateral or unilateral ocular nerve palsies (commonly of 6th nerve).
    b) Hypoplasia of tongue due to palsy of 12th nerve.
    c) Swallowing and speech difficulties due to 5th, 9th and 10th nerve palsies.
    d) Malformations of oro-facial structures (bifid uvula, micrognathia and ear deformities).
    e) Other anomalies of the musculoskeletal system like rib defects, absence of pectoralis major muscle (Poland anomaly) [3].

Moebius Syndrome is a condition which deprives people of the capacity to project their emotions through facial expressions. The lack of facial expressivity might lead to a decrease in the parental bonding [12]. A patient of moebius syndrome starts suffering from first day of

his/her life with difficulty in sucking milk being the first sign. These children require a multidisciplinary case and support and also the parental co-operation. Feeding difficulties and problems of aspiration often lead to failure to thrive during infancy [10].

Feeding tubes or special feeding bottles can be used for nursing to avoid swallowing and choking problems. Because of expression less face child may face & inability to convey reactions of joy or sorrow to her family and friends which ultimately may become a leading cause of his being interrupted as a mentally retarded child.

Thus psychological management and motivation should form the mainstay of our treatment for such children as they grow. Moreover genetic counseling of the parents should be done explaining them the possible pathogenesis, natural history of the disease and supportive measures for long term rehabilitation.

The treatment of patients with moebius syndrome is directed toward the restoration of motion secondary to the facial nerve palsy.This involves reconstructive plastic surgery with muscle transplantation ideally performed in patients just before they reach school age at 4 or 5 years.

Funding: Nil

Conflict of interest: Nil

Permission from IRB: Yes

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How to cite this article?

Singh P, Gupta SB, Kumar R, Agarwal R, Singh DP, Verma D. Moebius Syndrome: A Case Report. Int J Med Res Rev 2014;2(6):624- 626. doi:10.17511/ijmrr.2014.i06.21