Case Report

Diagnosing Möbius Syndrome on MR Imaging

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A B S T R A C T

Introduction: Möbius syndrome is a rare disorder with the preliminary diagnostic criteria of congenital facial and abducens nerve palsy.

Case report: We present a case of Möbius syndrome in a 9 year old boy with VI and VII cranial nerve palsy and delayed milestones which on MR imaging shows absence of bilateral VI and VII nerves. In addition, partial atresia of right external auditory canal was observed. The above findings led to a diagnosis of "Möbius syndrome".

Conclusion: Genetic anomalies are the primary cause for familial cranial nerve agenesis as seen in our case of Möbius syndrome.

Keywords: Facial nerve, Abducens Nerve, Möbius syndrome, Facial diplegia

INTRODUCTION

Möbius syndrome(MBS) is a rare congenital neurological disorder which is characterized by congenital complete or partial facial nerve paralysis with or without paralysis of other cranial nerves like abducens, occulomotor and hypoglossal.[1,2] The most frequent presentation is facial diplegia with bilateral lateral rectus palsy.

The prevalence of MBS is estimated to be 1/250,000 live births with equal incidence in both sexes of which 2% cases have familial inheritance.[3] Till now about 300 cases of MBS have been reported in the literature.[4]

CASE REPORT

A 9 year boy presented with congenital facial muscle weakness and restriction of outward eye movements which was non-progessive in nature. He had undergone many neurological and ophthalmic examination but no significant improvement seen. Developmental delay was noted in child from the very first year as he was unable to attain the respective milestone at the expected age. Familial inheritance was seen in the form of similar features in father and grandfather.

On physical examination, there was absence of facial wrinkles, bilateral convergent squint, high arched palate with both upper and lower lips everted, expressionless mask- like face were noted. Bilateral facial nerve and abducens nerve paralysis noted on neurological examination (Figure 1).

On MR imaging, following findings were seen:

- Non-visualisation of cisternal and canalicular segments of bilateral facial nerves (Figure 2 and 3).
- Absence of bilateral abducens nerve at the pontomedullary junction (Figure 4).
- Flattening of fourth ventricle suggesting absence of facial colliculus (Figure 5).
- Partial atresia of right external auditory canal (Figure 6).
- Clinical examination with MR imaging confirms a congenital cause of multiple cranial nerve palsy.

DISCUSSION

Paul Julius Möbius, a German neurologist, in 1892 was the first to describe this condition. It is also called as congenital facial diplegia, congenital oculo-facial paralysis, nuclear agenesis and congenital nuclear aphasia.[5,6]

There were many criteria to diagnose the MBS which vary among authors. According to Kumar et al[7], following criteria were used to diagnose the disease: (a) Complete or partial facial nerve paralysis is an essential criterion for the diagnosis of Möbius syndrome, (b) Limb malformations (syndactyly, brachydactyly, absent digits, talipes) are often present, (c) Bilateral or unilateral cranial nerve palsies (commonly VI, XII, also IX, X) may be seen, (d) Orofacial malformations, ear deformities, and musculoskeletal deformities may also be seen.

Recently, Verjzijl et al[8], reviewed the criteria and suggested facial palsy with impairment of ocular abduction as the primary criterion for diagnosing Mobius syndrome.

Möbius syndrome is a very rare entity which manifests soon after the birth with difficulty in sucking, inability to completely close the eyelids, expressionless face, loss of forehead wrinkles, facial asymmetry and restriction of outward eye movements.

The etiology of Möbius syndrome is although multifactorial, the genetic mechanisms links to loci 1p22 and 13q12.2 associated with hypoplasia or aplasia of the cranial nerve

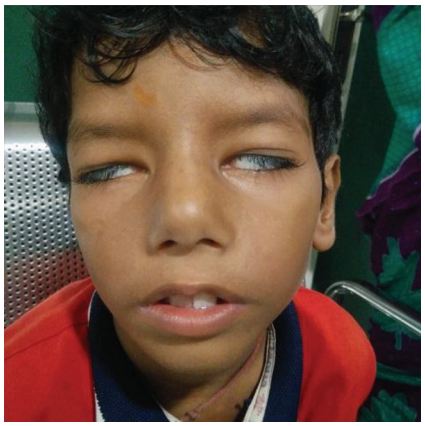

Figure-1: Partially closed eyes in the child while trying to close the eyes completely

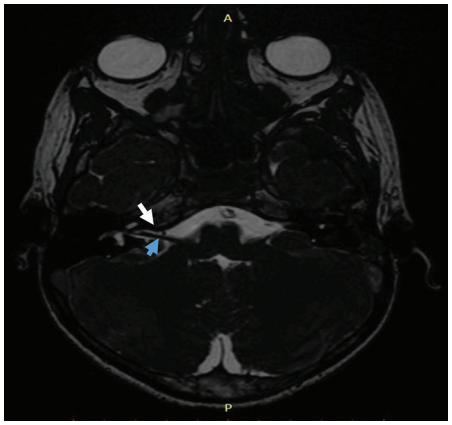

Figure-2: Axial FIESTA showing absence of cisternal and canalicular segments of right facial nerve

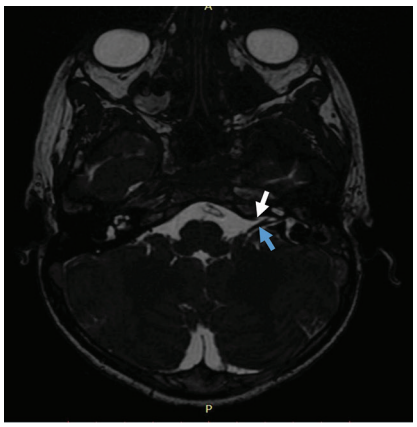

Figure-3: Axial FIESTA showing absence of cisternal and canalicular segments of left facial nerve

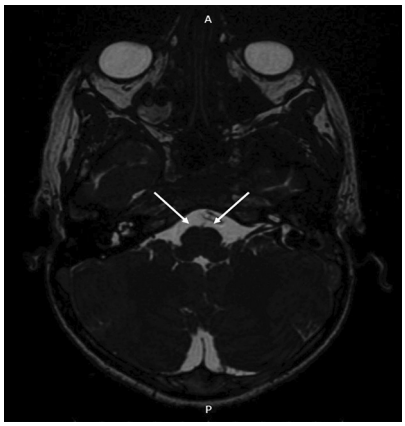

Figure-4: Axial FIESTA showing absence of bilateral abducens nerve at the pontomedullary junction

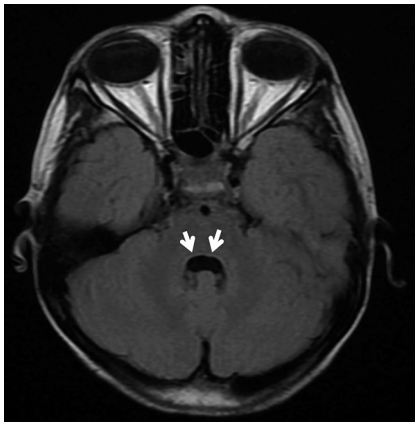

Figure-5: Axial T2 FLAIR showing flattening of fourth ventricle suggesting absence of facial colliculus

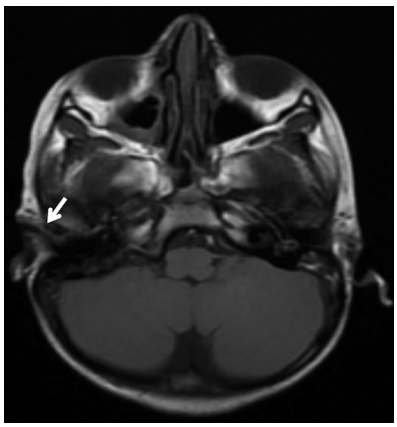

Figure-6: Axial T2WI showing partial atresia of right external auditory canal and normal left auditory canal

nuclei have been suggested.[9]

The characteristic finding on MR imaging is the absence of unilateral or bilateral facial and abducen nerve. Other finding like flattening of fourth ventricle indicates absence of facial colliculus which was also seen in our case.[10]

One of the most important differential of MBS is craniofacial syndrome which results from the abnormal growth pattern of face or skull, brought by birth defect or trauma. In 15% cases of Poland syndrome, MBS may present with congenital weakness of pectoral muscles. The seventh nerve palsy in MBS primarily affects the upper face, while other lower motor neuron palsies affect both upper and lower portions of the face. Similarly, supranuclear lesions of facial nerve affect only lower half of the face.

No definitive treatment is yet available, only supportive and symptomatic treatment can be given in the form of ocular care, surgical restoration of facial expression, speech therapy and occupational therapy to help in their daily activities.

CONCLUSION

MR imaging is very characteristic and has emerged as the gold standard modality in diagnosing cranial nerve anomalies

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and other associated syndromes.

Steady state free precession(SSFP) is the sequence of choice for visualizing cranial nerves due to it's ability to provide submillimetric spatial resolution highlighting the course of each nerve.

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