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Neuroimaging of a Rare Congenital Disorder- Moebius Syndrome

Case Study

Joshi A[1], Benjwal G[2]*, Sharma G[2] and Kala P[2]

[1]*Department of Anatomy, Himalayan Institute of Medical Sciences, Swami Rama Himalayan University, Dehradun, Uttarakhand, India*

[2]*Department of Radiodiagnosis, Himalayan Institute of Medical Sciences, Swami Rama Himalayan University, Dehradun, Uttarakhand, India*

***Corresponding author:** Gaurav Benjwal, Department of Radiodiagnosis, Himalayan Institute of Medical Sciences, Swami Rama Himalayan University, Dehradun, Uttarakhand, India. Email id: grv_22@ymail.com

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Abstract

Moebius syndrome(MBS) is a rare congenital neuromuscular disorder of the unilateral or bilateral sixth and seventh cranial nerve. It is mostly a clinical diagnosis but neuroimaging can aid in detecting its salient features.

This case report is a documentation of Magnetic resonance (MR) findings of a 9-year-old female diagnosed with MBS. Neuroimaging showed bilateral hypoplastic abducens nerves and bilaterally absent facial nerves with associated ventriculomegaly, hippocampal malrotation, and midbrain malformation.

Thus, neuroimaging using magnetic resonance can be used to capture the radiological features of MBS.

Keywords: Moebius Syndrome; MRI; Facial Nerve; Abducens Nerve; Ventriculomegaly

Introduction

Moebius syndrome (MBS) is a rare congenital neuromuscular disorder characterized by non-progressive weakness of the sixth (VI) and seventh (VII) cranial nerves, leading to ophthalmoplegia and facial paralysis. First described by Von Graefe and Saemisch in 1880 and later confirmed by Paul Julius Moebius in 1888, the prevalence of the syndrome is approximately 1 in 25,000 live births, with no gender predilection and mostly sporadic cases [1]. Since it can resemble several other neuromuscular conditions, neuroimaging can aid in better differentiating it from its clinical mimics.

Here, we report the case of a 9-year-old girl diagnosed with Moebius syndrome, focusing on its characteristic MRI findings.

Case Report

A 9year old girl presented to the pediatric OPD with complaints of the inability to move both eyes laterally and slurred speech. She lacked facial expressions since birth and had recurrent episodes of corneal ulcers, as informed by her parents. She also had drooling of saliva from the corners of her mouth since birth. On detailed physical examination, the patient had bilateral deficits of VI, VII and XII cranial nerves evidenced by the inability to move both her eyes laterally, bilateral absence of frowning, cheek blowing, proper eye closure, nasolabial folds and facial expressions and upward deviation of tongue respectively.

Multiplanar MR imaging of the cranium was done on a 1.5 Tesla magnet (SIEMENS 1.5T MAGNETOM) using a dedicated head coil. T1, T2 weighted images were obtained in axial, sagittal and coronal planes using Spin Echo and Gradient Echo sequences. Constructive Interference in Steady State (CISS) sequence showed bilaterally absent cranial nerves VII (Figure 1) and thinned-out

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hypoplastic cranial nerves VI (Figure 2). Ventriculomegaly (Figure 3) with predominantly prominent frontal horn without obstructive hydrocephalus was noted. Partial fusion of thalami was seen along a narrow band with mild reduction of the interpeduncular cistern diameter.T2W sagittal images showed Tectal beaking (Figure 4) and absent facial colliculus with resultant straightening of the fourth ventricle's floor (Figure 5).

T2W coronal inversion recovery sequence demonstrated malrotation of bilateral hippocampus (Figure 6) with deep and vertical collateral sulcus. The scan showed no calcifications or hemorrhages. The brain parenchyma, brainstem and cerebellum were normal

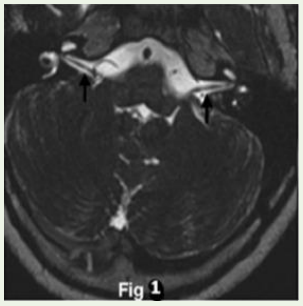

Figure 1: Enlarged image from 3D CISS sequence demonstrating the absence of facial (Cranial nerve VII) nerves bilaterally with single nerve (vestibulocochlear nerve; black arrows) coursing through bilateral cerebellopontine angle cisterns and internal acoustic meatuses supplying bilateral inner ear structures.

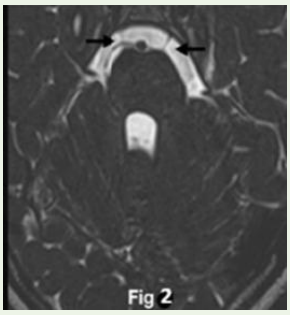

Figure 2: Enlarged image from 3D CISS sequence demonstrating thinning and reduced calibre of bilateral abducens nerves in the prepontine cistern (black arrows).

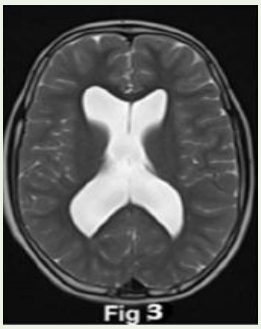

Figure 3: Axial T2W image showing dilatation of bilateral lateral ventricles (ventriculomegaly).

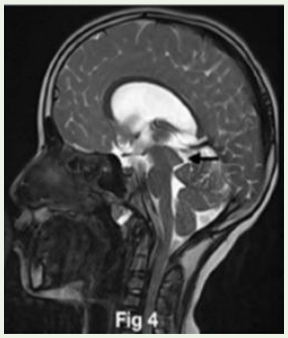

Figure 4: Midline Sagittal T2W image showing tectal beaking (black arrow).

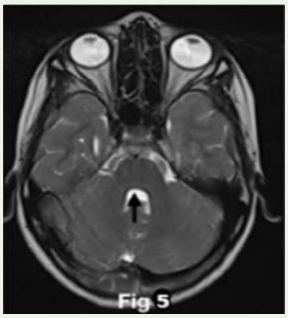

Figure 5: Axial T2W image displaying fourth ventricle floor straightening (black arrow) due to absence of facial colliculus.

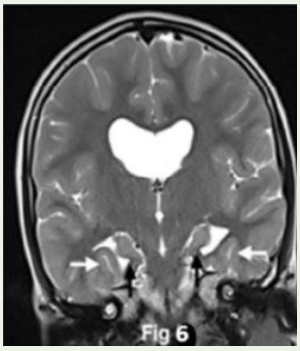

Figure 6: Coronal T2W image demonstrating bilateral hippocampal malrotation (black arrows) associated with the vertical orientation of the collateral sulci bilaterally (white arrows).

Informed consent, including permission about potential publication in a scientific journal was obtained from the father of the patient involved in the study.

Discussion

MBS develops due to faulty embryogenesis of the mesencephalon and rhombencephalon [2]. Histopathological evaluation identifies the primary pathology in the pontine tegmentum [3], where the nuclei of the VI cranial nerve and the posterior facial colliculus are located.

Among the various diagnostic criteria for MBS, Kumar et al. outlined the following: (a) partial or complete VII nerve paralysis, (b) associated limb defects such as syndactyly, brachydactyly, absent

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digits, or talipes, (c) unilateral or bilateral cranial nerve palsies (including VI, XII, IX, and X), and (d) potential orofacial, ear, and musculoskeletal deformities [3]. Meanwhile, Verzijl et al. emphasized facial palsy and impaired ocular abduction as key diagnostic features. [3] The present case satisfies both criteria.

Other cranial nerves frequently involved in MBS include V, IX, X, XI, and XII ,[4] with cranial nerve XII being the third most commonly affected.[3] Verzijl et al. proposed that MBS may arise from either a genetic defect affecting the VII nerve nuclei or intrauterine environmental and mechanical factors that disrupt brainstem vascularity.[3] However, Cocaine and Misoprostol use during pregnancy have also been associated with MBS ,[5,6] which suggests that MBS is a possible teratogenic effect of these drugs.

While MBS is primarily diagnosed clinically, neuroimaging modalities such as MRI allows direct imaging of cranial nerves and extraocular muscles. The MRI findings in this case align with those reported by Matsui et al. ,[7] showing ventriculomegaly without features of obstructive hydrocephalus. However, Matsui et al. did identify hydrocephalus in some patients with ventriculomegaly. In contrast, a cross-sectional study by Herrera et al. [8] found ventriculomegaly and hydrocephalus in only a few cases. Cerebral aqueductal stenosis was identified as the underlying cause of obstructive hydrocephalus in both studies.

The possible cause of ventriculomegaly without obstructive hydrocephalus in MBS can be white matter volume loss which can manifest as prominent frontal horns of lateral ventricle in the present case and other researches.[7,8]

Brainstem and cerebellar hypoplasia are commonly reported in MBS;[1,3,9] however, neuroimaging in this case revealed normal appearances of both structures. Infratentorial anomalies, including mesencephalic malformations with a reduced interpeduncular cistern diameter and tectal breaking, were noted in this case, consistent with the findings of Volpe et al. [10]

Thalamic fusion has been documented in MBS, with an established association between thalamic fusion and agenesis of the septum pellucidum, indicating a mild form of lobar holoprosencephaly.[8] The neuroimaging findings in this case corroborate this association.

The absence of the facial colliculus, a characteristic feature of MBS, results in a straightened floor of the fourth ventricle, a finding observed in this case. However, some studies describe the floor of the fourth ventricle as horseshoe-shaped.[3,8]

Hippocampal malrotation, as noted in this case, has also been reported in some subjects in a cross-sectional study by Herrera et al. [8]Calcification in the pontine region housing the VI nerve nuclei has been documented in MBS,[3,10]although it was not observed in this case.

The management of MBS is primarily supportive and symptomatic, requiring long-term multidisciplinary care, including physical, psychological, speech, and occupational therapy.

Conclusion/Summary

Neuroimaginging MBS particularly MRI shows bilateral hypoplastic abducens nerves and absent facial nerves with associated ventriculomegaly, hippocampal malrotation, and midbrain malformation. Thus, MRI using the CISS sequence can play an important role in evaluating brainstem and associated abnormalities in MBS which can be helpful for clinicians to differentiate it from other congenital neuromuscular disorders.

Acknowledgement

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