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Case Report

Möbius Syndrome

Serge Ouanounou, Gaurav Saigal, and Sherri Birchansky

Summary: We describe a case of Möbius syndrome in a 3-month-old infant. Striking imaging findings of pontine hypoplasia in the region of the 6th and the 7th nerve complexes were noted. In addition, absence of the middle cerebellar peduncles was noted, a finding that, to our knowledge, has never been reported before in the literature. Clinical presentations, other radiologic findings, and a possible pathogenesis are discussed.

The hallmark of Möbius syndrome is 6th and 7th nerve paresis. It may be associated with craniofacial, musculoskeletal, and cardiovascular defects, as well as other cranial nerve palsies.

Case Report

A 3-month-old male patient, born prematurely at 35 weeks, was noted to have several dysmorphic features, including retropositioning and low-set ears. Neurology and genetics consultations were obtained. Neurologic examination revealed an expressive face with an inability to move the facial muscles. Bilateral 6th and 7th cranial nerve deficits were noted. Generalized hypotonia was seen, with the muscle tone being more decreased around the shoulders than in the lower extremities. Vertical (holding the infant above the waist in a vertical direction to evaluate for axial tone) and horizontal (holding the infant at the waist at a horizontal level to see if the head can be supported) suspension also revealed hypotonia. No clonus was elicited. Chromosomal analysis and auditory tests were normal.

MR imaging demonstrated an abnormal configuration of the brain stem, especially the pons, which appeared hypoplastic particularly in its dorsal aspect in the region of the facial colliculus and 6th nerve complexes (Fig 1). Complete absence of the middle cerebellar peduncles was noted (Fig 2). In addition, the superior cerebellar peduncles appeared prominent bilaterally (Fig 3). The internal auditory canals appeared small. No cerebellar hypoplasia or atrophy was noted. The supratentorial brain appeared normal.

Discussion

Paul Julius Möbius, a German neurologist, first described a clinical entity of bilateral combined palsies of the 6th and the 7th cranial nerves that subsequently carried his name. Möbius syndrome is not only characterized by 6th and 7th nerve palsies, which

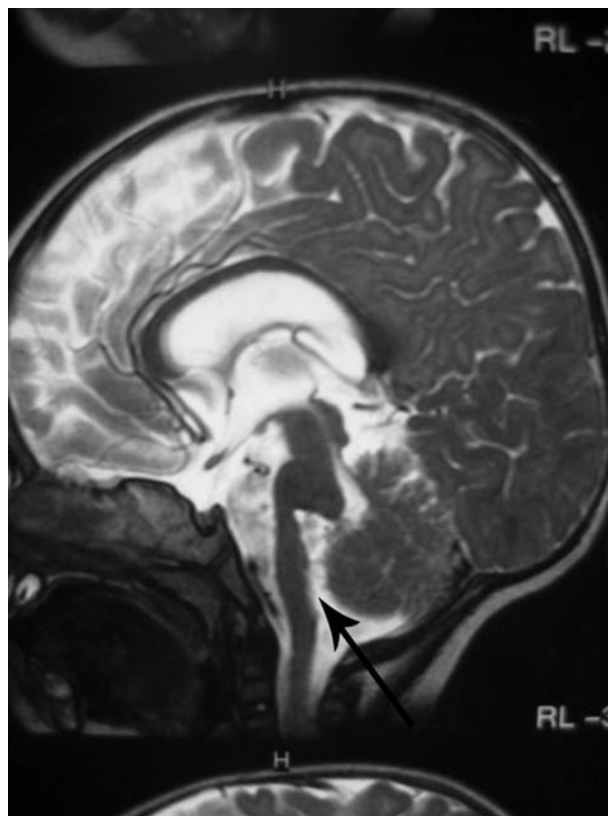


FIG 1. Sagittal T2-weighted MR image demonstrating an abnormal configuration of the brain stem, especially the pons, which appears hypoplastic, particularly in its dorsal aspect (arrow).

are its hallmark, but also involves abnormalities of the limbs, chest wall, spine, and soft tissues. Abramson et al (1) 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, or microtia; and thoracic findings of scoliosis, pectoral hypoplasia, or other chest wall deformity.

In another extensive review of 37 patients (largest series of Möbius patients to date), authors observed a uniform clinical picture characterized by facial diplegia of the upper and lower facial muscles, bilateral eye abduction impairment, hypoglossia, craniofacial and limb malformations, and long tract symptoms (2).

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From the Department of Radiology, University of Miami (S.O., G.S.), and the Department of Radiology, Miami Children's Hospital (S.B.), Miami, FL.

Address correspondence to Gaurav Saigal, MD, Department of Radiology, University of Miami, 1400 NW 12th Avenue, Miami, FL 33136.

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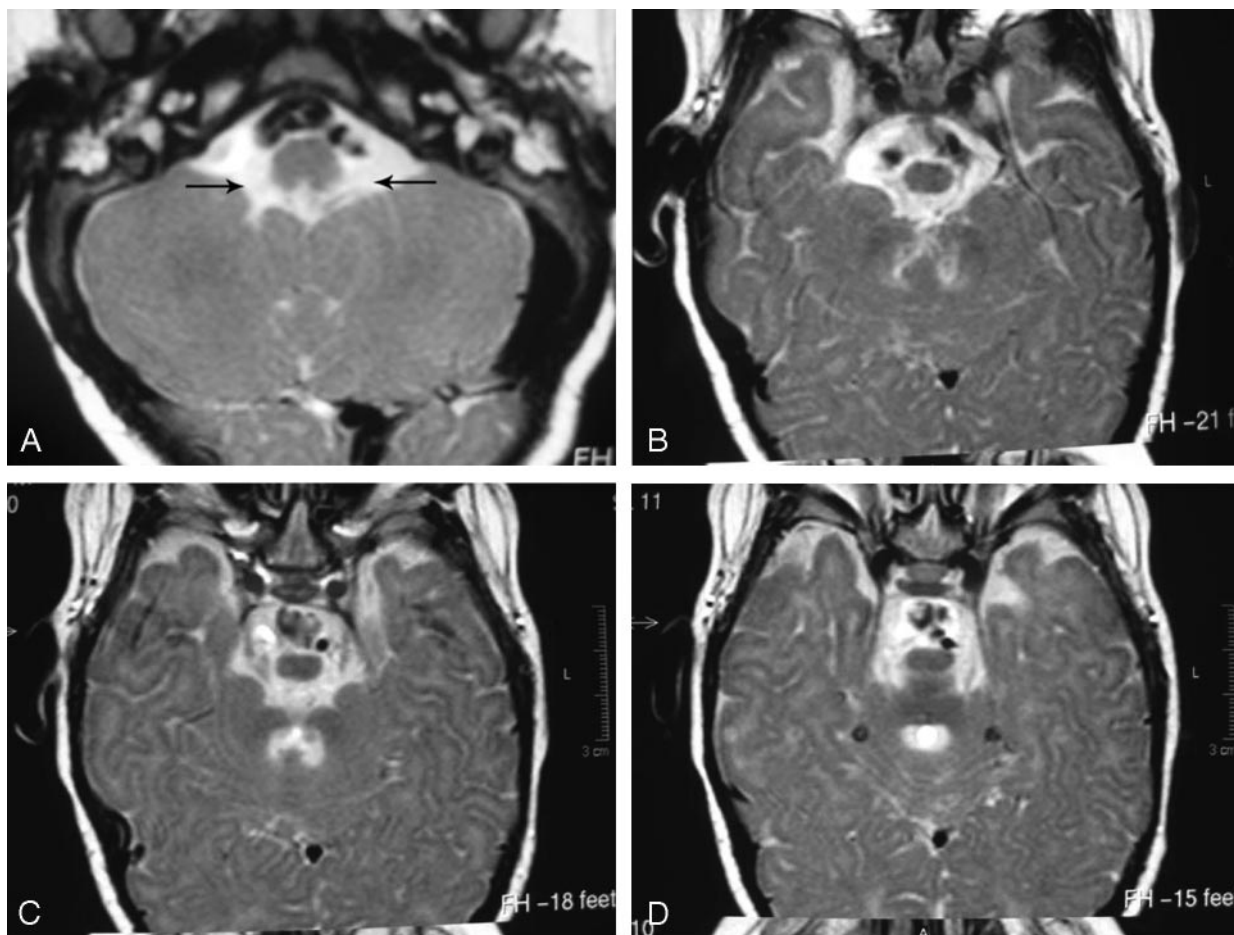


FIG 2. Axial T2-weighted MR images (A, caudal pons; B, midpons; C, junction of mid and rostral pons; D, rostral pons) demonstrating absence of the middle cerebellar peduncles bilaterally. The internal auditory canals appear small (arrows).

The more severe involvement of the upper facial muscles with relative sparing of the lower facial muscles, as well as the inability to abduct the eyes beyond the midline, bilaterally and symmetrically, were found to be highly characteristic of this syndrome. The lack of fine motor skills and poor coordination and balance performance seen in 80–90% of their patients were summarized as “clumsiness.” The authors concluded these findings to be due to maldevelopment of either the corticospinal or cortico-bulbo-cerebellar tracts.

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 suggestive of hypoglossal nuclei hypoplasia, calcification in the pons in the region of the abducens nuclei, and cerebellar hypoplasia (3–5). The case described here not only demonstrates the unusual morphological characteristics of the brain stem secondary to the pontine hypoplasia but also shows an associated absence of the middle cerebellar peduncles bilaterally. To the best of our knowledge, this finding of the absence of the middle cerebellar peduncles has never before been reported in the literature. The combination of the 6th

and 7th cranial nerve palsies with both upper and lower limb hypotonia can be attributed to the pontine hypoplasia as seen in our case, because the pons houses both the cranial nerve nuclei and the corticospinal tracts.

Towfighi et al (6) reported the neuropathologic findings in patients with Möbius syndrome. They divided the autopsy cases into four groups on the basis of neuropathologic findings: 1) Absence or decrease in the number of neurons in the affected cranial nerve nuclei without necrosis or degeneration of the actual nuclei. 2) In addition to neuronal loss, active neuronal degeneration was seen in the affected facial nerve nuclei. Primary peripheral nerve involvement was also seen. 3) In addition to a decrease in the number of neurons and reactive changes in the region of the involved cranial nerve nuclei, frank necrosis of the tegmentum of the lower pons was noted. 4) No lesions were found in the brain stem or the cranial nerves; however, severe atrophy of the facial muscles and creatinuria was seen, which suggests a myopathic disorder. Our case would fit in the group 3 of the neuropathologic findings as described by Towfighi et al.

The etiology of Möbius syndrome is multifactorial, and several theories have been proposed, with the

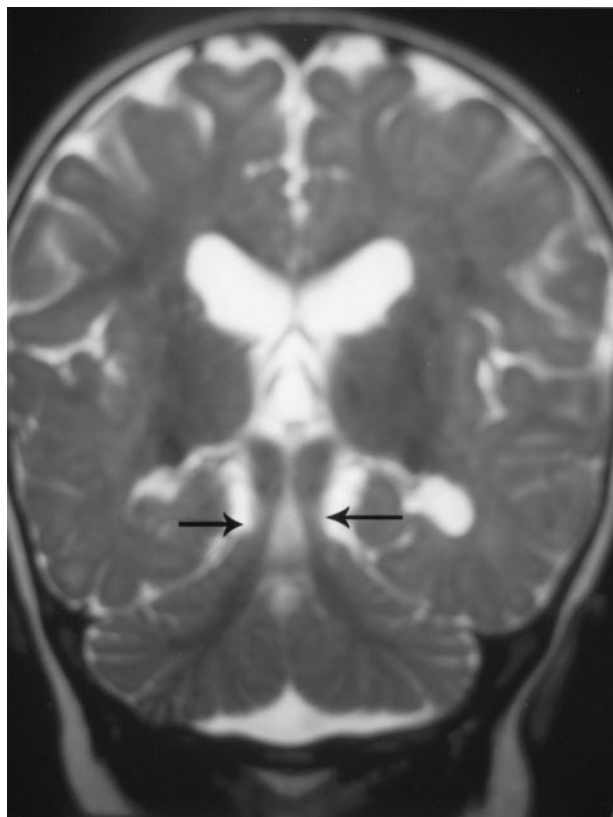


FIG 3. Coronal T2-weighted MR image, demonstrating prominence of the superior cerebellar peduncles (arrows).

most supported theory being that of transient ischemic or hypoxic insult to the fetus (1). Other infectious and genetic etiologies have also been proposed. In addition, the use of misoprostol, a prostaglandin-E1 analog and abortifacient during pregnancy, has also been implicated (7).

The treatment of patients with Möbius syndrome is directed toward the restoration of motion secondary to the facial nerve palsy, which results in mask-like facies and inability to smile. This involves reconstructive plastic surgery with muscle transplantation ideally performed in patients just before they reach school age at 4 or 5 years (8).

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