Möbius Syndrome as a Syndrome of Rhombencephalic Maldevelopment: A Case Report

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Möbius syndrome is a rare congenital disorder characterized by congenital facial weakness with impairment of ocular abduction. It is considered as a rhombencephalic disorder, and is often accompanied with hypoplasia of the pons and cerebellum. Here we report a male infant who had congenital facial asymmetry with absence of right-sided expression. Evident developmental delay was also found. The bilateral auditory brain stem response showed no response at 85 dB. A reconstructive brain magnetic resonance imaging (MRI) revealed the absence of the right facial nerve as well as hypoplasia of the pons and cerebellum of the same side. Some voluntary contraction over the patient’s right lower face was noted after facial muscle electrical stimulation.

1. Introduction

Congenital facial and abducens palsy was described by Von Graefe originally in 1880, and followed by other reports.¹ In 1888, Möbius drew attention to patients with congenital non-progressive bilateral facial and abducens palsy.² Congenital unilateral facial palsy might be caused by an obstetric trauma; while facial palsy in the absence of trauma, unilateral or bilateral appears to be a genetic condition.³ Dysfunction of other cranial nerves, orofacial malformation, limb malformation and musculoskeletal system defect are commonly associated features. We describe a young child who presented congenital unilateral facial palsy with hearing impairment after birth. Image study showed absence of the right facial nerve as well as hypoplasia of the pons and cerebellum of the same side.

2. Case Report

This male infant was born at 40 weeks of gestational age and birth weight 3100 g via normal vaginal delivery without perinatal or prenatal insults. Asymmetry of facial expression was found after birth, especially when he was crying. Feeding was relatively unremarkable. The patient was brought to our outpatient...
department (OPD) when he was 7 months old. The growth was within normal limits, with he had a head circumference of 41 cm (below 5th percentile). Upon examination, he was alert but his eyes did not follow moving objects. Mildly unsteady head control was still present. According to his mother’s description bilateral ocular movement in all directions seemed acceptable. There was marked incomplete closure of the right eye, a loss of right frontal wrinkling and the nasolabial fold, with the angle of the mouth deviating to the left when he was crying. Axial hypotonia and increased muscle tone over bilateral limbs, especially the lower ones, were found. There were neither other dysmorphic features, nor any extremity abnormality. The heart sound, liver size and abdomen were grossly intact and the family history didn’t reveal any hereditary diseases. We arranged a brain sonogram, which showed unremarkable findings. The auditory brainstem response showed no response bilaterally at 85 dB. Since there were no respiratory or feeding problems, we suggested rehabilitation programs for the patient’s congenital facial palsy, developmental delay and hearing impairment. A detailed brain MRI study was recommended after 1 year of age. When he was 1 year and 6 months old, a brain Magnetic resonance imaging (MRI) was performed which revealed hypoplasia of the right cerebellum and pons (Figure 1) and an absence of the right facial nerve (Figure 2). At this point he was more responsive and able to react to our stimulus, but still with of lack of meaningful words. He could walk without support, but had wide-based gait. Incomplete closure of the right eye and absence of the right frontal wrinkle persisted, but some degree of the nasolabial fold had developed along with a mouth angle deviation to the right. A neurodevelopmental test using the Bailey scale of infant development (second edition) showed the mental developmental index (MDI) was 52 and the performance developmental index (PDI) was below 50.

3. Discussion

In 1888, Möbius drew attention to patients with congenital non-progressive bilateral facial and abducens
palsy. The eponym Möbius syndrome has since been used for this condition. Isolated congenital facial palsy and the extended phenotype of congenital facial palsy with ocular muscle weakness, with or without craniofacial dysmorphisms and congenital abnormalities of the extremities, were criteria for the diagnosis of Möbius syndrome. To date, it is recognized as a syndrome of rhombencephalic maldevelopment with variable severity rather than an absence of cranial nerve nuclei. In the majority of the patients, we can observe a homogeneous clinical picture characterized by facial diplegia of the upper and lower facial muscles, bilateral abduction impairment, hypoglossia, craniofacial and limb malformations, and symptoms of the long tracts. In addition, hypotonia, developmental delay, hearing impairment, coordination dysfunction as well as other cranial nerve involvement presenting with feeding/respiratory problems are common associated symptoms. The radiologic features of Möbius syndrome are similarly variable, ranging from normal findings to variable degrees of hypoplasia of the brainstem, prevailing in the region of the pons, with or without intra-axial calcifications. A previous report demonstrated brainstem hypoplasia with straightening of the fourth ventricle floor, indicating an absence of the facial colliculus in the brain MRI.

Our patient presented obvious right facial palsy and hearing dysfunction since birth, and developmental delay and ataxic gait were evidenced as he grew up. The brain MRI showed hypoplasia of the right side of the pons and cerebellum and an absence of the right facial nerve. The clinical picture was compatible with the concept of a rhombencephalic maldevelopment. After physical therapy, mild active contraction in the right lower facial muscles was found. Collateral supply of other cranial nerves can be found in some situations. Although the facial nerve is absent, facial muscle function can recover to some degree after physical therapy. Other cranial nerves might aberrantly innervate some facial muscles, especially in the lower part of the face.

Teratogenicity is assumed to be an important factor in this syndrome, either by a genetic or ischemic mechanism. Genetic association has different cytogenetic loci in multiple studies including chromosome 1p22, 8q, 13q12.2-13, 10q, and 10q. The vascular hypothesis supposes a vascular event, which is the hypoperfusion or occlusion in the watershed zones of the lower brainstem. In conclusion, Möbius syndrome is a complex developmental disorder of the lower brainstem. Rather than a congenital absence of certain cranial nerves, it is clear that the associated deficiencies of the syndrome require a multidisciplinary approach by pediatricians, neurologists, ophthalmologists, genetic counselors, orthopedic, plastic and dental surgeons, audiologists, physical therapists, and speech therapists.

References