

**MOEBIUS SYNDROME WITH ATRIAL SEPTAL DEFECT**

Dear Sir,

Moebius syndrome comprises congenital facial nerve palsy with or without palsy of the other cranial nerves and the associated organ system malformations. Cardiovascular abnormalities, albeit rare, have been previously described. We describe a case of Moebius syndrome with an ostium secundum-type of atrial septal defect which, to the best of our knowledge, has not been reported in the existing literature.

A two-year-old male child, born to non-consanguineous parents, presented with delayed speech and motor milestones. The mother was concerned about the child's excessive but purposeless movements of the limbs and his facial asymmetry. He was born at term by spontaneous vaginal delivery. There was no history of birth asphyxia. The birth weight was 2.4 kg. At the time of admission, he weighed 13 kg and measured 87 cm in length with an appropriate upper to lower segment body ratio. The head circumference was 45 cm. He was dysmorphic in appearance; the dysmorphism comprised a peculiar mask-like, expressionless facies, an absent right depressor anguli oris, bilaterally low-set dysplastic ears, a stenotic right external auditory meatus, left-sided microphthalmia with a small palpebral aperture and bilateral lateral rectus palsy. A midline cleft palate was also noted. There was a depression overlying the right side of the chest.

Synbrachydactyly of the right upper limb was present. The left upper limb and both the lower limbs were normal (Fig. 1). The assessment of the individual domains of the developmental milestones revealed the following: gross motor at nine months, fine motor at 12 months, cognition at ten months, language at eight months, social/emotional awareness at eight months and feeding at 18 months. Aural screening using the brainstem-evoked response audiometry and visual screen utilising the visual-evoked potential were normal. A left-sided congenital hydrocoele was also present. An ejection systolic murmur of Grade III/VI was appreciated in the pulmonary area. The first heart sound was normal with a wide and fixed split-second heart sound. The purposeless movements did not conform to any particular type, such as chorea and athetosis. The neurological examination revealed the presence of bilateral facial palsy with the absence of the depressor anguli oris on the right side. The other systems were normal.



**Fig. 1** Photograph shows mask-like facies with an absent pectoralis (right) and synbrachydactyly (right) in a two-year-old boy.

The chest radiograph was non-contributory. The 2-D echocardiography done to detect the presence of systolic murmur showed a 6-mm ostium secundum-type of atrial septal defect. Ultrasonography of the chest showed the absence of a section of the pectoralis major muscle over the right chest wall. Magnetic resonance imaging of the brain was normal. Based on the abovementioned findings, a diagnosis of Moebius syndrome with an ostium secundum-type of atrial septal defect was made.

Moebius syndrome is a spectrum of congenital malformations, which essentially comprises a complete or partial facial nerve palsy, which may be associated with other cranial nerve paralysis. The associated neurological abnormalities may include mental retardation, speech problems, deafness and autism. The musculoskeletal abnormalities may include syndactyly, brachydactyly, polydactyly, talipes, arthrogryposis, Klippel-Feil anomaly, hypoplasia/aplasia of the radius, ulnar metacarpals and phalanges, and absent/hypoplastic latissimus dorsi, pectorals, serratus anterior and the intercostal muscles.<sup>(1)</sup> Goldenhar syndrome was excluded by the absence of vertebral abnormalities. The absence of coloboma, choanal atresia and microphallus ruled out CHARGE association. The phenotype of the child matched that of Poland syndrome, except for the presence of bilateral facial nerve palsy, which suggested Moebius syndrome.

The most commonly-accepted theory of embryopathogenesis is the vascular theory. An insufficient vascular supply in early gestation may explain the spectrum of malformations observed. A unifying theory to explain the findings of Moebius syndrome, Poland anomaly and Klippel-Feil defect, was proposed by Bavinck and Weaver.<sup>(2)</sup> They suggested that the interruption in the development of the subclavian artery and its tributaries (including the basilar, vertebral and internal thoracic arteries) in early gestation may be causative (subclavian artery disruption sequence). The cardiac anomalies reported thus far in Moebius syndrome include isolated dextrocardia,<sup>(3)</sup> dextrocardia with ventricular septal defect,<sup>(4)</sup> transposition of the great vessels,<sup>(5)</sup> ventricular septal defect<sup>(6)</sup> and a supracardiac variety of total anomalous pulmonary venous connection.<sup>(7)</sup> Our patient had an ostium secundum-type of atrial septal defect, which has not been reported thus far.

Yours sincerely,

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