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Case Report

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POLAND-MOBIUS SYNDROME-THE POSSIBLE COMMON DEVELOPMENTAL PATHOGENESIS

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Abstract

Introduction: Mobius syndrome is a rare disorder characterised by congenital non-progressive unilateral or bilateral VI and VII cranial nerve paresis with or without involvement of other cranial nerves. Poland syndrome is another rare congenital abnormality of the chest wall, characterised by unilateral partial or total absence of the pectoralis major muscle and ipsilateral symbrachydactyly. An infant with features of both Mobius syndrome and Poland syndrome is presented in this case report. **Case Report:** Two days old baby was brought with complains of drooling of milk on feeding. On examination, baby had left sided facial palsy, bilateral abducent nerve palsy, absent right pectoralis major muscle and homolateral symbrachydactyly. Baby also had congenital talipes equino varus of left foot. This work conforms to the values laid down in the Declaration of Helsinki (1964). The protocol of this study has been approved by the relevant ethical committee related to our institution in which it was performed. All subjects gave full informed consent to participate in this study. **Discussion:** The etiology of these syndromes is multifactorial. Both these syndromes have been attributed to vascular disruption sequence. The recurrent presentation of combination of these two syndromes provides further evidence to a possible common developmental pathogenesis referred to as the subclavian artery disruption sequence.

KEYWORD: Mobius syndrome characterized pectoralis major muscle and homolateral symbrachydactyly.

INTRODUCTION

Mobius syndrome

Unilateral or bilateral loss of ocular abduction, unilateral or bilateral complete or incomplete facial weakness, primary or secondary congenital anomalies of the extremities and possible other involvement of the branchial musculature are the principal clinical features of the Mobius syndrome[1], which was first described by Albrecht Von Gaefe and Saemisch in 1880.[2] Incidence of Mobius syndrome is 1: 50,000 to 100,000.[3]

Poland syndrome

Poland syndrome is a rare congenital abnormality of the chest wall, which was described for the first time by Alfred Poland in 1841, characterised by unilateral partial or total absence of the pectoralis major muscle and ipsilateral symbrachydactyly. The incidence of Poland syndrome is 1 per 32,000 live births. Boys are affected more than girls. The syndactyly is usually in the right hand as in this patient. The inheritance of Poland syndrome is entirely sporadic.[4,5]

The combination of Poland and Mobius syndrome is far rarer (first case to be reported from India), with an estimated prevalence 1:500,000.[4,5,6,7] An infant with features of both the Mobius syndrome and the Poland syndrome is presented in this case report and the developmental pathogenesis is discussed.

CASE REPORT

A two days old male baby was admitted with difficulty in feeding and deviation of mouth while crying. He is the first child born to a 26 years old lady. Baby was born at 38 weeks of gestation by spontaneous vaginal delivery in a hospital. There was no history of prolonged labour, instrumental delivery or birth asphyxia. The entire pregnancy was uncomplicated and there was no history of any drug ingestion, smoking/teratogens intake during pregnancy. His parents were not related and were healthy. There was no history of previous abortions or still births. There was no history of any dysmorphism in the family.

On examination, all growth parameters were above 10th centile; (weight: 2.5 kg, length: 51 cm, HC: 34 cm). Examination revealed several congenital abnormalities. He had hypertelorism, depressed nasal bridge, epicanthal folds, low set ears and deviation of mouth towards right side on crying. Neither eye could be abducted beyond the mid line. (Figure 1). The baby had drooling of milk while feeding. The breast bud on right side was imperceptible. There was decreased muscle mass over

the right side of chest wall, right sided symbrachydactyly (Figure 2) and congenital talipes equino varus of the left foot (Figure 3). Rest of his examination including other cranial nerves and limbs was normal.

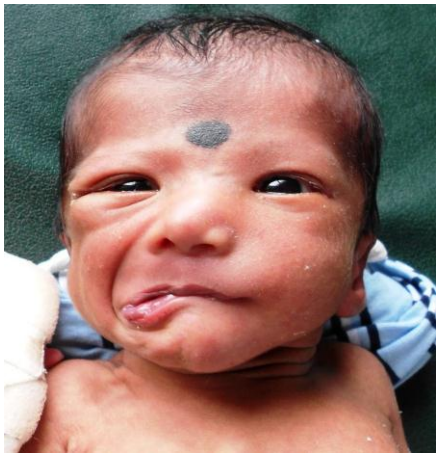

Figure 1

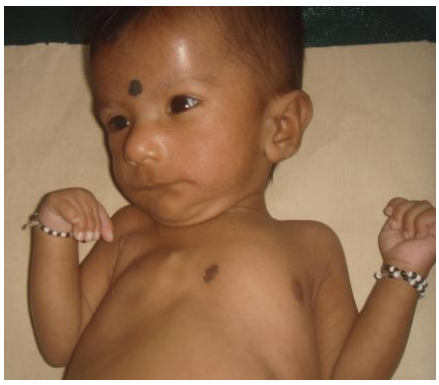

Figure 2

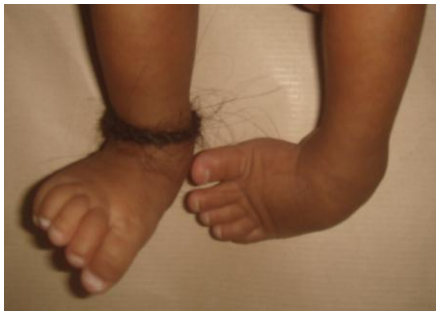

Figure 3

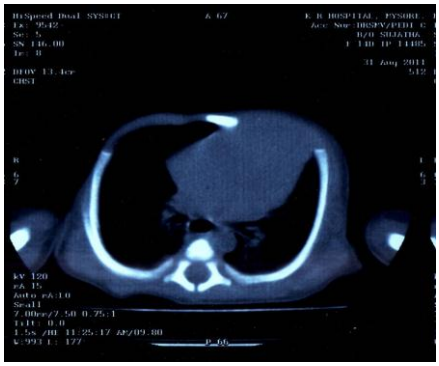

Figure 4

Both corneas were clear. He had no visceromegaly; the cardiovascular examination and genitals were normal. Other systemic examination was also normal.

Chest X-ray study showed lack of soft tissue development in the right pectoral region.

Computerised Tomography study of Thorax (Figure 4) showed decreased muscle mass over right side of chest wall suggestive of Poland Syndrome. MRI brain, echocardiography and abdominal ultrasound scan were normal. He was diagnosed as a case of Poland-Mobius syndrome on the basis of facial nerve palsy and bilateral VI nerve involvement along with associated limb abnormalities and absent pectoralis major muscle.

DISCUSSION

Mobius syndrome was defined by Paul Julius Mobius, a German neurologist, in 1888, who reported patients with congenital, non-progressive, bilateral VII and VI nerve palsy. Primary criteria for diagnosing Mobius syndrome are facial palsy with impairment of ocular abduction. Dysfunction of other cranial nerves and orofacial abnormalities are commonly associated but not necessary for the diagnosis.[7,8]

There have been around 500 cases of Mobius Syndrome described in the world medical literature; some of them have received surgical treatment. Mobius syndrome has also received other names such as congenital nuclear aplasia, childlike nuclear aplasia, oculofacial congenital paralysis and facial diplegia.[9]

Abramson et al. classified and graded the syndrome on the basis of clinical findings of cranial nerves and musculoskeletal anomalies using the acronym CLUFT (Cranial nerves, 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, longitudal, 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.[8]

Mobius syndrome can also be associated with diseases and disorders, such as hypogonadotropic hypogonadism[10], Poland syndrome[10], cataplexy[11], bilateral vocal cord paralysis[4,12], total anomalous pulmonary venous connection[13], narcolepsy and parasomnias.[14]

Etiology of Mobius syndrome is multifactorial with several proposed theories. The most supported one is transient ischemia or hypoxic insult to fetus, which is associated to a polygenic factor or to an interaction with teratogenic agents, such as Misoprostol.[3] Most cases of Mobius syndrome are sporadic but familial cases are also reported with autosomal dominant, auto-recessive and X-

linked recessive modes of inheritances. In addition to the involvement of chromosome 13, other loci map to 3q21-q22 and 10q21.3-q22.1.[15]

Poland syndrome was first described in 1841 by Alfred Poland who dissected the body of a criminal with unilateral symbrachydactyly associated with ipsilateral aplasia of the sternal head of the pectoralis muscle. Other anomalies associated with this syndrome include hypoplasia of the hand and forearm, hypoplasia of the breast, bilateral epicanthus, equinovarus, Mobius syndrome, upper musculature anomalies, rib cage deformities, and an absence of the ipsilateral kidney. Boys are affected more than girls. The syndactyly is usually in the right hand as in this patient.[4.5,6,7]

Many associations were described between this syndrome and a variety of diseases or other syndromes, such as Adams-Oliver syndrome which is characterised by defects of the limbs, scalp, and skull[16], Goldenhar syndrome which is facio-auriculo-vertebral syndrome[17], Wilms tumor[18], breast carcinoma[19], lung cancer[20], neuroblastoma[21], lymphoma[22], arterial septal defects[23] and eye abnormalities.[24]

The inheritance of Poland syndrome is entirely sporadic.[4] Despite the unclear pathogenesis of this syndrome, it is mostly believed that in the embryonic development, during the sixth week of pregnancy, a momentary stoppage or reduction in the circulation of the thoracic artery or one of its peripheral ramifications primes the pathogenic mechanism of the syndrome and results in different degrees of syndrome severity, depending on the length and intensity of the vascular interruption.[16,25]

Bavnik and Weaver suggested that Poland, Klippel-feil, and Mobius sequences should be grouped together on the basis of similar developmental pathogenesis referred to as the subclavian artery disruption sequence.[26]

According to this hypothesis, these conditions are caused by interruption of the early embryonic blood supply in the subclavian or vertebral arteries or their branches during the fifth through eighth weeks of fetal development. Vascular disruption in the subclavian artery may be due to internal obstruction of the vessel from edema, thrombi, emboli or spasm or to obstruction secondary to external pressure on the vessel from tissue edema, local hemorrhage, cervical rib, aberrant muscle, amniotic band, tumor, or embryonic intrauterine compression.[26] Exogenous factors (e.g., drugs, chemicals, generalised hypoxia, hyperthermia) may cause premature regression of vessels or a delay in vessel development.[26,27] Drugs implicated in the pathogenesis of these conditions are misoprostol, thalidomide and cocaine.[6, 3,28,29]

The recurrent presentation of combination of these syndromes supports the possibility of a common developmental pathogenesis referred to as the subclavian artery disruption sequence.[4,5,26,27,28] Also, based on the similar developmental pathogenesis these two syndromes may be grouped together and called as 'Poland-Mobius syndrome'[7] Further genetic and pathological studies of the involved blood vessels could lead to a better understanding of these syndromes.

Treatment is conservative consisting of reconstruction and plastic surgery of the breast, hands and fingers for cosmetic purposes. If the infant has feeding difficulties, physical and occupational therapy is required. Surgery can correct ophthalmological problems and smile surgery can be done which includes muscle transfer from the thigh to the corner of the mouth. Surgery is ideally performed in patients just before they reach school age at 4-5 years.[6,30] Fused fingers are separated as early as possible and breast or chest implants may be used after full physical development has been reached. Today, bioengineered cartilage can be implanted to help and give the chest a more normal appearance.[31]

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