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■ Case report ■

The first Korean case of poland-Möbius syndrome associated with dextrocardia

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= Abstract =

Poland syndrome is characterized by unilateral absence or hypoplasia of the pectoralis muscle and variable degree of ipsilateral hand anomalies. Möbius syndrome is a congenital neurological disorder characterized by complete or partial facial paralysis. Although the pathogeneses of these diseases are not well-characterized, diminished blood flow to the affected side is thought to play a role. A male infant weighing 2.670 g was born at 38+3 weeks of gestation with left facial paralysis, left chest wall defect with dextrocardia, and symbrachydactyly between the second and third fingers. The combination of Poland-Möbius syndrome is rare, and only 2 cases associated with dextrocardia have been reported worldwide. Here, we report the first case of Poland-Möbius syndrome associated with dextrocardia in Korea. **(Korean J Pediatr 2009; 52:1388-1391)**

Key Words : Poland-Möbius syndrome, Dextrocardia

Introduction

Poland syndrome consists of unilateral absence or hypoplasia of the pectoralis muscle, most frequently involving the sternocostal portion of the pectoralis muscle and a variable degree of ipsilateral hand and digit anomalies[1]. Möbius syndrome is characterized by absence or underdevelopment of the nerves that control the 6th, and 7th cranial nerves which manifests as facial paralysis at birth[5]. Oculomotor abnormalities cause the inability to abduct the eyes beyond midpoint. The combination of Poland and Möbius syndrome is described in the literature with an estimated prevalence of 1:500,000. These two syndromes should be grouped together on the basis of similar developmental pathogenesis referred to as the subclavian artery disruption sequence[9]. The combination of Poland-Möbius syndrome is rare and only two cases associated with dextrocardia have been reported worldwide. We hereby report the first neonatal case of Poland-Möbius syndrome associated with dextrocardia in Korea with the review of literature.

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Case report

A 2.670 g male infant was born at the gestational age of 38 weeks and 3 days to a 30-year-old gravida 2 para 1 woman by Cesarean section at a local obstetric clinic and was transferred to our hospital for evaluation of congenital anomalies. The mother was a non-smoker and had not taken alcohol or drugs during pregnancy. She had been diagnosed with IgA nephropathy two years prior to her pregnancy and has taken herbal medicine for 1 year. She had oligohydroamnios during the pregnancy. There was no other significant family history.

The birth weight 2,670 g (25th percentile), height 48 cm (25-50th percentile), and head circumference 33 cm (25-50th percentile) were checked. The vital signs were stable on admission.

On admission to the nursery, several congenital abnormalities were observed. He had left facial paralysis with difficulty in blinking and a downward retracted lip. And he was unable to abduct the left eye beyond midpoint (Fig. 1). The left chest wall had a defect in the muscular layer with

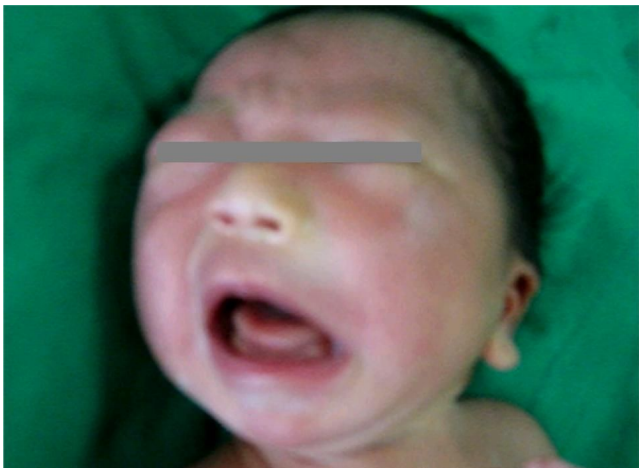

Fig. 1. Left facial paralysis is apparent when the child is crying.

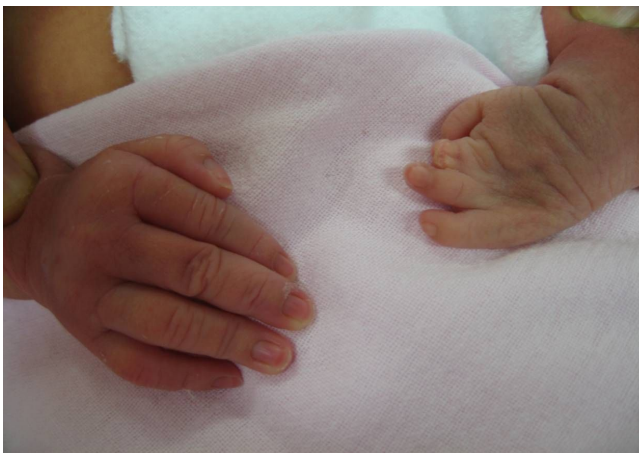

Fig. 2. Hypoplastic left hand with symbrachydactyly of the second and third fingers.

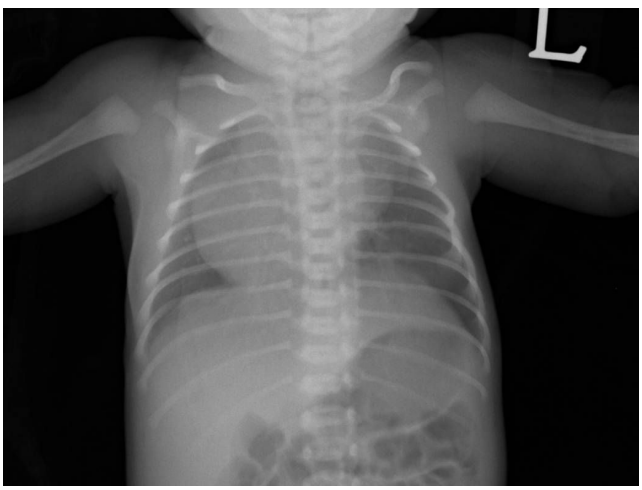

Fig. 3. Chest pulmonary angiography (PA) shows dextrocardia.

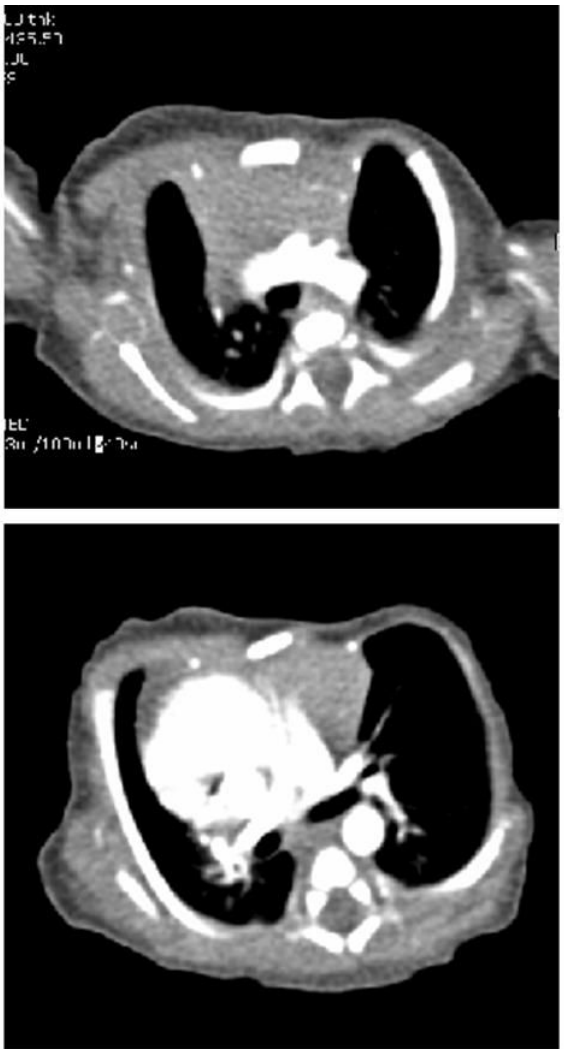

Fig. 4. Chest computed tomography (CT) scan reveals aplasia of the pectoralis major muscle, very thins or aplastic pectoralis minor muscle, and serratus anterior muscle. Irregular bulging of the anterior aspect of the left lung is noted within the deformed left thorax.

herniation of the underlying lung and nipple hypoplasia (Fig. 2). Depression of the chest wall and bulging of the herniated lung was seen with respiration. The heart sound were audible on the right side without murmur.

Dextrocardia with otherwise normal heart structure was diagnosed on the echocardiography. Symbrachydactyly between the 2nd and 3rd fingers on the left hand and toe anomaly on the left foot were noted (Fig. 3, 4). Radiologic findings demonstrated aplasia/hypoplasia of the phalanges and metacarpal bones of the left hand (Fig. 5). Chest PA, ultrasonography and chest CT revealed isolated dextrocardia with aplasia of the pectoralis major, minor, and serratus anterior muscles along with irregular bulging of the anterior

aspect of the left lung between the ribs (Fig 6, 7, 8).

Neonatal screening test for inborn errors of metabolism, chromosomal analysis of the peripheral blood, brain and abdominal sonography and auditory evoked test were normal. The patient is of good growth and development without feeding difficulty at seven months of age.

Discussion

Poland syndrome was first reported by Alfred Poland[1], a British anatomist in 1841 as the association of aplasia of the sternal head of the pectoralis major muscle with ipsilateral symbrachydactyly. Associated anomalies such as hypoplasia of the hand and forearm, hypoplasia of the breast, absent nipple, partial or total absence of other upper segment musculature, rib deformities, leukemia or radioulnar synostosis have been reported. In British Columbia, McGillivray and Lowry[2] found an incidence of Poland syndrome of 1 per 32,000 live births. It has even been estimated that 10% of patients with syndactyly of the hand have the Poland sequence. Poland syndrome is more common in males by three times and 75% involve the right side[3].

Möbius syndrome was first reported by von Graefe in 1880[4], by Paul J. Möbius in 1888[5]. It is a rare congenital neurological disorder (prevalence:1:50,000), which mainly related to the underdevelopment of the 6th, and 7th cranial nerves.

It is characterized by complete or partial facial paralysis and the inability to abduct the eyes beyond midpoint. Other associated malformations include congenital defects of the limbs such as arthrogryposis, limitations in the range of joint motion, multiplex congenita, motor delay from upper body weakness and mental retardation[6]. The cause of Möbius syndrome is unknown but intrauterine vascular, toxic, genetic and infectious factors have been proposed as the cause of a vascular disruption in the brain during prenatal development[7]. Möbius syndrome is classified into four groups according to the neuropathologic findings by Towfighi et al[8]. Type I includes those cases with hypoplasia to absence of the central brain nuclei, type II with peripheral nerve involvement, type III with destructive degeneration of the ventral brain nuclei and type IV with myopathy only.

The combination of Poland and Möbius syndrome is described in the literature with an estimated prevalence of 1:500,000. The association of these two relatively discordant syndromes arise from a common etiology. Bavinck and Weaver[9] presented that Poland, Klippel-Feil, and Möbius sequences should be grouped together on the basis of similar developmental pathtogenesis referred to as the subclavian artery disruption sequence.

The primary defect in the development of the proximal subclavian artery with early deficit of blood flow to the distal limb and pectoral regions, yielding partial loss of tissue in those regions is thought to be the main cause[10]. The two fold increase in incidence with maternal smoking supports the vascular pathogenesis. Use of drugs such as misoprostol used to promote uterine contractions and cocaine is known to increase the risk of Möbius syndrome. Furthermore, Bavink and Weaver[9] suggested that Poland, Klippel-Feil, and Möbius sequences may occur in various combinations in the same individual and the different pattern of defects depends on the specific area of diminished blood flow.

The majority of the cases are sporadic and the recurrence risk is negligible but reports of parent-to-child transmission as well as affected siblings born to unaffected parents were present. Marked variability in expression has been reported including two siblings in which one had the full Poland syndrome and the other only had absence of the pectoral muscle and in the other, only syndactyly of the hand[13]. The pathogenesis was not well-defined but Bouvet et al.[3] presented evidence of diminished blood flow to the affected side maybe in the proximal subclavian artery which cause distal limb and pectoral lesion underdevelopment.

Sugarman and Stark[11] reported a 5-year-old child with bilateral facial palsies with aplasia of sternal portion of the left pectoralis major and soft tissue syndactyly. In addition to theses malformations the child had dextrocardia. It was the first case for Poland-Möbius syndrome with dextrocardia. Since that paper, Bosch-Banyeras et al.[12] reported the second case of Poland-Möbius syndrome with dextrocardia. Our case is the first report of Poland-Möbius syndrome accompanying with dextrocardia in Korea. Conservative treatment consisting of reconstruction and plastic surgery of the breast, hands, and fingers is needed for cosmetic purposes and surgery is usually conducted after one year of age or before school age. If the infant experience difficulty in feeding, physical and occupation therapy is required. Surgery can correct ophthalmologic problems and smile surgery, muscle transfer from the thigh to the corner of the mouth leads to patient satisfaction.

한 글 요 약

Poland-Möbius syndrome 신생아 1례

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Poland 증후군은 일측 대흉근의 부재나 저형성과 다양한 동측 손기형을 특징으로 한다. Möbius 증후군은 완전 혹은 불완전의 안면마비를 특징으로 하는 선천적 신경학적 이상을 말한다. 이들의 병인은 명확히 밝혀지지 않았지만, 이환된 측의 혈류가 감소됨으로서 발생된다 여겨지고 있다. 2.670 g 남자아이가 38주 3일에 왼쪽 안면마비와 우심증이 동반된 왼쪽 흉벽 결손에 2번째, 3번째 손가락 합지증을 가지고 태어났다. Poland-Möbius 증후군은 드물며, 세계적으로 우심증과 관련된 증례는 단지 2례 밖에 없었다.

이에 우리는 우심증이 동반된 Poland-Möbius 증후군의 국내 첫 증례를 보고하는 바이다.

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