

Poland Syndrome in One Dizygotic Twin: A Case Report

Woo Hyun Yoo, M.D., Mi Hye Bae, M.D., Young Mi Han, M.D., Shin-Yun Byun, M.D., and Kyung Hee Park, M.D.

Department of Pediatrics, School of Medicine, Pusan National University, Busan, Korea

Poland syndrome, also known as pectoral aplasia-dysdactylia syndrome, is a rare anomaly, characterized by aplasia of the pectoralis major muscle with ipsilateral upper extremity anomalies. The cause of Poland syndrome is still unknown. Several twin patients were reported to propose a genetic or nongenetic hypothesis. We experienced a female baby showing absence of the pectoralis major muscle in a pair of mixed twin. The patient was referred to our hospital for evaluation of an asymmetric chest wall. She showed depression of the left chest wall, ipsilateral nipple hypoplasia, and axillary webbing. A chest CT scan showed aplasia of the left large pectoral muscle. Our patient had no abnormalities such as symbrachydactyly in an ipsilateral upper extremity. In comparison, a physical examination showed that the fraternal twin boy was completely normal.

Key Words: Poland Syndrome, Pectoralis Muscle Absence, Twin, Dizygotic

Poland syndrome, also known as pectoral aplasia-dysdactylia syndrome, is a rare anomaly, characterized by aplasia of the pectoralis major muscle with ipsilateral upper extremity anomalies.¹ In most cases, it is associated with absence of minor pectoral muscle, hypoplastic rib, pectus excavatum (funnel chest), pectus carinatum (pigeon breast), scoliosis, and ipsilateral breast and nipple hypoplasia. In shoulder joints, the disorder may also include absence or hypoplasia of serratus anterior, infraspinous, latissimus dorsi and trapezius muscle, hypoplastic/winged scapula and Sprengel's syndrome.^{2,3}

To the best of our knowledge, our patient is the first report of Poland syndrome in one of dizygotic twins

in Korea.⁴⁻⁷ Therefore, the authors herein describe a female baby showing absence of the pectoralis major muscle while the fraternal twin boy was completely normal.

Case Report

An 8-day-old female baby was referred to our hospital for evaluation of an asymmetric chest wall. The patient was born at 38⁺⁰ weeks gestation by cesarean section and weighed 2,800 g at birth. The patient was born as a second baby in dizygotic twins, and the first baby of nonconsanguineous parents. There was no family history of congenital anomaly.

The sibling was male and weighed 3,050 g at birth. The sibling was completely normal on physical examination. Physical examination of the patient revealed depression of the left chest wall, ipsilateral nipple hypoplasia, and axillary webbing, which is a wedge like fold between the left upper limb and anterior chest wall suggesting a defect of the pectoralis muscle (Fig. 1). Our patient had no abnormalities such

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Correspondence to: Kyung Hee Park, M.D.

Department of Pediatrics, Pusan National University School of Medicine, 179, Gudeok-ro, Seo-gu, Busan 49241, Korea

Tel: +82-51-240-7293, **Fax:** +82-51-248-6205

E-mail: kuma2002@hanmail.net

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Fig. 1. The patient showed depression of the chest wall, ipsilateral nipple hypoplasia and an axillary webbing.



Fig. 2. A chest CT scan showed aplasia of the left large pectoralis muscle (arrow).

as symbrachydactyly in an ipsilateral upper extremity.

Chest radiography and echocardiography showed no specific abnormal findings. A chest CT scan showed aplasia of the left large pectoralis muscle (Fig. 2). The patient's karyotype was normal 46, XX and screening for metabolic disease was unremarkable. Chromosomal microarray (Affymetrix® Cytoscan™ 750K array, Genome build : Hg 19) was performed and confirmed no microdeletion nor duplication of clinically important chromosomes.

Psychomotor development and growth of our patient was normal at 12 months of age. The patient is being monitored in the outpatient clinic to assess any functional problems.

Discussion

The cause of Poland syndrome is unknown, but genetic or non-genetic hypothesis has been proposed. Genetic origin is postulated due to patient reports of vertical transmission or affected siblings born to unaffected parents.^{8,9} However, most reported cases are sporadic while its exact cause is still subject to debate.⁹

Non-genetic hypothesis includes vascular pathogenesis, meaning that maldevelopment occurs due to diminished blood flow to the affected side in the proximal subclavian artery during the seventh week of gestation. Therefore it has been suggested that the degree of obstruction within the subclavian artery determines the severity of the anomaly.¹⁰⁻¹⁵

A combination of the blockade of various branches could lead to variant of Poland syndrome. Like our case, isolated aplasia of the pectoralis major muscle without ipsilateral upper extremity anomalies is thought to be a variant of Poland syndrome.¹⁴⁻¹⁶

The syndrome is present at an incidence of 1:30,000 with a higher frequency among males by a ratio of 3:1. And in 75% of the cases, it is located at the right hemithorax in the unilateral form.^{17,18} Interestingly, our patient was female and affected at the left side.

Among the previous patient reports, several twin patient cases have been reported, which might be helpful in clarifying the pathogenesis. Vaccari et al.⁹ reported on a couple of monozygotic twin girls who exhibited pectoral muscle hypoplasia. Both patients showed a de novo deletion of chromosome 11q12.3. Then they suggested that Poland syndrome might be due to genetic control. On the other hand, Poland syndrome in one identical twin was reported by Stevens et al.,¹⁹ who demonstrated only one affected

monozygotic twin, meaning that Poland syndrome is not determined by gene transmission.

Although unlike previous twin cases, our patient is a dizygotic and mixed twin, only one is affected and the other is completely normal. Therefore, the current case is in accordance with a previous report by Stevens supporting that Poland syndrome is sporadic. However further reports including twin cases are required in order to clarify the pathogenesis of Poland syndrome for genetic counseling and family planning.

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