CASE REPORT

Lung herniation: an uncommon presentation of Poland's syndrome in a neonate at birth

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SUMMARY

A term male infant was admitted to the neonatal intensive care unit with an asymmetric chest wall and a herniating mass through the left fourth intercostal space. While crying, the left fourth intercostal space revealed a mass which herniated on expiration and receded on inspiration. On auscultation, the heart sounds were audible on the right side of the chest. Systemic examination was otherwise unremarkable. A roentgenogram of the chest revealed dextrocardia and hyperlucent left lung fields. Echocardiogram showed dextrocardia with a structurally normal heart. A clinical diagnosis of Poland's syndrome was made based on the hypoplasia of the left pectoral muscles, ribs and nipple, dextrocardia and lung herniation. He was thriving well when reviewed at 2 years of age.

BACKGROUND

Poland's syndrome (PS) is a rare congenital anomaly, described as a combination of unilateral aplasia or hypoplasia of the sternal portion of pectoralis major muscle and less often pectoralis minor in an individual who also had syndactyly of the ipsilateral hand.¹ An essential feature of this syndrome is the absence of the sternal head of the pectoralis major muscle.²⁻³ Rib defects, dextrocardia and renal anomalies are uncommon in PS.²⁻⁴ Knowledge of the spectrum of defects seen in PS is important for the early diagnosis and appropriate intervention if required. We describe an interesting case, unique in its presentation, with lung herniation as the striking feature at birth. To the best of our knowledge, the left-sided PS with dextrocardia and lung herniation is unreported in the literature.

CASE PRESENTATION

A term male infant weighing 3100 g was born to a 22-year-old non-consanguineous Filipino mother by normal vaginal delivery. His Apgar scores were 9 at 1 and 5 min of life. The mother did not have antenatal scans and her medical and family histories were unremarkable. The infant's head circumference and length were age-appropriate.

At birth, an asymmetry of the chest wall (figures 1 and 2) with hypoplasia of the left nipple and depressed fourth left intercostal space were noted. While crying, a herniating mass was observed with expiration in the fourth left intercostal space (video 1). On palpation, there was softening of the third and fourth ribs towards the costochondral area with widening of the intercostal space and normal overlying skin. There was hypoplasia of the intercostal muscles and subcutaneous tissue in the fourth

Figure 1 Infant with Poland's syndrome showing the lung herniation on expiration.

intercostal space. Neither a thrill was felt nor a bruit was heard over the herniating mass. On auscultation, heart sounds were audible on the right side of the chest. He was haemodynamically stable with normal peripheral pulses and blood pressure. No other musculo-skeletal defects were noted. Systemic examination was otherwise unremarkable.

The baby was fed from day 2 of life and remained stable during the hospital stay before being discharged home in a stable condition on day 10 of life.

No musculo-skeletal deformities were found on parental examination; other systemic examinations were unremarkable.

INVESTIGATIONS

A chest roentgenogram revealed dextrocardia and hyperlucent left lung fields (figure 3). Echocardiography showed dextrocardia and a

Figure 2 Infant showing hypoplasia of the left pectoral muscles and areola. On expiration intercostal recession is noted.

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Video 1 Video shows the lung herniation on expiration and recession in inspiration.

structurally normal heart. Ultrasound of the fourth left intercostal space showed significant reduction in the bone density of the bordering ribs. Cranial, renal, hepatobiliary and spinal ultrasound scans were normal. Haemogram, renal and liver function tests were unremarkable.

A diagnosis of PS was made based on the hypoplasia of the left pectoral muscles and nipple, dextrocardia and lung herniation due to hypoplasia of left fourth intercostal muscles and bordering ribs.

TREATMENT

The infant was referred back to his country of origin. At the age of 2 years, except for the lung herniation during crying and occasional mild wheezy episodes, there were no significant medical problems. He had not undergone any surgical interventions. His growth and development were appropriate for his age.

Figure 3 X-ray of the chest with hyperlucent left lung fields and dextrocardia.

DISCUSSION

We present a case of PS with hypoplasia of the left chest wall, lung herniation and dextrocardia. PS is a rare congenital anomaly with sporadic presentation and is characterised by malformations of the chest wall due to hypoplasia of the pectoral muscles, nipple and ribs, with or without deformities of the ipsilateral upper limb and hand. Less often, latissimus dorsi and deltoid muscles are affected. Alfred Poland, a student-demonstrator in anatomy described this anomaly in 1841.¹ There were earlier reports of this anomaly by Lallemand (1826) and Froriep (1836). In 1895, Thompson summarised the full anatomical spectrum of PS.²

The incidence of PS is estimated to be 1 in 30 000 live births with the right side (75%) being affected more often than the left and the male-to-female ratio being 3 : 1.⁵

The aetiology of PS is still unclear. Several studies have suggested that genetic factors could play a role. Autosomal dominant inheritance and single gene defects have also been suggested as causes of PS.⁶ A widely accepted theory for the genesis of PS is the hypoplasia of the ipsilateral subclavian artery or one of its branches, caused by the interruption of the embryonic blood supply, at the end of the sixth week of gestation. Hence, PS is also known as the subclavian artery supply disruption sequence. Hypoplasia of the internal thoracic artery could result in the absence of the sternocostal portion of the pectoral muscles and hypoplasia of the brachial artery may result in hand abnormalities.⁴ Partial agenesis of two or more ribs during embryonic life and displacement of the heart to the right were observed by Torre *et al* in left-sided PS. So, they suggested that the mechanical factors during the time of organogenesis could explain the strong association between left-sided PS and dextrocardia.⁷

Clinical manifestations of PS are extremely variable. All patients with PS will have aplasia/hypoplasia of the sternocostal head of pectoral muscles.¹⁻³ Ipsilateral syndactyly was reported in 66% of patients with PS and the middle three fingers were the most affected.⁸ The areola and nipple are usually hypoplastic and elevated or even absent.² The overall incidence of thoracic cage defects in PS is 25%. PS is associated with hypoplasia of the II to IV or III to V ribs and cartilages.⁸ Hypoplasia or aplasia of the sternal portion of I and III ribs with severe chest wall deformities occurs in about 11–25% cases of PS.⁹ The rib defects seen in association with dextrocardia in PS are almost always on the left side with only 15% being reported on the right.¹⁰ The association of PS with dextrocardia, as seen in our case, has been reported in only 5.6% of 144 cases of PS that have been published.^{2-4,10} Isolated dextrocardia is always associated with cardiac defects, but in PS with dextrocardia the heart is always normal.¹¹ Lung herniation is extremely rare in PS and has been reported in only 9% of cases. Some of these cases with severe rib defects had paradoxical respiratory movements.¹² The infant in this case had a left-sided PS with isolated dextrocardia and structurally normal heart, with the rare presentation of lung herniation at birth.

The hand defects in PS are always ipsilateral to the side of the chest deformity. It varies from shortness of the middle phalanges (45%) to syndactyly or synbrachydactyly (89%) and rarely ectrodactyly. Hypoplasia of the upper limbs including the wrist and hands (89%), forearm (37%) and arm (7%) have been reported.²⁻³ The infant in this report did not have limb defects. Axillary web (7%), unilateral renal agenesis and duplication of urinary collecting systems were reported less often in PS.¹³ PS manifests mostly as an isolated anomaly, but there have been reports of this syndrome being associated with other anomalies like Moebius syndrome, Sprengel's deformity and Klippel-Feil

syndrome and malignancies like leukaemia, non-Hodgkin's lymphoma and lung cancer.^{2 4 14}

In infants, surgical treatment is indicated if a progression in the unilateral depression of the chest wall or a lack of adequate protection of the lungs and heart with paradoxical movement of the chest wall is noted. Surgical treatment involves the correction of ribs and cartilages using split rib grafts and staged soft tissue reconstruction using latissimus dorsi muscle.²

PS is generally sporadic and rarely inherited. Proband's all close relatives should undergo a thorough examination to look for the deformities seen in PS. There is no increased risk of recurrence of PS in families with only one affected child and normal parents as seen in our case. A risk of up to 50% must be foreseen if the immediate family member has any of the stigmata of PS.^{14 15}

Learning points

- ▶ Poland's syndrome (PS) is a rare congenital anomaly.
- ▶ Lung herniation is extremely rare and can be the presenting feature in PS.
- ▶ Dextrocardia and rib defects are always seen in left-sided PS. Isolated dextrocardia seen in PS is not associated with cardiac defects.
- ▶ Lung herniation due to rib defects may need early surgical intervention using split rib grafts to protect the heart and lungs.
- ▶ PS is mostly sporadic and less often reported as familial. A detailed examination of the proband's immediate relatives is recommended.

Contributors SC made the clinical diagnosis and conceptualised the idea of submitting as a case report, edited manuscript and finalised, and provided the video

clip. KGR wrote the basic draft, searched for the appropriate references and revised the manuscript. DA was involved in writing the case report and editing the manuscript; AR was involved in writing the case report, provided the clinical pictures and obtained the consent form from the father of the patient.

Competing interests None.

Patient consent Obtained.

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