Poland anomaly with contralateral ulnar ray defect

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Abstract

We report an atypical case of the Poland anomaly. Unreported features are that the hand abnormality is on the contralateral side to the chest wall defect, there is an ulnar ray predominance, and lack of syndactyly.

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The Poland anomaly comprises unilateral absence of the pectoralis major muscle with an ipsilateral hand defect.¹⁻⁸ We report a patient with atypical and previously unreported features.

Case report

The proband was the first child of healthy, non-consanguineous parents (father aged 27 years, mother aged 26 years). There was no family history of congenital malformations. Pregnancy was normal until the onset of pre-eclampsia at 32 weeks' gestation. The mother had not been taking oral contraceptives and there was no other history of maternal drug ingestion in pregnancy. Delivery was at 35 weeks' gestation by normal vaginal delivery, birth weight 2370 g (25th centile). Nasal continuous positive airway pressure was required for respiratory distress syndrome for 48 hours, but thereafter his progress was uneventful.

His abnormalities comprised a right sided chest wall defect and a left sided upper limb defect. There was absence of the sternocostal head of the right pectoralis major muscle, with a normal clavicular head and no axillary web (fig 1). The anterior portions of the right third

and fourth ribs were impalpable, associated with a depression of the chest wall in this area and paradoxical movement of the chest wall with respiration. The nipples were normal and there was no winging of the scapulae. The right hand was normal. The left arm was hypoplastic and shortened, especially the forearm. The left thumb and index finger were normal but the left middle finger was hypoplastic. The left little and ring fingers were absent (fig 2). At the age of 5 months, he is asymptomatic, growing and developing normally, and he has no detectable cranial nerve abnormalities.

Radiographs taken at the age of 2 days showed absence of the anterior ends of the third and fourth ribs on the right side and a hypoplastic left radius and ulna as well as absence of the left ring and little fingers.

Discussion

The clinical features of the Poland anomaly are variable in severity⁷ and in some cases the chest wall defect is not accompanied by any limb abnormality.^{4,7} The limb deformity typically comprises shortening of the fingers with cutaneous syndactyly of the index and middle fingers.^{4,7} In more severe cases, the fingers are all markedly shortened and webbed. Occasionally the index, middle, and ring fingers are completely absent with preservation of the little finger and the thumb.^{5,7}

The atypical features in this case are the lack of syndactyly, the ulnar predominance, and the contralateral side of the limb defect. These features represent an extension of the previously recognised spectrum of the Poland anomaly.

There is a recognised association between the Möbius syndrome and the Poland anomaly.² This is relevant to the present case because the Möbius syndrome has been reported in a child with an absent pectoralis major muscle on the right side and hypoplasia and syndactyly of the fingers on the left hand.⁹ The patient also had absent fingers on the right hand with a hypoplastic thumb. The case reported here had no features of the Möbius syndrome.

The aetiology of the Poland anomaly is uncertain. Bouvet *et al.*⁵ using plethysmography, showed a significant decrease of the velocity of the systolic increase in the arterial volume in affected arms of eight children with the Poland anomaly compared to eight control children with normal limbs. It was suggested that hypoplasia of the ipsilateral subclavian artery was the origin of the malformation.

Bouwes Bavinck and Weaver¹⁰ have

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Figure 1 A right sided chest wall deformity with absence of the sternocostal head of the pectoralis major muscle.

Figure 2 A left-sided hand abnormality with absent little and ring fingers, hypoplastic middle finger, and normal thumb.

suggested the term 'subclavian artery supply disruption sequence' (SASDS) to explain a common pathogenesis in Poland, Klippel-Feil, and Möbius syndromes, isolated terminal transverse limb defects, and the Sprengel anomaly. They propose that interruption of the early embryonic blood supply in the subcla-

vian arteries or vertebral arteries at about 6 weeks' gestation may produce predictable patterns of defects depending on where the restriction of blood flow occurs. However, a case such as the present one, in which the contralateral arm is affected, is not consistent with a vascular theory. It seems more probable that any hypoplasia of the subclavian artery is merely one feature of the upper limb hypoplasia, and there is no evidence for a causal relationship.

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