

Poland syndrome: neonatal presentation with axillary pterygium

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DESCRIPTION

Poland syndrome is a congenital syndrome, described for the first time in 1841 by Sir Alfred Poland.^{1 2} The estimated incidence is 1/30 000 to 1/32 000 live births,² predominantly affecting men, varying from 2:1 to 3:1.¹ The defect is more frequently unilateral and on the right side (75%).¹ The majority of cases are sporadic.^{1 2} However, there have been reports of familial cases.^{1 2}

We describe a female infant born at 38 weeks, second child of a 37-year-old mother with chronic high blood pressure and no relevant family history. Antenatal ultrasounds were described as normal. Maternal serologies (Venereal Disease Research Laboratory (VDRL), hepatitis C virus (HCV), hepatitis B virus (HBV), HIV and toxoplasmosis) were negative with immunity to rubella. Third trimester urine culture and streptococcus group b (GBS) were negative.

Uncomplicated, eutocic delivery with Apgar score 9/10/10. Birth weight was 3185 g (P₅₀), length 47,5 cm (P₁₅₋₅₀), head circumference 34 cm (P₅₀).

Physical examination at 15 hours of life showed right thoracic asymmetry and axillary pterygium related to *pectoralis* muscle agenesis (figure 1), posteriorly confirmed by ultrasound. There were



Figure 2 Right thoracic asymmetry.



Figure 1 Axillary pterygium related to *pectoralis* muscle agenesis.

Learning points

- ▶ Patients with Poland syndrome present with a spectrum of defects that includes a wide phenotypical variability.
- ▶ These patients should be followed up strictly, with special attention to pulmonary and muscle functions, as well as breast development. Subsequent surgical interventions may be necessary, including breast reconstruction in puberty.
- ▶ Early and accurate diagnosis allows adequate treatment and follow-up, as well as timely surgical correction.

no other associated clinical defects, namely rib cage or upper limb abnormalities. Chest X-ray and additional studies including echocardiogram, brain, abdominal and renal ultrasounds were normal.

She was discharged at 48 hours of life, maintaining a regular follow-up by a multidisciplinary team (physiatry, plastic surgery and pneumology). Currently at 22 months old, thoracic asymmetry with reduced right-sided subcutaneous fat and axillary pterygium are evident (figure 2). Ultrasound scan confirmed a residual sternoclavicular portion



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of the *pectoralis major* muscle, conditioning the tendinous axillary band. Surgical release will be considered when functional limitations occur. Future reconstructive breast surgery may also be an asset.

Poland syndrome is typically characterised by absence or hypoplasia of the *pectoralis major* muscle (mandatory feature), absence or hypoplasia of the *pectoralis minor* muscle, abnormalities of the chest wall, hypoplasia or absence of the nipple and ipsilateral upper extremity abnormalities.¹⁻³ Phenotypical presentation is wide and multiple combinations of these clinical defects are possible. In this case, the defect is isolated *pectoralis* muscle agenesis, representing the minimal form or type 1, according to recent classification of Poland syndrome.²

The pathogenesis is unknown, the most accepted hypothesis points to a dysembryological process caused by a vascular defect of the subclavian artery around the sixth week of pregnancy leading to an early insufficient blood flow to the chest and limb.¹⁻³

In most cases, functional disability is mild, treatment relies mainly in surgery and the main goal is to improve the symmetry of the chest wall and to correct breast hypoplasia.¹⁻³

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