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Poland syndrome – Atypical neonatal presentation

Síndrome de Poland – Apresentação neonatal atípica

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Keypoints

What is known

- Poland syndrome is a rare congenital disease characterized by the absence or hypoplasia of the pectoralis major muscle.
- The condition is benign, non-progressive, and may be asymptomatic, depending on the clinical defects.

What is added

- Patients can present with a wide phenotypical presentation and multiple combinations of defects.
- Although defects are more frequent on the right side, left-side and bilateral defects are also reported.
- Early diagnosis allows for appropriate treatment and psychological follow-up is also necessary due to the visible physical malformation.

Poland syndrome (PS) is a rare congenital condition. The estimated incidence is 1:30,000 live births[1,2]. Most cases are sporadic, with a higher prevalence in males[3]. PS diagnosis is clinical and the cardinal feature (essential for diagnosis) is the agenesis or hypoplasia of the pectoralis major muscle[3]. Other associated features include anomalies of the thoracic cage, agenesis or hypoplasia of the breast, areola and nipple, abnormalities of the upper limb and shoulder, genitourinary malformations, cardiac malformations, and hepatic or biliary tract malformations[3].

We describe a male infant born at 40 weeks and 5 days. The parents are not consanguineous and have no other children. The mother is a 25-year-old woman with generalized anxiety disorder, medicated with valerian root extract and ethyl loflazepate, and no other relevant family history.

Antenatal ultrasounds were described as normal. Maternal serologies were negative. The third-trimester urine culture and group B streptococcus test were negative. Assisted birth with vacuum extraction. Apgar score 8/9/10. Birth weight was 3010 g (P15-50; 0 < SD < –2 [WHO Growth Charts]), length was 50 cm (P50; SD 0 [WHO Growth Charts]) and head circumference was 36 cm (P85-97; 1 < SD < 2 [WHO Growth Charts]).

The newborn was admitted to the Special Neonatal Care Unit (UCEN) four hours after birth, with transient tachypnea of the newborn. Physical examination showed left thoracic asymmetry, hypoplastic homolateral nipple (Fig. 1), left hand brachydactyly (Fig. 2) and abnormal implantation of the 5th toe, bilaterally. There were no other associated congenital anomalies.

Thoracic ultrasound showed anterior thoracic muscle plan asymmetry and a left-hand X-ray confirmed the agenesis of the distal phalanx of the 2nd finger and hypoplasia of the middle phalanges of the 2nd and 3rd fingers (Fig. 3). A transthoracic echocardiogram revealed the median position of the cardiac apex, by the pushing of the left rib cage. Brain, abdominal, renal, and pelvic ultrasounds were normal.

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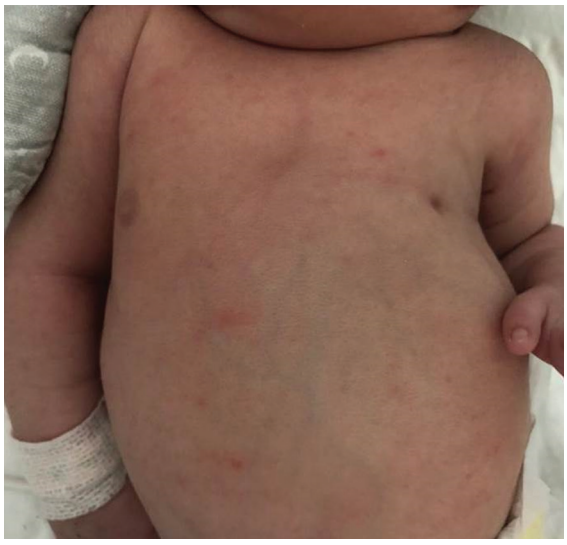

Figure 1. Left thoracic asymmetry and hypoplastic nipple.

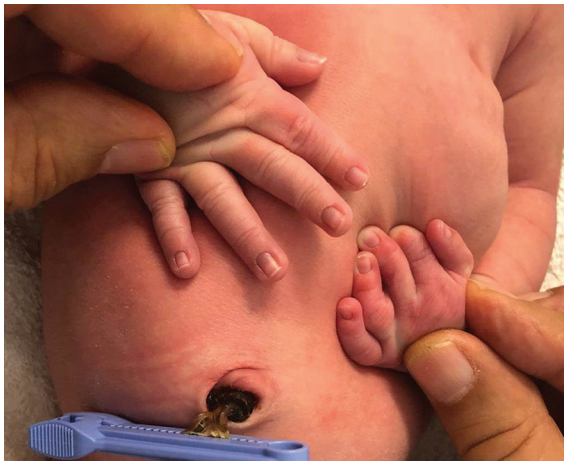

Figure 2. Left hand brachydactyly.

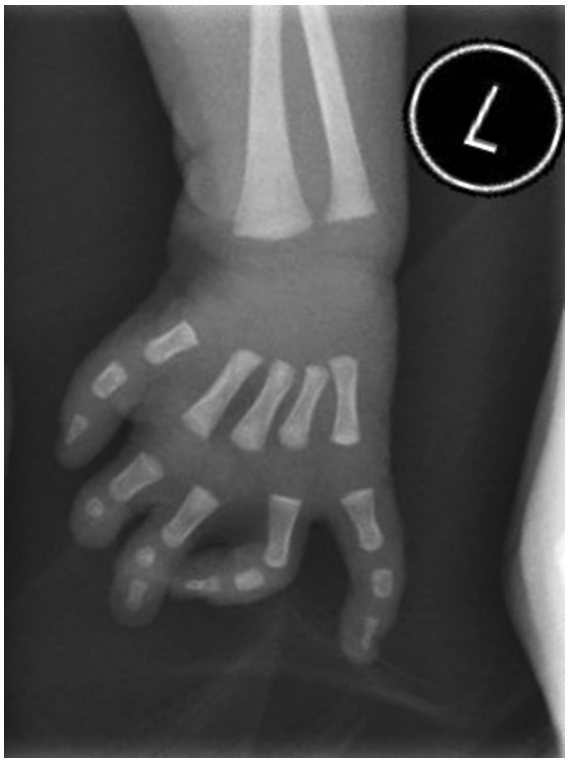

Figure 3. Left-hand x-ray showing hypoplasia of the middle phalanx and agenesis of the distal phalanx of the 2nd finger and hypoplasia of the middle phalanx of the 3rd finger.

The newborn was admitted for 7 days, due to the need for oxygen therapy in the first hours of life, followed by feeding difficulties with the need for nasogastric tube feeding.

Currently twelve months old, the patient weighs 12 kg (> P97; SD 2 [WHO Growth Charts]), has a length of 76.2 cm (P50; SD 0 [WHO Growth Charts]) and head circumference of 50.4 cm (> P97; SD 3 [WHO Growth Charts]). The patient has normal psychomotor development and is under close monitoring by a multidisciplinary team (pediatrics, physiatry, orthopedics, and genetics), with a conservative approach and no genetic testing for now.

Although the etiology is unknown, it has been suggested that a disruption in blood supply to the embryonic tissues that give rise to the chest wall and hand may play a role in the physiopathology of PS[1,4].

The phenotypical presentation of PS is wide and multiple combinations of clinical defects are possible[1]. According to the classification proposed by Romanini et al. in 2018[2], three types of PS are identifiable, based on the presence or absence of upper limb and rib cage abnormalities. Our patient presented with pectoral muscle defect associated with upper limb anomalies (with no rib anomalies), representing a type-2a or upper limb variant of PS.

Our patient's defects are located on the left hemithorax and hand, a less frequent location. The right side is usually more affected than the left (ratio 1.6 to 1) and there are also bilateral cases described[2].

It is important to underline that PS is not progressive and, in the absence of severe rib cage malformations, its survival rate is comparable with the general population. A normal psychomotor development is expected[3]. In some patients, plastic surgery may be performed to rebuild the chest wall or to construct a breast, and physical therapy may be beneficial in improving restricted mobility[4]. Due to the visible physical malformation,

which might be a source of distress, early psychological evaluation and follow-up is also important.

References

1. Mendo TDS, Almeida T, Maria AT, Tuna ML. Poland syndrome: neonatal presentation with axillary pterygium. *BMJ Case Rep*. 2021;14(3):2020-2021. doi:10.1136/bcr-2020-241395
2. Romanini MV, Calevo MG, Puliti A, Vaccari C, Valle M, Senes F, et al. Poland syndrome: A proposed classification system and perspectives on diagnosis and treatment. *Semin Pediatr Surg*. 2018;27(3):189-199. doi:10.1053/j.sempedsurg.2018.05.007
3. Baldelli I, Baccarani A, Barone C, Bedeschi F, Bianca S, Calabrese O, et al. Consensus based recommendations for diagnosis and medical management of Poland syndrome (sequence). *Orphanet J Rare Dis*. 2020;15(1):1-17. doi:10.1186/s13023-020-01481-x
4. Kennedy KR, Wang AL. Poland Syndrome. *N Engl J Med*. 2018;378(1):72-72. doi:10.1056/NEJMicm1709713