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A rare case report of left-sided Poland syndrome in females with hand anomalies and aplastic anemia with pancytopenia

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Abstract
Poland syndrome is a rare congenital disorder presenting with anomalies in the thorax and ipsilateral upper limb. Key features include agenesis or hypoplasia of the sternocostal head of the pectoralis major muscle, nipple hypoplasia, diminished subcutaneous fat, rib irregularities, and sometimes Sprengel deformity (elevated scapula). Symbrachydactyly, characterized by short fingers and cutaneous syndactyly (fused fingers), may also be present. The extent and combination of these features can vary widely. Treatment typically focuses on improving pulmonary function affected by thoracic deformities, with an emphasis on cosmetic improvement. Recent advancements involve using adipose-derived mesenchymal stem cells and fat transfer to correct chest defects and enhance breast appearance. This study reports a case of a 34-year-old woman who presented to the emergency room with bilateral lower limb pedal pitting edema, weakness, and abdominal distension. The diagnosis of Poland Syndrome was confirmed based on her left-sided fused ribs and brachysyndactyly of the same side. The patient chose not to undergo reconstructive surgery, as her primary concerns did not relate to cosmetic or functional issues associated with the syndrome.

Keywords: Reconstructive breast surgery, reconstructive surgery, congenital, surgery, Poland syndrome

Introduction

The most common genetic anomalies associated with hereditary congenital disorders include insertions, deletions, translocations, transpositions, and duplications. Poland syndrome, first described by Alfred Poland in 1841, is characterized by a congenital absence or underdevelopment of the pectoralis major muscle [1].

Poland syndrome is relatively uncommon, occurring in approximately 1 to 3 per 100,000 newborns, with a notable male predominance [2, 3]. It predominantly affects the right side in 75% of cases [2].

Poland syndrome (PS) is a rare developmental anomaly affecting the anterior chest wall. It is characterized by a range of features, including ipsilateral brachydactyly or syndactyly, hypoplasia of the sternocostal head of the pectoralis major muscle, various rib abnormalities, and an elevated, rotated scapula known as Sprengel deformity. Additional symptoms may include a lack of subcutaneous fat and digital abnormalities on the same side of the body. It is uncommon for an individual to exhibit all these symptoms simultaneously, and while Poland syndrome can affect both genders, it is notably prevalent among females [1, 4].

In severe and rare instances, Poland syndrome may involve abnormalities in internal organs like the lungs, kidneys, and heart, making the severity of the condition highly variable. [5] The condition is primarily identified by the absence of the sternocostal head of the pectoralis major muscle.

The exact etiology remains unclear, but it is believed to result from a disruption in the blood supply to the subclavian arteries, vertebral arteries, or their branches during early embryonic development. This disruption is thought to occur around the sixth week of embryonic development, leading to the characteristic abnormalities. The term "Subclavian Artery Supply Disruption Sequence" (SASDS) has been proposed to describe the pathogenesis of Poland syndrome along with related anomalies such as Klippel-Feil and Möbius syndromes [1].

Poland syndrome is a rare and poorly understood congenital anomaly that affects the anterior chest wall. The full spectrum of Poland syndrome's manifestations is not always present in every patient; some individuals may exhibit only a subset of these symptoms [1].

Despite its rarity, Poland syndrome's complexity underscores the need for more research to elucidate its etiology and improve management strategies. This case report seeks to enhance the current understanding of Poland syndrome by detailing the presentation and implications in a patient, offering insights into clinical management and potential associated abnormalities. By presenting a classic case featuring a combination of key symptoms, including brachysyndactyly, this report aims to provide a comprehensive overview of the syndrome in a female patient, contributing valuable information to the body of knowledge on Poland syndrome.

Case Report

A 34-year-old woman presented to the emergency room with bilateral pitting pedal edema, a mass in the left upper abdominal region, and abdominal distension. During the physical examination, we observed brachysyndactyly in her left hand, prompting further investigation. Examination of her chest revealed asymmetry in the left anterior wall with a depression, and the left nipple was positioned higher than usual. Notably, the sternocostal head of the pectoralis major muscle was absent when the shoulders were abducted.

Further investigation revealed no history of exposure to teratogenic drugs during her mother's pregnancy, and there was no family history of consanguinity or similar conditions.

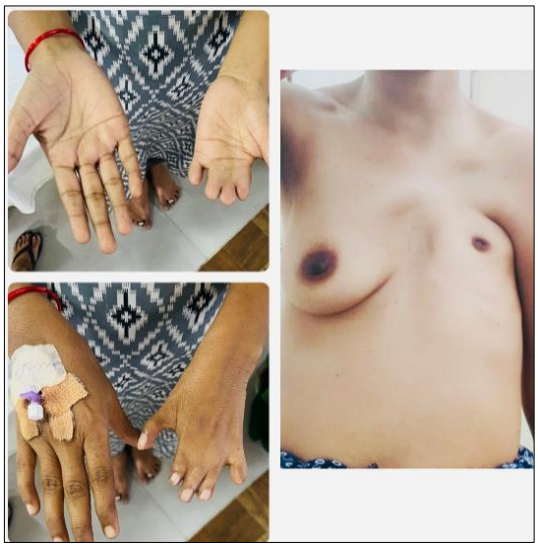

Fig 1: (Clinical Findings: A 34-year-old female with brachysyndactyly in her left hand and displaced left nipple)

Following a radiological examination, the chest X-ray indicated a rightward shift of the heart and a rightward deviation of the descending aorta, along with some bone abnormalities. Further evaluation with a CT scan of the thorax and abdomen revealed a hypoplastic sternum lacking a distinct xiphoid process, and fused ribs on the left side from the fifth to the ninth rib. The abdominal imaging showed a pronounced U-shaped configuration. The patient also presented with pancytopenia and aplastic anemia, and her spleen, though present, was misshapen with no noticeable notch. There was evidence of portal and splenic vein thrombosis. No neurological, urogenital, or respiratory issues were identified. The patient had not received any prior medical care or guidance related to her condition since birth.

A standard chest X-ray showed faint soft tissue in the left breast and hyperplastic changes on the left side of the rib cage. It also noted that the heart's apex was symmetrical. (Figure 2) These findings were confirmed with a Non-contrast CT scan of the thorax, which revealed that the heart had a normal shape but was shifted to the right. The apex of the heart was displaced to the right, and the left dome of the diaphragm was elevated. The costophrenic and costochondral angles were clearly visible and normal. No significant hilar abnormalities or pulmonary pathology were observed. The heart rate result was within normal limits. (Figure 3)

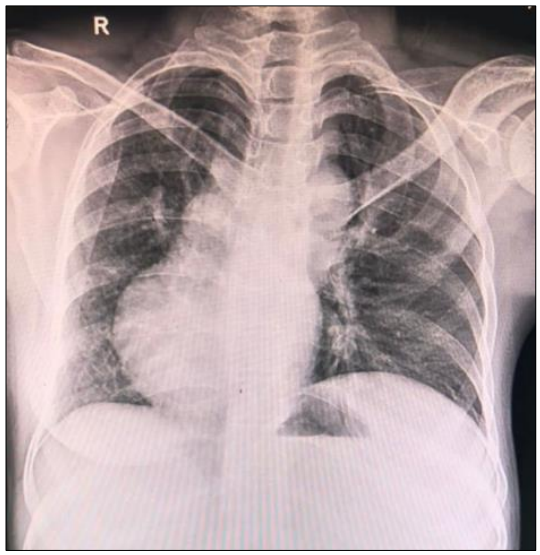

Fig 2: (PA view chest X-ray suggestive of left breast shadow faint left sided hypoplastic ribs)

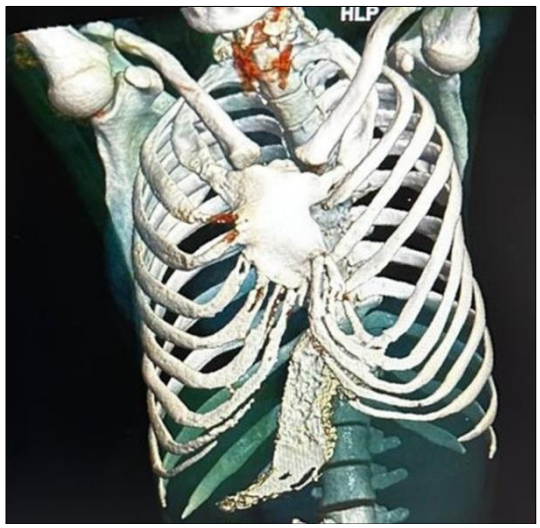

Fig 3: (Non-contrast CT SCAN (coronal view) thorax CT demonstrating a deformed sternum and fused fifth to ninth ribs

A CT scan of the chest revealed underdevelopment of the serratus anterior and pectoralis major and minor muscles on the left side. Rudimentary left breast tissue was also visible (Figure 4). The right breast appeared normal, and the serratus anterior, pectoralis major, and pectoralis minor

muscles on the right side were intact. Additionally, the chest X-ray showed hypoplastic fusion of the 5th to 9th ribs on the left side. The remaining visible lung fields were normal.

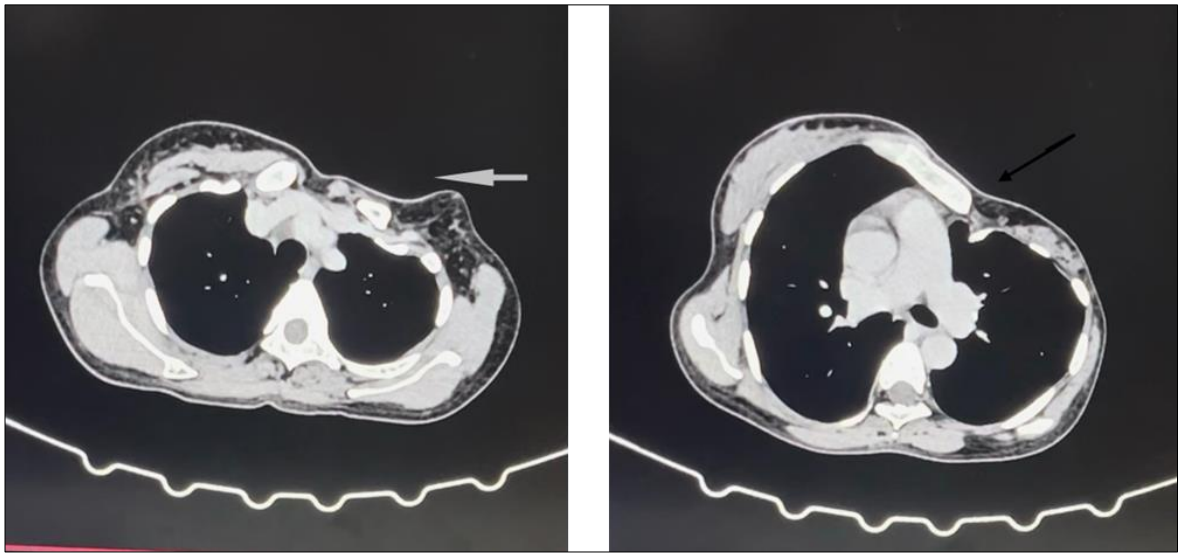

Fig 4: CT chest without contrast (axial view). The primitive pectoralis minor muscle and the undeveloped left pectoralis major muscle are depicted by the arrows)

The left-sided brachysyndactyly hand deformity was associated with a diagnosis of left-sided Poland syndrome. This condition also included splenomegaly, fused ribs on the left side, aplastic anemia, a right-sided displaced heart, and hypoplastic breast tissue on the left. Although the patient was advised against cosmetic surgery, no specific treatment for Poland syndrome was provided. Secondary concerns were addressed with additional research and care.

Discussion

Poland syndrome is a rare congenital condition characterized by musculoskeletal anomalies on one side of the body, specifically affecting the chest wall and upper limb. First described by Alfred Poland in 1841 during a cadaver dissection, the disorder is thought to arise from a partial or complete interruption of blood flow in the subclavian or vertebral arteries and their branches around embryonic days 17 to 18. This vascular disruption is believed to result in the absence of the pectoralis major muscle, abnormalities in interdigital tissue, and issues with cervical vertebrae segmentation [6, 7].

Key aspects of the condition include

- **Disruption of the internal thoracic artery:** Results in the absence of the pectoralis major muscle and underdevelopment of the breast on the affected side.
- **Disruption of the subclavian artery beyond the origin of the internal thoracic artery:** Leads to isolated malformations in the distal transverse limb.
- **Disruption of the subclavian artery before the origin of the internal thoracic artery:** Causes the Poland anomaly [7].

Some research suggests that exposure to ergot alkaloids during the first trimester of pregnancy may be a contributing factor to Poland syndrome [8]. Additionally, genetic factors involved in the development of the pectoral girdle might also influence the condition.

Poland syndrome can be associated with other conditions such as dextrocardia, vertebral anomalies, and undescended testes. Reports have also linked it with extracorporeal intercostal liver herniation, thoracic myelomeningocele, renal agenesis, and megacalycosis of the ipsilateral kidney. Although there have been associations with malignancies and other rare disorders, these are generally considered to be unrelated or not directly correlated [9, 10].

The estimated global prevalence of Poland syndrome, also known as Poland sequence, Poland anomaly, or Poland syndactyly, ranges from approximately 263,000 to 395,000 cases. It is more common in men than in women, with most cases being sporadic. The condition predominantly affects the right side of the body, with left-sided cases being rare, occurring in about 25% of instances [11].

Genetic inheritance of Poland syndrome is not well-documented, and it is generally thought to have a random origin. The most accepted theory is the Vascular Disruption hypothesis, which posits that interruption of blood supply from the subclavian artery during the sixth week of intrauterine life leads to underdevelopment of the pectoralis muscles and limb on the affected side. An alternative explanation, the Mesodermal Malformation Hypothesis, suggests that deficiencies in mesoderm, which is crucial for muscle, bone, and connective tissue development, could result in the observed anomalies. Despite various possible factors, the Vascular Disruption hypothesis remains the most widely supported. Studies indicate that males are affected about three times more frequently than females, and left-sided anomalies are less common due to a genetic predisposition favoring right-sided defects. The majority of male cases often go undetected until functional impairments necessitate diagnosis, complicating data collection [12].

Poland syndrome may present with visceral abnormalities such as hernia, renal agenesis, and atrial septal defect. Associations with blood dyscrasias, diffuse neurofibromas, psoriasis vulgaris, congenital hemangiomas, acquired perforating dermatosis, and café-au-lait spots have also been reported. [13] These findings suggest the need for thorough examination to understand the full spectrum of characteristics associated with Poland syndrome [14].

Conditions to consider in the differential diagnosis include scoliosis, multiple cranial nerve paralysis, cutaneous hamartoma or nevus, webbed neck with restricted movement, Mobius syndrome, Becker's deformity, high scapula, Klippel-Feil syndrome, and Amazon syndrome. Traumatic chest injuries should also be ruled out as they can present similarly [15].

Treatment for Poland syndrome varies based on the severity of the defects and individual factors such as age, gender, and additional health issues. Options may include surgical interventions for chest wall repair, breast reconstruction, rib cage surgery, and orthopedic procedures for hand and arm deformities. Physiotherapy can enhance mobility and function, and combined with surgical treatment, it can offer significant improvements. Prosthetic and orthodontic treatments may also enhance function and appearance [16-18].

Psychological support is essential due to the cosmetic and social impacts of the condition. Therapy focused on self-acceptance and managing cosmetic defects is crucial. Regular check-ups are recommended for early detection and intervention in children with Poland syndrome [19].

The timing and approach of treatment should be tailored to the individual's specific presentation, overall health, and personal preferences. An effective management plan involving a multidisciplinary team can improve outcomes and provide timely intervention.

Conclusion

In conclusion, this case report underscores a rare instance of left-sided Poland syndrome, a highly uncommon congenital condition characterized by a wide range of musculoskeletal malformations. It highlights the importance of early detection and strategic management of defects. The patient's distinctive clinical presentation illustrates the diverse ways in which the condition can manifest and enhances our understanding of its potential variations. This case emphasizes the necessity for tailored treatment plans to address the various manifestations of Poland syndrome, aiming to improve patient outcomes and overall quality of life.

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Conflict of interests

There are no conflicts of interests to declare. Confidentiality was maintained.

References

1. Chait F, Bahlouli N, Chehrastane R, Mrani Alaoui N, Boughaleb F, Zouheir EA, Tarik EM, Allali N, El Haddad S, Chat L. Clinical and Radiological Features in Poland Syndrome: Report of 3 Cases and Review of Literature. Glob Pediatr Health. 2023 Dec 25;10:2333794X231219388. Available from: https://journals.sagepub.com/doi/full/10.1177/2333794X231219388
2. Frioui S, Khachnaoui F. Poland's syndrome. Pan Afr Med J. 2015;21:294. Available from: https://www.panafrican-med-journal.com/content/article/21/294/full
3. Chichery A, Jalbert F, Foucras L, Grolleau J-L, Chavoin J-P. Syndrome de Poland. EMC, Techniques chirurgicales-Chirurgie plastique reconstructrice et esthétique. 2006;45-667-E:1-17.
4. Benzalim M, Berghalout L, Elfakir S, *et al.* Syndrome de Poland: à propos d'un cas et revue de la litterature. Pan Afr Med J. 2017;26:12. Available from: https://www.panafrican-med-journal.com/content/article/26/12/full
5. Assadi FK, Salem M. Poland syndrome associated with renal agenesis. Pediatr Nephrol. 2002;17:269-271.
6. Sparks DS, Adams BM, Wagels M. Poland's syndrome: an alternative to the vascular hypothesis. Surg Radiol Anat. 2015;37:701-702.
7. Bavinck JN, Weaver DD, Opitz JM, Reynolds JF. Subclavian artery supply disruption sequence: hypothesis of a vascular etiology for Poland, Klippel-Feil, and Möbius anomalies. Am J Med Genet. 1986;23:903-918.
8. Stylianos K, Constantinos P, Alexandros T, *et al.* Muscle abnormalities of the chest in Poland's syndrome: Variations and proposal for a classification. Surg Radiol Anat. 2012;34:57-63.
9. Baban A, Torre M, Bianca S, *et al.* Poland syndrome with bilateral features: case description with review of the literature. Am J Med Genet A. 2009;149A:1597-1602.
10. Baban A, Torre M, Costanzo S, *et al.* Familial Poland anomaly revisited. Am J Med Genet A. 2012;158A:140-149.
11. Seyfer AE, Fox JP, Hamilton CG. Poland syndrome: evaluation and treatment of the chest wall in 63 patients. Plast Reconstr Surg. 2010;126:902-911.
12. Slezak R, Sasiadek M. Poland's syndrome (Article in Polish). Pol Merkur Lekarski. 2000;9:568-571.
13. Bavinck JN, Weaver DD. Subclavian artery supply disruption sequence: hypothesis of a vascular etiology for Poland, Klippel-Feil, and Möbius anomalies. Am J Med Genet. 1986;23:903-918.
14. Vazirnia A, Cohen PR. Poland's syndrome: a concise review of the clinical features highlighting associated dermatologic manifestations. Am J Clin Dermatol. 2015;16:295-301.
15. Wright AR, Milner RH, Bainbridge LC, Wilsdon JB. MR and CT in the assessment of Poland syndrome. J Comput Assist Tomogr. 1992;16:442-447.
16. Ohn MH, Ohn KM. Poland's syndrome mimicking traumatic chest injury. BMJ Case Rep. 2021;14. Available from: https://casereports.bmj.com/content/14/2/e239810
17. Moir CR, Johnson CH. Poland's syndrome. Semin Pediatr Surg. 2008;17:161-166.
18. Delay E, Nachaoui H, Frobert P. Poland's syndrome (Article in French). Ann Chir Plast Esthet. 2022;67:358-373.
19. Sami H, Kivan H, Al Hussein S, Khawar A, Ashraf A. Left-Sided Poland Syndrome With No Hand Anomalies: A Case Report. Cureus. 2022 Dec 31;14(12). Available from: https://www.cureus.com/articles/115871-left-sided-poland-syndrome-with-no-hand-anomalies-a-case-report.