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CASE REPORTS

Poland Syndrome: A Case Report.

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ABSTRACT

Background: Poland syndrome is a rare congenital anomaly characterized by unilateral absence or hypoplasia of the pectoralis major muscle, often associated with ipsilateral thoracic and upper-limb abnormalities. Its etiology remains uncertain, with the most widely accepted hypothesis involving disruption of the subclavian artery supply during embryogenesis. Clinical presentation varies widely, ranging from isolated muscular defects to complex thoracic deformities.
Case Presentation: This report describes an 8-month-old male who presented with anterior chest wall asymmetry. Examination revealed unilateral hypoplasia of the left breast, absence of the anterior axillary fold, and hypoplasia of the pectoralis major and minor muscles.
Discussion: Poland syndrome is typically diagnosed on clinical grounds, with imaging serving to delineate the extent of musculoskeletal involvement and exclude associated anomalies. Management is individualized and depends on the severity of deformity, patient age, and functional or psychological impact. Reconstructive options include autologous tissue transfer, prosthetic implantation, or a combination of both. Early diagnosis and appropriate counseling are essential to optimize long-term outcomes.
Conclusion: Poland syndrome is a very rare congenital anomaly with few reports in the international literature. In adults, it needs breast reconstruction to address the asymmetry of the chest.
Keywords: Poland syndrome; congenital chest wall anomaly; pectoralis major agenesis; thoracic deformity; reconstructive surgery

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INTRODUCTION

Poland syndrome is a congenital abnormality characterized by deformities affecting the chest wall and the ipsilateral upper extremity. This condition has an incidence of 1 in 36,000–50,000 newborns. (1) It involves partial or complete absence of the pectoralis major and minor muscles, asymmetric chest wall deformity, and brachydactyly or syndactyly (2).
Potential etiologic factors include genetic predisposition and environmental influences such as maternal smoking. (1,2)

These factors may contribute to Poland syndrome, highlighting the need for further research into case-specific causes. This rare condition has been associated with cardiac angiosarcoma, breast cancer, hematological disorders, and dextrocardia. (5-8)

Case Report

This case involves an eight-month-old male with left-sided Poland's syndrome with total absence of the pectoralis major muscle but with the presence of the pectoralis minor muscle (Figure 1).

On examination, he had unilateral hypoplasia of the left breast, absence of the anterior axillary fold, and hypoplasia of the pectoralis major and minor muscles. (Figure 2).

The absence of hand anomalies in this case, despite other typical features, highlights the variability of Poland syndrome and underscores the need to consider a broad spectrum of presentations during diagnosis. (Figure 3).

This case presents primarily a cosmetic concern rather than a functional deficit. Follow-up is planned, with consideration of reconstructive options such as autologous tissue transfer or implants to improve aesthetic outcomes, as recommended in the literature.

DISCUSSION

Poland syndrome is a very rare congenital disorder in which there is hypoplasia or agenesis of the pectoralis muscles on the affected side. It can be accompanied by chest wall anomalies and ipsilateral upper-limb malformations. Poland syndrome has an incidence between 1/30,000 and 1/50,000 births, although its actual prevalence might be higher, due to undiagnosed cases with no functional disability or limb deformity. (1-3)

The cause of Poland syndrome is unknown. SASDS (subclavian artery supply disruption sequence) theory states that disruption of blood flow at week six of pregnancy leads to developmental problems in structures supplied by the subclavian artery. (1-3) This explains the wide range of symptoms, from mild muscular defects to serious malformations of the chest wall, bones, and upper limbs.

This is an unusual case of Poland syndrome in which there was complete lack of the pectoralis major muscle and preserved pectoralis minor with absence of hand malformation. This case demonstrates phenotypic

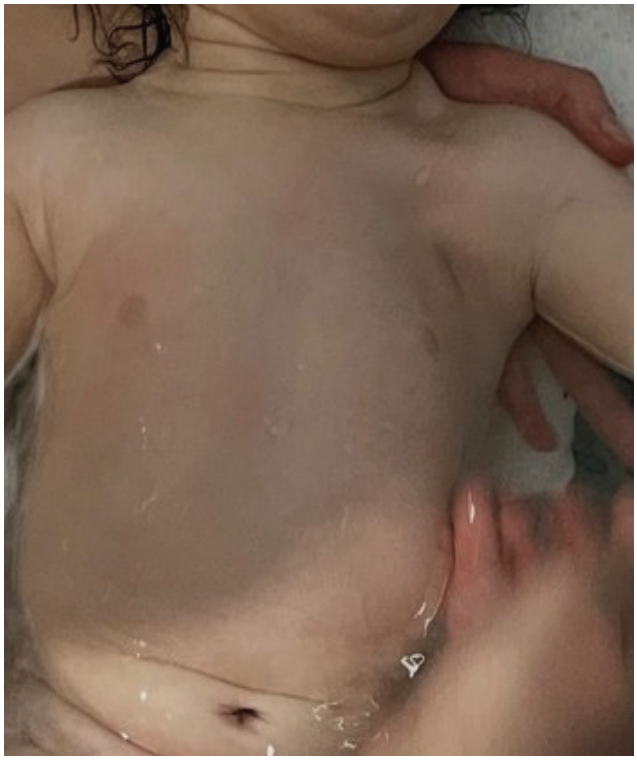

Figure 1. Total absence of the pectoralis major muscle. The pectoralis minor muscle is present.

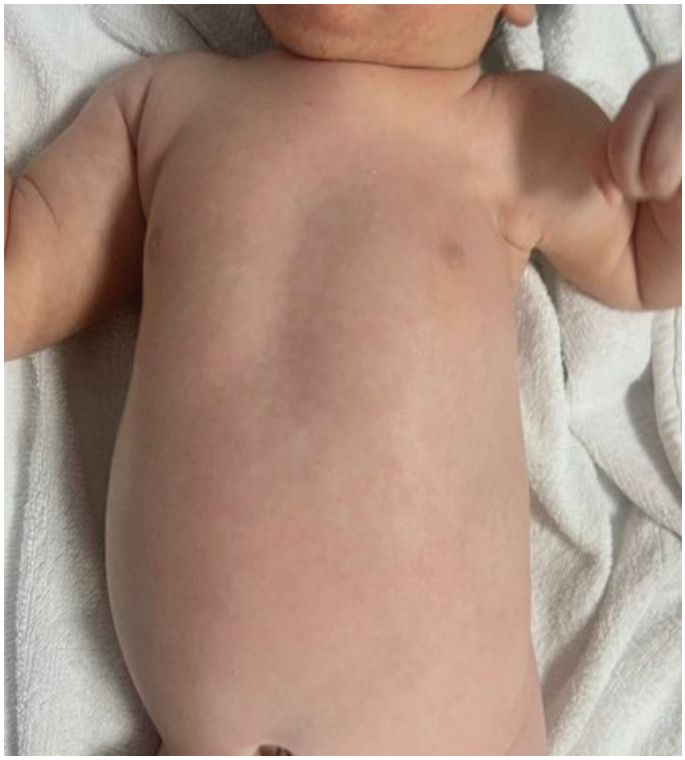

Figure 2. The nipple is present

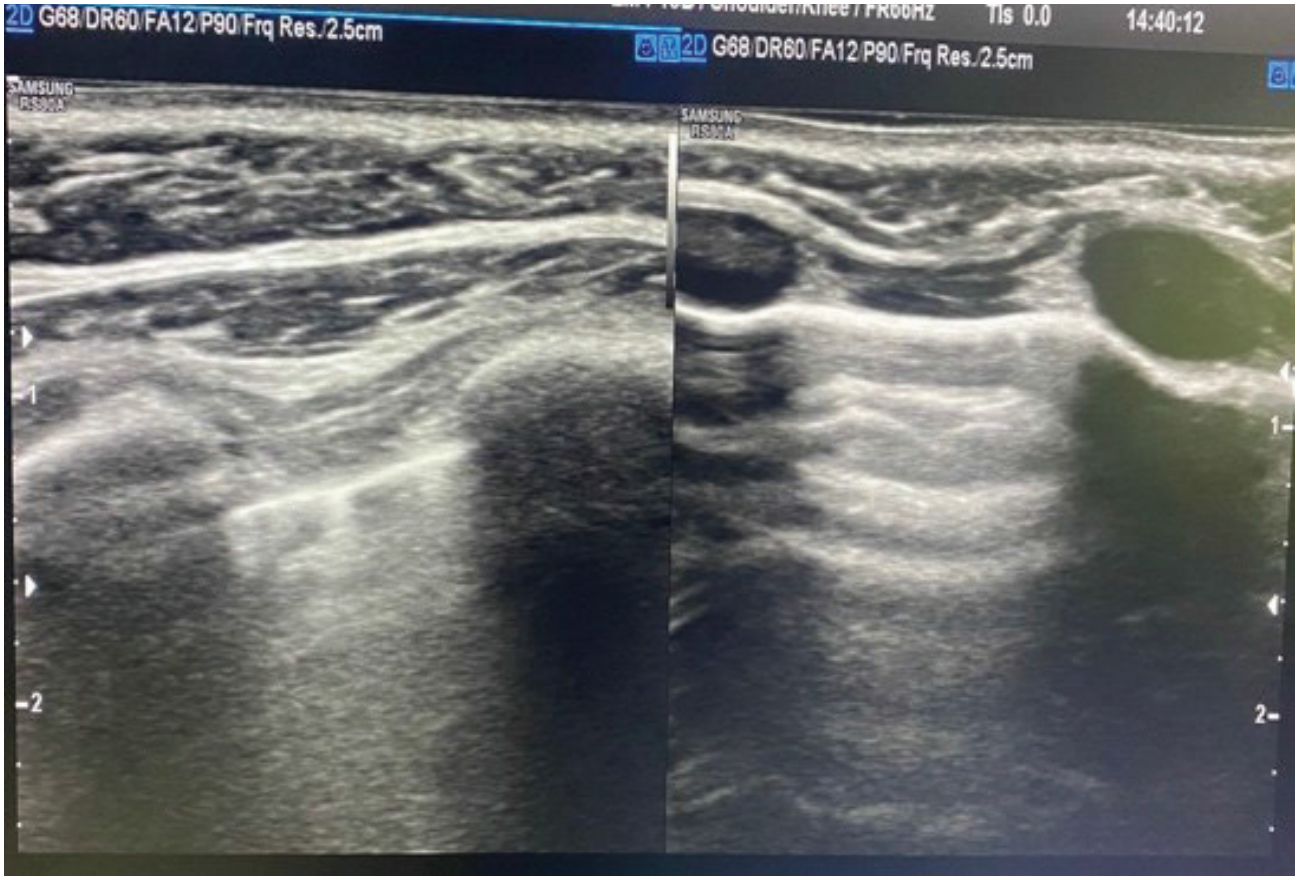

Figure 3. Ultrasound examination confirmed this muscle absence

variability in Poland syndrome. Hand deformities are common in Poland syndrome, such as syndactyly and brachydactyly; nevertheless, current studies reveal that there are patients with isolated chest malformations and absence of hand malformation. (9)

Taking all these factors into account, it is necessary to make a detailed assessment of children with unilateral chest wall differences in order to evaluate muscular and skeletal changes and any associated anomalies. It is worth noting that ultrasound is especially useful for examining chest muscles in infants because of its safety, availability, and the lack of exposure to ionizing radiation. (2-4)

The differential diagnosis consists of isolated deficiency of the pectoralis muscle, other congenital chest wall anomalies, Möbius syndrome, and very rare muscular or skeletal syndromes. In most cases, the combination of physical examination and specialized imaging is sufficient for diagnosing this condition.

Treatment should be planned individually and should take into consideration such factors as patient age, severity of symptoms, functional impairment, and cosmetic issues. Most infants do not require surgery immediately, as their upper limbs usually retain good functionality and chest wall discrepancy can become evident with growth. Follow-up is important in order to monitor the patient's development, detect complications, and choose the best time for surgery if needed. Surgical treatment options for adolescents and adults include muscle transplantations, prosthetic inserts, and various surgical methods. (10)

Poland syndrome usually presents itself as an isolated entity; however, it is important to create awareness among pediatricians, surgeons, radiologists, and geneticists. Early diagnosis helps in making proper diagnosis, counseling of the parents, and multidisciplinary treatment. It is important to have a particular focus on the patients presenting mild cases of Poland syndrome because they can easily be missed in childhood.

The above case adds to the very few cases of Poland syndrome without hand deformity reported in the literature. This shows the significance of diagnosing cases of Poland syndrome with symptoms only involving the muscles of the chest wall.

CONCLUSION:

Poland syndrome is a rare congenital anomaly, characterized by the unilateral absence of the pectoralis major muscle and associated chest wall deformities, which is infrequently documented in international medical literature. While the clinical presentation varies, adult patients primarily seek intervention to correct the aesthetic and functional asymmetry of the chest.

Breast reconstruction remains the definitive surgical approach to restore thoracic symmetry and improve the patient's quality of life. Given the scarcity of reported cases in adults, detailed case reports like this contribute significantly to a broader understanding of the syndrome's management and the refinement of reconstructive surgical techniques.

Editorial Statements

Ethics Approval and Consent to Participate

This study involved the analysis of data from human participants. The research protocol was reviewed and approved by the Institutional Ethics Committee and was conducted in accordance with institutional regulations, the principles of the Declaration of Helsinki, and internationally accepted standards for biomedical research ethics.

Data Availability Statement

The datasets generated and/or analyzed during the current study are available from the corresponding author upon reasonable request.

Author Contributions (CRediT): ES: Conceptualization, study design, and supervision. VR & LF: Data collection, analysis, and manuscript drafting. BT&AD: Literature review and critical revision. All authors approved the final manuscript.

Artificial Intelligence (AI) Statement

The authors used a generative artificial intelligence tool solely for language editing and grammar improvement. All scientific content, data analysis, interpretation, and conclusions were independently developed, verified, and approved by the authors.

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