Uncovering the Unusual: A Case Report of Poland Syndrome

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CASE REPORT

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Abstract

Poland syndrome (PS) is a rare congenital anomaly identified by the unilateral underdevelopment or absence of both the pectoralis major and minor muscles. We present a case of a 17-year-old male with right-sided chest flattening and shortness of breath for 3 years. Physical examination revealed reduced breath sounds on the affected side and chest X-ray showed right-sided hyperlucency. A noncontrast computed tomography (CT) thorax revealed absent pectoralis major and minor muscles on the right side, with visualized ribs appearing unremarkable, leading to a diagnosis of PS. Accurate diagnosis of PS requires imaging modalities such as chest X-rays, CT, and magnetic resonance imaging (MRI) scans. Treatment options for PS include reconstructive surgical procedures. PS affects chest development, leading to respiratory issues affecting an individual’s quality of life.

Keywords: Absent, Case report, Chest flattening, Congenital anomaly, Hyperlucency, Pectoralis major, Pectoralis minor, Poland syndrome, Shortness of breath, Ultrasound, Vascular disruption.

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Introduction

Poland syndrome (PS) is a rare congenital anomaly that affects the development of the chest wall, typically resulting in unilateral hypoplasia or absence of the pectoralis major and minor muscles. First described by Sir Alfred Poland in 1841, it remains an intriguing medical condition due to its rarity, unique clinical presentations, and the challenges it poses to affected individuals. With an estimated incidence of only one in 30,000 live births, PS is both underreported and underdiagnosed, making it a compelling subject for further exploration.

Poland syndrome (PS) remains an area of ongoing research and clinical interest. The etiological factors behind its development are not entirely elucidated, with the vascular disruption theory being the most widely accepted pathogenic mechanism. Understanding this condition is essential, as it can have a significant impact on an individual’s physical and psychological well-being, particularly concerning respiratory function and chest wall deformities. Therefore, this case contributes to the existing literature on PS by providing a comprehensive clinical example and highlighting the importance of accurate diagnosis and management in improving the quality of life for affected individuals.

Case Description

A 17-year-old male presented to the outpatient department with a complaint of right-sided chest flattening (Fig. 1) and shortness of breath on exertion for 3 years. Shortness of breath was not associated with any seasonal, diurnal variation, or wheezing. There were no complaints of cough for the patient. The patient reported no significant past medical or surgical history. There was no history of trauma or antitubercular drug intake. The patient was a nonsmoker and had no family history of similar complaints. On physical examination, the patient had right-sided chest flattening and reduced breath sounds on the affected side. A chest X-ray was done which showed right-sided hyperlucency (Fig. 2). A noncontrast computed tomography (CT) thorax was advised which reported absent pectoralis major and minor muscle on the right side, with visualized ribs appearing unremarkable (Fig. 3). Based on these findings, a diagnosis of PS was made.

Discussion

Poland syndrome (PS) is a rare congenital anomaly that is characterized by unilateral hypoplasia or absence of the pectoralis major and minor muscles. In 1841, Sir Alfred Poland initially documented this medical condition, which has since been estimated to occur in approximately one in every 30,000 live births. PS displays a higher prevalence in males compared to females, with a ratio of 3–1. Additionally, it has a tendency to affect the right side of the body more frequently than the left with a ratio of 1.7–1.1. The etiology of PS has been a subject of debate for >30 years, with various theories and factors proposed. Being a congenital...
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Poland syndrome (PS) is often suspected clinically when chest defects with or without an ipsilateral limb defect are detected in newborns or children. Diagnosis of PS can occur at any age and some cases may present with complaints unrelated to the syndrome itself. The varying age at which diagnosis occurs can be linked to the extensive spectrum of presentation severity, particularly in individuals with uncomplicated PS and normally developed hands. One noticeable characteristic of PS, affecting both males and females, is the absence of the anterior axillary fold. In females, this deformity can manifest as anything from underdeveloped breasts (breast hypoplasia) to the complete absence of a breast (amastia). The presence of accompanying defects in the same side of the chest and limb can signal a more serious form of PS. 4

Accurate diagnosis of PS often requires imaging modalities. A chest X-ray can show lung field hyperlucency on the affected side, which resembles a “postradical mastectomy picture” as described in the literature. Both CT and magnetic resonance imaging (MRI) scans are commonly used to determine the extent of musculoskeletal lesions and to aid in surgical decision-making for reconstruction. 5

The management of PS depends on the severity of the clinical features. In mild cases, no treatment may be required and affected individuals may not experience any significant functional or cosmetic issues. However, in moderate to severe cases, surgical intervention may be necessary to correct the chest wall deformities and improve respiratory function. Surgical reconstruction for PS typically involves the use of tissue expanders, autologous tissue transfer, or implants. 6

**CONCLUSION**

Poland syndrome (PS) is a rare congenital anomaly identified by unilateral hypoplasia or absence of the pectoralis major and minor muscles. It can present with a wide range of clinical features, including chest wall deformities, scoliosis, and respiratory distress. Diagnosis is based on clinical features and imaging studies, and management depends on the severity of the clinical features. Surgical intervention may be necessary in moderate to severe cases and affected individuals may also benefit from physical therapy, rehabilitation, and psychological counseling. Early diagnosis and...
appropriate management can help improve the quality of life of affected individuals.

**Clinical Significance**

This case is important because the underdevelopment of the chest wall can have a significant impact on the respiratory system, leading to shortness of breath or difficulty breathing. This is because the chest wall plays a critical role in the mechanics of breathing, particularly during inhalation. The chest muscles, ribs, and breastbone work together to expand the chest cavity and create negative pressure, allowing air to enter the lungs. When these structures are underdeveloped or absent, the chest cavity may not expand fully, leading to decreased lung capacity and difficulty breathing.

Shortness of breath in individuals with PS can be particularly pronounced during physical activity or times of stress. This is because the demand for oxygen increases during these times, and the respiratory system may struggle to meet this demand when lung function is impaired. In addition to the physical effects, shortness of breath can also have a psychological impact on individuals with PS, leading to anxiety, social isolation, and decreased quality of life.

**REFERENCES**