

Poland Syndrome: A Rare Case Report

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Abstract: Poland syndrome is a rare congenital anomaly characterized by unilateral absence or hypoplasia of the pectoralis major muscle, often associated with ipsilateral upper limb abnormalities. The condition shows wide phenotypic variability and is believed to result from disruption of the subclavian artery supply during embryogenesis.

Keywords: Poland syndrome; Congenital anomaly; Pectoralis major agenesis; Chest wall deformity; Rare disease

1. Introduction

Poland Syndrome is a rare congenital anomaly characterized by unilateral absence or hypoplasia of the pectoralis major muscle, often associated with ipsilateral upper limb abnormalities¹. The syndrome consists of multiple anatomical anomalies including, absence of sternocostal head of pectoralis major muscle, hypoplasia or absence of pectoralis minor muscle, as well as digital abnormalities such as syndactyly or ectrodactyly^{2,3}. Other common anomalies noted in Poland syndrome are malformations of the anterior chest wall and breast, dextrocardia, lung herniation and rib aplasia amongst others⁴.

Poland syndrome has an incidence of about 1 in 30,000 live births⁵, with a slight male preponderance⁶ and more commonly affecting the right side of the chest⁷.

Here, we report a rare case of Poland syndrome affecting the left side of the anterior chest wall in a 15 year old female.

2. Case Presentation

A 15-year-old female presented to the outpatient department with complaints of asymmetry of the chest wall, noticed since birth. There was no history of trauma, surgery, or similar conditions in the family. The patient had normal developmental milestones.

On physical examination, there was noticeable flattening of the left anterior chest wall. There was absence of anterior axillary fold as the left pectoralis major muscle was absent. There was hypoplasia of the left breast tissue and the left nipple appeared hypoplastic and slightly elevated compared to the right [Figure 1]. Posteriorly there was mild asymmetry of scapula; no rib deformities were noted [Figure 2]. Examination of the upper limbs revealed no abnormalities such as syndactyly and hand functions were normal. No vascular anomalies were identified.

Differential diagnoses considered included other congenital chest wall deformities such as, pectus excavatum and isolated muscular hypoplasia.

3. Discussion

Poland Syndrome is an uncommon congenital condition with an estimated incidence of 1 in 20,000–30,000 live births⁵. It is characterized by unilateral absence or underdevelopment of the pectoralis major muscle and may be associated with ipsilateral upper limb anomalies such as symbrachydactyly. The exact etiology remains unclear, though it is widely believed to result from disruption of the subclavian artery supply in the sixth week of gestation, during embryonic development⁸. Most cases are sporadic, with a higher prevalence in males and more frequent involvement of the right side. Due to its variable presentation, diagnosis may be delayed, especially in milder cases. This report describes a classical presentation without limb involvement in a young adolescent female.

Our case is notable for the absence of associated limb anomalies, and because the patient is a female with left sided affliction, as opposed to the common scenario. Imaging, particularly CT or MRI, plays a crucial role in confirming the diagnosis and planning management.

Management is primarily cosmetic and depends on the severity of deformity and patient preference. Surgical options include muscle transposition, implants, and reconstructive procedures, typically performed after puberty^{9,10}. Prognosis is generally excellent, especially in cases without functional impairment^{2,11}.

4. Conclusion

Poland Syndrome is a rare congenital anomaly with variable clinical presentation. Early recognition is essential for appropriate counseling and management planning. This case emphasizes that even in the absence of limb anomalies, Poland Syndrome should be considered in patients presenting with unilateral chest wall asymmetry.

Patient Consent: Written informed consent was obtained from the patient's guardian for publication of this case report and accompanying images.

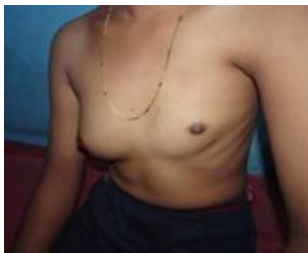


Figure 1

Anterior view showing unilateral flattening of the chest wall with hypoplasia of breast tissue and absence of anterior axillary fold.



Figure 2

Posterior view demonstrating mild asymmetry of the scapulae without spinal deformity.

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