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An Unusual Case of Left-Sided Poland Syndrome Presenting with Diaphragmatic Hernia and Mediastinal Shift with Absence of Hand and Scapular Deformity

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Learning Point of the Article:

Left-sided Poland Syndrome is rare, seen in only about 25% of cases. This case is unusual due to the absence of typical hand and scapular deformities and the presence of rare features such as diaphragmatic hernia and mediastinal shift. While often sporadic, consanguinity in this case suggests a possible genetic link. Early recognition and thorough evaluation are crucial to identify such rare associations and plan individualized treatment.

Abstract

Introduction: Poland syndrome (PS) is a rare congenital condition often defined by unilateral aplasia of the pectoralis major, predominantly affecting the right side of the body. The second most prevalent manifestation is symbrachydactyly, characterized by excessively short and webbed fingers.

Case Report: This report concerns an 18-year-old guy who presented to the orthopedic outpatient clinic with a complaint of left chest pain during exertion, identified with the rare left-sided PS characterized by the absence of the left antero-inferior pectoralis major muscle. The physical examination of the hands revealed no evidence of ipsilateral digital anomalies, a typical characteristic of PS. A significant characteristic is the concurrent presence of a little diaphragmatic hernia and a rightward shift of the mediastinum, frequently disregarded in standard presentations of PS; however, they affect the syndrome's variability and intensity. The consanguinity of the patient's parents necessitates an examination of possible genetic ramifications in an illness generally considered sporadic.

Conclusion: This case presents a rare left-sided PS with diaphragmatic hernia and mediastinal shift but without hand or scapular abnormalities. The atypical presentation underscores the need for high clinical suspicion and detailed evaluation. While typically sporadic, consanguineous parentage suggests a possible genetic link.

Keywords: Left-sided Poland syndrome, diaphragmatic hernia, no hand deformity, consanguineous marriage, no scapular deformity.

Introduction

Poland's syndrome (PS) is a rare birth condition that affects the chest area, usually showing up as missing or underdeveloped parts of the pectoralis major muscle on the one side, different shapes of the ribs, a shoulder blade that is higher and turned (called Sprengel's deformity), smaller or missing nipples or breasts, little fat under the skin, missing or deformed cartilage or ribs, hair loss in the armpit, and problems with fingers on the same side. Furthermore, the disease predominantly affects the right hemithorax unilaterally [3]. We share a unique case of left-

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Author's Photo Gallery

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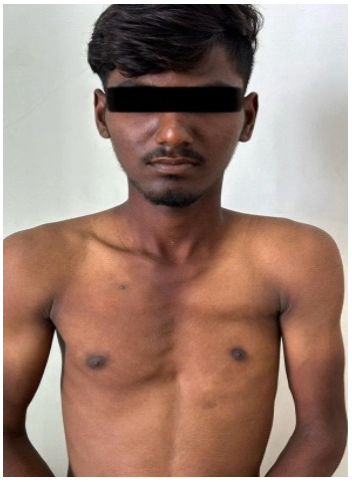

Figure 1: Front view absence of the pectoralis major sternocostal head.

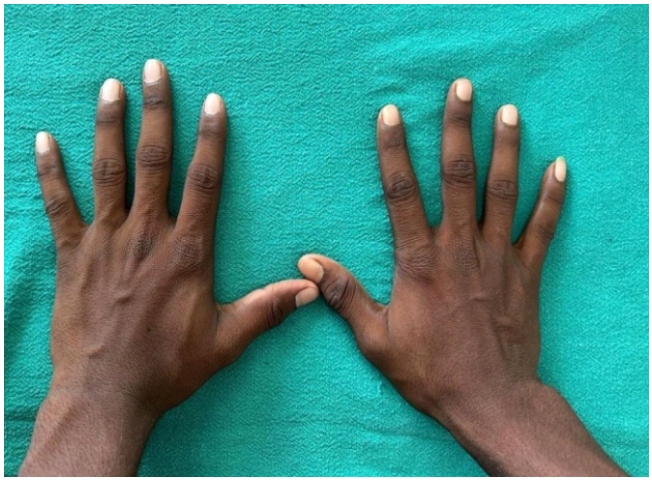

Figure 2: Examination of the hands did not show any signs of ipsilateral or contralateral digital abnormality.

sided PS that includes a diaphragmatic hernia, movement of the mediastinum, and missing hand and shoulder deformities.

Case Report

An 18-year-old male presented to the orthopedic outpatient department with a complaint of left-chest pain while playing sports from childhood for which he got symptomatic management from nearby hospitals. Furthermore, there is a history of occasional complaints of heartburn, left-sided weakness, and pain during physical exertion. Family history revealed that the patient's parents had a consanguineous marriage. There were no breathing or cardiac complaints during the review of the systems.

Upon physical examination, the left antero-inferior chest wall was depressed and flattened. During the physical examination, it was clear that the pectoralis major sternocostal head was missing when the arm was pushed against resistance while rotating inward and lifting the shoulder. The left nipple displayed hypoplasia in comparison to the right. During respiration, no paradoxical movement of the chest wall was observed. Examination revealed alopecia in the axillary and pectoral regions. Physical examination of the hands did not show any signs of ipsilateral or contralateral digital abnormality. Examination of the back indicated that there were no associated abnormalities such as scapular deformity or absence of latissimus dorsi/serratus anterior. There were no remarkable findings in the examination of other systems, namely the nervous, gastrointestinal, respiratory, and urogenital systems (Fig. 1-3).

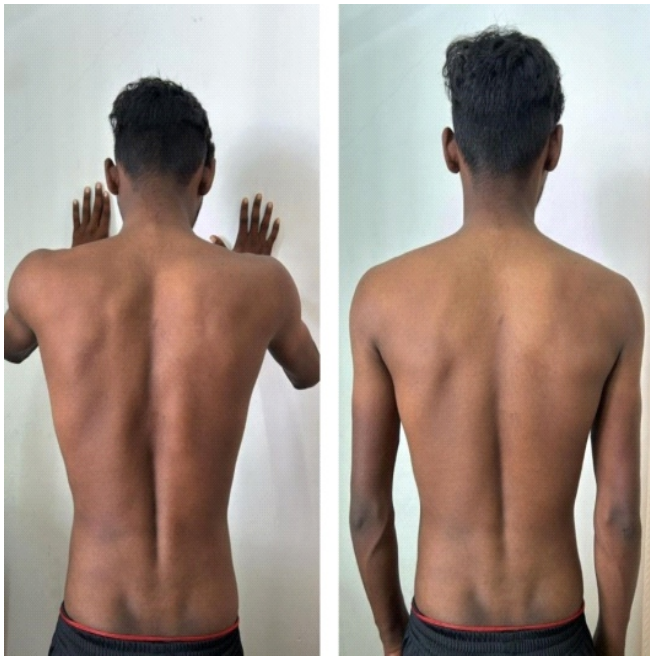

Figure 3: Dorsal view-no associated abnormalities such as scapular deformity or absence of latissimus dorsi/serratus anterior were noted.

Investigations

The chest X-ray showed hypertranslucent left-sided hemithorax with hypoplasia of 2–4 costal cartilages. A mild degree of diaphragmatic hernia was noted on the left side, and it explains the mild shift of the heart toward the right side. However, the heart was normal in size, and the apex of the heart was left sided. No gross active lung pathology was noted. The patient denied taking a computed tomogram or an ultrasound, citing monetary reasons. A normal electrocardiogram (ECG) ECG with sinus arrhythmia was recorded in the 12-lead ECG, taken for screening of any cardiac disease (Fig. 4 and 5).

Treatment

Given the mild nature of the patient's complaints, we initiated conservative management. Postural correction to prevent imbalances caused by asymmetry of the chest and upper limb started by strengthening the paraspinal and scapular stabilizing muscles. Strengthening exercises target the contralateral and compensatory muscles, particularly the shoulder and back muscles. Range of motion (ROM) exercises started to preserve full mobility of the shoulder and upper limb and to prevent

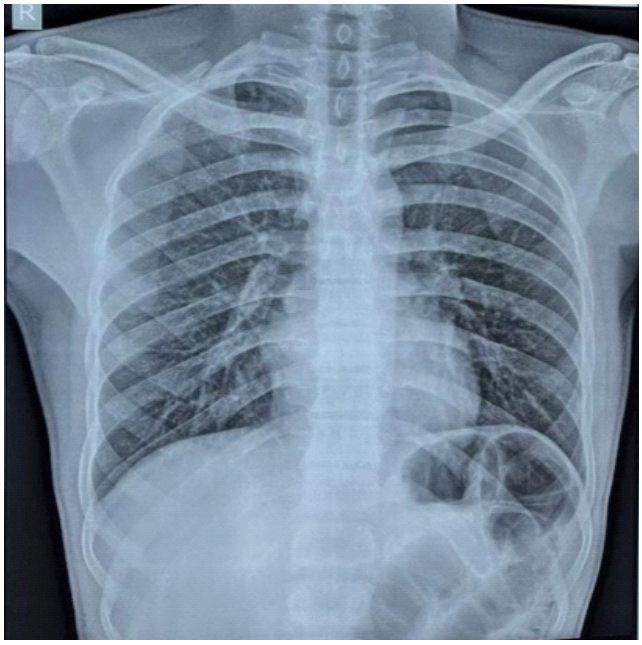

Figure 4: On chest X-ray, the left hemithorax is hyper-translucent, secondary to the absence of the left pectoralis muscle and anterior ends of four ribs (2nd–5th). The heart and mediastinum are shifted to the right due to chest wall deformity with mild degree of diaphragmatic hernia noted on the left side.

contractures or stiffness. Respiratory exercises can help improve lung expansion and chest movement, and training was begun to help the patient adapt to doing daily activities without certain muscles. The patient was referred to the general surgery department for diaphragmatic hernia management.

Outcome and follow-up

On follow-ups, the patient had good clinical improvement in terms of weakness and functional adaptation. The patient underwent regular physiotherapy as advised and showed improved ROM.

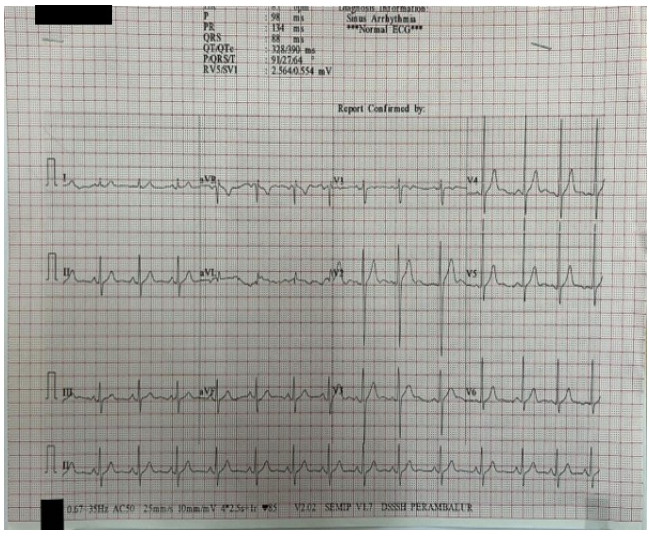

Figure 5: A normal electrocardiogram (ECG) with sinus arrhythmia was recorded in the 12-lead ECG, taken for screening of any cardiac disease.

Discussion

PS was initially described by Alfred Poland in 1840 and is characterized by ipsilateral agenesis or hypoplasia of the sternocostal head of the pectoralis major, hypoplasia of the nipple or breast, absence of subcutaneous fat, multiple rib anomalies, elevated and rotated scapula (Sprengel's deformity), and ipsilateral digit malformations [4]. When the upper part of the serratus anterior muscle loses blood supply from the suprascapular arteries, it leads to the shoulder blade being raised and sticking out, which is often linked to Sprengel's deformity in PS. PS may coexist with Klippel–Feil syndrome, noted for cervical shortening [5]. The patient showed very little hand involvement and did not have Sprengel's deformity, which is a rare hand issue seen in 13.5–56% of cases with PS and is used to help identify the condition [6]. Genetic inheritance of PS is rare and considered a sporadic occurrence. It has been proposed that the injury arises during the 6th week of gestation as a result of a localized vascular anomaly of the subclavian artery. This stage is characterized by the bifurcation of the pectoralis major's two heads and the formation of tissues between the digits [5]. The consanguineous origin of the patient's parents prompts an investigation into potential genetic implications in a condition typically regarded as sporadic.

This syndrome is typically unilateral and tends to affect the right side of the body. Only about 25% of recorded instances involve the left side of the body [1, 3]. Dextrocardia has been linked to left-sided PS, especially in patients who have two or more atrophied ribs [7]. In this case, we report this uncommon manifestation of left-sided PS without concomitant dextrocardia. The latissimus dorsi, external oblique, and serratus anterior muscles may also be impacted in certain instances of PS. Due to hypoplasia of the ribs and thin, malformed cartilages, the majority of patients have a depressed chest wall on the one side [8]. On the affected side, conventional anteroposterior radiography typically shows a hyperlucent lung [9]. In addition, the heart has a propensity to move toward the unaffected side.

Treatment for PS depends on factors such as age, sex, the type of deformity, and any associated functional limitations [10]. Often, correcting the thoracic abnormality only requires cosmetic surgery. Other indications for surgery include increased lung herniation and paroxysmal motions of the chest wall. Rebuilding a functional hand should be the main goal of surgical intervention for hand anomalies, depending on the specifics of the deformity [11]. We treated our patient

symptomatically because they complained of heartburn, mild left-sided weakness, and pain when exerting themselves. The patient received frequent physiotherapy to strengthen their muscles.

Conclusion

This case shows an uncommon left-sided PS with diaphragmatic hernia and mediastinal shift but no hand or scapular abnormalities. The absence of traditional symptoms highlights the requirement for high clinical suspicion, thorough physical examination, and radiographic assessment for appropriate diagnosis. As PS is usually sporadic, the consanguineous parenthood shows a genetic relationship, warranting further inquiry. Conservative care, including physiotherapy and respiratory exercises, improved patient function. Atypical PS patients need early detection and personalized, interdisciplinary therapy to improve quality of life.

Clinical Message

• Rare left-sided presentation

Left-sided Poland syndrome is much less common (~25% of cases). This case uniquely involved the absence of the left antero-inferior pectoralis major, no hand or scapular deformity, and left-sided diaphragmatic hernia.

• Uncommon clinical features

Absence of ipsilateral hand anomalies (usually a key identifying feature of PS) and scapular deformity (Sprengel's deformity), which is often associated with PS, were the uncommon features in our case report. The presence of diaphragmatic hernia and mediastinal shift, both uncommon and often overlooked features of PS.

• Genetic consideration

Though PS is generally sporadic, this case involved a consanguineous marriage, highlighting a potential genetic component that warrants further exploration.

• Importance of individualized treatment

Surgical intervention is often cosmetic unless there are functional impairments. Physiotherapy plays a crucial role in symptom management and improving quality of life when surgery is not pursued.

Declaration of patient consent: The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient has given the consent for his/ her images and other clinical information to be reported in the journal. The patient understands that his/ her names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

Conflict of interest: Nil **Source of support:** None

References

1. 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;27:189-99.

2. Mühlbauer W, Wangerin K. Zur embrologie und atiologie des Poland-und amazonensyndroms [Embryology and etiology of the Poland and amazone syndromes]. Handchirurgie 1977;9:147-52.

3. Seyfer AE, Fox JP, Hamilton CG. Poland syndrome: Evaluation and treatment of the chest wall in 63 patients. Plast Reconstr Surg 2010;126:902-11.

4. Bansal A, Reddy K, Dinsmore K, Gonzalez-Estrada A. Poland syndrome: A case report. BMJ Case Rep 2017;2017:bcr2017221564.

5. 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;23:903-18.

6. Ireland DC, Takayama N, Flatt AE. Poland's syndrome. J Bone Joint Surg Am 1976;58:52-8.

7. Lacorte D, Marsella M, Guerrini P. A case of Poland Syndrome associated with dextroposition. Ital J Pediatr 2010;36:21.

8. Walker JC Jr., Meijer R, Aranda D. Syndactylism with deformity of the pectoralis muscle. Poland's syndrome. J Pediatr Surg 1969;4:569-72.

9. Samuels TH, Haider MA, Kirkbride P. Poland's syndrome: A mammographic presentation. AJR Am J Roentgenol 1996;166:347-8.

10. Ohn MH, Ohn KM. Poland's syndrome mimicking

traumatic chest injury. BMJ Case Rep 2021;14:e241408.
11. Sami H, Kivan H, Al Hussein S, Khawar A, Ashraf A. Left-sided Poland syndrome with no hand anomalies: A case report. Cureus 2022;14:e33192.

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