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A CASE REPORT OF POLAND SYNDROME TREATED FOR SYNDACTYLY RELEASE AT KHYBER TEACHING HOSPITAL PESHAWAR

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ABSTRACT

Background: Poland syndrome (PS) is a rare congenital condition characterized by unilateral absence or underdevelopment of the pectoralis major muscle, often accompanied by ipsilateral hand anomalies such as syndactyly. The syndrome is more common in males, predominantly affecting the right side. Severity and associated anomalies vary widely, making early diagnosis and appropriate intervention essential.

Case Presentation: We report a case of a 16-year-old right-hand dominant male from a rural area of Peshawar, presenting with congenital webbing (syndactyly) of the index, middle, and ring fingers of the right hand. Clinical examination revealed an absent right pectoralis major muscle. Due to financial constraints, confirmatory imaging (CT scan) was not performed. The patient underwent successful surgical release of syndactyly between the middle and ring fingers. A second-stage release of the index and middle fingers was planned for a later date. There was no family history of PS, and no cardiac or other systemic anomalies were detected.

Discussion: Poland syndrome exhibits diverse phenotypic variability. This case aligns with the classical presentation in males, involving right-sided chest wall and upper limb anomalies. The absence of systemic involvement and mild severity allowed functional and aesthetic improvement via surgical intervention. Literature suggests vascular disruption during embryogenesis as a likely pathophysiological mechanism. Timely surgical correction in such patients enhances hand function and cosmetic outcomes, especially in cases with syndactyly.

Conclusion: This case highlights a mild variant of Poland syndrome involving functional impairment due to syndactyly, treated successfully at a tertiary care hospital in Pakistan. It emphasizes the importance of clinical evaluation for timely diagnosis and intervention, especially in resource-limited settings where imaging may not be feasible. To our knowledge, this is the first reported case of PS managed surgically in our institution.

INTRODUCTION

Poland's anomaly was first identified in 1841 by Sir Alfred Poland, who described it as a syndrome characterized by the absence or underdevelopment of the pectoralis major muscle. In some cases, this condition is also associated with breast hypoplasia, agenesis of the second, third, fourth, and fifth ipsilateral costal cartilages, athelia, and ipsilateral webbing of the fingers (cutaneous syndactyly) **[1]** The severity of Poland syndrome can vary among affected individuals. Presently, it is understood that this syndrome is specifically marked by the absence of the sternocostal portion of the pectoralis major muscle **[2]**.

Determining the exact incidence of Poland syndrome (PS) is challenging; however, current estimates suggest it occurs in approximately 1 in 7000 to 1 in 100,000 births. The condition is more commonly observed in males, with a reported male-to-female ratio of 2:1 to 3:1. In nearly 75% of unilateral cases, the syndrome affects the right hemithorax **[3,4]**. Affected individuals may exhibit additional variable characteristics, including the partial or complete absence of one nipple along with the areola and/or patchy hair loss in the axillary region **[1]**. In females, the syndrome may also result in the underdevelopment or complete absence of one breast along with its underlying subcutaneous tissues. In some instances, associated skeletal anomalies may be present, such as underdeveloped or missing upper ribs, elevation of the scapula (Sprengel deformity), or shortening of the arm due to the underdevelopment of the forearm bones, specifically the ulna and radius **[4]**.

We are presenting this case of PS from Khyber Teaching Hospital Peshawar Pakistan, treated for syndactyly release. PS has not been reported previously from our department and hospital.

Case Report:

A 16-year old male from a rural area of Peshawar, right hand dominant, helping his father in selling fruits, presented to outpatient department of Orthopedic Surgery, Khyber Teaching Hospital Peshawar, with complaints of webbing of his index, middle and ring fingers of the right hand since birth. His left hand was normal. On thorough examination, he had absent right pectoralis major muscle as well, as evident in the picture below. CT Scan was advised for confirmation of the absent pectoralis major and for literature reporting purpose but the patient was poor and he could not afford it, hence not performed. He was seen by the consultant orthopedics and hand surgeon of our department and after explaining the procedure to the patient and his parents, surgical release of his syndactyly was planned. Pre and post-operative pictures are also given below.

Figure 1: Absent right pectoralis major muscle.

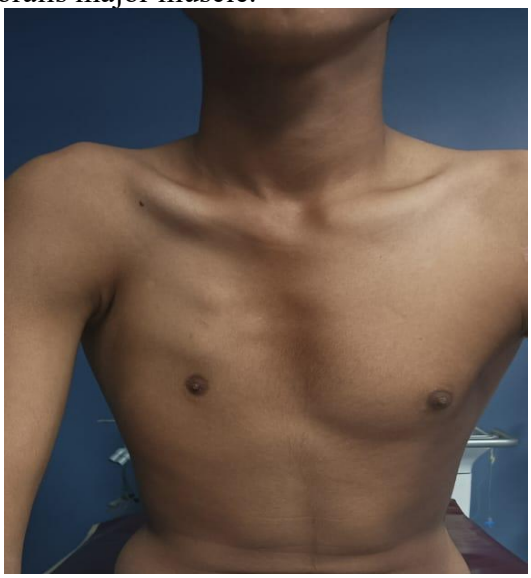

Figure 2: Syndactyly right index, middle and ring fingers.

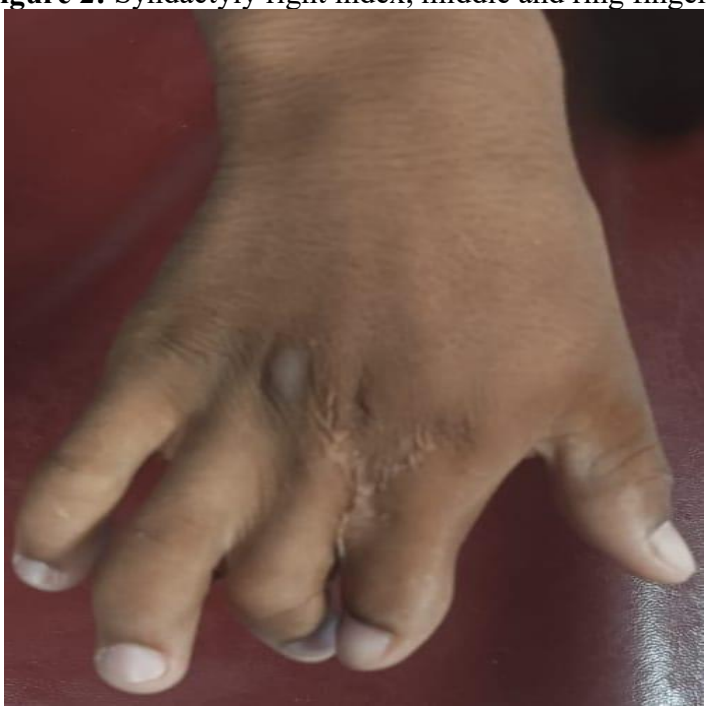

Figure 3: Frontal view of the syndactyly.

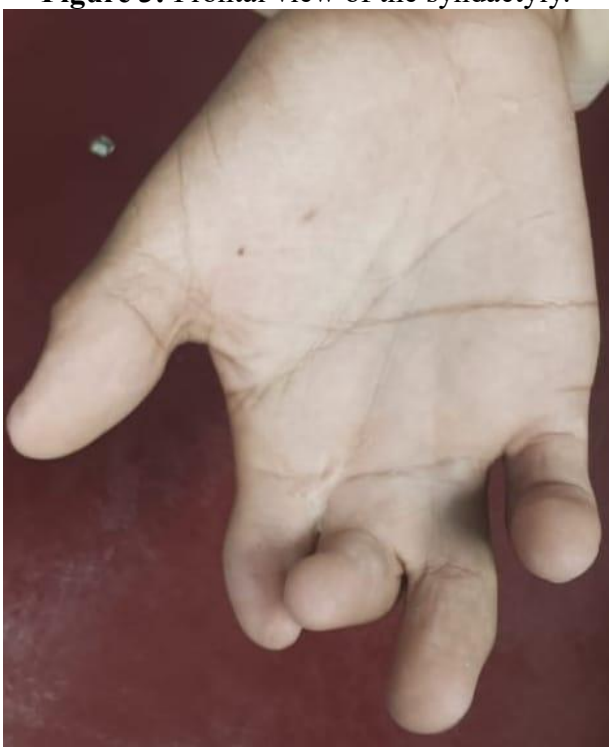

Figure 4: Post syndactyly release of middle and ring fingers.

Figure 5: Volar view of the syndactyly release.

Syndactyly of the index and middle finger was planned to release in second stage after a few months.

Discussion:

Poland syndrome is characterized by asymmetry in hand size, with one hand being smaller than the other, along with the unilateral absence of the sternocostal portion of the pectoralis major muscle. It also involves various deformities and malformations affecting muscles, bones, and veins in the affected chest wall and ipsilateral upper limb, as well as potential malformations of the heart and other internal organs **[5]**. It is observed in men two to three times more frequently than in women. Among males, unilateral deformities occur twice as often on the right side of the chest compared to the left, while in females, there is no clear predisposition toward one side **[1,5,6]**. The exact cause of Poland syndrome remains unknown, with most cases occurring sporadically **[2,7]**. However, a small number of cases have been reported to have a familial history **[8,9,10]**. One study suggested a lack of genetic inheritance by documenting a case where only one twin brother was affected by Poland syndrome, while the other was not **[11]**. In the present case, a detailed genealogical analysis was conducted, and no other family members were found to have Poland syndrome. In general, research indicates that the condition is three times more common in males than females and that the right side of the body is affected three times more frequently than the left **[1,5,6,11]**. The current case involved a male patient with right-side involvement. Another study investigated the relationship between Poland syndrome and dextrocardia in 16 reports, revealing that 23% of the 26 patients with left-sided involvement also had dextrocardia. However, among the 48 patients with right-sided Poland syndrome, no cases of dextrocardia were observed. This high incidence suggests that dextrocardia may be a component of the Poland complex **[12,13].** In the present case, although the right side was affected, no cardiac anomalies or isolated dextrocardia were detected upon telecardiographic, echocardiographic, and MRI examinations. In some cases of Poland syndrome, various anomalies can be observed, including nipple abnormalities such as hypoplasia or absence, as well as hypoplasia or aplasia of the arm. Other associated features include syndactyly, brachydactyly, absence of fingers or ribs on the affected side, skeletal anomalies involving the spine, scapula, sternum, and foot, as well as heart and kidney abnormalities. Additionally, pectoralis major anomalies may be present as part of Möbius syndrome, with varying frequencies among affected individuals. The severity of anatomical malformations varies significantly, as do the clinical manifestations of these anomalies. Nearly all patients with Poland syndrome lack the sternocostal head of the pectoralis major muscle, and most also have an absent pectoralis minor muscle. **[1,7,12,13].** Breast anomalies occur in approximately one-third of affected females, ranging from mild hypoplasia to complete amastia. In cases where breast abnormalities are subtle, Poland syndrome may be detected incidentally during radiological examinations at an estimated rate of 1 in 19,000 cases. Breast involvement can range from mild hypomastia to amastia. While the rib cage is typically normal in most patients, those with affected ribs often present with at least one area of rib collapse. Hand involvement occurs in less than half of patients with Poland syndrome **[5,13,14,15].** Additionally, approximately 6% of Poland syndrome patients present with dextrocardia without cardiac inversion **[13]**. Mild forms of the syndrome are more common than severe cases and may sometimes go undiagnosed. In the present case, there was no pectoralis major muscle on the right side, and hypoplasia of the anterior axillary line was noted. The case was classified as a mild form of Poland syndrome. Although the exact pathophysiology of Poland syndrome remains unclear, several hypotheses have been proposed. The most widely accepted theory suggests that during the sixth to seventh weeks of gestation, when the upper extremities begin to develop from the chest wall, exposure to teratogenic factors may cause mutations leading to malformations or spasms in the brachiocephalic arterial structures, reducing

blood flow. It has been reported that disruptions in subclavian artery blood flow can result in upper extremity abnormalities, while involvement of the internal thoracic artery may contribute to defects in the pectoralis major muscle, breast, and thoracic wall **[2,16,17]**. Another hypothesis suggests that malformations may result from impaired migration of the lateral plate mesoderm during the third and fourth weeks of intrauterine development **[18]**. In the present case, although pulmonary parenchyma and vascular structures were not evaluated via computerized thoracic tomography for the said reason, the absence of the pectoral muscle did not lead to any loss of strength or functional impairment in the patient. Patients with significant chest wall defects or hand anomalies may be candidates for surgical intervention. In males, depression of the pectoral region, and in females, breast abnormalities, often lead individuals to seek cosmetic solutions. Like in this case, the patient and his parents were concerned about his syndactyly both for functional and cosmetic reasons.

Conclusion:

Cases of Poland syndrome reported in the literature are often associated with anomalies in other organs or the presence of tumors at varying rates. As a result, patients with significant anomalies are typically diagnosed in childhood, while milder cases may go undiagnosed. Here, we present a rare case of Poland syndrome that involve functional impairment due to syndactyly, apart from an altered physical appearance.

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