

A case report of Poland Syndrome with Absent Limb Anomalies

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Learning Point of the Article:

Diagnosis of atypical presentations of Poland Syndrome requires a keen eye and a good understanding of both classical and varying presentations of such rare syndromes.

Abstract

Introduction: Poland Syndrome (PS) is a rare congenital condition associated with the absence of unilateral chest wall muscles and sometimes ipsilateral symbrachydactyly (abnormally short and webbed fingers). The condition typically presents with unilateral absence of the sternal portion of the pectoralis major muscle which may or may not be associated with a hypoplasia of the breast, an ipsilateral webbing of the fingers (cutaneous syndactyly) and agenesis of 2, 3, 4, and 5 ipsilateral costal cartilages, and athelia.

Case report: We report a 13-year-old patient with an atypical variant of PS without any limb anomalies. In view of the good function of the upper limb, no surgical treatment was offered, and the patient and his family were counseled regarding the condition. A follow-up of the patient at 2 years revealed that the patient is still asymptomatic with good functional status of the upper limb.

Conclusion: It is hoped that this paper will further improve our understanding of this rare syndrome and its atypical presentations.

Keywords: Poland syndrome, atypical Poland, congenital.

Introduction

In 1841, Sir Alfred Poland first described the Poland Syndrome (PS). In 1962, a British plastic surgeon, Patrick Clarkson, coined the term Poland syndactyly. PS is a term that is more accurate because the group of anomalies includes more than just syndactyly and was given by Baudinne et al. (1967) [1].

PS cases present with absence or underdevelopment of pectoralis major muscle, associated sometimes with or without a hypoplasia of the breast, an agenesis of 2, 3, 4, and 5 ipsilateral costal cartilages, an athelia, and an ipsilateral webbing of the fingers (cutaneous syndactyly) [2, 3]. Therefore, PS may occur with varied presentation. At present, it is assumed that PS is characterized by a missing sternocostal bundle of the pectoralis major muscle [4].

The incidence ranges between 1:7,000 and 1:100,000 births, with higher frequency among males (ratio: 2:1–3:1). In 75% of the cases, it is located on the right hemithorax in the unilateral form [2, 4]

Affected individuals may have variable associated features, such as underdevelopment or absence of one nipple including the areola and/or patchy absence of hair in the axilla [2, 5]. In females, there may be underdevelopment or aplasia of one breast and underlying (subcutaneous) tissues [8]. In some cases, associated skeletal abnormalities may also be present, such as underdevelopment or absence of upper ribs; elevation of the shoulder blade (Sprengel deformity); and/or shortening of the arm, with underdevelopment of the forearm bones (i.e., ulna and radius) [2, 5]. Rare association includes dextrocardia, renal hypoplasia, leukemia, and Moebius syndrome [3].

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Author's Photo Gallery



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Figure 1: X-ray posteroanterior view of chest showing no obvious bony abnormalities.

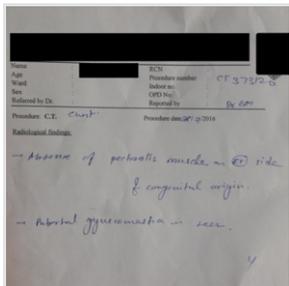


Figure 2: Computed tomography scan report confirming the diagnosis of Poland syndrome.

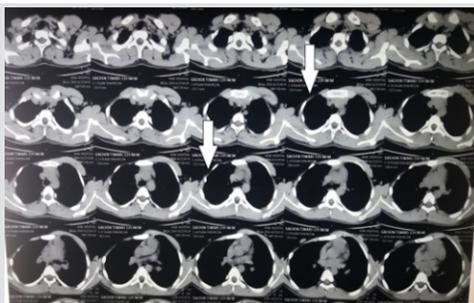


Figure 3: Axial section cut of computed tomography scan showing the absence of pectoralis major muscle.

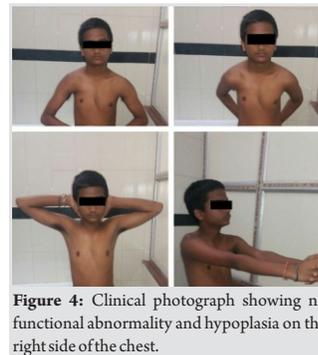


Figure 4: Clinical photograph showing no functional abnormality and hypoplasia on the right side of the chest.

Case Report

A 13-year-old male student presented to a tertiary care center with a flattening of the right anterior chest wall since birth. He was asymptomatic with only cosmetic concerns. He was the second child, and there were no similar presentations in the family or any other associated familial disorder.

Examination revealed normal growth parameters and IQ. There was no abnormal craniofacial finding. His chest wall was asymmetric with hypoplasia of the right side (Fig. 4). The patient had a normal power in ipsilateral upper limb with full range of motion of the shoulder joint (Fig. 4) His limbs were bilaterally symmetric with no finger anomalies. Auscultation of the chest revealed normal heart sounds, breath sounds, and respiration pattern.

Computed tomography scan (Fig. 2) revealed that the pectoralis major muscle was underdeveloped, but the pectoralis minor muscle and the anterior serratus muscle were present. Radiological examination of the chest showed no abnormalities of the ribs or heart (Fig. 1). Based on physical findings and the radiological picture, we diagnosed it as a case of PS.

In view of the good function of the upper limb, no surgical treatment was offered and the patient and his family were counseled regarding the condition. A follow-up of the patient at 2 years revealed that the patient is still asymptomatic with good functional status of the upper limb.

Discussion

The case presented here was one of the few cases with isolated pectoralis muscle involvement and had peculiar presentation as it involved only the unilateral aplasia of the pectoralis major muscle in the right hemithorax without any other associated defects [2, 4].

The pectoralis major has many actions primarily responsible for movement of the shoulder joint. These are flexion, adduction, and medial rotation of the humerus. It is also responsible for keeping the arm attached to the trunk. Aplasia of the pectoralis muscles and associated chest defects are consequences of an interruption of early embryonic blood supply of subclavicular

artery branches [6, 7]. A combination of the blockage of various branches could lead to different manifestations of PS. The interruption of the blood supply may be caused by a thrombus or embolus or due to maldevelopment of vessels.

However, there have been case reports of PS associated with unusual defects, which cannot be explained on the basis of compromised blood supply alone. Hence, the true pathogenesis of PS is still a matter of debate.

Most cases of PS are sporadic and occur in people with no history in their families. Rarely, this condition is passed through generations in families in an autosomal dominant pattern although no associated genes have been found. There are rare instances where more than one individual has been identified with PS either in the immediate [7, 10] or extended family [10]. Therefore, some authors believe that the central underlying mechanism for this condition may be an inherited abnormal vasculature formation. Several reconstructive procedures are available to correct the deformities such as customized silicone prosthesis and transposition of the latissimus dorsi which have given satisfactory esthetic and functional results [9].

Conclusion

It is hoped that this paper will further improve our understanding of this rare syndrome and its atypical presentations.

Clinical Message

Poland Syndrome is a rare syndrome of unknown cause with a classical presentation of underdeveloped chest muscle and upper limb anomalies typically on the right side. It may be associated with varying anomalies.

It is hoped that this paper will further improve our understanding of this rare syndrome and its atypical presentations.

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