

## Case Report

# Poland's Syndrome with Unusual Hand and Chest Anomalies: A Rare Case Report

G.K. Yadav<sup>1</sup>, S. Lal<sup>2</sup>, N. Dange<sup>1</sup>, K.G. Marwah<sup>1</sup> and J.P. Singh<sup>1</sup>

Department of Surgery, ESI-Dental College<sup>1</sup>, Rohini and ESI-PGIMS<sup>2</sup>, Basaidarapur, New Delhi, India

## Abstract

Poland's syndrome is a rare congenital anomaly consisting of a unilateral absence of the pectoralis major, ipsilateral muscle, hand anomaly and occasionally associated other malformations of the chest wall and breast. Many structural and functional abnormalities have been described in association with this syndrome. We report an incidentally diagnosed case in a 27-year-old male patient who presented to us with symbrachydactyly. In addition to this, anterior depression of 2nd, 3rd and 4th ribs and bifid (forked) 5th rib was present on radiological investigations. The body of sternum was short and deformed on the right side with absence of xiphoid process. All middle phalanges were absent on right hand. It is a rare variant of Poland's syndrome. [Indian J Chest Dis Allied Sci 2014;56:191-194]

**Key words:** Poland's syndrome, Bifid ribs, Congenital anomalies, Symbrachydactyly.

## Introduction

Poland described a case of unilateral absence of the pectoralis major muscle in 1841.<sup>1</sup> He was the first to report the abnormalities in this syndrome, without able to name it. Thompson (1895)<sup>2</sup> first described the full anatomical spectrum of the disease and Clarkson (1962)<sup>3</sup> coined the name Poland's syndactyly. Baudinne *et al*<sup>4</sup> described the term Poland's syndrome (PS) in 1967. This condition is also known as subclavian artery supply disruption sequence (SASDS), fissure thoracislateralis, pectoral-aplasia-dysdactyilia syndrome and unilateral chest-hand deformity. The clinical presentations are variable, and therefore, features are rarely present in the same patient. The typical components of this syndrome include ipsilateral hypoplasia or absence of the sternocostal portion of the pectoralis major muscle with associated hand defects.<sup>5</sup> The reported incidence of PS ranges from 1 in 7000 to 1 in 100,000. Most cases are sporadic and tend to occur in males (2-3:1) and is more commonly seen on the right side (2:1). Familial cases are rare, equal in both sexes with no side predominance.<sup>6</sup> Ten percent of these patients also express other features of PS.

We describe a case of PS in a 27-year-old male with absence of all middle phalanges of the right hand and other rare anomalies.

## Case Report

A 27-year-old male presented with a complaint of hand deformity on the right side that was present since birth. On physical examination, the pectoralis major muscle was absent and there was a reduction of hairs on the chest and in the axillary region on the right side.

Whereas the left chest and upper arm were found to be normal. Symbrachydactyly was present in the right hand (Figure 1).



**Figure 1.** Photograph of the patient showing absent anterior axillary fold, reduction of hair, and hypoplastic chest and hand on right side. Nipple on right side was higher than left.

The power was 4/5 and 5/5 in the right and left hand, respectively. Routine haematological and biochemical investigations were within normal limits. On hands radiograph, all the middle phalanges were found to be absent leading to single interphalangeal joints on the right side, while the left hand had a normal anatomy (Figure 2). A chest radiograph revealed hyperlucency of the hemithorax, with bifid (forked) 5th rib on the right side with a normal appearing heart shadow (Figure 3). Ultrasonography of the abdomen and electrocardiogram did not reveal any abnormality.

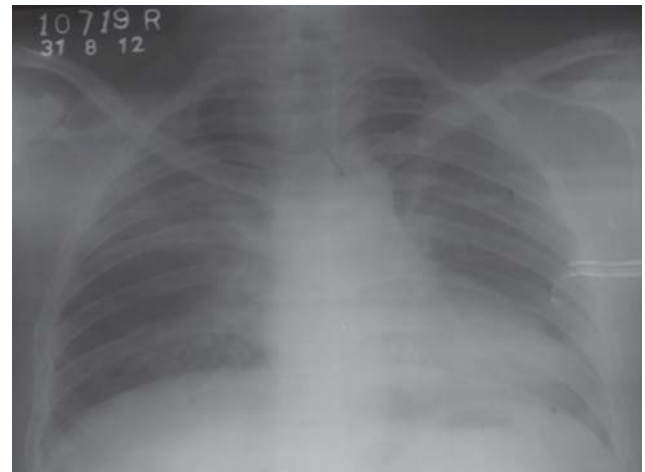
[Received: March 12, 2013; accepted after revision: May 2, 2014]

**Correspondence and reprint requests:** Dr S. Lal, 2/25 A, Moti Nagar, New Delhi-110 015; Phone: 91-9811155883; E-mail: slaldr@gmail.com



**Figure 2.** Hands radiograph shows absence of all middle phalanges, shortening of all metacarpals and phalanges on right.

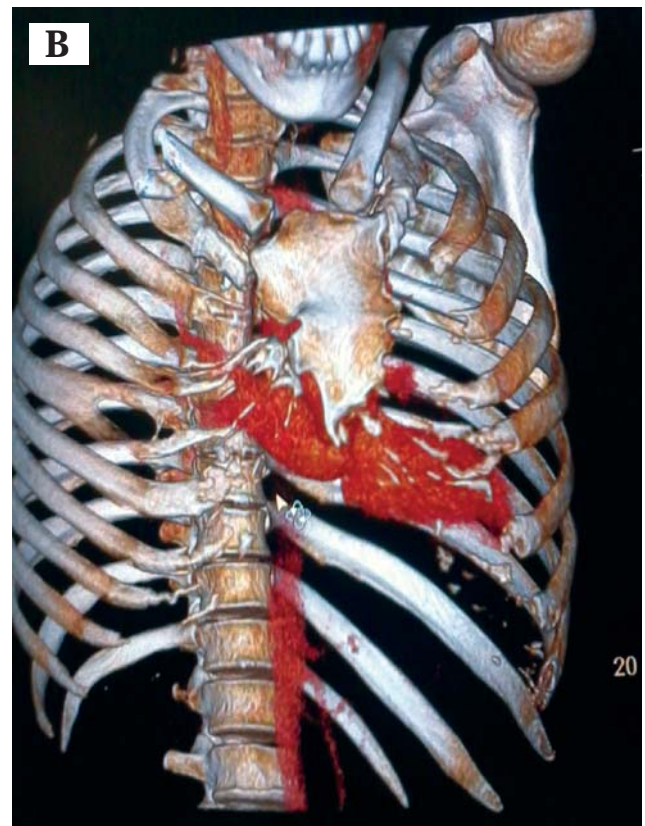
Computed tomography (CT) of the chest showed absence of the pectoralis major and minor muscles, an anterior depression on the 2nd, 3rd, and 4th ribs, and bifid (forked) 5th rib on the right side. Moreover, the body of sternum was short and deformed on the right side with absence of the xiphoid process (Figure 4 A & B). None of the family members had reported similar anomalies.



**Figure 3.** Chest radiograph postero-anterior view showing hyperlucency of the lung with bifid fifth rib on the right side.

### Discussion

Poland's syndrome is of unknown aetiology. Many theories have been proposed, but the most accepted one is the interruption of the embryonic blood supply in the sixth week of gestation to the subclavian artery/vertebral arteries and their branches leading to diminished blood flow to the affected side, with partial



**Figure 4 A and B.** Computed tomography of chest showing absence of pectoralis major and minor muscles, anterior depression on 2nd, 3rd, and 4th ribs, and bifid 5th (forked) rib on the right side. Moreover, the body of sternum was short and deformed on the right side with absence of xiphoid process.

loss of tissue in that region. Vascularisation of the fetal skin begins during the third month of intrauterine life, but these vessels do not anastomose with the deeper vasculature until late in gestation. These vascular disruptions may also lead to its relation with syndromes like Sprengel deformity (congenital elevation of the scapula), Klippel-Feil and Moebius syndrome (unilateral or bilateral facial palsy and abducens oculi palsy) and Adams-Oliver syndrome (aplasia cutis congenital and malformations of the limbs).<sup>9,10</sup> Another hypothesis is that a disruption of the lateral plate mesoderm (from which the pectoralis muscle develops) between 16 and 28 days after fertilisation may account for all the defects.<sup>10</sup> Exposure to ergot alkaloids and smoking in the first trimester of pregnancy are known to be associated with the occurrence of the PS.<sup>11</sup>

The clinical manifestations of PS are extremely variable. Bifid rib, sternal deformity with complete absence of all the middle phalanges are a rare variant. Absence of sternal head of pectoralis major muscle is the minimal expression. Involvement of adjacent muscles, including pectoralis minor, serratus anterior, latissimus dorsi, external oblique, infraspinatus and supraspinatus muscles have also been described. The skin of the area is hypoplastic with a thinned subcutaneous layer. Axillary hair may be absent on the affected side. Ipsilateral nipple is often smaller and higher. The breast is generally hypoplastic in females. Rare anomalies include absent ribs or costal cartilage, lung herniation and Sprengel deformity. In our case, there was a depression in the anterior portion of the right 2nd, 3rd and 4th ribs, and a bifid (forked) 5th rib. In addition, body of the sternum was short and deformed on the right side with an absence of xiphoid process. Bifid (forked) rib is extremely rare in PS. The overall prevalence of bifid rib is estimated to be 0.15% to 3.4% (mean 2%), and it accounts for up to 20% of all congenital rib anomalies.<sup>12</sup>

Upper limb abnormalities in PS vary from a shortness of the middle phalanges with cutaneous webbing (syndactyly), to a complete absence of the hand (ectrodactyly). Contralateral hand is rarely involved.<sup>13,14</sup> Hand malformations occur only in 12% of the patients with PS.<sup>13,15</sup>

Aplasia of the middle phalanges on the ulnar side of the hand is more common than on the radial side. In our case, there was aplasia of all the middle phalanges in the right hand leading to single interphalangeal joints.

Various associated anomalies including diaphragmatic hernia and those of the liver and biliary tract, kidney, testes, and heart (dextrocardia) have been reported and neoplasias such as leukemia, non-Hodgkin's lymphoma, cervical cancer, leiomyosarcoma, breast cancer, may occur.

The diagnosis is important as this condition may be associated with several visceral anomalies and neoplasia. Chest radiographs and abdominal ultrasound scan

should be obtained. A CT scan helps to demonstrate the extent of the muscle abnormalities and provides useful information for planning any reconstructive surgery. The syndrome can also be diagnosed prenatally using ultrasonography<sup>16</sup> and sometimes is detected as an incidental finding in patient of syndactyly.

The treatment of patients with PS, due to the variable clinical picture, is individualised and depends on the patient's age, gender, pathology and severity.<sup>17</sup> Functional disability is minimal, and therefore, a patient may seek a surgical opinion only for cosmetic reasons. Other indications are paroxysmal movements of the chest wall and progressive lung herniation. Several reconstructive procedures are available to correct the functional and structural deformities of the chest such as flaps (latissimus dorsi, rectus abdominus and omental) lipofilling, and custom-made silicone prosthesis and/or breast prosthesis.<sup>18</sup> The syndactyly of PS has many variations, as noted before, and is usually the first component to be repaired. The treatment of syndactyly should be carried out at the pre-school age.

To conclude, Poland's syndrome is an uncommon condition but easy to diagnose. Poland's syndrome may be an incidental finding because of mild functional disability. Patients may present for evaluation of other associated congenital anomalies or for aesthetic treatment. Presence of syndactyly should prompt a search for other features of PS. The well-known association between the developmental anomalies and tumours calls for awareness and regular follow-up. The treatment is individualised and depends on the patient's age, gender and severity of the deformity.

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