

Poland Syndrome: A Rare Case Report from Palestine

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Abstract

Poland syndrome (PS) is a rare chest wall congenital anomaly. PS was first described by Alfred Poland in 1841, characterised by ipsilateral agenesis/hypoplasia of the sternocostal head of pectoralis major, hypoplasia of nipple or breast, absence of subcutaneous fat, multiple rib abnormalities, elevated and rotated scapula (Sprengel deformity) and ipsilateral digit abnormalities (brachydactyly, syndactyly). The etiology is unknown, but speculation includes hypoplasia of the subclavian artery or its branches between the 6th and 7th week of gestation. The condition is generally not inherited from a person's parents. Treatment is primarily reconstructive surgery depending on the severity of the malformation, gender and patient preference. We present a 3^{6/12} female child with features of depressed right chest-wall, associated with nipple hypoplasia, small scapula with winging. But absent upper limb deformities. There is negative history of other family members of being affected.

Keywords: Limb Anomalies; Poland's Syndrome; Syndactyly

Abbreviation

PS: Poland Syndrome

Introduction

The Poland's syndrome is also known as (Poland's syndactyly, Poland sequence, and Poland's anomaly). Poland's syndrome was initially reported by Sir Alfred Poland in 1841 as an absence of the pectoralis major and minor muscles and malformation of the ipsilateral upper limb [1]. The true incidence of Poland anomaly is unknown, PS has an estimated incidence of 1/7,000 to 1/100,000 live births, this syndrome affects males three times as often as females and is sporadic in nature [2,3]. Because of its variability in presentation, PS is often undiagnosed at birth., milder forms of the disease may not be detected until late adolescence and puberty, when right- or left-sided differences of the body start to manifest [4]. PS cases present with absence or under-development of pectoralis major muscle, associated in some cases with or without a hypoplasia of the breast, athelia, patchy absence of hair in the axilla, deformity or aplasia of the costal cartilages or ribs II to IV or III to V, and an ipsilateral cutaneous syndactyly or brachysyndactyly [5-7]. The right side of hemithorax is affected in two thirds of patients [8,9] and bilateral absence of the pectoralis major muscle has been reported in the literature [10]. In females with PS, there may be irregular, asymmetric breast development or aplasia of one breast and underlying (subcutaneous) tissues, prompts them to seek treatment [11]. This syndrome is associated with various anomalies such as under-development or absence of upper ribs, elevation of the shoulder blade (Sprengel deformity), ipsilateral syndactyly, brachydactyly, dextrocardia, herniation of lung, aplasia or hypoplasia of breast, patchy absence of hair in the axilla and/or shortening of the arm, with under-development of the forearm bones [6,8,12]. Other associated abnormalities may include diaphragmatic hernia and renal anomalies, leukemia, neuroblastoma, leukemia, leiomyosarcoma, breast cancer and Wilms tumour and Mobius syndrome. etc. [7,13,14]. This syndrome has variable clinical features and rarely are all the features recognized in one individual [15]. Patients affected by this anomaly often experience body image disorders and decreased quality of life [16].



Case Report

We report a case of a 36/12 -years old female, second born of consanguineous parents, normal spontaneous delivery, developmentally normal, with weight of 12.3 kg below the 10th percentile, height 94 cm below the 25th percentile. Presented to our El-Doura Pediatric hospital because of unilateral “anterior thoracic depression” of the chest wall since birth associated with nipple hypoplasia, and subsequent asymmetry.

Patient had normal vital signs, pulse rate 92/min, respiratory rate 34/min, temperature 37C, BP 80/60 mmHg, oxygen saturation at room air 94%.

Physical examination showed chest asymmetry with right anterior chest wall depression and flattening of the right pectoral region with displaced nipple. The sternocostal part of pectoralis major was absent on left side, left nipple was placed at lower level and there was prominence of costochondral junction of the affected side. There was asymmetry of both shoulder, the right one is lower than the other. Hand examination did not show any signs of ipsilateral syndactyly or brachydactyly. Apex beat was in 5th intercostal space in midclavicular line. Heart sounds were audible and there was bilateral equal air entry in both lungs. No other systemic abnormality was found.

Investigations reveal: CBC, S/E, KFT, LFT, Cholesterol level (141 mg/dl), Triglyceride (87 mg/dl), RBS (80 mg/dl), uric acid (2 mg/dl), Phosphor (5,0 mg/dl), Ca (10.9 mg/dl). Chest x-ray was normal, Skeletal survey show normal study, abdominal U/S show normal study.

Discussion

Poland’s syndrome (OMIM 173800) is characterized by a great diversity in the clinical manifestations [17]. Two forms of PS are known, the complex one presents early and is associated with multiple chest wall and limb anomalies. The simple form is usually diagnosed later in life and presents with chest wall asymmetry without associated limb anomalies [18]. It was named after Sir Alfred Poland, who first published it in 1841 [19].

PS comprises a spectrum of absence or underdevelopment of the pectoralis muscles in combination with Syndactyly in the ipsilateral hand, which can be either complete or incomplete. Other deformities can include scoliosis, abnormalities of the ribs ranging from hypoplasia to complete absence of the ribs and lungs, upper extremity anomalies: Short upper arm, forearm or fingers (brachy symphalangism), hypoplasia or aplasia of breast or nipple, and deficiency of subcutaneous fat, deficiencies of the skin, sweat glands, radial nerve aplasia and hair of affected areas [20-23]. Other muscles may be affected at the same affected site including the serratus, latissimus dorsi, and the external oblique [21,23-26].

Etiology of Poland anomaly remains unclear but is thought to be secondary to defect of inadequate perfusion of subclavian artery branches or mechanical factors during the sixth week of gestation [27]. It is a period associated with splitting of the two heads of

pectoralis major and the development of tissues between the digits [19]. It may be associated with chest defects, as the athelia, aplasia of costal cartilages [27,28]. However fetal skin vascularization begins during the third month of intrauterine life, these vessels do not anastomose with the deeper vasculature until late in gestation [29]. Other etiologic factors of the PS are taken into account: genetic and also teratogenic effect e.g., exposure prenatally to noxious and physical environments such as smoking, viral infections [30,31], cocaine [32], misoprostol [33], disruption of the lateral mesodermal plate soon after fertilization [34] and failed abortions [10].

Geneticists agreed that PS is rarely inherited and in most cases are sporadic event. However familial recurrence with higher prevalence in males has been observed, which suggests a genetic basis of this congenital anomaly [10,35-37], although autosomal dominant, multifactorial and para-dominant inheritance also have reported [11]. Vaccari, *et al.* (2014) stated that phenotype concordance between the monozygotic twin probands provides evidence supporting the genetic control of PS. They postulated that the observed chromosome deletion 11q12.3, could be play a role in PS development [38].

Female 3^{6/12} patient has presented to our hospital due to inequality of both chest wall, her mother observed chest wall deformity since birth, in our patient we found hypoplasia of right chest wall, hypoplasia includes muscle, subcutaneous fat, right areola hypoplasia and associated with Sprengel deformity but no evidence of other deformities.

Skeletal deformities may include bone and cartilage defects of the ribs (II to IV, or III to V are most commonly involved), clavicle, and sternum [6,39], scapula may be smaller with winging and the mobility is always restricted a condition known as Sprengel deformity [10,34], scoliosis [9], lung herniation [8] dextrocardia [10] and absence or underdevelopment of the breast or areola.

Left-sided PS has been associated with dextrocardia particularly in patients with agenesis of two or more ribs and is thought to be secondary to reduced volume of the left hemithorax [14,40]. Other anomalies have been associated with Poland anomaly includes anterior thoracic wall, breast, diaphragm and vertebrae as hemivertebra and scoliosis [41].

The upper extremity also may be hypoplastic and can be classified according to the severity. The upper arm, forearm, and fingers may be shortened, which is termed brachy symphalangism. Hand anomalies occur from 13.5% to 56% in Poland anomaly [42,43].

In our patient we did not found hand deformity or upper limb shortening, but she has pectoral hypoplasia with the same side hypoplasia of the areola associated with Sprengel deformity. Hence, according to the diagnostic criteria our patient has Poland syndrome.

Conclusion

Classical PS is represented by unilateral aplasia of the sternocostal head of the pectoralis major muscle and ipsilateral hand abnormalities and other associated anomalies. However, the absence of upper limb deformities, in the presence of abnormal pectoralis muscle and ipsilateral rib anomalies, does not rule out PS.

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