

## Poland syndrome with some rare associations, and brief literature review

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### ABSTRACT

Poland syndrome (PS) is a rare congenital condition with predominant unilateral chest wall deformity due to hypoplasia of the pectoralis muscles. However, its clinical features are highly variable as all the features may not be present in one individual or it may present with some rare associations or complications as being reported here. A 6-year-old boy was diagnosed, for the first time, as a case of PS but, detailed examination and work up revealed two rare associations and/or incidental findings of this disorder, namely, Dandy-Walker variant and partial anomalous pulmonary venous return. Hence, a detailed clinical examination and a thorough workup are mandatory to quantify the disease spectrum of this rare disorder.

**Key words:** *Pectoralis, Poland syndrome, Rare associations*

Poland's syndrome (PS) is a rare congenital condition comprising a spectrum of chest wall deformities affecting the rib cage, the chest wall muscles, neurovascular structures of the ipsilateral arm and the overlying breast, to variable extent [1]. The disease is sporadic in nature, and usual incidence varies from 1 in 7000 to 1 in 100,000 live births with higher frequency among males (male:female ratio of 3:1) [2]. Due to the high variability in the clinical features and absence of all features in the same individual, it will be very much prudent to undertake a complete physical examination and accurate investigations to exclude other organ anomalies in each and every patient of PS.

Here, we are reporting a classical case of PS who presented to us for cosmetic reason but, on detailed investigations we found some rare associations and/or incidental findings of this disorder which have not been reported in previous literature.

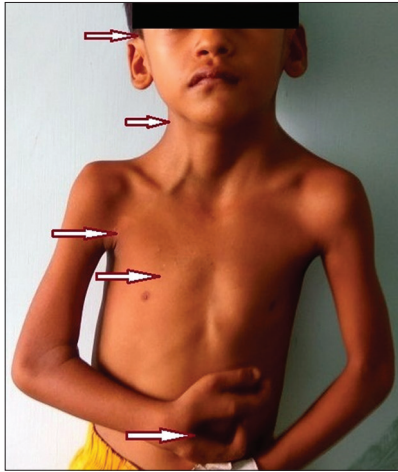
### CASE REPORT

A 6-year-old boy presented to our pediatric outpatient department with deformity of the chest wall, and vertebral column noticed by his mother since early infancy and became more prominent with an increase in the age. He had no chest pain or any limitation of chest movement. He was the second child and born of non-consanguineous parentage with an uneventful antenatal and postnatal period. Past history was unremarkable. There was no family history of any congenital disorder, specifically coagulation disorders. His siblings were all alive and healthy. He achieved all developmental milestones normally. His scholastic performance was not so satisfactory. On examination, the child had a weight of 16.5 kg, height of 122 cm, body mass index of 11.08 kg/m<sup>2</sup>

which were between 3<sup>rd</sup> and 15<sup>th</sup> percentiles, between 85<sup>th</sup> and 97<sup>th</sup> percentiles, and <3<sup>rd</sup> percentiles of the WHO growth charts, respectively. He had some pallor with stable vitals. His heart sounds, respiration, and breath pattern were normal as well. Head to toe examination showed microcephaly (head circumference 48 cm), low-set ear, webbing of neck, asymmetric chest with depression of the right anterior chest wall and absence of the right axillary fold, syndactyly of the index finger with the middle one and ring finger with the little finger of the right hand, winging of scapula on right side and kyphoscoliosis (Figs. 1 and 2). His neurological examination was normal except for the borderline mental retardation (intelligence quotient, IQ = 75) and the power of the muscles of the right and left hand was 4/5 and 5/5, respectively. The movements around the joints of the involved hand were restricted, although all the other joint movements showed a normal range. Radiological examination revealed an asymmetric chest with reduction of the third to fifth intercostal spaces with loss of lung volume on the right (Fig. 3). Left pectoralis muscle was hypoplastic in chest wall ultrasonography. Routine hematological and biochemical investigations did not reveal any abnormality except for mild anemia (Hb - 9.1 g%). 2D-echocardiography with color Doppler study showed a partial anomalous pulmonary venous return (PAPVR). Magnetic resonance imaging brain revealed Dandy-Walker variant (DWV) (Fig. 4).

### DISCUSSION

In 1841 Sir Alfred Poland first described this anomaly as a syndrome presenting with absence or underdevelopment of pectoralis major muscle, associated in some cases with a



**Figure 1:** Front view of the patient showing low set ear, webbing of the neck, poorly developed anterior axillary fold, depressed anterior chest wall, syndactyly of fingers on the right side



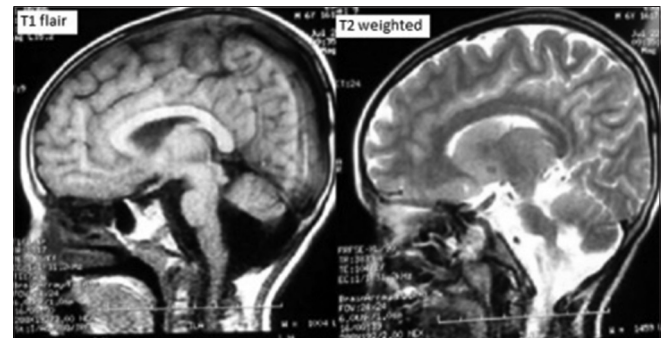
**Figure 2:** Image of the back of the patient showing Sprengel deformity and kyphoscoliosis

hypoplasia of the breast, agenesis of ipsilateral second, third, fourth, and fifth costal cartilage, anhelias, and ipsilateral webbing of the fingers (cutaneous syndactyly) [3]. At present, it is assumed that the missing sternocostal bundle of the pectoralis major muscle is the characteristic feature of PS [4]. In 75% of the cases, involvement of the right hemithorax is seen, particularly in the unilateral form [4,5]. The PS cases are increasingly being detected with various presentations and/or associations, such as underdevelopment or absence of one nipple including the areola and/or patchy absence of hair in the axilla [3]; underdevelopment or aplasia of one breast and underlying (subcutaneous) tissues in females. Other deformities that may be associated in few cases of PS are skeletal abnormalities, 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), preaxial polydactyly, hemivertebra may also be present [3]. Till date, there are no reports of cases who presented with kyphoscoliosis like ours.

Other organ system-related anomalies those are very rarely associated with PS include microcephaly, cerebral atrophy, encephalocele, abnormal morphology and function of



**Figure 3:** Chest X-ray (posteroanterior view) showing contracted lung volume with crowding of upper ribs over right lung field with loss of lung volume over lower lung zone



**Figure 4:** T1 flair and T2 weighted magnetic resonance imaging of brain showing small posterior fossa cyst communicating with 4<sup>th</sup> ventricle with hypoplasia of inferior vermis of cerebellum – suggestive of Dandy-Walker variant

hypothalamic-hypophyseal axis, paralysis of the cranial nerve or mental retardation, situs inversus or dextrocardia, gastroschisis, endocrine anomalies, melanosis, and congenital diaphragmatic hernia [6]. Urinary system anomalies such as agenesis/hypoplasia of kidneys, ureteric anomalies, and hypospadias have also been reported to occur with increased frequency in PS patients. PS cases are at increased risk of neoplasias such as leukemia, Non-Hodgkin's lymphoma, and childhood solid tumors such as neuroblastoma and Wilms' tumors [7]. Microcephaly was seen in our case, but DWV and PAPVR which were discovered in our patient have not been described in previous literature. The DWV may be associated with both extra- and intracranial anomalies, but isolated DWV usually carries a good developmental outcome [8] as seen in our case. Among intracranial associations, corpus callosal dysgenesis, ventriculomegaly, and vermian rotation may be observed. Observations of Paladini and Volpe (2006) that the degree of vermian hypoplasia correlates significantly with the occurrence and severity of mental retardation explain the lower IQ in our case [9].

Although the underlying etiology of PS is yet to be discovered; it is assumed that a momentary interruption or reduction in the circulation of the subclavian and vertebral arteries or one of their peripheral ramifications, during the 6<sup>th</sup> week of embryonic development, primes the pathogenetic mechanism

of the syndrome [4,5]. Depending on the length and intensity of the vascular interruption, different degrees of severity of the syndrome occurs [2]. The interruption of the blood supply may be caused by thrombus or embolus or the misdevelopment of vessels. However, this theory of compromised blood supply alone cannot explain some reported cases of PS associated with unusual defects. Moreover, Ferraro and colleagues [4] described an unusual presentation of the Poland's anomaly without any vascular alteration, making this theory in question. Current view of various genetic researchers on PS is that it is generally a sporadic event and rarely inherited. There are rare instances where PS has been reported to affect more than one individual either in the immediate or extended family [10]. Therefore, some authors believe that the principal underlying mechanism for the development of the condition may be an inherited abnormal vasculature formation.

To conclude, the exact etiology of the PS is unknown, but it can be diagnosed very easily, even only with good clinical eye. These cases may be detected incidentally due to minimal functional disability. Patients may present with the features of other associated congenital anomalies, or they come for cosmetic reasons. Our findings also reinforce that occurrence of PS is sporadic in nature, a thorough search for detection of other associated organ system anomalies should be carried out in all cases of PS, and more research is needed to quantify the disease spectrum of this syndrome.

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