INTRODUCTION

Poland’s syndrome (PS) is uncommon but not rare, occurring in 1:30,000 live births. It was named after Sir Alfred Poland, who first published it in 1841. Patrick Clarkson associated Poland’s name to the syndrome when he published his series of three similar cases 81 years later in the same institution, the Guy’s Hospital in London.

PS present as ipsilateral involvement of the chest muscles (absence of the costosternal portion of the pectoralis major muscle, lack of the pectoralis minor muscle), hypoplasia/aplasia of breast or nipple, deficiency of subcutaneous fat, aplasia or deformity of the costal cartilages or ribs II to IV or III to V and alopecia of the axillary and mammary region and upper extremity anomalies: Short upper arm, forearm or fingers (brachysymphalangism). However, clinical manifestations of PS are extremely variable and rarely are all the features are recognized in one individual.

Ipsilateral limb involvement is one of the most frequent signs of PS. However, we present a case of a newborn with the classical features of syndromes, but with no limb deformities.

Because clinical features are highly variable and not all present in the same individual, patients with PS should undergo an accurate physical examination and investigations to exclude renal, cardiac and other important anomalies.

CASE REPORT

We report the case of an Egyptian full term male neonate born to mother who had gestational diabetes controlled by diet. Baby was born via spontaneous vaginal delivery with Apgar score of 8 and 9 at 1st and 5th min. Birth weight was 3.44 kg (50th percentile), length was 52 cm (>50th percentile) and head circumference was 36 cm (50th percentile). He was apparently well at birth with no respiratory distress and was shifted to the postnatal ward at 1 h of age.

Initial physical examination showed right anterior chest-wall depression, lateral to the nipple. There were no other anomalies seen on further systemic evaluation. Lower limbs, nails and hair were normal. Neurological examination was normal. Skeletal survey was normal.

Computed tomography scan of the chest showed a small hypoplastic right pectoralis major [Figure 1] with underlying hypoplastic tapered anterior end of the right 4th, 5th and 6th ribs [Figure 2]. There is also a focal asymmetry with reduced subcutaneous fat at the right hemithorax anteriorly. There was no associated herniation of the lungs.

These anomalies were compatible with the diagnosis of PS.

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Echocardiography did not show associated cardiac anomalies. However, abdominal ultrasound was not done.

**DISCUSSION**

In PS, males predominate by at least 3:1 and usually the right side is affected in 75% of cases though there is a case report of a PS with bilateral features. It has a sporadic occurrence in the population without defined inheritance. A total of 15 reported cases of familial PS suggested an autosomal dominant inheritance with incomplete penetrance. This familial transmission usually arises from sporadic mutation during development.

The exact etiology of PS is unknown. Embryologically, at 6th week of gestation, the pectoral mass splits into a clavicular head and a costal head. This is also the time when tissues between the digits of the hand start to disappear to form web spaces. It appears that an insult during the 6th week of gestation may be responsible for anomalies in the pectoralis muscle and hand anomalies. The findings of regional vascular hypoplasia have suggested a vascular etiology. It results from an interruption of the early embryonic blood supply in the subclavian arteries, the vertebral arteries and/or their branches and hypothesize that the occlusions occur at specific locations in these vessels during or around the 6th week of embryologic development and produce predictable patterns of defects. The term subclavian artery supply disruption sequence is suggested for the group of birth defects represented by the above conditions. PS has been associated with other syndromes, including Möbius syndrome (congenital bilateral facial paralysis with an inability to abduct the eyes) and Klippel-Feil syndrome. Another hypothesis suggested disruption of the lateral embryonic plate mesoderm between 16 and 28 days after fertilization, which may account for all the defects.

Non-hodgkin's lymphoma and leukemia, unilateral peripheral facial palsy, ipsilateral renal hypoplasia have also been described in patients with PS. Our patient presents with hypoplasia of the right pectoralis muscle but absent limb anomalies. A similar case was reported in Taiwan, but the left side was affected. In Shamberger’s landmark review, 50 of 75 patients had ipsilateral syndactyly. The more severe brachydactyly was less common. Hand anomalies in PS were reported to occur from 13.5% to 56% respectively. Isolated pectoral hypoplasia (with normal hand) usually occur in familial PS. It is a variety of PS because, in familial cases, one family member may show an isolated pectoral hypoplasia and another may have combined hand-pectoral deformity. Those patients without hand involvement are defined as “partial Poland's sequence”. Hence, the diagnostic criteria of the syndrome should include isolated absence of the pectoralis major muscle with breast hypoplasia.

Hand anomalies in PS have been initially classified into four types. The isolated pectoral anomaly (with normal hand), which was well described in familial cases of PS, was not included in their classification. Al-Qattan in 2000, extended the classification into seven types. Type 1, having a normal hand; type 2, forme fruste deformity (one hand appears smaller than the contralateral side); type 3, brachysyndactyly (classic deformity); type 4, some functional rays still present; type 5, all digits are functionless or absent; type 6, transverse deficiency proximal to the metacarpophalangeal joints; and type 7, phocomelia-like deficiency.

The rib cage itself in PS may be entirely normal; however, in most patients the chest-wall is sunken on one side. The latter is caused by hypoplasia of the ribs and cartilages, which are deformed and thin. As stated before, ribs II to IV, or III to V are most commonly involved, but the
second rib is less frequently affected. Aplasia of the anterior portions of between one and three ribs, with severe chest-wall depression occurs in about 11-25% of patients.[4] Multiple rib anomalies can cause respiratory dysfunction, as was featured in one report.[17] In our case, the right 4th and 5th ribs were affected and even extended up to the 6th rib. Though multiple ribs were affected on the ipsilateral side, there was no compromise to his respiratory function.

CONCLUSION

Based on the above report it can be concluded that classical PS is represented by unilateral aplasia of the sternocostal head of the pectoralis major muscle and ipsilateral hand abnormalities[8,18] and a variety of 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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