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Case Report

Poland syndrome with associated synbrachydactyly and dextrocardia: a rare case

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ABSTRACT

Poland's Syndrome is a rare congenital condition. It is classically characterized by 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 or a breast portion of the pectoralis major muscle which may or may not be associated with the absence of nearby musculoskeletal structures. Authors report a 13-year-old male patient with typical features of Poland syndrome associated with synbrachydactyly and dextrocardia. This association is very rare, till 2010, only 19 cases have been described in scientific literature (Poland syndrome with dextrocardia), in this case synbrachydactyly also associated on ipsilateral side. To the best of our knowledge, this is the first documented case of the left sided Poland syndrome with dextrocardia and symbrachydactyly reported from the South East Asia Region (SEAR).

Keywords: Dextrocardia, Poland syndrome, Synbrachydactyly

INTRODUCTION

The Poland's anomaly was first described in 1841 by Sir Alfred Poland. Poland syndrome is a rare birth defect with an average incidence of Poland syndrome in the USA is estimated to range from one in 7,000 to one in 100,000 live births.[1] The main component of this syndrome is an underdevelopment or absence of the pectoralis major muscle on one side of the body.[2] There are many clinical variations of the syndrome including rib defects, absence of shoulder girdle muscle and breast hypoplasia or agenesis. Dextrocardia is rarely associated with Poland Syndrome with only 22 cases being previously reported in the worldwide literature and associated synbrachydactyly is even more rare. Whereas 'classical' Poland syndrome is predominantly right sided, all cases associated with dextrocardia have been left sided. Sometimes It is also associated with hand abnormalities like underdeveloped hands with abnormally short fingers (brachydactyly), fused fingers (syndactyly), or both (synbrachydactyly).[3,4] Other rare conditions associated with this syndrome are rib defects, scoliosis, dextrocardia and renal hypoplasia.[5] Poland syndrome affect the right side in approximately 65-75% of the cases and a male to female ratio 3:1 has been reported.[2,3,6,7]

CASE REPORT

A 13-year-old, school boy, presented to our institute with the chief complaints of dry cough from last 8 days. During a routine physical examination, we incidentally

found that he had a congenital deformity of chest wall and ipsilateral hand. His chest wall was found asymmetrical. This flattening and asymmetry of the left anterior chest wall were since birth. There was no familial history of similar disorder. The patient had normal growth parameters and was mentally alert. There were no unusual cranio-facial findings.

His chest was asymmetric, with hypoplasia on left side along with hypoplastic nipple, absence of axillary hair and axillary fold ipsilaterally (Figure 1).

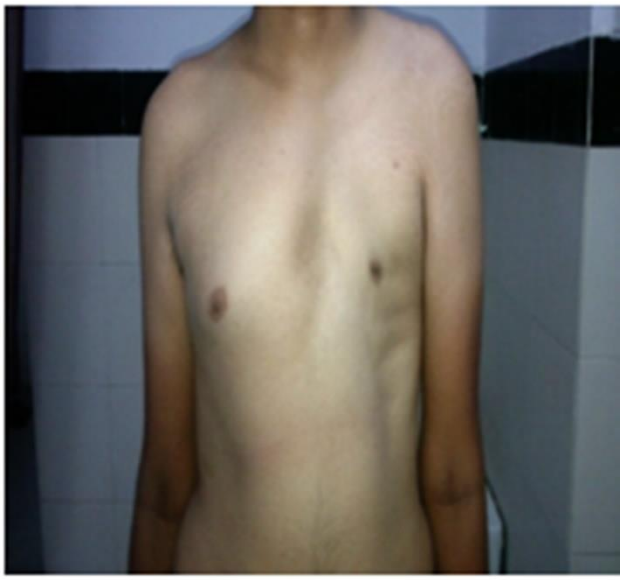

Figure 1: A 13 year old boy with Poland syndrome having asymmetrical chest wall, hypoplastic nipple and loss axillary hairs on left side.

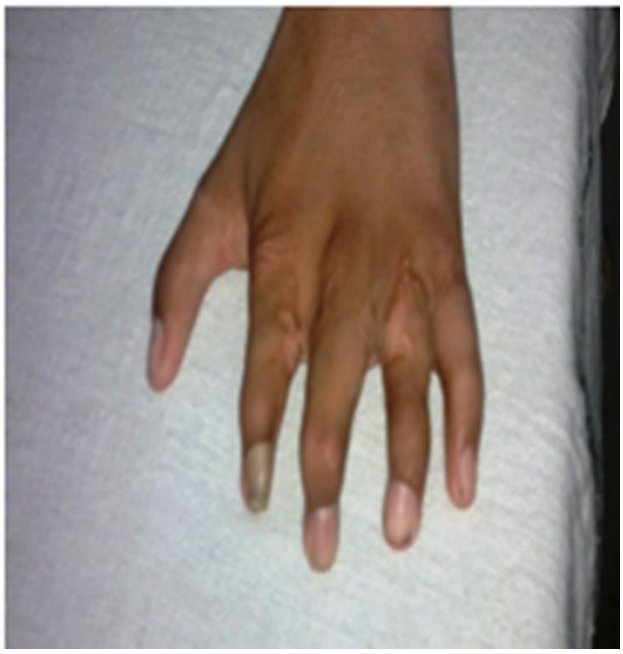

Figure 2: Short fingers of ipsilateral hand (operated), synbrachydactyly.

The limbs were normal and symmetric, but ipsilateral fingers were found to be short and separated. Past history of webbing of fingers was present. Seven years back, he was operated to release fingers (Figure 2).

Breath sounds were normal, but heart sounds were appreciated better on the right side. Chest x ray revealed a hyperluscent left lung with dextrocardia. CT scan of the chest revealed absence of the pectoralis major muscle on left side with hypoplasia of ipsilateral upper ribs with dextrocardia without any lung parenchymal abnormality (Figure 3).

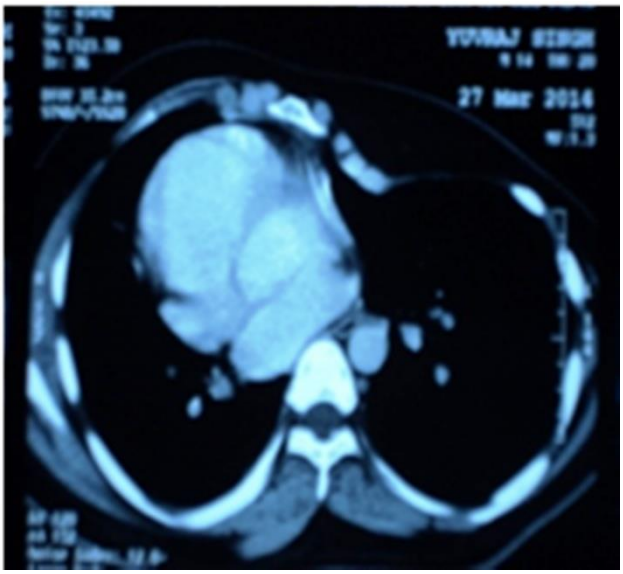

Figure 3: Computed tomographic section showing absence of pectoral muscle on left side with dextrocardia and asymmetrical chest wall.

No spinal deformity was found. Lymph nodes were not palpable. Blood picture did not reveal any abnormal cell. No musculoskeletal deformity was reported in his family. Based on these physical andradiological findings, a diagnosis of Poland's syndrome was made. The family was counselled, and the patient followed up on an outpatient basis.

DISCUSSION

Poland syndrome consists of a distinctive combination of congenital malformation. It includes various malformations of chest wall muscles, subcutaneous tissues, bones and upper extremity, usually on one-side of the body.[8] Many a times if expresses minimally with only absence of sternal head of pectoralis major muscle. Sometimes other adjacent muscles, like pectoralis minor, latissimus dorsi, serratus, external oblique, infraspinatus and supraspinatus muscles may also be involved. The involved is covered by hypoplastic skin with thinned subcutaneous layers and absent axillary hair on the affected side.[6] The anterior segments of the ribs and/or costal cartilages may be absent in some cases. The

Sprengel deformity with small and winged scapula may be present. One more specific anomaly names as brachysynphalangy with shortened arm, forearm and fingers may be present. Short and webbed fingers (cutaneous syndactyly) can be seen in few cases on the affected side. Moebius syndrome which consists of congenital bilateral facial paralysis with inability to abduct the eyesore Klippel-Feil syndrome may also be associated with it. Additionally, nipple is often smaller and higher on ipsilateral side in male while the breast is usually hypoplastic in females. In severe cases, anterior lung herniation may also be present.[9] Other body organs like spine and kidney malformations are occasionally reported in these cases. Most Poland syndrome cases have been reported sporadically with a trifling risk of recurrence in the same family. Presence of very few familial cases suggest an autosomal dominant inheritance with incomplete penetration. Geneticists currently hold the view that Poland syndrome is rarely inherited and generally is a sporadic event. There are rare instances where more than one individual has been identified with Poland syndrome either in the immediate or extended family.[8,10-14] Although several theories have been advanced to explain the etiology of Poland syndrome, most evidence indicates that it results from a vascular event during the critical sixth week of gestation with hypoplasia of the subclavian artery causing musculoskeletal malformations. Subclavian artery supply disruption sequence (SASDS) occurs when the medial and forward growth of ribs forces the artery into a U-shaped configuration.[7,10] To date the association of dextrocardia and Poland syndrome has been described only in 19 cases.[15] In all these cases, as in this, the syndrome was left sided and associated with rib defects, which occur only in about 15% of patients with pectoral defects on the right.[16,17]

Dextrocardia associated with Poland syndrome is always an isolated dextroposition: the normally connected heart is simply displaced to the right.[15-17] Leukemia and non-Hodgkin lymphoma have been described with Poland syndrome. CT scan and MRI can identify chest wall abnormalities and muscles involvement. Reconstructive surgery is helpful for correcting the chest deformity and breast asymmetry.[5,18]

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