A CASE OF POLAND SYNDROME IN A YOUNG FEMALE CHILD
WITHOUT ADDITIONAL MALFORMATION

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ABSTRACT
Poland syndrome is a rare congenital disorder characterized by absence of unilateral chest wall muscles. It typically presents with unilateral absence of sternal portion of the pectoralis major muscle with or without absence of pectoralis minor muscle, which may or may not be associated adjacent musculoskeletal abnormalities (Gashegu et al., 2009). Most of the Poland syndromes are sporadic incidence. We report a case of 5 year-old young female patient with absent pectoralis major and minor muscle on right side. To the best of our knowledge, this is one of the few documented cases of a patient with Poland Syndrome reported without any additional malformation reported from South India.

Keywords: Poland Syndrome

INTRODUCTION
The Poland anomaly was first described in 1841 by Sir Alfred Poland. In 1962, a New Zealand born British plastic surgeon, Patrick Clarkson, coined the term Poland syndactyly. Baudinne et al., (1967) reported a case of Poland's Syndrome, a term that is more accurate because the group of anomalies includes more than just syndactyly. Its incidence is difficult to determine, but current estimates range between 1:7,000 and 1: 100,000 births, with higher frequency among males (ratio, 2:1 – 3:1). In 75% of the cases, it involves the right hemithorax in the unilateral form. Other syndromes associated with Poland syndrome are Mobius syndrome and Klippel-Feil syndrome.

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<th>Classical Features</th>
<th>Additional Features</th>
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<td>Absence of sternal head of the pectoralis major muscle</td>
<td>Hypoplasia or aplasia of serratus, external oblique, pectoralis minor, latissimus dorsi, infraspinatus, and supraspinatus muscles</td>
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<td>Hypoplasia and/or aplasia of breast or nipple.</td>
<td>Total absence of anterolateral ribs and herniation of lung</td>
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<td>Decreased subcutaneous fat and axillary hair</td>
<td>Symphalangism with syndactyly and hypoplasia or aplasia of the middle phalanges</td>
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<td>Abnormalities of rib cage</td>
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<td>Upper extremity anomalies; short upper arm, forearm, or fingers</td>
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CASES
5 year old female child came with the complaint of unilateral chest wall asymmetry since birth. On examination of the child- CVS, RS and CNS were normal. Local examination of the chest shows depressed right chest wall with prominent ribs and supero-laterally displaced nipple with small size breast on right side. Scapula was normal bilaterally. Possibility of Poland syndrome was raised, patient was subject to ultrasound and CT. Ultrasound of the chest showed aplasia of the right pectoralis major muscle when compared to left side. Ultrasound of the abdomen shows no significant abnormality. CT scout film shows mild diffuse increased transclucency on the right side of the chest. Rest of the chest was normal. Computed tomography of the chest shows complete absence of right pectoralis major and minor muscles with normal subcutaneous fat in the right anterior chest wall.
Case Report

Figure 1: Clinical Photograph Showing Depressed Right Chest with Displaced Small Nipple

Figure 2: CT Scout Film of Young Female Patient Shows Mild Increased Right Hemi-Thorax Translucency

Figure 3: CT Thorax Axial Section Mediastinal Window Shows Complete Absence of Right Pectoralis Major and Minor Muscles
Figure 4: CT Thorax Sagittal Sections Mediastinal Window Shows Complete Absence of Right Pectoralis Major and Minor Muscles with Normal Left Pectoralis Major and Minor Muscles for Comparison

DISCUSSION
Poland 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 loss of tissue of the region. This condition is also known as subclavian artery supply disruption sequence (SASDS), fissure-thoracis-lateralis, pectoral-aplasia-dysdactyly-syndrome and unilateral chest-hand deformity (Yadav et al., 2014).

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) (Yadav et al., 2014) as well as with Morning glory syndrome and Pierre-Robin syndrome (Pisteljić et al., 1986).

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 in the immediate family. Therefore, some authors believe that an inherited abnormal vasculature formation may be the central underlying mechanism for this condition (Gashegu et al., 2009). Several twin patient cases have been reported, which might be helpful in clarifying the pathogenesis. In a couple of monozygotic twin girls who exhibited pectoral muscle hypoplasia, both the patients showed a de novo deletion of chromosome 11q12.3. Then, they suggested that Poland syndrome might be due to genetic control. On the other hand, Poland syndrome in one identical twin was reported by who demonstrated only one affected monozygotic twin, meaning that Poland syndrome is not determined by gene transmission (Yoo et al., 2015).

Associations have been found with leukemia, lymphoma, carcinoma of hypoplastic breast, ASD, neuroblastoma, carcinoma lung, wilms tumour which requires for oncological screening for the patients (Kaç et al., 2001).

Few renal anomalies has also been reported along with Poland syndrome including undescended testis, renal hypoplasia, renal agenesis, duplication of the renal collecting system and megacalycosis which is termed as acro-pectoral-rena field anomaly, which is sometimes associated with renal hypertension (Gude et al., 2001).

Poland syndrome had also been described with amastia-athelia with dextrocardia without abdominal situs inversion (Baltayiannis et al., 2011) and also often associated with a lung hernia [8] Ipsilateral limb
involvement is one of the most frequent signs of Poland syndrome. However, there are cases reported without any limb anamolies- similar to our study (Al Faleh et al., 2014).

One of the association of Poland syndrome is winging of scapula which is rare but potentially debilitating condition that impairs a person's ability to perform his or her activities of daily living. Postural deformities can also negatively impact one's quality of life during childhood and adulthood (Uludag et al., 2011) is also associated with Poland syndrome.

Association had also been described with facial asymmetry. Other anomalies in Poland syndrome including vertebral and lower limb malformations have also been described in rare cases. Furthermore, reports of Poland syndrome associated with other known syndromes in the same individual are reported. In fact, associations with Moebius syndrome, facio-auriculo-vertebral dysplasia and frontonasal dysplasia have been described (Ibrahim et al., 2013).

Conclusion

Our case represent a Poland syndrome without any associated malformation, we are reporting the case in female child -while Poland syndrome is most common in males. Right side presentation is concordance with previous studies. Further there are no associated anomalies in our case. Poland syndrome is a spectrum of anomalies not only we should think of reconstructive surgery, we should rule out other associated ocular-renal-musculoskeletal anomalies, we should look for occurrence in the family and ask the patient for follow up for oncological screening.

REFERENCES


