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Case Report

Poland syndrome: a case report

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ABSTRACT

Poland syndrome is characterized by the absence or hypoplasia of other chest muscles: small pectoral muscle, anterior tightened muscle, grandorsal muscle, and deltoid muscle. Anomalies of the thoracic cage: agenesis or hypoplasia of one or more costal segments, pectus carinatum, excavatum, clavicular hypoplasia, pulmonary herniation. Abnormalities of the mammary region: agenesis or hypoplasia of the breast, areola and nipple. The reported incidence of Poland's syndrome ranges from 1 in 7,000 to 1 in 100,000. Males are affected more frequently by a 2:1 to 3:1 ratio Poland's syndrome has also been diagnosed in 1 of 19,000 mammograms. The right side of the body was found to be involved in 60% to 75% of patients - associated malformations: genitourinary malformations, cardiac malformation, hepatic and biliary tract malformations. Case report Our case involves a 7-month-old boy with no medical history of cardiac or respiratory complaints. This is a male child with around 65cm height, and 7kg weight. There is no family history of consanguineous marriage and no family history of congenital anomalies. All siblings are alive and healthy. The child's milestones are within normal limits. There is aplasia of the right sternocostal head and clavicular heads and abdominal head of the pectoralis major muscle, there is a hypoplasia of the right nipple and breast and absence of subcutaneous fat on the same side. Ribs are visible on the right chest wall. Physical examination shows no history of hand and digit anomaly and no rib anomalies. Chest wall asymmetry with right chest wall showing depression on axillary floor. The anterior axillary fold is absent. On attempting abduction of the shoulder, it shows the absence of the sternocostal head of the pectoralis major.

Keywords: Poland syndrome, Cardiac, Nipple, Deltoid, Hepatic

INTRODUCTION

History-Alfred Poland, a student demonstrator in anatomy, described his case as similar to Poland syndrome in the year 1841; however, there were earlier reports of this anomaly by Lallemandin in 1826 and Froriep in 1839. The full anatomical spectrum of this syndrome was first summarised by Thompson in 1895. In 1900, Furst suggested a common aetiology. In 1962, Clarkson named it as Poland's syndactyly after more than 300 patients had been reported.

Epidemiology

The reported incidence of Poland's syndrome is 1 in 7,000 to 1 in 100,000. Males are more frequently affected by a ratio of 2:1 to 3:1. In 1 of every 19,000 mammograms Poland syndrome was diagnosed. The right side of the body was found to be involved in 60% to 75% of patients.

Vascular factors

Vascular disruption theory is the most widely used theory for Poland syndrome. This theory postulates that deficient blood flow in the proximal subclavian artery and its branches during the 6th week of gestation leads to regional tissue loss. Intrinsic factors, such as thrombi or emboli, and external mechanical factors, such as cervical ribs, aberrant muscles, and amniotic bands, can contribute to this disruption of blood flow to the subclavian artery and its branches. Maternal factors, such as smoking and

cocaine abuse, were also suggested as potential risk factors.[1]

Genetic

External factors may interfere with the migration of the pectoralis major muscle and digital separation in embryological development. Poland syndrome is usually sporadic signifying an autosomal dominant inheritance pattern with variable penetrance. Studies have identified de novo mutations and duplications in specific chromosomal regions leading to the development of Poland syndrome. Other coexisting syndromes associated with this are Moebius syndrome, Klippel–Feil syndrome and Pierre-Robin sequence.[2] Incidence of syndactyly is found to be from 2.5% to 13.5%, although hand deformities are not always present with Poland anomaly. Poland anomaly is more common in males, ranging from a ratio of 2:1to as high as 5:1 and more common on the right side than on the left side (3:2).

Analysis of the gender and sidedness indicates that males are more likely to have a right-sided anomaly, whereas females have an approximately equal distribution of left- and right-sided anomalies.[3] Agenesis or hypoplasia of the pectoralis major muscle is currently the cardinal feature to diagnose this syndrome. The pectoral muscle anomaly is generally easily observed by asking the patient to push the palms of the hands against each other with the arms positioned in front of the body.

In 1984, Slezak et al focused the eyes of hand surgeons that Poland syndrome may have a broad range of hand anomalies, and they classified these anomalies into four types: type 1, five digits present even if hypoplastic; type 2, functional border digits with the absence of central digits; type 3, more severe absence deformities with no functional digits; and type 4, radial ray defects with the absent thumb.[4]

Hand-pectoral deformity in Poland syndrome involves ipsilateral syndactyly or symbrachydactyly (89%), short middle phalanges (45%) to rarely ectrodactyly. Hypoplasia of the middle phalanges with cutaneous webbing is involved in syndactyly and is more often reported in the middle phalanges of the ulnar side of the hand than radial. Hypoplasia of the upper extremity involving the wrist and hand (89%), forearm (37%), arm (7%) and rarely phocomelia-like deficiency has been reported in Poland syndrome.[5] A 7-month-old girl with Poland syndrome also presented with ipsilateral renal agenesis as integral part of it. We recommend renal imaging studies be performed on all children with Poland syndrome.[6]

Recent mammogram studies have identified previously undiagnosed Poland anomaly. Although no long-term outcomes studies of patients with Poland anomaly exist, identifying all clinical manifestations of Poland anomaly is an important aspect of diagnosis, which may lead to better care through a better understanding of the condition. Furthermore, it is important to correctly identify this condition to avoid potential clinical mismanagement and possible litigation in cases of incidental trauma. Several authors have proved that chest radiographs are mandatory for all cases of Poland syndrome, complete blood count, urinalysis, and renal ultrasound in addition to the complete physical exam at the time of presentation, given reports of other systemic malformations coexisting with Poland syndrome.[7]

Treatment of Poland syndrome is planned by the severity of the defect and the resulting anatomical dysfunction. If a patient presents with minimal abnormalities or no functional deficit, then observation is needed and surgical intervention is not required. If surgery is indicated, it is usually performed within the first 18 to 24 months of life, to prevent progression of the deformity with growth of the skeleton and soft tissue.[8] Reconstructive surgery was performed under general anaesthesia with a skin-free latissimus dorsi flap (LD flap) and placement of a 215-ccm silicone implant. The incision is planned preoperatively for optimal positioning of the flap.[9] Several articles have reported on different kinds of neoplasms in Poland's syndrome patients and their association with an increased incidence of malignancy, such as leiomyosarcoma, neuroblastoma, leukaemia, and Wilms' tumour.

Summarizing 400 described cases of the syndrome, an increased risk of malignancy of the breast in this group.[10]

CASE REPORT

Our case involves a 7-month-old boy with no medical history of cardiac or respiratory complaints. This is a male child with around 65 cm height, and 7 kg weight. There is no family history of consanguineous marriage and no family history of congenital anomalies.

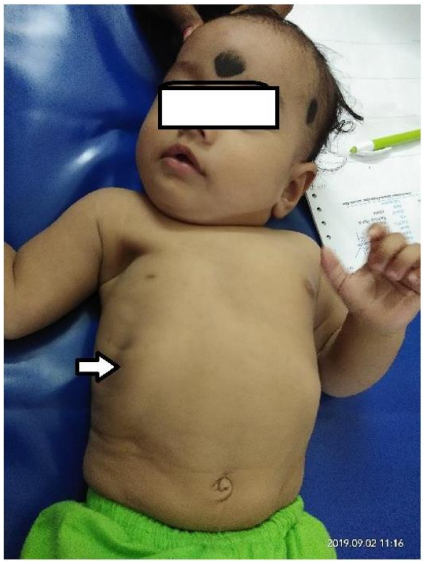

Figure 1: A child of Poland syndrome.

All siblings are alive and healthy. The child's milestones are within normal limits. There is aplasia of the right sternocostal head and clavicular heads and abdominal head of the pectoralis major muscle, there is a hypoplasia of the right nipple and breast and absence of subcutaneous fat on the same side. Ribs are visible on the right chest wall. Physical examination shows no history of hand and digit anomaly and no rib anomalies. Chest wall asymmetry with right chest wall showing depression on axillary floor. The anterior axillary fold is absent. On attempting abduction of the shoulder, it shows the absence of the sternocostal head of the pectoralis major.

DISCUSSION

Matsuura et al mentioned that there is a unilateral absence or hypoplasia of the pectoralis major muscle, typically affecting the sternal and costal portions.[2] Poland syndrome is characterized by varying severities of muscle involvement, accompanied by breast hypoplasia and chest wall deformities.

According to Romanini et al, pectoralis minor is mostly absent as compared to the normal presence of pectoralis minor muscle. In this case both the pectoral muscles are found to be absent.[10] They also mentioned that there are usually four varieties of Poland syndrome depending upon the presence or absence of a pectoral group of muscles like pectoralis major or minor muscles and congenital abnormalities of the upper limb and rib cage. In our study, this is type one variety.

According to Slezak et al, sporadic cases are more in number as compared to familial cases and these familial cases are very difficult to find.[4]

Baldelli et al stated that since a gene(s) responsible for PS has not yet been identified, the diagnosis of PS is clinical and based on the recognition of the characteristic recurrent pattern of features along with an appropriate differential diagnosis. Agenesis or hypoplasia of the pectoralis major muscle is currently the cardinal feature mandatory for the diagnosis. The pectoral muscle anomaly is generally easily observed by asking the patient to push the palms of the hands against each other with the arms positioned in front of the body.[11]

Garg et al established that Poland syndrome usually appears in a sporadic and unilateral form, even though there are bilateral cases also. Anomalies strictly confined to the right side three times more frequently than the left and it is more common in boys than in girls.[12]

CONCLUSION

From the aesthetic purpose of view asymmetry in the chest wall can be corrected by surgical reconstruction if the underlying anatomical cause can be found.

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