A Variant of Poland's Syndrome - A Case Report

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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. The condition typically presents with unilateral absence of the sternal portion of pectoralis major muscle which may or may not be associated with the absence of nearby musculoskeletal structures. We report a 12 year old girl with variant features of Poland syndrome.

Key Words: *Poland's anomaly, Poland sequence, Poland's syndrome*

Introduction

Poland syndrome has been estimated to occur in 1 to 3 per 100,000 newborns.[1] For unknown reasons, this disorder occurs more than twice as often in males than in females. Poland syndrome consists of unilateral absence or hypoplasia of the pectoralis muscle, most frequently involving the sternocostal portion of the pectoralis major muscle, and a variable degree of ipsilateral hand and digit anomalies, including symbrachydactyly, Sometimes called Poland sequence.[2] It was first described by Poland (1841). Poland syndrome is most commonly a sporadic condition, but familial cases have been reported.[2] The cause of Poland syndrome is unknown. Researchers have suggested that it may result from a disruption of blood flow during development before birth.[3] This disruption is thought to occur at about the sixth week (46th day) of embryonic developmentand affect blood vessels that will become the subclavian and vertebral arteries. These arteries normally supply blood to embryonic tissues that give rise to the chest wall and hand. Variations in the site and extent of the disruption may explain the range of signs and symptoms that occur in Poland syndrome. Rarely, this condition is passed through generations in families. In these families, the condition appears to be inherited in an autosomal dominant pattern. Mild cases of Poland syndrome without hand involvement may not be evident until puberty, when the differences (asymmetry) between the two sides of the chest become more apparent. By contrast, severely affected individuals have abnormalities of the chest, hand, or both that are apparent at birth. In rare cases, severely affected individuals have abnormalities of internal organs such as a lung, kidney, and the heart.[4-6] Poland syndrome may be underdiagnosed because mild cases without hand involvement may never come to medical attention.

Case report

A 12 year old female child was brought in outpatient clinic in pediatric department with complains of fever and cough.On clinical examination, it was found that the patient had hollowness in the left infraclavicular area. Child has hypoplasia of left breast and nipple with normal breast on right side along with accessory nipple (Fig.1). The anterior fold of left axilla was absent indicating the absence of the sternocostal portion of pectoralis major muscle. There was winging of left scapula i.e, sprengel deformity (Fig.2). The clavicle, acromion and contours of the shoulder were comparable on both sides. There was no evidence of polydactyly or syndactyly and no peripheral neurovascular deficit. The patient had no significant difficulty in performing his activities of daily living except for some difficulty in lifting heavy weights with left hand. The standard roentgenograms of chest-PA view revealed sprengel deformity of left scapula, fusion of ribs on both sides, hemivertebrae with fusion of D2–D6 vertebrae and scoliosis (Fig.3). The ultrasound of the abdomen and 2 D Echo was found to be normal.

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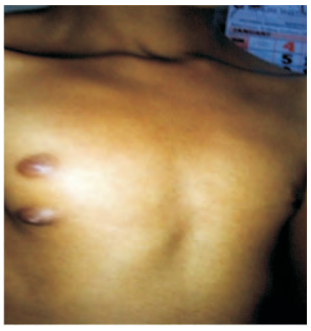

Figure 1: Hypoplasia of left breast with accessory nipple on right side

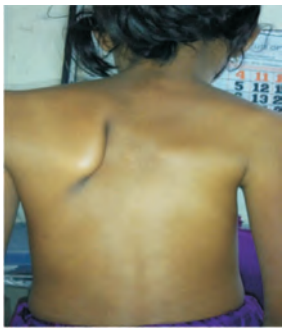

Figure 2: Sprengel deformity of left scapula

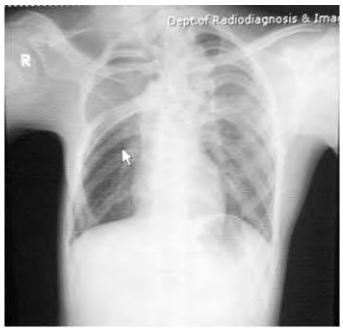

Figure 3: - The chest x-ray PA view shows fusion of ribs on both sides, hemi vertebrae with fusion of D2 –D6 vertebrae and scoliosis

Table 1: The principle abnormalities in Poland's syndrome

Category	Subcategory	Features
Inheritance	-	Autosomal dominant
Cardiovascular	Heart	Dextrocardia (in left-sided Poland sequence)
Chest	External Features	*Unilateral hypoplasia or absence of pectoralis major muscle Absence of pectoralis minor muscle
	Ribs, Sternum, Clavicles and Scapulae	*Sprengel anomaly *Hypoplastic ribs *Fused ribs
	Breasts	*Unilateral hypoplasia or absence of nipple Unilateral hypoplasia or absence of areola Unilateral absence of breast
Skeletal	Spine	*Hemivertebrae/Fusion of vertebrae
Hands	Unilateral syndactyly	Unilateral brachydactyly Unilateral oligodactyly
Muscle, Soft Tissue	-	Hypoplasia of latissimusdorsi muscle Hypoplasia of serratus anterior muscle Hypoplasia of infraspinatus muscle Hypoplasia of supraspinatus muscle Hypoplasia of deltoid muscle
Miscellaneous	-	All features are unilateral Occurs on right side in 75% of cases Three times more common in males Majority of cases are sporadic Pedigrees compatible with autosomal dominant inheritance have been reported Poland syndrome can be associated with Moebius syndrome Subclavian artery supply disruption in embryogenesis has been suggested as etiology

*findings present in our case

Discussion

The main features of Poland's syndrome are ipsilateral breast hypopolasia, deficiency of subcutaneous fat and axillary muscle. Absence of sternal head of pectoralis major and or serratius anterior and external oblique, winging of scapula i.e. sprengel deformity as described in table 1.[4-5] The right side is more often affected than the left, but in our case involvement was in the left side. The hereditary traits have been demonstrated for some anomalies of hands such as polydactyly. Other rare associations have been described including cardiac and urological abnormalities, acute leukemia, lymphoma, spherocytosis, mobius syndrome, inguinal hernia, knee flexion contracture, deformity of external ear, club foot and syndactyly of the toes.[6] The right side is more often affected than the left, but in our case involvement was in the left side. There were no features of syndactyly in our case. Syndactyly, believed to be constant feature in earlier reports may not be present.[4-6] Although patients with Poland's syndrome rarely have significant functional problems due to the muscle disorders, they may seek a surgical opinion for cosmetic reasons, particularly in the case of females with breast hypoplasia. These can be successfully corrected with an ipsilateral pedicled latissimus dorsi flap and submuscular augmentation.[7] Mild cases of Poland syndrome without hand involvement may not be evident until puberty. Sometimes due to social phobia especially in Indian setting, parents are hesitant to seek medical help. It our case also as child was female and parents belong to rural and low socio-economic setting, parents were quite hesitant for evaluation and further management. Due to following reasons, many cases are not reported or get late medical attention. CT scan accurately demonstrates the extent of the muscle abnormalities in Poland's syndrome and can be treated with planning reconstructive surgery.

References

1. McGillivray, B. C., Lowry, R. B. Poland syndrome in British Columbia: incidence and reproductive experience of affected persons. Am. J. Med. Genet 1977;1:65-74.
2. Czeizel, A., Vitez, M., Lenz, W. Birth prevalence of Poland sequence and proportion of its familial cases. Am. J. Med. Genet 1990;36:524.
3. Fraser, F. C., Ronen, G. M., O'Leary, E. Pectoralis major defect and Poland sequence in second cousins: extension of the Poland sequence spectrum. Am. J. Med. Genet 1989;33:468-470.
4. Mace JW, Kaplan JM, Schanberger JE and Gotlin RW. Poland's syndrome: report of seven cases and review of the literature. Clinical pediatrics 1972;11:98.
5. Ireland DCR, NoriaTakayama, Flatt AE. Poland's syndrome. A review of 43 cases. J. Bone and Joint Surg 1976;58:52-58.
6. Wilson MR, Louis DS, Stevenson TR. Poland's syndrome; variable expression and associated anomalies. J-Hand-Surg-Am 1998;13:880-82.
7. Hester TR. Jr. and Bostwick J. Poland's syndrome: Correction with latissimus muscle transposition. Plastic and Reconstructive Surgery 1982;69:226