Case Report

Poland sequence: Series of two cases and brief review of the literature

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Abstract:
Poland sequence is a rare congenital anomaly involving the chest wall and arm, displaying differing degrees of severity, functional and aesthetic impairments. Here we report a series of two cases that presented to us with this anomaly. These cases illustrate, for physicians, the importance of physical diagnosis and reinforce the practice of looking for additional anomalies when one is discovered.

Key words:
Aesthetic impairments, congenital anomalies, Poland sequence, Poland syndrome

Poland sequence, also known as Poland syndrome or Poland anomaly consists of congenital absence of the pectoralis major muscle; classically this sequence includes ipsilateral hand anomalies and it may also be associated with ipsilateral breast and nipple hypoplasia, and/or aplasia, deficiency of subcutaneous fat and axillary hair, and hypoplasia of the rib cage. It is a rare anomaly, and its incidence ranges from 1:20000 to 1:50000 as reported by different authors. Here we report a case series of this rare disorder.

Case Reports

Case 1
A 10-year-old male boy was admitted in our emergency ward with clinical and biochemical features suggestive of hepatitis probably caused by drugs (anti-tuberculosis drugs), started by a private practitioner on the basis of abnormal chest X-ray which showed increased radiodensity of left hemi-thorax [Figure 1]. On examination his chest was flat on the right side as compared to the left [Figure 2], and axillary hairs were absent. The radiodensity on the left side [Figure 1] was due to hyperlucent right lung, and absent pectoralis major on this side as revealed by computed tomography of thorax. It is a rare anomaly, and its incidence ranges from 1:20000 to 1:50000 as reported by different authors.[1] Here we report a case series of this rare disorder.

Case 2
A 16-year-old male came to our department with an asymmetrical chest configuration. He had flat left side as compared to right. There was no chest pain or any limitation of movement. There was no history of previous surgeries or any family history of congenital anomalies. On examination his left anterior chest wall was flat with absence of anterior axillary fold [Figure 3]. There was no visual anomaly on the left upper extremity and no significant difference in length and circumference between the two extremities. Pain and touch sensations were normal with normoactive biceps and triceps reflex. Chest skiagram revealed normal bones of chest wall and upper extremities but hyperlucency on the left side. Computed tomography of thorax revealed absence of muscular shadow on the left anterior chest wall because of absence of pectoralis major muscle [Figure 4]. Ultrasonography of the abdomen turned out to be normal. Again, parents were explained the nature of the abnormality, and advised regular follow-up of their son.

Discussion

It was in 1841 when the Poland syndrome was first described in a cadaver examined by Alfred Poland at Guy’s Hospital.[2] In his original description, titled Deficiency of the pectoral muscles he specifically noted absence of the sternocostal portion of the pectoralis major muscle with an intact clavicular origin, absence of the pectoralis minor, and hypoplastic serratus and external oblique muscles. Poland did not outline the breast hypoplasia or hand deformities in his original description. The definition has evolved, and the sequence is now thought to include shoulder-girdle anomalies with or without upper extremity involvement.[3] A compulsory diagnostic criterion of this anomaly is the presence of aplasia or hypoplasia of the pectoralis major muscle and at least one combined abnormality. Among these, the most frequent are costal aplasias/hypoplasias, depressions of the chest wall, atelia or amastia, absence of axillary hair, hypoplasia of subcutaneous fat, radius hypoplasia and hand malformations.
The malformations affecting the hand vary substantially and are not related to the severity of the chest malformation.[4] Only 400 cases of classic Poland Sequence have been reported in world literature.[4] It usually appears in a sporadic and unilateral form, even though a bilateral case[4] has also been described. The right side of the body is affected three times more frequently than the left and it is more common in boys than in girls. Its counterpart in the lower limb has also been described.[4]

The cause of this disease is unknown. This abnormality is congenital and sets in during the embryonic life.[5] A likely etiology appears to be vascular abnormality that causes failure of development. It is believed that Subclavian Artery Supply Disruption Sequence may be the likely etiopathogenic event. It is characterized by an intrauterine damage to the blood supply coming from the subclavian artery, and besides the Poland sequence it also includes the Klippel-Feil anomaly, the Möbius syndrome, transverse limb defects and the Sprengel anomaly. Poland sequence is usually sporadic, with a negligible risk of reoccurrence in the same family. However in a few cases, an autosomal dominant transmission with incomplete penetrance has been described.[5] Poland syndrome is an uncommon condition but there are certain key features that help to identify it. Because the functional disability in Poland syndrome is mild, patients usually present later for evaluation and discussion on aesthetic options. The sequence may be an incidental finding or found on specific examination as for patients having syndactyly. It may be found after an abnormal X-ray showing increased unilateral transradiancy or an abnormal electrocardiogram (ECG) showing left ventricular hypertrophy. The sternocostal head of pectoralis major is absent. Other involved muscles may include latissimus dorsi, serratus anterior, intercostals, infraspinatus, supraspinatus and deltoid. Associated hand anomalies may vary from micromelia, brachydactyly, oligodactyly to syndactyly. The breast may be absent or hypoplastic and can be associated with hypoplasia of ribs and scoliosis. The diagnosis is important as this condition may be associated with several visceral anomalies which may include renal -, cardiac -, liver - and biliary duct anomalies; central nervous system anomalies like encephalocele/exencephaly, microcephaly and other neural tube defects have been described.[6] Associated platelet disorders, leukemias and lymphomas have also been described.[7] Thus, radiological examination should be carried out in order to accurately evaluate all existing abnormalities. In particular, chest X-rays and abdominal ultrasound scanning are always to be carried out. Chest computed tomography is very...
useful in order to define the chest deformities and to evaluate indication for surgery.[3] The syndrome can also be diagnosed prenatally using ultrasonography.[8]

In general there is little physical disability from simple absence of the pectoralis muscle. Cosmetic appearance is a problem with the chest wall deformity, especially for women. Surgery is indicated mainly for aesthetic purposes.[10] Other indications are functional anomalies, paroxysmal movements of the chest wall and progressive lung herniation owing to the defect.[10] Chest wall reconstruction may be done using the lattisimus dorsi muscle, custom-made silicone prosthesis and/or a breast prosthesis.[10] Family physicians are encouraged to be alert in physical findings for improved diagnosis, appropriateness of care and patient satisfaction.

References

1. Czeizel A, Vitez M, Lenz W. Birth prevalence of Poland sequence and proportion of its familial cases. Am J Med Genet 1990; 36:524.
2. Poland A. Deficiency of the pectoral muscle. Guys Hosp Rep 1841;6:191.
3. McGillivray BC, Lowry RB. Poland syndrome in British Columbia.
4. Gausewetz SH, Meals RA, Seteguchi Y. Severe Limb deficiency in Poland Syndrome. Clin Orthop Relat Res 1984;185:9-13.
5. Bouwes Bavinck JN, Weaver DD. Subclavian artery supply disruption sequence: Hypothesis of a vascular etiology for Poland, Klippel-Feil, and Moebius anomalies. Am J Med Genet 1986;23:903-18.
6. David TJ. Nature and etiology of the Poland anomaly. New Eng J Med 1972;287:487-9.
7. Verzijl HT, van der Zwaag B, Cruysberg JR, Padberg GW. Moebius syndrome redefined: A syndrome of rhombencephalic maldevelopment. Neurology 2003;61:327-33.
8. Jeung MY, Gangi A, Gasser B, Vasiliscu C, Massard G, Wihlm JM, et al. Imaging of chest wall disorders. Radiographics 1999;19:617-37.
9. Paladini D, D’Armiento MR, Martinelli P. Prenatal ultrasound diagnosis of Poland syndrome. Obstet Gynecol 2004;104:1156-9.
10. Fodor PB, Khoury F. Latissimus dorsi muscle flap in reconstruction of congenital absent breast and pectoralis muscle. Ann Plast Surg 1980;4:422.

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