



Poland syndrome: rare presentation in two cases

Hayrettin Gocmen, Yücel Akkas, Selim Doganay

Abstract

Poland syndrome was first described in 1840 by Alfred Poland while still a medical student and the other components of the syndrome were described at London Guy's Hospital following the dissection of a cadaver's hand, which had hypoplasia and syndactyly. The incidence of Poland syndrome has been reported to be 1 in 30,000 live births. In the present case report, two Poland syndrome patients with ipsilateral hypomastia and a reduction in the axillary/pectoral hairs diagnosed during adulthood are presented; one patient was affected on the left side and had widespread café au lait spots, and the other patient had respiratory dysfunction due to multiple rib anomalies..

Poland syndrome was first described in 1840 by Alfred Poland while still a medical student and the other components of the syndrome were described at London Guy's Hospital following the dissection of a cadaver's hand, which had hypoplasia and syndactyly.¹

The main component of this anomaly is absence of the pectoralis major muscle.² This may also be accompanied by the absence or hypoplasia of the pectoralis minor, serratus anterior, latissimus dorsi, and deltoid muscles, and hypoplasia of the breast and the absence of nipples in girls. Rarer associations include rib defects, scoliosis, dextrocardia, renal hypoplasia, leukaemia, and Mobius syndrome.³ Some studies have reported the female-to-male ratio to be 1:3,⁴ whereas other studies have suggested that both genders are affected equally.⁵

The incidence of Poland syndrome has been reported to be 1 in 30,000 live births;⁶ Poland syndrome affects the right side in 67%–75% of cases.^{4,7} In the present case report, two Poland syndrome patients with ipsilateral hypomastia and reduction in the axillary/pectoral hairs diagnosed during adulthood are presented; one patient was affected on the left side and had widespread café au lait spots, and the other patient had respiratory dysfunction due to multiple rib anomalies.

Case report

Case 1—A 25-year-old male admitted with a complaint of congenital chest deformity involving the left side. The patient complained of a burning sensation on the left side of his chest and lack of strength in the left arm on extension. The physical examination revealed absence of the pectoral muscles. The physical examination also revealed café au lait spots on his chest (Figure 1).

Reduction of hairs in the left axillary region and on the left side of the chest was noted compared to the contralateral side. There was no oligosyndactyly (OS). Strength was 5/5 in the right arm and 4/5 in the left arm. Pectoral muscles in the left hemithorax were not observed on the thoracic computed tomography (CT) scan (Figure 2). There were no concomitant rib anomalies.

Figure 1. Pectoral muscles are absent on the left side (short arrow) and café au lait spot are seen on his right chest (long arrow)

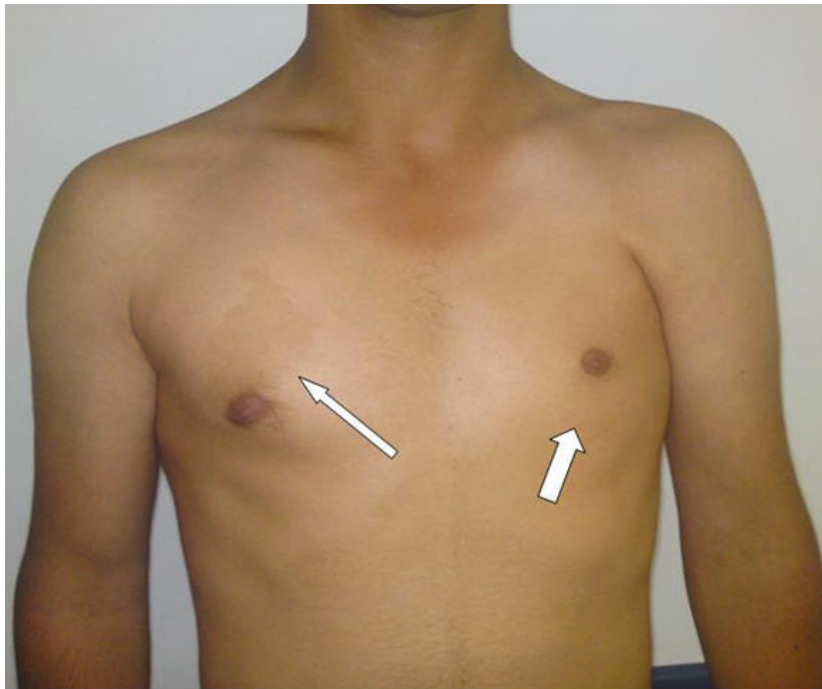
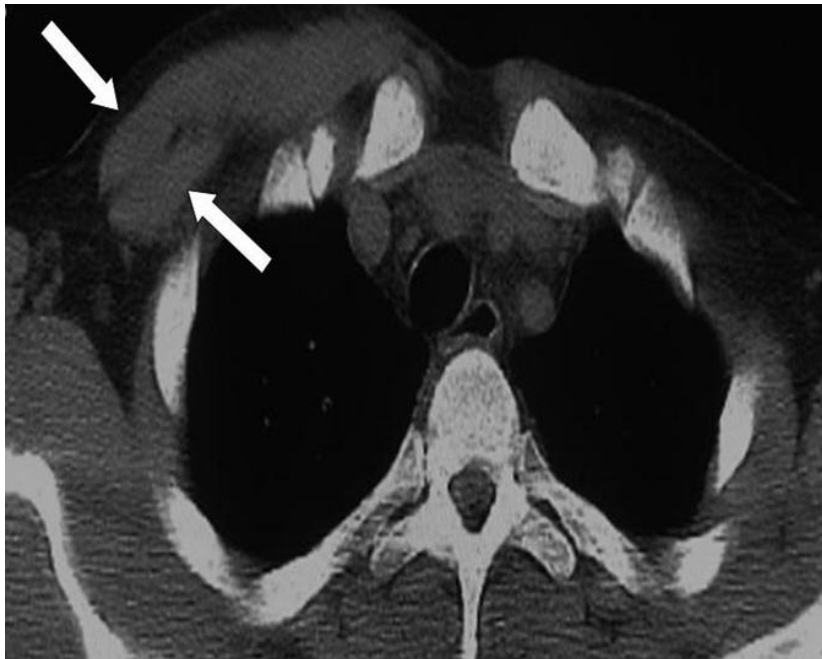


Figure 2. Axial plain CT scan shows pectoral muscles on the right chest (arrows) but pectoral muscles are absent on the left side



The abdominal ultrasonography and Doppler imaging of the neck were free of any pathologic findings. The results of the respiratory function tests were within normal limits (forced vital capacity [FVC]: 5.37 L 95%; forced expiratory volume1 [FEV1]: 4.82 L 102%; FEV1/FVC: 90%). The patient had two sisters and no disease-related abnormalities were reported in the family history

Case 2—A 21-year-old male admitted with complaints of a deformity of the right side of the chest and dyspnoea on exertion. The patient complained of lack of strength in the right arm and right shoulder. The physical examination revealed absence of the pectoral muscles on the right side and a marked volume loss within the right hemithorax (Figure 3). The patient had two sisters and no disease-related abnormalities were reported in the family history. There was hair reduction on the right side of the chest and in the axillary region.

Figure 3. The pectoral muscles on the right side are absent. There was hair reduction on the right side of the chest. The right breast was hypoplastic



The right breast was hypoplastic and there was no OS. The strength of the right and left arm was determined to be 3–4/5 and 5/5, respectively. Pectoral muscles of the right hemithorax were not observed on thoracic CT scan (Figure 4A). There were rib anomalies involving two levels and loss of volume within the right hemithorax (Figure 4B, 4C).

Figure 4A. Axial plain CT scan shows pectoral muscles on the left chest (arrow) but pectoral muscles are absent on the right side

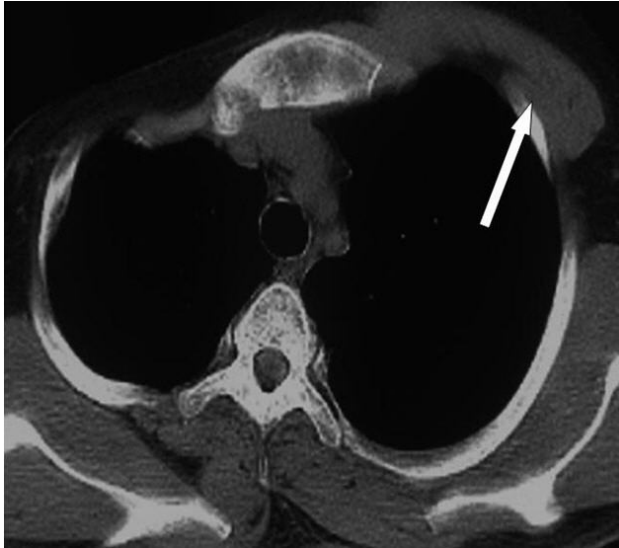


Figure 4B. Plain chest radiography shows rib anomalies (arrows) and loss of volume within the right hemithorax

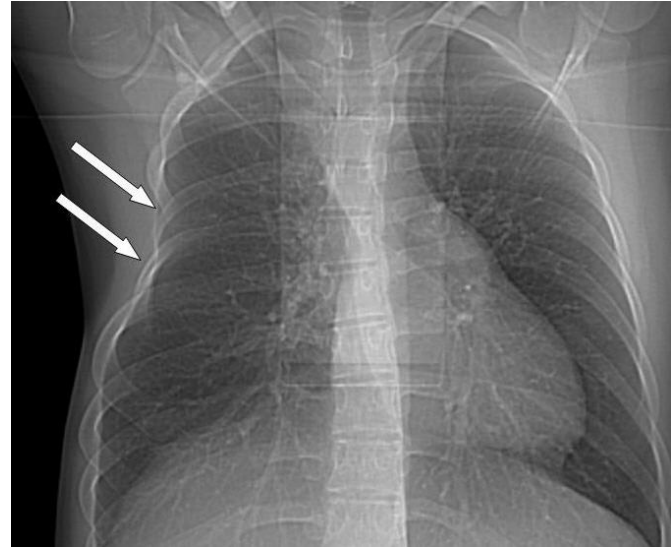
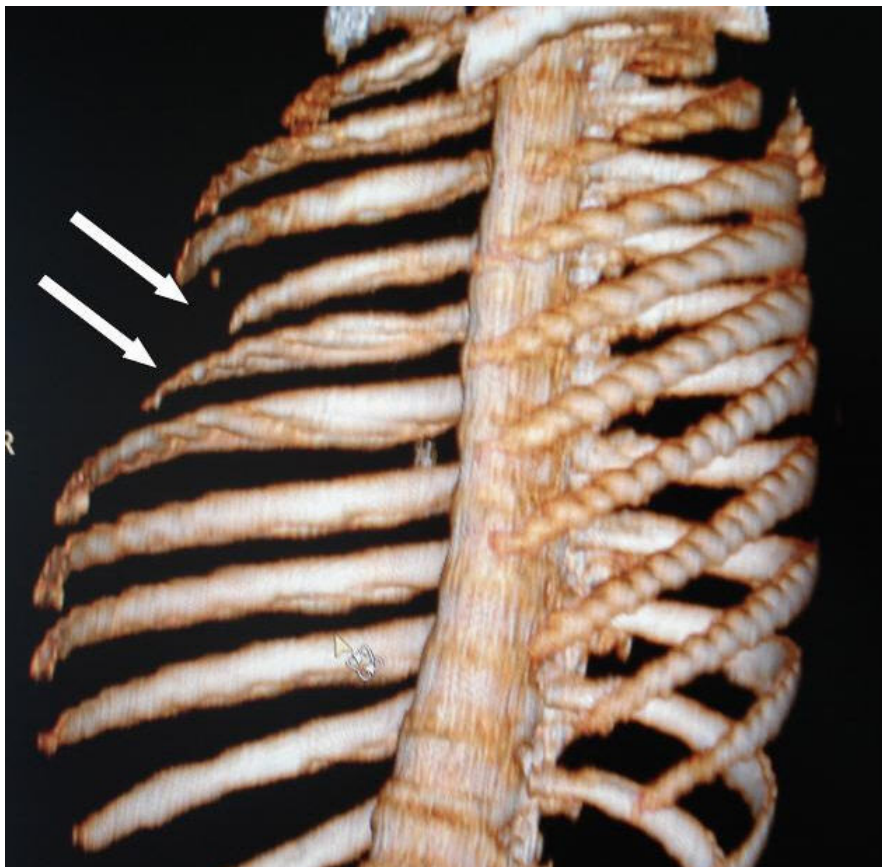


Figure 4C. On volume rendering three-dimensional computed tomography (3D-CT) there were rib anomalies (arrows)



The abdominal ultrasonography and Doppler imaging of the neck were free of any pathologic findings. The results of the respiratory function tests revealed mild restrictive respiratory insufficiency (FVC: 3.39 L 67%; FEV1: 2.88 L 63%; FEV1/FVC: 74%).

Discussion

Poland syndrome, as first described by Alfred Poland in a 26-year-old male, most often occurs sporadically.^{2,8} Some authors have suggested that Poland syndrome is a genetic disease which is inherited with an autosomal dominant pattern.⁹ In fact, Poland syndrome has even been reported in more than one member of a family and referred to as the Familial Poland syndrome⁹. In contrast, Stevens et al⁶ have reported the presence of Poland syndrome in one of two monozygotic twins; thus, there was no purely genetic transmission. Poland syndrome was not noted in the family members of our cases.

Although the disease pathology underlying Poland syndrome is not well understood, various hypotheses have been advanced. The most widely agreed-upon hypothesis is a decrease in blood flow during the intrauterine period due to development of malformations or spasm of the brachiocephalic arterial structures following mutation of the upper extremities while budding from the chest wall in the 6th–7th weeks of pregnancy.

Blood flow disruption in the subclavian artery is known to be a cause of upper extremity injury, while injuries of the pectoralis major muscle, breast, and the other chest wall structures has been reported to be due to an effect on the internal thoracic artery.^{4,10,11}

In addition to anomalies of the pectoralis major muscle, which is associated with this mechanism, syndactyly of the fingers, ipsilateral nipple anomalies (hypoplastic, aplastic, or inverted nipples), ipsilateral radius and ulnar anomalies (hypoplasia and aplasia), and anomalies of the ribs, are also rarely observed.

Ipsilateral breast hypoplasia and reduction in axillary and chest hairs was observed in both of our cases; however, there were no syndactyly or forearm anomalies. Nonetheless rib anomalies at two levels that led to ipsilateral volume loss in the right hemithorax, was observed in the second case.

A rare association between Poland syndrome with other organ system-related symptoms including microcephaly, cerebral atrophy, disorders in myelination, situs inversus or dextrocardia, hemivertebra, gastroschisis, paralysis of the cranial nerve or mental retardation, psychosocial retardation, hypospadias, and urinary system anomalies have also been reported⁶. No pathologic findings were observed on the abdominal ultrasonography and Doppler imaging of the neck in either of our cases. There was no dextrocardia and no psychiatric or neurologic complaints.

Absence of the pectoralis major muscle is usually unilateral and almost always observed on the right side. Our first case had involvement of the left side, which has been reported as a rare condition; whereas in our second case, the defect was on the right side.

In 1998, Karnak et al¹³ replaced this generally accepted opinion by publishing the first case of a 6-year-old girl who was described as having bilateral Poland syndrome anomalies due to absence of the pectoralis major muscles, symmetric chest wall deformities, and bilateral arm anomalies, together with hypoplasia of the breast tissues and nipples.

In spite of Poland syndrome generally causes aesthetic problems, surgical intervention has tried for only functional aims. In this context, many attempts at surgical intervention have been performed for problems, especially the forearm and fingers. Defects of the chest wall do not generally require treatment.

No cases of Poland syndrome associated with respiratory dysfunction verified with spirometry have been reported. Normal spirometric respiratory function was observed in a case with Poland syndrome reported by Deniz;¹⁴ however, mild decreases in the maximum inspiratory pressure (MIP) and maximum expiratory pressure (MEP) were detected. In our second case, there was volume loss in the ipsilateral hemithorax and restrictive respiratory insufficiency, accompanied by anomalies of two ribs.

Our cases admitted with complaints of loss of strength in the ipsilateral shoulder and arms. The physical examination revealed loss of strength of the affected side. Mysnysk et al¹⁵ have reported the loss of 20% and 29% of horizontal strength following measurement with a Cybex dynamometer in a study involving two professional wrestlers with Poland syndrome. Quantitative evaluation with electromyography (EMG) was also planned in our cases; however, the patients declined testing.

Endocrine anomalies, melanosis, and an increase in the incidence of benign and malignant tumors may also be observed in Poland syndrome. Although the most commonly encountered malignant tumors are lymphoreticular tumours, such as leukemia and lymphoma, childhood solid tumours, such as neuroblastoma and Wilms' tumours, may also be observed¹⁶.

A case of a 12-year-old girl who admitted with amenorrhea and absence of right breast enlargement has been reported¹⁷. No endocrinologic or oncologic problems were reported in our two cases.

The present report of two cases presented with left-sided involvement and rib anomalies with respiratory dysfunction, accompanied by café au lait spots is of clinical importance due to the rare occurrence of Poland Syndrome with an incidence of 1/30,000.

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References:

1. Upton J. Congenital anomalies of the hand forearm. In: McCarthy JG. Plastic Surgery. Philadelphia: WB Saunders Company, 1990:5301-5.

2. Aznar P, Urbano J, Laborda GE, et al. Breast and Pectoralis muscle hypoplasia (A mild degree of Poland's syndrome) *Acta Radiol* 1996;37:759-62.
3. Mace JW, Kaplan JM, Schanberger JE, et al. Poland's syndrome: report of seven cases and review of literature. *Clin Pediatr* 1972;11:98-102.
4. Fraser FC, Teebi AS, Walsh S, Pinky L. Poland sequence with dextrocardia: Which comes first? *Am J Med Genet* 1997;73:194-6.
5. Uster GD. Upper extremity. In: Mustarde' JC, Jackson IT. *Plastic Surgery in Infancy and Childhood*. Edinburgh: Churchill Livingstone, 1988:565-7.
6. Stevens DB, Fink BA, Prevel C. Poland's syndrome in one identical twin. *J Pediatr Orthop* 2000;20:392-5.
7. Smith DW. Poland sequence (Unilateral defect of pectoralis muscle and syndactyly of hand). Philadelphia. WB Saunders Company, 1982:224-7.
8. Shalev SA, Hall JG. Poland anomaly—report of an unusual family. *Am J Med Genet A* 2003;118:180-3.
9. Darian VB, Argenta LC, Pasyk KA. Familial Poland's syndrome. *Ann Plast Surg* 1989;23:531-7.
10. Beer GM, Kompatscher P, Hergan K. Poland's syndrome and vascular malformations. *Br J Plast Surg* 1996;49:482-4.
11. Donegan WL. Common Benign Conditions of the Breast In: Donegan WL, Spratt JS eds. 4th ed. *Cancer of the Breast* WB Saunders Company, 1995:88-9.
12. Jones KL. *Smith's Recognizable Patterns of Human Malformation*. 5th ed. Saunders, 1997:302.
13. Karnak I, Tanyel FC, Tuncbilek E, et al. Bilateral Poland anomaly *Am J Med Genet* 1998;75:505-7.
14. Deniz Ö, Tozkoparan E, Gümüş S, et al. Poland sendromu (olgu sunumu). *Tüberküloz ve Toraks Dergisi* 2005;53:275-9.
15. Mysnyk MC, Johnson DE. Congenital absence of the pectoralis muscles in two collegiate wrestling champions. *Clin Orthop* 1991;265:183-6.
16. Matsui A, Nakagawa M, Okuno M. Association of atrial septal defect with Poland-Moebius syndrome: vascular disruption can be a common etiologic factor. A case report. *Angiology* 1997;48:269-71.
17. Corona-Rivera JR, Corona-Rivera A, Totsuka-Sutto SE, Corona-Rivera E. Corroboration of the lower extremity counterpart of the Poland sequence. *Clin Genet* 1997;51:257-9.