Poland syndrome

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SUMMARY

Poland syndrome is a congenital anomaly with unilateral agenesis or hypoplasia of the pectoral muscles, deformity of the anterior chest wall and upper extremity anomalies. In patients with Poland syndrome, varying rates of breast asymmetry, from hypomastia to amastia, accompany. It is observed that the incidence of dextrocardia is increased in patients with left-sided Poland syndrome. Vital lung capacity may be reduced due to anterior chest wall deformity. This syndrome is thought to be due to a temporary impairment of circulation in the arteria subclavia or any of its branches or hypoplasia of these vessels during the development of the upper extremities in the intrauterine period. We aimed to emphasize the characteristics of Poland syndrome in a 26-year-old female case. On physical examination, pectus excavatum deformity was observed besides the absence of left pectoralis major and minor muscles with plica axillaris anterior. In terms of congenital or acquired pathologies that may accompany, the cases should be evaluated clinically in detail and followed up.

Key words: Poland syndrome – Pectus excavatum – Dextrocardia – Computed tomography – Poland sequence

INTRODUCTION

Poland syndrome is an anomaly characterized by hypoplasia or agenesis of the anterior chest wall muscles. It can be observed sporadically or congenitally. The most commonly affected muscle is the sternocostal part of the major pectoral muscle, followed by the minor pectoral muscle (Ergüven et al., 2011). In Poland syndrome, the agenesis of pectorals muscles may be accompanied by hypoplasia of the nipple, breast and subcutaneous adipose tissue, aplasia of costal cartilages or ribs (II-IV or III-V), alopecia of axilla and chest region, and ipsilateral upper extremity deformities (Ergüven et al., 2011; Fokin and Robicsek, 2002).

This syndrome was first described in 1841, when Sir Alfred Poland was a student at Guy's Hospital in London. He performed an autopsy on the cadaver of a 27-year-old prisoner named George Elt, and reported the case with left-sided complete absence of the major and minor pectoral muscles, as well as ipsilateral partial absence of the serratus anterior muscle and symbrachydactyly (Poland, 1841). In 1962, Patrick Clarkson, a hand surgeon, operated on a type of patient with similar features to those in Alfred Poland's case, and made the definition of "Poland syndactyly" in refrence to to that case (Clarkson, 1962). This syndrome affects men more often than women, and occurs usually on the right side (Yiğit et al.,

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2015). Poland's syndrome is mostly unilateral, but a few bilateral cases have been reported (Fokin and Robicsek, 2002; Yiğit et al., 2016).

Although the etiology is not known precisely, genetic factors, vascular causes, viral infections and teratogenic agents are thought to be effective (Yiğit et al., 2016). One of the generally accepted theories in its etiopathogenesis is insufficient blood flow in the intrauterine period (6-8 weeks). It is thought to be due to a temporary impairment of circulation in the arteria subclavia, or any of its branches or hypoplasia of these vessels during the developmentof the upper extremities. The degree of deterioration of arterial circulation determines the severity of this syndrome (Lasko et al., 2008). This syndrome may accompany Sprengel deformity, Klippel-Feil syndrome and Möbius syndrome (Van der Feen et al., 2006). In addition, case reports have been reported in the literature in which it is seen with various cardiac anomalies and organ malignancies (Ergüven et al., 2011). However, cases involving all the abnormalities associated with this syndrome are extremely rare.

CASE REPORT

A 26-year-old female patient was admitted to the clinic of chest diseases with complaint of shortness of breath. On physical examination, pectus excavatum deformity, agenesia of the left pectoral minor muscle, hypoplasia of the

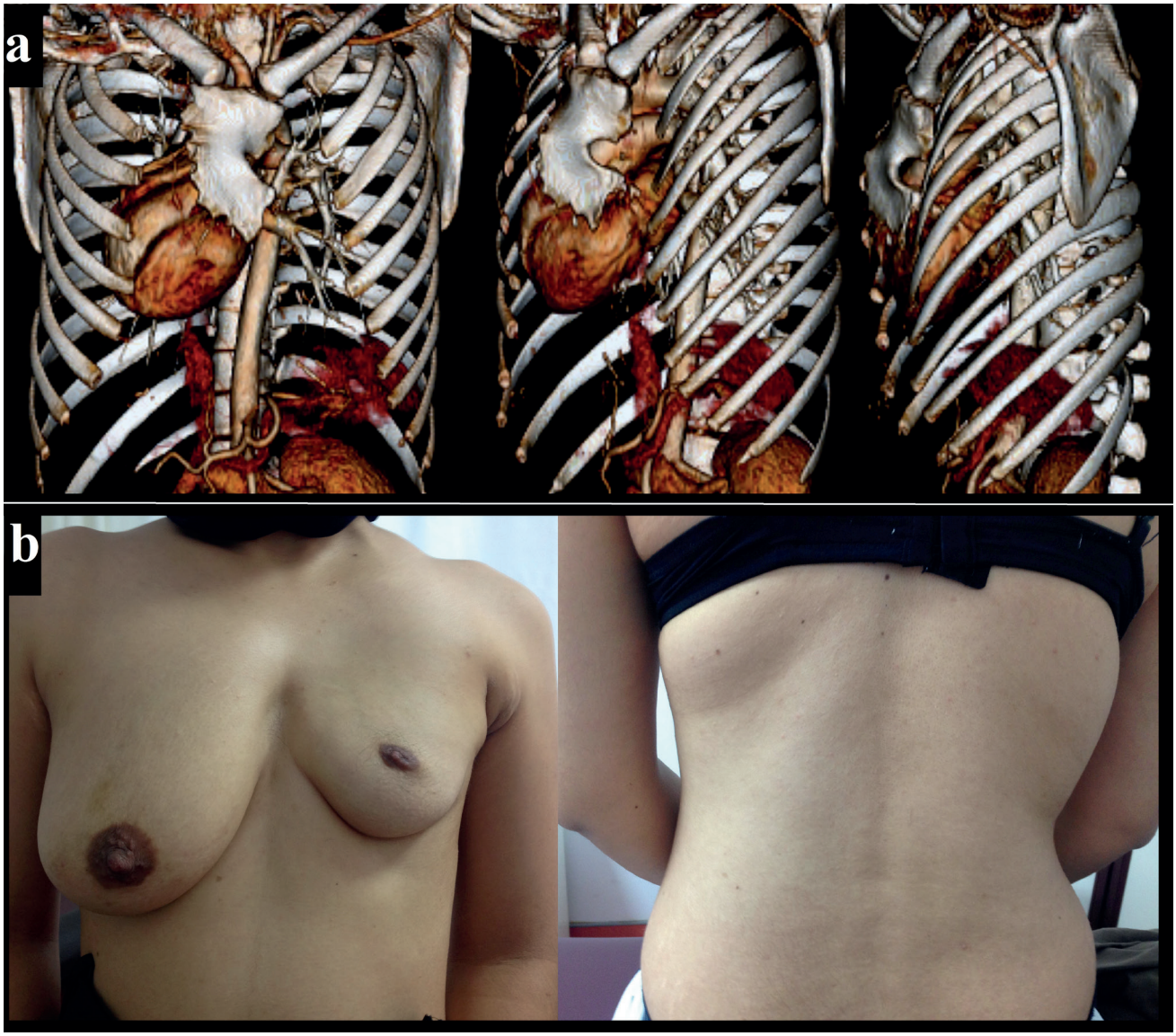

Fig. 1.- a) 3D-volume-rendered computed tomography images demonstrating pectus excavatum deformity and dextrocardia due to malformation of the sternum and costal cartilage on the left chest wall. **b)** Photos of the case show hypoplasia of the left breast and absence of the plica axillaris anterior, due to hypoplasia of pectoralis major muscle and hypoplasia of subcutaneous adipose tissue. The papillae mammaria and areola mammae are hypoplastic and superiorly sited.

pectoralis major muscle and breast asymmetry were observed (Fig. 1a). Furthermore, plica axillaris anterior was absent, but plica axillaris posterior was complete (Fig. 1b). In the left breast, glandular tissue, papillae mammaria, areola mammae, and subcutaneous adipose tissue were hypoplasic (Fig. 1b). Left axilla alopecia and axilla sweating disorder accompanied in the case. The patient's respiratory sounds were normal, and there were no additional pulmonary symptoms. There was no history of illness in his medical history. There were no any additional deformities in the left upper extremity, and the patient had full range of motion and muscle strength. The patient's liver and kidney function tests and complete hemogram tests were normal. Spirometric measurement results were borderline in terms of mild restrictive pathology [predicted VC max: 3.95 VC max: 3.08 L (%78), predicted FVC:3.90 FVC: 3.08 L (%79), predicted FEV1 %M: 84.35 FEV1 %M: 86.17 L].

Intravenous contrast-enhanced thorax CT was performed on 160-slice computed tomography (CT) device. Multiplanar reformed images (axial, sagittal and coronal planes) with a slice thickness of 3 mm were obtained from volumetric data (0.5 mm in the axial plane). CT images indicated that the pectoralis minor muscle is agenesic, and that the pectoralis major muscle is hypoplasic (Fig. 2). However, the costae, intercostales, serratus anterior, latissimus dorsi, external oblique and shoulder muscles were normal. CT confirmed that the left ribs were deformed and the volume in the left hemithorax was reduced. The patient had dextrocardia, because left hemithorax volume was reduced, but the heart structure was normal. The trachea was in the midline. The calibration and lumen of the main vascular

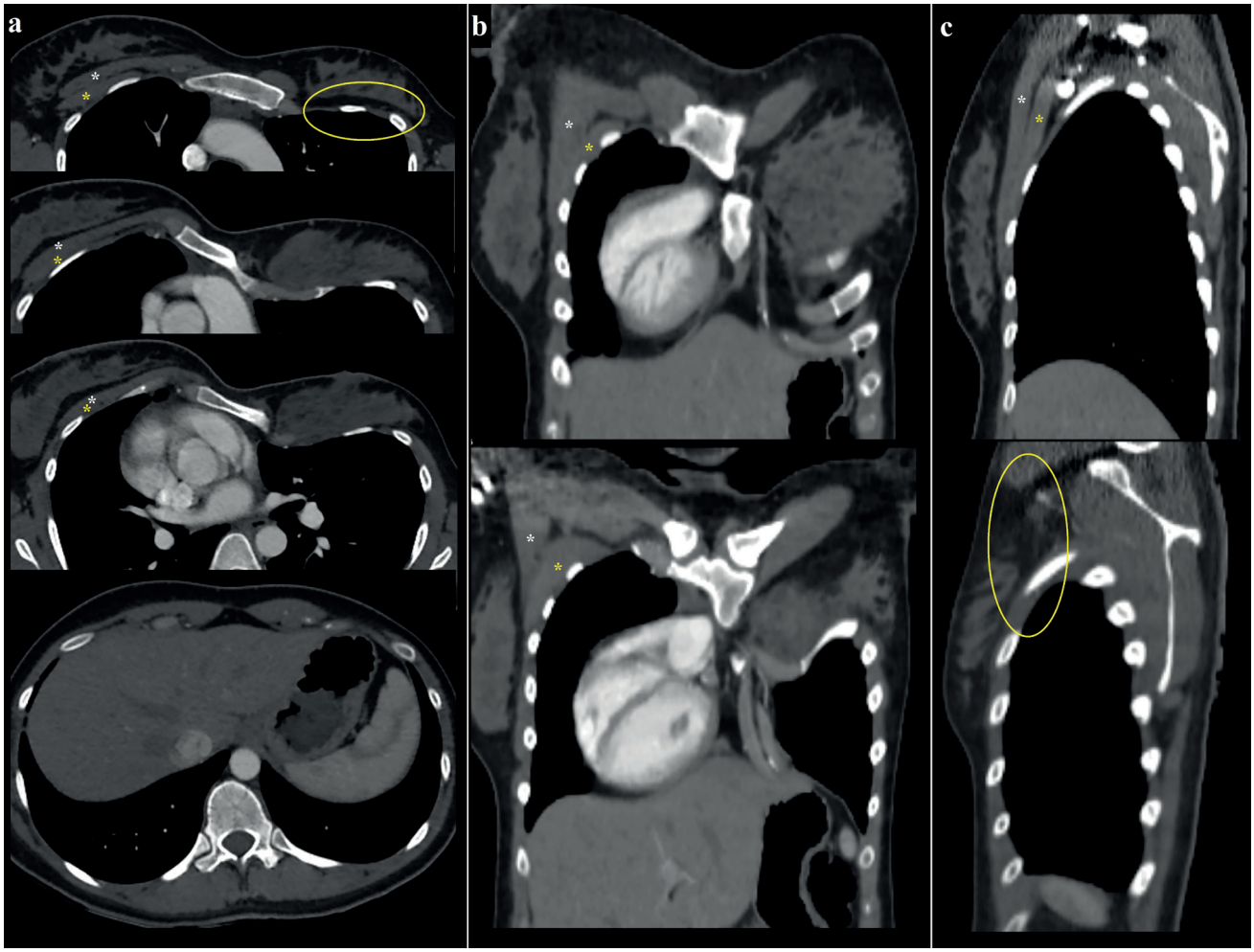

Fig. 2.- Chest computed tomography images obtained in axial (**a**), coronal (**b**) and sagittal (**c**) planes; It is noteworthy that the pectoral muscles are seen normally on the right side (white asterisk: pectoralis major; yellow asterisk: pectoralis minor) but the pectoralis major muscle is hypoplasic and the pectoralis minor muscle is agenesic on the left side. In addition, due to malformation in the sternum and left rib cartilages, pectus excavatum deformity and secondarily dextrocardia are observed.

structures in the mediastinum were normal. Both lung parenchymas were normal and the bronchovascular distribution was symmetrical and natural.

COMMENTS

Poland syndrome is a congenital or sporadic anomaly with agenesis or hypoplasia of major and minor pectoral muscles, hypoplasia of breast or nipple and of subcutaneous tissue, chest wall deformity, pectoral alopecia, and hand anomalies (Bıçakçı, 2010; Yiğit et al., 2015). The case had the agenesia of the left pectoral minor muscle and hypoplasia of the left pectoral major muscle, left breast, glandular tissue, papillae mammaria, areola mammae, and subcutaneous adipose tissue similar to the literature. Poland syndrome is a rare congenital anomaly. Its prevalence ranges from 1/7.000 to 1/100.000 live births. This syndrome is more common in men than in women, and the right side, more than the left side, is affected in males. However, left- and right-side is affected almost equal in females (Fokin and Robicsek, 2002). Our case was a woman with no family history of heredity, with left-sided involvement. Left axilla alopecia and axilla sweating disorder accompanied in the case. In Poland syndrome, ipsilateral syndactyly defects such as hypo / aplasia in the 3rd and 4th fingers of the upper extremity can be seen. Furthermore, serratus anterior, latissimus dorsi, and other shoulder muscles can be added to the agenesics of pectoral muscles (Torre et al., 2010). However, in our case, there was no other muscle affected except the pectoral muscles, and there was no deformity in the ipsilateral (left) upper extremity. Additionally, intercostales, serratus anterior, latissimus dorsi and shoulder muscles were complete.

In addition, it was noticeable that the heart and mediastinum were located in the right hemithorax in our case. Secondary dextrocardia cases associated with Poland syndrome have been reported in the literature, and this condition is called the 'Poland sequence' (Fraser et al., 1997). Torre et al. (2010) determined dextrocardia anomaly in 20 (14%) of 122 patients with Poland syndrome. This situation is explained as the heart displacement to the right side due to the narrowing of the left thoracic cavity during the intrauterine period. Due to the narrowing of the chest cavity, restrictive pulmonary symptoms can be observed and detected by spirometric measurements. In our case, spirometric measurements were borderline in terms of mildly restrictive pulmonary pathology (Deniz et al., 2005).

There are also publications reporting malignancy in patients with Poland syndrome in theliterature. In some cases, especially carcinomas in the hypoplasic breast, gastric carcinoma, lung tumors and intracranial solid tumors have been reported (Elli et al., 2009; Kurt et al., 2006; Ahn et al., 2000; Ji et al., 2008). However, there is no evidence that there is a genetic predisposition for cancer formation. For all that, cases of Poland syndrome should be followed up in terms of possible malignancy risk. We did not find any malignancy findings in our 26-year-old case.

CONCLUSION

Poland syndrome is a congenital or sporadic syndrome characterized by the absence of pectoral muscles. In terms of congenital or acquired pathologies that may accompany, the cases should be evaluated clinically in detail and followed up. Written informed consent was obtained from the patient for publication of this case report and accompanying images.

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