

POLAND'S SYNDROME IN WOMEN: CASE REPORT AND LITERATURE REVIEW

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Abstract: Introduction: Poland Syndrome is a rare congenital anomaly characterized mainly by hypoplasia or agenesis of the unilateral thoracic musculature and changes in the ipsilateral upper limb. **Goals:** This case refers to a female patient with Poland Syndrome, in an oncology hospital in the city of Manaus-AM-Brazil. **Methods:** The patient in the case reported presented breast asymmetry on physical examination and the study shows the correlation of imaging findings from digital mammography, radiography and computed tomography of the chest of this condition. **Conclusion:** The importance of recognizing this condition by the radiologist is to differentiate this benign anomaly from conditions such as unilateral hypertransparency on chest X-ray and breast asymmetry on mammography related to other causes.

Keywords: Poland's Syndrome, chest muscle hypoplasia, breast hypoplasia, mammography, computed tomography.

INTRODUCTION

First described in 1841 by surgeon Alfred Poland, Poland Syndrome is a congenital musculoskeletal malformation with an incidence of 1/32,000 of live births, being considered a rare condition. There is a predominance of males, and a higher frequency of cases involving the right side, which represents between 67% and 70% of the total. Most cases are sporadic, but it is believed that it may have an autosomal dominant hereditary character with variable penetration.

Some studies have suggested that a genetic factor or more likely extrinsic factors between the sixth and eighth weeks of pregnancy may interfere with the migration of the pectoralis major muscle and the separation of the fingers that occurs during this period. The recent vascular hypothesis for the etiology of the

syndrome implies that the interruption of the blood supply by hypoplasia of the ipsilateral subclavian artery or its branches is the origin of this anomaly.

The exact etiology is not known, but it is attributed to an anomaly in the development of the mesoderm, one of the three layers that make up the blastoderm, from which cardiac, skeletal and smooth muscles originate, causing damage to the development of the pectoral part and the distal part of one of the upper limbs, usually occurring on the right side of the body, with anomalies appearing in the proximal clavicular artery. Added to all these events, the result will be a partial or total decrease in the tissue of the pectoral region, which would explain the hypoplasia present in this region.

The main findings in this syndrome would be the partial or total absence of the pectoralis major, pectoralis minor, serratus anterior, latissimus dorsi, deltoid, infraspinatus and supraspinatus muscles, agenesis or hypoplasia of the breast and the nipple-areolar complex. Less commonly, cartilage and rib defects can be found, present in 15% of cases with involvement of the right side, hypoplasia of subcutaneous tissues of the chest wall, pulmonary herniation, skeletal hypoplasia affecting the hands, forearms, arms and scapula, syndactyly, polydactyly, ipsilateral brachysyndactyly and alopecia of the axillary and mammary region. Findings may be bilateral, non-simultaneous, more frequent on the right side.

In up to 15% of cases, Poland syndrome is associated with Moebius syndrome, Sprengel anomaly and, exceptionally, with perineal muscle atrophy (Charcot-Marie-Tooth disease), microcephaly, psoriasis, systemic lupus erythematosus, Basedow-Graves, Leukemia, Hodgkin's Lymphoma, Neurofibromatosis type I, Myasthenia gravis, Craniofacial dysplasias, Leiomyosarcoma,

Klippel-Feil Syndrome and dextrocardia.

As the muscles that make up the chest region are affected, it must be taken into account that respiratory changes may occur due to the abnormal functioning of these muscles, causing paradoxical movements in the chest wall, generating a decrease in lung capacity in up to 48% of cases.

Dextrocardia is one of the characteristic signs of Poland Syndrome, however, it is not present in all cases and it is believed that only 6% of people will develop this condition.

Unilateral hypoplasia of the breast and pectoralis major muscle without upper limb involvement are seen more often than other forms of the syndrome and are not uncommon conditions in women seeking breast augmentation due to breast asymmetry.

The pectoralis major muscle, also called pectoralis major, covers the upper portion of the thorax in a fan shape, being immediately inferior to the breast. Its lateral border forms the anterior axillary crease and most of the anterior wall of the axilla. The origin of the pectoralis major muscle is in the sternal half of the clavicle and anterior surface of the sternum, and its insertion is located at the crest of the greater tubercle of the humerus. The main function of this muscle is abduction and medial rotation of the humerus and anteroinferior movement of the scapula.

Imaging techniques play a fundamental role in the detection, localization and characterization of chest wall disorders, many of which have a characteristic radiological appearance that allows a definitive diagnosis. Sternal deformities can be seen with radiographic examinations and their severity quantified with computed tomography, which clearly shows many abnormalities. For chest wall disorders, chest radiography remains the primary imaging study, where it usually shows a hyperlucency on the affected side, which mimics a radical mastectomy. Mammography

is useful to show breast asymmetry or the absence of the pectoralis major muscle. Computed tomography also more clearly shows the absence of the pectoralis major muscle and allows for a better assessment of associated musculoskeletal abnormalities nearby.

CASE PRESENTATION

Female patient, 53 years old, mixed race, sought this service for routine mammography, without complaints. On physical examination, breast asymmetry was observed, with no other changes. Mammography revealed pectoral muscles visualized only on the left side and total absence of the pectoralis major and pectoralis minor muscles on the right side. A chest X-ray was performed, which showed hypertransparency in the right hemithorax: total absence of the pectoralis major and pectoralis minor muscles on the patient's right side.

Helical computed tomography of the chest was performed, without intravenous contrast, with the aim of evaluating soft tissues and bone structures, which showed agenesis of the pectoralis major and minor muscles on the right.

The vertebral bodies, costal arches, mediastinum and lung parenchyma were unaltered.

DISCUSSION

The incidence of Poland Syndrome varies from 1 in 7,000 to 1 in 100,000 live births and in 75% of cases the anomaly is located in the right half of the body.

There is no specific inheritance pattern, no defined risk factors, and familial recurrence is exceptional. The pathogenesis is not well understood, although it is attributed to the interruption of blood flow from the subclavian artery during the sixth week of gestation.

Most patients with Poland syndrome do

not require surgical treatment to repair the chest deformity. Thus, the surgical indication is reserved for cases where there is unilateral chest depression with risk of progression; lack of lung or heart protection; paradoxical chest wall motion; breast hypoplasia or aplasia in women; aesthetic defect secondary to the absence of the pectoralis major in men.

The radiological evaluation is justified to stage the spectrum of alterations present in addition to the classic findings, directing the treatment. Early diagnosis can contribute to pediatric, orthopedic, aesthetic (with a greater impact on women with breast cancer) and psychomotor management, as well as raising awareness among family members.

The importance of recognizing this condition by the radiologist is to differentiate this benign anomaly from conditions such as the unilateral hypertransparency on chest radiography and breast asymmetry on mammographic examination due to other causes. Poland syndrome has been diagnosed in 1 in 19,000 mammograms.

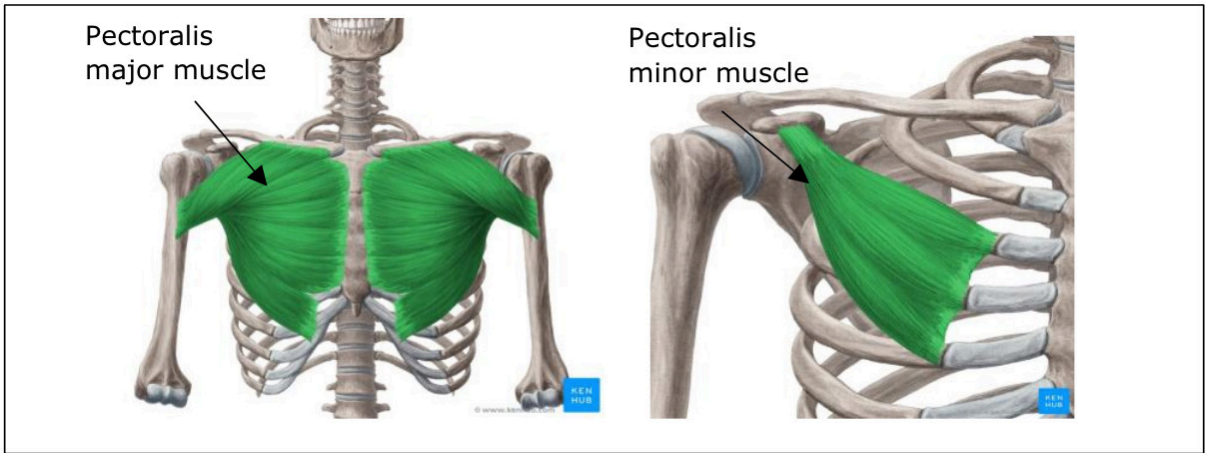


Figure 1. Anatomical schematic view of the pectoral musculature.
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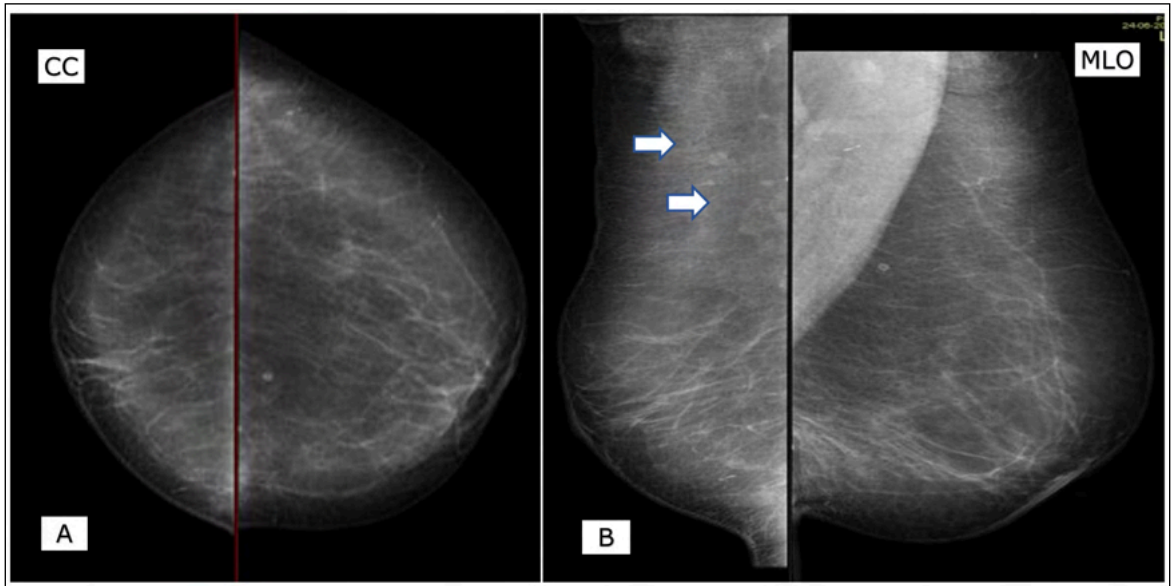


Figure 2. **A:** Mammography in craniocaudal view, showing benign calcification in the left breast and breast asymmetry. **B:** Mammography in oblique mediolateral view showing benign calcification in the left breast and complete absence of the pectoralis major muscle on the right side (arrows).

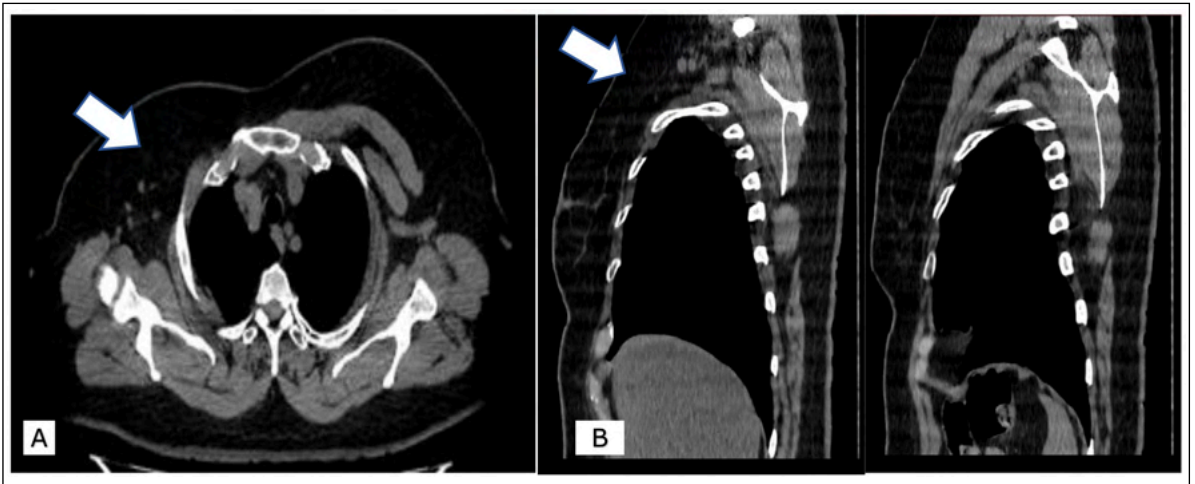


Figure 3. **A:** Computed tomography of the chest in the soft tissue window, in axial section, showing the absence of the pectoralis major and pectoralis minor muscles (arrow). **B:** Computed tomography of the chest in sagittal view confirming the absence of the pectoralis major and minor muscles on the right (arrow) compared to the left side, where the presence of these muscles is noted.

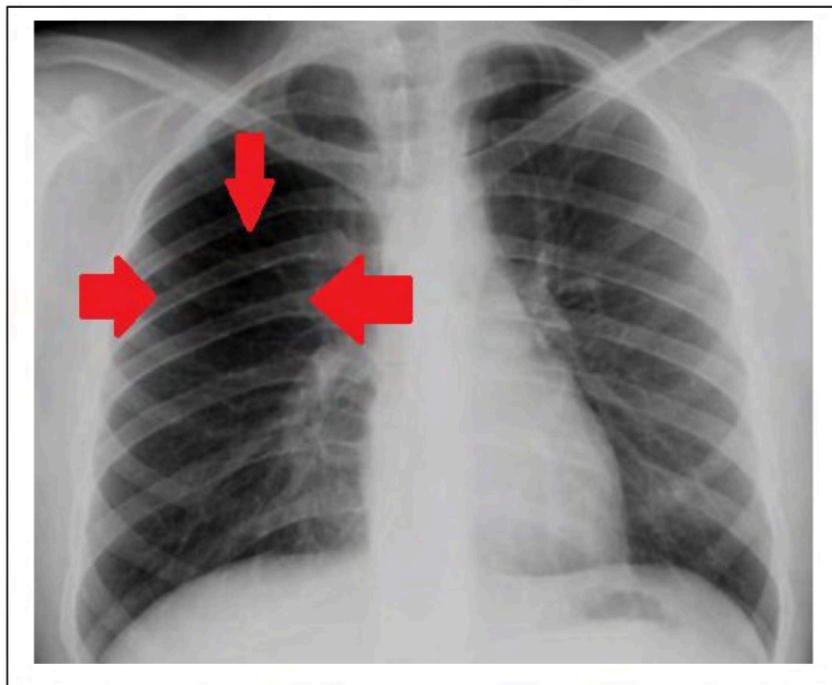


Figure 4. Chest X-ray: hypertransparency of the right hemithorax, due to the absence of the pectoral muscle.

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