



Poland's Syndrome and Breast Cancer: Coincidence or Not?

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Abstract

Poland's syndrome is an uncommon congenital anomaly of unknown etiology, the main characteristic of which is the absence of the major pectoral muscle. Thorax and upper limb malformations also may be present. Poland's syndrome has been observed in clinical cases connected to a variety of neoplasia, primarily hematological disorders. Patients with Poland's syndrome who have developed breast cancer have been reported incidentally. Here we report a case of Poland's syndrome associated with breast cancer.

Keywords

breast cancer, congenital anomaly, malformation, Poland's syndrome

INTRODUCTION

A rare congenital chest wall disorder, Poland's syndrome (PS) affects one in 20,000 to one in 32,000 newborns.^[1] The diagnosis is clinical, and the main criterion is the absence of the pectoralis major muscle, or, more concisely, its sternocostal head. Apart from that, underdevelopment of nipple-areola complex, breast hypoplasia, hypoplasia or absence of pectorals minor and ipsilateral ribs or upper limb anomalies may be present.^[2] Males are affected more often than females, with a ratio of 3:1 and the clinical features predominantly seen on the right side.^[3] The condition was named after Sir Alfred Poland, a British surgeon, who provided a precise description of the syndrome in 1841 and published it in a report entitled "Deficiency of the Pectoral Muscles". But it was Lallemand that first noticed this condition and documented it in 1826.^[3]

CASE REPORT

An 81-year-old woman with a history of chronic lymphocytic leukemia was admitted to our hospital with a palpable solid mass in her right breast. The mass was found by the patient during self-examination and adjoined right nipple-areola complex. A physical examination revealed breast asymmetry with hypoplasia of the right breast and a lump about 2 cm in diameter in the reduced breast. There were no signs of skin ulceration or nipple discharge, but nipple retraction was present. The right axillary lymph nodes were enlarged to 1.5 cm in diameter. It is worth mentioning that no changes were identified in the left breast, except lobular involution (Fig. 1).

Mammography showed heterogeneously dense mass with the size of about 2.2×2.0 cm with spiculated margins in the posterior third of the right breast, lower inner quadrant (Fig. 2).

Ultrasonography confirmed the presence of vertically oriented heterogeneous hypoechoic lesion, measuring

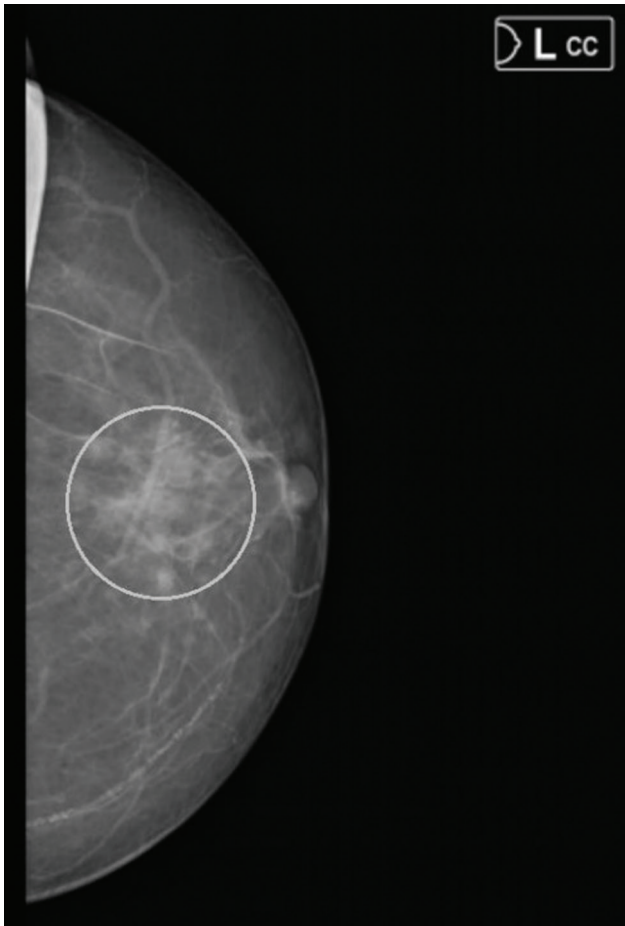


Figure 1. Left breast mammogram demonstrates fibrocystic changes and a zone of higher density (marked) which should be differentiated between fatty tissue or fibroadenoma.

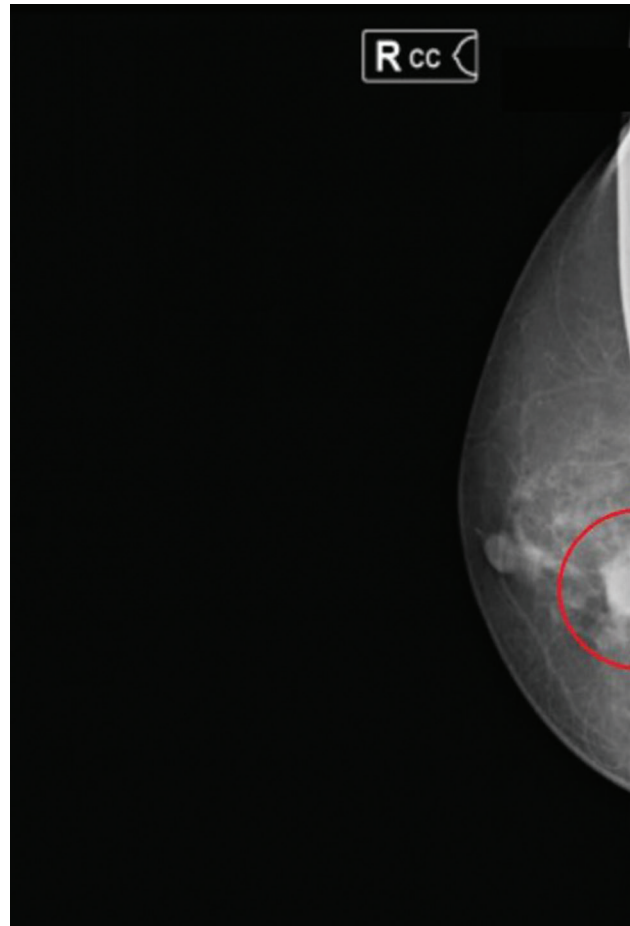


Figure 2. Right breast mammogram image illustrates extremely dense spiculated mass (marked).

1.8×1.5×1.7 cm, with irregular edges and distal acoustic shadow. Doppler ultrasound detected increased blood flow inside the mass and along its borders. A 5.9×4.1 cm right lower jugular lymph node and several lymph nodes (0.4×0.3 cm, 0.5×0.3 cm, 0.5×0.5 cm) in the right axillary fossa were also found, characterized by hypoechogenicity, disrupted corticomedullary differentiation and moderate blood circulation. A chest CT scan diagnosed hyperdense round mass with irregular edges in the right breast of the patient (Fig. 3).

The lesion size was 2.0×1.4 cm and it was adjacent to the retracted nipple. The anterior thoracic wall anatomy was affected, the right pectoralis major and minor were absent. The patient underwent right total mastectomy with lymph node dissection. During the operation, complete absence of the right pectoral muscles was confirmed (Fig. 4).

Pathological examination confirmed the presence of invasive breast carcinoma of non-specific type grade 2 (22 mm in diameter) with several foci of ductal carcinoma in situ (DCIS) grade 1. The tumor affected lymph and blood vessels with no signs of perineural invasion. Also, a metastasis in the right axillary lymph node was identified. Primary

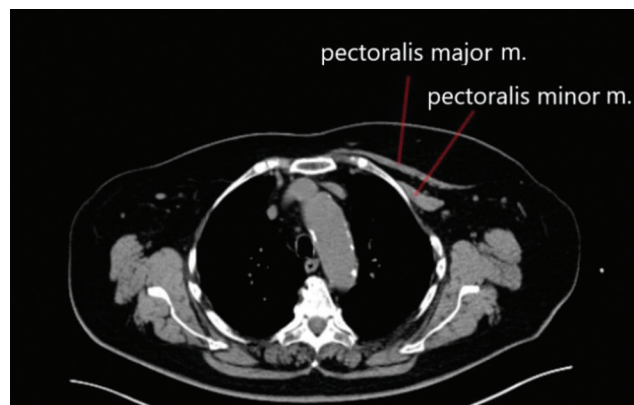


Figure 3. CT scan shows complete absence of the right pectoral muscles and hypoplasia of the right breast.

tumor and metastasis IHC detected luminal-A molecular subtype (ER/PR=100%/80%, HER2- negative). The final diagnosis was invasive breast cancer T2N1M0 with DCIS, luminal type A. The patient had no postoperative complications and was discharged, anastrozole 1 mg was prescribed. No relapse was observed during the 12-month follow-up.

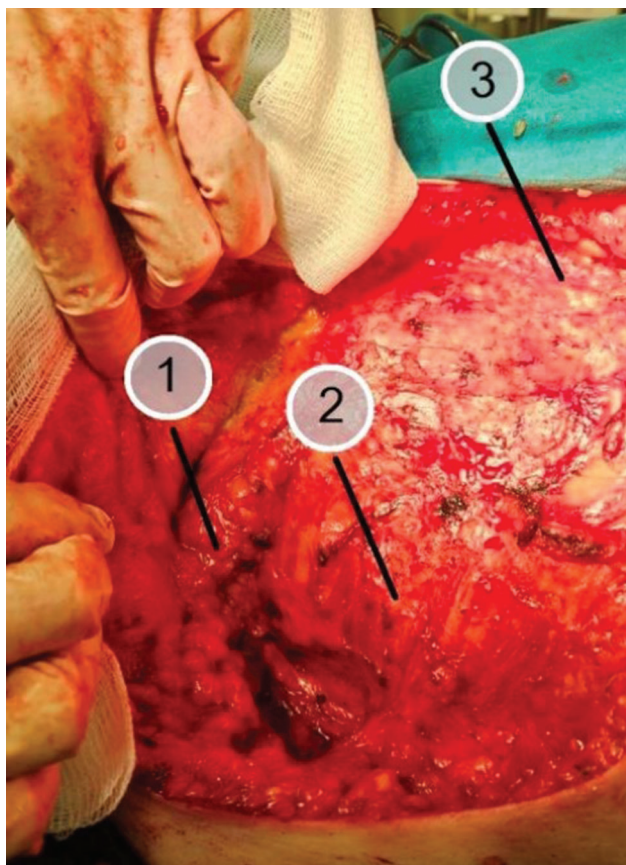


Figure 4. Intraoperative photo, sites of interest are numbered below: 1 – remnants of pectoral major muscle; 2 – intercostal space; 3 – sternum.

DISCUSSION

PS was named after Alfred Poland, who described pectoral muscles absence based on findings of an autopsy at Guy's Hospital. The first who introduced the notion of Poland's syndactyly and paired absence of pectoral muscles and fused fingers was a plastic and hand surgeon Patrick Clarkson. Later, Thompson structured and summarized the whole range of malformations associated with the PS.^[4]

There is still disagreement regarding the exact cause of PS, although vascular and genetic theories are the most prevalent ones. The vascular hypothesis states that what causes the PS is the disrupted blood flow distribution in the subclavian artery and its branches during the sixth week of gestation. Genetic predisposition to PS is supported by reports of familial cases and association with other congenital abnormalities, such as dextrocardia or Moebius syndrome.^[5] It is suggested that the PS, Klippel-Feil and the Moebius syndromes have common pathogenesis, subclavian artery supply disruption, and thus, they can be presented together.^[4]

PS is phenotypically variable. In both sexes, anterior axillary fold is poorly formed due to aplasia or apparent hypoplasia of the pectoralis major muscle. Usually, PS is not accompanied by functional restrictions of the affected upper limb. Wrist and hand hypoplasia is reported in

89% of the cases, forearm hypoplasia is present in 37% of the cases. Very often, such hand anomalies as symbrachydactyly (89%) and short middle phalanges (45%) are seen. Lower extremity involvement, rib aplasia, keeled (9.7%) or hollowed (0.8%) chest are rare.^[4] Apart from musculoskeletal deformities, craniofacial, dermatological and internal organs anomalies may be present. For example, there was a case of PS in a neonate with significant lung herniation through the left sided rib defect. Cases of unilateral renal agenesis, wandering spleen, hepatic exstrophy have also been reported. The dermatologic features in PS are not specific, and they include anhidrosis, axillary or pectoral alopecia, and, more rarely, café-au-lait spots.

Currently, there is a documented association with neuroblastoma, Wilms tumor, lymphoproliferative disorders, and various cancers such as breast and lung cancer.^[6] Hereditary gene abnormalities are assumed to be the cause of PS patients' predisposition to carcinomas. All the same, this is still up for debate. A case of monozygotic twins with PS is known – they were tested for genetic anomalies using array-comparative genomic hybridization (array-CGH). CGH detected deletion of 5 genes on chromosome 11. These genes were involved in regulating the cellular growth, differentiation, and apoptosis. Both twins presented with breast asymmetry, underdevelopment of pectoral muscles and one of them also had right hand hypoplasia.^[7]

Breast cancer (BC) is the most commonly diagnosed cancer in women. It has increased in incidence recently, due primarily to the increase of localized-stage and hormone receptor (HR)-positive cases. But the mortality rate of breast cancer has declined, reaching a 43% decrease since its peak in 1989. Interestingly, 30% of all BC cases can be attributed to the modifiable risk factors, such as excessive weight, sedentary lifestyle, malnutrition, tobacco smoking, and alcohol consumption.^[8] BC subtypes according to hormone receptor (HR) and HER2 status are HR(+)/HER2(-), HR(+)/HER2(+), HR(-)/HER2(+), and HR(-)/HER2(-). The HR(+)/HER2(-) subtype is the most common and has the best prognosis. Overall, HR(+) patients have higher OS and breast cancer specific survival (BCSS) than HR(-) patients.^[9]

So far, only approximately 20 cases of PS associated with breast cancer have been reported. The median patients' age is 50 years. Mostly, mammary hypoplasia and ipsilateral BC relative to the PS manifestations are presented. The vast majority of cases comprise invasive ductal carcinoma, or, more rarely, a combination of several histological types. No clinical cases of such patients with distant metastases have yet been reported.

The absence of costal cartilage in second to fifth ribs (18% in right-sided PS vs. 40% in left-sided PS) makes chest wall organs more vulnerable to damage. Therefore, musculoskeletal defects should be diagnosed during ultrasonography, CT or MRI before performing invasive procedures, such as core-needle biopsy or surgery. The surgical option in these cases remains controversial.^[10]

Therefore, anatomical anomalies should be thoroughly examined and concisely interpreted using diagnostics im-

aging, since their presence may change tactics of surgical intervention and increase risk of heart and lung traumatization. Skeletal and soft tissue malformations may require use of reconstruction techniques.

CONCLUSION

It was shown that patients with Poland's syndrome are at a risk of developing malignancy, including breast cancer. Breast cancer diagnostics and treatment are the same as in patients without the disease. More clinical data is needed to identify the main cause of breast cancer in patients with the Poland's syndrome.

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Competing Interests

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Author contributions

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Синдром Поланда и рак груди: совпадение или нет?

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Резюме

Синдром Поланда – редкая врождённая аномалия неизвестной этиологии, главной характеристикой которой является отсутствие большой грудной мышцы. Могут также присутствовать пороки развития грудной клетки и верхних конечностей. Синдром Поланда наблюдался в клинических случаях, связанных с различными неоплазиями, в первую очередь гематологическими расстройствами. Пациенты с синдромом Поланда, у которых развился рак молочной железы, были зарегистрированы случайно. Здесь мы сообщаем о случае синдрома Поланда, связанного с раком молочной железы.

Ключевые слова

рак молочной железы, врождённая аномалия, порок развития, синдром Поланда