

Breast Carcinoma Associated with Poland's Syndrome: One Case Report and Literatures Review

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Introduction

Poland's syndrome is a rare congenital anomaly, characterized by abnormalities of the chest wall, breast, spine and upper limb. The incidence of this syndrome has been estimated to be 1:30000. The pathogenesis is still unclear^[1].

Case Report

A 46 year-old female was admitted to our hospital presenting with a right breast mass that was noted 2 years ago. She had suffered from a right-sided Poland's syndrome from birth. Two years ago prior to being admitted, she discovered a breast mass about 1 cm in diameter. There had been no pain or nipple discharge, but recently the mass grew rapidly over the last 6 months reaching about the size of an egg.

Chest X-rays showed scoliosis, a congenital malformation, malformation of ribs 3, 4 and 5, confluence of ribs 4 and 5, and extenuation of rib 8 (Fig.1). On physical examination the patient showed an asymmetrical thoracic skeleton, and that her left shoulder was lower than her right shoulder. Her breasts were asymmetrical, as the right breast was smaller than the left. A mass of $5 \times 5 \times 4$ cm in size that was hard, irregular and inflexible was noted in the lower outer quadrant of the right breast. The ipsilateral nipple was retracted and an ipsilateral axillary lymph node of 1.0 cm was noted.

Mammography disclosed an irregular mass, the retracted nipple and no calcification in the right breast and no mass or calcification in the left breast. Mammographic diagnosis: right breast cancer, left breast hyperplasia. Ultrasonography demonstrated an irregular solid mass with a size of 3.7×2.0 cm suggesting a diagnosis of right breast cancer. A core needle biopsy confirmed the diagnosis of invasive breast cancer.

The patient received two cycles of neoadjuvant chemotherapy with administration of FEC (cyclophosphamide 1 g, Epirubicin 80 mg, Tegafur 1 g) and then underwent a modified radical mastectomy. During the operation a malformation of ribs 3 to 5 was noted and ribs 6 and 7 were absent. Pectoralis major and pectoralis minor muscles showed hypogenesis. The pathological diagnosis revealed a right breast invasive ductal carcinoma with a histological grade of II, and skin and nipple were also invaded. Reaction to chemotherapy showed degree I. LN: level II 0/5, level I 3/17. ER and PR were positive, but CerbB2 negative. The patient received 4 further cycles of chemotherapy with FEC post-operation, followed by radiation therapy, and now

is being treated with tamoxifen and living without any other health problems during 2.5 years of follow-up.



Fig.1. Chest X-ray: spine inflection and malformation of the right ribs.

Discussion

Poland's syndrome is a congenital malformation that was first reported by Alfred Poland in 1841 in the "Guy Hospital Report"^[2]. The report noted that the pectoralis major and pectoralis minor muscles were less developed or completely absent, and an upper extremity was malformed. In 1862 Clarkson confirmed these anatomic malformations during an operation and named them as Poland's syndrome^[3].

Subsequent reports on the anatomic diversity were added as additional components to this syndrome. Poland's syndrome includes congenital malformations of the breast, chest, trunk and upper limb. Malformations of the chest and trunk include hypoplasia or complete absence of the breast and/or nipple; costal cartilage defects and rib defects; lack of fat tissue on the chest wall; malformations or absence of trunk muscles, which include the latissimus dorsi, deltoid, oblique externus abdominis, serratus anterior, rectus abdominis, supra spinalis and inferior spinalis. It is uncommon that a patient has all these defects.

Poland's syndrome is a typical unilateral disease, with the exception of a case report on bilateral congenital malformation. The male to female ratio of this syndrome is 3 to 1 and 75% are found on the right side. The estimated incidence of this syndrome varies from 1 in 20,000 to 1 in 50,000 new births. The most commonly described finding is a mammary anomaly or hypoplasia.

Poland's syndrome is often diagnosed through a clinical examination, X-ray, computerized X-ray tomography (CT) or nuclear magnetic resonance imaging (MRI). A physician should take notice of an anomaly of the chest skeleton and/or pectoralis muscles when a patient is seen with mammary asymmetry or unilateral hypoplasia. The etiopathogenesis of Poland's syndrome is unclear, and it is without a specific definition. All reported cases have been sporadic, and no clear genetic evidence has been

found. An identical twin was reported with this syndrome, but the other child was not effected. One pathogenic hypothesis is that there was a disruption in the distribution of vessels supplying the trunk and upper limb in the fetus at 6 weeks of gestation. Local ischemia may cause malformation of the supraclavicular artery, vertebral artery or others followed by subsequent hypoplasia. Another hypothesis is that smoking by pregnant women increases the risk of Poland's syndrome in newborns.

There have been a number of reports on various malignancies in patients with Poland's syndrome, which include leukemia^[4], lymphoma^[5], breast cancer^[6–11], lung cancer^[12], stomach cancer^[13] and head and neck tumors^[14]. Seven studies including 1 report from China^[15], reported breast cancer in a Poland's syndrome patient. Among these 7 cases, 6 patients had ipsilateral breast cancer on the side with mammary hypoplasia. Only one case reported left breast cancer with right mammary hypoplasia. All of these patients showed anomalies of bone, cartilage or pectoralis muscles defects in addition to mammary hypoplasia. In our case the patient had right side mammary hypoplasia and also rib malformation. Women with Poland's syndrome often have received breast reconstruction^[16]. Katz et al.^[11] reported on a 42-year old women who received left breast reconstruction because of hypoplasia. Two years later an ipsilateral mammary mass was found. Mammography was unable to identify the tumor, but ultrasonography showed a solid and hypoechoic mass anterior to the saline implant. The final diagnosis of breast cancer was confirmed by pathology.

The relationship between hypoplasia and cancer is still unclear. However there have been numerous reports of cancer in Poland's syndrome patients and physicians must be aware of a cancer risk in these cases. Breast cancer may occur even in patients with unilateral mammary hypoplasia. Furthermore, Poland's syndrome patients who receive breast reconstruction must be followed-up by additional examinations in order to achieve early cancer detection and treatment.

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