

## Case Report

# Carcinoma of Breast Associated with Poland Syndrome: A Rare Case Report and Literature Review

Diwan Shrestha<sup>\*1</sup>, Manish Roy<sup>1</sup>, Utsab Man Shrestha<sup>1</sup>, Punyaram Kharbuja<sup>1</sup>, Bikash Bhaila<sup>1</sup> and Diksha Karki<sup>2</sup>

<sup>1</sup>Department of Surgical Oncology, Bhaktapur Cancer Hospital, Bhaktapur, Nepal.

<sup>2</sup>Department of Pathology, Bhaktapur Cancer Hospital, Bhaktapur, Nepal.

**Corresponding Author:** Diwan Shrestha, Department of Surgical Oncology, Bhaktapur Cancer Hospital, Bhaktapur, Nepal.

Received: 📅 2023 Dec 02

Accepted: 📅 2023 Dec 21

Published: 📅 2024 Feb 26

## Abstract

Poland syndrome is a rare congenital malformation, characterized by a congenital defect in the pectoralis muscle. It is associated with various ipsilateral upper extremity anomalies and homolateral breast hypoplasia. Here in, we report a case of Poland syndrome associated with breast cancer in a 66 years female and review the literature.

**Keywords:** Breast Cancer, Poland Syndrome, Pectoralis Muscle Defect and Breast Hypoplasia.

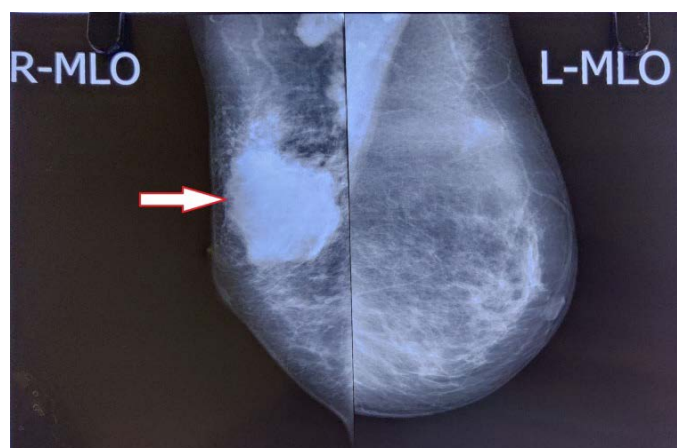
## 1. Introduction

Poland syndrome is characterized by the absence of pectoralis major and minor muscles, hypoplasia or complete absence of the breast, costal cartilage and rib defects, and hypoplasia of the subcutaneous chest wall tissue. Lallemand described Poland syndrome for the first time in 1826, but Alfred Poland precisely defined it in 184 [1]. Other components of Poland syndrome include ipsilateral syndactyly and monodactyl, dextrorotation of the heart, lung hypoplasia, and lung herniation as well as vertebral and subclavian vein (and its tributaries) abnormalities [2]. Poland syndrome has been reported to be associated with various malignancies such as leukemia, Wilms tumor, malignant lymphoma, and leiomyosarcoma [3, 4]. However, there are only a few reports of Poland syndrome associated with breast carcinoma. This case report describes a case of Poland syndrome associated with invasive ductal carcinoma of the breast.

## Case Report

A 66-year female presented with a painless, palpable lump in her right breast. The patient had no family history of breast cancer. Upon examination, her breasts were asymmetrical, with hypoplasia of the right breast. There was an approximately 5 x 6 cm irregular, hard mass in the upper outer quadrant of the right breast without skin ulceration. No lymphadenopathies were observed in the axillary or supraclavicular regions. Ultrasound examination showed a heterogenous space occupying lesion with calcification and

mammography revealed a lobulated irregular margined radio dense lesion in the right breast with asymmetrical breast (Figure 1). Computed tomography of the chest showed a heterogeneously enhancing multilobulated lesion in the lateral half of the right breast. PET CT showed a soft tissue lesion with speculated margins in the outer quadrant of the right breast with missing right pectoralis major and minor muscles, anterior part of the right third and fourth ribs, and intercostal muscles (Figure 2).

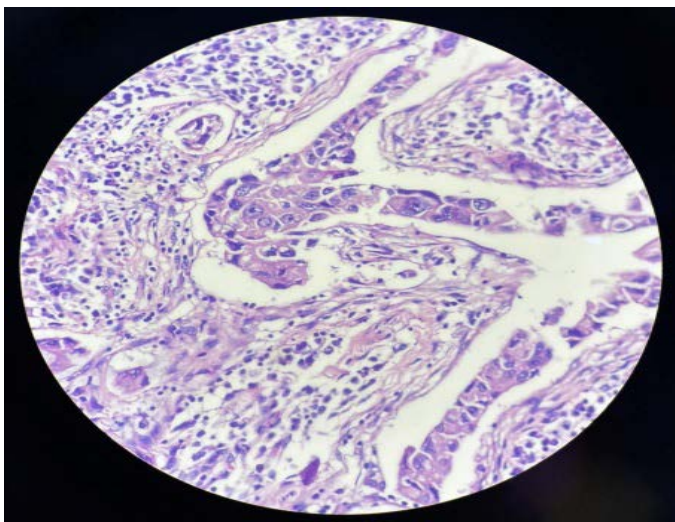


**Figure 1:** Mammography Showing a Lobulated Irregular Margined Radiodense Lesion in Right Breast with Asymmetrical Breasts.

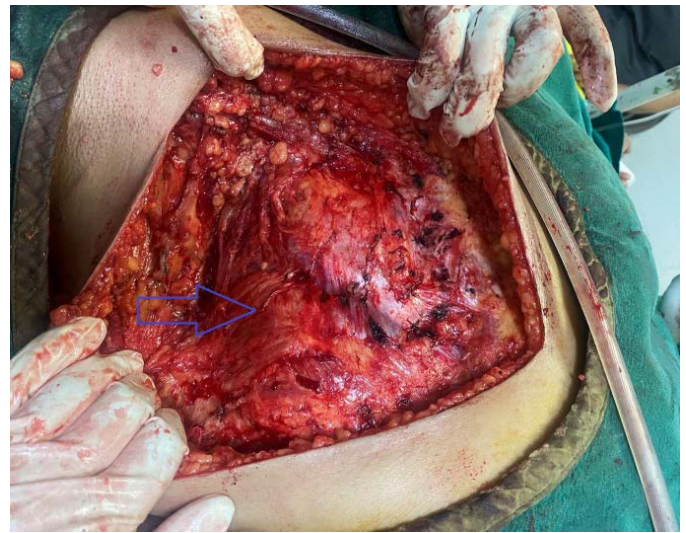


**Figure 2:** PET CT Showing Soft Tissue Lesion with Speculated Margins in Outer Quadrant of Right Breast with Missing Right Pectoralis Major and Minor Muscles, Anterior Part of Right Third and Fourth Ribs and Intercostal Muscles.

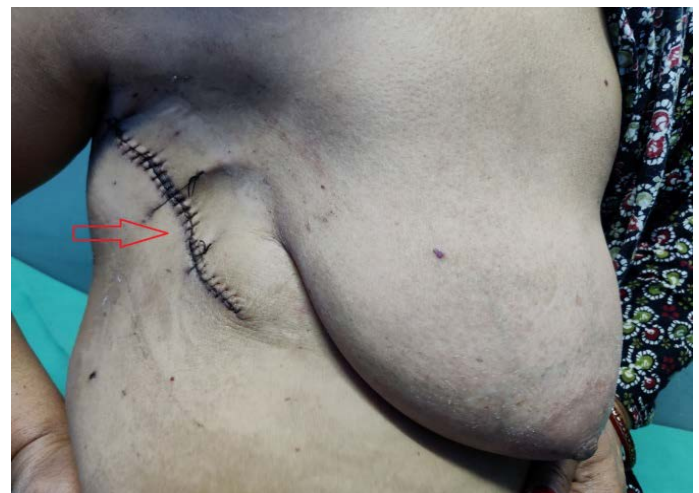
Trucut biopsy showed invasive ductal carcinoma. Immunohistochemical staining of the tumor cells were negative for estrogen and progesterone receptors, positive for HER 2 Neu expression (3+), and ki 67 (index 60%). She received six cycles of neoadjuvant chemotherapy (Docetaxel, Carboplatin, and Trastuzumab). Figure 3 shows the residual tumor following neoadjuvant chemotherapy. Right modified radical mastectomy was performed. The intraoperative findings revealed a 3 x 2 cm hard lump in the upper outer quadrant of the right breast with few enlarged axillary lymph nodes, absent right pectoralis major and minor muscles and absent anterior part of the right third and fourth ribs (Figure 4). Pathological staging was ypT1bN0M0. Her postoperative course was uneventful (Figure 5).



**Figure 3 :** Residual tumor post NACT.



**Figure 4 :** Intraoperative picture showing absent right pectoralis major and minor muscles and absent anterior part of right third and fourth ribs.



**Figure 5:** Postoperative picture.

## 2. Discussion

Poland syndrome is a rare congenital anomaly and its incidence varies from 1:36,000 to 1:50,000 [5]. This syndrome has a male predominance with a male to female ratio of 3:1, and 75% of the deformities occur on the right side [6]. This syndrome has unclear etiology, but the results of an epidemiological study showed that the risk of Poland's anomaly increased with maternal smoking during pregnancy [7]. It is also reported to be caused by obstruction of blood flow in the subclavian arteries and vertebral arteries around the sixth week of fetal gestational development [8].

Until now, only 18 cases of breast carcinoma associated with Poland syndrome are reported. Although there is a male dominance in Poland syndrome, all cases of breast cancer with Poland syndrome occurred in females within the age range between 33 to 74 years (including the patient in this report). Among these 19 cases (including those in this report), 18 patients had ipsilateral breast carcinoma on the side of mammary hypoplasia, and only one patient had contralateral breast carcinoma associated with Poland syn-

drome (Table 1). Although all reported cases had absence of pectoralis muscle, hypoplasia of the upper extremities were present only in 10 cases. Histologically, invasive ductal carcinoma (IDC) was predominant. Estrogen receptor and / or progesterone receptor were positive in most of the cases.

**Table 1: Reported Cases of Breast Carcinoma with Poland Syndrome.**

Refer-ence	Age	Side	Absence of pec-toralis muscle	Breast hy-poplasia	Hypoplasia of upper extremity	Histology	Stage	ER	PR
Khandel-wal et al. (9)	71	R	Yes	Ips	No	IDC, DCis	Ia T1N0M0	+	-
Havlik et al.(10)	33	R	Yes	Ips	Yes	IDC, ILC	IIA T2N0M0	+	-
Katz et al. (11)	42	L	Yes	Ips	No	IDC	Ia T1N0M0	-	+
Fukushi-ma et al. (12)	57	R	Yes	Ips	Yes	IDC	IIa T1N1M0	+	+
Fukushi-ma et al. (12)	53	L	Yes	Ips	Yes	IDC	IIa T2N0M0	-	-
Tamio-lakis et al. (13)	53	L	Yes	Ips	Yes	IDC	Ia T1N0M0	-	+
Okamo et al. (14)	59	L	Yes	Con	Yes	IDC	IIA T1N1bM0	-	+
Wong et al. (15)	51	L	Yes	Ips	No	DCis	0 TisN0M0	N/A	N/A
Caussa et al. (16)	43	L	Yes	Ips	No	IDC	IIIa T3N1M0	+	N/A
Un~a et al. (17)	39	R	Yes	Ips	Yes	IDC	Ia T1cN0M0	+	+
Salhab et al. (18)	52	L	Yes	No	No	IDC, DCis	Ia T1N0M0	N/A	N/A
Ji et al. (19)	58	L	Yes	No	No	IDC	IIa T2N0M0	N/A	N/A
Zhang et al. (20)	43	L	Yes	Ips	Yes	IDC	IIIc T1N3M0	+	+
Wang et al. (21)	46	R	Yes	Ips	Yes	IDC	IIb T2N1M0	N/A	N/A
Zhou et al. (22)	42	R	Yes	Ips	Yes	IDC	IIIc T2N3M0	N/A	N/A
Huang et al. (23)	60	L	Yes	Ips	No	IDC	IIIb T4N1M0	-	-
Huang et al. (23)	74	L	Yes	Ips	Yes	IDC	IIb T3N0M0	+	+
Yesilkaya et al. (24)	39	L	Yes	No	No	IDC	IIa T2N0M0	N/A	N/A
This report	66	R	Yes	Ips	No	IDC	I T1b- N0M0	-	-

L: left breast cancer; R: right breast cancer; is: ipsilateral; con: contralateral; IDC: invasive ductal carcinoma; ILC: invasive lobular carcinoma; DCis: ductal carcinoma in situ; ER: estrogen receptor; PR: progesterone receptor; (+): positive; (-): negative; N/A: not available.

Compared to the general population, women with Poland syndrome have a greater risk of breast carcinoma [25]. Therefore, self-examination of the hypoplastic breast should be done in these patients. Most tumors in Poland syndrome occur in the ipsilateral hypoplastic breast, but ipsilateral or contralateral normal breast can also be involved. So, in the examination of the breasts of Poland syndrome, both hypoplastic and normal breast should not be ignored for the possibility of neoplasia. In Poland syndrome, fat tissue on the chest wall is deficient, and the pectoralis muscles are usually absent, resulting in the thinness of the involved chest. During traumatic procedures such as fine needle aspiration cytological (FNAC) examination and when a breast tumor is excised from a patient with Poland syndrome, thoracic organs and axillary vein may be injured, hence it should be done with caution.

Based on the histopathological diagnosis, breast cancers that developed in Poland syndrome did not differ from normal breast cancers, but lymph node metastases (31%) occurred more commonly in Poland syndrome [26]. Hence, adjuvant radiotherapy may be necessary. As there is decreased protection by chest wall muscles in Poland syndrome, they have an increased risk of heart and lung complications and adjuvant radiotherapy should be offered with caution.

### 3. Conclusions

Carcinoma of the breast associated with Poland syndrome is rare, but physicians must be aware of cancer risks in these patients. In Poland syndrome, there is a congenital defect of the pectoralis muscle which can affect the treatment decisions in breast carcinoma.

### Funding

The authors received no financial support for the research, authorship, and publication of this article.

### References

- Poland, A. (1841). Deficiency of the pectoral muscles. *Guy's Hosp Rep*, 6, 191.
- Avci, G., Mısırlıoğlu, A., Eker, G., Aköz, T. (2003). Mild degree of Poland's Syndrome reconstruction with customized silicone prosthesis. *Aesthetic plastic surgery*, 27, 112-115.
- Shaham, D., Ramu, N., Bar-Ziv, J. (1992). Leiomyosarcoma in Poland's syndrome: a case report. *Acta Radiologica*, 33(5), 444-446.
- Hoefnagel, D., Rozycki, A., Wurster-Hill, D., Stern, P., Gregory, D. (1972). Leukaemia and Poland's syndrome. *The Lancet*, 300(7785), 1038-1039.
- ÇAKSEN, H., PATIROGLU, T., ÖZDEMİR, M. A., PATIROGLU, T. E., POYRAZOGLU, M. H., et al. (1997). Neuroblastoma and Poland syndrome in a 15-year-old boy. *Pediatrics International*, 39(6), 701-704.
- Fokin, A. A., Robicsek, F. (2002). Poland's syndrome revisited. *The Annals of thoracic surgery*, 74(6), 2218-2225.
- Martínez-Frías, M. L., Czeizel, A. E., Rodríguez-Pinilla, E., Bermejo, E. (1999). Smoking during pregnancy and Poland sequence: Results of a population-based registry and a case-control registry. *Teratology*, 59(1), 35-38.
- Bavinck, J. N. B., Weaver, D. D., Opitz, J. M., Reynolds, J. F. (1986). Subclavian artery supply disruption sequence: Hypothesis of a vascular etiology for Poland, Klippel-Feil, and Möbius anomalies. *American journal of medical genetics*, 23(4), 903-918.
- Khandelwal, A., O'Hea, B. J., Garguilo, G. (2004). Breast cancer in a patient with Poland's syndrome. *The American Surgeon*, 70(6), 491-495.
- Havlik, R. J., Sian, K. U., Wagner, J. D., Binford, R., Broadie, T. A. (1999). Breast cancer in Poland syndrome. *Plastic and reconstructive surgery*, 104(1), 180-182.
- Katz, S. C., Hazen, A., Colen, S. R., Roses, D. F. (2001). Poland's syndrome and carcinoma of the breast: a case report. *The Breast Journal*, 7(1), 56-59.
- Fukushima, T., Otake, T., Yashima, R., Nihei, M., Takeuchi, S., et al. (1999). Breast cancer in two patients with Poland's syndrome. *Breast cancer*, 6, 127-130.
- Tamiolakis, D., Venizelos, I., Antoniou, C., Tsiminikakis, N., Alifieris, E., et al. (2004). Breast cancer development in a female with Poland's syndrome. *Oncology Research and Treatment*, 27(6), 569-571.
- Okamoto, H., Miura, K., Yamane, T., Fujii, H., Matsumoto, Y. (2002). Invasive ductal carcinoma of the breast associated with Poland's syndrome: report of a case. *Surgery today*, 32, 257-260.
- Wong, T. C. M., Lim, J., Lim, T. C. (2004). A case of ductal carcinoma in situ of breast with Poland syndrome. *ANNALS-ACADEMY OF MEDICINE SINGAPORE*, 33, 382-384.
- Caussa, L., Kirova, Y. M., Campana, F., Fourchette, V., Salmon, R. J. (2009). Poland syndrome, breast cancer: the importance of the radiotherapy technique after mastectomy. *Radiotherapy and Oncology*, 91(1), 138-139.
- Uña, J., Vega, V., Gutierrez, I., Herrera, J., Hernández-Briz, M. J. (2007). Breast cancer, Poland's syndrome, and sentinel lymph node involvement. *Clinical Nuclear Medicine*, 32(8), 613-615.
- Salhab, M., Al Sarakbi, W., Perry, N., Mokbel, K. (2005, December). Pneumothorax after a clinical breast fine-needle aspiration of a lump in a patient with Poland's syndrome. In *International Seminars in Surgical Oncology (Vol. 2, No. 1, pp. 1-5)*. BioMed Central.
- Ji, J., Zhang, S., Shao, C., Xu, M., Chen, S., Lu, C., et al. (2008). Poland's syndrome complicated with breast cancer: mammographic, ultrasonographic, and computed tomographic findings. *Acta Radiologica*, 49(4), 387-390.
- Zhang, F., Qi, X., Xu, Y., Zhou, Y., Zhang, Y., et al. (2011). Breast cancer and Poland's syndrome: a case report and literature review. *The breast journal*, 17(2), 196-200.
- Wang, X., Ning, L. (2008). Breast carcinoma associated with Poland's syndrome: One case report and literatures review. *Chinese Journal of Clinical Oncology*, 5(3), 223-225.
- Zhou, B., Li, H., Fan, Z. (2010). A case of Poland's syndrome with breast cancer. *Chin J Breast Dis (Electronic Edition)*, 2, 227-9.
- Huang, Y., Pang, H., Jin, S., Han, X., Liu, X., Y et al. (2018).

- Clinical characteristics of Poland's syndrome associated with breast cancer: Two case reports and a literature review. *Journal of Cancer Research and Therapeutics*, 14(7), 1665-1669.
24. Yesilkaya, Y., Dizdar, O., Altundag, K. (2011). Ipsilateral breast cancer in a patient with Poland's syndrome. *The American Surgeon*, 77(2), 234-235.
25. Huang, Y., Pang, H., Jin, S., Han, X., Liu, X., et al. (2018). Clinical characteristics of Poland's syndrome associated with breast cancer: Two case reports and a literature review. *Journal of Cancer Research and Therapeutics*, 14(7), 1665-1669.
26. DeFazio, M. V., Dervishaj, O. A., Bozzuto, L. M., Pittman, T. A., Olding, M. J., et al. (2018). Delayed recurrent and bilateral breast cancer in patients with partial Poland's anomaly: report of 2 rare cases and review of the literature. *Clinical breast cancer*, 18(3), e285-e290.