

Primary rhabdomyosarcoma of the breast in a 17-year-old girl

Case report

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Abstract

Rationale: Primary rhabdomyosarcoma of the breast is very rare disease with poor prognosis and no definitive treatment has yet been established.

Patient concerns: A 17-year-old girl presented with right breast mass without distant metastasis in image study.

Diagnosis: The result of core needle biopsy was intraductal carcinoma; however, histopathologic finding after mastectomy was primary rhabdomyosarcoma of breast.

Interventions: Adjuvant chemotherapy was recommended because resection margin was involved by tumor cells, but she did not visit the clinic anymore. Five months later, tumor recurred with local invasion and chemotherapy of vincristine, doxorubicin, and cyclophosphamide alternating with ifosfamide and etoposide (VAC/IE) was done.

Outcomes: In the course of chemotherapy and sequential follow-up, there was no tumor growth until now.

Lessons: Primary breast rhabdomyosarcoma is an uncommon disease, as a result diagnosis is often delayed. For the same reason, there is little information about treatment. This report may be helpful for managing the disease.

Abbreviations: IRS = Intergroup Rhabdomyosarcoma Study Group, NSM = nipple sparing mastectomy, PET-CT = positron emission tomography-computed tomography, VAC/IE = vincristine, doxorubicin and cyclophosphamide alternating with ifosfamide and etoposide.

Keywords: alveolar type, breast cancer, chemotherapy, rhabdomyosarcoma

1. Introduction

Most breast masses in children and adolescents are benign and self-limited.^[1,2] However, even in very rare cases, primary breast cancer may occur in child and adolescents. Juvenile secretory carcinoma is the most common (more than 80% of cases), followed by intraductal carcinoma. Although rare, rhabdomyosarcoma and lymphoma also occur as a primary tumor of the breast.^[3] We report a case of primary breast rhabdomyosarcoma in a 17-year-old girl.

2. Case presentation

A 17-year-old post-menarche girl referred to our center with palpable mass of right breast. The result of core needle biopsy performed at local clinic was intraductal carcinoma. In imaging study, a 3.1 cm irregular heterogeneous enhancing mass at right upper inner quadrants without lymph node enlargement in both axilla was identified (Fig. 1A). Laboratory test was normal and no clinical evidence of distant metastasis at positron emission tomography-computed tomography (PET-CT) and systemic examination.

The patient underwent right nipple sparing mastectomy (NSM) and breast reconstruction with implant. The sentinel lymph node biopsy performed during surgery was negative for malignancy. Histopathologic finding of resected mass was an alveolar rhabdomyosarcoma with lympho-vascular invasion and medial resection margin was involved by tumor. When immunohistochemical stained, the specimen was myogenin, desmin, and vimentin positive, indicating that the tumor originated from muscle tissue (Fig. 2A–D). She was referred to oncology part and adjuvant chemotherapy was recommended. However, she did not visit the clinic anymore.

Five months later, she appeared with right chest mass and symptoms caused by SVC syndrome. The tumor extended beyond the sternum to the left side and both axilla lymph nodes (Fig. 1B). Chemotherapy of vincristine, doxorubicin, and cyclophosphamide alternating with ifosfamide and etoposide (VAC/IE) regimen began in January and the mass remained stable until June when the 51-week schedule was completed. Since the end of chemotherapy, she has remained stable until now.

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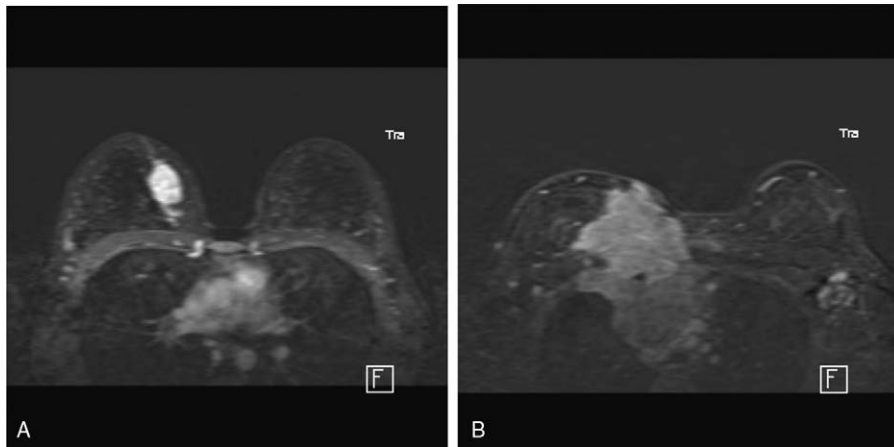


Figure 1. Magnetic resonance imaging (A) at diagnosis showed 3.1 cm irregular heterogeneous enhancing mass at right breast, (B) at recur showed huge heterogenous irregular enhancing mass at right upper central to right inner breast area. Mass invaded to right chest wall directly and extended beyond the sternum to mediastinum.

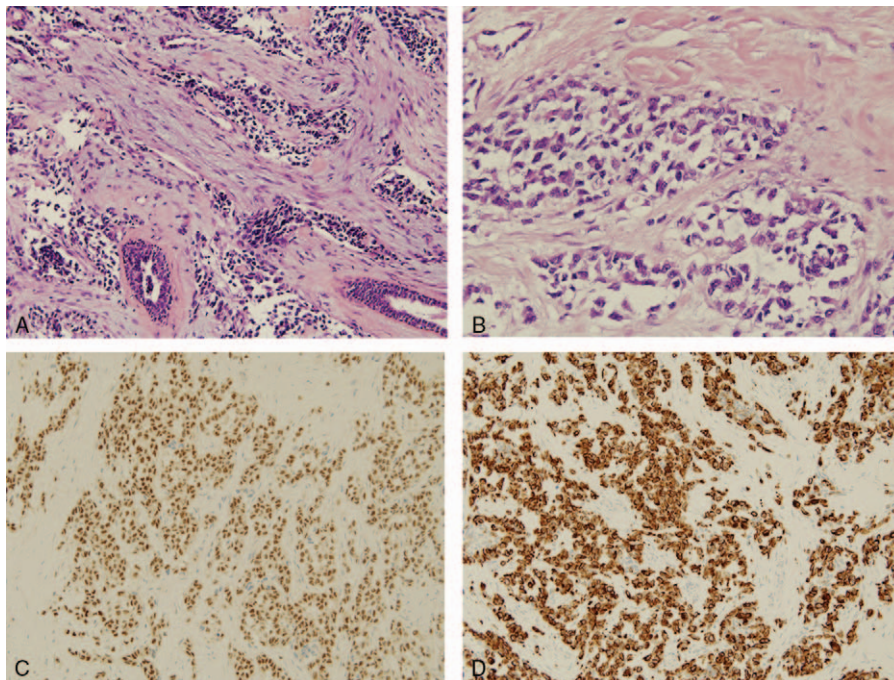


Figure 2. Hematoxylin and eosin stain of tumor tissue showed (A) tumor cells arranged in alveolar pattern ($\times 200$) and the (B) tumor consisted of loose nests of highly malignant round blue cells with scanty amount of cytoplasm ($\times 400$). Immunohistochemical study using (C) vimentin and (D) desmin revealed positive staining of the tumor cells ($\times 200$).

The patient provided written informed consent and ethics approval was not required for this paper as it is a case report.

3. Discussion

Breast cancer is the one of most commonly diagnosed cancer in women worldwide.^[4] In most cases, it is an epithelial origin and nonepithelial origin account for less than 5% of all breast neoplasms. Breast nonepithelial neoplasm usually rapidly increase in size and larger than epithelial origin neoplasm. However, it is difficult to distinguish nonepithelial breast malignancy from other breast neoplasms only by clinical features.

Histologically, H&E stain is not enough for diagnosis and immunostaining may helpful. Precise diagnosis is important because they are different in prognosis and management.^[5]

In children and adolescents, soft tissue tumors are more common than adult and among soft tissue tumor, rhabdomyosarcoma is the most common (4–8%) malignancy.^[6] However, primary breast rhabdomyosarcoma is relatively rare. According to the previous reports from Intergroup Rhabdomyosarcoma Study Group (IRS) of the United States, only 0.2% of rhabdomyosarcoma patients were diagnosed breast origin. On the other hand, when confined to age between 10 and 21 years of age, 1.6% had breast origin rhabdomyosarcoma.^[7] Among the 2

histological types of rhabdomyosarcoma, alveolar type accounts for about 20%. It has a worse prognosis than embryonal type and develops in the extremities of children over 10 years old with tendency of early metastasis and poor response to chemotherapy.^[8,9]

Even some causative factors, for example, prior RT and conditions causing chronic lymphedema known as associated with secondary breast sarcoma, there is no proven association between causative factor of soft tissue sarcoma and primary breast sarcomas.^[10] Predisposing genetic conditions, including Li-Fraumeni syndrome, familial adenomatous polyposis and its variants or neurofibromatosis type 1 can sometimes be identified, but these are uncommon.^[11]

There are no large-scale studies or established guideline about the treatment of breast sarcoma because it is a rare disease. However, from several case reports and other soft tissue sarcoma except breast origin, we can consider some options for treatment according to stage, histologic grade, and tumor size. Multidisciplinary approaches involving surgeon, radiation, and medical oncologists are needed for treatment.^[12] Surgery remains the only curative modality and complete mastectomy is recommended because ensuring adequate resection margin is an important prognostic factor. As regional lymph node involvement is rare in breast sarcoma, prophylactic lymph node dissection is not indicated. In local recurred case, salvage mastectomy should be done. Recommended regimens of chemotherapy are vincristine, doxorubicin and cyclophosphamide (VAC), or vincristine, doxorubicin and ifosfamide (VAI).^[9] However, the role of chemotherapy for breast sarcoma is uncertain. Radiotherapy is also controversial and no benefit for improve disease-free survival. Only for high-grade or large tumors, it is suggested for reduce the rate of local recurrence. In this case, VAC/IE regimen was effective in tumor growth inhibition and patient survival.

The IRS has presented extent of the lesion, site of primary onset, histological type and age at onset as a prognostic factor based on clinical studies. In breast origin rhabdomyosarcoma, the majority of cases are alveolar type and the age at onset was over 10 years old. For this reason, the prognosis is still poor for alveolar rhabdomyosarcoma cases; 5-year survival rate 43%.^[7]

Compared with other breast lesions, breast sarcoma is characterized by unilateral, well-defined, large, painless, firm mass within the breast. However, most of the breast mass in young age is benign and rarely malignant; this leads to a delay in diagnosis and a worse prognosis. Physicians who deal with breast disease should be aware of breast sarcoma and this case may be helpful for understanding and management.

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