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Initial presentation and recurrence of metastatic rhabdomyosarcoma as breast mass

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Rhabdomyosarcoma rarely metastasizes to the breast. We report a case of a pediatric patient who initially presented with a right breast mass and pancytopenia, which was subsequently diagnosed as alveolar rhabdomyosarcoma. Despite initial favorable response to chemotherapy, a new metastatic focus was found in the contralateral breast 10 months later.

Introduction

In the pediatric and adolescent population, the overwhelming majority of breast masses are due to benign etiologies. When malignant, they tend to be metastatic disease from lymphoma, leukemia, and rhabdomyosarcoma. In both primary and metastatic cases of rhabdomyosarcoma to the breast, the histologic subtype is most commonly alveolar. Our case of metastatic alveolar rhabdomyosarcoma initially presented as a right breast mass and pancytopenia. This is an extremely rare presentation of rhabdomyosarcoma. Following an initial favorable response to chemotherapy, a new left breast metastasis was found on repeat PET/CT 10 months after diagnosis.

Case report

A 15-year-old female presented to urgent care with diffuse swelling of the right breast. She initially palpated a lump in her breast one week before presentation. The lump then quickly enlarged and became more firm, such that the right breast was noticeably larger than the left breast. The patient also had symptoms of an upper respiratory tract infection and two episodes of epistaxis. Physical exam was positive for a large firm mass involving most of the outer...
right breast. In addition, laboratory analysis was remarkable for pancytopenia (WBC 4, Hgb 10, Plt 56). A subsequent mammogram showed a unilateral dense right breast (Fig. 1). Breast ultrasound revealed a 5cm hypoechoic mass at 10 o’clock (not shown). The patient was started on Keflex for possible mastitis.

One week later, the patient did not notice any improvement in her right breast swelling. Repeat laboratory evaluation confirmed persistent pancytopenia. Due to concern for malignancy, she was admitted to the hospital and referred to surgical oncology. The working diagnosis was acute myelogenous leukemia with granulocytic sarcoma of the breast. Rhabdomyosarcoma was considered but was felt to be unlikely due to the presence of pancytopenia. Bone marrow biopsy was nonspecific; immunophenotyping for myeloid leukemia was negative. The patient then underwent a palpation-guided core biopsy of the breast mass. Pathology was consistent with alveolar rhabdomyosarcoma with a FOX01 (FKHR) gene rearrangement on chromosome 13q14.

On further questioning, the patient reported several weeks of right foot swelling and tenderness following minimal trauma. Subsequent foot radiographs demonstrated a healing second metatarsal fracture. She also complained of developing pain in her right thigh and knee. A chest radiograph was remarkable for intrathoracic lymphadenopathy and diffuse pulmonary opacities (Fig. 2). A PET scan revealed diffuse infiltration of the right breast with multiple hypermetabolic masses. Moreover, there was diffuse bone-marrow activity plus innumerable hypermetabolic pulmonary opacities, pathologically enlarged lymph nodes throughout the chest/abdomen/pelvis, a large right popliteal fossa mass, and multiple soft-tissue masses in the right foot. The largest and most hypermetabolic of these soft-tissue masses was located along the plantar surface at the level of the metatarsals (Figs. 3-4). The patient was diagnosed with stage 4 rhabdomyosarcoma, with the largest right foot mass presumed to be the primary tumor site.

The patient was started on chemotherapy with etoposide, ifosfamide, and mesna per COG protocol ARST08P1 Arm P2. She also received radiation therapy to the right foot, popliteal fossa, and hemipelvis. Ten months later, a PET/CT performed after 13 cycles of chemotherapy showed decreased activity in the previous areas of involvement. However, a new hypermetabolic focus in the lateral left breast was identified (Fig. 5). Ultrasound confirmed the presence of a 2.2cm left axillary tail mass with suspicious features (Fig. 6). Histopathology from subsequent core-needle biopsy was

Figure 1. 15-year-old female with metastatic rhabdomyosarcoma. Maximum intensity projection (MIP) image reveals extensive hypermetabolic activity within the right breast, lungs, axial and proximal appendicular skeleton, right foot, and lymph nodes of the chest, abdomen, pelvis, right inguinal region, and right popliteal fossa.

Figure 2. 15-year-old female with metastatic rhabdomyosarcoma. Frontal chest radiograph shows multiple findings including asymmetrically enlarged right breast, mediastinal lymphadenopathy, small bilateral pleural effusions, and pulmonary opacities.

Figure 3. 15-year-old female with metastatic rhabdomyosarcoma. Maximum intensity projection (MIP) image reveals extensive hypermetabolic activity within the right breast, lungs, axial and proximal appendicular skeleton, right foot, and lymph nodes of the chest, abdomen, pelvis, right inguinal region, and right popliteal fossa.

Figure 4. 15-year-old female with metastatic rhabdomyosarcoma. Fusion image of the chest better details hypermetabolic activity in a diffusely enlarged and dense right breast, extensive pulmonary involvement, and intrathoracic lymphadenopathy.
consistent with metastatic alveolar rhabdomyosarcoma (Figs. 7-8). The chemotherapy regimen was then altered to vincristine, irinotecan, and temozolomide. The left breast mass initially decreased in size. Unfortunately, the response was transient; followup ultrasound a month later revealed that it had grown to 4 cm. One week after the ultrasound study, the patient presented to the emergency department with dizziness and hypoesthesia on the right side of her body. MRI of the brain showed multiple intra-axial hemorrhagic metastases, the largest of which was in the left inferior parietal lobule (Fig. 9).

Discussion

In the pediatric population, breast malignancies are quite rare. The etiologies of breast enlargement and masses are most often benign. These include hormonal, infectious, or traumatic etiologies. In addition, benign tumors such as fibroadenomas outnumber malignancies. Unlike in adult women, primary breast carcinoma in children and adolescents is exceedingly rare, with phyllodes tumor being the most common primary malignancy. Most malignancies are metastases, commonly from neuroblastoma, rhabdomyosarcoma, or hematologic malignancies (1).

The most common soft-tissue sarcoma of childhood, rhabdomyosarcoma accounts for 5% of all childhood malignancies (2). These tumors arise from primitive muscle cells that fail to differentiate into normal skeletal muscle; they also occur in organs that do not contain skeletal muscle. Most cases arise from the head and neck, genitourinary organs, and extremities. The main subtypes of rhabdomyosarcoma are embryonal (with botryoid and spindle-cell variants) and alveolar. Other less common subtypes include...
Metastases to the breast are seen in 6% of patients with rhabdomyosarcoma (1). These are most commonly of the alveolar subtype. In one analysis that examined 19 cases of rhabdomyosarcoma with initial metastases to the breasts, all of those (18) in which the histology could be accurately determined were of the alveolar subtype. Both primary and metastatic breast rhabdomyosarcomas have been reported to most commonly occur in adolescent females or young adults, with an age range of 11.5 to 20.2 years and a median of 15.2 years (4). However, since 1997, other cases have been reported in adult women. More recently, Li et al reported five patients with either primary or secondary breast rhabdomyosarcomas whose mean age was 30 years (5).

Clinically, breast metastases often present as rapidly enlarging masses that may be tender. Their radiologic appearances are nonspecific. Although they can be bilateral and multiple, breast metastases more commonly initially present as a single mass. Mammographically, they can also (as in the case of our patient) manifest as diffusely increased density (1). The combination of pancytopenia and breast mass, seen with our patient, is an exceedingly rare presentation of rhabdomyosarcoma (6).

Due to the radiosensitivity of developing fibroglandular tissue in pediatric patients and their dense breasts, ultrasound is preferred over mammography as the initial imaging modality in this population. The sonographic appearance of a metastatic lesion is most commonly a solid mass with variable echogenicity. As in adults, breast masses in the pediatric population require thorough workup. Although the appearances of benign and malignant masses may overlap, those with suspicious features (for example, irregular margins, posterior shadowing, antiparallel orientation) warrant further evaluation with tissue sampling (7). However, the lack of a discrete mass on ultrasound does not negate biopsy, as metastatic rhabdomyosarcoma can sonographically mimic normal fibroglandular tissue (8).

To the best of our knowledge, there is a paucity of long-term survival data in the literature for rhabdomyosarcoma with initial metastases to the breasts. Hays et al reported a 5-year Kaplan-Meier estimated survival rate of 35% in patients with breast metastases from rhabdomyosarcoma. Nevertheless, the same study reported two patients with no evidence of active disease for more than 15 years (4).

Our case emphasizes the importance of recognizing that, although rare, malignancies do occur in the pediatric and adolescent breasts. In addition, metastatic disease is significantly more common than primary breast malignancies. In these young patients, especially those with known primary malignancies such as rhabdomyosarcoma or lymphoma/leukemia, a new palpable breast mass should be viewed with suspicion. Although benign entities occur with much greater frequency in children than adult females, a breast mass should warrant careful workup in all patients.

References

1. Chung EM, Cube R, Hall GJ, Gonzalez C, Stocker JT, Glassman LM. From the archives of the AFIP: breast masses in children and adolescents: Radiologic-pathologic correlation. Radiographics 2009;29(3):907-31. [PubMed]
2. McCarville MB, Spunt SL, Pappo AS. Rhabdomyosarcoma in pediatric patients: The good, the bad, and the unusual. AJR American Journal of Roentgenology. 2001;176(6):1563-9. [PubMed]
3. Parham DM, Ellison DA. Rhabdomyosarcomas in adults and children: An update. Archives of Pathology & Laboratory Medicine. 2006;130(10):1454-65. [PubMed]
4. Hays DM, Donaldson SS, Shimada H, et al. Primary and metastatic rhabdomyosarcoma in the breast: Neoplasms of adolescent females, a report from the Intergroup Rhabdomyosarcoma Study. Medical and Pediatric Oncology. 1997;29(3):181-9. [PubMed]
5. Li DL, Zhou RJ, Yang WT, et al. Rhabdomyosarcoma of the breast: A clinicopathologic study and review of
the literature. *Chinese Medical Journal.* 2012;125(14):2618-22. [PubMed]

6. Hayashi Y, Kikuchi F, Oka T, et al. Rhabdomyosarcoma with bone marrow metastasis simulating acute leukemia. Report of two cases. *Acta Pathologica Japonica.* 1988;38(6):789-98. [PubMed]

7. Weinstein SP, Conant EF, Orel SG, Zuckerman JA, Bellah R. Spectrum of US findings in pediatric and adolescent patients with palpable breast masses. *Radiographics.* 2000;20(6):1613-21. [PubMed]

8. Ahn SJ, Kim SK, Kim EK. Metastatic breast cancer from rhabdomyosarcoma mimicking normal breast parenchyma on sonography. *Journal of Ultrasound in Medicine.* 2010;29(3):489-92. [PubMed]