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IMAGING INSIGHTS SUGGESTING A SINISTER CAUSE OF A BREAST MASS IN ADOLESCENTS: A CASE OF RHABDOMYOSARCOMA IN A TEENAGER

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ABSTRACT

Background: Sarcomas of the breast are exceedingly rare, accounting for less than 1% of malignant breast tumors, with primary rhabdomyosarcomas being even rarer. Due to the scarcity of reported cases, the imaging characteristics of breast rhabdomyosarcoma are not well-defined, making diagnosis challenging, especially in adolescents.

Case description: We present the case of a 17-year-old female diagnosed with embryonal rhabdomyosarcoma following a comprehensive workup for right breast masses. Initial imaging showed no distant metastasis, and the patient underwent a right mastectomy followed by adjuvant chemoradiotherapy. A few months post-treatment, she developed recurrent nodules in the chest wall. Further investigation confirmed the recurrence of embryonal rhabdomyosarcoma.

Conclusions: This case underscores the importance of considering primary rhabdomyosarcoma as a differential diagnosis in adolescent breast lesions. Given its rare occurrence and potential imaging overlap with more common tumors like cystosarcoma phyllodes, awareness and careful evaluation are critical for accurate diagnosis and timely management.

KEYWORDS

Primary breast rhabdomyosarcoma, embryonal rhabdomyosarcoma, pediatric breast tumors

LEARNING POINTS

- *Critical imaging insights:* The report provides valuable imaging characteristics that can help differentiate rhabdomyosarcoma from more common breast tumors like fibroadenoma and cystosarcoma phyllodes, resulting in more accurate and timely diagnosis.

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INTRODUCTION

Rhabdomyosarcomas (RMS) are the most common soft tumor sarcomas of childhood. Their cells appear similar to skeletal muscle progenitor cells although they may originate from non-skeletal origins and constitute 3 to 4% of all the pediatric tumors[1]. The subtypes embryonal (ERMS) and alveolar (ARMS) comprise 70% and 20% of all the cases of RMS[2]. Other subtypes include spindle cell/ sclerosing and pleomorphic. With advances in cytogenetic testing, RMS subtypes are predominantly categorized by the presence or absence of fusion involving the *FOXO1* gene[3]. ERMS is associated with good prognosis while ARMS is associated with poor prognosis and increased incidence of metastasis. Common sites of origin of primary RMS are head and neck (40% of the cases), genitourinary system (22% of the cases), extremities (18% of the cases), chest wall, trunk, biliary tract, retroperitoneum and perineum[4]. Sarcomas of the breast account for less than 1% of malignant breast tumors and primary rhabdomyosarcomas of breast are extremely rare[5]. There is paucity of data regarding exact number of cases of rhabdomyosarcoma arising in the breast; only a few cases have so far been reported hence imaging characteristics have not been clearly described. Breast RMS may be confused with fibroadenoma which is the most common benign breast tumor in adolescent population and differentiation between the two is essential.

CASE DESCRIPTION

We report a 17- year-old female who presented to our radiology department with recurrent palpable nodules in the right chest wall. Her complaints started at the age of 15 when she developed rapidly enlarging lumps in her right breast

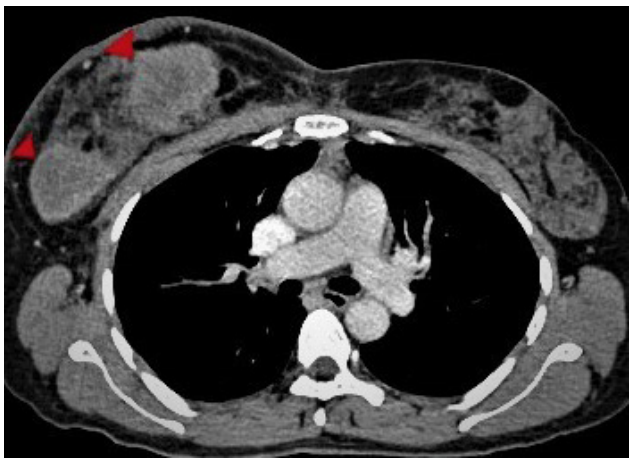


Figure 1. Contrast enhanced computed tomography scan of the chest: axial section showing two soft tissue density lobulated masses in the right breast (red arrow heads).

that were evaluated and diagnosed as embryonal RMS. Her initial work-up contrast-enhanced computed tomography (CECT) scan, which was done at an outside facility, showed multicentric soft tissue density enhancing masses in right breast *(Fig. 1)*. No distant metastasis was reported.

A complete work-up of the lesion confirmed primary ERMS of the right breast. The patient was advised to undergo mastectomy because of the multicentric nature of the disease however the patient delayed treatment and presented a few months later with a confluent large mass replacing the right breast almost in its entirety. Subsequently the patient underwent mastectomy. Images of the surgical specimen are shown in *Fig. 2*. Post mastectomy the patient was referred to oncology for adjuvant chemoradiotherapy.

After the completion of chemoradiotherapy the patient

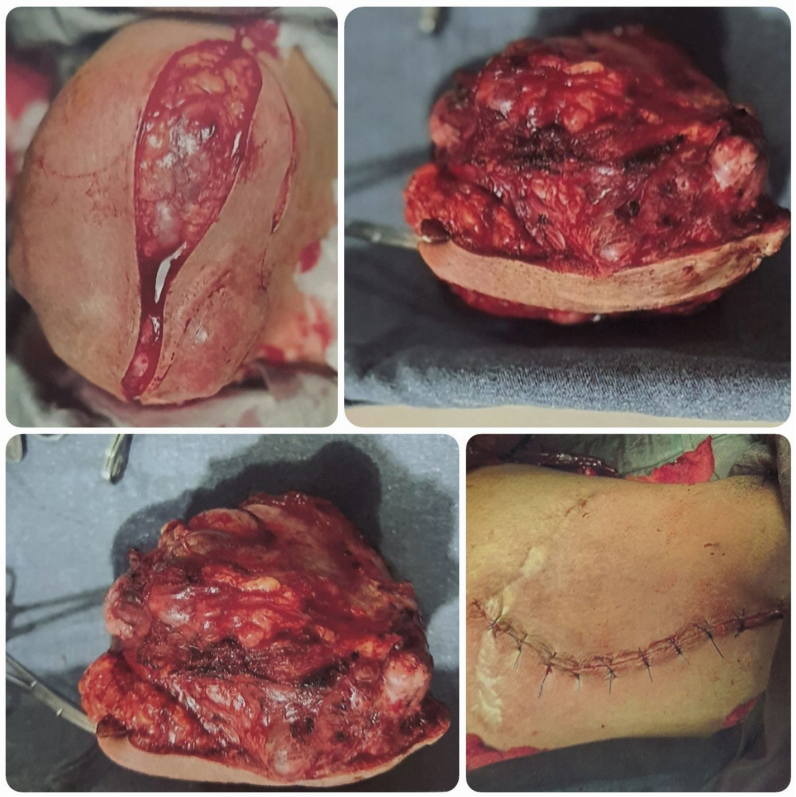


Figure 2. Right breast mastectomy specimen showing large mass replacing the right breast entirely.

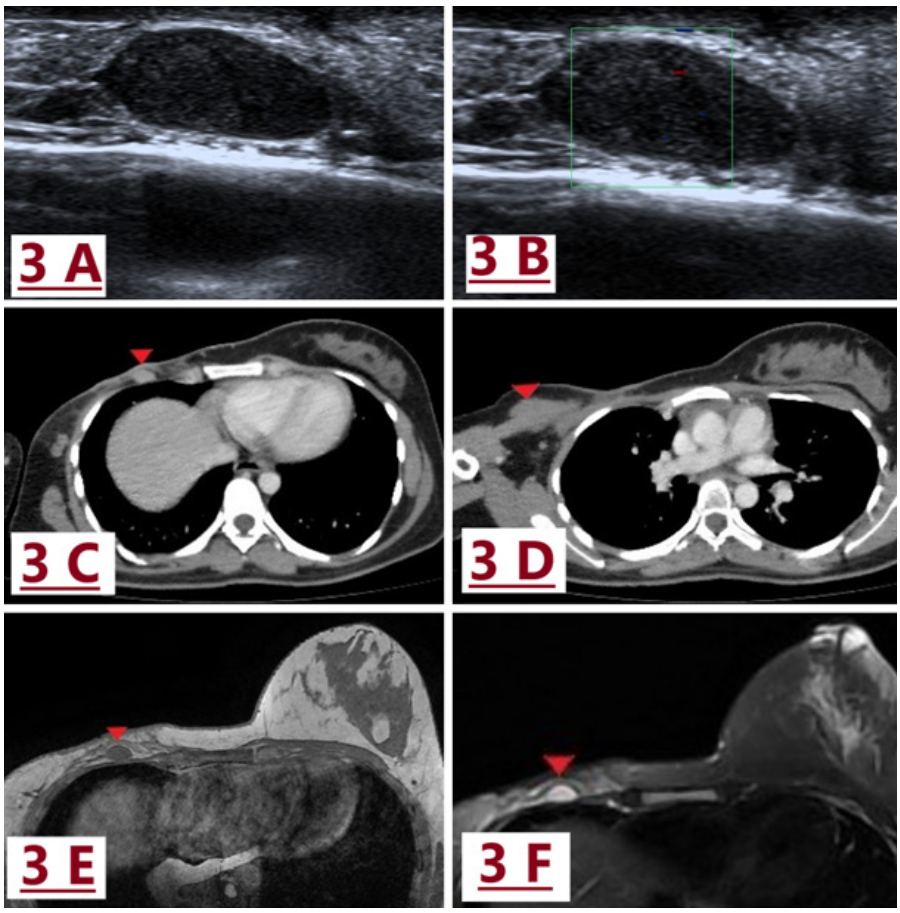


Figure 3. The grey scale ultrasound image (A) demonstrates a well-circumscribed nodule while color Doppler image (B) demonstrates intralesional flow. Figures C and D are CECT axial sections and E and F are axial T1 and axial T2 fat suppressed MRI images, respectively, showing the nodules (red arrow heads).

remained well for almost 10 months after which she felt nodules at the surgical bed. She presented to our radiology department for ultrasound that showed at least three well-circumscribed hypoechoic lesions centered at right chest wall (mastectomy bed) as shown in *Fig. 3A and 3B*. Contrast enhanced CT was subsequently done and confirmed the soft tissue density lesions at the mastectomy bed *(Fig. 3C and 3D)*. No distant metastasis was identified. Magnetic resonance imaging (MRI) was also done however patient refused to receive contrast hence a non-contrast study was performed. Three well circumscribed lesions were noted again, that returned T1 low and T2 hyperintense signals. One of the lesions is shown in *Fig 3E and 3F*.

Ultrasound guided core biopsy of one the recurrent lesions at the chest wall was performed for histopathological confirmation of recurrent disease. Histopathology confirmed the diagnosis of recurrent ERMS. Histological examination of sections revealed a malignant neoplasm arranged in nests and sheets of primitive rounded cells present around the blood vessels. Individual tumor cells showed abundant cytoplasm and hyperchromatic nuclei *(Fig. 4)*. Immunohistochemistry stained positive for myogenin *(Fig. 5A)* and desmin *(Fig. 5B)*. The patient was referred to the oncology clinic for further management with chemotherapy.

DISCUSSION

Cystosarcoma phyllodes is the most commonly observed primary breast malignancy in children[4]. The age of presentation of RMS is typically 5 years and in adolescence, while primary breast RMS is most often seen between the age of 10 and 19 years and alveolar is the most common subtype in these patients[4].

Clinical examination reveals a mobile non-tender lesion. Mammography is considered the primary imaging modality for the evaluation of breast pathologies. Since the breast tissue in adolescents displays high density on mammography and because of radiosensitivity of the breast, ultrasound and color Doppler are the radiological modalities of choice

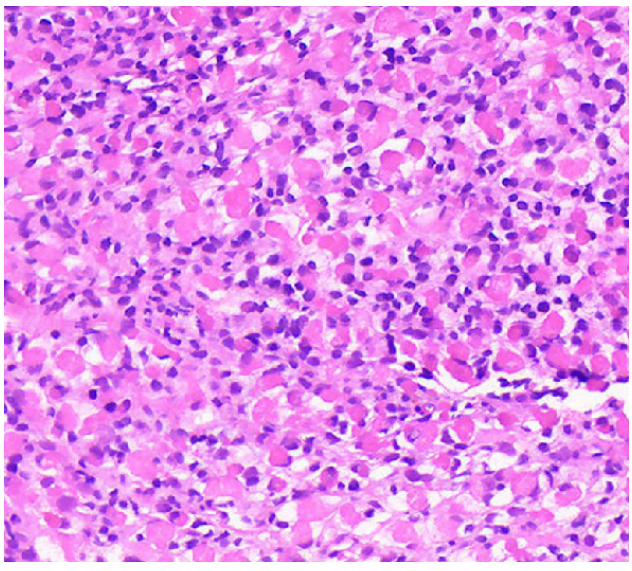


Figure 4. Hematoxylin and eosin-stained section demonstrates nests and sheets of primitive rounded cells with abundant cytoplasm and hyperchromatic nuclei.

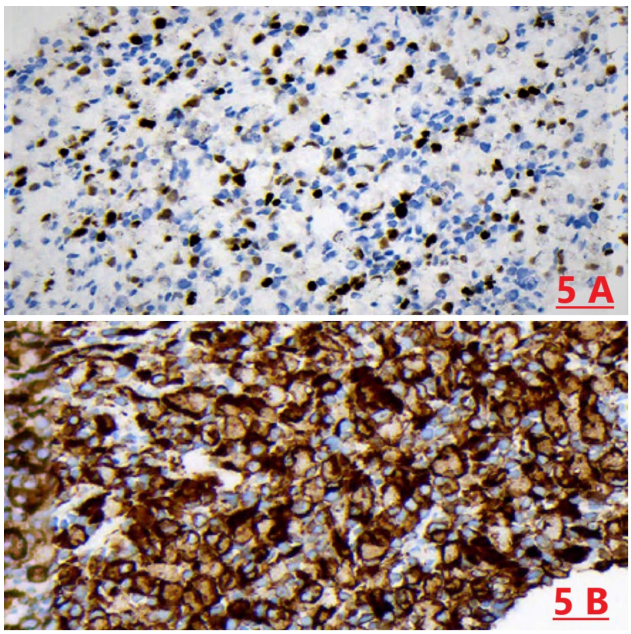


Figure 5. Immunohistochemistry stains positive for myogenin (A) and desmin (B).

for evaluation. In case of diagnostic ambiguities, contrast-enhanced MRI may be used as a problem-solving tool. Contrast-enhanced CT scan is primarily used to stage the malignant diseases. Most of the breast lesions encountered in adolescents are benign, however it is essential to keep in mind the uncommon pathologies like RMS. Increased breast vascularity on color Doppler ultrasound must raise a concern for primary or secondary neoplastic etiology. Documented sonographic features of RMS include a nodular lesion, lobulated well-circumscribed hypoechoic mass with irregular margins, diffuse nodular infiltration with no obvious normal breast parenchyma, intralesional heterogeneity, the long axis of the lesion perpendicular to overlying skin and bilateral diffuse involvement with lobulated infiltrative hypoechoic pattern[4]. Besides the aforementioned imaging patterns, sonographic imaging features of other malignant breast pathologies including echogenic peripheral rim of the lesion, posterior acoustic shadowing, satellite nodules and axillary lymphadenopathy can also be seen in RMS. MRI of RMS shows a hypointense to isointense signal on T1, hyperintense signal on T2 with diffuse post contrast enhancement and type III kinetic curves. There may be intralesional areas of necrosis. Intralesional hemorrhage may also be present that returns T1 hyperintense signal. The imaging features of RMS on various radiological modalities are summarized in *Table 1*. On follow-up, an intralesional increase in the necrotic component and a reduction in the degree of enhancement in comparison with the initial examination are useful criteria for evaluation of response to treatment. Early diagnosis improves the response to treatment and prognosis.

Histopathology of RMS reveals fusiform, polygonal or epithelioid cells which allows differentiation from other neoplastic etiologies like invasive lobular carcinoma, leukemia and lymphoma. Pathologic, immunohistochemical and molecular biology studies aid in the diagnosis of RMS with tumor cells of RMS diffusely positive for desmin, vimentin and alpha smooth muscle actin (SMA).

Treatment of primary breast RMS is via surgery, chemotherapy and radiotherapy with a range of duration of follow-up at 2 months to 7 years. Wide local excision with negative margins is usually sufficient however patients with local recurrence are treated with salvage mastectomy. Axillary clearance is

Imaging Modality	Imaging Features
Mammography	Not the first line modality for adolescents and young adults due to dense breast parenchyma
B-mode ultrasound	Variable appearance including: • Nodular lesion • Lobulated well-circumscribed hypoechoic mass with irregular margins • Diffuse nodular infiltration with no obvious normal breast parenchyma • Intralesional heterogeneity • The long axis of the lesion perpendicular to overlying skin and bilateral diffuse involvement with a lobulated infiltrative hypoechoic pattern • Echogenic peripheral rim of the lesion • Posterior acoustic shadowing • Satellite nodules • Axillary lymphadenopathy (rare)
Color Doppler	Intralesional increased vascularity that helps differentiate from benign breast pathologies
Contrast-enhanced MRI	• RMS returns T1 hypointense to isointense signal, T2 hyperintense signal with diffuse post contrast enhancement and type III kinetic curves • Intralesional areas of necrosis • Intralesional hemorrhage may also be present that returns T1 hyperintense signal
Contrast-enhanced CT Scan	Not very helpful in characterization of breast pathologies however this is the primary modality to stage the malignant disease process

Abbreviation: RMS, rhabdomyosarcoma; MRI, magnetic resonance imaging; CT, computed tomography.

Table 1. Summary of the imaging features of RMS on various radiological modalities.

not indicated unless there is palpable lymphadenopathy. Our patient received a combined modality treatment with neoadjuvant chemotherapy followed by mastectomy and radiotherapy. Unfortunately, recurrent disease was seen on follow-up ultrasound, MRI and CECT, therefore the patient was referred to the oncology department for treatment with chemotherapy.

CONCLUSION

Primary breast RMS is a rare entity that may be confused with fibroadenoma and cystosarcoma phyllodes on initial work-up. Therefore, a rapidly enlarging lesion with suspicious characteristics must be correlated with histopathology for definite characterization. Early diagnosis allows a better response to treatment and favorable prognosis. Treatment options include surgery, chemotherapy and radiotherapy.

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