



# Primary alveolar rhabdomyosarcoma of breast with bone marrow metastasis mimicking acute leukemia: a diagnostic challenge



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## Introduction:

Rhabdomyosarcoma (RMS) is the most common soft tissue sarcoma in children. RMS of the breast is most often metastatic from another site. Here, we describe one case of primary breast alveolar rhabdomyosarcoma with bone marrow metastasis mimicking an acute myeloid leukemia.

## Case Features:

- 15-year-old girl presents with a rapidly-enlarging right breast mass.

- Her CBC showed wbc: 4.4, hb: 10.5, plt: 56 and leukoerythroblastosis (left shifted granulocytes, rare blasts and occasional nucleated RBC).

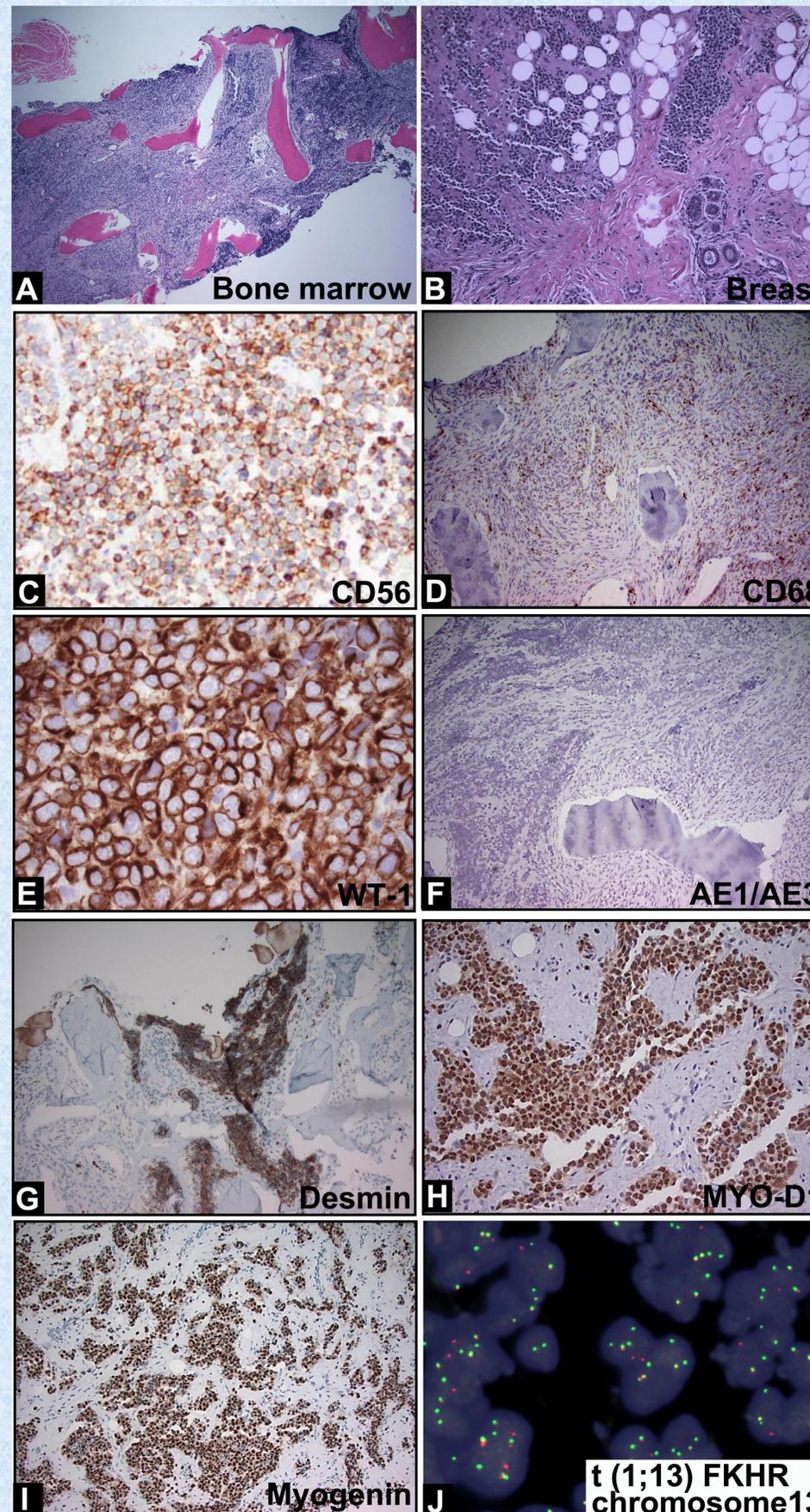
- The ultrasound and Mammogram showed an infiltration of right breast with 5.2 cm hypoechoic mass at the 10'clock position.

- The clinical differential diagnosis includes acute mastitis, hemorrhage/hematoma and myeloid sarcoma/AML.

- PET Scan showed diffuse bone marrow infiltrate with possible lung lesion and hot spot in the left foot.

## Pathology and Ancillar Tests:

Biopsy of the breast mass (B) revealed sheets of homogenous small round blue cell infiltrate positive for CD56, CD68 and cytoplasmic WT-1 (C-E), but negative for CD34, CD117, CD45,



MPO, S-100 and cytokeratin (F). The bone marrow biopsy (A) demonstrated sheets of PAS positive immature cells replacing the marrow space, and markedly diminished trilineage hematopoiesis. Therefore, an acute leukemia and possible myeloid sarcoma involving the breast was the main differential diagnosis. Further immunohistochemistry revealed cells positive for desmin, MyoD1, and myogenin (G-I).

## Molecular Properties:

FISH study confirmed the presence of a t (1;13) translocation involving the FKHR gene on chromosome 13 in breast lesion (J).

## Conclusions:

Overall, the findings are diagnostic for an alveolar RMS. Myelophthistic bone marrow metastasis, however, can mimic the clinical features of acute leukemia, which is diagnostically challenging. This case emphasizes the importance of judicious application of immunohistochemistry and molecular studies, and the expression of WT-1 and CD56 are useful markers to differentiate RMS from other small round blue cell tumors.

## References:

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