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SHORT REPORT

Alveolar rhabdomyosarcoma mimicking lymphoma with bone marrow involvement

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Abbreviations

CWS	German cooperative soft tissue sarcoma study
BFM	acute lymphatic leukemia study Berlin-Frankfurt-Muenster
FKHR	FKHR gene
LDH	lactate dehydrogenase
MRI	magnetic resonance imaging
NHL	non-Hodgkin lymphoma
PAX3/7	PAX gene
PET	positron emission tomogram

Short report

Rhabdomyosarcoma is the most common soft tissue sarcoma of childhood [1]. Two major subtypes, embryonal and alveolar rhabdomysoarcoma, can be distinguished by histology. Alveolar rhabdomyosarcoma are small, blue, round-cell tumors requiring aggressive oncologic treatment. Depending on the location and stage, they can mimic other neoplasms, thus making the diagnostic process challenging [4, 5, 7].

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We report on a 4-year-old boy who presented with an acute swelling at his right groin. The blood count was unremarkable, but the LDH was increased to 2,073 U/l. Bone marrow aspirate showed vacuolated blasts matching a L3-morphology (Fig. 1). MRI revealed a right-sided lymphatic inguinal mass and multiple enlarged abdominal lymph nodes. Because of the clinical and morphological picture, the diagnosis of NHL was assumed, and chemotherapy according to the NHL-BFM 95 protocol was started. Two days later, immunocytochemistry on the bone marrow cells became available: all B-cell markers were negative, but there was expression of cytokeratin, vimentin, α-actin, myoD1, and myogenin (Myf-4). Since the patient had already experienced an excellent clinical response with no lymph node enlargement for biopsy left, the decision was made to continue with chemotherapy for NHL. Four months after completion of chemotherapy, the patient presented again with a right-sided inguinal swelling. Inguinal lymph node biopsy revealed alveolar rhabdomyosarcoma. MRI of the leg and PET showed a suspicious 9-mm lesion in the right calf. Surgical exploration confirmed the presence of alveolar rhabdomyosarcoma, suggesting that this lesion was the primary tumor site. Lymph node metastases were observed extending from the right knee into the thorax. At this time, the bone marrow was not infiltrated. Rhabdomyosarcoma cells showed the PAX7-FKHR fusion transcript on RT-PCR. Retrospective analysis of the bone marrow from the initial presentation was also positive for this genetic aberration by RT-PCR, confirming the diagnosis of alveolar rhabdomyosarcoma with the occult primary site at the right calf, extensive lymph node and bone marrow infiltration. Chemotherapy according to the CWS-IV protocol was given, accompanied by 45-Gy radiation of the primary site. Although remission was

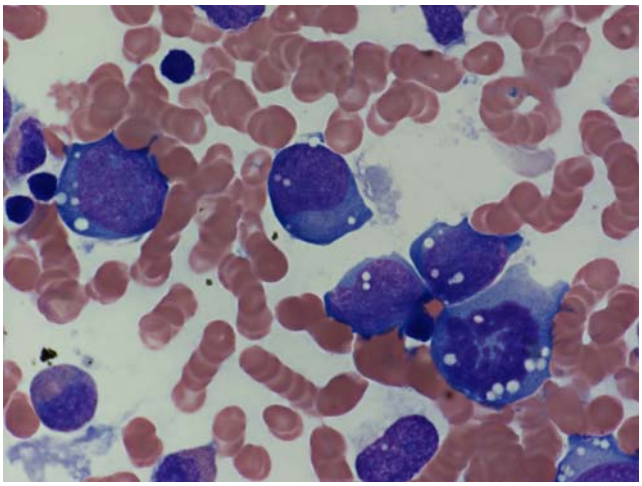

Fig. 1 Bone marrow aspirate: vacuolated blasts with dark basophile cytoplasm and large nucleoli matching a L3 morphology

achieved, 1 month after therapy and 27 months after the initial presentation, a second relapse was diagnosed.

Rhabdomyosarcoma with an occult primary site is known to be a very challenging diagnostic problem, especially if presenting as leukemia [2, 5]. Even rarer is the combination of bone marrow and superficial lymph node involvement perfectly mimicking the presenting picture of lymphoma. This case underlines the importance of awaiting results of immunophenotyping of tumor cells before starting therapy and performing molecular testing for RMS fusion transcripts. PAX7-FKHR fusion transcripts (t(1;13)) are found in about 20% of alveolar rhabdomyosarcoma and are associated with younger age and primary sites in the lower extremities, as seen in our patient [3, 6]. If metastatic disease is present, these patients may have a better outcome than patients with PAX3-FKHR fusion transcripts (t(2;13)) [6]. This possibly reflects differences in the pattern of metastatic disease as bone marrow involvement, which is associated with an extremely poor prognosis, has so far only been published in patients with PAX3-FKHR fusion transcripts. To our knowledge this is the first reported case of alveolar rhabdomyosarcoma with bone marrow involvement and PAX7-FKHR fusion.

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