Case Report

Rhabdomyosarcoma masquerading as acute leukemia with lymphoid phenotype expression: A diagnostic trap

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ABSTRACT

Rhabdomyosarcoma (RMS) can mimic acute leukemia both clinically and morphologically and can express a few surface markers characteristic of leukemic blasts. We report a case of a 21-year-old male patient who presented with pancytopenia. Bone marrow studies showed the atypical cells resembling blasts, expressing CD19 on flow cytometry and desmin by immunocytochemistry in bone marrow aspirate smears. A diagnosis of RMS infiltrating the bone marrow was made.

Key words: Acute leukemia, lymphoid phenotype expression, rhabdomyosarcoma

INTRODUCTION

Bone marrow infiltration by the nonhematopoietic malignancies has always been a source of misdiagnosis. Neoplastic cells especially of small round cell neoplasms resemble the blasts of acute leukemia.[1-3] Cases of rhabdomyosarcoma (RMS) presenting with disseminated disease with no obvious primary tumor always present a diagnostic problem. There are also reports of RMS confined to the bone marrow.[4] We herein present a case of RMS with bone marrow metastasis in the initial presentation itself thus clinically and morphologically mimicking an acute hematological malignancy.

CASE REPORT

A 21-year-old male patient had pain, edema and weakness of left leg of 2 weeks duration. He developed fever, urinary retention and consulted a local hospital. The routine investigations revealed pancytopenia. On examination, there was generalized lymphadenopathy and hepatosplenomegaly. Peripheral smear examination revealed leukoerythroblastic blood picture. The bone marrow aspirate was reported as high-grade malignancy possibly erythroleukemia and referred to our center. On examination, his general condition was poor. He had high-grade fever, severe pallor, generalized lymphadenopathy and bilateral pedal edema. Abdomen was distended, and a vague mass was palpable in the right suprapubic area. He had right sided pleural effusion, hematuria, and low urine output. Laboratory investigations revealed hemoglobin - 3.8 g%, total count - 8800/mm^3, platelet count - 8000/mm^3. Renal function tests revealed elevated urea and creatinine levels. Computerized tomography scan revealed soft tissue mass in the paraaortic and ischiorectal fossa inseparable from prostate. Peripheral smear revealed a leukoerythroblastic blood picture. Bone marrow aspirate revealed atypical cells including few plasmacytoid, binucleate and multinucleate cells arranged mainly singly and in focal clusters. Individual cells had scanty cytoplasm and round to oval nucleus with fine granular chromatin and inconspicuous nucleoli [Figure 1a]. Flow cytometry revealed the tumor cells positive only for CD19 [Figure 1b]. Other lymphoid and myeloid markers including CD56 were negative. Immunocytochemistry in bone marrow aspirate showed the positivity of tumor cells for desmin [Figure 1c]. Thus, diagnosis of RMS infiltrating bone marrow was given. He

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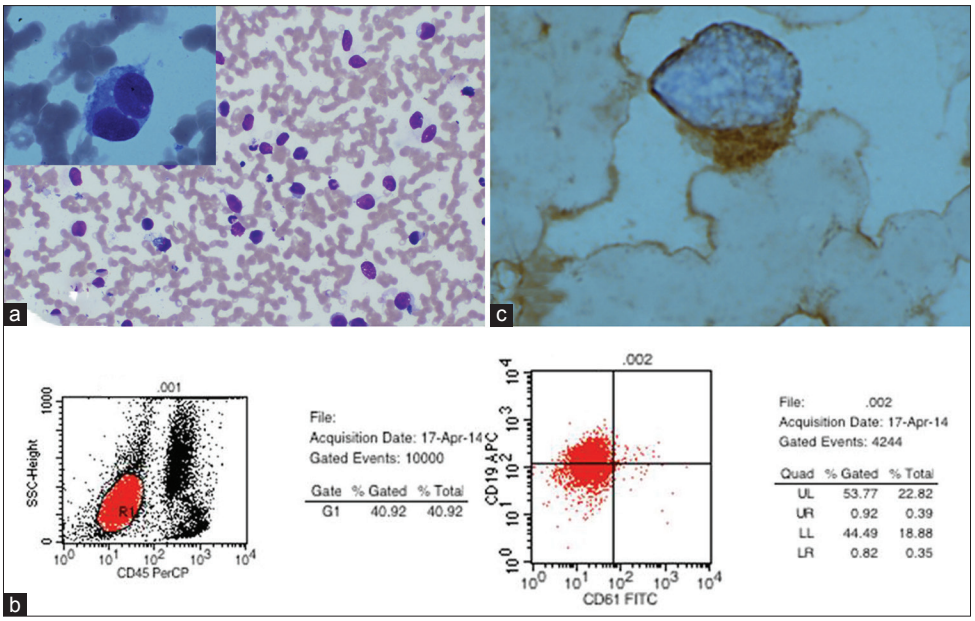

Figure 1: (a) Atypical cells arranged mainly singly and in small clusters (Giemsa, ×200). Inset showing the cell morphology (Giemsa, ×1000). (b) Tumor cells are positive for CD19 on flow cytometry. (c) Tumor cells are positive for desmin by immunocytochemistry (×1000)

was started on combination chemotherapy with vincristine, dactinomycin, cyclophosphamide and adriamycin. His hematuria subsided and blood counts improved. The patient is currently stable and is doing well.

DISCUSSION

Clinical and morphological resemblance of RMS and acute leukemia can result in misdiagnosis. RMS is the most common soft tissue sarcoma of children and adolescents commonly involving head and neck region, urogenital tract and extremities. Distant metastasis has been reported in fewer than 25% of the patients with lungs being the most common site followed by bone marrow.[5] RMS has a predilection for involving unusual sites like breast, testis, and subcutaneous tissue. Autopsy studies revealed pancreatic metastasis in around 67% of patients who died from metastatic RMS.[6] There are case reports of RMS in unusual locations such as upper thoracic spine with resultant fluctuating neurological status of the patient.[7] Cardiac RMS can present as severe mitral stenosis requiring emergency surgery and revealed as RMS only by histopathology.[8]

Bone marrow metastasis of RMS reveals a loosely arranged distribution of cells resembling blasts thus simulating acute leukemia. There are case reports of RMS misdiagnosed and treated as poorly differentiated leukemia based on morphology and cytochemistry alone and then proved as RMS based on demonstration of t(2,13) by cytogenetics study.[4] Nonhematolymphoid tumors presenting in leukemic phase, diffuse infiltration of bone marrow or inapparent primary site can be mistaken for hematolymphoid malignancies. Patients with widespread RMS can present with systemic symptoms, anemia, thrombocytopenia, disseminated intravascular coagulation, tumor lysis syndrome and thus clinically mimicking a hematologic malignancy.[9,10] RMS can also express B cell associated antigens like CD19, CD10, CD20 and immunoglobulins. It can also express CD56 and CD36 that are commonly used to assess hematologic malignancies.[11] Clustering of neoplastic cells is also seen in acute leukemias.[12]

CONCLUSION

Thus to conclude, we are presenting this case to emphasize the need to include RMS in the differential diagnosis in patients who present with clinical impression of acute leukemia with atypical features in bone marrow morphology and flow cytometry.

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