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CASE REPORT

Acute Myeloid Leukemia with Myelodysplasia - Related Changes after Isolated Myeloid Sarcoma

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SUMMARY

Background: **As a tumor mass, a myeloid sarcoma consists of myeloid blasts and presents at an anatomical site other than the bone marrow. In about one quarter of cases, myeloid sarcoma happens without an underlying acute myeloid leukemia or other myeloid neoplasm, and it may precede or coincide with AML or form acute blastic transformation of MDSs, MPNs, or MDS/MPNs.**
Methods: **Herein, we described a rare case of acute myeloid leukemia with myelodysplasia-related changes (AML-MRC), with WT1 mutation and high expression of TP53 after isolated myeloid sarcoma of lymph nodes showing a higher proportion of blasts, dysplasia of both megakaryocytes and granulocytes.**
Conclusions: **The case highlights the importance of a bone marrow examination, including morphology, immunophenotyping, cytogenetic, and molecular examination in all cases to exclude the possibility of myeloid sarcoma, especially the morphological feature of bone marrow dysplasia in the early stage before AML.**
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KEYWORDS

acute myeloid leukemia with myelodysplasia-related changes, myeloid sarcoma, dysplasia, WT1 mutation

INTRODUCTION

Myeloid sarcoma is defined as a tumor mass of myeloid blasts, which occurs at an anatomical site other than the bone marrow, in most cases concomitant with acute myeloid leukemia, rarely without bone marrow involvement. With a slight male predominance, myeloid sarcoma may occur at any site of the body and at any age. Organs most commonly involved including lymph nodes, genitals, skin, breast, gastrointestinal (GI) tract, peritoneum, bone, and central nervous system (CNS). Besides being preceded or coinciding with AML, myeloid sarcoma may constitute acute blastic transformation of preceding myelodysplastic, myelodysplastic/myeloproliferative, or myeloproliferative neoplasms [1]. In this rare case of AML-MRC, obvious dysplasia of megakaryocytes and granulocytes suggested that the patient may experience a long course of MDS. Due to the

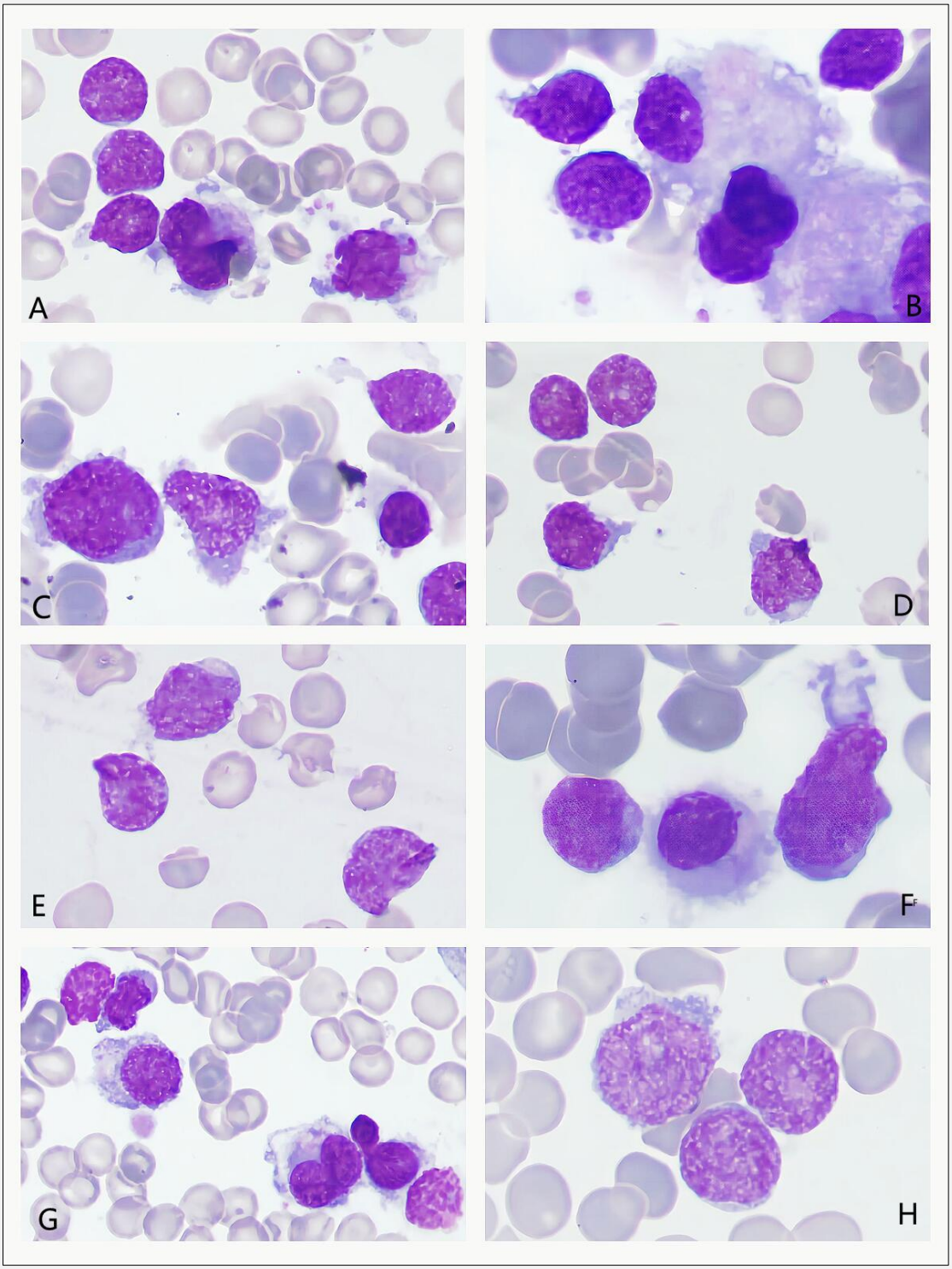

Figure 1. Bone marrow smears showed myeloid blasts 63% with a higher proportion of micromegakaryocytes and small megakaryocytes, indicating AML accompanied with obvious dysplasia of megakaryocytes.

rarity of myeloid sarcoma and difficulties in its diagnosis and treatment, the morphological features of dysplasia in bone marrow may be an important clue pointing towards the diagnosis of myeloid sarcoma, which should be paid attention to in the diagnosis to avoid misdiagnosis.

CASE REPORT

A 57-year-old man was admitted to another center with multiple masses in the neck, bilateral axilla, and bilateral groin. He was initially misdiagnosed with non-Hodgkin T-cell lymphoma based on a cervical lymph node biopsy a year ago. After comprehensive diagnosis, the diagnosis of myeloid sarcoma with high expression of TP53 was confirmed based on the morphological and immunohistochemical results, his bone marrow morphology and flow immunotyping were reported as normal, and 56 leukemia fusion genes were all negative. Complete remission was achieved after chemotherapy. The patient was admitted to our hematology department due to intermittent fever, weakness, and skin ecchymosis on the left upper limb. Color ultrasound showed no abnormal lymph nodes. The complete blood cell count of the patient indicated white blood cells 0.79 x 10^9/L, hemoglobin 88 g/L, and platelets 17 x 10^9/L. Blasts can be seen in the peripheral blood smear. Bone marrow smears showed myeloid blasts 63% with a higher proportion of micromegakaryocytes and small megakaryocyte, (Figure 1) admixed with hypogranular neutrophils with clumped nuclear chromatin. Peroxidase of the blasts was positive, indicating AML accompanied with obvious dysplasia of megakaryocytes and granulocytes. Flow-cytometric immunophenotyping of the bone marrow blasts showed partial positivity for c myeloperoxidase (cMPO) and positivity for CD33, CD13, CD36, CD64, CD14, CD117, CD11c, and HLA-DR and also negativity for cCD3, cCD79a, CD34, CD19, CD15, and CD16, which was consistent with the immunophenotype of AML. The results of morphology and immunophenotype supported the diagnosis of AML-MRC. The test of 56 leukemia genes showed that WT1 was positive. The patient was diagnosed with recurrent and progressive myeloid sarcoma with WT1 mutation and high expression of TP53. Due to the adverse prognostic factors, such as hypertension, hepatitis B, and diabetes, the patient had repeated symptoms of myelosuppression and infections during treatment with multiple chemotherapy regimens. The patient was in critical condition. After the symptoms slightly improved, he voluntarily asked to be discharged.

DISCUSSION

Myeloid sarcoma is a kind of distinct myeloid neoplasm defined as a rare tumor mass of myeloid or monocytic blasts or rarely erythroid or megakaryocytic blasts occurring in an extramedullary site other than the bone marrow [2]. In most cases, AML occurs simultaneously. Myeloid sarcomas may present with the form of AML, may precede or coincide with AML, or may represent a blast transformation of a preceding MDS, MPN, or MDS/MPN [3]. Almost any site in patients with myeloid sarcomas may be involved, but the most commonly involved sites are the skin, gastrointestinal tract, lymph nodes, soft tissue, and bone [4]. The diagnosis of myeloid sarcoma is difficult and easily misdiagnosed, especially when a history of AML is absent, myeloid sarcoma diagnosis can be challenging. When myeloid sarcoma occurs as an isolated mass, its histopathological diagnosis will be hard and confusing. It could easily be misdiagnosed as a malignant lymphoproliferative disorder, Ewing sarcoma, round blue cell tumors, thymoma, poorly differentiated carcinoma, or other rare hematopoietic tissue malignancy. Inadequate immunophenotyping of myeloid sarcoma commonly leads to misdiagnosis, which will not be corrected until an AML diagnosis is later established by bone marrow examination [5]. In the case we report here, obvious dysplasia of megakaryocytes and granulocytes suggested that the patient may experience a long course of MDS. Therefore, the morphological features of dysplasia in bone marrow may be an important clue pointing towards the diagnosis of myeloid sarcoma, which should be taken into account in diagnosis and differential diagnosis of myeloid sarcoma.

This case focuses on the importance of a bone marrow evaluation, including morphology, immunophenotyping, cytogenetic and molecular examination in all the cases to exclude the possibility of myeloid sarcoma, especially the morphological feature of bone marrow dysplasia in the early stage before AML.

Declaration of Interest:
The authors declare no competing interests.

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