Myeloid sarcoma with acute myeloid leukaemia: a case report of rare presentation

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Abstract

Myeloid sarcoma (MS) is a rare extramedullary manifestation of acute myeloblastic leukaemia (AML). The presentation of MS depends on the anatomical location being affected. Myeloid sarcoma can also occur in the absence of AML, but when they occur simultaneously, they are often associated with a poorer prognosis. We report the case of an adolescent patient of AML with MS of cervical lymph nodes presenting with vague and non-specific complaint of abdominal pain for two months with history of a non-specific and non-healing skin lesion on the right lower extremity. Detailed history and physical examination were a major help. Bone marrow biopsy and cervical lymph node biopsy were performed to make the definitive diagnosis. Chemotherapy and supportive care were administered. Since such cases are rarely encountered, they tend to be misdiagnosed. Reporting cases with such variable presentations can help physicians not to miss such important differential diagnoses in their evaluation.

Keywords: Acute myeloblastic leukaemia, Acute myeloid sarcoma, Cervical lymphadenopathy.

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Introduction

Myeloid sarcoma is a rare extramedullary proliferation of immature myeloid cells as a solid tumour, occurring in any anatomical location. It mostly (2-9% of the times) occurs as the natural course of acute myeloblastic leukaemia (AML).[1] The pathophysiology of AML involves accumulation of immature cells of myeloid lineage in the blood. MS presents with a variable clinical presentation depending on the anatomical location, thus creating diagnostic dilemma for the clinicians. The importance of early detection of MS and early initiation of chemotherapy cannot be adequately emphasised. We present a case of myeloid sarcoma of cervical lymph nodes with AML in an adolescent male manifesting as rare and non-specific symptoms.

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Case report

A 16-year-old male presented in the out-patient department (OPD) of Jinnah Hospital, Lahore, on June 14, 2024, with the complaint of mild but continuous left upper quadrant (LUQ) pain for two months. The patient denied having diarrhoea, haematemesis, or any association with food intake. There was also history of a non-specific skin lesion on the dorsum of the right foot one year ago that took around two months to heal. Upon detailed history taking, the patient stated that he had also experienced mild intermittent fever, night sweats, and fatigue on and off for the past two months.

On physical examination, right-sided cervical lymphadenopathy, splenomegaly, and mild anaemia were noted. The rest of the physical examination was unremarkable.

The complete blood count (CBC) showed elevated white blood cells (WBCs) at 147.82 × 10^9/L [Normal (N): 4.5 - 11.0 × 10^9/L], haemoglobin (Hb) was 9.2 g/dL (N; male: 130 - 180g/L, female: 3.5 - 5.5 x 1012/L), and platelets were 136 × 10^9/L (N: 140 - 400 x10^9/L) with differentials showing increased number of blasts. After the abnormal CBC, the patient was admitted to the oncology ward of Jinnah Hospital Lahore. The haematopathologic analysis of the bone marrow aspirate taken from the right posterior superior iliac crest showed marked infiltration of atypical WBCs (Figure 1) with the following differential leukocyte count (DLC): myelocytes 1% (N: 5% - 7%), metamyelocytes 2% (N: 8% - 20%), neutrophils 4% (N: 40% - 60%), lymphocytes 5% (11% - 23%), atypical cells 76% (N: none), monocytoid cells 12% (N: 0% - 0.8%), and nucleated red blood cells 5/100 WBCs (N: 15 to 30 per 100 cells). The atypical cells were medium- to large-sized with a moderate nuclear-to-cytoplasmic (N:C) ratio,

Table-1: Serial Complete Blood Counts (CBCs) following the start of chemotherapy.

Days after commencing Chemotherapy	1st	3rd	5th	Reference Range
WBCs	204.1x10^9/L	8.5x10^9/L	1.6x10^9/L	4.5 - 11.0 × 10^9/L
LYM	36.9x10^9/L	1.8x10^9/L	0.7x10^9/L	1 - 4.8 × 109/L
MID	57.1x10^9/L	2.0x10^9/L	0.20x10^9/L	Variable
GRAN	*******	4.7x10^9/L	0.7x10^9/L	1.5 - 8.5 x 10^9/L
RBC	2.5x10^12/L	2.91x10^12/L	2.79x1010^12/L	M: 4.3 - 5.9 x 1012/L F: 3.5 - 5.5 x 1012/L
HGB	8.1g/dL	7.1g/dL	7.0g/dL	M: 130 - 180g/L F: 115 - 165 g/L
PLT	148x10^9/L	42x10^9/L	29x10^9/L	140 - 400 x10^9/L)

WBC – White Blood Cell, LYM – Lymphocyte, MID – Middle Cell Absolute Count, GRAN – Granulocytes, RBC – Red Blood Cell, HGB – Haemoglobin, PLT – Platelets, F – Female, M – Male.

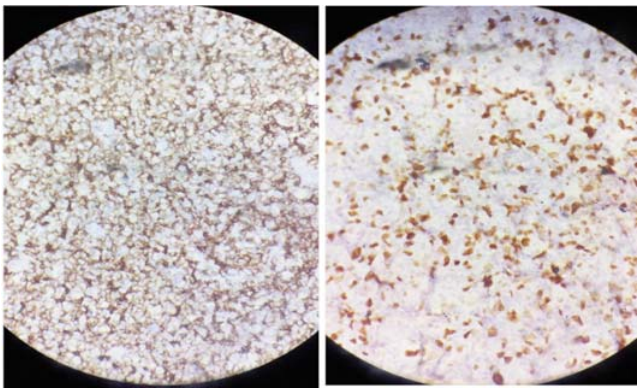

Figure-1: Photomicrograph of bone marrow aspirate taken from right posterior iliac crest of the patient showing extensive infiltration by atypical cells (100X).

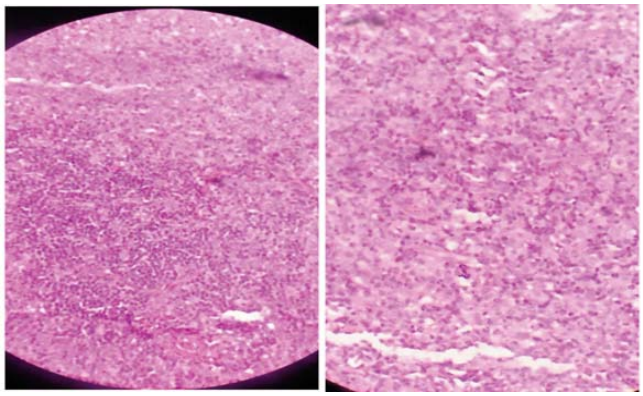

Figure-2: H & E-stained section of cervical lymph node biopsy showing partially effaced architecture. Inter-follicular area is expanded by sheets of large atypical cells with scant cytoplasm, round to oval indented nuclei with vesicular chromatin and inconspicuous nucleoli (100X).

homogeneous nuclear chromatin, irregular nuclear contours, indistinct nucleoli, and abundant agranular cytoplasm. RBCs showed microcytosis, hypochromia, and a few spherocytes (transfused cells). Flow cytometry and immunophenotyping of the lysed bone marrow sample showed cells positive for myeloid markers, i.e. myeloperoxidase (MPO), cluster of differentiation (CD) 13, and CD33, along with human leukocyte antigen-DR (HLA-DR). They also showed positivity for CD4, CD34, and CD117 (62%, N: 2% to 4%), which is consistent with AML.

The cervical lymph node biopsy showed atypical cellular infiltrate (Figure 2). Immunohistochemical staining revealed CD117, MPO, leukocyte common antigen (LCA), and terminal deoxynucleotidyl transferase (TdT) positivity in tumour cells. The Ki-67 staining indicated a 70% (N: 10%-30%) proliferation index, consistent with MS.

After the diagnosis was confirmed, treatment was started with the primary aim to cure the patient and to immediately begin cytoreduction therapy, that is, to reduce the number of abnormal cells in the patient's blood using medication. The induction regimen comprised Daunorubicin and Cytarabine which resulted in profound neutropenia with three days of chemotherapy and it was hence stopped. The patient's response to therapy can be observed in Table 1. The patient developed typhlitis about 5-6 days after initiation of therapy, so he was managed on those lines besides neutropenic prophylaxis and treatment.

Despite broad-spectrum antibiotic cover, he developed pneumonia simultaneously with typhlitis. Medications were modified along with supportive care as per patient's changing needs. Despite best efforts to maintain his oxygen saturation, he became tachypnoeic, drowsy, and lost consciousness. He could not be shifted to intensive care unit due to unavailability of bed at that time. Unfortunately, the patient couldn't recover and expired shortly afterwards.

Discussion

Myeloid sarcoma, also known as chloroma (because of the green colour due to MPO) or granulocytic sarcoma, is a tumour of myeloblasts in tissues other than bone marrow or blood.[2,3] AML with MS is associated with a low complete remission rate and a high induction death rate.[4] Its clinically variable signs and symptoms, depending on which part of the body has being invaded, make it difficult for clinicians to detect it, especially when the constitutional symptoms of malignancy are not so prominent. According to a review study, the most common part affected by MS was head and neck region (44.4%), followed by anterior mediastinum (22.2%) and gastrointestinal tract (22.2%).[5] In the current case, the onset of symptoms was very non-specific. It highlights the significance of identifying the insidious onset of this malignancy.

Physical examination and ultrasound revealed no abnormality other than splenomegaly, which was likely the cause of abdominal pain. Splenomegaly may or may not be present in myeloid neoplasms.[6] The differential diagnoses can be extensive for myeloid sarcoma of the head and neck region, making misdiagnosis common. In a historical study, the rate of misdiagnosing MS was observed to be 75%, with large-cell B lymphoma being the most commonly mistaken diagnosis.[7] The novel techniques of using specific tumour markers and flow cytometry studies have lowered this percentage. In the present case, the diagnosis of MS was confirmed with immature blasts in the cervical lymph nodes showing positivity for MPO, CD117, CD33, TdT, LCA, and Ki-67, thus excluding the lymphoid origin. The diagnosis of AML was made according to the World Health Organisation classification of myeloid neoplasms.[8]

The recommended treatment for MS is the same as that for AML with or without its co-existence. Neiman et al. demonstrated the importance of systemic chemotherapy for preventing progression of MS to AML in non-leukaemic patients. About 81% of the patients treated with surgical resection or radiotherapy progressed to AML within 11 months. On the other hand, 58% of the patients treated with chemotherapy remained non-leukaemic for 11 months.[9] Similarly, Meis et al. reported delay in development of AML in patients of MS with 25% remaining non-leukaemic for 3.5-16 years of follow-up post-diagnosis.[7] In the present case, with co-instancing MS and AML, the treatment plan was aimed at immediate cytoreduction followed by clearance of blasts and cure. However, due to sudden dip in his counts he was not given the recommended 7+3 regimen of Cytarabine and Daunorubicin. Instead, a 2+3 regimen was employed to prevent the patient from deteriorating further. Still, the patient expired due to neutropenic sepsis despite adequate antibiotic cover.

Studies have shown that patients treated with induction therapy and in some cases with high risk disease, stem cell transplantation (SCT) have a better survival rate compared to those treated with chemotherapy alone for AML with MS.[5] But, SCT is majorly considered mostly in case of relapse, which usually occurs after a median period of seven months.[3] Early diagnosis through biopsy and prompt initiation of chemotherapy are associated with a better prognosis.[10] In the present case, earlier diagnosis could have potentially led to a more effective treatment plan and better prognosis.

Conclusion

The variable clinical presentation and rare occurrence of myeloid sarcoma makes it challenging to diagnose. This case report highlights how subtly it can present. Therefore, emphasising the importance of early diagnosis and treatment is crucial for improving its prognosis.

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References

1. Phatharacharukul P, Fayad N, Siwiec R. Colonic Myeloid Sarcoma as a Rare Presentation of Relapsed Acute Myeloid Leukaemia. ACG Case Rep J 2020;7:e00407. doi: 10.14309/crj.0000000000000407.
2. Rochate D, Pavão C, Amaral R, Viveiros C, Cabeçadas J, Carneiro V, et al. Extramedullary Acute Leukemia—Still an Unforeseen Presentation. Hematol. Rep 2022;14:143-8. doi: 10.3390/hematolrep14020021.
3. Magdy M, Abdel Karim N, Eldessouki I, Gaber O, Rahouma M, Ghareeb M. Myeloid Sarcoma. Oncol Res Treat 2019;42:224-9. doi: 10.1159/000497210.
4. Xu LH, Wang Y, Chen ZY, Fang JP. Myeloid sarcoma is associated with poor clinical outcome in pediatric patients with acute myeloid leukemia. J Cancer Res Clin Oncol 2020;146:1011-20. doi: 10.1007/s00432-020-03128-7.
5. Lee JY, Chung H, Cho H, Jang JE, Kim Y, Kim SJ, et al. Clinical characteristics and treatment outcomes of isolated myeloid sarcoma without bone marrow involvement: a single-institution experience. Blood Res 2017;52:184-92. doi: 10.5045/br.2017.52.3.184.
6. Prakash S, Padilla O, Tam W. Myeloid, mast cell, histiocytic and dendritic cell neoplasms and proliferations involving the spleen. Semin Diagn Pathol 2021;38:144-53. doi: 10.1053/j.semdp.2020.07.003.
7. Meis JM, Butler JJ, Osborne BM, Manning JT. Granulocytic sarcoma in non-leukaemic patients. Cancer 1986;58:2697–709. doi:.10.1002/1097-0142(19861215)58:12<2697::AID-CNCR2820581225>3.0.CO;2-R
8. Arber DA, Orazi A, Hasserjian R, Thiele J, Borowitz MJ, Le Beau MM, et al. The 2016 revision to the World Health Organization classification of myeloid neoplasms and acute leukemia. Blood 2016;127:2391-405. doi: 10.1182/blood-2016-03-643544.
9. Neiman RS, Barcos M, Berard C, Bonner H, Mann R, Rydell RE, et al. Granulocytic sarcoma: a clinicopathologic study of 61 biopsied cases. Cancer 1981;48:1426-37. doi: 10.1002/1097-0142(19810915)48:6<1426::aid-cncr2820480626>3.0.co;2-g.
10. Lan TY, Lin DT, Tien HF, Yang RS, Chen CY, Wu K. Prognostic factors of treatment outcomes in patients with granulocytic sarcoma. Acta Haematol 2009;122:238-46. doi: 10.1159/000253592.

AUTHOR'S CONTRIBUTION:
AJ: Concept, design, data acquisition, analysis, interpretation and agreement to be accountable for all aspects of the work.
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HS: Drafting and revision.
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