Case Report

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Rare Presentation of Diffuse Osteosclerosis in Acute Myeloid Leukaemia

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Abstract:

Diffuse osteosclerosis is a rare complication of hematological malignancies. While primary myelofibrosis and acute megakaryocytic leukemia are known associations, acute myeloid leukemia (AML) is rarely reported. We present a case of a 45-year-old male with disabling backache due to diffuse osteosclerosis and an underlying diagnosis of AML.

Key words: diffuse osteosclerosis, AML, rare disease

Introduction

Acute myeloid leukaemia (AML) is a haematological malignancy characterized by the proliferation and infiltration of immature myeloid cells in the bone marrow and peripheral blood. The clinical presentation of AML can vary widely, and bone involvement in AML is rare, with only a few cases reported in the literature

Case Presentation

A 45-year-old male with a history of Type 2 Diabetes mellitus for 4 years with good compliance presented with lower back pain for a week and pain at the large joints for past 3 months. He also reported low-grade fever, weight loss, reduced appetite, letharginess, exertional tiredness and a history of recurrent upper respiratory tract infections. Physical examination revealed pallor, multiple sub-centimeter submandibular and inguinal lymphadenopathy without organomegaly. Initial blood investigations revealed pancytopenia, which later progressed to leukocytosis with suppression of other cell lines, and elevated inflammatory markers, Including Erythrocyte sedimentation rate and C-reactive protein. Ultrasound scan of the abdomen revealed moderate splenomegaly. X-ray spine, skull, and Non-contrast computed tomography (NCCT)-whole spine and CECT CAP (Contrast enhanced computed tomography of chest, abdomen, pelvis) revealed diffuse bone sclerosis.

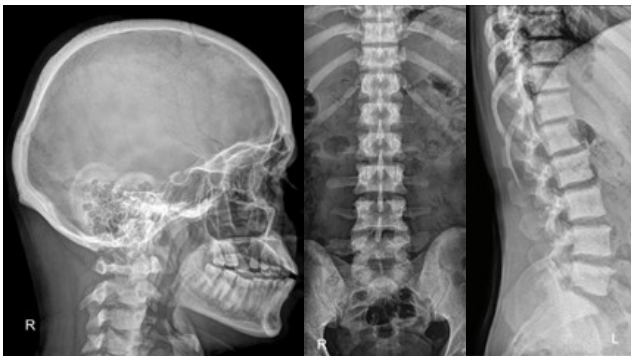

Figure 1: X-rays showing diffuse sclerosis of bones.

Initial bone marrow biopsy was a dry tap but repeat biopsy revealed that, around 50% of the marrow nucleated cells are small to medium sized blasts with high nuclear cytoplasmic ratio, diffuse nuclear chromatin pattern and distinct nucleoli and immunophenotyping confirmed the diagnosis of acute myeloid leukaemia (AML) with a rare presentation of diffuse osteomyelosclerosis.

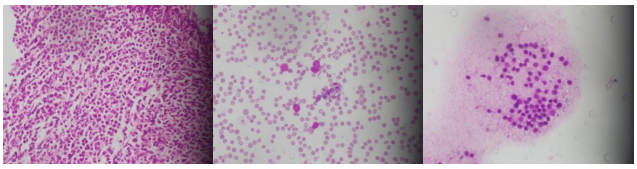

Figure 2:Bone marrow biopsy

He was started on induction chemotherapy. With improvement in blood counts, planned for consolidation chemotherapy. Later he developed a relapse of AML and neutropenia and succumbed to illness after 1 year of diagnosis.

Discussion

Diffuse osteosclerosis is a rare presentation in AML and can pose a diagnostic challenge. It is characterized by increased bone density due to abnormal accumulation of immature myeloid cells in the bone marrow, leading to decreased bone marrow space and increased bone density on imaging studies. This can manifest as diffuse

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sclerosis involving the spine, pelvis, and other skeletal sites.

The exact mechanism underlying diffuse osteosclerosis in AML is not well understood. It is believed to result from the infiltration of leukemic blasts into the bone marrow, leading to the disruption of normal bone remodeling and formation. The clinical presentation of diffuse osteosclerosis in AML can vary and may include bone pain, tenderness, and pathological fractures or non-specific symptoms such as back pain, fever, weight loss, and fatigue.

Bone marrow biopsy is the gold standard for diagnosing AML, but in cases of diffuse osteosclerosis, it can be challenging to obtain an adequate sample due to the decreased bone marrow space. In our case, the initial bone marrow biopsy resulted in a dry tap, but repeat biopsy after three weeks revealed the diagnosis of AML.

In cases where osteosclerosis or myelofibrosis is prominent, it may be difficult to aspirate liquid marrow or obtain a trephine core biopsy of the bone marrow. Osteosclerosis is not infrequently associated with hematologic malignancy and can be seen in certain myeloid and lymphoid neoplasms including, but not limited to, chronic myelogenous leukemia, primary myelofibrosis, acute megakaryoblastic leukemia, and hairy cell leukemia(1). It may also be seen in some myelodysplastic syndromes and have a worse prognostic outcome (6)

Management of diffuse osteosclerosis in AML involves treating the underlying leukaemia with chemotherapy and in some cases, bone marrow transplantation.

Conclusion

Diffuse osteosclerosis is a rare presentation in AML and can pose a diagnostic challenge. Our case stresses the importance of including AML as a differential diagnosis for diffuse osteosclerosis and underscores the importance of considering underlying hematological disease in pancytopenia and highlights the pathophysiological complexity of systemic osteosclerosis.Further research is needed for better understanding of the pathogenesis and optimal management of diffuse osteosclerosis in AML.

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