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WHEN BLAST CELLS MEET AUTOANTIBODIES: A RARE CASE OF MIXED PHENOTYPE ACUTE LEUKEMIA IN SYSTEMIC LUPUS ERYTHEMATOSUS

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INTRODUCTION

Mixed phenotype acute leukemia (MPAL) is a rare type of leukemia where leukemic blasts express features of both acute lymphoblastic leukemia (ALL) and acute myeloid leukemia (AML). Up to date, its association with Systemic Lupus Erythematosus (SLE) is unknown.

CASE REPORT

We report a case of 29-year-old lady diagnosed in 2020 with Systemic Lupus Erythematosus and secondary Anti-phospholipid syndrome (APS). Her initial presentations were autoimmune hemolytic anemia (AIHA) and unprovoked deep vein thrombosis. Immunological findings revealed positive anti nuclear antibody, elevated anti dsDNA antibody, hypocomplementemia, lupus anticoagulant and anti cardiolipin antibodies. Her disease was complicated by a macerated stillbirth due to placental insufficiency and a right lacunar infarction. She was treated with steroids, hydroxychloroquine and lifelong warfarin. A second flare of AIHA in May 2023 led to the addition of Azathioprine.

After a year of remission, she presented in November 2024 with fever, sore throat, odynophagia and submandibular swelling. Blood counts revealed leucocytosis and bicytopenia. Bone marrow aspiration confirmed acute myeloid leukemia with 80 % blast (Figure 1) and computed tomography (CT) neck suggested presence of deep abscess.

Antibiotics and cytoreduction therapy were started. Peripheral blood immunophenotyping confirmed MPAL and GMALL induction was commenced. Shortly after, she developed left eye proptosis and ophthalmoplegia. Contrast enhanced CT brain showed presence of cerebral dural venous sinus thrombosis (Figure 2). Despite anticoagulant and supportive care, she deteriorated with massive cerebral edema and succumbed to complications of her illness.

CONCLUSION

The aim of this case report is to highlight the extremely rare hematological malignancy in SLE patients which need to be identified early and treated aggressively as it is a complex disease with a poor survival rate.

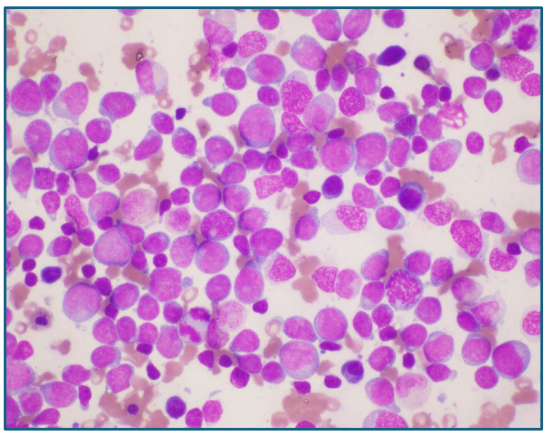

Figure 1 shows the presence of moderate to large blast cells with granular cytoplasm, cytoplasmic projection, irregular nuclear outline, open chromatin pattern with prominent nucleoli.

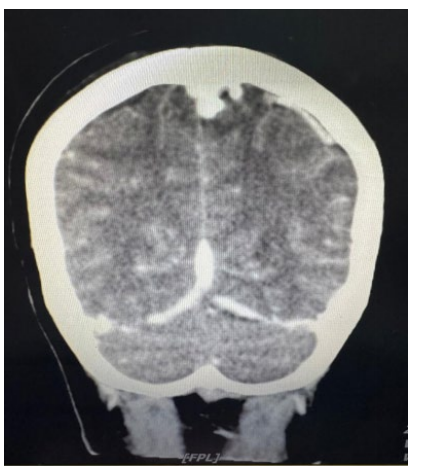

Figure 2 demonstrates filling defect over bilateral transverse sinus consistent with cerebral dural venous sinus thrombosis.

DISCUSSION

Mixed phenotype acute leukemia (MPAL) is characterized as biphenotypic or bilineal blasts which represents 1–3% of acute adult leukemias.[1]
The pathophysiology of acute leukemia in systemic lupus erythematosus is unclear but exposure to immunosuppressive therapy may increase risk of hematological malignancy.[2]
Until now, the prognosis of MPAL is poor but there is a trend towards improved survival rate in recent years, due to improved adoption of ALL-like regimens, the availability of targeted therapies and allogeneic hematopoietic stem cell transplantation.[3]

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