

Hemelmage

VEXAS Syndrome

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An 83-year-old man presented with macrocytic anaemia, vasculitis-appearing skin rash, Sweet syndrome and unprovoked subclavian deep venous thrombosis (DVT). A complete blood count showed Hgb 102 g/L, MCV 117 fL, WBC $3.6 \times 10^9/L$, neutrophil $2.5 \times 10^9/L$, and platelet $160 \times 10^9/L$. Bone marrow aspirate showed cytoplasmic vacuoles in myeloid and erythroid precursor cells. There was no excess of blasts. Some dysplastic neutrophils with abnormal nuclear lobation including hypolobation were present. These morphological findings raised the diagnostic consideration of vacuoles, E1 enzyme, X-linked, autoinflammatory, and somatic (VEXAS) syndrome. Next generation sequencing (NGS) testing identified pathogenic variant *UBA1* p.Met41Val [variant allele frequency (VAF) 72%], confirming the diagnosis of VEXAS syndrome. Trephine biopsy sample was hypercellular with no evidence of increased blasts, lymphoma or plasma cell neoplasm. The patient was treated with rivaroxaban, prednisolone and azacytidine with resolution of symptoms and full recovery of blood count.

VEXAS syndrome is an adult-onset autoinflammatory disease. It is caused by somatic mutation in *UBA1* gene [1]. This case highlights the importance of identifying VEXAS syndrome-related bone marrow morphology and testing for *UBA1* mutation.

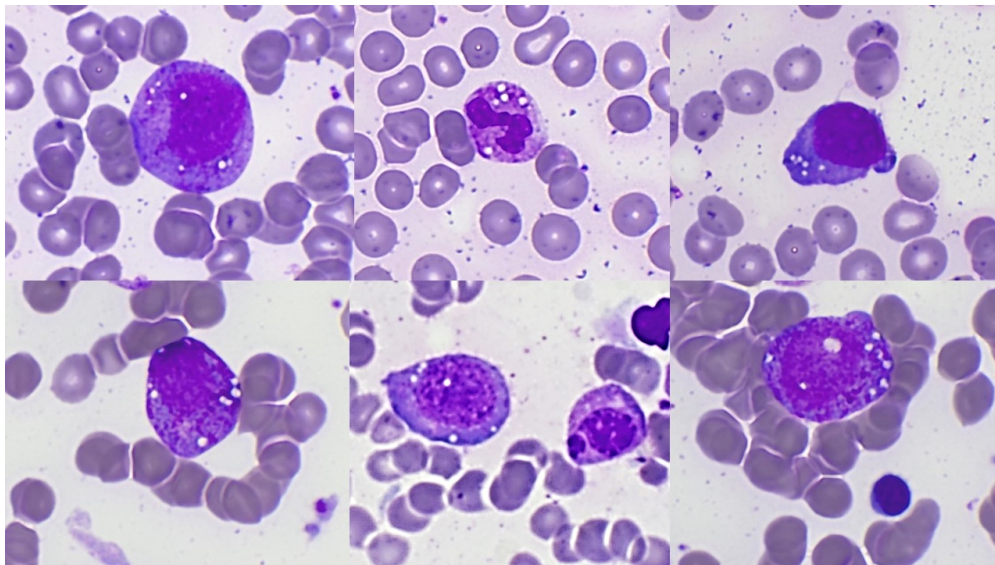


Figure 1: Bone marrow aspirate showing vacuolation in maturing myeloid and erythroid precursor cells (May-Grünwald-Giemsa stain x 600 magnification).

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References

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