ORBITAL/OCULAR INFLAMMATORY INVOLVEMENT IN VEXAS SYNDROME

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Aims:
To present the new diagnostic syndrome VEXAS involving the eye and the adnexa.
Methods:
A case of a 75-year-old male presented with fever and postseptal cellulitis of the left orbit for three weeks and was put on antibiotics. There was only minor improvement and a left orbital biopsy was performed demonstrating unspecific inflammation. Few days later the patient developed postseptal cellutis of the right eye and vitritis of the left eye.
Results:
The bone marrow was with myelodysplastic characteristics and a DNMT3A clone was discovered. Cytoplasmic vacuoles are localized in promyelocytes and myelocytes. A blood sample was used for Sanger sequencing and demonstrated a missense in the UBA1 gene (c.121A>G;p.(Met41Val) characteristic for the VEXAS Syndrome.
VEXAS syndrome (acronym for vacuoles, E1 enzyme, X-linked, autoinflammatory, somatic) is a monogenic disease first described by Beck et al. in 2020.
Conclusion:
Vexas Syndrome should be considered in an older male patient as differential in atypical presentations of inflammatory conditions especially if they are associated with haematological abnormalities, inflammation of an atypical organ or are treatment refractory. A genetic test for Vexas should be undertaken.

Key words: VEXAS Syndrome, inflammation, orbital/ocular involvement