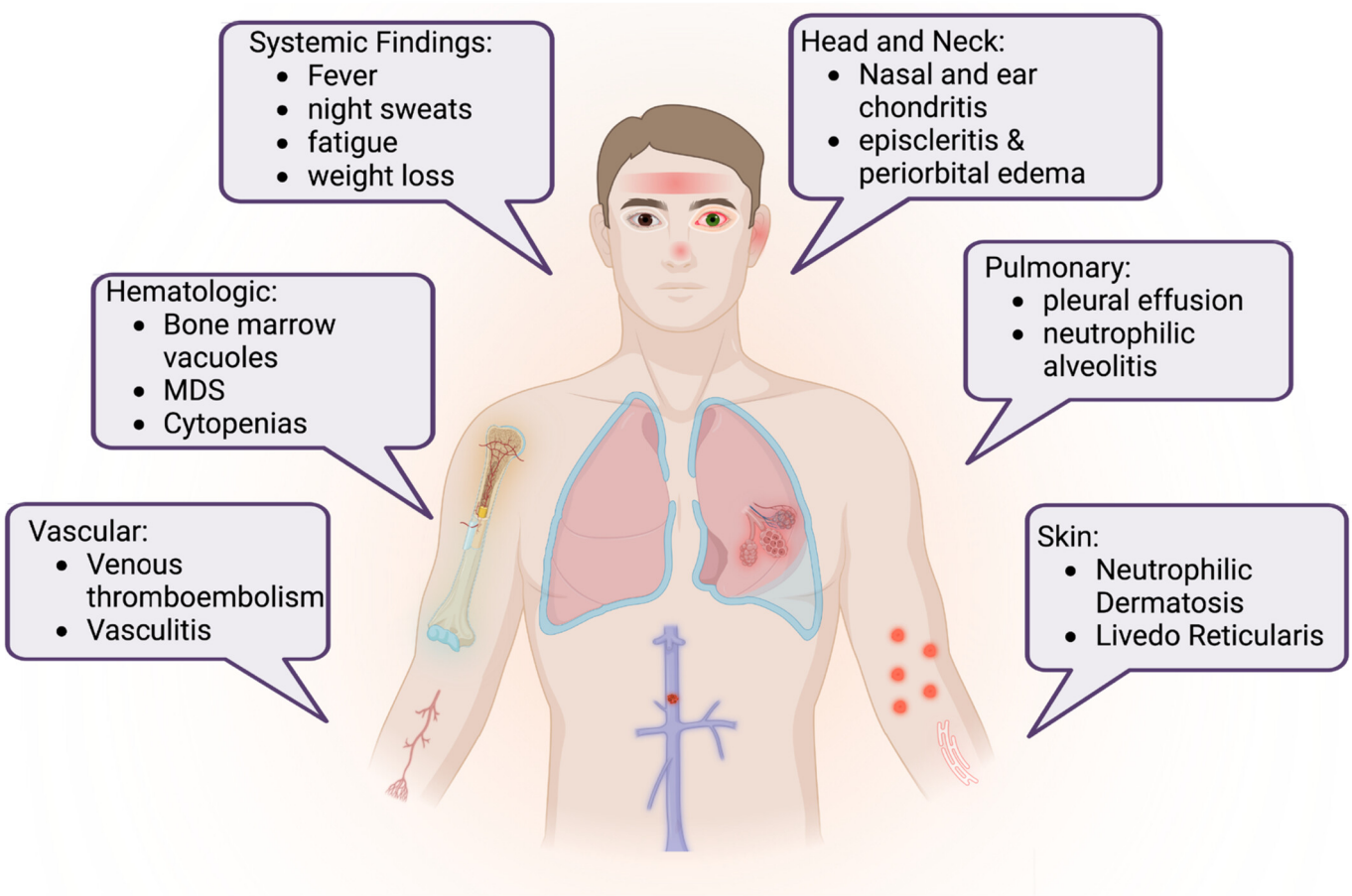
DISEASE PROFILE

VEXAS

VEXAS SYNDROME WAS initially described in December 2020 by Beck et al. using a genome-driven approach to screen peripheral blood exome data to identify 25 patients with a mutation at codon 41 (p.Met41) in UBA1, a gene located on the X chromosome encoding E1. These patients, all elderly males, showed overlapping clinical manifestations often with systemic inflammation and associated haematologic abnormalities.



References

1. Anis J. Saad, Mihir K. Patil, Nicolas Cruz, Chloe S. Lam, Connor O'Brien, Vinod E. Nambudiri. VEXAS syndrome: A review of cutaneous findings and treatments in an emerging autoinflammatory disease 2 March 2024 https://doi.org/10.1111/exd.15050

Sample positive report

Test Description:

This is a comprehensive molecular profile which uses next generation sequencing (NGS), Sanger Sequencing and fragment analysis testing performed on cell-free DNA (cfDNA) to identify molecular abnormalities (including SNVs, INDELS and CNVs) in 179 genes implicated in hematologic neoplasms, including leukemia, lymphoma, myeloma, and MDS. Whenever possible, clinical relevance and implications of detected abnormalities are described below.

Detected Genomic Alterations				
UBA1	TET2	SUZ12	No evidence of chromosomal structural gain or loss	

Heterogeneity
There is a dominant abnormal clone with UBA1 mutation. The TET2 and SUZ12 mutations are detected in very small subclones.

Diagnostic Implications	
UBA1, TET2, SUZ12	These abnormalities are consistent with VEXAS (Vacuoles, E1 enzyme, X-linked, Autoinflammatory, Somatic) syndrome

Therapeutic Implications	
SUZ12	SUZ12 mutation may suggest sensitivity to bromodomain inhibitors.

Prognostic Implications	
UBA1	Unknown
TET2	Neutral