MACHAON DIAGNOSTICS

TEST SUMMARY:
VEXAS GENETIC PANEL BY NGS

This high-sensitivity somatic sequencing test enables accurate and rapid—**48h STAT turnaround time**—diagnosis, helping clinicians identify VEXAS, ultimately guiding more effective management. This test sequences 100% of UBA1 exons plus 5-bp of flanking intronic DNA, detecting novel variants plus all 22 single nucleotide variants and small insertions/deletions (indels) so far associated with VEXAS.

VEXAS syndrome was only "discovered" in 2020 and is caused by pathogenic somatic variants in the UBA1 gene. Recent studies suggest VEXAS may be more common than initially believed, estimating a prevalence of ~1 in 4,000 men over age 50. Thus, physicians should consider VEXAS in older men who have a refractory adult-onset autoinflammatory syndrome with multiorgan involvement. Because pathogenic VEXAS variants are somatic and often mosaic in blood cells, they can be missed by conventional germline testing methods.

TURNAROUND TIMES
Routine TAT: <1 week
STAT: <48 hours (M-F)

SPECIMEN REQUIRMENTS
3mL EDTA Whole Blood

CPT CODES
81403

METHODOLOGY
Next-generation sequencing
Read depth ~5000x
Sensitivity: 2% variant allele frequency

ASSOCIATED TESTING
Cytokine Release Syndrome (12-test) Panel
Somatic Inborn Errors of Immunity (IEI) Genetic Panel

Please visit MachaonDiagnostics.com for more information.

References:
1. Beck et al (2020) N Engl J Med. 383(27):2628–38.
2. Beck et al (2023) JAMA. 329(4):318-324.
3. UBA1 Infevers Database.
https://infevers.umaimontpellier.fr/web/search.php#ancre2920

V:29MAY2025

WHY CHOOSE US?
- Fastest turnaround in the US
- Weekend testing for STATs
- Draw kits (includes free shipping)
- Clinical consultation
- Critical Results called to physician

ABOUT US:
Machaon Diagnostics is a clinical reference laboratory, specializing in coagulation, platelets, complement, genetics and rare disease.

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