MAYO CLINIC LABORATORIES

1-800-533-1710

UBA1Q

UBA1 Mutation, Quant, ddPCR, V

Patient ID	Patient Name		Birth Date	Sex	Age
SA00170572	TESTING, UBA1Q		2001-01-01	F	23
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00170572	SA00170572	CLIENT,CLIENT			
Account Information		**Collected**			
C7028846 DLMP Rochester		17 Sep 2024 00:00			

UBA1 Mutation, Quant, ddPCR, V

Specimen Type

MCR

Peripheral blood

Interpretation

[1] MCR

Peripheral blood, UBA1 Mutation, Quantitative detection:

Positive. UBA1 mutation is detected. The mutation identified is . The fractional abundance of the detected mutation is % (mutated/total copies). See comment.

Comment:
Somatic mutations in the E1-activating enzyme UBA1 are associated with VEXAS syndrome (vacuoles, E1 enzyme, X-linked, autoinflammatory, somatic). Pathogenic alterations most commonly involve the p.Met41 codon and intron 2 acceptor splice region, with rare occurrences at codon p.Ser56 (Patel et al., 2021, 34802540; Beck et al., 2020, 33108101; Georgin-Lavialle et al., 2022, 34632574; Ferrada et al., 2021, 33779074). Specific p.Met41 variants have been associated with various clinical features (Ferrada et al., 2022, 35973467). The presence of a known pathogenic UBA1 mutation detected by this assay is consistent with a diagnosis of VEXAS syndrome in the appropriate clinical and pathologic context. Clinical correlation is suggested.

ADDITIONAL INFORMATION

Method Summary: This assay detects specific UBA1 mutations (p.Met41Val/Leu/Thr, the c.118 intron 2 splice region and p.Ser56Phe) using a droplet digital polymerase chain reaction (ddPCR) system. DNA is extracted from patient samples and is PCR-amplified using oligonucleotide primers and mutant- and wildtype-specific fluorescently labeled probes directed to the genomic target regions. Results are analyzed using dedicated software and Poisson statistics to provide absolute quantification of mutant target and wild type copies. Calculated results are reported as mutant fractional abundance (variant allele fraction %). The analytical sensitivity of the assay is 0.5% (mutated/total UBA1 copies). This assay will not detect other potential genetic alterations outside the specified target regions and mutation types. The reproducibility of this assay is such that results within 0.5 log change should be considered equivalent.

Signing Pathologist

MCR

LAUREN SCHULTZ

Received: 17 Sep 2024 13:45 **Reported:** 17 Sep 2024 13:48

Laboratory Notes

[1] This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. This test has not been cleared or approved by the U.S. Food and Drug Administration.

Performing Site Legend

Code	Laboratory	Address	Lab Director	CLIA Certificate
MCR	Mayo Clinic Laboratories - Rochester Main Campus	200 First Street SW, Rochester, MN 55905	Nikola Baumann Ph.D.	24D0404292