MAYO CLINIC LABORATORIES

1-800-533-1710

UBA1Q

UBA1 Mutation, Quant, ddPCR, V

Patient ID	Patient Name		Birth Date	Sex	Age
SA00170571	TESTING, UBA1Q ATLAS		2001-01-01	M	23
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00170571	SA00170571	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:

Negative. No mutation is detected in the specific assay target regions (p.Met41, intron 2 splice region, p.Ser56). See comment.

Comment: This assay evaluates the known hotspot mutations in UBA1 including the p.Met41 codon, intron 2 splice acceptor region and p.Ser56 codon. The analytic sensitivity of the assay is 0.5% (mutated/total UBA1 copies). Samples containing a lower percentage of mutated nucleic acid or other possible genetic variants outside the targeted regions of the assay will not be detected by this method. A negative result does not exclude the presence of a pathologic process. Correlation with clinical and other laboratory findings is recommended.

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 13:37 **Reported:** 17 Sep 2024 13:40

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