MAYO CLINIC LABORATORIES

1-800-533-1710

KITQ

KIT p.Asp816Val Variant Analysis, Quantitative, Varies

Patient ID	Patient Name		Birth Date	Sex	Age
C7028846-0006242	KITQ, VALI NORMAL		2001-01-01	M	23
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
X100486255	X100486255	unknown,physician			
Account Information		**Collected**			
C7028846 DLMP Rochester		23 Dec 2024 03:00			

KIT D816V Variant Analysis Quant, V

Specimen Type

MCR

blood

Interpretation

[1] MCR

Peripheral blood, KIT p.Asp816Val mutation (D816V) analysis:

Negative. No mutation is detected in the specific assay target region for KIT p.Asp816Val (D816V). See comment.

Comment: This assay evaluates for the presence of the p.Asp816 Val (D816V) hotspot mutation in the KIT gene. The analytic sensitivity of the assay is 0.1% (mutated/total KIT copies). Samples with a fraction of mutated nucleic acid near or below this limit may not be detected due to sampling effects. Suboptimal DNA quantity or quality may also adversely affect the optimal assay sensitivity. Other genetic variants in KIT outside the targeted region of the assay will not be detected by this method. A negative result does not necessarily exclude the presence of a pathologic hematologic process. Correlation with clinical, pathologic and other pertinent laboratory findings is recommended.

ADDITIONAL INFORMATION

This test is performed using droplet digital polymerase chain reaction (ddPCR) to detect the KIT:c.2447A>T, p.Asp816Val (NM_000222.3:g.55599321A>T) variant. DNA extracted from patient samples is PCR-amplified using oligonucleotide primers and mutant- and wild type-specific fluorescently-labeled probes. 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 Calculated results are reported as KIT p.D816V mutant fractional abundance (variant allele fraction %) (Unpublished Mayo method). The analytical sensitivity of this assay is 0.1%; however, sensitivity may be adversely impacted by variability in tumor cell distribution, or limited overall DNA quantity or quality.

Signing Pathologist

MCR

LAUREN SCHULTZ

Received: 24 Dec 2024 11:20 **Reported:** 26 Dec 2024 10:39

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