MAYO CLINIC LABORATORIES

1-800-533-1710

KITQ

KIT p.Asp816Val Variant Analysis, Quantitative, Varies

Patient ID	Patient Name		Birth Date	Sex	Age
C7028846-0006243	KITQ, VALI ABNORMAL		2001-01-01	M	23
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
X100486256	X100486256	unknown,physician			
Account Information		**Collected**			
C7028846 DLMP Rochester		23 Dec 2024 04:00			

KIT D816V Variant Analysis Quant, V

Specimen Type

MCR

blood

Interpretation

[1] MCR

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

Positive. KIT p. Asp816Val mutation (D816V) is detected. The mutation identified is D816V. The fractional abundance of the detected mutation is 20.6% (mutated/total copies). See comment.

Comment: The p.Asp816Val (D816V) mutation in the KIT gene is identified in > 80% of systemic mastocytosis (SM) and less commonly in cutaneous mastocytosis. It represents a minor diagnostic criterion for systemic mastocytosis in the World Health Organization (WHO) and International Consensus Classification (ICC) systems. Presence of the KIT D816V mutation confers resistance to some tyrosine kinase inhibitors such as imatinib, nilotinib and masitinib but has shown response to midostaurin (PMID: 28031180; 31372066; 28744009). Avapritinib is a selective KIT D816V inhibitor that has shown activity in advanced SM (PMID: 35877999; 35640224; 34873345). Both midostaurin and avapritinib have received U.S. Food and Drug Administration (FDA) approval status for use in adult patients with advanced SM. Clinical and pathologic correlation is suggested.

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 Calculated results are reported as KIT p.D816V mutant fractional abundance (variant allele fraction %) (Unpublished Mayo method).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 11:21 **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