

***KIT* Asp816Val Mutation Analysis, Varies**

Test ID: KITVS

Explanation:

On the effective date, KITVS will become obsolete and will be replaced with KITQ, which is a more robust assay with a quantitative component for positive samples.

Recommended Alternative Test:

***KIT* p.Asp816Val Variant Analysis, Quantitative, Varies**

Test ID: KITQ

Useful for:

Diagnosing systemic mastocytosis

Genetics Information:

This test uses droplet digital polymerase chain reaction to detect the *KIT* NM_000222.3:c.2447A>T(p.Asp816Val) variant.

Testing Algorithm:

For more information see:

[-Mast Cell Disorder: Diagnostic Algorithm, Bone Marrow](#)

[-Eosinophilia: Bone Marrow Diagnostic Algorithm](#)

Methods:

Droplet Digital Polymerase Chain Reaction (ddPCR)

Reference Values:

An interpretive report will be provided indicating the status as positive or negative for *KIT* p.Asp816Val (D816V). *KIT* gene (NCBI accession NM_000222.3)

Necessary Information:

Specimen type is required to perform testing.

Specimen Requirements:

Submit only 1 of the following specimens:

Specimen Type:	Whole blood
Preferred:	Lavender top (EDTA)
Acceptable:	Yellow top (ACD)
Specimen Volume:	3 mL
Collection Instructions:	1. Invert several times to mix. 2. Send specimen in original tube. Do not aliquot. 3. Label specimen as blood.
Specimen Stability Information:	Ambient (preferred) 14 days/Refrigerate 14 days
Specimen Minimum Volume:	1 mL

Specimen Type:	Bone marrow aspirate
Preferred:	Lavender top (EDTA)
Acceptable:	Yellow top (ACD)
Specimen Volume:	3 mL
Collection Instructions:	1. Invert several times to mix. 2. Send specimen in original tube. Do not aliquot. 3. Label specimen as bone marrow.
Specimen Stability Information:	Ambient (preferred) 14 days/Refrigerate 14 days
Specimen Minimum Volume:	1 mL

Specimen Type:	Extracted DNA
Container/Tube:	1.5 to 2-mL tube
Specimen Volume:	Entire specimen
Collection Instructions:	1. Label specimen as extracted DNA 2. Provide indication of volume of DNA.
Specimen Stability Information:	Frozen (preferred)/Refrigerate/Ambient
Additional Information:	We cannot guarantee that all extraction methods are compatible with this test. If testing fails, one repeat will be attempted, and if unsuccessful, the test will be reported as failed, and a charge will be applied.
Specimen Minimum Volume:	50 mcL at 10 ng/mcL concentration

Specimen Type: Paraffin-embedded bone marrow aspirate clot

Container/Tube: Paraffin block

Specimen Stability Information: Ambient

Specimen Type: Tissue (FFPE)

Container/Tube: Paraffin block

Collection Instructions: Submit a formalin-fixed, paraffin-embedded tissue block.

Specimen Stability Information: Ambient

Additional Information: Decalcified core biopsies are not accepted.

Forms:

[Hematopathology Patient Information](#) (T676)

Specimen Stability Information:

Specimen Type	Temperature	Time	Special Container
Varies	Varies		

Cautions:

- Some cases of systemic mastocytosis (SM) may have sparse representation in bone marrow aspirate or blood samples containing too few neoplastic mast cells to successfully detect the *KIT* p.D816V (below limit of assay detection). Paraffin embedded tissues may be prone to sampling effects and DNA degradation, which may also compromise assay performance. Rare cases of SM may have other *KIT* mutations involving D816 (such as p.D816Y), or in other exon 17 codons that are not specifically targeted by this D816V ddPCR assay.
- Assay sensitivity may be impacted by variability in tumor cell distribution or limited overall DNA quantity or quality.

CPT Code:

81273

Day(s) Performed: Monday through Friday

Report Available: 4 to 7 days

Questions

Contact Michelle Rath, Laboratory Resource Coordinator at 800-533-1710.