

## ***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 Raths, Laboratory Resource Coordinator at 800-533-1710.