

# IT'S TIME TO UNCOVER KIT D816V, THE CAUSE OF SYSTEMIC MASTOCYTOSIS<sup>1</sup>

Information for Hematopathologists

## UNCOVERKIT

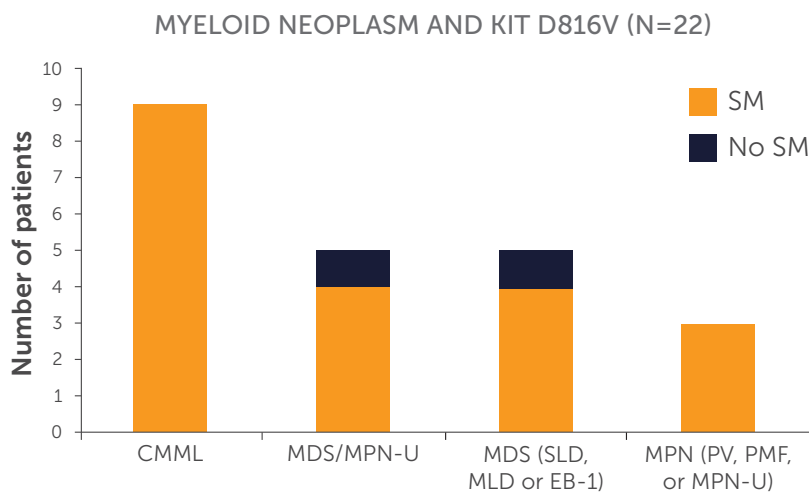
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## The KIT D816V mutation drives systemic mastocytosis in ~95% of patients<sup>1-3</sup>

Systemic mastocytosis is a myeloid neoplasm that unleashes uncontrolled mast cell proliferation and activation throughout the body.<sup>2</sup>

## Systemic mastocytosis with an associated hematological neoplasm (SM-AHN) may be missed in patients with myeloid neoplasms<sup>4</sup>

8-year retrospective review of 64 patients with suspected or confirmed myeloid malignancies with pathogenic KIT mutations:



- In one study, nearly all patients with the KIT D816V mutation initially diagnosed with a chronic myeloid neoplasm (20/22, 91%) were later confirmed to have SM-AHN
- In 35% of patients with chronic myeloid neoplasms confirmed with SM-AHN (7/20), there was no clinical or pathological suspicion of a systemic mast cell disorder, and a diagnosis of SM-AHN was not rendered upon initial pathological review

## Up to 70% of patients with advanced systemic mastocytosis have an associated hematologic neoplasm.<sup>5</sup>

CMML, chronic myelomonocytic leukemia; EB-1, excess blasts-1; KIT, KIT proto-oncogene, receptor tyrosine kinase; MDS, myelodysplastic syndrome; MDS/MPN, myelodysplastic syndrome/myeloproliferative neoplasm; MLD, multilineage dysplasia; MPN, myeloproliferative neoplasm; PMF, primary myelofibrosis; PV, polycythemia vera; SM-AHN, SM with associated hematologic neoplasm; SLD, single lineage dysplasia; U, unclassifiable.

Craig JW, et al. *Mod Pathol.* 2020;33(6):1135-1145.

## Complete a full diagnostic workup

Accurately diagnosing a patient with systemic mastocytosis requires a full diagnostic workup, which can include<sup>6-8</sup>:

- Bone marrow biopsy (top markers: CD117 and CD25)
- Serum tryptase test
- KIT D816V mutation testing

SEE BACK FOR MORE DETAILS ON  
SCREENING FOR SYSTEMIC MASTOCYTOSIS

# SUSPECT SYSTEMIC MASTOCYTOSIS? TEST FOR KIT D816V

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## Test using a high-sensitivity KIT D816V assay if SM is suspected<sup>8</sup>



Myeloid mutation panels alone are not recommended for the detection of KIT D816V. Next-generation sequencing assays can exhibit low sensitivity and high-sensitivity assays should always be performed. **If systemic mastocytosis is suspected, order mutational testing with a high-sensitivity KIT D816V assay.**

## Testing recommendations from the NCCN Clinical Practice Guidelines in Oncology (NCCN Guidelines<sup>®</sup>)<sup>8</sup>

- Use a highly sensitive assay, such as ASO-qPCR or digital droplet PCR, on the peripheral blood to detect KIT D816V
- A KIT mutational analysis may also be performed on the bone marrow aspirate following a positive test on the peripheral blood

## High-sensitivity testing is available through Blueprint's Biomarker Testing Program<sup>8</sup>

No-charge\* high-sensitivity  
KIT D816V testing for eligible patients  
is sponsored by Blueprint Medicines

[Access Blueprint's Biomarker Testing Program for Systemic Mastocytosis\\*](#) >

You can also learn about independent labs offering high-sensitivity (<1%) assays on peripheral blood.

[Learn More](#) >

\*Terms and conditions apply. Testing performed by Labcorp. While Blueprint Medicines provides financial support, Blueprint Medicines assumes no liability and provides no warranties for the testing services provided by independent third parties. See website for full program details and program eligibility.

**References:** **1.** Garcia-Montero AC, et al. *Blood*. 2006;108(7):2366-2372. **2.** Gilreath JA, et al. *Clin Pharmacol*. 2019;11:77-92. **3.** Verstovsek S. *Eur J Haematol*. 2013;90(2):89-98. **4.** Craig JW, et al. *Mod Pathol*. 2020;133(6):1135-1145. **5.** Reiter A, et al. *Blood*. 2020;135(16):1365-1376. **6.** Metcalfe DD. *Blood*. 2008;112(4):946-956. **7.** Theoharides TC, et al. *N Engl J Med*. 2015;373(2):163-172. **8.** Referenced with permission from the NCCN Clinical Practice Guidelines in Oncology (NCCN Guidelines<sup>®</sup>) for Systemic Mastocytosis V.3.2021. © National Comprehensive Cancer Network, Inc. 2021. All rights reserved. Accessed July 12, 2021. To view the most recent and complete version of the guideline, go online to NCCN.org. NCCN makes no warranties of any kind whatsoever regarding their content, use or application and disclaims any responsibility for their application or use in any way.

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