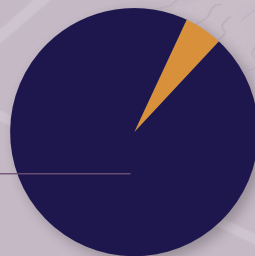


This pocket card belongs to the SM diagnostic test pocket card series.

High-Sensitivity *KIT* D816V Testing Can Help You Investigate a Systemic Mastocytosis Diagnosis

The most common *KIT* mutation in systemic mastocytosis (SM) is *KIT* D816V, which is found in

~95%
of cases of SM^{1,2}



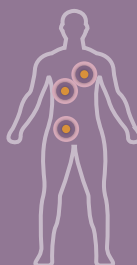
KIT is a tyrosine kinase receptor that plays a key role in regulating mast cell proliferation. SM is driven by activating mutations of *KIT*^{3,4}



KIT D816V is a reliable diagnostic and prognostic marker for SM^{3,4}



The presence of *KIT* mutation(s) at codon 816 or in other regions of *KIT* is one of the WHO minor criteria for SM⁵



ECNM recommends the use of high-sensitivity methods (e.g. ddPCR) for screening *KIT* D816V in peripheral blood upon clinical suspicion of SM⁶

DISCOVERSM

 **blueprint™**
MEDICINES
a sanofi company

Blueprint Medicines and associated logos are trademarks of Blueprint Medicines Corporation. All rights reserved. © 2025 Blueprint Medicines (Netherlands) B.V. INTBP-PRXSM-24.004v2.0, 08/25

Suspecting SM: *KIT* D816V Testing

Common symptoms of SM include

typical skin lesions, unexplained osteoporosis, anaphylaxis, histamine-induced symptoms (cramping, headache, hypotension, diarrhoea), cytopenia and eosinophilia, and splenomegaly⁶

Consider screening for *KIT* D816V mutation in combination with or after serum tryptase determination (>11.5 ng/mL) in patients with suspected SM⁶



Highly sensitive PCR methods (e.g. ddPCR with ~0.01% sensitivity) are recommended for the detection of *KIT* D816V in peripheral blood^{3,6}

In a study of ISM patients (n=39):⁷



95%
detection of *KIT* D816V
mutation with ddPCR assay



Next-generation sequencing assays have low sensitivity and are not sufficient to detect *KIT* D816V mutation^{2,3}



28%
detection of *KIT* D816V
mutation with NGS assay

Note: The high-sensitivity *KIT* D816V assay is one of several tests that can be used for SM suspicion. Further workup involves assessing if the patient meets WHO criteria for SM diagnosis⁵

For more information, visit: **www.systemicmastocytosis-hcp.com**

ddPCR=droplet digital polymerase chain reaction; ECNM=European Competence Network on Mastocytosis; WHO=World Health Organization.

References: 1. Pardanani A, et al. *Am J Hematol*. 2023;98(7):1097–1116. 2. Kristensen T, et al. *J Mol Diagn*. 2011;13(2):180–188. 3. Hoermann G, et al. *J Allergy Clin Immunol Pract*. 2022;10(8):1953–1963. 4. Verstovsek S, et al. *Eur J Haematol*. 2013;90(2):89–98. 5. Valent P, et al. *Hemasphere*. 2021;5(11):e646. 6. Valent P, et al. *J Allergy Clin Immunol Pract*. 2022;10(8):1999–2012.e6. 7. George TI, et al. *Blood*. 2020;136(Suppl 1):7.