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

 $\sim$ 95% of cases of SM<sup>1,2</sup>





KIT is a tyrosine kinase receptor that plays a key role in regulating mast cell proliferation. SM is driven by activating mutations of KIT<sup>3,4</sup>



KIT D816V is a reliable diagnostic and prognostic marker for SM<sup>3,4</sup>



The presence of *KIT* mutation(s) at codon 816 or in other regions of *KIT* is one of the WHO minor criteria for SM<sup>5</sup>



ECNM recommends the use of highsensitivity methods (e.g. ddPCR) for screening *KIT* D816V in peripheral blood upon clinical suspicion of SM<sup>6</sup>

## **DISCOVERSM**



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## 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 splenomegaly<sup>6</sup>

Consider screening for KIT D816V mutation in combination with or after serum tryptase determination (>11.5 ng/mL) in patients with suspected SM<sup>6</sup>



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

In a study of ISM patients (n=39):<sup>7</sup>



detection of *KIT* D816V mutation with ddPCR assay



Next-generation sequencing assays have low sensitivity and are not sufficient to detect *KIT* D816V mutation<sup>2,3</sup>



28%

detection of KIT D816V mutation with NGS assav

**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 diagnosis<sup>5</sup>

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

 $\label{lem:ddPCR} \mbox{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.