KIT D816 Mutation Analysis

Versiti offers highly sensitive detection of KIT D816 mutations for the diagnosis of systemic mastocytosis and prognosis in acute myeloid leukemia

KIT is located on chromosome 4q12 and encodes for the mast/stem cell growth factor receptor, a type III receptor tyrosine kinase.[1] The presence of somatic, activating mutations in KIT codon 816, predominantly D816V (A2447T), can be identified in the mast cells of 95% or more of patients with systemic mastocytosis (SM),[2] and represent clonal markers in the disease. D816 mutations serve as a WHO minor criterion for the diagnosis of SM[3] and appear to confer relative resistance to tyrosine kinase inhibitors such as imatinib.[4] KIT D816 mutations are also detectable in acute myeloid leukemia (AML), where they represent an adverse prognostic factor in combination with core binding factor (CBF) fusion gene.[5]

Indications for testing:

Diagnosis of systemic mastocytosis

Prognosis in AML

Test method:

KIT D816 mutations are detected by allele specific PCR.

Assay sensitivity and limitations:

The lower limit of detection of this assay is approximately 0.25% allele proportion for the D816V mutation. Other rare D816 mutations cross react with this assay.

Rare polymorphisms within primer or probe regions may interfere with the detection of KIT D816 mutations.

Reporting of Results

KIT D816V mutations are reported as mutation detected or not detected.

Specimen requirements:

3-5 ml EDTA (lavender top) whole blood or EDTA bone marrow.

SHIP

Shipping requirements:

Place the room temperature specimen and requisition in plastic bag, seal and insert in a Styrofoam container. Seal the Styrofoam container, place in a sturdy cardboard box and tape securely. Ship the package in compliance with your overnight carrier guidelines. Label with the following address:

Client Services/ Molecular Oncology Laboratory
Diagnostic Laboratories
638 N. 18th Street
Milwaukee, WI 53233

ORDER

Required forms:

Versiti Wisconsin Molecular Oncology Laboratoy Requisition

CPT Codes/Billing/Turnaround time:

Turnaround Time: 3-6 days

CPT Codes: For recommended CPT codes, visit the Versiti.org test catalog

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References

1. Garcia-Montero AC, Jara-Acevedo M, Teodosio C, et al. KIT mutation in mast cells and other bone marrow hematopoietic cell lineages in systemic mast cell disorders: a prospective study of the Spanish Network on Mastocytosis (REMA) in a series of 113 patients. Blood 2006;108:2366-72.
2. Horny HP, Metcalfe DD, Bennett JM, et al. WHO Classification of Tumours of Haematopoietic and Lymphoid Tissues. 4th ed. Lyon, France: WHO Press, 2008:54-63.
3. Roskoski R. Structure and regulation of Kit protein-tyrosine kinase--the stem cell factor receptor. Biochem Biophys Res Commun 2005:338:1307-15.
4. Ustun C, DeRemer DL, Akin C. Tyrosine kinase inhibitors in the treatment of systemic mastocytosis. Leuk Res 2011;35:1143-52.
5. Jawhar M, Dohner K, Kreil S, et. al. KIT D816 mutated/CBF-negative acute myeloid leukemia: a poor-risk subtype associated with systemic mastocytosis. Leukemia 2019; 33:1124–1134

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