

UCLA DIAGNOSTIC MOLECULAR PATHOLOGY LABORATORY
Department of Pathology and Laboratory Medicine
David Geffen School of Medicine at UCLA
Phone: (310) 794-2781, Fax: (310) 794-2765

c-kit Mutations Detection for Systemic Mastocytosis

↑CPT

83890; 83907; 84311; 83898; 83904; 83912

↑Synonyms

SM; Systemic Mastocytosis

↑Laboratory

Molecular Pathology

↑Availability

Monday-Friday, 0700-1700

↑Turnaround Time

3-14 days

↑Specimen

Blood, bone marrow, fresh frozen or paraffin tissue

↑Volume

4 mL of blood or 10 mm³ of tissue

↑Minimum Volume

1 mL of blood or 5 mm³ of tissue

↑Container

Lavender top (EDTA) tube, or fresh tissue, or formalin-fixed paraffin-embedded tissue

↑Storage Instructions

All specimens should be sent to the Laboratory immediately after collection, preferably by overnight delivery. Blood specimens should be kept at room temperature. Fresh tissue should be refrigerated but not frozen.

↑Causes for Rejection

Specimens inadequately identified; clotted blood; broken tube

↑Reference Range

No mutation observed

↑Use

Nucleotide substitutions at and adjacent to codon 816 in the catalytic domain of c-kit are common in SM. Detection of a codon 816 c-kit mutation is included as a minor diagnostic criterion in the WHO's diagnostic criteria for SM. Determining mutational status of the c-kit gene also has pharmacogenetic implications in patients considered for investigational mast cell cytoreductive therapies and targeted small-molecule tyrosine kinase inhibitors

↑Limitations

Absence of mutations in exon 17 of c-kit does not exclude a diagnosis of systemic mastocytosis. The sequencing method used will detect c-kit mutations around codon 816 as long as they are present in at least 20% of the total cells in the submitted specimen. At lower proportions in a mixed specimen, they will not be reliably detected. Other mutations in c-kit are associated with gastrointestinal stromal tumor (GIST). They are in different codons and are not detected by this test.

↑Methodology

Sequencing analysis. Polymerase Chain Reaction (PCR) is performed for DNA amplification. Primers are designed specifically for exon 17 of the c-kit gene. PCR products are sequenced and analyzed on the ABI 3130xl Genetic Analyzer.
