



Test Catalog

Diagnostic. Prognostic. Predictive. Predisposition.



CXCR4 Mutation Analysis

Alternative Name

CXCR4

Methodology

Molecular

Test Description

Bi-directional sequencing to detect nonsense, frameshift, and other mutations encoding the C-terminus of CXCR4. Analyzed range includes detection of the C1013G mutation and spans amino acids L301 to S352. Testing is available separately or as part of the [NeoTYPE® CLL Prognostic Profile](#). Testing is approved for specimens from the state of New York.

Clinical Significance

CXCR4 activates AKT1/MAPK pathways in B-lineage cells and facilitates cell migration in Waldenstrom macroglobulinemia (WM). Mutations are detected in nearly 30% of WM cases, and are associated with primary resistance and initial lack of response to BTK, PI3K, and mTOR inhibitors. The majority of these cases with CXCR4 mutations have concurrent MYD88 L265P mutations. The common C1013G mutation in CXCR4 and other somatic frameshift and nonsense mutations detected by this test are the same as or similar to the germline mutations associated with WHIM syndrome. Therapeutic antagonists to CXCR4 are in clinical trials.

Specimen Requirements

- **Peripheral blood:** 5 mL in EDTA tube.
- **Bone marrow:** 2 mL in EDTA tube.
- **FFPE tissue:** Paraffin block is preferred. Alternatively, send 1 H&E slide plus 5-10 unstained slides cut at 5 or more microns. Please use positively-charged slides and 10% NBF fixative. Do not use zinc fixatives.

Note: Test is DNA-based. Please select Extract & Hold - DNA if specimen hold service is desired.

Storage & Transportation

Use cold pack for transport. Make sure cold pack is not in direct contact with specimen.

CPT Code(s)*

81479

New York Approved

Yes

Level of Service

Global

Turnaround Time

10 days

*The CPT codes provided with our test descriptions are based on AMA guidelines and are for informational purposes only. Correct CPT coding is the sole responsibility of the billing party.

Please direct any questions regarding coding to the payor being billed.

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Committed to research as the means to improve patient care, we provide Pharma Services for pharmaceutical companies, in vitro diagnostic manufacturers, and academic scientist-clinicians. We promote joint publications with our client physicians. NeoGenomics welcomes your inquiries for collaborations. Please contact us for more information.

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