

MYD88, L265P, Somatic Gene Mutation, DNA Allele-Specific PCR, Varies

Overview

Useful For

Establishing the diagnosis of lymphoplasmacytic lymphoma/Waldenstrom macroglobulinemia

Helping to distinguish lymphoplasmacytic lymphoma/Waldenstrom macroglobulinemia (low-grade B-cell lymphoma) from other subtypes

Special Instructions

Hematopathology Patient Information

Method Name

Allele-Specific Polymerase Chain Reaction (PCR)

NY State Available

Yes

Specimen

Specimen Type

Varies

Shipping Instructions

Whole blood or bone marrow specimens must arrive within 10 days of collection.

Specimen Required

Submit only 1 of the following specimens:

Preferred:

Specimen Type: Bone marrow

Container/Tube:

Preferred: Lavender top (EDTA)
Acceptable: Yellow top (ACD)
Specimen Volume: 2 mL
Collection Instructions:

- 1. Invert several times to mix bone marrow.
- 2. Send bone marrow specimen in original tube. **Do not aliquot**.
- 3. Label specimen as bone marrow.

Specimen Stability: Ambient (preferred)/Refrigerated



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Specimen Type: Paraffin-embedded tissue

Container/Tube: Paraffin block Specimen Stability: Ambient

Specimen Type: Paraffin-embedded bone marrow aspirate clot

Container/Tube: Paraffin block Specimen Stability: Ambient

Acceptable:

Specimen Type: Whole blood

Container/Tube:

Preferred: Lavender top (EDTA)
Acceptable: Yellow top (ACD)
Specimen Volume: 3 mL
Collection Instructions:

1. Invert several times to mix blood.

2. Send whole blood specimen in original tube. **Do not aliquot.**

3. Label specimen as blood.

Specimen Stability: Ambient (preferred)/Refrigerated

Specimen Type: Frozen tissue
Container/Tube: Plastic container

Specimen Volume: 100 mg

Collection Instructions: Freeze tissue within 1 hour of collection.

Specimen Stability: Frozen

Specimen Type: Unstained slides

Container/Tube: Unstained tissue slides

Specimen Volume: 10 to 20 Slides

Additional Information: Tissue must demonstrate involvement by a hematologic neoplasm (eg, acute myelocytic

leukemia), not solid tumors.

Specimen Stability: Ambient

Specimen Type: Extracted DNA
Container/Tube: 1.5- to 2-mL tube
Specimen Volume: Entire specimen

Collection Instructions:

Label specimen as extracted DNA and source of specimen.
 Indicate volume and concentration of the DNA on the label.

Specimen Stability: Frozen (preferred)/Refrigerated

Specimen Type: Methanol-acetic acid (MAA) fixed pellets

Container/Tube: Plastic container

Specimen Stability: Ambient (preferred)/Refrigerated



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Forms

- 1. Hematopathology Patient Information (T676)
- 2. If not ordering electronically, complete, print, and send a <u>Hematopathology/Cytogenetics Test Request</u> (T726) with the specimen.

Specimen Minimum Volume

Whole blood, Bone marrow: 1 mL; Extracted DNA: 50 mcL at 20 ng/mcL; Other specimen types: See Specimen Required

Reject Due To

Gross	Reject
hemolysis	
Bone marrow	Reject
core biopsies	
Paraffin	
shavings	
Moderately to	
severely	
clotted	

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Varies	Varies	10 days	

Clinical & Interpretive

Clinical Information

The single point alteration in *MYD88*, L265P, is present in 67% to 100% of patients with lymphoplasmacytic lymphoma, and these patients typically have clinical manifestations of Waldenstrom macroglobulinemia (often designated LPL/WM).

Reference Values

Variant present or absent based on expected alteration polymerase chain reaction product size. Concurrent amplification of wild type *MYD88* fragment determined for sample amplification integrity. *MYD88* gene (NCBI accession NM_002468.4)

Interpretation

Mutation present or not detected; an interpretive report will be issued.

Cautions

This MYD88 test is a targeted assay and will not detect any alteration at MYD88 codon 265 that does not result in the L>P (leucine to proline) amino acid change. It will also not detect additional MYD88 alterations, including insertion or



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deletion events.

The analytical sensitivity of the assay (1% MYD88 L265P in a wild-type background) can be affected by a variety of factors, including biologic availability (ie, tumor burden), fixation of paraffin-embedded specimens, or nonspecific polymerase chain reaction interferences.

Rare cases of lymphoplasmacytic lymphoma/Waldenstrom macroglobulinemia (LPL/WM) have been reported lacking the *MYD88* L265P abnormality, so a negative result would not completely exclude this diagnosis but would make the possibility of LPL/WM more unlikely.

Clinical Reference

- 1. Treon SP, Xu L, Yang G, et al. MYD88 L265P somatic mutation in Waldenstrom's macroglobulinemia. N Engl J Med. 2012;367(9):826-833
- 2. Varettoni M, Arcaini L, Zibellini S, et al. Prevalence and clinical significance of the MYD88 (L265P) somatic mutation in Waldenstrom's macroglobulinemia and related lymphoid neoplasms. Blood. 2013;121(13):2522-2528
- 3. Xu L, Hunter ZR, Yang G, et al. MYD88 L265P in Waldenstrom macroglobulinemia, immunoglobulin M monoclonal gammopathy, and other B-cell lymphoproliferative disorders using conventional and quantitative allele-specific polymerase chain reaction. Blood. 2013;121(11);2051-2058
- 4. Poulain S, Roumier C, Decambron A, et al. MYD88 L265P mutation in Waldenstrom macroglobulinemia. Blood. 2013;121(22);4504-4511
- 5. Gachard N, Parrens M, Soubeyran I, et al. IGHV gene features and MYD88 L265P mutation separate the three marginal zone lymphoma entities and Waldenstrom macroglobulinemia/lymphoplasmacytic lymphomas. Leukemia. 2013;27(1):183-189. doi: 10.1038/leu.2012.257
- 6. Ondrejka SL, Lin JJ, Warden DW, Durkin L, Cook JR, Hsi ED. MYD88 L265P somatic mutation: its usefulness in the differential diagnosis of bone marrow involvement by B-cell lymphoproliferative disorders. Am J Clin Pathol. 2013;140(3):387-394

Performance

Method Description

Extracted DNA from the clinical specimen is subjected to a single-tube allele-specific polymerase chain reaction (PCR) using MYD88 exon 5 primers that simultaneously amplify both a wildtype sequence fragment and a fragment containing the specific nucleotide change resulting in L265P if present. PCR products are visualized by capillary electrophoresis, and the presence of altered and wildtype amplicons is determined according to the expected specific PCR product sizes.(Unpublished Mayo method)

PDF Report

No

Day(s) Performed

Monday through Friday

Report Available



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5 to 8 days

Specimen Retention Time

Blood, bone marrow: 2 weeks; Extracted DNA: 3 months

Performing Laboratory Location

Rochester

Fees & Codes

Fees

- Authorized users can sign in to <u>Test Prices</u> for detailed fee information.
- Clients without access to Test Prices can contact <u>Customer Service</u> 24 hours a day, seven days a week.
- Prospective clients should contact their account representative. For assistance, contact <u>Customer Service</u>.

Test Classification

This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. It has not been cleared or approved by the US Food and Drug Administration.

CPT Code Information

81305

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
MYD88	MYD88 L265P Gene Mutation	82140-5
	Analysis	

Result ID	Test Result Name	Result LOINC® Value
MP021	Specimen Type	31208-2
36308	Final Diagnosis	82140-5
621251	MYD88 Cancel	77202-0