

## 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:**

1. Label specimen as extracted DNA and source of specimen.
2. 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

## Forms

1. [Hematopathology Patient Information](#) (T676)
2. If not ordering electronically, complete, print, and send a [Hematopathology/Cytogenetics Test Request](#) (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 hemolysis	Reject
Bone marrow core biopsies Paraffin shavings Moderately to severely clotted	Reject

## 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 [Test Prices](#) for detailed fee information.
- Clients without access to Test Prices can contact [Customer Service](#) 24 hours a day, seven days a week.
- Prospective clients should contact their account representative. For assistance, contact [Customer Service](#).

**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 Analysis	82140-5

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