

# MYD88 L265P Mutation Detection by PCR, Quantitative

## Indications for Ordering

- Useful in distinguishing lymphoplasmacytic lymphoma (LPL) from other low-grade B-cell lymphoproliferative disorders which may be in the differential diagnosis
- Monitoring of individuals with LPL diagnosis and previously identified *MYD88* L265P mutation

## Test Description

- Real-time PCR
  - Performed on whole blood, bone marrow, and formalin-fixed, paraffin-embedded (FFPE) tissue
- Quantitation of *MYD88* L265P mutant alleles

## Tests to Consider

[MYD88 L265P Mutation Detection by PCR, Quantitative 2009318](#)

## Disease Overview

**Prevalence** – 3-4/million

- Mostly affects older individuals

## Diagnostic/treatment issues

- *MYD88* L265P mutations are present in the majority of LPL cases
  - Includes Waldenström's macroglobulinemia
  - Marker for risk of progression from monoclonal gammopathy of undetermined significance (MGUS) IgM to Waldenström macroglobulinemia
  - Mutation also detected in a low percentage of chronic lymphocytic leukemia (CLL) and diffuse large B-cell lymphoma (DLBCL) cases
- Detection of *MYD88* L265P mutation can aid in differentiation between LPL and other low-grade B-cell lymphoproliferative disorders which may appear similar to LPL
  - May be crucial for treatment decisions
  - LPL may be treated with chemotherapy or rituximab

## Genetics

**Gene** – *MYD88*

## Structure/function

- *MYD88* encodes an adaptor protein that acts as a signal transducer in the interleukin-1 and toll-like receptor signaling pathways
- *MYD88* L265P mutation augments cell survival through increased NF-κB activity and JAK-STAT3 signaling

## Test Interpretation

**Analytic sensitivity** – 0.5% mutant allele

## Results

- Detected – *MYD88* L265P mutation detected
  - Quantitated as % of *MYD88* L265P mutant allele
  - Strongly supports LPL in the presence of appropriate clinical and histologic setting
- Not detected – no mutation detected

## Limitations

- Does not detect mutations in other regions of the *MYD88* gene
- Does not detect *MYD88* codon 265 mutations other than L265P
- Results of this test must be interpreted in the context of morphological and other relevant data
- Test should not be used alone to diagnose malignancy