

Digital PCR (dPCR) for Detection of MYD88 L265P Variants

MYD88^{L265P} is the hallmark mutation in Waldenström Macroglobulinemia (WM) and IgM-monoclonal gammopathy of undetermined significance (IgM-MGUS). Its significance as prognostic and predictive factor is already demonstrated.

Initially identified on Bone Marrow (BM) CD19 selected cells (CD19+) using allele-specific quantitative polymerase chain reaction (AS-qPCR) or Sanger sequencing, in recent years, the advent of highly sensitive techniques, such as digital PCR (dPCR), have facilitated the MYD88 mutational screening also on unselected BM cells, peripheral blood (PB) samples and on plasmatic or cerebrospinal fluid cell-free DNA (cfDNA). Moreover, MYD88^{L265P} detection was proposed as reliable minimal residual disease (MRD) marker in BM samples. This presentation will uncover the advantages and feasibility of dPCR for MYD88^{L265P} detection in different specimens' type; the diagnostic and prognostic implications of MYD88 at baseline as well as its role as MRD marker for MRD monitoring. Finally, standardization experience for the implementation of MYD88^{L265P} ddPCR assay in clinical practice will be reported.