DIGITAL PCR IS A HIGHLY SENSITIVE METHOD FOR THE DETECTION OF MYD88^{L265P} MUTATION IN BONE MARROW TREPHINE BIOPSIES OF LYMPHOMAS WITH PLASMACYTIC/PLASMACYTOID DIFFERENTIATION

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Introduction: Though recognition of myeloma and Waldenstroem macroglobulinemia in bone marrow trephine biopsies is usually straight forward, IgM myelomas, lymphocytic/plasmacytoid lymphomas and plasmacytic/plasmacytoid subsets of other low-grade non-Hodgkin lymphomas may lead to diagnostic difficulties. The presence of MYD88^{L265P} mutation in the vast majority of Waldenstroem macroglobuinaemia proved to be a useful adjunct in the differential diagnosis of lymphomas with plasmacytic/plasmacytoid differentiation. Thus, we analysed and re-evaluated a broad spectrum of B-cell-derived lymphoproliferative disorders with plasmacytic/plasmacytoid differentiation diagnosed at the University Hospital Innsbruck between 2000 and 2012 for MYD88^{L265P} mutation.

Material and Methods: Patients and bone marrow trephine biopsies were be selected from the University Hospital Database and the tissue repository of the Department of Pathology. Biopsies were analysed by conventional histochemistry, immunohistochemistry and molecular analysis using allele specific and digital PCR. Results were correlated with pathological characteristics, clinical data and outcome reports.

Conclusions: We aimed to establish a distinct diagnostic algorithm for the differential diagnosis of plasmacytic/plasmacytoid variants of lymphoproliferative diseases using digital PCR for detection of MYD88^{L265P} mutation to enable an accurate diagnostic workup and proper patient allocation to definite treatment pathways.