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Myeloproliferative Neoplasms: Laboratory Workup in the Era of Next-Generation Sequencing

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Introduction

Myeloproliferative neoplasms (MPNs) are clonal hematopoietic stem cell disorders characterized by proliferation of at least one hematopoietic lineage, with minimal defects in maturation. The emergence of next-generation sequencing (NGS) technique has expanded the genetic landscape of MPNs. As a result, most MPNs are now known to carry well-defined molecular abnormalities, such as BCR-ABL1 rearranged chronic myeloid leukemia (CML), JAK2-mutated polycythemia vera (PV), JAK2, CALR or MPL mutated essential thrombocythemia (ET) and primary myelofibrosis (PMF), and CSF3R-mutated chronic neutrophilic leukemia (CNL). In addition, somatic mutations in genes that regulate DNA methylation, histone modification, mRNA splicing, transcription, and signal transduction have been shown to play important roles in subsequent disease progression. We share our experience in clinical applications of molecular testing in the diagnosis, risk stratification, monitoring of measurable/minimal residual disease (MRD) and target therapy of MPNs in the NGS era.

Recent publications

1.Wu L, Xia M, Sun X, Han X, Zu Y, Jabbour EJ, You MJ, Lin P, Li S, Xu J, Bueso-Ramos CE, Medeiros LJ, Qiu X, Yin CC. High levels of immunoglobulin expression predict shorter overall survival in patients with acute myeloid leukemia. Eur J Haematol. 2020;105:449-459.

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4. Yin CC and Jones D. Myeloid Neoplasms: Molecular Diagnostics. In: Cheng L, Zhang DY, Eble JN (Eds): Molecular Genetic Pathology (2nd edition). Springer 2013. p865-886.

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