Standards for the diagnosis and treatment of chronic myeloid leukemia

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Myeloproliferative disorders are a group of clonal myeloid neoplasms characterized by increased proliferation of myeloid cells with preserved cell differentiation. The molecular features of myeloproliferative neoplasms have been efficiently mapped in the last decades(1). Chronic myeloid leukemia (CML) is a myeloproliferative neoplasm defined by the presence of t(9;22)(q34;q11), a BCR-ABL1 gene fusion, and characterized by three distinct clinical-laboratorial phases: a chronic phase (CP) with leukocytosis, left shift, basophilia, eosinophilia, thrombocytosis, and no increase in blast counts; an accelerated phase (AP) characterized by increasing in blast count and additional cytogenetic changes; and a blastic phase (BP) characterized by more than 20% blasts in the bone marrow. The management of CML represents one of the major advances in the history of medicine with the transformation of a highly lethal condition into a chronic disease managed, most of the times, with one pill a day. Although rare, with an incidence of 1.6 per 100,000(2), the estimated 8-year survival of CML in CP used to be 6% before 1975. After the discovery of tyrosine kinase inhibitors for CML treatment the 8-year survival became 87% since 2001(3). The correct diagnosis and monitoring of CML involve distinct laboratorial techniques such as complete blood counts, bone marrow morphological analysis, conventional cytogenetics, fluorescence in situ hybridization and real time polymerase chain reaction (PCR). In order to achieve this highly successful treatment strategy, the correct diagnosis and laboratorial monitoring are crucial.

In this edition of the Jornal Brasileiro de Patologia e Medicina Laboratorial (JBPML), the paper of Dorfman et al. (2018)(4) presents a review of several aspects of CML. The paper covers diagnostic approaches, the importance of cytogenetic and molecular analysis, the three clinical phases of the disease, a summary of the three generations of tyrosine kinase inhibitors approved for CML treatment and monitoring strategies, including molecular criteria for determining treatment success and failure. The authors also include the cytogenetic and molecular anatomy of t(9;22)(q34;q11) and BCR-ABL1 gene fusion with some of their variants. This information is essential for professionals working with diagnosis and monitoring of these patients, since discrepancies between cytogenetic and molecular analysis often pose a challenge to clinical pathologists in the diagnosis and also for treatment definitions by hematologists(5), and a deep understanding of the possible molecular variations can be helpful in this context.

In summary, the review presented by Dorfman et al. (2018)(4) is a helpful resource for hematologists and pathologists dealing with diagnosis, monitoring and treatment of CML patients. It also serves as a useful source for students and professionals in need of a first contact with CML, since it comprehensively describe the main clinical and laboratorial aspects of this disease.

REFERENCES


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