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Acute myeloid leukemia with *CEBPA, NPM1, RUNX1* gene mutations

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Cytogenetic studies have traditionally played one of the most important roles for the classification and risk stratification of acute myeloid leukemia (AML). AML patients with a favorable risk cytogenetic profile are associated with relatively good responses to chemotherapy-based regimens and high complete remission rates, and AML patients with unfavorable cytogenetic risk profiles require allogeneic stem cell transplantation to improve prognosis. However, most AML cases have an intermediate prognosis and include approximately 50% of AML cases that have a normal karyotype at diagnosis and cannot be further subclassified based on cytogenetics. In recent years, a number of recurrent molecular mutations discovered in AML have been proposed to establish better classification of prognosis in the intermediate cytogenetic risk category. Recent revisions in the current 2008 WHO classification of tumours of haematopoietic and lymphoid tissues have resulted in adopting the provisional diagnostic entities AML with mutated *CEBPA* and AML with mutated *NPM1* as distinct entities, and adding AML with mutated *RUNX1* as a provisional diagnostic entity. During the past decade, numerous molecular genetic tests, Sanger DNA sequencing, and more recently, next-generation sequencing (NGS) approaches, have been employed to identify specific gene mutations that could be used to further divide cytogenetically normal AML (CN-AML) cases into clinically relevant prognostic subsets. Continued refinement of CN-AML cases within the AML classification using NGS-based testing will improve the identification of molecularly defined subsets of AML patients with different risk categories and will also provide the possibility of minimal residual disease monitoring using such mutations in specific quantitative molecular assays. Among the most frequent CN-AML-associated mutations, those affecting the *CEBPA*, *NPM1*, and *RUNX1* genes are associated with distinct biologic and clinical features and gene expression profiles. Therefore, this presentation will focus on the characteristic features of AML with mutations in the *CEBPA*, *NPM1*, and *RUNX1* genes.

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