Acute myeloid leukemia with characteristic molecular mutations

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. 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, several 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 WHO- ‘Classification of tumors of hematopoietic 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. This presentation will focus on the characteristic clinicopathologic features of AML with mutations in the CEBPA, NPM1 and RUNX1 genes.

Biography
Ryan S Robetorye has received his MD and PhD degrees from Baylor College of Medicine in Houston, Texas. He is board certified in Clinical Pathology, Hematology and Molecular Genetic Pathology and currently works as a Consultant at the Mayo Clinic in Phoenix, Arizona. He currently serves as the Medical Director of several clinical laboratories at the Mayo Clinic.

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