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MDS/acute leukemia predisposition syndromes

Within the past decade, nearly a dozen adult-onset inherited myelodysplastic syndrome (MDS) and leukemia predisposition syndromes have been identified. Individuals with inherited forms of hematologic malignancies are currently underdiagnosed due to the low frequency of cases and low level of clinician awareness of these syndromes. However, these individuals are increasing and likely to be encountered in clinical practice with wider adoption of NGS-based testing for the detection of prognostically significant or targetable genomic alterations in hematologic malignancies. Clinicians must increasingly recognize the possibility that some gene mutations identified in some genes may represent pathogenic germline mutations and initiate appropriate follow-up germline genetic testing. The increasing importance of recognition of germline mutations is evidenced by the inclusion of a new category in the 2016 revision of the WHO classification of myeloid neoplasms and acute leukemia designated classification of myeloid neoplasm with germline predisposition.

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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