DDX41 and its unique contribution to myeloid leukemogenesis

July 22 2024

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In an editorial paper published in *Oncotarget* titled, "DDX41 and its unique contribution to myeloid leukemogenesis," researcher Hirotaka Matsui from the National Cancer Center Hospital in Tokyo, Japan, and Kumamoto University discusses myeloid neoplasms.

Until the early 2000s, myeloid neoplasms attributable to genetic

backgrounds were considered exceedingly rare, with notable exceptions limited to those arising as components of systemic syndromes such as Fanconi anemia and Li-Fraumeni syndrome.

Historically, no hematopoietic-specific tumor syndromes had been identified until 1999, when RUNX1 was implicated as the causative gene for familial platelet disorder with a predisposition to acute myeloid leukemia (AML).

Subsequently, in 2004, CEBPA was recognized as another critical gene responsible for inherited AML. The subsequent advent and widespread application of comprehensive genetic analysis facilitated the identification of germline pathogenic variants in genes such as ANKRD26, ETV6, and GATA2 among patients with myeloid neoplasms that developed against a background of inherited thrombocytopenia or systemic disorders.

It is now established that genetic predisposition is present in approximately 10% of myeloid neoplasms, underscoring the fact that myeloid neoplasms with a genetic background are by no means exceptional.

"Among these, myeloid neoplasms caused by DDX41 variants are particularly noteworthy due to their distinct disease phenotype and pathogenesis," Matsui writes.

More information: Hirotaka Matsui, DDX41 and its unique contribution to myeloid leukemogenesis, *Oncotarget* (2024). DOI: 10.18632/oncotarget.28603

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