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Cryptic t(11;17) is a recurrent translocation resulting in *NUP98-PHF23* fusion that shares gene expression signaturesof *NUP98-HOXA9* fusion and leukemic stem cells

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Chromosome translocations are recurrent features of various types of hematological malignancies. These translocations produce specific fusion genes that play crucial roles in leukemogenesis. Translocations involving Nucleoporin 98 gene (*NUP98*) have been found in a wide array of hematopoietic malignancies. Due to its participation in the formation of fusion with at least 30 different genes, *NUP98* is considered to be one of the most promiscuous fusion partner genes. Although *NUP98* fusions were initially considered to be infrequent events, application of fluorescence *in situ* hybridization (FISH) and microarray-based molecular cytogenetic analysis has revealed cryptic rearrangements involving NUP98. With application of molecular technology, a single *NUP98* fusion (*NUP98-NDS1*) was detected in 16.1% of pediatric AML with apparent normal karyotype and in 2.3% of adult AML with apparent normal karyotype. In a previous study a cryptic t(11;17) was identified resulting in *NUP98-PHF23* fusion in a patient with AML. Expression of *NUP98-PHF23* fusion gene cloned from this patient has led to the development of myeloid, erythroid, T-cell, and B-cell leukemia in mice. Recently, it wasfound that t(11;17) is a recurrent translocation in AML. RNA-seq was applied to characterize and compare the t(11;17) translocation breakpoints in the two AML patients and also to detect differentially expression genes that are common in both patients with *NUP98-PHF23* fusion. It was found that *NUP98-PHF23* fusion shares gene expression signatures of *NUP98-HOXA9* fusion as well as leukemic stem cells.

Biography

Yi Ning is an American Board of Medical Genetics certified Cytogeneticist and an Associate Professor of Pathology in Johns Hopkins University. She earned her MD from Shanghai Medical University, Shanghai, China in 1984, and PhD from Baylor College of Medicine, Houston, Texas, in 1991. After completing her fellowship training in National Human Genome Research Institute, Bethesda, MD, she served as Director of Cytogenetics Lab at University of Maryland for 12 years. Currently she serves as Director of Cytogenetics Lab of Johns Hopkins Pathology providing cytogenetic diagnosis and conducting research to delineate molecular mechanisms of chromosome rearrangements in oncogenesis.

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