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Clonal cytogenetic abnormalities of undetermined significance

Myelodysplastic syndromes are a group of hematopoietic stem cell diseases characterized by cytopenia(s), morphological dysplasia, and clonal hematopoiesis. In some patients, the cause of cytopenia(s) is uncertain, even after thorough clinical and laboratory evaluation. Evidence of clonal hematopoiesis has been used to support a diagnosis of myelodysplastic syndrome in this setting. In patients with cytopenia(s), the presence of clonal cytogenetic abnormalities, except for +8, del (20q) and –Y, can serve as presumptive evidence of myelodysplastic syndrome. Recent advances in next generation sequencing have detected myeloid neoplasm-related mutations in patients who do not meet the diagnostic criteria for myelodysplastic syndrome. Various terms have been adopted to describe these cases, including clonal hematopoiesis of indeterminate potential and clonal cytopenia of undetermined significance. Similarly, studies have shown that certain chromosomal abnormalities, including ones commonly detected in myelodysplastic syndrome, may not be associated necessarily with an underlying myelodysplastic syndrome. These clonal cytogenetic abnormalities of undetermined significance (CCAUS) are similar to clonal hematopoiesis of indeterminate potential and clonal cytopenia of undetermined significance. Here, we review the features of CCAUS, distinguishing CCAUS from clonal cytogenetic abnormalities associated with myelodysplastic syndrome, and the potential impact of CCAUS on patient management..

Recent Publications

1. Zuo W, Wang S A, DiNardo C, Yabe M, Li S, et al. (2017) Acute leukemia and myelodysplastic syndromes with chromosomal rearrangement involving 11q23 locus, but not MLL gene. J Clin Pathol 70:244–249.
2. Goswami R S, Wang S A, DiNardo C, Tang Z, Li Y, et al. (2016). Newly emerged isolated del(7q) in patients with prior cytotoxic therapies may not always be associated with therapy-related myeloid neoplasms. Mod Pathol 29:727–34.
3. Tang Z, Li Y, Wang S A, Hu S, Li S, et al. (2016). Clinical significance of acquired loss of the X chromosome in bone marrow. Leuk Res 47:109–13.

Biography

Guilin Tang is a Hematopathologist and Cytogeneticist, Section Chief of Clinical Cytogenetic Laboratory in the Department of Hematopathology, and Adjunct Medical Director of the Department of School of Health Professions. Her clinical interests include diagnosis of hematologic neoplasms (both leukemia and lymphomas) and cancer cytogenetics. Her major research interest is the characterization and risk stratification of cytogenetic abnormalities in various types of hematological malignancies, to better understand the pathogenesis, identify new clinicopathologic entities and predict patient prognosis. She is also very interested in characterization of clinically indolent cytogenetic clones (clonal cytogenetic abnormalities of undetermined significance), especially those emerged following cytotoxic therapies.

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