Polysomy 8 defines a clinico-cytogenetic entity representing a subset of myeloid hematologic malignancies associated with a poor prognosis: report on a cohort of 12 patients and review of 105 published cases

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Tetrasomy, pentasomy, and hexasomy 8 (polysomy 8) are relatively rare compared to trisomy 8. Here we report on a series of 12 patients with acute myeloid leukemia (AML), myelodysplastic syndrome (MDS), or myeloproliferative disorder (MPD) associated with polysomy 8 as detected by conventional cytogenetics and fluorescence in situ hybridization (FISH). In an attempt to better characterize the clinical and hematological profile of this cytogenetic entity, our data were combined with those of 105 published patients. Tetrasomy 8 was the most common presentation of polysomy 8. In 60.7% of patients, polysomy 8 occurred as part of complex changes (16.2% with 11q23 rearrangements). No cryptic MLL rearrangements were found in cases in which polysomy 8 was the only karyotypic change. Our study demonstrates the existence of a polysomy 8 syndrome, which represents a subtype of AML, MDS, and MPD characterized by a high incidence of secondary diseases, myelomonocytic or monocytic involvement in AML and poor overall survival (6 months). Age significantly reduced median survival, but associated cytogenetic abnormalities did not modify it. Cytogenetic results further demonstrate an in vitro preferential growth of the cells with a high level of aneuploidy suggesting a selective advantage for polysomy 8 cells.

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