Identity

GTG-banded (left) and RHG-banded (right) metaphases on blood cells showing the isolated tetrasomy 13. In the present case, the specific 13ps+ polymorphism of one of the chromosomes 13 revealed that tetrasomy resulted in the triplication of the same parental chromosome 13 rather than a double-duplication mechanism.

Clinics and pathology

**Disease**
Acute myeloid leukaemia, poorly differentiated (AML-M0).

**Epidemiology**
Only 4 cases of primary acquired isolated tetrasomy have been described in patients with undifferentiated acute myeloid leukaemia.

**Prognosis**
The possibility that isolated tetrasomy 13 may represent an independent poor prognostic factor could be suggested by the poor outcome under therapy in our patients and those reported previously. However, responses to intensive chemotherapy in older patients (under 60 year of age) are lower than with younger patients, and all described cases of isolated tetrasomy 13 occurred in elderly subjects.

Genetics

**Note:** Two candidate genes mapped on chromosome 13 whose deregulated function might contribute to the development of transformation of undifferentiated myeloid cells are FLT1 and Rb1. However, their involvement in acute leukemia with trisomy 13 / tetrasomy 13 have to be determined, and the mechanism whereby the increased gene dose alone or in association with other additional mutation(s) confers neoplastic potential of undifferentiated phenotype is unknown.
Cytogenetics

Note: Tetrasomy 13 can occur in different cases of acute leukemia with trisomy 13 as the primary cytogenetic abnormality, or can be associated with additional abnormalities following transformation.

References


This article should be referenced as such: