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Leukaemia Section

Short Communication

1q triplication in hematologic malignancies

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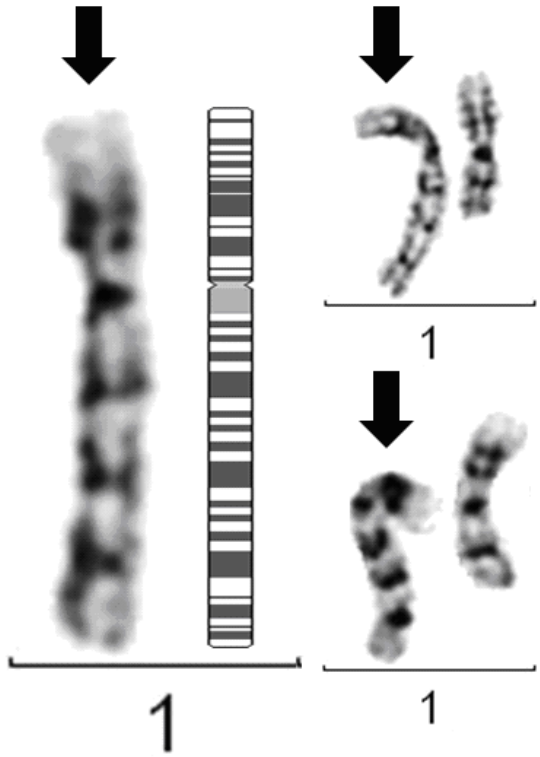

Giemsa-banding partial karyograms of 1q triplication and its representative ideogram. The arrows indicate a trp(1)(q21q32) chromosome.

Clinics and pathology

Disease

Acute myeloid leukemia (AML), myelodysplastic syndrome (MDS), Burkitt lymphomas or non-Burkitt type lymphomas, acute lymphoblastic leukemia (ALL), multiple myeloma (MM), myeloproliferative neoplasm (MPN) and Fanconi anemia.

Note

29 cases have been reported in the literature.

Phenotype/cell stem origin

Rare secondary karyotypic event in various hematologic malignancies; AML/MDS (8 cases), Lymphoma (9 cases), ALL (6 cases), MM (3 cases), MPN (2 cases), Fanconi anemia without other hematologic malignancies (1 case).

Epidemiology

Male predominance (71%), patients ranged in age from 14 to 69 (median 41.5 years).

Prognosis

Most of 1q triplication cases did not provide detailed information for the patients' survival. Although partial duplication/triplication of 1q or trisomy 1 was reported to be correlated with a poor outcome, further studies are needed for the evaluation of prognosis in such patients.

Cytogenetics

Cytogenetics morphological

1q triplication is a distinct secondary chromosomal abnormality. Most repeated region (tandem triplication) of 1q is q21-q32 (33% of total cases).

Additional anomalies

Most cases showed a complex karyotype except three solitary abnormalities of trp(1)(q) cases.

Genes involved and proteins

Note

The gene involved in trp(1)(q) is unknown. However, it was suggested that the most common region of duplication, 1q23-q24, harbors genes associated with tumor cell invasiveness.

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