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Leukaemia Section
Short Communication

t(Y;1)(q12;q12)

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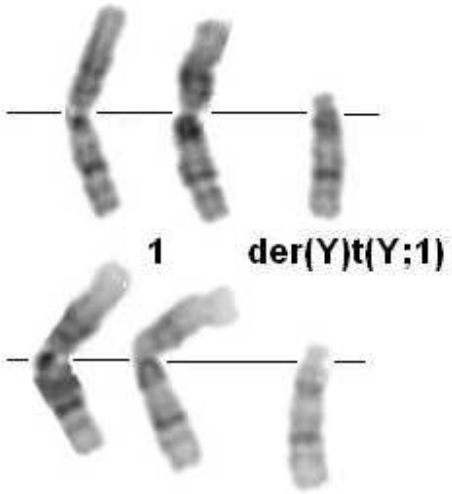

der(Y)t(Y;1)(q12;q12) G-banding.

Clinics and pathology

Disease

10 cases of hematological malignancy with der(Y)t(Y;1) had been reported to date. There were 8 cases of myelodysplastic syndrome, 1 case of polycythemia vera and 1 case of myelofibrosis.

Phenotype/cell stem origin

Suggested involvement of a pluripotent stem cell or a myeloid progenitor cell.

Etiology

Presence of der(Y)t(Y;1)(q12;q12) abnormality is relatively restricted to myelodysplastic syndrome.

Prognosis

Owing to the small number of cases reported, the prognostic implication of der(Y)t(Y;1) remains to be defined. It is however known to be compatible with long survival of up to 13-15 years. This aberration occurs as a transient abnormality in one case.

Cytogenetics

Cytogenetics morphological

Found it the unbalanced form + der(Y)t(Y;1)(q12;q12).

Genes involved and proteins

Note

Genes involved are unknown. Whether an increased dosage of gene products located at 1q12-qter or the breakpoint at chromosomal location of Yq12 is important in the pathogenesis of MDS remains to be elucidated. Furthermore, since juxtaposition of heterochromatin and euchromatin has been shown to affect gene function, this may contribute to the pathogenic mechanism underlying der(Y)t(Y;1) as the heterochromatin at Yq12 is involved in the translocation.

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