t(1;18)(q10;q10)

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Identity

der(1;18)(q10;q10) G- banding.

Clinics and pathology

Disease

Only two cases of hematological malignancy with der(1;18) are reported in the literature. In both instances der(1;18) occurs as the sole karyotypic abnormality. They are found in two patients suffering from myeloid disorders.

Phenotype/cell stem origin

Unknown, but may involve a myeloid progenitor cell as both reported cases can be grouped under myeloid malignancy.

Clinics

The first case was a 23-year old male who presented as myelodysplastic syndrome that rapidly progressed to acute myeloid leukemia, and died of neutropenic sepsis at induction phase. The second case was a 65-year old female diagnosed as chronic myeloproliferative disorder, unclassifiable, and run a chronic stable clinical course for years. She however suffered from recurrent pyogenic cutaneous infection.

Prognosis

Owing to the small number of cases reported, the prognostic implication of der(1;18)(q10;q10) remains to be defined. The clinical outcome of the two reported cases were markedly different, with one having rapid downhill course and short survival whereas the other one having chronic disease again if necessary.

Cytogenetics

Cytogenetics morphological

Found in the unbalanced form -18, + der(1;18), with trisomy for 1q and monosomy for 18p.

Genes involved and proteins

Note

Genes involved are unknown. Mechanistically, either trisomy 1q or monosomy 18p that results from the unbalanced translocation may potentially contribute to leukemogenesis. Trisomy 1q, arising through duplication or unbalanced translocations, is a recurrent theme in the myeloid disorders. Examples of such rearrangements include der(1;7)(q10;q10) and der(Y)t(Y;1)(q12;q12). Chromosomes with large constitutive heterochromatin bands such as chromosome 1 may be at risk of centromeric instability and be predisposed to centromeric fusion with other chromosomes.
References


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