t(1;22)(p13;q13)

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Identity

t(1;22)(p13;q13) G- and R- banding.

Clinics and pathology

Disease

Only found so far in M7 ANLL (acute megakaryocytic leukaemia); not found in Down syndrome (DS), and yet, DS is a disease with highly elevated risk of M7 (see leukaemia and Down Syndrome); misdiagnoses of a solid tumour have been documented.

Phenotype/cell stem origin

Megakaryocytic.

Etiology

No known toxic exposure.

Epidemiology

About 40 known cases; 0% to 3% of paediatric ANLL; 70 to 100% of infants M7; age: infants: median age 4 months; 20% are <1 month; 80% are <1 year; 95% are <2 years; sex ratio: 15M/24F (non significant).

Clinics

No preceeding myelodysplasia, and no history of transient leukemoid reaction; prominent organomegaly; blood data: moderate WBC; thrombocytopenia; myelofibrosis and fibrosis of other organs.

Cytology

Platelet-specific markers: platelet-peroxidase by electron microscopy, or platelet glycoproteins IIb/IIIa (CD41) or IIIa (CD61).

Treatment

Bone marrow transplantation is indicated.

Prognosis

Complete remission in only 50% of cases; median survival: 8 months; a few long survivors; absence of a prognostic indicator.

Cytogenetics

Additional anomalies

60% of cases (mostly patients under 6 months of age) have the t(1;22) as a single anomaly; the remaining third of cases (mainly patients above the age of 6 months) exhibit complex and hyperploid clones, with a highly monomorph pattern: +2, +19, +der(1)t(1;22), +6, +21 were found in more than 50% of cases each.
+10, +7, +15, +18, +20, del(1p), +4, +9, +14, +17, add(21p) are also recurrent; survival was equivalent in cases with or without a complex karyotype; the frequent presence of an additional der(1) indicates that the crucial event is likely to lie on the der(1)t(1;22).

**Variants**

One case of complex t(1;22) with a third chromosome has been described.

**Genes involved and proteins**

**Note**

Genes involved in this leukaemia are still unknown.

**To be noted**

**Note**

Individual data on the 39 published cases of t(1;22) and a complete bibliography can be found in our t(1;22) study group page.

**References**


Lion T, Haas OA. Acute megakaryocytic leukemia with the t(1;22)(p13;q13). Leuk Lymphoma. 1993 Sep;11(1-2):15-20


*This article should be referenced as such:*