Identity

Epidemiology

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

Clinics

No preceding myelodysplasia, and no history of transient leukemoid reaction; prominent organomegalia; 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 mths of age) have the t(1;22) as a single anomaly; the remaining third of cases (mainly patients above the age of 6 mths) 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, +8, +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**

2 cases of complex t(1;22) with a third chromosome have been described.

**Genes involved and proteins**

**OTT (one twenty-two)**

**Location:** 1p13

**Protein**

Contains RNA recognition motif consensus.

**MAL (megakaryocytic acute leukemia)**

**Location:** 22q13

**Protein**

931 amino acids; could attach DNA to nuclear scaffold and be involved in chromatin organization

**Result of the chromosomal anomaly**

**Hybrid gene**

**Description**

5' OTT - 3' MAL, comprising most of OTT fused to most of MAL; the reciprocal 5' MAL - 3' OTT may or may not be present.

**Fusion protein**

Oncogenesis

May modulate chromatin organization, HOX differentiation pathways, or extracellular signalling.

**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

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**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: