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Leukaemia Section
Short Communication

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 mths; 20% are < 1mth; 80% are < 1 yr; 95% are < 2 yrs; 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 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, comprisng 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 bibiography can be found in our t(1;22) study group page

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