

t(1;22)(p13;q13) (updated: old version not available)

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 preceding 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

Gene Name [OTT \(one twenty-two\)](#)

Location 1p13

Protein contains RNA recognition motif consensus

Gene Name [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 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
Description

Fusion Protein may modulate chromatin organization, HOX differentiation pathways, or extracellular signaling
Oncogenesis

External links

Other database [t\(1;22\)\(p13;q13\)](#) [Mitelman database \(CGAP - NCBI\)](#)

Other database [t\(1;22\)\(p13;q13\)](#) [CancerChromosomes \(NCBI\)](#)

To be noted

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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