

12p rearrangements in ALL

Clinics and Pathology

Disease	acute lymphocytic leukemia (ALL)
Phenotype / cell stem origin	lack of specificity for particular immunophenotype, although more stem origin frequent in B-lineage cases
Epidemiology	approximately 10-15% of pediatric ALL cases, and 5% of adult ALL
Prognosis	recent data indicate no difference in overall outcome between childhood ALL cases with versus without 12p abnormalities, although there was an improved outcome for pseudodiploid patients with versus without a cytogenetic 12p abnormality; although a dic(9;12) has been reported to be associated with an excellent outcome, in a recent study, there was no difference in outcome between those patients with a dic(9;12) versus patients lacking an abnormal 12p.

Cytogenetics

Cytogenetics	various aberrations result in an abnormal 12p; these include
Morphological	morphological balanced translocations with 12p breakpoints, del(12p), add(12p), monosomy 12, der(12)t(V;12)(V;p), and dic(V;12)(V;p); an abnormal 12p usually occurs as part of a more complex karyotype, and occurs as the sole aberration in less than 20% of cases with an abnormal 12p; in greater than 10% of cases both 12p homologues are abnormal; few cases with an abnormal 12p have more than 50 chromosomes
Additional anomalies	del(6q), del(13q) or monosomy 13, acquired +21 ; few recurring anomalies

Genes involved and Proteins

Note	approximately half of patients with an abnormal 12p have a rearranged TEL gene
Gene Name	TEL (or ETV6)
Location	12p13
Protein	TEL proteins belong to the ETS family transcription factors; important in the vitelline angiogenesis and in the bone marrow hematopoiesis

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