12p rearrangements in ALL

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Identity

![del(12)(p12) G-banding - Courtesy Diane H. Norback, Eric B. Johnson, and Sara Morrison-Delap, UW Cytogenetic Services.]

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 morphological**
Various aberrations result in an abnormal 12p; these include 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.
**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.

**References**


*This article should be referenced as such:*