t(12;17)(p13;q11-21) in ALL
David Betts
Department of Oncology, University Children’s Hospital, Steinwiesstr. 75, CH-8032 Zürich, Switzerland

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Disease
Acute lymphoblastic leukaemia (ALL).
Note: Identical or similar translocations have been reported rarely in acute myeloid leukaemia (AML) and acute mixed lineage leukaemia (t(12;17)(p13;q11-21) in AML).

Phenotype / cell stem origin
Most reports indicate an early pre-B immunophenotype, frequently characterised by low CD10 and positivity of the myeloid marker CD33.

Epidemiology
Rare; non-random translocation that predominantly occurs in children and young adults. No definable sex bias.

Prognosis
Early reports suggested that prognosis may be poor, but there are currently too few reported cases to define a robust association.

Cytogenetics

Cytogenetics morphological
The chromosome 17q breakpoint has been defined in different reports to be between q11-q21. The chromosome 12 breakpoint has been confirmed to be located in 12p13 telomeric to the ETV6/TEL locus. The translocation occurs as the sole or primary event in approximately 50% of cases.

Additional anomalies
No consistent picture and only +21 has been reported in more than one case.

Genes involved and Proteins
Note: Breakpoint on 12p13 telomeric to TEL. Currently the genes involved on both chromosome 12 and 17 are unidentified.

References

This article should be referenced as such: