

Leukaemia Section

Short Communication

t(7;12)(q36;p13)

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Clinics and pathology

Phenotype/cell stem origin

Acute lymphoblastic leukemia (ALL) and acute non lymphocytic leukemia (ANLL); ANLL cases are of various subtypes: M0, M1, M3V (with t(15;17) FISH negative), M4, M5, M7, and RAEB-t.

Epidemiology

At least 9 cases known; may be overlooked, and therefore underestimated; was found in 3% of children cases of ANLL, that was also 15% of infant cases of ANLL under 18 mths; sex ratio 3M/3F, age: 0-20 mths (n=9), median 8 mths (n=6).

Clinics

WBC range 8-230 x 10⁹/L, median 12 x10⁹/L; organomegaly, central nervous system involvement in 3 of 6 cases.

Prognosis

Poor prognosis: of 6 cases, one case had no remission and died at 7 mths; 4 cases had relapse (duration first remission 1-20 mths), 2 cases are still alive (16 mths + and 33 mths +).

Cytogenetics

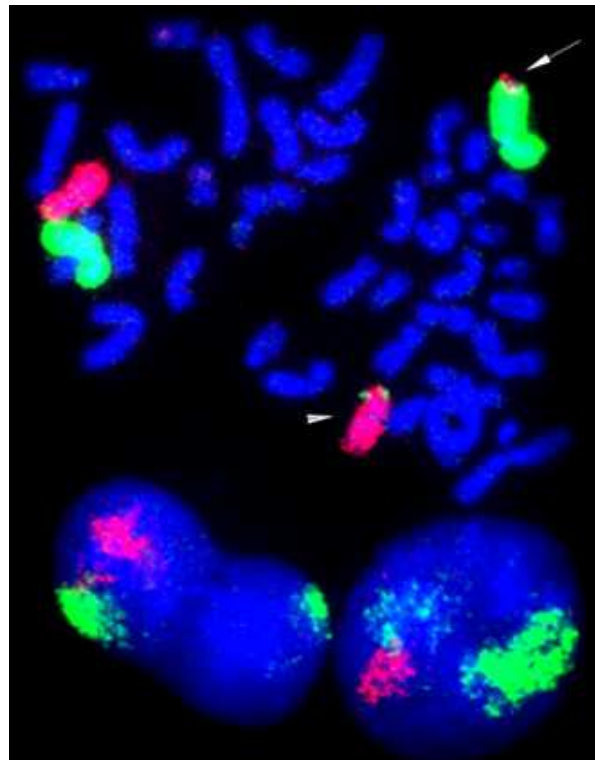
Cytogenetics morphological

Not always visible by chromosome banding techniques alone; may also be misdiagnosed as del(12)(p13).

Cytogenetics molecular

Detectable by dual colour FISH with chromosome 7 and 12 paints, although the translocation of chromosome 7 material onto the der(12) is not always visible. FISH using the ETV6 containing YAC 964c10 shows a split signal on the der(12) and der(7). Also the

commercial probe LSI TEL/AML1 (ES) for the detection of the t(12;21) shows a split signal on the der(7) and the der(12) in the t(7;12) cases since the breakpoint in these cases falls within the first three exons, which are contained in this probe. FISH using the PAC H_DJ1121A15 mapping to 7q36 shows a split signal on the der(7) and der(12).



Example of FISH performed on bone marrow metaphase from a patient with t(7;12)(q36;p13). Dual colour FISH using whole chromosome paint specific for chromosome 7 (in green) and chromosome 12 (in red) shows the reciprocal translocation. The arrow indicates the der(7) and the arrowhead indicates the der(12) - Sabrina Tosi.

Probes

- Chromosome 7 paint, wcp7 directly labelled with SpectrumGreen.
- Chromosome 12 paint, wcp12 directly labelled with SpectrumOrange.
- YAC 964c10 (CEPH, Paris).
- LSI TAL/AML1 (ES).
- PAC H_DJ1121A15.

Additional anomalies

+19 in 8 of 8 cases.

Genes involved and proteins**Note**

The gene in 7q36 is still unknown.

ETV6

Location: 12p13

DNA/RNA

9 exons; alternate splicing.

Protein

Contains a Helix-Loop-Helix and ETS DNA binding domains; wide expression; nuclear localisation; ETS-related transcription factor.

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