

Leukaemia Section

Short Communication

t(4;12)(q11-q21;p13)

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Identity

Note: it is likely that breakpoints are heterogeneous, with 2 distinct entities: t(4;12)(q11-12;p13) in ANLL, and t(4;12)(q13-21;p13) in ALL to be delineated.

Clinics and pathology

Disease

ANLL and therapy related AL cases with t(4;12)(q11-12;p13); B-cell ALL cases seem to have a more distal breakpoint in 4q13 or 21.

Phenotype / cell stem origin

ANLL cases: M0, M1, and other subtypes; often CD7+; a stem cell may be involved; ALL cases are CD10+.

Epidemiology

Only 13 available cases: 9 ANLL and 4 ALL; so far, ANLL cases with a proximal breakpoint in 4q11 or 12 are adult cases (43-77 yrs), and ALL cases are children cases (3-14 yrs); balanced sex ratio.

Prognosis

Adult cases: response to therapy is poor and median survival might be a year.

Cytogenetics

Additional anomalies

None in 5/12 cases (all 5 cases are ANLL); del(6q) has recurrently been found in ALL; -7 would be recurrent if only 1 entity exists; the karyotype in ALL cases can be complex.

Genes involved and Proteins

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.

References

Harada H, Harada Y, Eguchi M, Dohy H, Kamada N. Characterization of acute leukemia with t(4;12). Leuk Lymphoma 1997 Mar;25(1-2):47-53.

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Huret JL, Lafage-Pochitaloff M. t(4;12)(q11-q21;p13). Atlas Genet Cytogenet Oncol Haematol.1998;2(1):26.