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
at least 17 available cases: 13 ANLL and 4 ALL; so far, ANLL cases with a proximal breakpoint in 4q11 or 12 are adult cases (43-81 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 7/14 cases (all 7 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

CHIC2
Location
4q11-q12
Note
CHIC2 is involved in the ANLL cases; there is no data concerning ALL cases
Protein
Contains a transmembrane domain; member of a family of proteins with function in exocytosis.

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.

Result of the chromosomal anomaly

Hybrid gene
Description
5’ CHIC2 - 3’ ETV6
Fusion protein
Description
N term CHIC2 with the first 110 amino acids (exons 1 to 3) fused to the C term amino acids (exons 2 to 8) of ETV6.

To be noted
Case Report
The rare t(4;12)(q11;p13) in an elderly patient with de novo AML with multilineage dysplasia co-expressing stem cell markers.
t(4;12)(q11;p13) in an acute myeloid leukemia without maturation with myelodysplasia.

References

This article should be referenced as such: