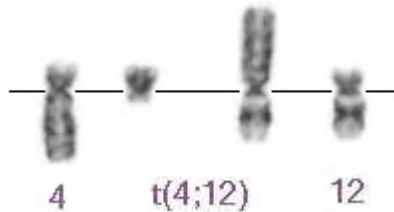


t(4;12)(q11-q21;p13)

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



t(4;12)(q11;p13) G-banding - Courtesy Jacqueline Beerworth and Sarah Moore

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 4 q11 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

Gene Name [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.

Gene Name [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 5' CHIC2 - 3' ETV6
Description

Fusion Protein N term CHIC2 with the first 110 amino acids (exons 1 to 3) fused to the 441 C term amino acids (exons 2 to 8) of ETV6.
Description

To be noted

Additional cases are needed to delineate the epidemiology of this rare entity:

you are welcome to submit a paper to our new [Case Report section](#).

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](#)

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URL : <http://www.infobiogen.fr/services/chromcancer/Anomalies/t0412.html>

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