t(5;14)(q33;q32) PDGFRB/TRIP11

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Published in Atlas Database: February 1998
Online updated version: http://AtlasGeneticsOncology.org/Anomalies/t0514ANL.html
DOI: 10.4267/2042/37418

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Clinics and pathology

Disease
Yet poorly known: 1 case of ANLL.

Clinics
Found at relapse with eosinophilia of a M2 ANLL with t(7;11).

Cytogenetics

Cytogenetics, morphological
So far found as an additional anomaly in a clone bearing a t(7;11)(p15;p15).

Genes involved and Proteins

PDGFRB
Location: 5q33
Protein
PDGFRB is the receptor for PDGFB (platelet-derived growth factor-b); membrane protein; belongs to the immunoglobulin superfamily.

CEV14
Location: 14q32
Protein
Contains a N-term leucine zipper and a C-term putative thyroid hormone receptor interacting domain.

Results of the chromosomal anomaly

Hybrid gene
Description
5' CEV14 - 3' PDGFRb

Transcript
10 kb fusion transcript (major) and other (minor) transcripts.

Fusion protein
Description
N-term leucine zipper from CEV14 fused to the transmembrane domain and the Tyr kinase domain of PDGFRb in C-term; the reciprocal transcript is not expressed; breakpoints at amino acids 1936 of PDGFRb and 567 of CEV14.

Oncogenesis
Ectopic constitutive tyrosine kinase activation of PDGFRb may occur.

To be noted
The above t(5;14)(q33;q32) with PDGFRb and CEV14 rearrangements must not be confused with the t(5;14)(q31;q32) with IL3 and IgH involvements found in ALL.

References