Leukaemia Section

Short Communication

t(5;14)(q33;q32) PDGFRB/KIAA1509

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Clinics and pathology

Disease
Atypical chronic myeloid leukemia (a-CML).

Epidemiology
Only one case to date with ascertainment of the genes involved (another case was a 18 year old female patient with a M2 acute non lymphocytic leukemia (ANLL) and a t(7;11)(p15;p15); the t(5;14) appeared at relapse, 5 months before death, 27 months after diagnosis).

Clinics
The sole case of t(5;14)(q33;q32) with certain PDGFRB/KIAA1509 involvement was a 42 year old male patient, BCR-ABL negative, treated with imatinib, and maintained in complete remission 18 mths after diagnosis.

Results of the chromosomal anomaly

Hybrid gene
Description
5' KIAA1509 - 3' PDGFRB; breakpoint in PDGFRB intron 10, identical to most PDGFRB breakpoints.

Fusion protein
Description
934 amino acids composed of the 355 amino acids from KIAA1506 in N-term and 579 amino acids from PDGFRB C-term.

Oncogenesis
The coiled coil domain may mediate PDGFRB homodimerization and constitutive activation.

Genes involved and Proteins

PDGFRB
Location: 5q33
Protein
PDGFRB is the receptor for PDGFB (platelet-derived growth factor-b); Ig like, transmembrane and tyrosine kinase domains; membrane tyrosine kinase; can homodimerize.

KIAA1509
Location: 14q32
Protein
Poorly known; 1935 amino acids; possess a coiled coil domain.

References


This article should be referenced as such:
Huret JL, t(5;14)(q33;q32) PDGFRB/KIAA1509. Atlas Genet Cytogenet Oncol Haematol.2006;10(3):188.