t(5;15)(q33;q22)

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Leukaemia Section
Short Communication

Clinics and pathology

Disease
Atypical chronic myelogenous leukemia (a-CML) (BCR-ABL negative chronic myeloproliferative disease).

Epidemiology
Only one case to date, a 79 year old male patient.

Prognosis
The disease was sensitive to imatinib, but the patient developed resistance to imatinib and died 14 months after diagnosis.

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.

TP53BP1
Location: 15q22
Protein
Component of the cellular response to DNA damage.

Results of the chromosomal anomaly

Hybrid gene
Description
5' TP53BP1 - 3' PDGFRB; breakpoint in PDGFRB intron 10, identical to most PDGFRB breakpoints; exon 23 of TP53BP1 fused in frame to PDGFRB exon 11; reciprocal product not detectable.

Fusion protein

Description
247 kDa; composed of the N-term TP53BP1 including the coiled coil domains and the kinetochore binding domain from TP53BP1 fused to the transmembrane and the tyrosine kinase domains of PDGFRB C-term.

Oncogenesis
The coiled coil domains from TP53BP1 may mediate PDGFRB homodimerization and constitutive activation of its tyrosine kinase activity; on the other hand, the DNA damage response of TP53BP1 may be perturbed.

References


This article should be referenced as such: