t(5;15)(q33;q22)

Clinics and Pathology

Disease	Atypical chronic myelogenous leukemia (a-CML) (BCR-ABL negative chronic myeloproliferative disease)
Epidemiology	Only one case to date, a 79 yr old male patient
Prognosis	The disease was sensitive to imatinib, but the patient developped resistance to imatinib and died 14 mths after diagnosis

Genes involved and Proteins

Gene Name	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
Gene Name	TP53BP1
Location	15q22
Protein	Component of the cellular response to DNA damage

Result 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.

External links

Other database	t(5;15)(q33;q22)	Mitelman database (CGAP - NCBI)
Other database	t(5;15)(q33;q22)	CancerChromosomes (NCBI)

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.

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