Leukaemia Section
Short Communication

t(2;13)(p16;q12)
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Clinics and pathology

Disease
Myeloproliferative disorder (atypical chronic myelogenous leukaemia (a-CML)).
Note: BCR-ABL negative myeloproliferative disease undistinguishable from CML otherwise.

Epidemiology
Only one case to date, a 32-year-old female patient.

Prognosis
Unknown; the patient received bone marrow transplantation, relapsed 4 years later, and was in complete remission 6 months after treatment of the relapse.

Genes involved and Proteins

SPTBN1
Protein
Scaffold protein; forms homo-tetramer; non-erythrocytic beta-spectrin; joins the actin cytoskeleton to the plasma membrane.

FLT3
Location: 13q12
Protein
Class III receptor tyrosine kinase (RTK); promotes signalling through phosphorylation of multiple proteins and activation of several downstream signalling pathways, such as the Ras/Raf/MAPK and PI3 kinase cascades.

Results of the chromosomal anomaly

Hybrid gene
Description
Fusion of exon 3 of SPTBN1 and exon 13 of FLT3.

Fusion protein
Description
Encodes a 66 kDa protein which retains the 2 coiled-coil domains of SPTBN1 and the tyrosine kinase domain of FLT3.

Expression localisation
Expression of the fusion protein transformed Ba/F3 cells to growth factor independance.

Oncogenesis
Constitutive phosphorylation.

References

This article should be referenced as such: