Leukaemia Section
Short Communication

(t4;12)(p16;p13)
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Published in Atlas Database: August 2005
Online updated version: http://AtlasGeneticsOncology.org/Anomalies/t0412p16p13ID1343.html
DOI: 10.4267/2042/38268
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Clinics and pathology

Disease
Peripheral T-cell lymphoma.

Epidemiology
Only one case to date, a 63 year old female patient.

Prognosis
No data.

Cytogenetics

Additional anomalies
Were found.

Genes involved and Proteins

FGFR 3
Location: 4p16.3
Protein
115 kDa; contains, from N-term to C-term: an extracellular domain with a signal sequence and 3 Ig-like loops, a transmembrane domain, and an intracellular domain with 2 tyrosine kinase domains. FGFR3 is a fibroblast growth factor receptor with tyrosine kinase activity; binding of ligand (FGF) induces receptor dimerization, autophosphorylation and signal transduction.

ETV6
Location: 12p13
Protein
53 and 57 kDa; contains, from N-term to C-term: an helix -loop-helix (HLH) domain (or 'sterile alpha domain' SAM) responsible of dimerization, and a sequence specific DNA-binding domain (ETS domain); binds to 5' CCGGAAGT 3'; ETV6 is a member of the ETS family of transcription factors; transcriptional repressor.

Results of the chromosomal anomaly

Hybrid gene
Description
5' ETV6 - 3' FGFR3; splicing variants (1618 and 1767 bp); the 1767 bp variant is an in frame fusion of ETV6 exon 5 to FGFR3 exon 10; both breakpoints are within exons; no reciprocal transcript.

Fusion protein
Description
Contains, from N-term to C-term, the HLH domain of ETV6 fused to the tyrosine kinase domains of FGFR3.

References


This article should be referenced as such: