

t(4;12)(p16;p13)

Clinics and Pathology

Disease Peripheral T-cell lymphoma

Epidemiology only one case to date, a 63 yr old female patient

Prognosis no data

Cytogenetics

Additional anomalies were found

Genes involved and Proteins

Gene Name

[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

Gene Name

[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 ofr 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

Result 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 contains. from N-term to C- term. the HLH domain of ETV6 fused to the

Protein tyrosine kinase domains of FGFR3
Description

External links

Other database [t\(4;12\)\(p16;p13\)](#) [Mitelman database \(CGAP - NCBI\)](#)
Other database [t\(4;12\)\(p16;p13\)](#) [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](#).

Bibliography

Fusion of ETV6 to fibroblast growth factor receptor 3 in peripheral T-cell lymphoma with a t(4;12)(p16;p13) chromosomal translocation.

Yagasaki F, Wakao D, Yokoyama Y, Uchida Y, Murohashi I, Kayano H, Taniwaki M, Matsuda A, Bessho M.

Cancer Res 2001; 61 : 8371-8374.

Medline [11731410](#)

Contributor(s)

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Citation

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