

t(X;11)(q13;q23)

Clinics and Pathology

- Disease** Rare type of acute leukemia: acute non lymphocytic leukemia (ANLL), and acute lymphoblastic leukemia (ALL)
- Note** Peripheral leucocytes at diagnosis of Case 2 were cultured and are presently known as the KARPAS-45 cell line.
- Epidemiology** Two cases known in the literature: Case 1: 7 mths, ANLL, male; Case 2: 30 mths, T-ALL, male. CLINICS Clinics at presentation: Case 1: Fever, bloody stool. WBC: 72.000 X 10⁹/l; Case 2: Mediastinal mass, dyspnoea. No hepatosplenomegaly. WBC: 5.600 X 10⁹/l.
- Prognosis** Grave, both cases died within a year after diagnosis.

Cytogenetics

- Probes** Pooled cDNA FISH-probes from AFX1: AFX 12, 115, 106, 108, 114.
- Additional anomalies** in KARPAS 45: Hypotetraploidy. Y, -3, +6, -14, -18 t(1;5)(q21;q12.2)x2, del4(4)(q22), del(16)(q22)

Genes involved and Proteins

Gene Name [AFX1 \(All-1 fusion partner on chromosome X\)](#)

Location Xq13

Gene Name [MLL \(Mixed Lineage Leukemia\)](#)

Location 11q23

Result of the chromosomal anomaly

Hybrid gene

5' MLL-AFX1 3' and the reciprocal: 5' AFX1-MLL 3'

Description

Fusion Protein Hybrid transcript MLL-AFX1 contains the code for the following domains:

Description AT-hook + DNA methyltransferase (from MLL) + part, aa 147-187 of the

DNA-binding domain (from AFX1)

External links

Other database [t\(X;11\)\(q13;q23\)](#) [Mitelman database \(CGAP - NCBI\)](#)

Other database [t\(X;11\)\(q13;q23\)](#) [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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Contributor(s)

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