Atlas of Genetics and Cytogenetics in Oncology and Haematology

OPEN ACCESS JOURNAL AT INIST-CNRS

Leukaemia Section
Short Communication

t(X;11)(q13;q23)

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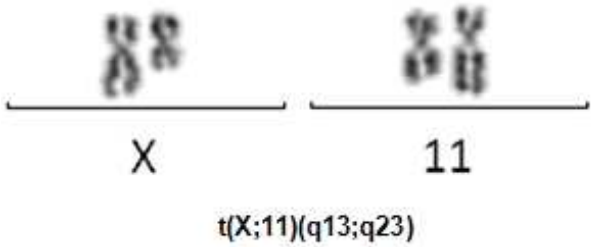
Published in Atlas Database: August 2001

Online updated version : http://AtlasGeneticsOncology.org/Anomalies/t0X11ID1127.html
DOI: 10.4267/2042/37793

Identity



t(X;11)(q13;q23) G- banding - Courtesy Melanie Zenger and Claudia Haferlach.

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 litterature: 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 10^9/l; Case 2: Mediastinal mass, dyspnoea. No hepatosplenomegaly. WBC: 5.600 X 10^9/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

AFX1 (All-1 fusion partner on chromosome X)

Location

Xq13

MLL (Mixed Lineage Leukemia)

Location

11q23

Result of the chromosomal anomaly

Hybrid gene

Description

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

Fusion protein

Description

Hybrid transcript MLL-AFX1 contains the code for the following domains: AT-hook + DNA methyltransferase (from MLL) + part, aa 147-187 of the DNA-binding domain (from AFX1).

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This article should be referenced as such:

Bojesen SE. t(X;11)(q13;q23). Atlas Genet Cytogenet Oncol Haematol. 2001; 5(4):291-292.