t(X;11)(q13;q23)

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Identity

Identity

Disease

Described in infants and young children; 4 cases of acute myeloid leukemia (AML) (Pui et al., 1987; Raimondi et al., 1989; Pui et al., 1989; Harrison et al., 1998) and one case of acute lymphoblastic leukemia (ALL) (Smith et al., 1973). With one exception, the FAB types in cases of AML were M4. Peripheral leucocytes at diagnosis of this ALL case were cultured and are presently known as the KARPAS-45 cell line (Karpas et al., 1977). In addition, MLL/AFX1 fusion was confirmed in an AML case with highly complex change originally published involving the Xq22 locus (Nacheva et al., 1982; Parry et al., 1994; Borkhardtet al., 1997).

Note

This translocation has also been found in 2 cases of CLL (Bentz et al., 1995; Kalla et al., 2005). In one case a t(X;11)(q13;q23) was cloned revealing the involvement of BRWD3 gene recently located on Xq21.1 (Kalla et al., 2005).

Phenotype/cell stem origin

Suggested involvement of a pluripotent stem cell or a myeloid progenitor cell; very rarely in lymphoid lineage.

Etiology

No known prior exposure; case of AML M2 developed in a 6 years old male previously treated by chemotherapy and radiotherapy for acute lymphoblastic leukemia (Harrison et al., 1998).

Epidemiology

6 cases to date, children aged 6 months to 5 years, male predominance; sex ratio 4M/2F.

Clinics

From the known data: WBC: 21.6 to 91x10⁹/L, case with a complex t(X;11) associated with fever, enlargement of the liver, spleen and parotid glands, blood in the stool (Karpas et al., 1977); mediastinal mass, dyspnoea, no hepatosplenomegaly, WBC: 5x10⁹/L in T-ALL (Smith et al., 1973).

Prognosis

Survival: poor prognosis; 3 patients died within a year after diagnosis, and one patient died after 24.5 months.

Genetics

Note

Breakpoints difficult to ascertain in suboptimal preparations.
Cytogenetics

Probes

Additional anomalies
Part of a highly complex change in one case; 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

Note
Cloning and characterization of AFX the gene that fuses to MLL in one case of AML and in the leukemic cell line.

AFX1 (All-1 fusion partner on chromosome X, MLLT7)
Location
Xq13
DNA/RNA
AFX consists of two exons and encodes for a protein of 501 amino acids.

Protein
Transcription factor; high degree of homology between AFX1 and the forkhead protein family and highly homologous to the human FKHR protein.

MLL (Mixed lineage leukemia gene, ALL1, HRX, and Hrtx)
Location
11q23
DNA/RNA
The Mixed-Lineage Leukemia gene consists of at least 36 exons, encoding a 3969 amino-acid nuclear protein with a molecular weight of nearly 430 kDa.

Protein
Multidomain molecule; shares homology with the Drosophila trithorax protein; function as a positive regulator of gene expression in embryonic development and hematopoiesis.

Result of the chromosomal anomaly

Hybrid gene
Note
5' MLL - AFX 3' as well as the 5' AFX - MLL 3'.

Fusion protein
Note
Chimeric proteins that contain the N-terminus of MLL; 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).

References

This article should be referenced as such: