t(17;21)(q11.2;q22)

Clinics and Pathology

Disease	acute non lymphoblastic leukemia (ANLL) and myelodysplastic syndromes (MDS); de novo ANLL and treatment related leukemias (t-ANLL)
Phenotype / cell stem origin	one M2, one treatment related RAEBt/M4, one t-ANLL
Etiology	two cases are secondary to treatment with topoisomerase II inhibitors for Hodgkin disease and neuroblastoma
Epidemiology	3 cases to date; 1M/2F, aged 2yrs, 39 yrs and 76 yrs
Prognosis	unknown

Cytogenetics

Additional anomalies	sole anomaly in one case; one case was also -7, +8, one case showed also a t(11;12)(p15;q13) with NUP98 rearrangement

Genes involved and Proteins

Note	the gene involved in 17q11 is unknown; the breakpoint on chromosome 17 is between the loci for NF1 and RARA
Gene Name	AML1
Location	21q22
Dna / Rna	transcription is from telomere to centromere
Protein	contains a Runt domain and, in the C-term, a transactivation domain; forms heterodimers; widely expressed; nuclear localisation; transcription factor (activator) for various hematopoietic-specific genes

Result of the chromosomal anomaly

Hybrid gene Description	5 prime AML1-3 prime unknown; breakpoint in intron 5 or 6 of AML1
Fusion Protein Description	the N-term is provided by AML1, as in the t(3;21) and in the t(8;21)associated with ANLLs, whereas, in the ALL with t(12;21), the fusion protein comprises the C-term part of AML1.

External links

Other database	t(17;21)(q11.2;q22)	Mitelman database (CGAP - NCBI)
Other database	t(17;21)(q11.2;q22)	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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