

t(5;21)(q13;q22)

Clinics and Pathology

Disease myelodysplastic syndrome (MDS) and acute non lymphocytic leukemia (ANLL)

Phenotype / cell stem origin 1 case of refractory anemia with excess of blasts in transformation (RAEB-t), 1 MDS evolving towards a M4-ANLL, 2 M2-ANLL, and 1 ANLL not otherwise specified

Epidemiology 5 cases to date; 3M/2F, aged 58 yr (median, range: 31-75)

Cytogenetics

Cytogenetics sole anomaly in 2 cases, complex karyotypes in 2 other cases.
Morphological

Genes involved and Proteins

Note The gene in 5q13 is yet unknown, and, because cryptic [t\(12;21\) ETV6/AML1](#) are not rare, it is therefore uncertain whether this translocation involve a new AML1 partner

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

External links

Other database [t\(5;21\)\(q13;q22\)](#) [Mitelman database \(CGAP - NCBI\)](#)

Other database [t\(5;21\)\(q13;q22\)](#) [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.

Bibliography

Chromosome pattern, occupation, and clinical features in patients with acute non-lymphocytic leukemia.

Mitelman F, Nilsson PG, Brandt L, Alimena G, Gastaldi R, Dallapiccola B. Cancer Genet Cytogenet 1981; 4: 197-214.

A new translocation, t(5;21)(q13;q22) in acute myelogenous leukemia.

Gogineni SK, da Costa M, Verma RS. Cancer Genet Cytogenet 1996; 88: 167-169.

CBFA2(AML1) translocations with novel partner chromosomes in myeloid leukemias: association with prior therapy.

Roulston D, Espinosa III R, Nucifora G, Larson RA, Le Beau MM, Rowley JD.
Blood 1998; 92:2879-2885.

Combined spectral karyotyping and DAPI banding analysis of chromosome abnormalities in myelodysplastic syndrome.

Kakazu N, Taniwaki M, Horiike S, Nishida K, Tatekawa T, Nagai M, Takahashi T, Akaogi T, Inazawa J, Ohki M, Abe T.
Genes Chromosomes Cancer 1999; 26: 336-345.

Concurrent translocations of MLL and CBFA2 (AML1) genes with new partner breakpoints in a child with secondary myelodysplastic syndrome after treatment of acute lymphoblastic leukemia.

Mathew S, Head D, Rubnitz JE, Raimondi SC.
Genes Chromosomes Cancer 2000; 28: 227-232.
Medline [10825008](#)

Testicular infiltration in acute myeloid leukemia with complex karyotype including t(8;21).

Giagounidis AAN, Hildebrandt B, Braunstein S, Aivado M, Germing U, Heinsch M, Aul C.
Ann Hematol 2002; 81: 115-118

Contributor(s)

Written 02-2003 Jean-Loup Huret

Citation

This paper should be referenced as such :

Huret JL . t(5;21)(q13;q22). Atlas Genet Cytogenet Oncol Haematol. February 2003 .

URL :

<http://www.infobiogen.fr/services/chromcancer/Anomalies/t0521q13q22ID1174.html>

© Atlas of Genetics and Cytogenetics in Oncology and Haematology
