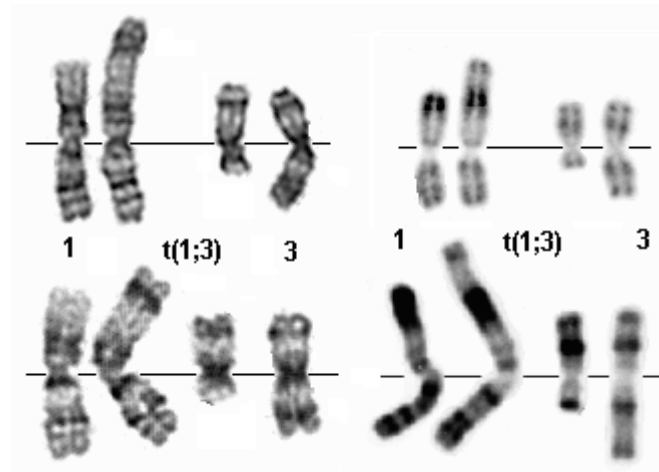


t(1;3)(p36;q21)

Identity



t(1;3)(p36;q21) G-banding (left) - Courtesy Diane H. Norback, Eric B. Johnson, and Sara Morrison-Delap, [Cytogenetics at the Waisman Center](#); R-banding (right) Courtesy Pascale Cornillet-Lefebvre and Stéphanie Struski (above) and Christiane Charrin (below)

Clinics and Pathology

Disease	Myeloid lineage (MDS, ANLL, therapy related ANLL, CML, MPD); features similar to those of the 3q21q26 syndrome including normal or elevated platelet count at diagnosis, megakaryocytic hyperplasia and dysplasia. Very rarely in lymphoid lineage
Phenotype / cell stem origin	of 39 cases, there were: 22 myelodysplastic syndromes (MDS) (17/22 transformed into refractory acute non lymphoblastic leukemia (ANLL) of -M1 or -M4 type), 8 de novo ANLL, 3 therapy-related MDS, 2 polycythemia vera, 1 essential thrombocythemia, 1 chronic myelogenous leukemia (CML), 1 multiple myeloma, 1 waldenstrom's macroglobulinemia
Epidemiology	patients are aged: 30-80 yrs
Clinics	Roughly 50% of patients present with MDS, another 10% with therapy associated MDS, 25% with de novo AML, and the remainder with a range of other myeloproliferative disorders. The majority of MDS patients transform into AML with a short preleukemic phase. Blood data: frequent thrombocytosis or normal platelet count
Cytology	frequently characterized by dysmegakaryocytopoiesis

Pathology	The pathology is typical of MDS, often with a prominent monocytic component. Trilineage dysplasia. Acute leukemias that evolve usually show the morphology of M4 AML.
Treatment	Patients are treated with conventional chemotherapy for AML.
Prognosis	Very poor so far: from 16 cases, median survival was 6 mths in ANLL, 20 mths in MDS

Cytogenetics

Note Other rearrangements showing similar clinical features include [inv\(3\)\(q21q26\)](#), [t\(3;3\)\(q21;q26\)](#), t(3;5)(q21;q31), t(3;8)(q21;q24), and [t\(3;21\)\(q26;q22\)](#). The breakpoints in 3q21 cluster in a 50 kb region centromeric to the breakpoint in inv(3)(q21;q26) and the ribophorin gene (RPN1). The breakpoints at 1p36 are clustered in a 90 kb region at 1p36.3.

Additional anomalies del (5q) in 5 of 20 cases (1/4)

Genes involved and Proteins

Note Mechanisms of Oncogenesis : The available data suggest that transcription of [MEL1](#) (MDS1/EVI1 -like gene) is activated as a result of translocation bringing the gene just 3' to RPN1 gene at 3q21. MEL1 is a 1257 amino acid protein that is homologous (63% similar in amino acid sequence) to EVI. The mechanism of activation of MEL1 is similar to [EVI1](#) that is activated by juxtaposition 3' to RPN1 in the t(3;3)(q21;q26) and 5' to RPN1 in the inv(3)(q21;q26). It appears that MEL1 is normally expressed in uterus and kidney and not in normal hematopoietic cells or in leukemias that lack the t(1;3)(p36;q31). The MEL1 protein contains 2 DNA binding domains (7 C2H2 zinc finger repeats at the amino terminus and 3 zinc finger repeats at the carboxyl terminus). The amino terminal domain of MEL1 contains a PRD domain, a motif also found in the same location in the MDS1/EVI1 protein but not in MDS1). This is of interest because this domain is also found in RIZ, PRDI-BF1, and egl-43 and is homologous to the SET (Suvar3-9, Enhancer of zeste, Trithorax) domain that present in [MLL](#). Inclusion of this domain in EVI1 appears to convert EVI1 from a transcriptional repressor to an activator. Therefore MEL1 may be a transcriptional activator. The target genes of MEL1 have not been identified.

External links

Other database	t(1;3)(p36;q21)	Mitelman database (CGAP - NCBI)
Other database	t(1;3)(p36;q21)	CancerChromosomes (NCBI)

Bibliography

A new translocation , t(1;3)(p36;q21), in myelodysplastic disorders.

Moir DJ, Jones PA, Pearson J, Duncan JR, Cook P, Buckle VJ.
Blood 1984; 64: 553-555.
Medline [6743828](#)

Rearrangements of chromosome 3 involving bands 3q21 and 3q26 are associated with normal or elevated platelet counts in acute non-lymphocytic leukemia.

Bittner MA, Neilly ME, Le Beau MM, Pearson MG, Rowley JD.
Blood 1985; 66: 1362-1370.
Medline [4063525](#)

t(1;3)(p36;q21) in acute nonlymphocytic leukemia: a new cytogenetic-clinicopathologic association.

Bloomfield CD, Garson OM, Volin L, Knuutila S, de la Chapelle A.
Blood 1985; 66: 1409-1413.
Medline [4063527](#)

Diagnostic and prognostic significance of t(1;3)(p36;q21) in the disorders of hematopoiesis.

Welborn JL, Lewis JP, Jenks H, Walling P
Cancer Genet Cytogenet. 1987; 28: 277-285.
Medline [87301329](#)

Acute leukemia with t(1;3)(p36;q21), evolution to t(1;3)(p36;q21) , t(14;17)(q32;q21) and loss of red cell A and Le(b) antigens.

Marsden KA, Pearse AM, Collins GG, Ford DS, Heard S, Kimber RI.
Cancer Genetics Cytogenetics 1992; 64: 80-85.
Medline [1458454](#)

Clinical, haematological and cytogenetic features in 24 patients with structural rearrangements of the Q arm of chromosome 3.

Grigg AP, Gascoyne RD, Phillips GL, Horsman DE
Br J Haematol. 1993; 83: 158-165.
Medline [93168610](#)

Abnormalities of 3q21 and 3q26 in myeloid malignancy: a United Kingdom cancer cytogenetic group study.

Secker-Walker LM, Mehta A, Brain B.
Br J Haematol. 1995; 91: 490-501.
Medline [96027684](#)

The PR domain of the Rb-binding zinc finger protein RIZ1 is a protein binding

interface and is related to the SET domain functioning in chromatin mediated gene expression.

Huang S, Shao G, Limin L.

J Biological Chem 1998; 273: 15933-15939.

Medline [9632640](#)

A novel gene MEL1, mapped to 1p36.3 is highly homologous to the MDS1/EVI1 gene and is transcriptionally activated in t(1;3)(p36;q21)-positive leukemia cells.

Mochizuki N, Shimizu S, Nagasawa T, Tanaka H, Taniwaki M, Yokota J, Morishita K. Blood 2000; 96: 3209-3214.

Medline [11050005](#)

Identification of breakpoint cluster regions at 1p36.3 and 3q21 in hematologic malignancies with t(1;3)(p36;q21).

Shimizu S, Suzukawa K, Kodera T, Nagasawa T, Abe T, Taniwaki M, Yagasaki F, Tanaka H, Fujisawa S, Johansson B, Ahlgren T, Yokota J, Morishita K.

Genes Chromosom Cancer 2000; 27: 229-238.

Medline [20146274](#)

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URL : <http://www.infobiogen.fr/services/chromcancer/Anomalies/t0103.html>

Cornillet-Lefebvre P, Daliphard S, Struski S . t(1;3)(p36;q21). Atlas Genet Cytogenet Oncol Haematol. November 2000 .

URL : <http://www.infobiogen.fr/services/chromcancer/Anomalies/t0103.html>

Hess JL . t(1;3)(p36;q21). Atlas Genet Cytogenet Oncol Haematol. May 2002 .

URL : <http://www.infobiogen.fr/services/chromcancer/Anomalies/t0103.html>

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