Atlas of Genetics and Cytogenetics
in Oncology and Haematology

OPEN ACCESS JOURNAL INIST-CNRS

Leukaemia Section
Short Communication

t(2;3)(p16;q26) BCL11A/MECOM

Jean-Loup Huret

Genetics, Dept Medical Information, University of Poitiers, CHU Poitiers Hospital, F-86021 Poitiers, France (JLH)

Published in Atlas Database: September 2013

Online updated version : http://AtlasGeneticsOncology.org/Anomalies/t0203p16q26ID1664.html
DOI: 10.4267/2042/53539

Abstract

Review on t(2;3)(p16;q26) BCL11A/MECOM, with data on clinics, and the genes implicated.

Identity

Note
This translocation is found in a subset of cases described in the card t(2;3)(p15-23;q26-27).
Other subsets involve other genes, such as THADA in the t(2;3)(p21;q26) THADA/MECOM.

Clinics and pathology

Disease

Acute myeloid leukemia (AML)

Phenotype/cell stem origin

There were one M1-AML, two M2-AMLs, and one M5-AML; patients presented with dysplasia of at least two myeloid cell lineages.

Epidemiology

Four cases to date; there were three male and one female patients; patients were aged 36, 36, 36, and 55 (in years) (Trubia et al., 2006).

Prognosis

Clinical outcome in cases with the t(2;3)(p16;q26) BCL11A/MECOM and the case with the t(2;3)(p21;q26) THADA/MECOM (plotted together) was severe: "One patient is alive with active disease at 12 months, five patients died after 4-14 months" (Trubia et al., 2006).

Genetics

Note
MECOM was overexpressed.

Cytogenetics

Cytogenetics morphological

The t(2;3)(p16;q26) was the sole anomaly in two of four cases (at least in a subclone), accompanied with -7 in one case, and +14 in another case.

Genes involved and proteins

BCL11A

Location
2p16

Protein
BCL11A is a Krüppel zinc-finger transcription factor, which has been shown to be essential for pre-B-cell development, thymocyte maturation, and globin switching, expressed in haematopoietic and neural tissues. BCL11A controls FLT3 and IL7R expression in early hematopoietic progenitors (Wu et al., 2013).

MECOM

Location
3q26

Note
MECOM is also known as EVI1 or PRDM3; MECOM symbol means: "MDS1 and EVI1 complex locus".

Protein

"EVI1" contains two domains of seven and three zinc finger motifs, respectively, a repression domain between the two sets of zinc fingers, and an acidic domain at its C-term. Sequence specific DNA binding protein.

Interacts with transcriptional coactivators, corepressors, and other sequence specific transcription factors. MECOM ("MDS1-EVI1") also contains a PR domain from "MDS1" in N-term (Wieser, 2008).

Result of the chromosomal anomaly

Hybrid gene

Description

Regulatory elements were transferred at the 5' of MECOM.

Fusion protein

Description

The t(2;3) brings about the juxtaposition at 3q26 of the MECOM locus with regulatory elements normally located in proximity of the 2p breakpoints, with consequent EVI1 overexpression, without the formation of a fusion protein.

References

Trubia M, Albano F, Cavazzini F, Cambrin GR, Quarta G, Fabbiano F, Ciambelli F, Magro D, Hernandezo JM, Mancini M, Diverio D, Pelicci PG, Coco FL, Mecucci C, Specchia G, Rocchi M, Liso V, Castoldi G, Cuneo A. Characterization of a recurrent translocation t(2;3)(p15-22;q26) occurring in acute myeloid leukaemia. Leukemia. 2006 Jan;20(1):48-54

Wieser R.. MECOM (Ecotropic Viral Integration Site 1 (EVI1) and Myelodysplastic Syndrome 1 (MDS1)-EVI1). Atlas Genet Cytogenet Oncol Haematol. 2008;12(4):306-310. http://documents.irevues.inist.fr/bitstream/handle/2042/38551/12-2007-EVI103q26ID19.pdf?sequence=2

Wu X, Satpathy AT, Kc W, Liu P, Murphy TL, Murphy KM.. Bcl11a controls Flt3 expression in early hematopoietic progenitors and is required for pDC development in vivo. PLoS One. 2013 May 31;8(5):e64800. doi: 10.1371/journal.pone.0064800. Print 2013.

This article should be referenced as such:

Huret JL. t(2;3)(p16;q26) BCL11A/MECOM. Atlas Genet Cytogenet Oncol Haematol. 2014; 18(4):276-277.