

# Leukaemia Section

## Short Communication

## inv(3)(p24q26) ?/MECOM

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### Abstract

Review on inv(3)(p24q26), with data on clinics, and the genes involved.

#### Keywords

chromosome 3; inv(3)(p24q26); MECOM

### Clinics and pathology

#### Note

Ten cases available, but with almost no data (Haferlach et al., 2012).

#### Disease

##### Phenotype/cell stem origin

Out of ten cases, there were eight acute myeloid leukemia (AML) cases, and two myelodysplastic syndromes (MDS)

##### Epidemiology

inv(3)(p24q26) represented about 1% of a cohort of 606 AML and 377 MDS patients with normal karyotypes (n = 594) or chromosome 7 alterations (-7/7q-; n = 389). Median age was 60 years (range 20-76)

##### Cytogenetics

Cryptic rearrangement.

##### Prognosis

Survival outcomes in 22 patients with cryptic MECOM rearrangements (inv(3)(p24q26), t(3;21)(q26;q11), and der(7)t(3;7)(q26;q21) altogether) and were compared with inv(3)(q21q26)/t(3;3)(q21;q26) cases. Median overall survival was 9.4 months in the subgroup

with cryptic MECOM rearrangements which was not significantly different from the 21.8 months in patients with an inv(3)(q21q26)/t(3;3)(q21;q26)

### Genes involved and proteins

#### Note

The partner of MECOM is unknown.

#### MECOM (Ecotropic Viral Integration Site 1 (EVI1) and Myelodysplastic Syndrome 1 (MDS1-EVI1))

#### Location

3q26.2

#### Note

MECOM is a nuclear transcription factor that plays an essential role in the proliferation and maintenance of hematopoietic stem cells and can inhibit myeloid differentiation. Two alternative forms exist, one generated from EVI1, the other MECOM (MDS1 and EVI1 complex locus) through intergenic splicing with MDS1 (myelodysplasia syndrome 1), a gene located 140 kb upstream of EVI1.

#### Protein

The protein encoded by this gene is a transcriptional regulator involved in cell differentiation and proliferation, and apoptosis. The encoded protein can interact with transcriptional coactivators (P/CAF, CBP) and corepressors (CTBP1, HDAC) as well as other transcription factors (GATA1, Smad3) (de Braekeleer et al., 2012)

## Result of the chromosomal anomaly

### *Fusion protein*

#### **Oncogenesis**

Increased MECOM expression was noted.

## References

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