t(8;22)(p11;q13)

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Identity

Note
Two distinct clinical syndromes have been associated with the 8p11-p12 region:
Stem-cell myeloproliferative disorder with FGFR1 rearrangement.
AML M4 or M5 erythrophagocytosis-associated, with MOZ rearrangement.
The t(8;22)(p11;q13) involves MOZ.
The partners of 8p11 are 8q13, 14q11, 16p13, 19q13, 22q13 and 3q27, 17q12 in a complexe translocation t(3;8;17)(q27;p11;q12).

Clinics and pathology

Disease
Acute myelomonocytic or monocytic leukemia (M4, M5a, M5b) associated with erythrophagocytosis by blasts noted to various degree, one case is probably a therapy-related leukemia.

Epidemiology
Rare.

Cytology
Erythrophagocytosis by blasts cells is occasionally found but not marked.

Prognosis
Probably poor.

Cytogenetics

Cytogenetics morphological

Additional anomalies
In one case association with trisomy 8.

Genes involved and proteins

MOZ

Location
8p11

Note
MOZ contains a LAP (Leukemia associated protein) zinc finger domain, a HAT domain (Histone acetyltransferase) and a acidic domain. Detection by FISH: YAC 176C9.

Protein
ZNF220

Monocytic leukemia zinc finger protein 2004 amino acids and 225 kDa nuclear protein, with 2 PHD-type zinc fingers. MOZ is a histone acetyltrans-ferase (HAT) and the founding member of the MYST family of HATs, a family that includes proteins involved in cell cycle regulation, chromatin remodeling and dosage compensation. MOZ plays an important role during hematopoiesis with his transcriptional coregulator activity.

P300

Note
Detection: FISH with the bacterial artificial chromosome clone H59D10.

Protein
Adenoviral E1A-associated protein p300 with acetyltransferase activity.

Result of the chromosomal anomaly

Hybrid gene

Note
Gene fusion MOZ/P300.
**Description**

MOZ is disrupted within the sequences encoding the acidic domain. Both fusion transcripts are expressed. The t(8;22) breakpoints occur in MOZ codon 1117 within an exon of 4 kb.

**Fusion protein**

**Description**

MOZ-p300 fusion is similar to MOZ-CBP described in the t(8;16) but involve p300 instead of CBP. The translocation creates in-frame fusion proteins (MOZ-p300 and p300-MOZ). The two fusion proteins retain the N-terminus portion of MOZ including the HAT domain.

**Oncogenesis**

How the MOZ-p300 fusion protein is involved in acute leukemia is not known, but it probably affects the chromatin condensation.

**References**


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