inv(16)(p13q22)
t(16;16)(p13;q22)
del(16)(q22)

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Identity

Note: the three chromosome anomalies are variants of each other, and they share identical clinical features.

Clinics and pathology

Disease
ANLL/MDS

Phenotype / cell stem origin
Nearly pathognomonic of M4eo: M4 with marked eosinophilia; rarely: M2 or M5, M4 without eo, MDS; known cases of BC-CML with M4eo phenotype and inv(16). CD2 (T-cell marker) may be co-expressed.

Epidemiology
5% of ANLL, 20% of M4.

Clinics
CNS involvement is frequent, according to some authors.

Cytology
Most often: eosinophils > 5%, with large immature basophilic granules, NASCA+, in the bone marrow (but normal in blood: this M4 do not show the ‘eo’ characteristic in blood).

Prognosis
High CR rate; better prognosis than most other ANLL; median survival may be 5 yrs.

Cytogenetics

Cytogenetics, morphological
May be overlooked, especially with R-banding; best seen without banding procedure (‘giemsa’) for some workers.

Additional anomalies
None in 2/3 of cases; +8, +22 in 15% each, del(7q), +21.

Variants
Are known:
1- t(16;16)(p13;q22); - del(16)(q22): may be associated with less typical phenotype and preceding MDS, older age, complex karyotype, worse prognosis;
2- but also: translocations of 16q22 with: 1p31-32, 3q21, 5q33, associated with eosinophils anomalies.

Genes involved and Proteins

MYH11
Location: 16p13
Protein
Contains a N-term ATPase head responsible for actin binding and mechanical movement, and a C-term long repeat of coil-coil domain to facilitate filament aggregates; member of the myosin II family.

CBFb
Location: 16q22
Protein
Subunit of the transcription factor complex CBF; CBFb by itself does not contain any DNA binding motif or transcriptional activation domain, but forms a dimer with CBFa: → transcription factor.

Results of the chromosomal anomaly

Hybrid gene
Description
Breakpoint in CBFB intron 5.
Transcript
No reciprocal MYH11-CBFB.

Detection protocol
RT-PCR.

Fusion protein

Description
N-term -the first 165 (or 133 in a few cases) amino acids of CBFb, removing only 17 or 22 amino acids to the tail of MYH11- C-term; also variable breakpoint in MYH11; identical in the cases of RAEBT and BC-CML.

Expression localisation
Localized in the nucleus.

References


This article should be referenced as such: