t(3;8)(q26;q24)

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Phenotype / cell stem origin
Mostly AML FAB-M2 or FAB-M-4 subtype.

Etiology
Unclear, may be secondary to chemotherapy.

Epidemiology
10 cases reported so far in the literature, less than 1% of AML cases.

Cytology
Acute myeloid leukemia of mostly M2, M4 or M5 FAB subtype or high grade MDS. Marked trilineage dysplasia and megakaryocytic hyperplasia, may be associated with peripheral blood thrombocytosis giving the so-call 3q21q26 syndrome.

Treatment
Chemotherapy; may responds to thalidomide or arsenic better than conventional chemotherapy.

Evolution
Myelodysplastic syndrome progress to acute myeloid leukemia.

Prognosis
Poor.

Identity

Clinics and pathology

Disease
Acute myeloid leukemia, de novo myelodysplastic syndrome or therapy related myelodysplastic syndrome.
Dysplastic myeloid elements.

Increased dysplastic megakaryocytes and increased blasts in the interstitium.
Cytogenetics

Note: The breakpoint on 3q26 may lie in EVI1 or MDS1 genes. The breakpoint on 8q24 is distal to the PVT1 gene, a MYC activator gene in mice. The t(3;8) is frequently associated with -7. It also can be an isolated finding.

Probes
EVI1/MDS1
RP11-115B16, RP11-114D6; Vysis C-MYC; Break-apart distal; Probe (green).

Genes involved and Proteins
EVI1/MDS1

Location: 3q26.2

Note: Aberrant EVI1 expression usually occurs in AML, MDS or CML-BC as a result of translocation involving 3q26. The most common ones are inv(3)(q21q26), t(3;3) and t(3;21)(q26;q22). The partner genes of EVI1 are identified as Ribophorin I in inv(3)(q21q26) and t(3;3), AML/ MDS1 /EAP in t(3;21), and ETV6 in t(3;12), respectively. Others involving t(3;12), t(2;3)(p13;q26), t(3;17)(q26;q22) and t(3;13)(q26;q13-14) are uncommon. Aberrant EVI1 expression also occurs in 10% of acute myeloid leukemia without involving 3q26 and is also correlated with an adverse outcome.

DNA / RNA
16 exons spanning 64.2 Kb. Transcriptional orientation is from telomere to centromere. EVII gene may be transcribed in different isoform which may have different oncogenic effect.

Protein
1051 amino acids; 118335 Da. Nuclear location, contains 10 C2H2-type zinc fingers.

PVT1/C-MYC (pvt-1 (murine) oncogene homolog, MYC activator)

Location: 8q24

Note: The RNA function of pvt1 is unknown.

References


This article should be referenced as such: