

t(16;21)(p11;q22)

Clinics and Pathology

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|------------------------------|--|
| Disease | de novo acute non lymphocytic leukemia (ANLL); to be noted is one case of chronic myelogenous leukemia (CML) -blast crisis. |
| Phenotype / cell stem origin | ANLL cases: mainly M1, M2, M4, M5a, M5b, or M7 ANLL; may be preceded by a myelodysplastic syndrome (MDS). |
| Epidemiology | about 40 reported cases, mainly found in young adults; children cases are described; median age is about 30 yrs; balanced sex ratio |
| Clinics | blood data: anemia, thrombocytopenia, mild hyperleucocytosis; with high monocytic cell count at times |
| Cytology | myelocytic and monocytoid features are often present; eosinophils in the bone marrow are sometimes abnormal and/or elevated; erythrophagocytosis may be found |
| Prognosis | seems poor: complete remission may not be achieved; there is high incidence of relapse within a year and a median of survival is about 22 months (cases herein reviewed) |

Disease [Ewing tumours](#)

Note t(16;21)(p11;q22) has been found in rare cases of Ewing tumours, a paediatric neoplasm with small round-cells derived from neural crests cells usually associated with translocations involving [EWSR1](#)

Cytogenetics Ewing tumours are usually associated with a t(11;22)(q24;q12) with 5' [EWSR1](#) - 3' FLI1 involvement, less often associated with t(21;22)(q22;q12) with 5' EWSR1 - 3' ERG involvement, rarely associated with t(2;22)(q36;q12) (5' EWSR1 - 3' FEV) or with t(17;22)(q21;q12) (5' EWSR1-3' ETV4)

Prognosis recent treatments have improved the prognosis of Ewing's tumours. The prognosis is mainly determined by the presence of metastases at the time of diagnosis

Cytogenetics

Additional anomalies ANLL cases: found solely in about 60% of cases in at least a subclone; associated with [+10](#), [+8](#), or [de\(9q\)/-9](#) in about 10% of cases each

Genes involved and Proteins

Gene [FUS](#)

Name

Location 16p11

Protein RNA binding protein; member of the TET family, like EWSR1

Gene Name

ERG

Location 21q22

Protein ETS transcription factor

Result of the chromosomal anomaly**Hybrid gene**

Description

5' FUS including exons 1 to 6, 7 or 8 - 3' ERG from exon 7, 8 or 9 to C-term.

Fusion Protein

Description

N-term FUS transactivation domain fused to the C-term DNA binding ETS domain of ERG

Oncogenesis seems to act as a transcriptional activator

External links

Other

database

[t\(16;21\)\(p11;q22\)](#)

[Mitelman database \(CGAP - NCBI\)](#)

Other

database

[t\(16;21\)\(p11;q22\)](#)

[CancerChromosomes \(NCBI\)](#)

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