Atlas of Genetics and Cytogenetics in Oncology and Haematology

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Solid Tumour Section

Mini Review

t(16;21)(p11;q22)

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Clinics and pathology

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.

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).

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

FUS

Location

16p11

Protein

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

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.

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