t(11;21)(q13;q22)

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Clinics and pathology

Disease
Myeloid malignancies

Epidemiology
Three cases of t(11;21)(q13;q22) in myeloid malignancies are available to date: a 63-year-old female patient with AML evolving from myelodysplastic syndrome (MDS) (Imagama et al., 2007), a 65-year-old male patient with a M2 acute myeloid leukemia (M2-AML) (Dai et al., 2007), and a 70-year-old male patient with a chronic myelogenous leukemia (CML). The t(11;21) was found during transformation into blastic crisis (BC-CML) and in the subsequent reversion to the chronic phase (Wang et al., 1988).

Prognosis
The MDS-to-AML case relapsed two years after remission. The M2-AML case died 10 months after diagnosis. The CML case was remaining in remission of blast crisis for 4 months at the time of the report.

Cytogenetics

Cytogenetics morphological
The t(11;21) was part of a complex karyotype in the MDS-to-AML case, found with an additional anomaly in the M2-AML case, and accompanying the classical t(9;22)(q34;q11) in the CML case.

Genes involved and proteins

Note
The involvement of RUNX1 was proved in the two AML cases; the involvement of MACROD1/LRP16 as the partner of RUNX1 was established in the case described by Imagama et al.

MACROD1
Location
11q13.1
Note
MACROD1 is also known as LRP16.

Protein
MACROD1/LRP16 gene has been characterized as an estrogen-responsive gene. LRP16 is required for ERalpha signaling transduction by functioning as an ERalpha coactivator (Han et al., 2007; Han et al., 2008). MACROD1/LRP16-overexpression promotes the cell cycle, and cell proliferation (Yang et al., 2009).

RUNX1
Location
21q22
DNA/RNA
Transcription from telomere to centromere.

Protein
Contains the RUNT binding domain at 5' portion and the transactivation domain at 3' portion. Forms heterodimers; widely expressed; nuclear localization; a transcription factor and critical regulator of hematopoietic-cell development.
Result of the chromosomal anomaly

Hybrid gene

Description
In the case described by Imagama et al. 2007, the translocation fuses RUNX1 exon 5 or exon 6 to MACROD1 exon 2, suggesting that the RUNX1 breakpoint lies in intron 6 and that alternative fusion splice variants are generated. The reciprocal MACROD1-RUNX1 fusion was also detected.

References


This article should be referenced as such: