t(8;19)(p12;q13)

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Clinics and pathology

Disease
Acute myeloid leukemia, M0 type (M0 AML)

Epidemiology
Only one case to date, 70-year-old male patient.

Prognosis
The patient died 21 months after diagnosis.

Genes involved and proteins

FGFR
Location
8p12
Protein
FGF receptor; membrane associated tyrosine kinase. Signal transduction.

ERV/HERV-K
Location
19q13
Note
ERVK/HERV-K are disseminated throughout the whole genome; one of these, located in 19q13, was found implicated in the t(8;19).

Protein
ERV/HERV sequences are thousands of endogenous retroviruses. Most -if not all- are defective, containing deletions or nonsense mutations. The ERVK/HERV-K family is the most recently inserted family, after chimpanzees and men diverged. ERV element consists of two identical, nontranslated long terminal repeats (LTRs) flanking an internal region that encodes proteins required for viral replication and assembly. Defective ERV have lost their internal region and LTRs often remain solos.

These retroelements (RE) could be agents of genomic instability. They can cause host DNA rearrangements due to recombination events, by transduction of RE flanking sequences into new genomic loci, by creating pseudogenes, or by causing RNA recombination. The HERV-K subgroup has been suspected to be involved in cancer (including seminomas), autoimmune diseases, and neuronal diseases such as schizophrenia.

Result of the chromosomal anomaly

Hybrid gene
Description
5’ sequences from an ERV element - 3’ FGFR1 (starting at exon 9).

Fusion protein
Description
Open reading frame from ERV sequences fused to part of the juxtamembrane domain and the tyrosine kinase-encoding regions of the FGFR1 gene.

References


Belshaw R, Dawson AL, Woolven-Allen J, Redding J, Burt A, Tristem M. Genomewide screening reveals high levels of...


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