Leukaemia Section
Mini Review

\( t(12;22)(p13;q12) \)
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Identity

Schematic representation of the reciprocal \( t(12;22)(p13;q12) \) translocation producing the EWSR1-ZNF384 fusion gene.

Clinics and pathology

**Disease**
Acute leukemias.

**Phenotype / cell stem origin**
Pro-B acute lymphoblastic leukemia with expression of myeloid antigens; biphenotypic leukemia.

**Prognosis**
Relatively good prognosis.

Genes involved and Proteins

**ZNF384 (Zinc Finger protein 384)**

**Protein**
Belongs to the Kruppel C2H2-type zinc-finger protein family; contains 8 C2H2-type zinc fingers. Nucleocytoplasmic shuttling protein and transcription factor.

**EWSR1 (Ewing sarcoma region 1)**

**Location:** 22q12

Results of the chromosomal anomaly

**Hybrid gene**

**Description**

5' EWSR1 - 3' ZNF384
Schematic representation of the EWSR1-ZNF384 fusion protein. SYQG, Ser-Tyr-Gln-Gly transactivating domain; LZ, leucine-rich domain; SR, serine rich domain; PR, Proline rich domain; NLS, Nuclear Localization signal; ZFs, Kruppel-type C2H2 zinc finger domains; QA, Gln-Ala repeat (see also Martini et al., 2002).

References


This article should be referenced as such: