

Leukaemia Section

Mini Review

t(12;22)(p13;q12)

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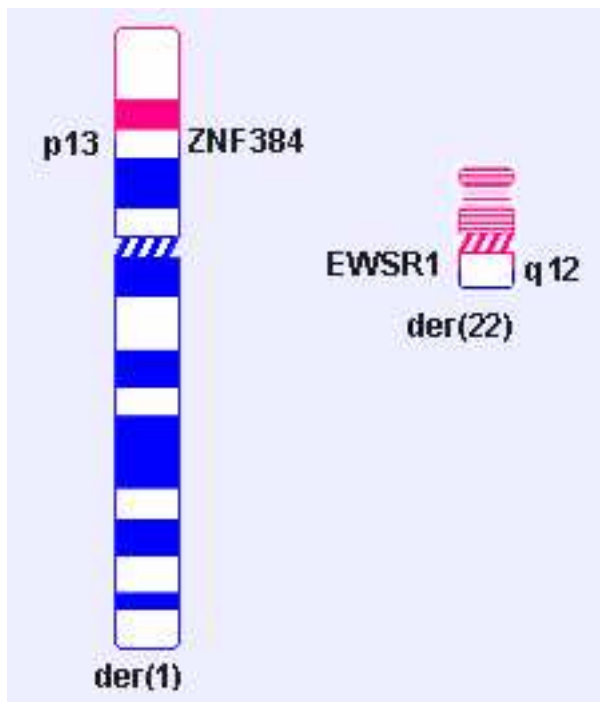
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Identity



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).

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