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Leukaemia Section
Short Communication

inv(19)(p13q13) TCF3/TFPT, t(19;19)(p13;q13) TCF3/TFPT

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Clinics and pathology

Disease

Childhood pre-B cell acute lymphoblastic leukemia.

Note

There is no clinical data in the only paper describing the fusion TCF3-TFPT, nor cytogenetic indications (Brambillasca et al., 1999); inv(19)(p13q13) has also be found in 3 cases of hairy cell leukemia (Haglund et al., 1994); the genes involved in these three patients are unknown; this inv(19) is likely to represent another entity.

Cytogenetics

Cytogenetics morphological

This chromosome rearrangement is cryptic.

Genes involved and proteins

TCF3

Location

19p13.3

Protein

TCF3, better known as E2A, is a member of the basic helix-loop-helix (bHLH) 1 family of transcription factors that are ubiquitously expressed during development.

TFPT

Location

19q13.4

Protein

Role in cell cycle inhibition and apoptosis.

Result of the chromosomal anomaly

Hybrid gene

Description

5' TCF3 - 3' TFPT; the translocation joins TCF3 exon 13 or 14 to part of TFPT; the junction is in frame in some cases, and out of frame in others. The reciprocal transcript was not found.

Fusion protein

Description

Variable junction between TCF3 and TFPT, retaining the transactivation domain of TCF3, but with a truncation in TFPT, due to the frequent occurrence of a stop codon.

References

Haglund U, Juliusson G, Stellan B, Gahrton G. Hairy cell leukemia is characterized by clonal chromosome abnormalities clustered to specific regions. Blood. 1994 May 1;83(9):2637-45

Brambillasca F, Mosna G, Colombo M, Rivolta A, Caslini C, Minuzzo M, Giudici G, Mizzi L, Biondi A, Privitera E. Identification of a novel molecular partner of the E2A gene in childhood leukemia. Leukemia. 1999 Mar;13(3):369-75

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