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Leukaemia Section
Short Communication

t(5;14)(q35;q11)

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Clinics and pathology

Disease

T-cell acute lymphoblastic leukemia (T-ALL).

Epidemiology

In only one case of t(5;14)(q35;q11) was the involvement of NKX2-5 and TRD ascertained. It was the case of a 3-year-old male patient with a common T-ALL, herein described (Przybylski et al., 2006). Another case of t(5;14)(q35;q11), but without NKX2-5-TRD ascertainement, is the case of a 45-year-old male patient with an acute monoblastic leukemia (FAB type M5). In this second case, the t(5;14) appears to be secondary to a t(6;11)(q27;q23). Although molecular studies were not available at that time, it is likely that the patient, who have had a history of possible toxic exposure, who presented with a high blood count (above $50X10^9$/l), and who did not enter complete remission, had a MLL-MLLT4 hybrid gene (Welborn et al., 1993).

Genes involved and proteins

TRD

Protein

T cell receptor.

NKX2-5

Location

5q35.2

Protein

Homeodomain protein; belongs to the NK2/NKX family. Transcriptional activator; regulates beta-catenin and GATA4 in human cardiomyocytes (Riazi et al., 2009). Involved in cardiac morphogenesis. NKX2-5 mutations may cause congenital heart diseases, although it does not appear to be the main aetiologic factor (Draus et al., 2009).

Result of the chromosomal anomaly

Fusion protein

Description

The break on chromosome 5 occurred between NKX2-5 and BNIP1, and downstream the D3 diversity segment of TRD on chromosome 14. NKX2-5 was placed under influence of the TRD enhancer, resulting in strong ectopic NKX2-5 expression.

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