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 ascertainment, 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).

**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.

**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).

**References**


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