Leukaemia Section
Mini Review

**t(12;13)(p13;q14)**

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**Clinics and pathology**

**Disease**

Acute lymphoblastic leukemia (ALL) and myeloid malignancies.

**Note:** This translocation is likely to be molecularly heterogeneous.

**Epidemiology**

Only one case to date with identification of ETV6 and TTL as genes involved in the translocation: a 46 year-old male patient with ALL (blasts were CD10+/-, CD19+) (Qiao et al., 2003).

**Clinics**

Altogether, 15 cases are available:

- Pre-B or early pre-B ALL cases, with an unbalanced sex ratio (7M/2F), and a median age of 6 years (range: 2-46), 7 of 9 patients being children (Raimondi et al., 1989; Pui et al., 1991; Raimondi et al., 1991; Chan et al., 1994; Kobayashi et al., 1994; Raimondi et al., 1997; Coignet et al., 1999; Qiao et al., 2003).
- Myeloid cases: 1 chronic myelomonocytic leukaemia (CMML), 1 chronic myelogenous leukaemia in blast crisis (BC-CML), 1 M3 acute myeloid leukaemia (AML), 1 M0-AML, and 2 treatment related AML (t-AML). In contrast with lymphoid cases, the sex ratio was balanced (3M/3F), and median age was 58 years (range: 51-70) (Zitzelsberger et al., 1990; Abeliovich et al., 1993; Fugazza et al., 1997; Tosi et al., 1998; Castro et al., 2000; Temperani et al., 2000).

**Prognosis**

The patient with ETV6/TTL hybrid gene attained complete remission, but relapsed and died of refractory disease 4 years after diagnosis.

**Cytogenetics**

**Additional anomalies**

The t(12;13) accompanied a del(5q) in two (t-AML) cases, del(7q) in one of these two cases, +8 in one myeloid case, t(9;22)(q34;q11) in the BC-CML case, a PML / RARA hybrid without any apparent t(15;17) in the M3 case, an additional 21 in two ALL cases (+21 once, i(21q) once). The t(12;13) was the sole anomaly in four cases (2 ALL and two AML).

**Genes involved and Proteins**

**Note:** ETV6 was found implicated in 4 ALL cases and 1 AML case; most other cases have been published before ETV6/TEL was known.

**ETV6**

**Location:** 12p13

**Protein**

HLH domain responsible for hetero- and homodimerization in N-term, and an ETS domain responsible for sequence specific DNA-binding in C-term. Transcriptional regulator; involved in bone marrow hematopoiesis.

**TTL**

**Location:** 13q14

**Protein**

This gene/protein remains poorly known: there has been no study on it since the princeps paper by Qiao et al. (2003).
Results of the chromosomal anomaly

Hybrid gene

Description
Both reciprocal transcripts, TTL/ETV6 and ETV6/TTL, were detected. ETV6/TTL fusion transcript comprises 5’ ETV6 exon 1, fused to TLL exon 9, resulting in a potential 31 amino acids peptide. The other transcript, TTL/ETV6, comprises 5’ TLL exons 1 to 5 or to 8a, fused to ETV6 from exon 2. The predicted 530 amino acids fusion protein consists mostly of ETV6 with both HLH and ETS domains, and could have modified transcriptional activities. On the other hand, a loss of function of ETV6 and/or of TLL could play the critical role in leukemogenesis.

References


This article should be referenced as such: