**Leukaemia Section**

**Mini Review**

**t(7;14)(p15;q11)**

Julie Bergeron, Elizabeth Macintyre, Vahid Asnafi

Laboratoire d’Hématologie and INSERM EMI0210, Hôpital Necker-Enfants Malades, Université Paris-Descartes, AP-HP, Paris, France

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**Disease**

T- Acute lymphoblastic leukemia (T-ALL).

**Phenotype / cell stem origin**

T lineage TCR gamma delta +, CD4/8 double positive (DP), CD1a- immunophenotype.

**Epidemiology**

1 case reported.

**Clinics**

FABL1 or L2. The index case had hepatosplenomegaly without mediastinal involvement.

**Cytogenetics**

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**Cytogenetics, molecular**

Balanced t(7;14).

Der(7): Intrinsic region of HOXA locus on 7p15 between HOXA6 and HOXA7 genes fused with Jd1 segment of TCRD on 14q11.

Der(14): DREC segment on chromosome 14q11 rearranged with Dd2 and Dd3 segments and fused to the telomeric part of HOXA locus on 7p15.

**Additional anomalies**

This case also expressed (by RQ-PCR) a CALM-AF10 fusion transcript ((t(10;11)(p13;q14-21)).

**Variants**

Variant translocation cases are reported: 9 cases of T-ALLs having the HOXA locus translocated to TCRB in a t(7;7). The breakpoints on 7p15 in those HOXA-TCRB cases are more centromeric, close to HOXA9.

**Genes involved and Proteins**

**HOXA (intrinsic region)**

**Location:** 7p15

**Note:** HOXA6 and HOXA7 lie at 6,9kb from each other on 7p15.

**Protein**

Various HOXA genes act as transcription factors playing important roles in the differentiation and commitment processes of embryonic and hematopoietic cells.

**TCRD**

**Location:** 14q11

**Note:** Breakpoint on der(7) lie 5’ from Jd1. Breakpoint on der(14) lies 12 nucleotides 5’ of the 3’ end of the DREC segment.

**Protein**

Protein encoded by the TCRD locus are the T-cell receptor chains.

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**Fig1:** FISH hybridization result using a TCRA/D distal (Green) and HOXA proximal (orange) FISH probes showing a fusion signal in 6 of 8 mitosis.
Results of the chromosomal anomaly

**Hybrid gene**

Germline chromosome 14

Fig2: Sequence of events leading to the final translocation. Exons are represented by boxes. Triangles represent RSS and show their orientation. E=enhancer.

**Fusion protein**

![Diagram](image)

Fig3: The nucleotide sequence of both derivatives implicated in the t(7;14) translocation. Underscored are RSS or RSS-like sequence in the vicinity of the breakpoints. In lower case letters: non templated nucleotides at the junction.

**Description**

No fusion protein. Overexpression of HOXA genes as a result of the translocation with TCRD was expected, as it was demonstrated to be the case in HOXA-TCRB T-ALLs. However this case had a CALM-AF10 fusion in the same leukemic clone. CALM-AF10 is already known to be associated with HOXA cluster global overexpression. The HOXA pattern of expression in this case was similar to other CALM-AF10 T-ALL.

**Oncogenesis**

Probable, as several HOX/HOXA genes have been implicated in leukemic processes.
References


This article should be referenced as such: