Leukaemia Section

Mini Review

\textbf{t(1;11)(q21;q23)}

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\textbf{Identity}

\begin{center}
\includegraphics[width=\textwidth]{t111.png}
\end{center}

\textbf{t(1;11)(q21;q23) G- banding: - Courtesy Charles D. Bangs.}

\textbf{Clinics and pathology}

\textbf{Disease}

ANLL, ALL at times.

\textbf{Phenotype / cell stem origin}

30 cases were reported. 25 of them were ANLL: mostly M4/M5 (16), 2 M1, 1 M2, 3 secondary (s) LAM, in 3 cases FAB type were not described. The other cases were: 2 ALL, 1 biphenotypic ALL, 1 sALL and 1 sMSD.
**Epidemiology**
Most cases were infants (10/23) and children (7/23), range is 4 months - 62 years, balanced sex ratio (14F/12M on 26 cases).

**Prognosis**
Yet unknown.

**Cytogenetics**

**Cytogenetics morphological**
Presents as der(11)t(1;11)(q21;q23) in 9 of the 30 cases. Unbalanced form is identified in the 4 ALL and in all of the secondary cases.

**Additional anomalies**
Balanced translocation is present as sole anomaly in 16/21 cases, and as part of simple karyotype in 5/21 cases; +19, +22 were recurrent. Additional abnormalities were observed in 8 of the 9 cases showing the der(11); karyotype of 5 cases were highly complex.

**Variants**
Two three-way translocations were identified: t(1;11;3)(q21;q23;q21) and t(1;11;4)(q21;q23;p16).

**Genes involved and Proteins**

**AF1q**
- **Location:** 1q21
- **DNA / RNA:** 1.8 kb mRNA.
- **Protein:** 9 kDa.

**MLL**
- **Location:** 11q23
- **DNA / RNA:** 21 exons, spanning over 100 kb; 13-15 kb mRNA.
- **Protein:** 431 kDa; contains two DNA binding motifs (a AT hook, and Zinc fingers), a DNA methyl transferase motif, a bromodomain; transcriptional regulatory factor; nuclear localisation transcriptional regulatory factor; nuclear localisation.

**Results of the chromosomal anomaly**

**Hybrid gene**
- **Description:** 5’ MLL - 3’ AF1q; breakpoints: between exons 6 and 7 in MLL and within the 5’ untranslated region in AF1q.

**Fusion protein**
- **Description:** N-term -- AT hook (DNA binding) and DNA methyltransferase motif from MLL fused to the entire AF1q on the der(11); the reciprocal on der(1) is out of frame.

**References**

Tse W, Zhu W, Chen HS, Cohen A. A novel gene, AF1q, fused to MLL in t(1;11) (q21;q23), is specifically expressed in leukemic and immature hematopoietic cells. Blood 1995 Feb 1;85(3):650-6.


This article should be referenced as such: