t(11;11)(q13;q23)

Jean-Loup Huret

Genetics, Dept Medical Information, University of Poitiers, CHU Poitiers Hospital, F-86021 Poitiers, France (JLH)

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Clinics and pathology

Epidemiology

The involvement of MLL in 11q23 and ARHGEF17 in 11q13 was ascertained in only 1 case (Teuffel et al., 2005). It was an unusual case of treatment-related MLL rearrangement in the absence of leukemia.

Clinics

The case reported by Teuffel et al. (2005), was a five-year-old girl, who experienced an acute myeloid leukemia (AML) with a variant t(8;21) and achieved remission under treatment. Four years later, a follow-up control of her karyotype revealed a t(11;11)(q13;q23), in the absence of any sign of leukemia in the bone marrow, over a period of 30 months following the discovery of the t(11;11).

Other cases of t(11;11)(q13;q23) were:
- A 13-year-old girl, who have had a M4eo AML with inv(16)(p13q22). Eleven month later, a t(11;11)(q13;q23) was found, but bone marrow remained normal; however, an overt M5b AML was diagnosed 6 months later (Leblanc et al., 1994). This case resembles the case of Teuffel.
- There was also the case of a 69-year-old male patient with a primary M4 AML, who died 5 months after diagnosis, and an AML (not classified) female patient (Testa et al., 1985; Mackinnon and Campbell, 2007).

Cytology

In the case reported by Teuffel, the MLL-ARHGEF17 was only seen in the myeloid lineage. The myeloid differentiation was not perturbed by the presence of the chimeric protein, and normal mature myeloid cells carrying the chimeric protein were found in normal amounts.

Cytogenetics

Cytogenetics morphological

The t(11;11) was apparently the sole anomaly in 3 of the 4 cases; a complex karyotype with del(5q), a marker chromosome, and other anomalies was found in the case reported by Mackinnon and Campbell, 2007.

Genes involved and proteins

ARHGEF17

Location
11q13

Protein
Guanine nucleotide exchange factor (GEF) for RhoA GTPases. Involved in transduction of various signals into downstream signaling cascades.

MLL

Location
11q23

DNA/RNA
36 exons, multiple transcripts 13-15 kb.

Protein
3969 amino acids; 431 kDa; contains two DNA binding motifs (a AT hook and a CXXC domain), a DNA methyl transferase motif, a bromodomain. MLL is cleaved by taspase 1 into 2 proteins before entering the nucleus, called MLL-N and MLL-C. The FYRN and FRYC domains of native MLL associate MLL-N and MLL-C in a stable complex; they form a multiprotein complex with transcription factor TFIID. MLL is a transcriptional regulatory factor. MLL can be associated with more than 30 proteins, including...
the core components of the SWI/SNF chromatin remodeling complex and the transcription complex TFIID. MLL binds pro-motors of HOX genes through acetylation and methylation of histones. MLL is a major regulator of hematopoiesis and embryonic development.

**Result of the chromosomal anomaly**

**Hybrid gene**

**Description**

The fusion between MLL and ARHGEF17 occurred in introns 12 and 1 respectively.

**References**


Mackinnon RN, Campbell LJ. Dicentric chromosomes and 20q11.2 amplification in MDS/AML with apparent monosomy 20. Cytogenet Genome Res. 2007;119(3-4):211-20

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