Identity

No anomaly was found in the karyotype, apart a possible 17p deletion. FISH revealed the translocation: BAC RP11-186B7 (green signal) hybridized to both chromosomes 17, indicating that it is centromeric to the translocation breakpoint. BAC RP1-4G17 (red signal) hybridized to both chromosomes 17 as well as one chromosome 11, indicating that the translocation breakpoint on 17p is within this BAC clone. The split red signal is also evident in an interphase cell (Reprinted by permission from Macmillan Publishers Ltd: Leukemia, 2007 Apr;21(4):842, Reader et al., copyright (2007)).

Clinics and pathology

Disease
Acute myeloid leukaemia

Epidemiology
Only one case to date, a 42 year old male patient (Reader et al., 2007).

Evolution
The patient achieved complete remission for 11 months, but relapsed and died 3 months after.

Cytogenetics

Cytogenetics morphological
Sole anomaly; as a matter of fact, the karyotype appeared normal, with a doubt on one chromosome 17: Cryptic translocation.

Genes involved and proteins

NUP98
Location
11p15
Protein
Nucleoporin: associated with the nuclear pore complex. Role in nucleocytoplasmic transport processes.

PHF23
Location
17p13
**Protein**
Recently discovered gene, which possess a plant homeodomain. Possible effect on chromatin structure and transcriptional regulation.

**Result of the chromosomal anomaly**

**Hybrid gene**

**Transcript**
NUP98 exon 13 fused in frame to PHF23 exon 4.

**Fusion protein**

**Description**
NH2-term NUP98 including the Phe-Gly repeats, the GLEB domain fused to the coiled-coil domain and the plant homeodomain of PHF23 C-term.

**References**

This article should be referenced as such: