Leukaemia Section
Mini Review

**t(11;12)(p15;p13)**
Laura JCM van Zutven, H Berna Beverloo

Department of Clinical Genetics, Erasmus MC, Dr. Molewaterplein 50, 3015 GE Rotterdam, The Netherlands

Published in Atlas Database: April 2006


DOI: 10.4267/2042/38355

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### Identity

**Note:** t(11;12)(p15;p13) should be the recurrent translocation; however, the only known case was in fact a variant/complex t(11:21;12)(p15:p13:p13).

### Clinics and pathology

**Disease**
Acute megakaryoblastic leukemia (AML-M7).

**Epidemiology**
So far only 1 case known, an infant case. However, the translocation might be missed using conventional banding techniques, and therefore additional cases might exist.

**Prognosis**
Remains in complete remission for at least 5 years.

### Cytogenetics

**Cytogenetics, morphological**
Not visible with conventional banding techniques alone: misdiagnosed as add(11)(p15) and der(21)(t(11:21)(p15;p13). Chromosome 12 was cytogenetically normal by conventional banding techniques and only identified as a partner in this translocation after FISH.

**Variants**
t(11:21;12)(p15;p13;p13). A t(11;12)(p15;p13), resulting in the same fusion product, might also exist, but none have been identified so far.

### Genes involved and Proteins

**NUP98**

**Location:** 11p15

**Protein**
920 amino acids; 97 kDa; contains repeated motifs (GLFG and FG) in N-term and a RNA binding motif in C-term.

### Results of the chromosomal anomaly

**Hybrid gene**

**Description**
In-frame fusion of the first 13 exons of NUP98 to exons 28-31 of JARID1A.

**Transcript**
5' NUP98 - 3' JARID1A

**Detection protocol**
FISH: BAC clones RP11-348A20 (NUP98) and RP11-283I3 (spanning JARID1A exon 11-31) colocalize.

**Fusion protein**

**Description**
The NUP98-JARID1A fusion protein contains the Phe-Gly (FG) repeats of the N-terminal part of NUP98. The JARID1A sequence starting with exon 28 still contains the sequence encoding the C-terminal PHD domain.
Schematic representation of the fusion NUP98-JARID1A. From up to down: NUP98, JARID1A and the putative chimeric NUP98-JARID1A structure. FG-repeats, phenylalanine-glycine repeats; JMj, Jumonji domains; ARID, AT-rich interaction domain; PHD, plant homeodomain fingers or LAP domains. The arrow indicates the position of the fusion.

**References**


This article should be referenced as such: