

# Leukaemia Section

## Mini Review

### t(1;2)(p36;p21)

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## Clinics and pathology

### Disease

Myelodysplastic syndrome (MDS) in most cases, acute lymphoblastic leukemia (ALL) in one case.

### Phenotype/cell stem origin

At least 3 of the 6 available cases were treatment related myelodysplastic syndromes (t-MDS) (Roulston et al., 1998; Mauritzson et al., 2002; Masuya et al., 2002), and 2 other cases were MDS (Horiike et al., 1988; Storlazzi et al., 2008).

### Clinics

A 38-year-old male patient presented with a treatment related myelodysplastic syndrome (t-MDS) evolving towards an acute myeloid leukemia (t-AML). Previous treatment included topoisomerase inhibitors for a Hodgkin disease 36 months before diagnosis of the t-MDS (Roulston et al., 1998). A t-MDS was diagnosed in a 76-year-old female patient previously treated with radiotherapy for uterine cancer 29 years ago. She died 26 months after diagnosis of the t-MDS (Mauritzson et al., 2002). A 49-year-old female patient was diagnosed with t-MDS (FAB refractory anemia (RA)); she had been treated with etoposide 2 years previously for M1-AML; the patient died 6.5 years after onset of the t(1;2). Other chromosome anomalies appeared during course of the disease, as well as an unrelated clone (Masuya et al., 2002). A 67-year-old female patient had a chronic myelomonocytic leukemia (CMML) with a normal karyotype; she received hydroxyurea. Three years later, a refractory anemia with excess of blasts-2 (RAEB-2) and a t(1;2) was diagnosed. The patient died one month later (Storlazzi et al., 2008).

Refractory anemia with excess of blasts (RAEB) was diagnosed in a 69-year-old male patient. The patient was still alive 15 months after diagnosis (Horiike et al.,

1988). A T-cell acute lymphoblastic leukemia (T-ALL) was found in a 1-year-old child (Mathew et al., 2001).

## Cytogenetics

### Cytogenetics morphological

In two cases, the t(1;2) was the sole anomaly (Horiike et al., 1988; Storlazzi et al., 2008). In contrast, complex karyotype were present in the 4 other cases. inv(14)(q11q32) was present in the T-ALL case (Mathew et al., 2001); del(5q) was found in two cases (Roulston et al., 1998; Mauritzson et al., 2002) and del(7q) in one case (Masuya et al., 2002). Other remarkable anomalies were: t(14;21)(q22;q22) with RUNX1 involvement (Roulston et al., 1998), +8, +12, +13 appearing during course of the disease (Masuya et al., 2002); there was also, in the latter case, an unrelated clone with t(11;12)(p15;q13).

## Genes involved and proteins

### Note

In only one case were the genes involved in the translocation studied (Storlazzi et al., 2008).

### PRDM16

#### Location

1p36

#### Protein

Transcription activator; PRDM16 forms a transcriptional complex with CEBPB. PRDM16 plays a downstream regulatory role in mediating TGFB signaling (Bjork et al., 2010). PRDM16 induces brown fat determination and differentiation (Kajimura et al., 2010).

### FLJ42875

#### Location

1p36

**DNA/RNA**

2 transcript variants; non-coding RNA of unknown function.

## Result of the chromosomal anomaly

**Fusion protein****Description**

PRDM16 (both long and short isoforms) and FLJ42875 are overexpressed. The sequence on chromosome 2 upregulating these 2 genes is unknown.

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