

# Leukaemia Section

## Short Communication

### Unbalanced whole-arm translocation der(1;10)(q10-q11;p10-p12)

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## Abstract

### Abstract

Unbalanced whole-arm chromosome translocations (WAT) involving the long arm of chromosome 1 are relatively rare anomalies in hematologic malignancies. They may involve acrocentric chromosomes (e.g. der(15)t(1;15)(q10;p10), der(21)t(1;21)(q10;p13) and der(22)t(1;22)(q10;p10)) or telomeric regions of nonacrocentric chromosomes (e.g. der(12)t(1;12)(q10;q24.3) and der(19)t(1;19)(q10;q13.4)). The most common WAT of 1q to centromeric or pericentromeric regions of other recipient chromosomes (indicated as p10 or q10;p10 or q10) are the der(1;7), der(1;13), der(16)t(1;16) and der(19)t(1;19) that are non-randomly distributed among hematological malignancies.

The derivatives of these translocations have been shown to give rise to a dicentric chromosome in a few cases (Sawyer et al., 1998; Fogu et al 2012).

### Keywords

chromosome 1; chromosome 10; hematologic malignancies; multiple myeloma; myeloid malignancies.

## Clinics and pathology

### Disease

Myeloid malignancies and multiple myeloma

## Epidemiology

Four recurrent translocations of 1q10-q11 to centromeric or pericentromeric regions of 10p have been identified, including an 61-year old male patient who was diagnosed with refractory anemia with excess blasts-1 (Odish et al., 2007), a 1-year old acute myelomonocytic leukemia (AML- M4) infant (Brown et al., 2012), a 49-years old chronic myeloid leukemia (CML) male patient and a male multiple myeloma (MM) patient with (Calasanz et al., 1997). Because of the close breakpoint in the 10p centromeric region, a case with der(10)t(1;10)(q11;p12) is included (Christodoulidou et al., 1994) (Table 1).

In addition, an unbalanced der(1)t(1;10) that resulted in trisomy of the long arm of chromosome 1 and monosomy of the short arm of chromosome 10 have been described in a 36-years old male case diagnosed with refractory cytopenia with multilineage dysplasia.

The rearranged chromosome contained the centromeres of both chromosomes 1 and 10, leading to a dic(1;10)(p11;p11) chromosome (Sambani et al., 2002).

## Prognosis

Only sporadic cases with uncertain clinical importance.

	Sex/Age	Disease	Karyotype
1.	M/49	CML	48,XY,+8,t(9;22),der(10)t(1;10)(q11;p12)
2.	M	MM	46,XY,del(5)(q13q22),der(10)t(1;10)(q11;p11),t(11;14)(q13;q32), der(14)t(11;14)/46,XY,del(5),t(11;14),der(12)t(1;12)(q11;p11),der(14)t(11;14)
3.	M/ 36	MDS	46,XY,+1,dic(1;10)(p11;p11)
4.	61/M	MDS	46,XY,+1,der(1;10)(q10;p10)
5.	1/F	AML	46,XX,t(8;16)(p11;p13)/46,idem,der(10)t(1;10)(q11;p11)/ 46,idem,add(7)(p21)/46,idem,add(7),der(10)

**Table 1.** Reported patients with der(1;10)(q10-q11;p10-p12) in hematologic malignancies.

1. Christodoulidou et al., 1994; 2. Calasanz et al., 1997 ; 3. Sambani et al., 2002; 4. Odish et al., 2007; 5. Brown et al., 2012

## Cytogenetics

### Cytogenetics morphological

Characterized by the presence of two copies of normal chromosome 1, a single copy of normal chromosome 10 and a der(10) chromosome containing 1q and 10p.

### Additional anomalies

Appears as a sole chromosomal abnormality in a patient with MDS (Odish et al., 2007), found in association with +8 and in the Ph-positive CML, complex karyotype in the remaining cases.

## Result of the chromosomal anomaly

### Fusion protein

#### Oncogenesis

Unbalanced whole-arm chromosome translocations with involvement of the 1q heterochromatin are non-random chromosomal rearrangements, detectable in both hematologic neoplasms and lymphomas. WAT of 1q to centromeric or pericentromeric regions of a partner chromosome involve breakage of non-homologous chromosomes at their centromeres leading to aberrant heterochromatin/euchromatin junctions. The main consequence of unbalanced WAT 1q translocation is a genomic imbalance resulting from the gain of the long arm of chromosome 1 and loss of the entire whole arm of the partner chromosome. Genomic imbalances leading to gene dosage abnormalities

are likely to play an important role in neoplastic processes that are associated with chromosome 1q WAT rearrangements.

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