Leukaemia Section
Short Communication

der(2)t(1;2)(q12-21;q37)
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Published in Atlas Database: March 2018
Online updated version : http://AtlasGeneticsOncology.org/Anomalies/t0102q12q37ID1821.html
Printable original version : http://documents.irevues.inist.fr/bitstream/handle/2042/70208/03-2018-t0102q12q37ID1821.pdf
DOI: 10.4267/2042/70208

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Abstract
The unbalanced der(2)t(1;2)(q12-21;q37) is a rare anomaly in hematological malignancies with unknown clinical significance.

Keywords
Unbalanced 1q translocations, der(2)t(1;2)(q12-21;q37), genomic imbalance, secondary genetic events.

Clinics and pathology

Disease
Acute myeloid leukemia (AML), multiple myeloma (MM) and less frequently lymphoid malignancies.

Phenotype/cell stem origin
Myeloid malignancies in 5 (3 males and 2 females): 1 Fanconi anemia patient with refractory anemia (RA) (Huret et al., 1988), 1 AML (Raimondi et al. 1999), 1 acute myeloblastic leukemia with minimal differentiation (AML-M0) and 2 acute monoblastic leukemia patients (AML-M5) (Busson Le Coniat et al., 2000; Cerveira et al., 2012).
Multiple myeloma in 8 (3 males and 5 females) (Smadja et al., 2001; Sawyer et al., 2014; Rack et al., 2016).

Partial karyotypes showing the unbalanced rearrangement between chromosomes 1 and 2.
**Epidemiology**

10 males and 8 females aged 21 to 81 years (median 69 years); male prevalence in patients with lymphoid malignancies (2 males in ALL and 3 males/1 female in lymphomas), aged 51, 69 and 71 years, 2 unknown. Patient with myeloid malignancies were aged 21, 68, 77 and 81 years, 1 unknown; the age of MM patients is unknown.

**Prognosis**

The Fanconi anemia patient received androgen therapy. He developed refractory anemia 4 years later that was treated with corticosteroids and antibiotics, but after recurrent infections he died from pneumonia 4 years later. The patient with acute monoblastic leukemia and t(11;19)(q23;p13) was treated with chemotherapy but failed to achieve remission; after receiving allogenic bone marrow transplant he is alive 23 months. 2 AML patients had highly complex karyotypes, considered as an adverse prognostic factor in AML. Similarly, in patients with MM or lymphoid malignancies and highly complex karyotypes, the prognosis is likely unfavorable.

**Cytogenetics**

**Cytogenetics morphological**

Unbalanced rearrangement resulting in 1q trisomy.

**Additional anomalies**

Found in association with del(5q) in MDS, 11q23/ KMT2A (MLL)/ MLLT1 in acute monoblastic leukemia, as an additional anomaly to t(9;22) in 1 AML and in both ALL cases and as an additional aberration to t(14;18) in 2 lymphoma patients; found as part of highly complex karyotypes in the remaining patients.

**Result of the chromosomal anomaly**

**Fusion protein**

**Oncogenesis**

A non-random structural gain of 1q is a known structural anomaly in hematopoietic malignancies, and it is often the result of an unbalanced chromosome translocation. The breakpoints within 1q show considerable variation from 1q11 to 1q43, with a clustering to 1q12-23. Various unbalanced 1q translocation partners have been described, among them, the der(2)t(1;2)(q12-21;q37) has been detected accompanied with other chromosome abnormalities, therefore it is likely a secondary genetic event. The major consequence of this unbalanced chromosome translocation is the genomic imbalance resulting from the gain of the long arm of chromosome 1q.

**References**


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