Leukaemia Section
Short Communication

der(Y)t(Y;1)(q11-12;q12-21)
Adriana Zamecnikova
Kuwait Cancer Control Center, Kuwait annaadria@yahoo.com

Published in Atlas Database: March 2018
Online updated version : http://AtlasGeneticsOncology.org/Anomalies/t0Yq01qID1818.html
Printable original version : http://documents.trevues.inist.fr/bitstream/handle/2042/70459/03-2018-t0Yq01qID1818.pdf
DOI: 10.4267/2042/70459

This work is licensed under a Creative Commons Attribution-Noncommercial-No Derivative Works 2.0 France Licence.
© 2019 Atlas of Genetics and Cytogenetics in Oncology and Haematology

Abstract

Structural abnormalities involving sex chromosomes are uncommon in hematological malignancies. The unbalanced translocation between the long arm of chromosomes 1 and Y results in a partial trisomy of the 1q region and has been described mainly in chronic myeloproliferative disorders.

Keywords
Chronic myeloproliferative disorders; unbalanced translocation; genomic gains; disease evolution.

Clinics and pathology

Disease
Myeloid disorders mainly, rarely B-cell lymphoid malignancies.

Note
Chronic myeloid neoplasm in 15 patients (15 males aged 15 to 75 years; median 63 years). Among them, there were 7 cases of myelodysplastic syndrome (MDS) (Hollings et al., 1988; Thompsonet al., 1991; Wei et al., 1993; Raymakersetal al., 1996; Djordjevicet al., 2008; Wan et al., 2001; Sanford et al., 2015), 1 polycythemia vera (Testa et al., 1981), 1 idiopathic myelofibrosis (Michaux et a., 1996), 1 post-polycthyemic myelofibrosis (Manabe et al., 2013), 1 essential thrombocytethemia (Lim et al., 2016), 3 chronic myelomonocytic leukemia (Hollings et al., 1988; Michaux et al., 1996; Chen et al., 2010) and 1 atypical chronic myeloid leukemia (Ohsaka Hisa 2002).

Acute myeloid leukemia in 5 (aged 0 to 86 years, median 63 years): 2 acute myeloblastic leukemia with maturation (AML-M2) (Haupt et al., 1991; Singh et al., 1993), 2 acute myelomonocytic leukemia (AML-M4) (Bao et al., 2006; Brown et al., 2012) and 1 acute monoblastic leukemia (AML-M5) (Tuborgh et al., 2013).

Acute lymphoblastic leukemia in 2 pediatric patients aged 0 and 10 years (Dayton et al., 1994; Heerema et al., 1999).

Multiple myeloma in 3 (1 aged 73 years, 2 unknown age) (Mugneret et al., 1995; Wang et al., 2010; Sawyer et al., 2014) and.

Mature B-cell neoplasm in 4 (aged 10,11,36 and 60 years) (Tanaka et al., 1990; Dayton et al., 1994; Lones et al., 2004; Havelange et al., 2013).

Epidemiology
About 28 male patients harboring der(Y)t(Y;1)(q11-12;q12-21) have been reported (aged 1 to 86 years, median 60 years). Among them, there were 7 pediatric patients aged 0 to 15 years (median 10 years).

Prognosis
The acquisition of an unbalanced 1q rearrangement in patients with myeloid malignancies appears to be a late event associated with disease transition and poor response to treatment. der(Y)t(Y;1) in multiple myeloma and lymphoid malignancies is part of complex karyotypes with coexisting other high-risk genetic abnormalities such as 11q23 and 8q24 rearrangements that have an additional impact leading to shorter survival in these patients.
**Cytogenetics**

**Cytogenetics morphological**
Unbalanced translocation; a trisomic 1q juxtaposed to Yq12-21 replacing the terminal segment of chromosome Y.

**Additional anomalies**
Sole karyotype aberration in 10 patients, while it was accompanied by a limited number of additional chromosomal changes in 5 myeloid malignancies: +9 in 2 and +8, del(20q), del(11)(q13) sole cases. Found with 11q23 rearrangements as part of complex karyotypes in 2 AML patients. Association with t(8;14)(q24;q32)/8q24 rearrangement in 1 ALL in all the 4 patients with mature B-cell neoplasm; highly complex rearrangements in MM.

**Result of the chromosomal anomaly**

**Fusion protein**
Oncogenesis
Complete or partial trisomies of 1q are well-known in hematological malignancies, but involvements of sex chromosomes are uncommon. Among them, der(Y)t(Y;1)(q11-12;q12-21) is most common in chronic myeloproliferative disorders, but cases of other diseases, such as AML or Burkitt's lymphoma have also been reported. While it is unknown which genes on 1q are responsible for the development and/or progression of these diseases, it is likely that 1q trisomy results in deregulation of several genes implicated in leukemogenesis.

**References**


karyotype are distinct from other aggressive B-cell lymphomas with MYC rearrangement. Genes Chromosomes Cancer. 2013 Jan;52(1):81-92


Ohsaka A, Hira T. Spectral karyotyping refined the identification of a der(Y)(t(Y;1)(q11 1 or 2;q12)) in the blast cells of a patient with atypical chronic myeloid leukemia


Thompson PW, Standen GR, Geddes AD. Transient t(Y;1)(q12;q21) in a patient with Fanconi anemia and myelodysplastic syndrome Cancer Genet Cytogenet 1991 Apr;52(2):201-2

Tuborgh A, Meyer C, Marschalek R, Preiss B, Hasle H, Kjeldsen E. Complex three-way translocation involving MLL, ELL, RREB1, and CMAHP genes in an infant with acute myeloid leukemia and t(6;19;11)(p22 2;p13 1;q23 1)

