

Leukaemia Section

Short Communication

der(1;7)(q10;p10)

Hélène Bruyère

Cytogenetics Laboratory, VGH, Department of Pathology and Laboratory Medicine, 899 12th Avenue West, Vancouver, BC, V5Z 1M9, Canada; Helene.Bruyere@vch.ca

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Abstract

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Review on der(1;7)(q10;p10), with data on clinics and cytogenetics.

Clinics and pathology

Note

Preponderant in myeloid disorders.

Disease

Myelodysplastic syndromes (MDS) in 60 % of cases.

Acute myeloid leukemia (AML) in 30%, most commonly AML-M2 but reported in all FAB subtypes; frequently with preceding MDS.

Myeloproliferative neoplasms (MPN) represent the remaining 10% of cases.

Lymphoid disorders: very rare, half of cases being multiple myeloma with the der(1;7) as the sole abnormality (raising the possibility of an underlying MDS), the other half in various lymphoid disorders chronic lymphocytic leukemia (CLL), Burkitt lymphoma/leukemia, follicular lymphoma), most often part of a complex karyotype (Geisler et al., 1997; Hsiao et al., 2005; Al-Bahar et al., 2010).

Others: reported in only one case each of bilineage or biphenotypic acute leukemia with a t(9;22) (Sanada et al. 2007), sarcoma (Roberts et al., 2008) and carcinoma (Jin et al., 2002) with complex karyotypes and in two cases of aplastic anemia (Kim et al., 2010).

Phenotype/cell stem origin

MDS cases: most commonly RA (60%).

AML cases: most commonly M2 but has been reported in all FAB subtypes.

MPN cases: reported in a few cases of polycythemia vera, essential thrombocythemia, chronic myelomonocytic leukemia and idiopathic myelofibrosis.

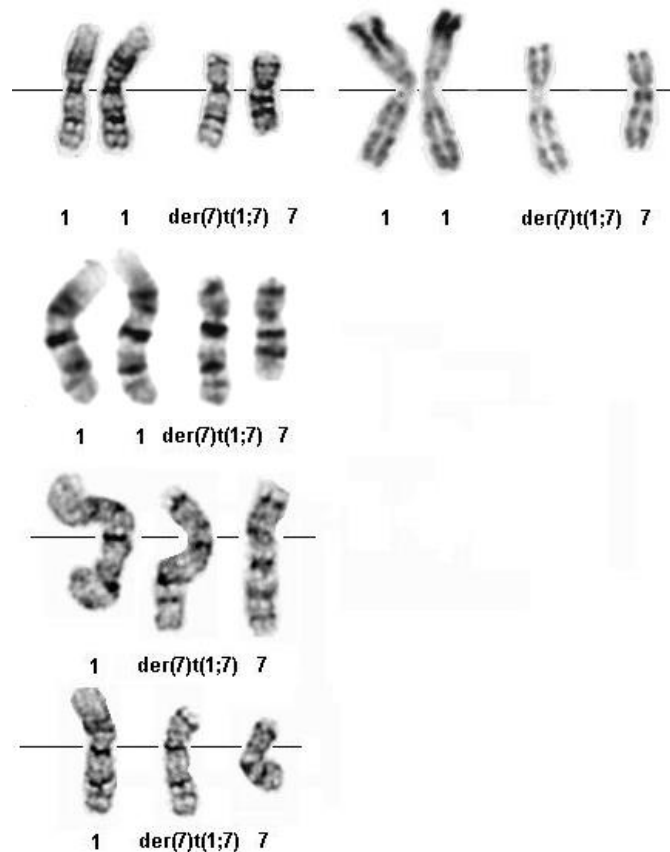
Epidemiology

Found in 1.5-6% of MDS, 0.2-2% of AML and rarely in MPN. Older adults mostly (median late 50's, early 60's. male predominance (4M/1F), influenced partly by the preponderance of males in a large study of Japanese MDS cases with der(1;7) (Sanada et al., 2007).

Prognosis

MDS: There is some controversy as to the prognosis of the der(1;7) with trisomy 1q and monosomy 7q. A better outcome of der(1;7) compared to -7/del(7q) cases was shown in a retrospective study including 77 cases (Sanada et al., 2007), while there was no statistical difference in overall survival between der(1;7) versus del(7q) versus -7 in several studies including a smaller number of patients (Slovak et al., 2009; Hussain et al., 2012).

AML: In the UKMRC trials, the der(1;7) may be included in the "-7/del(7q)" group, associated with a poor prognosis (Grimwade et al., 2010). In the CALGB 8461 study, loss of 7q was associated with an intermediate prognosis (Byrd et al., 2002).



der(1;7)(q10;p10) G- (left) and R- (right) bandings - top: Courtesy Jean-Luc Lai; middle top: - Courtesy Sun Yuan; middle and bottom: - Courtesy Diane H. Norback, Eric B. Johnson, and Sara Morrison-Delap, UW Cytogenetic Services

Cytogenetics

Cytogenetics morphological

Unbalanced whole-arm translocation with two chromosomes 1, a derivative chromosome including the long arm of chromosome 1 and the short arm of chromosome 7, and a chromosome 7 resulting in trisomy for 1q / monosomy for 7q. The balanced form may have been reported once in a secondary AML-M2 case (Higuchi et al., 1995).

Additional anomalies

85% of cases are not complex if the unbalanced der(1;7) with extra chromosome 1 is considered as a single abnormality ("single abnormality" in 50% of cases, one additional abnormality in the remaining 35%). The most frequent additional abnormalities are: +8 (50%); del(20q) (20%); +21 or +9 (3% each). A cytogenetically unrelated, abnormal clone is found in 5% of cases, 80% in MDS, 20% in AML. Loss of Y, -7 and +8 are the most common abnormalities.

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