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Leukaemia Section
Short Communication

der(9)t(1;9)(q12;q12)

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Abstract

Review on der(9)t(1;9)(q12;q12), with data on clinics.

Clinics and pathology

Disease

Myeloid malignancies mainly.

Phenotype/cell stem origin

6 patients were diagnosed with chronic myeloproliferative disorders (MPD): polycythemia vera (PV) 5 cases (Rege-Cambrin et al., 1991; Najfeld et al., 2002; Jacob et al., 2003; Sambani et al., 2005) and myelofibrosis (MF) 1 patient (Sambani et al., 2005). 1 patient was diagnosed with plasma cell leukemia (Heller et al., 2004) and 1 with follicular lymphoma (Mohamed et al., 2001) (Table 1).).

Epidemiology

Sporadic cases; found in 4 male and 4 female patients (sex ratio 1:1) aged 53 to 73 years.

Prognosis

Unknown, rare occurrence.

Cytogenetics

Cytogenetics morphological

Presents as 2 normal chromosomes 1 and 9 and an extra der(9)t(1;9) chromosome in 7 out of 8 cases.

Additional anomalies

Sole abnormality in 2 MPD patients (Sambani et al., 2005), associated with limited additional anomalies in the remaining MPD patients (each case 1): +8 (Jacob et al., 2003), +9 (Najfeld et al., 2002), del(5)(q14q32) (Rege-Cambrin et al., 1991) and del(11)(q22) (Sambani et al., 2005); complex karyotypes in plasma cell leukemia and lymphoma patients.

Result of the chromosomal anomaly

Fusion protein

Oncogenesis

The unbalanced der(9)t(1;9)(q12;q12) is a rare but recurrent chromosome translocation, found mainly in chronic myeloproliferative neoplasms. The breakpoints in chromosomes 1 and 9 are within the constitutive heterochromatin bands that are notoriously unstable chromosomal regions and permit formation of complex and unstable chromosomal translocations. Mostly, these rerrangements are unbalanced and accompanied by genomic imbalances, casually implicated in disease initiation or progression. The presence of an extra der(9)t(1;9)(q12;q12) results in trisomy of both 1q and 9p arms, therefore altered dosages of genes are likely to be involved in neoplastic processes. der(9)t(1;9)(q12;q12) is apparently secondary as mostly coexists with well-known primary abnormalities such as trisomy 8 or 9 or 5q deletion. The presence of either trisomy 9 or +9p suggests that gene-dosage effect of JAK2, located at 9p24.1 may contribute to MPD phenotype. In addition, the observation that MPD patients with 9/+9p trisomy are invariably JAK2V617F-positive (Campell et al., 2006) imply a role for JAK2 cooperating mutation in myeloproliferative disorders.

Ref	Sex/Age	Disease	Karyotype
1.	F/53	Polycythemia vera	47,XX,del(5)(q14q32),+der(9)t(1;9)(q12;q12)
2.	F/63	Follicular lymphoma	
3.	M	Polycythemia vera	46,XY,der(9)t(1;9)(q12;q12),+9
4.	F/60	Polycythemia vera	48,XX,+8,+der(9)t(1;9)(q12;q12)
5.	M	Plasma cell leukemia	51,XY,-1,-1,+3,+der(5)t(5;11)(q13-14;q24)t(1;11)(q12;q25),+7 or der(7)t(1;7)(p31;p15),+8, +der(9)t(1;9)(q12;q12),der(11)t(1;11)(p31;p15)t(1;11)(q12;q25),-13,der(14)t(X;14)(q21;p13),+15,+18,der(19)t(9;19)(q12;q11),+i(19)(q10)
6.	M/73	Polycythemia vera	47,XY,+der(9)t(1;9)(q12;q12),del(11)(q22)/47,idem,der(17)t(13;17)(q13;q25)
7.	F/53	Idiopathic myelofibrosis	47,XX,+der(9)t(1;9)(q12;q12)
8.	M/70	Polycythemia vera	47,XY,+der(9)t(1;9)(q12;q12)

Table 1. Reported patients with der(9)t(1;9)(q12;q12).
Abbreviations: M., male; F., female; LN., lymph node. 1. Rege-Cambrin et al., 1991; 2. Mohamed et al., 2001; 3. Najfeld et al., 2002; 4. Jacob et al., 2003; 5. Heller et al., 2004; 6-8. Sambani et al., 2005.

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