

Leukaemia Section

Short Communication

der(9)t(1;9)(q11-12;q34)

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Abstract

Abstract

Review on der(9)t(1;9)(q11-12;q34), with data on clinics, and the genes involved.

Clinics and pathology

Disease

Myeloid malignancies, multiple myeloma (MM) and plasma cell leukemia (PCL).

Phenotype/cell stem origin

3 patients were diagnosed with myeloid malignancies: 1 patient with acute myelomonocytic leukemia (Piccaluga et al., 2004), 1 with polycythemia vera (PV) (Andrieux et al 2003) and 1 with chronic myelomonocytic leukemia (CMML) (Suh et al., 2009). There were 4 patients with MM (Smadja et al., 2001; Mohamed et al., 2007; Sawyer et al., 2014) and 1 case was diagnosed with plasma cell leukemia (Ueshima et al., 1983).

6 additional cases were found with various 1q21-25 and 9q34 breakpoints: 4 AML cases (Lai et al., 1987; Smadja et al 1992; Raimondi et al., 1999; Yoshida et al., 2013), 1 Chronic myelogenous leukemia (CML) patient (Erdag et al., 2009) and 1 patient with biphenotypic leukemia (Matsuda et al., 2009). (Table 1) leukemia.

Epidemiology

There was 7 male and 7 female patients (sex ratio 1:1) aged 0 to 66 years including 2 infant patients, one of them with Down syndrome.

Prognosis

Unknown, very rare occurrence in patients with hematologic malignancies.

Cytogenetics

Cytogenetics morphological

Presents as 2 normal chromosomes 1, one normal chromosome 9 and a der(9)t(1;9) chromosome, resulting in 1q trisomy associated with partial monosomy of 9q.

Additional anomalies

All the 3 reported patients with myeloid malignancies showed this translocation either as the sole abnormality (Piccaluga et al., 2004; Suh et al., 2009) or as part of a simple karyotype (Andrieux et al., 2003). A complex karyotype was present in all MM patients, among them the 14q32 rearrangement was found as an accompanying anomaly in 3 out of 4 patients (Smadja et al., 2001; Mohamed et al., 2007; Sawyer et al., 2014).

Result of the chromosomal anomaly

Fusion protein

Oncogenesis

The unbalanced der(9)t(1;9)(q11-12;q34) is created by fusion in the centromeric region of 1q to a distant telomeric region of chromosome 9. It is a rare anomaly in patients with hematologic malignancies that results in trisomy of 1q associated with partial monosomy of 9q.

		der(9)t(1;9)(q11-12;q34)	
M/35	PCL	44,X,-Y,t(6;8)(q13;p11),der(9)t(1;9)(q12;q34)dup(1)(q12q44),-13	1 .
M	MM	45,XY,del(1)(p21p31),der(9)t(1;9)(q11;q34),del(12)(q13q24),-13,add(14)(q32),del(16)(q21),+19/44,idem,del(6)(q16),der(14;15)(q10;q10)add(14)(q32)/90,idemx2	2 .
M	MM	46-56,XY,+2,+3,+5,+6,del(6)(q23)x2,+7,+9,der(9)t(1;9)(q12;q34)x2,+11,+15,+17,+19,+mar	3 .
F/52	PV	46,XX,del(3)(p21),der(9)t(1;9)(q11;q34), 46,XX,der(9)t(1;9)	4 .
F/56	AML-M4	46,XX,der(9)t(1;9)(q11;q34)	5 .
M/57	MM	44,XY,der(3)t(1;3)(q21;q29),t(4;14)(p16;q32),der(9)t(1;9)(q12;q34),-10,-13	6 .
M/45	CMML	46,XY,der(9)t(1;9)(q11;q34)BCR-ABL1 negative	7 .
M	MM	82-92,XXY,-Y,-1,-1,der(1)add(1)(p36)add(1)(q42),-2,-2,del(3)(q21)x2,-4,-4,add(5)(q15),der(5 t(1;5)(q12;q15),add(6)(q21)x2,+7,+7,+7,add(7)(p22),add(7)(q36),der(7)t(1;7)(q12;q32)x3,+8,der(8)t(1;8)(q12;q22)x2,+9,+9,add(9)(p24),der(9)t(1;9)(q12;q34),der(10)t(1;10)(q12;q26),add(11)(p15)x2,-12,-13,del(?13)(q12q22)x3,-14,der(14)t(1;14)(p11;q32)x3,-15,15,add(15)(p11),add(16)(q24),-17,add(17)(p11)x2,+19,+19,-20,+21,add(21)(p11),+22,add(22)(q13),inc	8 .
		der(9)(1;9)(q21-q25;q34)	
F/59	AML-M4	46,XX,t(8;16)(p11;p13)/46,idem,der(3)t(3;11)(q27;q13),der(9)t(1;9)(q24;q34)	9 .
F/54	AML-M7	48,XX,+5,t(9;22)(q34;q11),+der(22)t(9;22)47,XX,t(9;22),+der(22)/47,idem,der(9)t(1;9)(q21;q34)/46,XX,del(1)(p11),add(3)(q26),+8,t(9;22),-2147,XX,t(8;10)(q21;q23),der(9),t(9;22),add(12)(q24),del(13)(q13q32),+der(22)	1 0 .
F	AML	48,XX,der(9)t(1;9)(q23;q34),+11,+21	1 1 .
M/66	CML	46,XY,t(2;17)(p13;q22),der(9)t(1;9)(q21;q34),t(9;22)(q34;q11),der(19)t(19;20)(p13;p11),-20,der(22)t(9;22)/46,X,t(X;20)(q22;q13),t(6;12)(p21;q24). 5 months after PBSCT	1 2 .
F/0	BAL	46,XX,+1,t(1;15;11;10)(p36;q11;q23;q24),der(1;12)(q10;q10)/46,idem,t(1;12)(p10;p10)/46,idem,t(1;12)(p10;p10),der(9)t(1;9)(q25;q34)	1 3 .
F/1	AML-M7	46,XX,i(7)(q10),der(9)t(1;9)(q25;q34),-16,add(19)(q13),+21c	1 4 .

Table 1. Reported patients with der(9)(q11-q12;q34) including additional cases with 1q21-q25 and 9q34 breakpoints.

Abbreviations: M, male; F, female; PCL, plasma cell leukemia; MM, multiple myeloma; PV, polycythemia vera; AML-M4, acute myelomonocytic leukemia; CMML, chronic myelomonocytic leukemia; AML-M7, acute megakaryoblastic leukemia; CML, chronic myelomonocytic leukemia; PBSCT, peripheral blood stem cell transplantation; BAL, bilineage or biphenotypic leukemia.

1. Ueshima et al., 1983; 2-3. Smadja et al., 2001; 4. Andrieux et al., 2003; 5. Piccaluga et al., 2004; 6. Mohamed et al 2007; 7. Suh et al., 2009; 8. Sawyer et al 2014; 9. Lai et al 1987; 10. Smadja et al 1992; 11. Raimondi et al., 1999; 12. Erdag et al 2009; 13. Matsuda et al 2009; 14. Yoshida et al 2013.

The formation of an extra copy of the entire 1q suggest that trisomy of genes located on 1q, rather than partial 9q monosomy is involved in disease pathogenesis. This is further supported by the occurrence of additional cases of der(9)t(1;9) with various 1q21-25 breakpoints, suggesting that altered gene dosages in a part or the entire long arm of chromosome 1 may be important for disease initiation or progression.

Alternatively, the breakpoint at 9q34, where the ABL1 gene is located rises the possibility of its

involvement in the translocation, at least in some patients.

However, the absence of ABL1 rearrangement or the BCR / ABL1 fusion gene was excluded only in sporadic cases (Piccaluga et al., 2004; Suh et al., 2009).

Because of the rare occurrence, further studies are needed to evaluate the genes involved in the oncogenesis as well as the suggested association of this rearrangement with monocytic-lineage leukemia (Suh et al., 2009).

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