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Leukaemia Section
Short Communication

der(20)t(1;20)(q10-21;q11-13)

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Abstract

Review on t(1;20)(q10-21;q11-13), with data on clinics.

Clinics and pathology

Disease

Acute myeloid leukemia (AML), acute lymphoblastic leukemia (ALL), myeloproliferative neoplasm (MPN), Myelodysplastic syndrome (MDS), multiple myeloma (MM), Burkitt lymphomas and non-Burkitt type lymphomas.

Phenotype/cell stem origin

Suggested involvement of a pluripotent stem cell.

Epidemiology

Rare karyotypic event in various hematologic malignancies; AML/MDS (5 cases), ALL (4 cases), MPN (2 cases), MM (4 cases), lymphoma (4 cases). Male predominance (15 males/ 3 females); patients ages ranged from 1 to 73 years; described mainly in adults (aged 28 to 73 years); all the 4 ALL patients were children (aged 1 to 7 years) (Table 1).

Prognosis

Seems to confer a poor prognosis.

	Sex	Age	Karyotype	Diagnosis	Reference
1	M	49	46,XY,t(3;11)(p13;q21)/46,XY,der(20)t(1;20)(q21;q13) biclonal clones	PV	Wan et al; 2001
2	F		46,XX,dup(1)(q21q25),dup(1)(q21q42),del(7)(q31),del(11)(q21q25),add(17)(q25),der(20)t(1;20)(q10;q13) 46,XX,dup(1)(q21q42),del(7),del(11),der(20)t(1;20)	MDS Fanconi anemia	Alter et al; 2000
3	M	38	47,XY,+?der(1)t(1;20)(q21;q11)del(1)(p11),-9,t(9;22)(q34;q11),+18,der(20)t(1;20)(q21;q11)	CML	Mori et al; 1997
4	M	30	46,XY,t(11;12)(q13;p13)/46,idem,der(9)t(1;9)(q12;p24)/ 46,idem,der(14)t(1;14)(q12;p10)/ 46,idem,der(20)t(1;20)(q12;q13)/46,idem,der(21)t(1;21) (q12;q10)	AML-M5	Itzhar et al; 2011
5	F		46,XX,der(20)t(1;20)(q21;q13)	AML	Raimondi et al;1999
6	M	34	47,XY,t(9;22)(q34;q11),t(10;21)(p11;q22),der(20)t(1;20)(q21;q13),+der(22)t(9;22)/48,idem,+8	AML-M1	Sasaki et al; 1983
7	F	7	56,XX,+X,+X,t(2;16)(p12;q12),+4,+5,+6,+10,+18,der(20)t(1;20)(q12;q13),+21,+21,+mar	ALL	Busson-Le Coniat et al; 1999
8	M		56,XY,+X,+Y,+5,+6,i(7)(q10),+9,+10,+11,+18,der(20)t(1;20)(q12;q13),+21,+22	ALL	Hereema et al; 2004
9	F	1	46,XX,t(4;11)(q21;q23),der(20)t(1;20)(q11;q13)	ALL	Prigogina et al; 1998

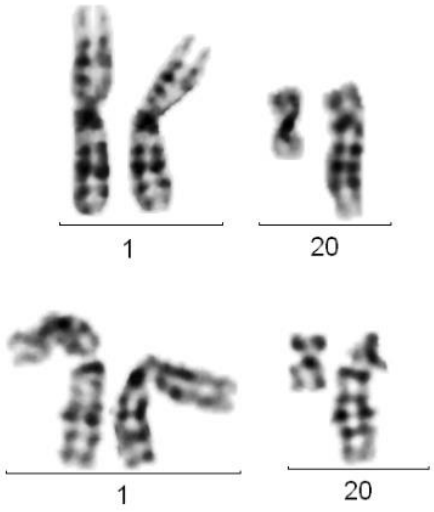
10	M	4	45,X,-Y,der(20)t(1;20)(q21;q13)	B-ALL	Raimondi et al; 2003
11	M	59	47,XY,der(16)t(1;16)(q21;q11),+der(19)t(1;19)(q21;q13),der(20)t(1;20)(q21;q11)	MM	Keung et al; 1999
12	M	73	42,X,-Y,del(1)(p13p22),der(1;7)t(1;7)(p13;?p22)ins(1;?)(p13;?),del(2)(q31q37),dic(7;9)(p15;q34),-8,-9,der(10)t(8;10)(q11;p12),-13,add(15)(q26), der(20)t(1;20)(q12;q13)	MM	Mohamed et al; 2007
13	M	54	54,XY,+3,+5,+9,+9,+15,+15,del(18)(q22),+19,der(20)t(1;20)(q21;q13),+21	MM	Mohamed et al; 2007
14	M		51-54,XY,+1,der(1;16)(q10;p10),+3,t(4;18)(p14;p11),del(6)(q25),del(6)(q23),+del(6)(q11),+7,+9,add(11)(q23),-13,+15,+18,der(20)t(1;20)(q12;q13),+mar	MM	Sawyer et al; 1998
15	F	28	46,XX,t(14;18)(q32;q21)/47,idem,+12/47,idem,t(5;7)(q22;q32),+12/47,idem,+12,der(20)t(1;20)(q21;q13) lymph node	FL B-cell	Horsman et al; 2001
16	M	2	47,XY,del(2)(q21q31),t(3;22)(q27;q11),del(6)(q13q15),der(8)t(2;8)(q21;q24),+11,der(20)t(1;20)(q21;q13) lymph node	DLBCL	Itoyama et al; 2002
17	M	73	46,XY,i(6)(p10),t(8;14)(q24;q32),der(20)t(1;20)(q21;q13) lymph node	BL	Lones et al; 2004
18	M		48,Y,t(X;1)(q28;p22),+Y,t(5;12;16)(p14;q24;p13),der(6)t(6;18)(q13;q21),-8,del(8)(p21),+add(9)(q22),add(12)(p11),der(18)t(8;18)(q11;q21),der(19)t(12;19)(p11;q13),del(20)(q13),der(20)t(1;20)(q21;q11),+mar lymph node	B-cell lymphoma	Shimazaki et al; 1999

Abbreviations: PV, Polycythemia vera; MDS, myelodysplastic syndrome; CML, Chronic myeloid leukemia; ALL, acute lymphoblastic leukemia; FL, follicular lymphoma, DLBCL, diffuse large B-cell lymphoma; BL, Burkitt lymphoma/leukemia; MM, multiple myeloma.

Cytogenetics

Cytogenetics morphological

Cytogenetically heterogeneous, the breakpoints in 1q varied from 1q10 to 1q21, with a clustering to 1q21, and the 20q breaks occurred in 20q10 to 20q13, mainly in the 20q13 region.



Partial karyotypes showing the unbalanced t(20)t(q10;q11).

Additional anomalies

Usually present with additional chromosomal abnormalities; may be found together with well-known primary abnormalities such as t(9;22)(q34;q11), t(4;11)(q21;q23), and t(14;18)(q32;q21), t(8;14)(q24;q32).

Result of the chromosomal anomaly

Fusion protein

Oncogenesis

Unbalanced translocations involving all or part of the long arms of chromosomes 1 and 20 are found in both hematologic neoplasms and lymphomas. The abnormality is usually present with complex pattern of rearrangements or occurring in a subclone; indicating that der(20)t(1;20) might be a secondary aberration. The extra copy of 1q segment and/ or 20q monosome may directly or indirectly provide a proliferative advantage leading to clonal evolution associated with tumor progression and advanced disease.

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