Table 1 Clinical and laboratory findings at the time of diagnosis

Feature (n=24)	DS-AMKL (n=21)	non-DS-AMKL	*P*
Median age, mo (range)	21 (8-38)	15 (2-185)	0.52
Sex			
M/F ratio	12:12	8:13	0.42
Clinical findings			
Fever, no. (%)	12 (50)	14 (67)	0.33
Lymphadenopathy, no. (%)	2 (8.3)	6 (29)	0.12
Hepatomegaly, no. (%)	16 (67)	14 (67)	1.00
Splenomegaly, no. (%)	11 (46)	9 (43)	1.00
Laboratory findings			
Hemoglobin, g/dL (median, range)	8.3 (5.2-13.0)	8.2 (3.6-11.0)	0.38
Platelets, $\times 10^9$/L (median, range)	26.0 (2-143)	24.0 (3-492)	0.72
Leukocytes, $\times 10^9$/L (median, range)	6.8 (2.4-107.0)	17.9 (0.1-134.8)	0.07
Circulating blast cells, % (median, range)	15.0 (0-88)	13.0 (0-82)	0.95
Bone marrow blast cells, % (median, range)	25.0 (2-90)	50.0 (10-81)	0.09
Serum LDH, IU/L (median, range)	662 (260-6450)	1639 (338-8660)	0.11

Abbreviations: DS-AMKL, Down syndrome-associated acute megakaryoblastic leukemia; LDH, lactic dehydrogenase.

Table 2 Morphological classification, immunophenotype and karyotype of the 24 patients with DS-AMKL

Patient No.	Age (months)	Sex	Morphological classification	Immunophenotype (%) CD7	CD13	CD36	CD41	CD42	GlyA	Karyotype
A: Constitutional trisomy 21 only										
1	32	F	3+Tri	57	19	42	10	14	NA	47,XX,+21c
2	21	F	1A	68	36	42	12	NA	13	47,XX,+21c
3	21	M	2B	72	17	55	56	NA	20	47,XY,+21c
4	21	F	3+Ep	18	45	NA	31	44	NA	47,XX,+21c
5	18	M	2B	53	78	81	43	47	24	47,XY,+21c
B: Numerical abnormalities only										
6	21	M	1A	83	20	NA	41	42	2	51,XY,+8,+19,+21c,+21,+22
7	8	M	1A	56	10	NA	16	23	NA	48,XY,+8,+21c
C: −5/del(5q) and/or −7/del(7q)										
8	20	F	1B	78	30	35	24	34	NA	47,XX,del(6)(q23q25),-7,+ring(7?),+21c
9	23	F	1A	36	29	83	76	75	NA	46,XX,-1,+der(1)t(1;?),-5,+der(5)t(5;7),-7,+21c
10	24	F	1A+dE	90	30	35	24	34	NA	47,XX,del(7)(q32),+21c
11	17	F	3+m	40	75	NA	14	12	12	47,XX,add(5)(p15),-7,+8,+21c
12	20	F	2B	14	79	NA	4	25	99	90,idem × 2,-3,-7,-9,del(11)(q?),-18
13	38	M	2B+dE	81	83	NA	15	20	4	47,XY,-7,+8,+21c,+rl
D: Other structural changes										
14	20	F	1A	83	27	NA	59	87	NA	48,X,der(X)t(X;1)(q28;q25),+11,+21c
15	18	M	1A	70	12	34	2	2	38	47,XY,der(3)t(3;3)(p25;p10),i(7q), der(17)t(1;17)(q25;q25),+21c
16	17	F	1A	51	4	NA	11	9	28	48,idem,add(5)(q1?),+11
17	12	M	1A	89	36	NA	74	19	15	47,XY,der(7)t(1;7)(q23;q36),del(20)(q11q13.1),+21c
18	29	M	2B	60	43	69	14	34	62	47,XY,del(11)(p?),+21c
19	25	F	1A+dE	71	23	NA	41	NA	84	47,XX,t(5;12)(p15;q21),add(7)(p11),+21c,add(22)(q13)
20	18	M	1B	52	37	NT	10	26	32	48,idem,+add21
21	22	F	1B+Ep	99	78	88	59	66	93	47,XX,add(7)(p11),add(19)(p13),+21c
22	8	M	1A	72	53	64	72	75	4	NA
23	21	M	2A	87	12	33	20	23	4	NA
24	19	F	1A	87	26	NA	40	NA	11	NA

Abbreviations: GlyA indicates glycophorin A; Tri, trilineage dysplasia; NA, not available; Ep, emperipolesis; dE, dyserythropoieis; m, micromegakaryocytes.

Table 3 Morphological classification, immunophenotype and karyotype of the 21 patients with non-DS-AMKL

Patient No.	Age (months)	Sex	Morphological classification	Immunophenotype (%) CD7	CD13	CD36	CD41	CD42	GlyA	Karyotype
A: Normal karyotypes										
1	147	F	3+m	66	95	42	53	19	1	46,XX
2	18	M	2A+Tri	25	54	40	4	2	4	46,XY
B: Numerical abnormalities only										
3	3	F	NA	1	88	NA	77	54	8	48,XX,+21,+22
4	15	F	2A	42	9	NA	68	10	NA	58,XX,+X,+2,+2,+6,+7,+8,+10,+13,+15,+19,+19,+22
5	38	F	1A	38	30	NA	10	4	NA	49,XX,+12,+18,+22
6	12	F	NA	NA	30	NA	95	NA	NA	57,XX,+2,+4,+6,+7,+8,+10,+14,+15,+19,+19,+22
7	45	M	1A	95	3	NA	86	3	1	48,idem,+8
8	2	M	2A	19	32	NA	46	18	5	51,XY,+6,+7,+8,+19,+21
C: t(1;22)(p13;q13)										
9	2	F	2A+dE	NA	NA	NA	NA	NA	NA	46,XX,t(1;22)(p13;q13)
10	12	M	1A	5	13	NA	67	70	NA	51,XY,+der(1)t(1;22)(p13; q13),t(1;22)(p13;q13), +6,+7,+10,+19
D: 3q21q26 abnormalities										
11	12	M	1A	14	3	64	48	47	NA	46,XY,-11,der(11)t(3;11)(q21;p15)
E: t(16;21)(p11;q22)										
12	38	M	3+Ep	3	69	NA	33	33	NA	46,XY,t(16;21)(p11;q22)
F: −7										
13	8	F	1A	30	7	NA	82	80	NA	46,XX,-7,-7,del(11)(p11),+2mar,inc
14	41	M	3+m	66	43	NA	28	32	8	47,XY,-7,+21,+ring(1)
G: Other structural changes										
15	7	M	1A	12	65	NA	57	NA	NA	46,XY,t(2;7)(p12;p22)
16	28	F	1A	NA	NA	NA	30	17	1	51,XX,+X,6p+,6p+,-13,+21,+21,+2mar
17	35	F	3+m	3	18	2	78	1	NA	47,XX,add(16)(p13),+21
18	12	F	2A+dE	30	5	62	67	55	5	46,XX,t(2;11;19)(q31;q13;q13.4),del(3)(q23),13q-
19	6	F	NA	NA	10	NA	52	53	NA	51,XX,4p+,11q+,14q+,+17,+19,+21,+22 ,+mar
20	33	F	2A	5	2	12	5	1	1	46,XX,del(2)(q11),del(2)(q31),der(5)t(2;5)(q11;q22), der(5)t(5;13)(q35;q14),-13,add(16)(p13),+mar
21	185	F	2A	59	90	NA	16	4	NA	49,XX,+5,+8,i(17)(q10),+21

Abbreviations: GlyA indicates glycophorin A; Tri, trilineage dysplasia; NA, not available; Ep, emperipolesis; dE, dyserythropoieis; m, micromegakaryocytes.