

Supplementary data 1

Table S I Characteristics of 43 patients with myelodysplastic syndromes harboring BCOR mutations

Case No.	Gender	Age (year)	subtype	BM Blast (%)	Karyotype	BCOR mutations (VAF %)	Concomitant mutations
1	F	23	RAEB-2	18.0	Normal	BCOR/4/C925Y (51)	DHX9, WT1
2	M	80	RAEB-1	8.4	47,XY,+8[10]/46,XY[10]	BCOR/11/K1452X (60)	DNMT3A, ROBO1, STAG2
3	M	40	RAEB-2	3.6 (Auer)	46,XY,del(20)(q12q13)[20]	BCOR/8/R1323W (50)	NPM1
4	F	36	RAEB-1	5.0	Normal	BCOR/8/K1339N (33)	ASXL1, MPL, TET2, UPF3A
5	F	56	RAEB-1	8.0	Normal	BCOR/4/A619fs (16)	PHF6, RUNX1, TET2
6	M	77	RCMD	4.0	Normal	BCOR/4/Y361fs (85)	NRAS, ROBO1, RUNX1, U2AF1
7	M	56	RCMD	2.2	Normal	BCOR/12/R1547*(49)	ASXL1, RUNX1, SRSF2
8	M	62	RAEB-1	9.5	Normal	BCOR/4/p819-819del (74)	DNMT3A, IDH2, ROBO1, RUNX1
9	M	69	RAEB-2	11.2	Normal	BCOR/7/R1165X (29) /BCOR/11/C4326+1C>A (56)	CEBPA, DNMT3A, RUNX1, STAG2, TET2, U2AF1
10	M	57	RAEB-2	13.2	Normal	BCOR/7/P1101fs (65)	ITIH3
11	F	69	RAEB-2	17.0	Normal	BCOR/4/V912fs (32)	ASXL1, STAG2
12	F	43	RCMD	3.5	47,XX,add(3)(q28),+8[4] /46,XX[5]	BCOR/4/P483L (48)	EZH2, IDH1, U2AF1

13	F	73	RAEB-2	12.0	Normal	BCOR/4/T738fs (46)	EZH2, MPL, RUNX1
14	M	51	RAEB-1	8.2	46,XY,-11,+mar[10]/46,XY,del(7)(q31),inc[3]/46,XY[2]	BCOR/4/P483L (100)	No
15	F	20	RAEB-1	5.0	Normal	BCOR/10/P1367fs (30)	RBM, TET2,
16	F	44	MDS-U	0	Normal	BCOR/9/L1299fs (30)	CEBPA, TET2
17	M	21	RT	0.6	Normal	BCOR/8/L1299fs (30)	ASXL1, CALR, MPL
18	F	46	RAEB-2	15.0	Normal	BCOR/4/T591fs (45)	DNMT3A; IDH2,
19	M	70	RAEB-1	7.8	46,XY,inv(1)(q21q25),del(5)(q22q35[9])/46,XY[9]	BCOR/4/P483L (100)	DNMT3A, JAK2, RUNX1, TET2, TP53, U2AF1
20	M	68	RAEB-1	7.5	Normal	BCOR/4/T870fs (44)	ASXL1, DHX9, STAG2
21	F	52	RCMD	1.0	Normal	BCOR/7/R1165X (24)	No
22	M	46	RCMD	0.1	47,XY,+8[10]/46,XY[10]	BCOR/4/P483L (100)	ASXL1, JAK2, ANKRD11
23	M	18	RCMD	0.4	Normal	BCOR/8/G1342X (14)	IDH1
24	F	35	RCMD	0.5	Normal	BCOR/4/R810X (12)	ASXL1, IDH1
25	M	70	RCMD	1.8	Normal	BCOR/8/R1183X (39)	JAK2, STAG2
26	F	54	RCMD	0.4	Normal	BCOR/4/S526L (52)	EZH2, KIF20B, SETBP1
27	M	64	RT	2.0	46XY, +8[5]/46XY[15]	BCOR/15/K1672* (13)	ASXL1, STAG2, TET2
28	M	84	RAEB-1	6.0	Normal	BCOR/13/O1606X (21)	ASXL1, SRSF2
29	F	41	RCMD	3.0	Normal	BCOR/6/P1083fs (21.4)	No
30	F	54	RCMD	2.5	Normal	BCOR/4/P483L (46)	No
31	M	44	RT	2.5	Normal	BCOR/4/I501fs (60)	IDH2, U2AF1
32	F	77	RCMD	3.6	Normal	BCOR/4/P483L (80)	DNMT3A

33	M	61	RAEB-1	7.4	Normal	BCOR/10/1380C (62)	ASXL1
34	M	72	RCMD	1.0	Normal	BCOR/4/T819fs (91)	MPL, ROBO1
35	F	54	RAEB-2	19.0	44,XX,-2,-4,del(5)(q13q33),der(11),-16,-17,del(20)(q11),+2mar,inc[15]	BCOR/4/P483L (50)	SETBP1, TET2, TP53, ROBO2
36	F	65	RAEB-2	18.0	46,XX,del(20)(q12)[10]	BCOR/9/V1351M (54)	ASXL1, U2AF1
37	M	67	RAEB-1	5.0	Normal	BCOR/4/P483L (100)	ASXL1, U2AF1
38	M	77	RAEB-1	7.0	48,XY,+8,+2[2]/46,XY[18]	BCOR/13/Y1567fs (28)	ASXL1, TET2, RUNX1
39	M	69	RAEB-1	9.0	Normal	BCOR/4/G154fs (76)	CEBPA, DNMT3A, MPL, STAG2, WT1
40	M	69	RAS	0.6	Normal	BCOR/4/P483L (99.5)	DNMT3A, SF3B1
41	F	63	RT	0	47,XX+8[3]	BCOR/4/P483L (50)	No
42	M	73	RAEB-2	10.0	46,XY,t(9;21)(q22;q22)[8]/47,idem,+8[7]/46,XY[2]	BCOR/4/S637fs (40)	ASXL1, EZH2, DHX9, TET2
43	F	28	RCMD	2.6	47, XX, del (1)(p22), der(2), del(7)(q31), +8, der(17)[8]/46,XX[10]	BCOR/4/R861H (49)	No

Figure S1

