Unique ethnic features of *DDX41* mutations in patients with idiopathic cytopenia of undetermined significance, myelodysplastic syndrome, or acute myeloid leukemia

Eun-Ji Choi,[1*] Young-Uk Cho,[2*] Eun-Hye Hur,[1] Seongsoo Jang,[2] Nayoung Kim,[3] Han-Seung Park,[1] Jung-Hee Lee,[1] Kyoo-Hyung Lee,[1] Si-Hwan Kim,[2] Sang-Hyun Hwang,[2] Eul-Ju Seo,[2] Chan-Jeoung Park[2] and Je-Hwan Lee[1]

[1]Department of Hematology, Asan Medical Center, University of Ulsan College of Medicine; [2]Department of Laboratory Medicine, Asan Medical Center, University of Ulsan College of Medicine and [3]Asan Institution for Life Sciences and Department of Convergence Medicine, Asan Medical Center, University of Ulsan College of Medicine, Seoul, Korea

**E-JG and Y-UC contributed equally as co-first authors.*

 doi:10.3324/haematol.2020.270553

Received: August 27, 2020.

Accepted: February 2, 2021.

Pre-published: February 25, 2021.

Correspondence: *JE-HWAN LEE* - jhlee3@amc.seoul.kr

Supplementary Appendix

Supplementary Table 1. List of genes selected for targeted next-generation sequencing

Gene	Transcript	Gene	Transcript
ABL1	NM_005157.5	*KIT*	NM_000222.2
ANKRD26	NM_014915.2	*KMT2A*	NM_005933.3
ASXL1	NM_015338.5	*KRAS*	NM_033360.3
ATM	NM_000051.3	*LUC7L2*	NM_016019.5
BCOR	NM_001123383.1	*MALT1*	NM_006785.3
BCORL1	NM_021946.4	*MPL*	NM_005373.2
BRAF	NM_004333.4	*MYD88*	NM_002468.4
CALR	NM_004343.3	*NF1*	NM_000267.3
CBL	NM_005188.3	*NOTCH1*	NM_017617.4
CDKN2A	NM_000077.4	*NPM1*	NM_002520.6
CEBPA	NM_004364.4	*NRAS*	NM_002524.4
CSF3R	NM_156039.3	*PHF6*	NM_001015877.1
DDX41	NM_016222.3	*PTEN*	NM_000314.6
DNMT1	NM_001379.3	*PTPN11*	NM_002834.4
DNMT3A	NM_022552.4	*RAD21*	NM_006265.2
EED	NM_003797.4	*RUNX1*	NM_001754.4
EP300	NM_001429.3	*SETBP1*	NM_015559.2
ETNK1	NM_018638.4	*SETD2*	NM_014159.6
ETV6	NM_001987.4	*SF1*	NM_004630.3
EZH2	NM_004456.4	*SF3B1*	NM_012433.3
FBXW7	NM_033632.3	*SMC1A*	NM_006306.3
FLT3	NM_004119.2	*SMC3*	NM_005445.3
GATA1	NM_002049.3	*SRSF2*	NM_003016.4
GATA2	NM_032638.4	*STAG2*	NM_001042749.2
HRAS	NM_005343.3	*STAT3*	NM_139276.2
IDH1	NM_005896.3	*TET2*	NM_001127208.2
IDH2	NM_002168.3	*TP53*	NM_000546.5
IKZF1	NM_006060.5	*U2AF1*	NM_001025203.1
JAK2	NM_004972.3	*WT1*	NM_024426.4
JAK3	NM_000215.3	*ZRSR2*	NM_005089.3
KDM6A	NM_021140.3		

Supplementary Table 2. *DDX41* variants, concurrent mutations in other genes, and karyotypes at diagnosis in the 28 patients with germline *DDX41* mutations

Patient	Sex	Age	Diagnosis, 2016 WHO criteria	BM blast (%)	Probable germine *DDX41* variants			Somatic *DDX41* variants			Concurrent mutations (VAF, %)	Karyotype
					Nucleotide change	AA change	VAF (%)	Nucleotide change	AA change	VAF (%)		
1	M	54	ICUS	3.6	c.776A>G	p.Y259C	44.7	c.1574G>A	p.R525H	5.3		46,XY[20]
2	M	54	ICUS	4.0	c.455T>G	p.V152G	48.7	c.1126G>A	p.A376T	10.2		46,XY[20]
3	M	54	ICUS	2.0	c.1496dup	p.A500fs	50.8	c.1679C>T	p.P560L	41.1	DNMT3A p.R604fs (38.2)	46,XY[20]
4	M	65	ICUS	3.4	c.455T>G	p.V152G	40.7	c.1480A>G	p.K494E	25.4	CBL p.I383M (5.8), KMT2A p.T548A (2.6)	46,XY[20]
5	M	67	ICUS	2.0	c.776A>G	p.Y259C	50.0	c.1759G>C	p.G587R	3.3	ETNK1 p.Q28H (7.8), ASXL1 p.H633fs (3.3)	46,XY[20]
6	M	64	MDS-MLD	1.6	c.455T>G	p.V152G	49.6	c.1574G>A	p.R525H	27.4	ASXL1 p.Q1142* (4.3), TP53 p.G266V (2.1)	46,Y,t(X;3)(q28;q12)[3]/46,XY[17]
7[a]	M	76	MDS-MLD	4.0	c.455T>G	p.V152G	53.5	c.680C>T	p.T227M	14.5	EZH2 p.R684C (3.9)	46,XY[20]
8	M	58	MDS-EB-1	6.6	c.776A>G	p.Y259C	45.5	c.1574G>A	p.R525H	14.5	NF1 p.L1411V (14.3), TP53 p.R273C (11.2), PHF6 p.M1T (8.5), PHF6 p.R116* (3.6)	46,XY[20]
9[a]	M	63	MDS-MLD	3.2	c.455T>G	p.V152G	46.9	c.680C>T	p.T227M	12.1	PHF6 p.G248D (6.8), CBL p.P6825fs (5.1)	46,XY[20]
10	M	76	MDS-EB-1	6.5	c.455T>G	p.V152G	47.9	c.1574G>A	p.R525H	10.6	EZH2 c.625+1G>A (2.2)	46,XY[20]
11	M	71	MDS-EB-1	7.0	c.776A>G	p.Y259C	47.6	c.1574G>A	p.R525H	13.6	CBL p.R420L (10.8), NF1	46,XY,add(7)(11.2)[4]/46,XY[16]

											p.A2625T (10.4), ASXL1 p.D784fs (7.7)	
12	M	61	MDS-EB-2	18.2	c.19G>T	p.E7*	50.4	c.682G>T	p.G228C	14.0	SRSF2 p.P95H (11.9), ETNK1 p.N244S (8.6)	46,XY[20]
13	M	78	MDS-EB-1	6.8	c.455T>G	p.V152G	55.6	c.680C>T	p.T227M	25.4	ETNK1 p.G245A (19.4)	45,X,-Y[6]/46,XY[19]
14[a]	M	61	MDS-MLD	0.8	c.776A>G	p.Y259C	43.5	c.680C>T	p.T227M	6.5	NF1 p.K1423Q (5.0)	46,XY[20]
15[a]	M	65	MDS-EB-1	8.0	c.776A>G	p.Y259C	48.9	c.1574G>A	p.R525H	14.5		46,XY[10]
16[a]	M	79	MDS-EB-1	5.6	c.983T>G	p.L328R	48.1	c.1588G>A	p.G530S	22.5		46,XY[20]
17[a]	M	71	MDS-EB-1	6.2	c.776A>G	p.Y259C	53.2	c.1574G>A	p.R525H	7.7	DNMT3A p.W893fs (13.6), NF1 p.H1605dup (7.2), DNMT1 p.Y940N (3.0), PHF6 p.C20F (2.6)	46,XY,del(7)(q22q31)[16]/46,XY[14]
18	M	75	MDS-EB-1	6.0	c.19G>T	p.E7*	48.1				KDM6A p.C1331S (5.0), TP53 p.R280I (3.1)	46,XY,del(20)(q11.2q13.3)[3]/46,XY[17]
19[a]	M	41	MDS-EB-2	19.2	c.1496dup	p.A500fs	46.0	c.1574G>A	p.R525H	7.7	CBL p.K389M (6.4)	46,XY[4]
20[a]	M	62	MDS-MLD	3.4	c.455T>G	p.V152G	43.4	c.1574G>A	p.R525H	18.7	DNMT3A p.F752del (21.7), IKZF1 p.N159S (3.1)	46,XY[20]
21[a]	M	72	MDS-SLD	3.4	c.455T>G	p.V152G	48.0	c.1127C>T	p.A376V	28.9		46,XY[20]
22[a]	M	65	MDS-MLD	4.8	c.455T>G	p.V152G	50.8	c.680C>T	p.T227M	35.8		46,XY[20]
23[a]	M	75	MDS-EB-2	13.8	c.1496dup	p.A500fs	46.9	c.1032C>G	p.D344E	3.2	TET2 p.E1728* (3.3), PHF6 p.M1T (2.9)	46,XY[20]
24	M	75	MDS-EB-1	7.2	c.776A>G	p.Y259C	43.6	c.1574G>A	p.R525H	5.7	TET2 p.H1921R (6.6), JAK2 p.V617F (4.1)	46,XY[20]

25	M	74	AML NOS	24.0	c.1496dup	p.A500fs	45.5	c.1574G>A	p.R525H	2.0	EZH2 c.2196-1G>C (2.0)	46,XY[20]
26	M	63	AML NOS	20.2	c.19G>T	p.E7*	49.2	c.1574G>A	p.R525H	2.7	PHF6 p.M1V (3.8)	46,XY[20]
27	F	71	AML NOS	23.6	c.1496dup	p.A500fs	50.9	c.1574G>A	p.R525H	15.8	SETBP1 p.D868N (15.4), ASXL1 p.W796fs (15.1)	46,XX[20]
28	M	69	AML NOS	65.2	c.1496dup	p.A500fs	47.9	c.1574G>A	p.R525H	23.2	ASXL1 p.R693* (25.7), NRAS p.G12D (7.1)	48,XY,+1,+8[8]/46,XY[12]

WHO: World Health Organization; BM: bone marrow; AA: amino acid; VAF: variant allele frequency; M: male; F: female; ICUS: idiopathic cytopenia of undetermined significance; MDS: myelodysplastic syndrome; MDS-MLD: MDS with multilineage dysplasia; MDS-EB: MDS with excess blasts; MDS-SLD: MDS with single lineage dysplasia; AML NOS, acute myeloid leukemia not otherwise specified.

[a]These patients were confirmed to have germline *DDX41* variants by targeted NGS assay using sorted blood T-cells.

Supplementary Table 3. Interpretation of probable germline *DDX41* mutations according to the Genome Aggregation Database[1] and the recommendation of the American College of Medical Genetics and Genomics[2]

Patient	Nucleotide change	AA change	Somatic *DDX41*	gnomAD, total	KRGDB	dbSNP	SIFT/PolyPhen-2/ MutationTaster	ACMG recommendation		Causality
								Criteria	Interpretation	
1	c.776A>G	p.Y259C	Present	2.84e-5	5.82e-4	rs139780256	D/ProD/DC	PS4	Uncertain significance	Causal
2	c.455T>G	p.V152G	Present	7.96e-6	2.94e-4	rs758775538	D/B/DC	PS4	Uncertain significance	Causal
3	c.1496dupC	p.A500fs	Present	7.96e-6	0			PVS1 + PS4	Pathogenic	Causal
4	c.455T>G	p.V152G	Present	7.96e-6	2.94e-4	rs758775538	D/B/DC	PS4	Uncertain significance	Causal
5	c.776A>G	p.Y259C	Present	2.84e-5	5.82e-4	rs139780256	D/ProD/DC	PS4	Uncertain significance	Causal
6	c.455T>G	p.V152G	Present	7.96e-6	2.94e-4	rs758775538	D/B/DC	PS4	Uncertain significance	Causal
7	c.455T>G	p.V152G	Present	7.96e-6	2.94e-4	rs758775538	D/B/DC	PS4	Uncertain significance	Causal
8	c.776A>G	p.Y259C	Present	2.84e-5	5.82e-4	rs139780256	D/ProD/DC	PS4	Uncertain significance	Causal
9	c.455T>G	p.V152G	Present	7.96e-6	2.94e-4	rs758775538	D/B/DC	PS4	Uncertain significance	Causal
10	c.455T>G	p.V152G	Present	7.96e-6	2.94e-4	rs758775538	D/B/DC	PS4	Uncertain significance	Causal
11	c.776A>G	p.Y259C	Present	2.84e-5	5.82e-4	rs139780256	D/ProD/DC	PS4	Uncertain significance	Causal
12	c.19G>T	p.E7*	Present	0	0	rs749405703		PVS1 + PS4 + PM2	Pathogenic	Causal
13	c.455T>G	p.V152G	Present	7.96e-6	2.94e-4	rs758775538	D/B/DC	PS4	Uncertain significance	Causal
14	c.776A>G	p.Y259C	Present	2.84e-5	5.82e-4	rs139780256	D/ProD/DC	PS4	Uncertain significance	Causal
15	c.776A>G	p.Y259C	Present	2.84e-5	5.82e-4	rs139780256	D/ProD/DC	PS4	Uncertain significance	Causal
16	c.983T>G	p.L328R	Present	0	0		D/ProD/DC	PM2	Uncertain significance	Causal
17	c.776A>G	p.Y259C	Present	2.84e-5	5.82e-4	rs139780256	D/ProD/DC	PS4	Uncertain significance	Causal
18	c.19G>T	p.E7*	Absent	0	0	rs749405703		PVS1 + PS4 + PM2	Pathogenic	Causal

19	c.1496dupC	p.A500fs	Present	7.96e-6	0			PVS1 + PS4	Pathogenic	Causal
20	c.455T>G	p.V152G	Present	7.96e-6	2.94e-4	rs758775538	D/B/DC	PS4	Uncertain significance	Causal
21	c.455T>G	p.V152G	Present	7.96e-6	2.94e-4	rs758775538	D/B/DC	PS4	Uncertain significance	Causal
22	c.455T>G	p.V152G	Present	7.96e-6	2.94e-4	rs758775538	D/B/DC	PS4	Uncertain significance	Causal
23	c.1496dupC	p.A500fs	Present	7.96e-6	0			PVS1 + PS4	Pathogenic	Causal
24	c.776A>G	p.Y259C	Present	2.84e-5	5.82e-4	rs139780256	D/ProD/DC	PS4	Uncertain significance	Causal
25	c.1496dupC	p.A500fs	Present	7.96e-6	0			PVS1 + PS4	Pathogenic	Causal
26	c.19G>T	p.E7*	Present	0	0	rs749405703		PVS1 + PS4 + PM2	Pathogenic	Causal
27	c.1496dupC	p.A500fs	Present	7.96e-6	0			PVS1 + PS4	Pathogenic	Causal
28	c.1496dupC	p.A500fs	Present	7.96e-6	0			PVS1 + PS4	Pathogenic	Causal

AA: amino acid; gnomAD: Genome Aggregation Database; KRGDB: Korean Reference Genome Database; B: benign; D: deleterious; DC: disease-causing; P: polymorphism; ProD: probably damaging; T: tolerated; ACMG, American College of Medical Genetics and Genomics.

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Supplementary Table 4. Odds ratio of probable germline *DDX41* mutations for developing ICUS/MDS/AML in the Korean population

Variants	Patients (n=457)	Controls (n=1,722)[a]	Odds ratio (95% CI)
Total	29[b]	15	7.7 (4.1–14.5)
p.V152G	10	1	38.5 (4.9–301.6)
p.Y259C	9[b]	2	17.3 (3.7–80.2)
p.A500fs	6	0	49.6 (2.8–882.1)
p.E7*	3	0	26.5 (1.4–514.5)
p.L328R	1	0	11.3 (0.5 – 278.4)
ICUS/MDS[c]	25[b]	15	10.9 (5.7 – 21.0)
p.V152G	10	1	62.6 (8.0 – 490.8)
p.Y259C	9[b]	2	28.0 (6.0 – 130.5)
p.A500fs	3	0	42.7 (2.2 – 828.5)
p.E7*	2	0	30.4 (1.5 – 634.5)
p.L328R	1	0	18.2 (0.7 – 447.0)
AML[d]	4	15	2.7 (0.9 – 8.3)
p.A500fs	3	0	71.1 (3.7 – 1383.0)
p.E7*	1	0	30.1 (1.2 – 742.5)

Abbreviations: AML, acute myeloid leukemia; CI, confidence interval; ICUS, idiopathic cytopenia of undetermined significance; MDS, myelodysplastic syndrome.

[a]Frequencies of *DDX41* variants in healthy controls were based on the Korean Reference Genome Database (http://coda.nih.go.kr/coda/KRGDB/index.jsp). [b]A patient of MDS harboring p.Y259C was considered as non-causal owing to an absence of the somatic *DDX41* mutation was included. [c]The number of patients was 285. [d]The number of patients was 172.

Supplementary Table 5. Types of germline *DDX41* mutations and the concurrent somatic *DDX41* mutations according to the type of hematologic malignancy

	ICUS/MDS (n=24)	**AML (n=4)**
Type of germline *DDX41* mutations		
p.V152G (%)[a]	10 (41.7)	0
p.Y259C (%)[a]	8 (33.3)	0
p.A500fs (%)	3 (12.5)	3 (75.0)
p.E7* (%)	2 (8.3)	1 (25.0)
p.L328R	1 (4.2)	0
Concurrent somatic *DDX41* mutations (%)	23 (95.8)	4 (100.0)
p.R525H (%)	10 (43.5)	4 (100.0)
p.T227M (%)	5 (21.7)	0
Others (%)	8 (34.8)	0

AML: acute myeloid leukemia; ICUS: idiopathic cytopenia of undetermined significance; MDS: myelodysplastic syndrome; VAF: variant allele frequency.

Bold indicates the significant differences. [a]The combined frequency of p.V152G and p.Y259C was significantly higher in the ICUS/MDS (75.0%) than in the AML (0%) patients (P=0.01).

Supplementary Table 6. Concurrent somatic *DDX41* mutations according to the three most common germline *DDX41* mutations

	Germline *DDX41* mutation			***P*[a]**
	p.V152G (n=10)	**p.Y259C (n=8)**	**p.A500fs (n = 6)**	
p.R525H (%)	3 (30.0)	6 (75.0)	4 (66.7)	**0.032**, 0.129, 1.000
p.T227M (%)	4 (40.0)	1 (12.5)	0	0.314, 0.234, 1.000
Others (%)	3 (30.0)	1 (12.5)	2 (33.3)	0.588, 1.000, 0.538

VAF: variant allele frequency.

Bold indicates the significant differences. [a]*P* values were derived from comparisons between p.V152G and p.Y259C, p.V152G and p.A500fs, and p.Y259C and p.A500fs, respectively.

Supplementary Table 7. Clinical courses of the patients with *DDX41* mutations

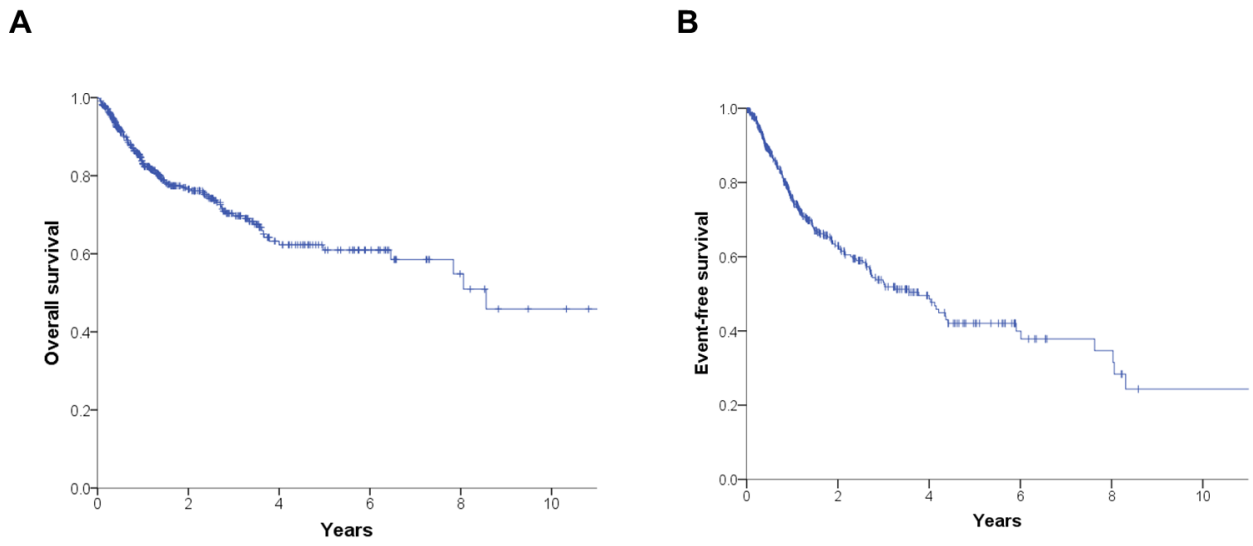
No.	Sex	Age	Dx	Subtype	Probable germline	Somatic	Clinical course
1	M	54	ICUS		p.Y259C	p.R525H	Evolved to MDS-EB-1 77.9 months after ICUS diagnosis; Scheduled for allogeneic HCT.
2	M	64	ICUS			p.T227M p.Q63fs	Lost to follow-up; Died of unknown cause after 35.9 months from diagnosis.
3	M	54	ICUS		p.V152G	p.A376T	Evolved to MDS-EB-1 17.6 months after ICUS diagnosis; Progressed to MDS-EB-2 two months after MDS-EB-1 diagnosis; Alive without relapse four years after allogeneic HCT from a haploidentical donor (patient's son).
4	M	54	ICUS		p.A500fs	p.P560L	Lost to follow-up.
5	M	65	ICUS		p.V152G	p.K494E	Alive without evidence of disease evolution 31.3 months after ICUS diagnosis; A family history of Hodgkin lymphoma (patient's son).
6	M	67	ICUS		p.Y259C	p.G587R	Evolved to MDS-EB-2 nine months after ICUS diagnosis with a gain of *PTPN11* mutation; One course of azacitidine; After that, best supportive care for four months.
7	M	54	MDS	MDS-MLD	p.V152G	p.R525H	Died of pneumonia 3.4 months after allogeneic HCT from a matched unrelated donor.
8	M	52	MDS	MDS-U	p.Y259C		Received two courses of azacitidine (SD) before HCT; Alive without relapse 44 months after allogeneic HCT from a haploidentical donor (patient's son).
9[1]	M	76	MDS	MDS-MLD	p.V152G	p.T227M	Received danazol for 1.5 years; Alive in SD without HI.
10	M	58	MDS	MDS-EB-1	p.Y259C	p.R525H	Progressed to MDS-EB-2 49.8 months after MDS diagnosis; Alive without relapse 16.5 months after allogeneic HCT from a matched unrelated donor.
11[1]	M	62	MDS	MDS-MLD	p.V152G	p.T227M	Not responded to cyclosporine, erythropoietin-stimulating agent, and one course of decitabine.
12	M	76	MDS	MDS-EB-1	p.V152G	p.R525H	Lost to follow-up.
13	M	71	MDS	MDS-EB-1	p.Y259C	p.R525H	Achieved HI with decitabine treatment; Progressed to MDS-EB-2 after 23 courses of decitabine; Died of unknown cause one year after progression.

14	M	60	MDS	MDS-EB-2	p.E7*	p.G228C	Received eight courses of decitabine (SD) before HCT; Alive without relapse 8.7 months after allogeneic HCT from a haploidentical donor (patient's son without *DDX41* mutation).
15	M	78	MDS	MDS-EB-1	p.V152G	p.T227M	Progressed to MDS-EB-2 50.2 months after MDS diagnosis; Achieved PR after six courses of azacitidine; Loss of the response after 21 courses of azacitidine; Died of infection.
16[1]	M	61	MDS	MDS-MLD	p.Y259C	p.T227M	Progressed to MDS-EB-1 91.5 months after MDS diagnosis; Received six courses of decitabine (SD).
17[1]	M	64	MDS	MDS-EB-1	p.Y259C	p.R525H	Alive in stable disease 13.6 months after MDS diagnosis; Scheduled for allogeneic HCT.
18[1]	M	79	MDS	MDS-EB-1	p.L328R	p.G530S	Received danazol for one year; Alive in SD without HI.
19[1]	M	71	MDS	MDS-EB-1	p.Y259C	p.R525H	Received 13 courses of azacitidine; Alive in SD.
20	M	75	MDS	MDS-EB-1	p.E7*		Lost to follow-up.
21[1]	M	41	MDS	MDS-EB-2	p.A500fs	p.R525H	Received four courses of decitabine (SD) before HCT; Alive without relapse four months after allogeneic HCT from a matched unrelated donor.
22[1]	M	62	MDS	MDS-MLD	p.V152G	p.R525H	Died of pneumonia after one course of decitabine.
23[1]	M	72	MDS	MDS-SLD	p.V152G	p.A376V	Alive without disease progression seven months after MDS diagnosis.
24[1]	M	65	MDS	MDS-MLD	p.V152G	p.T227M	Progressed to MDS-EB-1 33.6 months after MDS diagnosis; Alive without relapse eight weeks after allogeneic HCT from a matched unrelated donor.
25[1]	M	75	MDS	MDS-EB-2	p.A500fs	p.D344E	Received nine courses of azacitidine (SD).
26	M	74	MDS	MDS-MLD	p.Y259C	p.R525H	Alive without disease progression six months after MDS diagnosis.
27	M	53	MDS	MDS-EB-1	p.D139G		Progressed to MDS-EB-2 29.6 months after MDS diagnosis; Progressed to AML 31.0 months after MDS diagnosis.
28	M	76	AML	NOS	p.A500fs	p.R525H	Achieved CR with Guadecitabine; Relapsed; Died of disease progression
29[2]	F	59	AML	RGA	p.E3K		Relapsed 7.7 months after the end of consolidation chemotherapy; Achieved the second CR with salvage chemotherapy; Alive without relapse 15 months after allogeneic HCT from a matched unrelated donor.
30	M	57	AML	NOS	p.Y33C		Achieved CR with intensive chemotherapy; Alive without relapse 13.6 months after allogeneic HCT from a matched unrelated donor.
31	F	66	AML	RGA	p.D139G		Relapsed 13 months after the end of consolidation chemotherapy; Achieved the second CR with salvage chemotherapy;

							Underwent allogeneic HCT from a haploidentical donor (patient's son).
32	M	20	AML	APL	p.K187R		Relapsed 11 months after the end of consolidation chemotherapy; Achieved the second CR with arsenic trioxide; Alive without relapse 15.8 months after allogeneic HCT from a haploidentical donor (patient's father).
33	F	62	AML	NOS		p.R525H	Relapsed 4 years after allogeneic HCT in the first CR from a matched sibling donor; Achieved the second CR with salvage chemotherapy; alive without relapse 9.8 months after the second allogeneic HCT from a haploidentical donor (patient's son).
34	M	79	AML	MRC		p.A488T	Received four courses of decitabine; Died of pneumonia and persistent disease.
35	M	67	AML	NOS	p.E7*	p.R525H	Relapsed 4 years after allogeneic HCT in the first CR from a haploidentical donor (patient's son); Achieved the second CR with salvage chemotherapy; Alive without relapse 3.4 months after the second allogeneic HCT from a matched unrelated donor.
36[3]	F	47	AML	t-AML		p.P560L	History of alkylating agents-containing chemotherapy for metastatic ovarian cancer; Achieved CR with intensive chemotherapy;
						p.P321L	Under consolidation chemotherapy.
37	F	71	AML	MPAL	p.A500fs	p.R525H	Refractory to intensive induction chemotherapy; Achieved CR after 2 courses of LDAC; Under LDAC chemotherapy.
38	M	69	AML	NOS	p.A500fs	p.R525H	Lost to follow-up; Died of unknown cause one month after diagnosis
39	M	54	AML	RGA		p.A100E	Achieved CR with intensive chemotherapy; Under consolidation chemotherapy.

Abbreviations: AML, acute myeloid leukemia; APL, acute promyelocytic leukemia; CR, complete remission; Dx, diagnosis; EB, excess blasts; HCT, hematopoietic cell transplantation; HI, hematologic improvement; ICUS, idiopathic cytopenia of undetermined significance; LDAC, low-dose cytarabine; MDS, myelodysplastic syndrome; MLD, multilineage dysplasia; MRC, myelodysplasia-related changes; No., number; NOS, not otherwise specified; PR, partial remission; RGA, recurrent genetic abnormalities; SD, stable disease; SLD, single lineage dysplasia; t-AML, therapy-related AML; U, unclassifiable.

Supplementary Figure 1. (A) Overall survival curves according to the presence of *DDX41* mutations. (B) Event-free survival curves according to the presence of *DDX41* mutations.



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