

Sample	Leukemia type	Age/Sex	Haplogroup	No. of cells	Total haplotype	Haplotype diversity
CD34+ and blast						
LAA	AML M0	61/F	H	95	16	0.593 ± 0.046
AGM	AML M1	62/M	B4b	96	11	0.645 ± 0.030
MFSS	AML M1	36/F	L2a	96	10	0.180 ± 0.053
JCS	AML M1	40/M	L3f	96	15	0.631 ± 0.030
DC	AML M1	15/M	L2a	96	18	0.741 ± 0.033
OAM <sup>a</sup>	AML M1	33/M	D1	92	11	0.280 ± 0.062
EMB	AML M1	16/M	D1	95	34	0.576 ± 0.063
ERR	AML M1	53/F	A2	94	6	0.124 ± 0.047
UPN21	AML	16/F	D1	82	7	0.164 ± 0.056
UPN1	AML M2	47/F	T2	146	10	0.286 ± 0.049
UPN16	2nd AML/MDS	74/F	J1c	92	5	0.183 ± 0.053
UPN22	Relapsed AML	11/F	H	65	20	0.833 ± 0.036
OAM <sup>b</sup>	Relapsed AML	36/M	D1	96	6	0.449 ± 0.046
UPN18	AML & prior NHL	62/F	A2	92	8	0.294 ± 0.061
UPN20	CML	53/M	A2	85	3	0.248 ± 0.056
UPN17	CML	27/F	X2b	80	17	0.829 ± 0.030
UPN2	AML from CMML	58/M	J1c	96	14	0.496 ± 0.055
UPN3	CLL	64/F	H	89	9	0.307 ± 0.064
UPN19 <sup>c</sup>	APML	21/F	H	94	22	0.765 ± 0.037
UPN19 <sup>d</sup>	APML	21/F	H	96	10	0.620 ± 0.039
Subtotal	F	F	F	1873	252	F
Donor 1	F	48/F	U5b	85	14	0.561 ± 0.052
Donor 2	F	44/M	L2a	92	16	0.558 ± 0.059
Donor 3	F	36/M	L0a1	93	12	0.558 ± 0.052
Donor 4	F	55/M	T2	93	14	0.661 ± 0.050
Donor 5	F	35/M	U5b	93	13	0.558 ± 0.053
Donor 6	F	35/M	M7a1	95	15	0.627 ± 0.049
Donor 7	F	32/M	U6	93	40	0.805 ± 0.041
Donor 8	F	25/M	H	93	38	0.800 ± 0.036
Donor 9	F	57/M	K1a	93	31	0.650 ± 0.059
Donor 10	F	39/M	pre-V	94	17	0.622 ± 0.041
Subtotal	F	F	F	924	210	F
Granulocyte						
UPN21	AML	16/F	D1	24	8	0.562 ± 0.120
UPN22	Relapsed AML	11/F	H	70	42	0.953 ± 0.016
UPN20	CML	53/M	A2	92	17	0.487 ± 0.062
UPN19	APML	21/F	H	79	23	0.748 ± 0.040

Subtotal	F	F	F	265	90	F
Donor 1	F	48/F	U5b	64	28	0.809 ± 0.048
Donor 2 <sup>e</sup>	F	44/M	L2a	82	27	0.706 ± 0.056
Donor 2 <sup>f</sup>	F	45/M	L2a	96	16	0.570 ± 0.055
Donor 3	F	36/M	L0a1	72	26	0.657 ± 0.065
Donor 4	F	55/M	T2	76	27	0.756 ± 0.049
Donor 5	F	35/M	U5b	82	31	0.645 ± 0.063
Subtotal	F	F	F	472	155	F

Note: The data of peripheral blood CD34+ cells and granulocytes for healthy donors 1 to 5 are from Ogasawara et al.<sup>2</sup> Single blasts at <sup>a</sup>diagnosis and <sup>b</sup>relapse were analyzed for patient OAM. Both <sup>c</sup>blasts (CD33+CD34-) and <sup>d</sup>normal CD34+ cells were analyzed for patient UPN19. Granulocytes from <sup>e</sup>peripheral blood and <sup>f</sup>bone marrow were analyzed for healthy donor 2. We counted the numbers of total hapotype (including the aggregate mtDNA type and the types that differ from aggregate sequence by substitution and/or indels) based on the mutations detected in each sample (see supplementary online Table 3). We utilized haplotype diversity ( $h$ )<sup>8</sup>, which was widely used in population genetic study of human and animal mtDNAs for measuring genetic diversity (e.g. references<sup>6,9</sup>), to assess the probability that two randomly chosen cells were different in order to measure the heterogeneity level in each sample according to

$$h = \frac{n}{n-1} \left( 1 - \sum_{i=1}^k p_i^2 \right)$$

, where  $n$  is the total number of cells analyzed for the donor or patient,  $k$  is the number of haplotypes, and  $p_i$  is sample frequency of the  $i$ -th haplotype.<sup>8</sup> There is statistically difference on haplotype diversity between the leukemia group and the normal control group (F test to compare variances,  $P = 0.004$ ; unpaired t-test with Welch's correction,  $P = 0.007$ ; supplementary online Figure 1).