Sample & Cell type	Haplotype [a]	No. of Cell
LAA	Aggregate sequence	56
CD34+	+16068T>Y	1
	+16099C>Y	1
	+16354C>Y	1
	+16391G>R	1
	+16459C>Y	1
	+16473G>R	1
	+369C>Y	1
	+441C>Y	1
	+557C>Y	1
	+568C>Y	1
	+8CT6C/9CT6C, 571C>Y	1
	7CT6C/8CT6C	1
	8CT6C/9CT6C	24
	8CT6C/9CT6C/10CT6C	2
	9CT6C	1
AGM	Aggregate sequence [b]	42
CD34+	+16339C>Y, 9CT6C/10CT6C/11CT6C	1
	+16378C>Y, 9CT6C/10CT6C, 378C>Y	1
	+129T>Y, 9CT6C/10CT6C	1
	+140C>Y, 9CT6C/10CT6C	2
	+183A>A/T, 9CT6C/10CT6C	1
	+268C>Y, 9CT6C/10CT6C	1
	+9CT6C/10CT6C, 378C>Y	1
	9CT6C, 523-524insAC/non-ins	1
	9CT6C	6
	9CT6C/10CT6C	39
MFSS	Aggregate sequence [c]	87
CD33+CD34+	+16131T>Y	1
	+16190C>Y	1
	+52T>Y	1
	+71G>R	1
	+135T>Y	1
	+139T>C	1
	+422T>Y	1
	+556A>R	1
	523-524delAC/non-del	1
JCS	Aggregate sequence	40
CD33+CD34+	+16025T>Y	1
	+16148C>Y	1
	+16331A>R, 8CT6C/9CT6C	1
	+16353C>Y	1

	+16365C>Y, 8CT6C/9CT6C	1
	+16449C>Y	1
	+16465C>Y, 588T>Y	1
	+56A>R	1
	+112C>Y, 8CT6C/9CT6C	1
	+185G>R, 8CT6C/9CT6C	1
	+8CT6C/9CT6C, 384A>R	1
	+532A>R	1
	523-524delAC/non-del	1
	8CT6C/9CT6C	43
DC CD33+CD34+	Aggregate sequence	40
	+16087A>R, 16150C>Y	1
	+16110G>R	1
	+16123T>Y, 16150C>Y, 8CT6C/9CT6C	1
	+16150C>Y	27
	+16150C>Y, 523-524insAC/non-ins	1
	+16150C>Y, 268C>Y	1
	+16150C>Y, 8CT6C/9CT6C	2
	+16213G>R, 16539A>R	1
	+16228C>Y, 8CT6C/9CT6C	1
	+16257C>Y	1
	+16343A>R	1
	+16359T>Y	1
	+59T>Y	1
	+541C>Y	1
	523-524insAC/non-ins	2
	7CT6C/8CT6C	3
	8CT6C/9CT6C	10
OAM CD33+CD34+	Aggregate sequence	78
	+16098A>R	1
	+16237A>R	1
	+16321C>Y	1
	+16488C>C/A	5
	+16494C>Y	1
	+16498C>Y	1
	+16533T>Y	1
	+64C>Y	1
	+460T>Y	1
	523-524insAC/non-ins	1
relapsed OAM CD33+CD34+	Aggregate sequence [d]	67
	+16103A>R, 16530A>R	1
	+16164A>A/C	1
	+16188insC/non-ins	1
	+16295C>Y, 16488C	1
	16488C	25
EMB	Aggregate sequence	62

CD33+CD34+	+16064T>Y, 523-524delAC/non-del	1
	+16204G>R	1
	+16249T>Y, 403T>Y	1
	+16277A>R, 75G>R, 142T>Y	1
	+16290C>Y	1
	+16292C>Y	1
	+16292delC/non-del	1
	+16297T>Y	1
	+16298T>Y	1
	+16324T>Y	1
	+16325T>Y	1
	+16366delC/non-del	1
	+16380delC/non-del	1
	+16381T>Y	1
	+16443T>Y	1
	+16510A>R	1
	+16550T>Y	1
	+10T>Y	1
	+42T>Y	1
	+89T>Y	1
	+99T>Y	1
	+152T>Y, 460T>Y	1
	+267T>Y	1
	+321T>Y	1
	+383T>Y	1
	+390A>A/C	1
	+399T>Y	1
	+399T>Y, 495C>Y	1
	+411C>Y	1
	+431C>Y	1
	+538A>R	1
	+556A>R	1
	+583G>R	1
ERR	Aggregate sequence	88
CD33+CD34+	+16262C>Y	2
	+16353C>Y	1
	+16424T>Y	1
	+16429C>Y	1
	+16508C>Y	1
UPN21	Aggregate sequence	75
CD34+	+16380insC/non-ins	1
	+16560C>Y	1
	+146T>Y	1
	+415A>R, 470A>R	1
	523-524insAC/non-ins	1
	8CT6C/9CT6C	2
UPN21	Aggregate sequence	16

granulocyte	+16132A>R	1
	+16222C>Y	1
	+16253A>R	1
	+16293A>R	1
	+13A>R	1
	+343C>Y	1
	8CT6C/9CT6C	2
UPN1 CD13+CD33+	Aggregate sequence	123
	+16123T>Y	1
	+16286C>Y	1
	+16291C>Y	1
	+143G>R	1
	+390A>R, 493A>R	1
	8CT6C	1
	8CT6C/9CT6C	4
	9CT6C/10CT6C	10
	8CT6C/9CT6C/10CT6C	3
UPN16 CD34+	Aggregate sequence	83
	+16356T>Y, 16542C>Y	1
	+405T>Y	1
	+577G>R	1
	7CT6C/8CT6C	6
UPN22 CD34+	Aggregate sequence	23
	+16037A>R, 16320C>T	1
	+16320C>T	2
	+16320C>T, 146T>Y, 195T>C	1
	+16320C>T, 195T>C	1
	+16320C>T, 195T>C, 8CT6C/9CT6C	1
	+16320C>T, 9CT6C	1
	+16320C>Y	4
	+16320C>Y, 16380delC/non-del	1
	+16320C>Y, 195T>Y, 8CT6C/9CT6C	1
	+16320C>Y, 8CT6C/9CT6C	1
	+16320C>Y, 8CT6C/9CT6C, 515A>R	1
	+16222C>Y, 16239C>Y, 319T>Y	1
	+(16222), 16239C>T, (319)	1
	+(16222), 16239C>T, 9CT6C, (319), 415A>R	1
	+15C>Y, 9CT6C/10CT6C	1
	9CT6C	6
	8CT6C/9CT6C	12
	8CT6C/9CT6C/10CT6C	4
	9CT6C/10CT6C	1
UPN22 granulocyte	Aggregate sequence	12
	+16029T>Y, 378C>C/A	1
	+16030C>Y	1
	+16068T>Y	1

	+16122A>R, 9CT6C	1
	+16167C>Y, 8CT6C/9CT6C	1
	+16189T>C	1
	+16285A>R, 9CT6C/10CT6C	1
	+16320C>T	1
	+16320C>T, 146T>C, 195T>C	1
	+16320C>T, 195T>C	1
	+16320C>T, 195T>C, 204T>C	1
	+16320C>T, 386C>Y, 550A>R	1
	+16320C>T, 7CT6C	1
	+16320C>T, 8CT6C/9CT6C	1
	+16320C>Y	3
	+16320C>Y, 16329G>G/C, 60T>Y, 7CT6C/8CT6C	1
	+16320C>Y, 16452T>Y, 8CT6C/9CT6C	1
	+16320C>Y, 515A>A/C	1
	+16320C>Y, 8CT6C/9CT6C	2
	+16357T>C, 461C>Y	1
	+16394C>Y, 8CT6C/9CT6C, 456C>Y	1
	+16397T>Y	1
	+16409T>Y	1
	+16450G>G/T, 8CT6C/9CT6C	1
	+16456G>R, 16466A>R, 8CT6C/9CT6C	1
	+16568T>Y	1
	+(16222), 16239C>T, 16368T>Y, (319)	1
	+(16222), 16239C>T, 8CT6C/9CT6C, (319)	2
	+(16222), 16239C>T, 9CT6C, (319)	1
	+(16222), 16239C>T, 9CT6C, (319), 645A>R	1
	+16222C>Y, 16239C>Y, 319T>Y	2
	+16222C>Y, 16239C>Y, 319T>Y, 8CT6C/9CT6C	1
	+186C>T	1
	+195T>C	1
	+266T>Y, 9CT6C	1
	7CT6C/8CT6C	1
	8CT6C/9CT6C	9
	8CT6C/9CT6C, 523-524insAC/non-ins	1
	8CT6C/9CT6C/10CT6C	3
	8CT6C/9CT6C/10CT6C, 523-524insAC/non-ins	1
	9CT6C	3
UPN18	Aggregate sequence	77
CD34+	+16094T>Y, 16158A>R	1
	+16179C>Y, 16186C>Y	1
	+16188C>Y	1
	+16484T>C	1
	+573insC/non-ins	1
	7CT6C/8CT6C	2
	8CT6C/9CT6C	8
UPN20	Aggregate sequence	73
CD34+	+16462T>Y	1

	523-524insAC/non-ins	11
UPN20 granulocyte	Aggregate sequence	65
	+16259C>C/A, 523-524insAC/non-ins	1
	+16053C>Y, 16500T>Y, 480T>Y	1
	+16150C>T	1
	+16245C>Y	1
	+16432A>R, 30T>Y	1
	+58T>Y	1
	+98C>T	1
	+139T>Y	1
	+438delC/non-del	1
	+466T>Y	1
	+522C>Y	1
	+571C>Y	1
	+598A>R	1
	+602C>Y	1
	523-524insAC/non-ins	12
	523-524non-del	1
UPN17 CD34+	Aggregate sequence [e]	28
	+16081A>R, 189G, 559C>Y	1
	+16183A>Y, 8CT6C/9CT6C/10CT6C	1
	+16348C>Y	1
	+16359T>Y, 189A, 424T>Y	1
	+16452T>Y, 189A, 454T>Y, 591C>Y, 8CT6C/9CT6C/10CT6C	1
	+189G, 7CT6C/8CT6C/9CT6C	1
	+189G	11
	+189G, 514C>Y	1
	+189G, 8CT6C/9CT6C/10CT6C	3
	+192T>Y, 8CT6C/9CT6C/10CT6C	1
	+442T>Y, 189A	1
	189A	7
	189A, 8CT6C/9CT6C/10CT6C	6
	189A, 9CT6C/10CT6C	1
	8CT6C/9CT6C/10CT6C	12
	9CT6C/10CT6C	3
UPN2 CD33+CD34+	Aggregate sequence	66
	+16052C>Y	1
	+16132A>R, 260G>R	1
	+16188C>Y	1
	+16203A>R, 281A>R	1
	+16264C>Y, 260G>R	1
	+16353C>Y	1
	+16467C>Y	1
	+16565C>Y	1
	+185G>R	1
	+185G>R, 260G>R	1

	+260G>R	18
	+260G>R, 313C>Y	1
	+260G>R, 455T>Y	1
UPN3	Aggregate sequence [f]	74
CD5+CD33-	+16278C>Y	1
	+16328C>Y, 9CT6C/10CT6C	1
	+16565C>Y	1
	+42T>Y	1
	8CT6C	3
	9CT6C	3
	9CT6C/10CT6C	1
	8CT6C/9CT6C/10CT6C	4
UPN19	Aggregate sequence	52
CD34+	+16025T>Y	1
	+16439C>T	1
	+16546C>Y	1
	+185G>R	1
	8CT6C	2
	8CT6C/9CT6C	8
	9CT6C/10CT6C	28
	8CT6C/9CT6C/10CT6C	1
	523-524insAC/non-ins	1
UPN19	Aggregate sequence	41
CD33+CD34-	+16068T>Y	1
	+16088T>Y, 9CT6C/10CT6C	1
	+16101T>Y	1
	+16152T>Y	1
	+16173C>Y, 9CT6C/10CT6C	1
	+16191C>Y	1
	+16271T>Y, 9CT6C/10CT6C	1
	+16471G>R	1
	+27C>Y	1
	+30T>Y	1
	+40T>Y, 62G>R, 8CT6C/9CT6C	1
	+95A>R, 9CT6C/10CT6C	1
	+133T>Y, 9CT6C/10CT6C	1
	+135T>Y, 9CT6C/10CT6C	1
	+269C>Y, 9CT6C/10CT6C	1
	523-524insAC/non-ins	2
	8CT6C/9CT6C	10
	8CT6C/9CT6C, 523-524insAC/non-ins	2
	9CT6C/10CT6C	18
	8CT6C/9CT6C/10CT6C	4
	9CT6C/10CT6C/11CT6C	2
UPN19	Aggregate sequence	34
granulocyte	+16146A>R	1

	+16189T>Y	1
	+16223C>Y	1
	+16233A>R, 472A>R	1
	+16237A>R	1
	+16239C>Y, 146T>Y, 8CT6C/9CT6C/10CT6C	1
	+16257C>Y	1
	+16258A>R	1
	+16278C>Y	1
	+147C>Y	1
	+230A>R, 9CT6C/10CT6C	1
	+374A>G	1
	+498delC	1
	+508A>R	1
	+511C>Y	1
	+645A>R	1
	523-524insAC/non-ins	3
	8CT6C	1
	8CT6C/9CT6C	2
	9CT6C/10CT6C	21
	10CT6C/11CT6C	1
	9CT6C/10CT6C/11CT6C	1
Donor 6 [g]	Aggregate sequence	55
CD34+	+16039G>R	1
	+16128C>T	1
	+16131T>Y	1
	+16172T>Y	1
	+16214C>Y	1
	+16240A>R	1
	+16267C>C/A	1
	+(16324)	1
	+412G>R	1
	+480T>Y	1
	8CT6C/9CT6C	7
	9CT6C/10CT6C	18
	8CT6C/9CT6C/10CT6C	4
	9CT6C/10CT6C/11CT6C	1
Donor 7	Aggregate sequence	40
CD34+	+16022T>Y, 8CT6C/9CT6C	1
	+16025T>Y	1
	+16076C>Y, 16409T>Y, 16502T>Y	1
	+16086T>Y	1
	+16087A>R, 8CT6C/9CT6C	1
	+16096G>R	1
	+16100A>R	1
	+16131T>Y	1
	+16191C>Y	1
	+16211C>Y	1
	+16224T>T/G, 8CT6C/9CT6C	1

	+16288T>Y	1
	+16309A>R	1
	+16378C>Y, 16409T>Y	1
	+16381T>Y	1
	+16408C>Y, 8CT6C/9CT6C	1
	+16468T>Y, 379A>R	1
	+16490G>R	1
	+16507C>Y, 8CT6C/9CT6C	1
	+16539A>A/C	1
	+16544T>Y	1
	+10T>Y, 146T>Y, 7CT6C/8CT6C	1
	+14T>Y	1
	+59T>Y, 145C>Y	1
	+146T>Y	1
	+161T>Y	1
	+204T>Y, 7CT6C/8CT6C	1
	+230A>R	1
	+234A>R	1
	+305C>Y, 8CT6C/9CT6C	1
	+372T>Y	1
	+390A>R	1
	+578T>Y	1
	+8CT6C/9CT6C, 379A>R	1
	+8CT6C/9CT6C, 410G>R, 549C>Y	1
	523-524delAC/non-del	1
	7CT6C	2
	7CT6C/8CT6C	5
	8CT6C/9CT6C	10
Donor 8	Aggregate sequence	37
CD34+	+16038A>R, 8CT6C/9CT6C	1
	+16082C>Y, 7CT6C	1
	+16189T>Y	1
	+16238T>Y	1
	+16275A>R	1
	+16282C>Y	1
	+16282C>Y, 16391G>R	1
	+16296C>Y	1
	+16308T>Y, 525C>Y	1
	+16315T>Y, 8CT6C/9CT6C	1
	+16387A>R	1
	+16391G>R	1
	+16392T>Y	1
	+16392T>Y, 16508C>C/A, 260G>R	1
	+16400C>Y, 8CT6C/9CT6C	1
	+16403C>Y, 8CT6C/9CT6C	1
	+16502T>Y, 8CT6C/9CT6C	1
	+16533T>Y, 8CT6C/9CT6C	1
	+16537C>Y	1
	+11C>Y, 8CT6C/9CT6C	1

	+12T>Y	1
	+16A>R, 8CT6C/9CT6C	1
	+39C>Y, 8CT6C/9CT6C	1
	+42T>Y, 154T>Y, 491C>Y, 523-524delAC/non-del	1
	+77A>R, 8CT6C/9CT6C	1
	+131T>Y	1
	+142T>Y	1
	+146T>Y	1
	+273C>Y, 8CT6C/9CT6C	1
	+345C>Y	1
	+346T>Y	1
	+441C>Y	1
	+538A>R	1
	+570C>Y	1
	+8CT6C/9CT6C, 378C>Y	1
	8CT6C/9CT6C	20
	8CT6C/9CT6C/10CT6C	1
Donor 9[h] CD34+	Aggregate sequence	55
	+16086T>Y, 588T>Y	1
	+16095C>C/A, 16354C>T	1
	+16129G>A, 523-524insAC/non-ins	1
	+16131T>Y	1
	+16144T>Y	1
	+16356T>Y	1
	+16368T>C	1
	+16390G>R, 523-524insAC/non-ins	1
	+16526G>R	1
	+56insT, 460T>Y	1
	+56A>T	3
	+56A>T, 545G>R	1
	+64C>T	1
	+(73)	1
	+150C>Y, 292T>Y	1
	+152T>C	1
	+185G>R, 195T>A	1
	+195T>A	1
	+195T>A, 7CT6C/8CT6C	1
	+195T>T/A	4
	+195T>T/A, 383T>Y	1
	+251G>R	1
	+291A>A/T	1
	+292T>Y	2
	+292T>Y, 588T>Y, 523-524insAC/non-ins	1
	+370C>Y	1
	+535C>Y	1
	+541C>Y	1
	+587C>Y	1
	523-524insAC/non-ins	3

Donor 10	Aggregate sequence		51
CD34+	+16124T>Y		1
	+16217T>Y		1
	+16239C>Y, 8CT6C/9CT6C		1
	+16293A>G	1	
	+16378C>Y, 8CT6C/9CT6C		1
	+16506T>Y		1
	+16537C>Y, 16565C>Y, 8CT6C/9CT6C		1
	+182C>Y, 8CT6C/9CT6C		1
	+343C>Y, 8CT6C/9CT6C		1
	+410G>R		1
	+571C>Y		1
	+8CT6C/9CT6C, 593T>Y		1
	7CT6C		1
	7CT6C/8CT6C		1
	7CT6C/8CT6C/9CT6C		1
	8CT6C/9CT6C		28
Donor 2	Aggregate sequence		61
Granulocyte from BM	+16028T>Y, 16372T>Y		1
	+16054A>G	1	
	+16105T>Y		1
	+16114C>Y		1
	+16127A>R		1
	+16131T>Y		1
	+16208G>R		1
	+16232C>Y		1
	+16331A>R		1
	+16390G>R		1
	+16419C>Y, 16468T>Y		1
	+485T>Y		1
	8CT6C		2
	8CT6C/9CT6C		5
	9CT6C/10CT6C		16

[a] Mutation was scored relative to the revised CRS [1]. A site heterogeneous for both thymidine (T) and cytidine (C) was abbreviated as Y, and heterogeneous for adenosine (A) and guanosine (G) was abbreviated as R. We highlight the status of the length mutations of the C-tract at region 303-309 and the dinucleotide AC repeat at region 515-524 in these cells differ from the aggregated sequence by listing all of the status, e.g. 8CT6C means region 303-315 is CCCCCCCCTCCCCCC in the cell, 8CT6C/9CT6C means region 303-315 has heteroplasmy of CCCCCCCCTCCCCCC and

CCCCCCCCCTCCCCCC in the cell, 523-524delAC means this cell contains 4 repeats of AC at region 515-524, while 523-524insAC/non-ins means co-existing of 6 and 5 repeats of AC at region 515-524 in the cell. +, extra mtDNA nucleotide changes compared with the aggregate sequence. Back mutations relative to the sequence variations in aggregate sequence are in brackets.

[b] The aggregate sequence of AGM contains 9CT6C/10CT6C/11CT6C in region 303-315 in HVS-II. The length mutations of the C-tract in region 16184-16193 due to 16189T>C mutation were not scored.

[c] The length mutations of the C-tract in region 16184-16193 due to 16189T>C mutation in MFSS were discarded.

[d] The aggregate sequence of the relapsed patient OAM contains 16488C>C/A compared with the consensus sequence of this individual in supplementary online Table 2. The status of no mutation at site 16488 was demonstrated as 16488C.

[e] The aggregate sequence of UPN17 contains 189A>R and 8CT6C/9CT6C. The status of a homoplasmic mutation or no mutation at site 189 was highlighted as 189G and 189A, respectively. The length mutations of the C-tract in region 16184-16193 due to 16189T>C mutation were not scored.

[f] The aggregate sequence of UPN3 contains 8CT6C/9CT6C in region 303-315 in HVS-II.

[g] The list of nucleotide substitutions in donor 6 is a consensus result of triplicates of independent amplifications using the original single cell lysate.

[h] The length mutations of the C-tract in region 16184-16193 due to 16189T>C mutation in donor 9 were not counted.