Clinical significance of chromatin-spliceosome acute myeloid leukemia: a report from the Northern Italy Leukemia Group (NILG) randomized trial 02/06

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Supplemental Appendix

to C. Caprioli et al.:

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Supplemental Methods

Molecular analyses

Molecular analyses were performed on mononuclear cells obtained by Ficoll-gradient centrifugation from peripheral blood and/or bone marrow containing at least 20% blasts.

Libraries for targeted NGS were sequenced and demultiplexed on a MiSeq or MiniSeq instrument (Illumina). Variants not reaching the minimum target coverage of 1000x were confirmed by conventional methods. The detection limit/sensitivity for identified variants was set to 5% variant allele frequency, as per manufacturer indication. Frameshift and nonsense variants were always considered as relevant mutations. Single nucleotide variants were retained in the absence of description as genetic polymorphism into public databases of human polymorphisms (NCBI dbSNP (http://www.ncbi.nlm.nih.gov/snp; Build 137) and Exac (http://exac.broadinstitute.org/), taking into account both allele frequency and ethnicity of patients. Functional interpretation for missense variants was performed using SIFT 1.03 (http://sift.jcvi.org) and PolyPhen2.0 (http://genetics.bwh.harvard.edu/pph2). For alterations of splicing sites and splicing related regions, we used the Human Splicing tool (http://www.umd.be/HSF3) to predict alterations in the splicing process. Indeed, the description of the identified mutations in literature was checked in COSMIC database (http://cancer.sanger.ac.uk/cancergenome/projects/cosmic).

Supplemental Data

Supplemental Table 1. Gene panels and regions investigated by targeted NGS using Trusight Myeloid (Illumina, San Diego, California, USA) and Sophia Myeloid Solution (Sophia Genetics SA, Saint Sulpice, Switzerland) (indicated with *).

Gene	Target Region (Exons)	Gene	Target Region (Exons)	Gene	Target Region (Exons)
*ABL1**	4-6 4-9*	*FLT3**	14-15, 20 13-15, 20*	*NRAS**	2, 3*
*ASXL1**	12 9,11,12*	*GATA1*	2	*PDGFRA*	12, 14, 18
ATRX	8-10, 17-31	*GATA2*	2-6	*PHF6*	Full
BCOR	Full	*GNAS*	8, 9	*PTEN*	5, 7
BCORL1	Full	*HRAS**	2, 3*	*PTPN11*	3, 13 3, 7-13*
*BRAF**	15*	*IDH1**	4	*RAD21*	Full
*CALR**	9*	*IDH2**	4	*RUNX1**	Full*
*CBL**	8, 9*	*IKZF1*	Full	*SETBP1**	4 (partial) 4*
CBLB	9, 10	*JAK2**	12, 14 Full*	*SF3B1**	13-16 10-16*
CBLC	9, 10	*JAK3*	13	*SMC1A*	2, 11,16-17
CDKN2A	Full	*KDM6A*	Full	*SMC3*	10, 13, 19, 23, 25, 28
*CEBPA**	Full*	*KIT**	2, 8-11, 13, 17 2, 8-11, 13, 17, 18*	*SRSF2**	1*
*CSF3R**	14-17 Full*	*KRAS**	2, 3*	*STAG2*	Full
CUX1	Full	*MLL*	5-8	*TET2**	3-11 Full*
*DNMT3A**	Full*	*MPL**	10*	*TP53**	2-11 Full*
*ETV6**	Full*	*MYD88*	3-5	*U2AF1**	2, 6*
*EZH2**	Full*	*NOTCH1*	26-28, 34	*WT1**	7, 9 6-10*
FBXW7	9-11	*NPM1**	12 11-12*	*ZRSR2**	Full*

Supplemental Table 2. Annotation of gene mutations in the prospective study cohort.

UPN	AML category	Gene	Chr	Coordinate (Hg19)	Variant	Type	Consequence	VAF	Transcript	c.DNA	protein	Sift	PolyPhen
6	CS-AML	ASXL1	20	31022904	GA>GA/G	deletion	frameshift	40.12	NM_015338.5	c.2390delA	p.Ser798ValfsTer20		
46	sAML	ASXL1	20	31022628	G>G/T	snv	stop gained	47.88	NM_015338.5	c.2113G>T	p.Glu705Ter		
62	CS-AML	ASXL1	20	31022441	AGGGGGGGG>AGGGGGGGG/AGGGGGGGGG	insertion	frameshift	33.74	NM_015338	c.1934dupG	p.Gly646Trpfs*12		
126	CS-AML	ASXL1	20	31023014	TC>TC/T	deletion	frameshift	46.21	NM_015338.5	c.2500delC	p.His834IlefsTer4		
149	CS-AML	ASXL1	20	31022441	AGGGGGGGG>AGGGGGGGG/AGGGGGGGGG	insertion	frameshift	35.17	NM_015338	c.1934dupG	p.Gly646Trpfs*12		
226	sAML	ASXL1	20	31022441	A>A/AG	insertion	frameshift	38.09	NM_015338.5	c.1926_1927insG	p.Gly646TrpfsTer12		
265	CS-AML	ASXL1	20	31022441	A>A/AG	insertion	frameshift	35.01	NM_015338.5	c.1926_1927insG	p.Gly646TrpfsTer12		
369	sAML	ASXL1	20	31022441	A>A/AG	insertion	frameshift	30.89	NM_015338.5	c.1926_1927insG	p.Gly646TrpfsTer12		
470	CS-AML	ASXL1	20	31022441	A>A/AG	insertion	frameshift	27.65	NM_015338.5	c.1926_1927insG	p.Gly646TrpfsTer12		
548	*de novo*	ASXL1	20	31023033	C>C/A	snv	missense	11.08	NM_015338.5	c.2518C>A	p.Pro840Thr	deleterious (0.04)	possibly damaging (0.452)
605	CS-AML	ASXL1	20	31022441	A>A/AG	insertion	frameshift	33.73	NM_015338.5	c.1926_1927insG	p.Gly646TrpfsTer12		
786	CS-AML	ASXL1	20	31024023	TTA>TTA/T	deletion	frameshift	40.49	NM_015338.5	c.3509_3510delTA	p.Leu1170Ter		
1148	CS-AML	ASXL1	20	31022441	A>A/AG	insertion	frameshift	10.25	NM_015338.5	c.1926_1927insG	p.Gly646TrpfsTer12		
1201	sAML	ASXL1	20	31022441	AGGGGGGGG>AGGGGGGGG/AGGGGGGGGG	insertion	frameshift	27.96	NM_015338	c.1934dupG	p.Gly646Trpfs*12		
1308	CS-AML	ASXL1	20	31023383	T>T/TA	insertion	frameshift	42.74	NM_015338.5	c.2868_2869insA	p.Thr957AsnfsTer13		
110	*de novo*	BCOR	X	39914620	C>C/T	snv	splicing	44.59	NM_001123385.1	c.4741+1G>A	p.?		
258	CS-AML	BCOR	X	39933862	TAGAG>TAGAG/T	deletion	frameshift	20.2	NM_001123385.1	c.733_736delCTCT	p.Leu245ThrfsTer20		
635	*de novo*	BCOR	X	39933853	G>G/A	snv	missense	21.83	NM_001123385.1	c.746C>T	p.Pro249Leu	deleterious (0)	probably damaging (0.999)
846	CS-AML	BCOR	X	39914693	TG>TG/T	deletion	frameshift	12.09	NM_001123385.1	c.4668delC	p.Thr1557ProfsTer11		
997	CS-AML	BCOR	X	39911623	GA>GA/G	deletion	frameshift	28.87	NM_001123385.1	c.5006delT	p.Val1669AlafsTer5		
1110	sAML	BCOR	X	39922127	T>T/A	snv	stop gained	49.21	NM_001123385.1	c.4045A>T	p.Lys1349Ter		
1148	CS-AML	BCOR	X	39921466	CCGG>CCGG/C	deletion	inframe deletion	20.16	NM_001123385.1	c.4351_4353delCCG	p.Pro1451del		
1148	CS-AML	BCOR	X	39922057	T>T/C	snv	missense	12.05	NM_001123385.1	c.4115A>G	p.Glu1372Gly	tolerated (0.13)	possibly damaging (0.359)
1270	CS-AML	BCOR	X	39933749	C>C/T	snv	missense	54.79	NM_001123385.1	c.850G>A	p.Asp284Asn	deleterious (0.01)	possibly damaging (0.725)
1343	sAML	BCOR	X	39923086	GT>GT/G	deletion	frameshift	25.25	NM_001123385.1	c.3621delA	p.Lys1207AsnfsTer31		
548	*de novo*	BCORL1	X	129185889	A>A/G	snv	missense	27.91	NM_021946.4	c.4751A>G	p.Asp1584Gly	deleterious (0.01)	possibly damaging (0.712)
548	*de novo*	BCORL1	X	129148079	C>C/G	snv	missense	13.4	NM_021946.4	c.1331C>G	p.Thr444Ser	tolerated (0.38)	benign (0.018)
1110	sAML	BCORL1	X	129148456	G>G/GC	insertion	frameshift	28.71	NM_021946.4	c.1708_1709insC	p.Ser572GlnfsTer31		
1110	sAML	BCORL1	X	129148479	A>A/AG	insertion	frameshift	18.46	NM_021946.4	c.1731_1732insG	p.Pro578AlafsTer25		
1110	sAML	BCORL1	X	129148480	C>C/G	snv	missense	17.68	NM_021946.4	c.1732C>G	p.Pro578Ala	tolerated (0.61)	benign (0.028)
1343	sAML	BCORL1	X	129189840	G>G/A	snv	stop gained	31.9	NM_021946.4	c.4865G>A	p.Trp1622Ter		
166	CS-AML	CBL	11	119149251	G>G/A	snv	missense	32.29	NM_005188.3	c.1259G>A	p.Arg420Gln		probably damaging (0.998)
319	*de novo*	CBL	11	119149241	C>C/T	snv	missense	91.96	NM_005188.3	c.1249C>T	p.Pro417Ser		probably damaging (1)

UPN	AML category	Gene	Chr	Coordinate (Hg19)	Variant	Type	Consequence	VAF	Transcript	c.DNA	protein	Sift	PolyPhen
854	*de novo*	CBL	11	119148931	G>G/A	snv	missense	42.68	NM_005188.3	c.1151G>A	p.Cys384Tyr		probably damaging (0.999)
1285	*de novo*	CBL	11	119149251	G>G/A	snv	missense	31.25	NM_005188.3	c.1259G>A	p.Arg420Gln		probably damaging (0.998)
1324	sAML	CBL	11	119148931	G>G/A	snv	missense	93.46	NM_005188.3	c.1151G>A	p.Cys384Tyr		probably damaging (0.999)
319	*de novo*	CDKN2A	9	21974792	G>G/T	snv	stop gained	51.41	NM_001195132.1	c.35C>A	p.Ser12Ter		
66	*de novo*	CEBPA	19	33793265	AGGTGGCT>AGGTGGCT/A	deletion	frameshift	48.57	NM_004364.3	c.49_55delAGCCACC	p.Ser17CysfsTer141		
66	*de novo*	CEBPA	19	33792378	G>G/GCACCTTCTGCTGCGTCTC	insertion	inframe insertion	36.72	NM_004364.3	c.925_942dupGAGACGCAGCAGAAGGTG	p.Glu309_Val314dup		
80	*de novo*	CEBPA	19	33792392	G>G/GTCTTAGACT	insertion	inframe insertion	41.08	NM_004364.3	c.928_929insAGTCTAAGA	p.Thr310_Gln310insLysSerLys		
86	*de novo*	CEBPA	19	33793238	C>C/CAG	insertion	frameshift	46.57	NM_004364.3	c.82_83insCT	p.Ser28ThrfsTer133		
86	*de novo*	CEBPA	19	33792384	TCTG>TCTG/T	deletion	inframe deletion	45.94	NM_004364.3	c.934_936delCAG	p.Gln312del		
94	*de novo*	CEBPA	19	33792387	G>G/GCTGCGTCTCCACGTTGCGCTGCTTGGC	insertion	inframe	44	NM_004364	c.907_933dup	p.Ala303_Gln311dup		
94	*de novo*	CEBPA	19	33793082	TC>TC/TCC	insertion	frameshift	40.64	NM_004364	c.238dupG	p.Asp80Glyfs*28		
96	*de novo*	CEBPA	19	33793205	GGGCCCGCGCCCCGGGGAAA>GGGCCCGCGCCCCGGGGAAA/G	deletion	frameshift	59.64	NM_004364.3	c.97_115delTTTCCCCGGGGCGCGGGCC	p.Phe33ProfsTer121		
96	*de novo*	CEBPA	19	33792734	G>G/A	snv	missense	10.74	NM_004364.3	c.587C>T	p.Pro196Leu	tolerated (0.62)	benign (0.028)
196	*de novo*	CEBPA	19	33793154	C>C/CA	insertion	frameshift	44.81	NM_004364.3	c.166dupT	p.Cys56LeufsTer52		
196	*de novo*	CEBPA	19	33792437	G>G/T	snv	missense	43.72	NM_004364.3	c.884C>A	p.Ala295Glu	deleterious (0)	probably damaging (0.997)
241	*de novo*	CEBPA	19	33792731	G>G/GGCGGGT	insertion	inframe insertion	40.96	NM_004364.3	c.584_589dupACCCGC	p.His195_Pro196dup		
269	*de novo*	CEBPA	19	33792384	T>T/TCTG	insertion	inframe insertion	90.42	NM_004364.3	c.934_936dupCAG	p.Gln312dup		
274	*de novo*	CEBPA	19	33792391	C>C/CGAG	insertion	missense	46.23	NM_004364.3	c.929_930insCTC	p.Thr310_Gln311insSer		
274	*de novo*	CEBPA	19	33793252	C>C/CG	insertion	frameshift	29.47	NM_004364.3	c.68dupC	p.His24AlafsTer84		
296	*de novo*	CEBPA	19	33792381	C>C/CCTT	insertion	inframe insertion	36.92	NM_004364.3	c.937_939dupAAG	p.Lys313dup		
297	*de novo*	CEBPA	19	33792391	C>C/CA	insertion	frameshift	44.34	NM_004364.3	c.929_930insT	p.Gln311AlafsTer10		
297	*de novo*	CEBPA	19	33792392	G>G/GAC	insertion	frameshift	44.7	NM_004364.3	c.928_929insGT	p.Thr310SerfsTer9		
297	*de novo*	CEBPA	19	33793203	CG>CG/C	deletion	frameshift	55.43	NM_004364.3	c.117delC	p.Ala40ArgfsTer120		
362	*de novo*	CEBPA	19	33792393	T>T/TCTC	insertion	inframe insertion	45.99	NM_004364.3	c.925_927dupGAG	p.Glu309dup		
402	*de novo*	CEBPA	19	33792350	A>A/T	snv	missense	53.69	NM_004364.3	c.971T>A	p.Leu324Gln	deleterious (0.01)	probably damaging (1)
402	*de novo*	CEBPA	19	33792381	C>C/CCTT	insertion	inframe insertion	37.69	NM_004364.3	c.937_939dupAAG	p.Lys313dup		
413	CS-AML	CEBPA	19	33793242	TGGGCGCGTGCG>TGGGCGCGTGCG/T	deletion	frameshift	27.49	NM_004364.3	c.68_78delCGCACGCGCCC	p.Pro23GlnfsTer81		
441	*de novo*	CEBPA	19	33792833	TCC>TCC/TCCCC	insertion	frameshift	46.37	NM_004364	c.486_487dupGG	p.Glu163Glyfs*156		
447	sAML	CEBPA	19	33792891	CCAGCCTGCCGTCCAGGTAGCCGGCGGCCGCG>CCAGCCTGCCGTCCAGGTAGCCGGCGGCCGCG/C	deletion	frameshift	29	NM_004364	c.399_429del	p.Cys133Trpfs*17		
508	*de novo*	CEBPA	19	33792393	TCTCCACGTTGCGCTG>TCTCCACGTTGCGCTG/T	deletion	inframe deletion	42.8	NM_004364.3	c.913_927delCAGCGCAACGTGGAG	p.Gln305_Glu309delinsdel		
521	*de novo*	CEBPA	19	33792381	C>C/CCTT	insertion	inframe	43.29	NM_004364	c.937_939dupAAG	p.Lys313dup		
521	*de novo*	CEBPA	19	33793033	GCCC>GCCC/GCC	deletion	frameshift	41.37	NM_004364	c.287delG	p.Gly96Alafs*64		

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835	*de novo*	CEBPA	19	33792384	T>T/TCTG	insertion	inframe insertion	53.02	NM_004364.3	c.934_936dupCAG	p.Gln312dup		
835	*de novo*	CEBPA	19	33793115	T>T/TCGTAAGA	insertion	frameshift	51.59	NM_004364.3	c.205_206insTCTTACG	p.Asp69ValfsTer41		
865	*de novo*	CEBPA	19	33792781	AGGG>AGGG/AGGGG	insertion	frameshift	31.11	NM_004364	c.539dupC	p.Tyr181Leufs*140		
866	*de novo*	CEBPA	19	33793252	CGGGGGG>CGGGGGG/CGGGGG	deletion	frameshift	41.05	NM_004364	c.68delC	p.Pro23Argfs*137		
866	*de novo*	CEBPA	19	33792381	C>C/CCTT	insertion	inframe	37.85	NM_004364	c.937_939dupAAG	p.Lys313dup		
877	*de novo*	CEBPA	19	33793141	C>C/CG	insertion	frameshift	53.09	NM_004364.3	c.179dupC	p.Ser61ValfsTer47		
924	*de novo*	CEBPA	19	33793073	TG>TG/T	deletion	frameshift	53.04	NM_004364.3	c.247delC	p.Gln83SerfsTer77		
924	*de novo*	CEBPA	19	33792386	T>T/TGCC	insertion	inframe insertion	45.34	NM_004364.3	c.934_935insGGC	p.Gln312_Lys312insArg		
1137	*de novo*	CEBPA	19	33792381	C>C/CCTT	insertion	inframe insertion	43.41	NM_004364.3	c.937_939dupAAG	p.Lys313dup		
1234	*de novo*	CEBPA	19	33793174	CG>CG/C	deletion	frameshift	26.03	NM_004364.3	c.146delC	p.Pro49ArgfsTer111		
1234	*de novo*	CEBPA	19	33792381	C>C/CCTT	insertion	inframe insertion	25.07	NM_004364.3	c.937_939dupAAG	p.Lys313dup		
1294	*de novo*	CEBPA	19	33793174	CGGGG>CGGGG/CGGG	deletion	frameshift	45.32	NM_004364	c.146delC	p.Pro49Argfs*111		
1294	*de novo*	CEBPA	19	33792381	C>C/CCTT	insertion	inframe	43.57	NM_004364	c.937_939dupAAG	p.Lys313dup		
1352	*de novo*	CEBPA	19	33792391	C>C/CCTT	insertion	missense	39.55	NM_004364.3	c.929_930insAAG	p.Thr310_Gln311insArg		
1352	*de novo*	CEBPA	19	33793258	G>G/GCT	insertion	frameshift	38.66	NM_004364.3	c.61_62dupAG	p.Ser21ArgfsTer140		
1375	*de novo*	CEBPA	19	33793242	TGGGCGCGTGCG>TGGGCGCGTGCG/T	deletion	frameshift	61.75	NM_004364.3	c.68_78delCGCACGCGCCC	p.Pro23GlnfsTer81		
1375	*de novo*	CEBPA	19	33792380	A>A/AT	insertion	frameshift	42.76	NM_004364.3	c.940_941insA	p.Val314AspfsTer7		
1375	*de novo*	CEBPA	19	33792381	C>C/CTT	insertion	frameshift	42.55	NM_004364.3	c.939_940insAA	p.Val314LysfsTer5		
66	*de novo*	CSF3R	1	36933434	G>G/A	snv	missense	39.27	NM_156039.3	c.1853C>T	p.Thr618Ile	deleterious (0)	probably damaging (0.999)
505	CS-AML	CSF3R	1	36932125	G>G/A	snv	missense	13.24	NM_156039.3	c.2425C>T	p.Pro809Ser	deleterious (0)	probably damaging (0.999)
1236	*de novo*	CSF3R	1	36932296	G>G/A	snv	stop gained	11.32	NM_156039.3	c.2254C>T	p.Gln752Ter		
102	CS-AML	CUX1	7	101837153	G>G/A	snv	missense	48.46	NM_001202543.1	c.1141G>A	p.Glu381Lys	tolerated (0.55)	possibly damaging (0.629)
548	*de novo*	CUX1	7	101821800	G>G/A	snv	missense	18.85	NM_001202543.1	c.913G>A	p.Ala305Thr	deleterious (0.02)	probably damaging (0.998)
548	*de novo*	CUX1	7	101840469	C>C/T	snv	missense	11.17	NM_001202543.1	c.1811C>T	p.Pro604Leu	deleterious (0)	probably damaging (0.971)
548	*de novo*	CUX1	7	101844649	C>C/T	snv	missense	10.78	NM_001202543.1	c.2105C>T	p.Ala702Val	tolerated (0.22)	benign (0.003)
10	*de novo*	DNMT3A	2	25457242	C>C/T	snv	missense	30.53	NM_022552.4	c.2645G>A	p.Arg882His	deleterious (0.04)	probably damaging (0.956)
14	*de novo*	DNMT3A	2	25458598	A>A/AGATGTCC	insertion	frameshift	45.1	NM_022552.4	c.2568_2574dupGGACATC	p.Leu859GlyfsTer7		
20	*de novo*	DNMT3A	2	25457242	C>C/T	snv	missense	50.51	NM_022552.4	c.2645G>A	p.Arg882His	deleterious (0.04)	probably damaging (0.956)
32	*de novo*	DNMT3A	2	25457242	C>C/T	snv	missense	40.59	NM_022552	c.2645G>A	p.Arg882His	tolerated (0.998)	benign (0.043)
43	*de novo*	DNMT3A	2	25457242	C>C/G	snv	missense	40.45	NM_022552.4	c.2645G>C	p.Arg882Pro	deleterious (0)	probably damaging (0.988)
64	*de novo*	DNMT3A	2	25457242	C>C/T	snv	missense	45.06	NM_022552.4	c.2645G>A	p.Arg882His	deleterious (0.04)	probably damaging (0.956)
92	*de novo*	DNMT3A	2	25463184	G>G/A	snv	missense	27.39	NM_022552	c.2309C>T	p.Ser770Leu	tolerated (0.996)	probably damaging (1)
101	*de novo*	DNMT3A	2	25457242	C>C/T	snv	missense	40.95	NM_022552.4	c.2645G>A	p.Arg882His	deleterious (0.04)	probably damaging (0.956)

UPN	AML category	Gene	Chr	Coordinate (Hg19)	Variant	Type	Consequence	VAF	Transcript	c.DNA	protein	Sift	PolyPhen
110	*de novo*	DNMT3A	2	25457242	C>C/T	snv	missense	44.31	NM_022552.4	c.2645G>A	p.Arg882His	deleterious (0.04)	probably damaging (0.956)
124	*de novo*	DNMT3A	2	25457242	C>C/T	snv	missense	38.19	NM_022552.4	c.2645G>A	p.Arg882His	deleterious (0.04)	probably damaging (0.956)
131	CS-AML	DNMT3A	2	25470585	A>A/T	snv	missense	49.18	NM_022552.4	c.889T>A	p.Trp297Arg	deleterious (0)	probably damaging (0.964)
131	CS-AML	DNMT3A	2	25466805	G>G/T	snv	missense	46.5	NM_022552.4	c.1898C>A	p.Pro633His	deleterious (0)	probably damaging (0.985)
132	sAML	DNMT3A	2	25467476	G>G/A	snv	nonsense	41.6	NM_022552	c.1600C>T	p.Gln534*		
137	*de novo*	DNMT3A	2	25463181	C>C/T	snv	missense	45.4	NM_022552.4	c.2312G>A	p.Arg771Gln	deleterious (0.04)	probably damaging (0.969)
163	*de novo*	DNMT3A	2	25457243	G>G/T	snv	missense	41.67	NM_022552.4	c.2644C>A	p.Arg882Ser	deleterious (0)	probably damaging (0.975)
170	*de novo*	DNMT3A	2	25457242	C>C/T	snv	missense	43.76	NM_022552	c.2645G>A	p.Arg882His	tolerated (0.998)	benign (0.043)
182	*de novo*	DNMT3A	2	25470484	C>C/T	snv	stop gained	43.44	NM_022552.4	c.990G>A	p.Trp330Ter		
185	*de novo*	DNMT3A	2	25467172	CCCA>CCCA/C	deletion	inframe deletion	36.66	NM_022552.4	c.1700_1702delTGG	p.Val567del		
213	*de novo*	DNMT3A	2	25457242	C>C/T	snv	missense	40.35	NM_022552.4	c.2645G>A	p.Arg882His	deleterious (0.04)	probably damaging (0.956)
241	*de novo*	DNMT3A	2	25458694	A>A/T	snv	missense	41.88	NM_022552.4	c.2479T>A	p.Phe827Ile	deleterious (0.02)	probably damaging (0.902)
242	CS-AML	DNMT3A	2	25457242	C>C/T	snv	missense	45.35	NM_022552.4	c.2645G>A	p.Arg882His	deleterious (0.04)	probably damaging (0.956)
247	*de novo*	DNMT3A	2	25463541	G>G/C	snv	missense	48.45	NM_022552.4	c.2141C>G	p.Ser714Cys	deleterious (0.02)	probably damaging (0.975)
272	*de novo*	DNMT3A	2	25457242	C>C/T	snv	missense	48.7	NM_022552.4	c.2645G>A	p.Arg882His	deleterious (0.04)	probably damaging (0.956)
275	*de novo*	DNMT3A	2	25463266	GCC>GCC/GC	deletion	frameshift	42.53	NM_022552	c.2226delG	p.Lys744Argfs*35		
277	*de novo*	DNMT3A	2	25457242	C>C/T	snv	missense	43.28	NM_022552.4	c.2645G>A	p.Arg882His	deleterious (0.04)	probably damaging (0.956)
319	*de novo*	DNMT3A	2	25457242	C>C/T	snv	missense	43.46	NM_022552.4	c.2645G>A	p.Arg882His	deleterious (0.04)	probably damaging (0.956)
320	*de novo*	DNMT3A	2	25457242	C>C/T	snv	missense	36.53	NM_022552.4	c.2645G>A	p.Arg882His	deleterious (0.04)	probably damaging (0.956)
325	*de novo*	DNMT3A	2	25457242	C>C/T	snv	missense	49.23	NM_022552.4	c.2645G>A	p.Arg882His	deleterious (0.04)	probably damaging (0.956)
342	*de novo*	DNMT3A	2	25457242	C>C/T	snv	missense	20.9	NM_022552	c.2645G>A	p.Arg882His	tolerated (0.998)	benign (0.043)
342	*de novo*	DNMT3A	2	25470968	C>C/T	snv	missense	19.7	NM_022552	c.793G>A	p.Val265Met	tolerated (0.99)	possibly damaging (0.593)
352	*de novo*	DNMT3A	2	25467449	C>C/A	snv	missense	10.7	NM_022552	c.1627G>T	p.Gly543Cys	tolerated (1)	probably damaging (0.999)
377	*de novo*	DNMT3A	2	25457243	G>G/A	snv	missense	17.3	NM_022552	c.2644C>T	p.Arg882Cys	tolerated (1)	probably damaging (0.936)
379	*de novo*	DNMT3A	2	25467495	CT>CT/C	deletion	frameshift	36.63	NM_022552.4	c.1580delA	p.Gln527ArgfsTer124		
406	*de novo*	DNMT3A	2	25457243	G>G/A	snv	missense	43.37	NM_022552.4	c.2644C>T	p.Arg882Cys	deleterious (0)	probably damaging (0.986)
407	*de novo*	DNMT3A	2	25457159	CA>CA/C	deletion	frameshift	47	NM_022552.4	c.2727delT	p.Phe909LeufsTer13		
421	*de novo*	DNMT3A	2	25457243	G>G/A	snv	missense	45.7	NM_022552	c.2644C>T	p.Arg882Cys	tolerated (1)	probably damaging (0.936)
447	sAML	DNMT3A	2	25463568	A>A/T	snv	missense	42.02	NM_022552	c.2114T>A	p.Ile705Asn	tolerated (1)	probably damaging (0.978)

UPN	AML category	Gene	Chr	Coordinate (Hg19)	Variant	Type	Consequence	VAF	Transcript	c.DNA	protein	Sift	PolyPhen
447	sAML	DNMT3A	2	25464428	T>T/C	snv	splicing	41.65	NM_022552	c.2082+3A>G	p.?		
477	*de novo*	DNMT3A	2	25457243	G>G/A	snv	missense	45.85	NM_022552	c.2644C>T	p.Arg882Cys	tolerated (1)	probably damaging (0.936)
507	*de novo*	DNMT3A	2	25457243	G>G/A	snv	missense	12.26	NM_022552.4	c.2644C>T	p.Arg882Cys	deleterious (0)	probably damaging (0.986)
527	*de novo*	DNMT3A	2	25469028	C>C/A	snv	splicing	48.18	NM_022552.4	c.1429+1G>T	p.?		
548	*de novo*	DNMT3A	2	25468915	A>A/G	snv	missense	43.18	NM_022552.4	c.1448T>C	p.Val483Ala	deleterious (0.02)	possibly damaging (0.703)
548	*de novo*	DNMT3A	2	25457243	G>G/A	snv	missense	36.13	NM_022552.4	c.2644C>T	p.Arg882Cys	deleterious (0)	probably damaging (0.986)
583	*de novo*	DNMT3A	2	25457242	C>C/T	snv	missense	44.96	NM_022552.4	c.2645G>A	p.Arg882His	deleterious (0.04)	probably damaging (0.956)
616	*de novo*	DNMT3A	2	25467023	C>C/T	snv	splicing	32.01	NM_022552	c.1851+1G>A	p.?		
666	*de novo*	DNMT3A	2	25457243	G>G/A	snv	missense	38.39	NM_022552.4	c.2644C>T	p.Arg882Cys	deleterious (0)	probably damaging (0.986)
683	*de novo*	DNMT3A	2	25461998	C>C/T	snv	splicing	48.04	NM_022552.4	c.2408+1G>A	p.?		
683	*de novo*	DNMT3A	2	25505534	G>G/A	snv	missense	17.57	NM_022552.4	c.224C>T	p.Ser75Phe	deleterious (0.05)	benign (0.012)
733	*de novo*	DNMT3A	2	25457243	G>G/A	snv	missense	45.01	NM_022552.4	c.2644C>T	p.Arg882Cys	deleterious (0)	probably damaging (0.986)
753	CS-AML	DNMT3A	2	25462020	C>C/T	snv	missense	55.14	NM_022552.4	c.2387G>A	p.Gly796Asp	deleterious (0)	probably damaging (0.998)
753	CS-AML	DNMT3A	2	25470498	G>G/A	snv	missense	43.7	NM_022552.4	c.976C>T	p.Arg326Cys	deleterious (0.03)	probably damaging (1)
761	*de novo*	DNMT3A	2	25470461	AC>AC/A	deletion	frameshift	37.05	NM_022552.4	c.1012delG	p.Val338TrpfsTer7		
766	*de novo*	DNMT3A	2	25466797	C>C/T	snv	missense	48.45	NM_022552.4	c.1906G>A	p.Val636Met	deleterious (0)	probably damaging (0.999)
790	*de novo*	DNMT3A	2	25457242	C>C/T	snv	missense	33.87	NM_022552.4	c.2645G>A	p.Arg882His	deleterious (0.04)	probably damaging (0.956)
803	*de novo*	DNMT3A	2	25457242	C>C/T	snv	missense	50.8	NM_022552.4	c.2645G>A	p.Arg882His	deleterious (0.04)	probably damaging (0.956)
824	*de novo*	DNMT3A	2	25457242	C>C/G	snv	missense	34.41	NM_022552	c.2645G>C	p.Arg882Pro	tolerated (0.969)	possibly damaging (0.738)
830	*de novo*	DNMT3A	2	25467433	A>A/G	snv	missense	44.87	NM_022552.4	c.1643T>C	p.Met548Thr	deleterious (0)	probably damaging (0.96)
847	*de novo*	DNMT3A	2	25457242	C>C/T	snv	missense	46.23	NM_022552.4	c.2645G>A	p.Arg882His	deleterious (0.04)	probably damaging (0.956)
852	*de novo*	DNMT3A	2	25458626	AG>AG/A	deletion	frameshift	46.48	NM_022552.4	c.2546delC	p.Pro849LeufsTer4		
865	*de novo*	DNMT3A	2	25464465	T>T/ TACATGATCTTCC CCTGGTGCCGCA CCATGCCCACCG TGATGGAGTCC	insertion	frameshift	37.0	NM_022552	c.2001_2047dup	p.Tyr683Trpfs*38		
867	*de novo*	DNMT3A	2	25468174	T>T/C	snv	missense	51.22	NM_022552.4	c.1502A>G	p.Asn501Ser	tolerated (0.1)	benign (0.037)
931	*de novo*	DNMT3A	2	25457242	C>C/T	snv	missense	42.63	NM_022552.4	c.2645G>A	p.Arg882His	deleterious (0.04)	probably damaging (0.956)
981	*de novo*	DNMT3A	2	25457242	C>C/T	snv	missense	47.74	NM_022552.4	c.2645G>A	p.Arg882His	deleterious (0.04)	probably damaging (0.956)
997	CS-AML	DNMT3A	2	25463308	G>G/A	snv	missense	26.13	NM_022552.4	c.2185C>T	p.Arg729Trp	deleterious (0)	probably damaging (1)
1042	*de novo*	DNMT3A	2	25457242	C>C/T	snv	missense	45.7	NM_022552	c.2645G>A	p.Arg882His	tolerated (0.998)	possibly damaging (0.043)

UPN	AML category	Gene	Chr	Coordinate (Hg19)	Variant	Type	Consequence	VAF	Transcript	c.DNA	protein	Sift	PolyPhen
1095	*de novo*	DNMT3A	2	25467478	T>T/C	snv	missense	43.5	NM_022552	c.1598A>G	p.Tyr533Cys	tolerated (0.983)	probably damaging (0.993)
1100	*de novo*	DNMT3A	2	25467209	T>T/G	snv	splicing	44.98	NM_022552.4	c.1668-2A>C	p.?		
1110	sAML	DNMT3A	2	25467428	C>C/T	snv	missense	58.7	NM_022552.4	c.1648G>A	p.Gly550Arg	deleterious (0.01)	probably damaging (0.893)
1110	sAML	DNMT3A	2	25463181	C>C/G	snv	missense	22.51	NM_022552.4	c.2312G>C	p.Arg771Pro	deleterious (0)	probably damaging (0.996)
1140	*de novo*	DNMT3A	2	25457242	C>C/T	snv	missense	45.92	NM_022552.4	c.2645G>A	p.Arg882His	deleterious (0.04)	probably damaging (0.956)
1147	*de novo*	DNMT3A	2	25457243	G>G/A	snv	missense	44.77	NM_022552	c.2644C>T	p.Arg882Cys	tolerated (1)	probably damaging (0.936)
1166	CS-AML	DNMT3A	2	25457242	C>C/T	snv	missense	24.63	NM_022552.4	c.2645G>A	p.Arg88His	deleterious (0.04)	probably damaging (0.956)
1168	*de novo*	DNMT3A	2	25457242	C>C/T	snv	missense	46.89	NM_022552.4	c.2645G>A	p.Arg882His	deleterious (0.04)	probably damaging (0.956)
1191	*de novo*	DNMT3A	2	25466797	C>C/A	snv	missense	42.16	NM_022552.4	c.1906G>T	p.Val636Leu	deleterious (0)	probably damaging (0.974)
1199	*de novo*	DNMT3A	2	25457242	C>C/T	snv	missense	44.12	NM_022552.4	c.2645G>A	p.Arg882His	deleterious (0.04)	probably damaging (0.956)
1222	*de novo*	DNMT3A	2	25457242	C>C/T	snv	missense	30	NM_022552	c.2645G>A	p.Arg882His	tolerated (0.998)	benign (0.043)
1270	CS-AML	DNMT3A	2	25463181	C>C/T	snv	missense	35.8	NM_022552.4	c.2312G>A	p.Arg771Gln	deleterious (0.04)	probably damaging (0.969)
1285	*de novo*	DNMT3A	2	25457243	G>G/A	snv	missense	24.18	NM_022552.4	c.2644C>T	p.Arg882Cys	deleterious (0)	probably damaging (0.986)
1290	*de novo*	DNMT3A	2	25457242	C>C/T	snv	missense	37.14	NM_022552	c.2645G>A	p.Arg882His	tolerated (0.998)	benign (0.043)
1314	*de novo*	DNMT3A	2	25457242	C>C/T	snv	missense	41.19	NM_022552.4	c.2645G>A	p.Arg882His	deleterious (0.04)	probably damaging (0.956)
1335	CS-AML	DNMT3A	2	25457231	G>G/C	snv	missense	40.92	NM_022552.4	c.2656C>G	p.Gln886Glu	deleterious (0)	possibly damaging (0.871)
1338	*de novo*	DNMT3A	2	25457243	G>G/A	snv	missense	45.05	NM_022552.4	c.2644C>T	p.Arg882Cys	deleterious (0)	probably damaging (0.986)
1351	*de novo*	DNMT3A	2	25468135	C>C/T	snv	missense	48.96	NM_022552.4	c.1541G>A	p.Cys514Tyr	deleterious (0)	probably damaging (0.992)
1364	*de novo*	DNMT3A	2	25470971	G>G/GCTCA	insertion	frameshift	44.59	NM_022552.4	c.786_789dupTGAG	p.Pro264Ter		
1370	*de novo*	DNMT3A	2	25457242	C>C/T	snv	missense	40.94	NM_022552	c.2645G>A	p.Arg882His	tolerated (0.998)	benign (0.043)
1376	*de novo*	DNMT3A	2	25463541	G>G/C	snv	missense	44.06	NM_022552.4	c.2141C>G	p.Ser714Cys	deleterious (0.02)	probably damaging (0.975)
168	*de novo*	ETV6	12	11992218	G>G/GCCC	insertion	inframe insertion	13.71	NM_001987.4	c.308_309insCCC	p.Arg103_Tyr104insPro		
1308	CS-AML	FBXW7	4	153249384	C>C/T	snv	missense	38.73	NM_033632.3	c.1394G>A	p.Arg465His	deleterious (0)	probably damaging (1)
14	*de novo*	FLT3	13	28608263	T>T/TCATATTCTCTGAAATCAACGTAGC	insertion	inframe insertion	30.51	NM_004119.2	c.1792_1793insGCTACGTTGATTTCAGAGAATATG	p.Glu598_Tyr598insGlyTyrValAspPheArgGluTyr		
20	*de novo*	FLT3	13	28592641	T>T/G	snv	missense	36.61	NM_004119.2	c.2504A>C	p.Asp835Ala	deleterious (0)	probably damaging (0.999)
32	*de novo*	FLT3	13	28592623	T>T/G	snv	missense	13.69	NM_004119	c.2522A>C	p.Asn841Thr	tolerated (0.998)	possibly damaging (0.176)
43	*de novo*	FLT3	13	28592623	T>T/A	snv	missense	40.73	NM_004119.2	c.2522A>T	p.Asn841Ile	deleterious (0)	probably damaging (0.99)

UPN	AML category	Gene	Chr	Coordinate (Hg19)	Variant	Type	Consequence	VAF	Transcript	c.DNA	protein	Sift	PolyPhen
62	CS-AML	FLT3	13	28608257	T>T/TCATATTCATATTCTCTGAAATCAACGTAGAAGTACTCATTATCTGAGC	insertion	inframe	43	NM_004119	c.1798_1799ins48	p.Tyr599_Asp600ins16		
149	CS-AML	FLT3	13	28592640	A>A/C	snv	missense	43.32	NM_004119	c.2505T>G	p.Asp835Glu	tolerated (0.998)	probably damaging (0.951)
241	*de novo*	FLT3	13	28592642	C>C/A	snv	missense	39.8	NM_004119.2	c.2503G>T	p.Asp835Tyr	deleterious (0)	probably damaging (1)
247	*de novo*	FLT3	13	28592642	C>C/A	snv	missense	37.4	NM_004119.2	c.2503G>T	p.Asp835Tyr	deleterious (0)	probably damaging (1)
277	*de novo*	FLT3	13	28608262	T>T/TTCATATTCTCTGAAA	insertion	inframe insertion	35.81	NM_004119.2	c.1779_1793dupTTTCAGAGAATATGA	p.Asp593_Tyr597dup		
325	*de novo*	FLT3	13	28592642	C>C/A	snv	missense	29.3	NM_004119.2	c.2503G>T	p.Asp835Tyr	deleterious (0)	probably damaging (1)
325	*de novo*	FLT3	13	28608268	T>T/TTCTCTGAAATCAACGTAG	insertion	inframe insertion	7.24	NM_004119.2	c.1770_1787dupCTACGTTGATTTCAGAGA	p.Glu596_Tyr596insAspTyrValAspPheArg		
342	*de novo*	FLT3	13	28608272	C>C/CTGAAATCAACGTAGAAGTACTCATTATCTGAGGAGCCGGTCT	insertion	inframe	15.2	NM_004119	c.1783_1784ins42	p.Phe594_Arg595ins14		
342	*de novo*	FLT3	13	28608262	T>T/TTCATATTCTCTGAAATCAACGTAGAAGTCC	insertion	inframe	5.8	NM_004119	c.1793_1794ins30	p.Glu598_Tyr599ins10		
378	CS-AML	FLT3	13	28608320	A>A/C	snv	missense	23.94	NM_004119.2	c.1736T>G	p.Val579Gly	deleterious (0)	possibly damaging (0.712)
405	*de novo*	FLT3	13	28608262	T>T/TTCATATTCTCTGAAATCAACG	insertion	inframe insertion	28.25	NM_004119.2	c.1773_1793dupCGTTGATTTCAGAGAATATGA	p.Glu598_Tyr598insAspValAspPheArgGluTyr		
421	*de novo*	FLT3	13	28608275	A>A/AAATCAACGTAGAAGTACTCATTATCCG	insertion	inframe	34.9	NM_004119	c.1780_1781ins27	p.Ser585_Asp593dup		
527	*de novo*	FLT3	13	28592642	C>C/A	snv	missense	45.98	NM_004119.2	c.2503G>T	p.Asp835Tyr	deleterious (0)	probably damaging (1)
548	*de novo*	FLT3	13	28592642	C>C/G	snv	missense	24.89	NM_004119.2	c.2503G>C	p.Asp835His	deleterious (0)	probably damaging (0.999)
552	*de novo*	FLT3	13	28608221	A>A/AACTCTAAATTTTCTCTTGGAAACTCCCATTTGAGATCATATTCATATTCTCTGAAATCAACGTAGAAGT	insertion	inframe	41	NM_004119	c.1766_1834dup	p.Tyr589_Glu611dup		
582	*de novo*	FLT3	13	28592642	C>C/A	snv	missense	42.22	NM_004119.2	c.2503G>T	p.Asp835Tyr	deleterious (0)	probably damaging (1)
588	*de novo*	FLT3	13	28608272	C>C/CTGAAATCAACGTAGAAGTACTCATTATCTT	insertion	inframe	9	NM_004119	c.1783_1784ins30	p.Phe594_Arg595ins10		
616	*de novo*	FLT3	13	28608262	T>T/TTCATATTCTCTGAAATCAACGTAGAAGTACTCATTATCTGAGGAGCCGGTC	insertion	inframe	14	NM_004119	c.1743_1793dup	p.Thr582_Glu598dup		
683	*de novo*	FLT3	13	28608278	T>T/G	snv	missense	15.64	NM_004119.2	c.1778A>C	p.Asp593Ala	deleterious (0.01)	possibly damaging (0.698)
733	*de novo*	FLT3	13	28608254	A>A/AGATCATATTCATATTCTAGGG	insertion	inframe insertion	80.28	NM_004119.2	c.1801_1802insCCCTAGAATATGAATATGATC	p.Leu601_Lys601insProLeuGluTyrGluTyrAsp		

UPN	AML category	Gene	Chr	Coordinate (Hg19)	Variant	Type	Consequence	VAF	Transcript	c.DNA	protein	Sift	PolyPhen
865	*de novo*	FLT3	13	28608249	A>A/ ATTTGAGATCATA TTCATATTCTCTG AAATCAACGTAG AAGTACTCATTAT CTGAGGAGCC	insertion	inframe	47.0	NM_004119	c.1747_1806dup	p.Gly583_Lys602dup		
877	*de novo*	FLT3	13	28592628	A>A/T	snv	missense	40.53	NM_004119.2	c.2517T>A	p.Asp839Glu	deleterious (0)	probably damaging (0.996)
931	*de novo*	FLT3	13	28592642	C>C/A	snv	missense	19.65	NM_004119.2	c.2503G>T	p.Asp835Tyr	deleterious (0)	probably damaging (1)
940	CS-AML	FLT3	13	28592642	C>C/A	snv	missense	23.4	NM_004119	c.2503G>T	p.Asp835Tyr	tolerated (0.999)	probably damaging (0.999)
940	CS-AML	FLT3	13	28608256	A>A/ ATCATATTCATATT CTCTGAAATCAAT GAGT	insertion	inframe	10.3	NM_004119	c.1799_1800ins30	p.Tyr599_Asp600ins 10		
1025	*de novo*	FLT3	13	28592642	C>C/A	snv	missense	37.75	NM_004119.2	c.2503G>T	p.Asp835Tyr	deleterious (0)	probably damaging (1)
1042	*de novo*	FLT3	13	28608218	C>C/ CCAAACTCTAAAT TTTCTCTTGGAA ACTCCCATTTGA GATCATATT	insertion	coding	64.7	NM_004119	c.1793_1837dup	p.Glu598_Phe612du p		
1095	*de novo*	FLT3	13	28608262	T>T/ TTCATATTCTCTG AAATCAACGTAG	insertion	inframe	27	NM_004119	c.1770_1793dup	p.Tyr597_Glu598ins 8		
1100	*de novo*	FLT3	13	28592642	C>C/A	snv	missense	29.27	NM_004119.2	c.2503G>T	p.Asp835Tyr	deleterious (0)	probably damaging (1)
1168	*de novo*	FLT3	13	28592620	T>T/C	snv	missense	39.62	NM_004119.2	c.2525A>G	p.Tyr842Cys	deleterious (0)	probably damaging (1)
1222	*de novo*	FLT3	13	28608219	C>C/ CAAACTCTAAATT TTCTCTTGGAAA CTCCCATTTGAG	insertion	inframe	15	NM_004119	c.1801_1836dup	p.Leu601_Phe612du p		
1236	*de novo*	FLT3	13	28592642	C>C/A	snv	missense	38.28	NM_004119.2	c.2503G>T	p.Asp835Tyr	deleterious (0)	probably damaging (1)
1290	*de novo*	FLT3	13	28608218	C>C/ CCAAACTCTAAAT TTTCTCTTGGAA ACTCCCATTTGA GATCATATTCATA TTCTCTGAAATC GG	insertion	splicing	17.0	NM_004119	c.1837_1837+1ins63	p.?		
1379	*de novo*	FLT3	13	28608246	C>C/ CCCATTTGAGAT CATA	insertion	inframe insertion	41.11	NM_004119.2	c.1795_1809dupTAT GATCTCAAATGG	p.Tyr599_Trp603dup		
82	sAML	GATA1	X	48649520	G>G/T	snv	stop gained	15	NM_002049.3	c.4G>T	p.Glu2Ter		
126	CS-AML	GATA1	X	48649533	T>T/TG	insertion	frameshift	61.96	NM_002049.3	c.17_18insG	p.Ser8ValfsTer32		
28	*de novo*	GATA2	3	128202731	C>C/T	snv	missense	27.16	NM_032638.4	c.989G>A	p.Arg330Gln	deleterious (0)	probably damaging (0.902)
96	*de novo*	GATA2	3	128200720	C>C/T	snv	missense	42.24	NM_032638.4	c.1085G>A	p.Arg362Gln	deleterious (0)	probably damaging (0.973)
158	*de novo*	GATA2	3	128200744	G>G/C	snv	missense	13.89	NM_032638.4	c.1061C>G	p.Thr354Arg	deleterious (0)	probably damaging (0.996)
269	*de novo*	GATA2	3	128202794	T>T/A	snv	missense	48.93	NM_032638.4	c.926A>T	p.Asp309Val	deleterious (0)	probably damaging (0.998)
274	*de novo*	GATA2	3	128202767	G>G/A	snv	missense	45.49	NM_032638.4	c.953C>T	p.Ala318Val	deleterious (0)	probably damaging (0.991)
297	*de novo*	GATA2	3	128202810	G>G/C	snv	missense	51.02	NM_032638.4	c.910C>G	p.Pro304Ala	deleterious (0)	probably damaging (0.991)
402	*de novo*	GATA2	3	128202731	C>C/A	snv	missense	24.82	NM_032638.4	c.989G>T	p.Arg330Leu	deleterious (0)	probably damaging (0.942)

UPN	AML category	Gene	Chr	Coordinate (Hg19)	Variant	Type	Consequence	VAF	Transcript	c.DNA	protein	Sift	PolyPhen
635	*de novo*	GATA2	3	128202759	G>G/A	snv	missense	42.8	NM_032638.4	c.961C>T	p.Leu321Phe	deleterious (0)	probably damaging (0.99)
703	*de novo*	GATA2	3	128204906	T>T/A	snv	stop gained	11.8	NM_032638.4	c.535A>T	p.Lys179Ter		
1352	*de novo*	GATA2	3	128202759	G>G/A	snv	missense	13.93	NM_032638.4	c.961C>T	p.Leu321Phe	deleterious (0)	probably damaging (0.99)
548	*de novo*	GNAS	20	57484414	C>C/T	snv	missense	12.61	NM_080425.2	c.2524C>T	p.Arg842Cys	deleterious (0)	probably damaging (1)
633	CS-AML	HRAS	11	533878	C>C/T	snv	missense	14.34	NM_005343.2	c.178G>A	p.Gly60Ser	deleterious (0)	probably damaging (0.998)
32	*de novo*	IDH1	2	209113112	C>C/T	snv	missense	38.46	NM_005896	c.395G>A	p.Arg132His	tolerated (0.986)	benign (0.06)
43	*de novo*	IDH1	2	209113113	G>G/C	snv	missense	47.73	NM_005896.2	c.394C>G	p.Arg132Gly	deleterious (0)	probably damaging (0.997)
64	*de novo*	IDH1	2	209113113	G>G/A	snv	missense	49.06	NM_005896.2	c.394C>T	p.Arg132Cys	deleterious (0.02)	possibly damaging (0.791)
92	*de novo*	IDH1	2	209113113	G>G/C	snv	missense	21.81	NM_005896	c.394C>G	p.Arg132Gly	tolerated (0.999)	probably damaging (0.907)
131	CS-AML	IDH1	2	209113113	G>G/A	snv	missense	55.52	NM_005896.2	c.394C>T	p.Arg132Cys	deleterious (0.02)	possibly damaging (0.791)
137	*de novo*	IDH1	2	209113113	G>G/A	snv	missense	48.39	NM_005896.2	c.394C>T	p.Arg132Cys	deleterious (0.02)	possibly damaging (0.791)
163	*de novo*	IDH1	2	209113112	C>C/T	snv	missense	42.46	NM_005896.2	c.395G>A	p.Arg132His	deleterious (0)	possibly damaging (0.629)
185	*de novo*	IDH1	2	209113112	C>C/T	snv	missense	30.29	NM_005896.2	c.395G>A	p.Arg132His	deleterious (0)	possibly damaging (0.629)
192	*de novo*	IDH1	2	209113113	G>G/T	snv	missense	49.33	NM_005896.2	c.394C>A	p.Arg132Ser	deleterious (0)	probably damaging (0.996)
312	*de novo*	IDH1	2	209113112	C>C/T	snv	missense	42.98	NM_005896.2	c.395G>A	p.Arg132His	deleterious (0)	possibly damaging (0.629)
319	*de novo*	IDH1	2	209113112	C>C/T	snv	missense	48.14	NM_005896.2	c.395G>A	p.Arg132His	deleterious (0)	possibly damaging (0.629)
407	*de novo*	IDH1	2	209113112	C>C/T	snv	missense	46.14	NM_005896.2	c.395G>A	p.Arg132His	deleterious (0)	possibly damaging (0.629)
432	*de novo*	IDH1	2	209113113	G>G/T	snv	missense	35.74	NM_005896	c.394C>A	p.Arg132Ser	tolerated (1)	probably damaging (0.878)
511	*de novo*	IDH1	2	209113112	C>C/T	snv	missense	47.21	NM_005896.2	c.395G>A	p.Arg132His	deleterious (0)	possibly damaging (0.629)
588	*de novo*	IDH1	2	209113112	C>C/T	snv	missense	35.27	NM_005896	c.395G>A	p.Arg132His	tolerated (0.986)	benign (0.06)
684	*de novo*	IDH1	2	209113112	C>C/T	snv	missense	21.11	NM_005896.2	c.395G>A	p.Arg132His	deleterious (0)	possibly damaging (0.629)
703	*de novo*	IDH1	2	209113113	G>G/T	snv	missense	43.46	NM_005896.2	c.394C>A	p.Arg132Ser	deleterious (0)	probably damaging (0.996)
867	*de novo*	IDH1	2	209113112	C>C/T	snv	missense	11.83	NM_005896.2	c.395G>A	p.Arg132His	deleterious (0)	possibly damaging (0.629)
3	CS-AML	IDH2	15	90631838	C>C/T	snv	missense	41.6	NM_002168.2	c.515G>A	p.Arg172Lys	deleterious (0)	probably damaging (1)
101	*de novo*	IDH2	15	90631934	C>C/T	snv	missense	50	NM_002168.2	c.419G>A	p.Arg140Gln	deleterious (0)	probably damaging (1)
126	CS-AML	IDH2	15	90631934	C>C/T	snv	missense	44.77	NM_002168.2	c.419G>A	p.Arg140Gln	deleterious (0)	probably damaging (1)
258	CS-AML	IDH2	15	90631934	C>C/T	snv	missense	20.98	NM_002168.2	c.419G>A	p.Arg140Gln	deleterious (0)	probably damaging (1)

UPN	AML category	Gene	Chr	Coordinate (Hg19)	Variant	Type	Consequence	VAF	Transcript	c.DNA	protein	Sift	PolyPhen
352	*de novo*	IDH2	15	90631838	C>C/T	snv	missense	13.8	NM_002168	c.515G>A	p.Arg172Lys	tolerated (0.999)	probably damaging (0.999)
380	CS-AML	IDH2	15	90631934	C>C/T	snv	missense	22.76	NM_002168.2	c.419G>A	p.Arg140Gln	deleterious (0)	probably damaging (1)
406	*de novo*	IDH2	15	90631934	C>C/T	snv	missense	44.78	NM_002168.2	c.419G>A	p.Arg140Gln	deleterious (0)	probably damaging (1)
507	*de novo*	IDH2	15	90631934	C>C/T	snv	missense	42.83	NM_002168.2	c.419G>A	p.Arg140Gln	deleterious (0)	probably damaging (1)
527	*de novo*	IDH2	15	90631934	C>C/T	snv	missense	47.56	NM_002168.2	c.419G>A	p.Arg140Gln	deleterious (0)	probably damaging (1)
616	*de novo*	IDH2	15	90631934	C>C/T	snv	missense	31.93	NM_002168	c.419G>A	p.Arg140Gln	tolerated (1)	probably damaging (0.998)
753	CS-AML	IDH2	15	90631838	C>C/T	snv	missense	45.11	NM_002168.2	c.515G>A	p.Arg172Lys	deleterious (0)	probably damaging (1)
847	*de novo*	IDH2	15	90631934	C>C/T	snv	missense	18.59	NM_002168.2	c.419G>A	p.Arg140Gln	deleterious (0)	probably damaging (1)
853	*de novo*	IDH2	15	90631934	C>C/T	snv	missense	46.99	NM_002168.2	c.419G>A	p.Arg140Gln	deleterious (0)	probably damaging (1)
858	*de novo*	IDH2	15	90631839	T>T/A	snv	missense	39.22	NM_002168.2	c.514A>T	p.Arg172Trp	deleterious (0)	probably damaging (1)
867	*de novo*	IDH2	15	90631934	C>C/T	snv	missense	23.71	NM_002168.2	c.419G>A	p.Arg140Gln	deleterious (0)	probably damaging (1)
919	*de novo*	IDH2	15	90631934	C>C/T	snv	missense	47.53	NM_002168.2	c.419G>A	p.Arg140Gln	deleterious (0)	probably damaging (1)
997	CS-AML	IDH2	15	90631838	C>C/T	snv	missense	31.43	NM_002168.2	c.515G>A	p.Arg172Lys	deleterious (0)	probably damaging (1)
1042	*de novo*	IDH2	15	90631934	C>C/T	snv	missense	49.7	NM_002168	c.419G>A	p.Arg140Gln	tolerated (1)	probably damaging (0.998)
1110	sAML	IDH2	15	90631934	C>C/T	snv	missense	28.99	NM_002168.2	c.419G>A	p.Arg140Gln	deleterious (0)	probably damaging (1)
1191	*de novo*	IDH2	15	90631838	C>C/T	snv	missense	43.26	NM_002168.2	c.515G>A	p.Arg172Lys	deleterious (0)	probably damaging (1)
1308	CS-AML	IDH2	15	90631838	C>C/T	snv	missense	46.6	NM_002168.2	c.515G>A	p.Arg172Lys	deleterious (0)	probably damaging (1)
1335	CS-AML	IDH2	15	90631838	C>C/T	snv	missense	43.8	NM_002168.2	c.515G>A	p.Arg172Lys	deleterious (0)	probably damaging (1)
1337	CS-AML	IDH2	15	90631934	C>C/T	snv	missense	40.35	NM_002168.2	c.419G>A	p.Arg140Gln	deleterious (0)	probably damaging (1)
1373	*de novo*	IDH2	15	90631934	C>C/T	snv	missense	25	NM_002168.2	c.419G>A	p.Arg140Gln	deleterious (0)	probably damaging (1)
132	sAML	JAK2	9	5073770	G>G/T	snv	missense	14.2	NM_004972	c.1849G>T	p.Val617Phe	tolerated (0.998)	probably damaging (0.934)
222	sAML	JAK2	9	5073770	G>G/T	snv	missense	46.69	NM_004972.3	c.1849G>T	p.Val617Phe	deleterious (0.01)	probably damaging (0.967)
377	*de novo*	JAK2	9	5090452	G>G/A	snv	missense	51.1	NM_004972	c.2768G>A	p.Arg923His	tolerated (0.962)	possibly damaging (0.689)
623	CS-AML	JAK2	9	5055741	A>A/T	snv	missense	49.1	NM_004972	c.1009A>T	p.Asn337Tyr	tolerated (0.981)	benign (0.081)
1148	CS-AML	JAK3	19	17947985	G>G/T	snv	stop gained	27.38	NM_000215.3	c.1739C>A	p.Ser580Ter		
582	*de novo*	KDM6A	X	44879925	C>C/T	snv	stop gained	42.61	NM_021140.2	c.514C>T	p.Arg172Ter		
852	*de novo*	KDM6A	X	44919270	C>C/T	snv	stop gained	15.77	NM_021140.2	c.1198C>T	p.Gln400Ter		
1148	CS-AML	KDM6A	X	44910996	C>C/T	snv	stop gained	15.62	NM_021140.2	c.697C>T	p.Gln233Ter		

UPN	AML category	Gene	Chr	Coordinate (Hg19)	Variant	Type	Consequence	VAF	Transcript	c.DNA	protein	Sift	PolyPhen
877	*de novo*	KMT2A	11	118352549	C>C/A	snv	missense	50.32	NM_001197104.1	c.3754C>A	p.Pro1252Thr	deleterious (0.01)	benign (0.008)
32	*de novo*	KRAS	12	25398284	C>C/G	snv	missense	14.81	NM_004985	c.35G>C	p.Gly12Ala	tolerated (0.997)	possibly damaging (0.773)
91	*de novo*	KRAS	12	25380256	T>T/A	snv	missense	24.12	NM_033360.2	c.202A>T	p.Arg68Trp	deleterious (0)	probably damaging (1)
163	*de novo*	KRAS	12	25398284	C>C/G	snv	missense	25.24	NM_033360.2	c.35G>C	p.Gly12Ala	deleterious (0.02)	possibly damaging (0.739)
220	*de novo*	KRAS	12	25398281	C>C/T	snv	missense	17.32	NM_033360.2	c.38G>A	p.Gly13Asp	deleterious (0.03)	possibly damaging (0.517)
274	*de novo*	KRAS	12	25398284	C>C/A	snv	missense	30.78	NM_033360.2	c.35G>T	p.Gly12Val	deleterious (0)	probably damaging (0.984)
275	*de novo*	KRAS	12	25398281	C>C/T	snv	missense	31.86	NM_004985	c.38G>A	p.Gly13Asp	tolerated (0.988)	possibly damaging (0.506)
387	CS-AML	KRAS	12	25398284	C>C/T	snv	missense	43.84	NM_033360.2	c.35G>A	p.Gly12Asp	deleterious (0)	possibly damaging (0.387)
766	*de novo*	KRAS	12	25380238	T>T/G	snv	missense	14.02	NM_033360.2	c.220A>C	p.Thr74Pro	deleterious (0.02)	probably damaging (0.996)
824	*de novo*	KRAS	12	25398284	C>C/G	snv	missense	22.82	NM_004985	c.35G>C	p.Gly12Ala	tolerated (0.997)	possibly damaging (0.773)
1147	*de novo*	KRAS	12	25398278	A>A/ACGC	insertion	inframe	29.37	NM_004985	c.38_40dupGCG	p.Gly13dup		
1343	sAML	KRAS	12	25398284	C>C/T	snv	missense	29.71	NM_033360.2	c.35G>A	p.Gly12Asp	deleterious (0)	possibly damaging (0.387)
852	*de novo*	NOTCH1	9	139390622	CG>CG/C	deletion	frameshift	15.26	NM_017617.3	c.7568delC	p.Ser2523CysfsTer66		
10	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	28.11	NM_002520.6	c.859_860insTCTG	p.Trp288CysfsTer12		
14	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	43.29	NM_002520.6	c.859_860insTCTG	p.Trp288CysfsTer12		
20	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	36.45	NM_002520.6	c.859_860insTCTG	p.Trp288CysfsTer12		
32	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	14.52	NM_002520	c.860_863dupTCTG	p.Trp288CysfsTer12		
43	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	39.36	NM_002520.6	c.859_860insTCTG	p.Trp288CysfsTer12		
64	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	40.48	NM_002520.6	c.859_860insTCTG	p.Trp288CysfsTer12		
91	*de novo*	NPM1	5	170837544	T>T/TCTGC	insertion	frameshift	41.16	NM_002520.6	c.860_861insCTGC	p.Trp288CysfsTer12		
92	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	15.3	NM_002520	c.860_863dupTCTG	p.Trp288CysfsTer12		
108	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	36.95	NM_002520.6	c.859_860insTCTG	p.Trp288CysfsTer12		
110	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	39.21	NM_002520.6	c.859_860insTCTG	p.Trp288CysfsTer12		
124	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	42.7	NM_002520.6	c.859_860insTCTG	p.Trp288CysfsTer12		
137	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	39.52	NM_002520.6	c.859_860insTCTG	p.Trp288CysfsTer12		
158	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	41.25	NM_002520.6	c.859_860insTCTG	p.Trp288CysfsTer12		
163	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	38.67	NM_002520.6	c.859_860insTCTG	p.Trp288CysfsTer12		
168	*de novo*	NPM1	5	170837545	C>C/CTGCA	insertion	frameshift	18.36	NM_002520.6	c.861_862insTGCA	p.Trp288CysfsTer12		
170	*de novo*	NPM1	5	170837544	T>T/TCTGC	insertion	frameshift	24.04	NM_002520	c.863_864insCCTG	p.Trp288CysfsTer12		
182	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	38.67	NM_002520.6	c.859_860insTCTG	p.Trp288CysfsTer12		
185	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	37.13	NM_002520.6	c.859_860insTCTG	p.Trp288CysfsTer12		
192	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	34.29	NM_002520.6	c.859_860insTCTG	p.Trp288CysfsTer12		
213	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	39.68	NM_002520.6	c.859_860insTCTG	p.Trp288CysfsTer12		
220	*de novo*	NPM1	5	170837545	C>C/CTGCA	insertion	frameshift	39.86	NM_002520.6	c.861_862insTGCA	p.Trp288CysfsTer12		
241	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	41.66	NM_002520.6	c.859_860insTCTG	p.Trp288CysfsTer12		
246	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	36.27	NM_002520.6	c.859_860insTCTG	p.Trp288CysfsTer12		
247	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	42.01	NM_002520.6	c.859_860insTCTG	p.Trp288CysfsTer12		

UPN	AML category	Gene	Chr	Coordinate (Hg19)	Variant	Type	Consequence	VAF	Transcript	c.DNA	protein	Sift	PolyPhen
261	*de novo*	NPM1	5	170837545	C>C/CTGCA	insertion	frameshift	34.74	NM_002520.6	c.861_862insTGCA	p.Trp288CysfsTer12		
272	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	47.94	NM_002520.6	c.859_860insTCTG	p.Trp288CysfsTer12		
275	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	19.2	NM_002520	c.860_863dupTCTG	p.Trp288CysfsTer12		
277	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	42.07	NM_002520.6	c.859_860insTCTG	p.Trp288CysfsTer12		
312	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	44.04	NM_002520.6	c.859_860insTCTG	p.Trp288CysfsTer12		
319	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	41.8	NM_002520.6	c.859_860insTCTG	p.Trp288CysfsTer12		
320	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	38.21	NM_002520.6	c.859_860insTCTG	p.Trp288CysfsTer12		
323	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	36.46	NM_002520.6	c.859_860insTCTG	p.Trp288CysfsTer12		
325	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	45.32	NM_002520.6	c.859_860insTCTG	p.Trp288CysfsTer12		
342	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	15.7	NM_002520	c.860_863dupTCTG	p.Trp288CysfsTer12		
352	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	7.8	NM_002520	c.860_863dupTCTG	p.Trp288CysfsTer12		
377	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	16.4	NM_002520	c.860_863dupTCTG	p.Trp288CysfsTer12		
379	*de novo*	NPM1	5	170837544	T>T/TCTGC	insertion	frameshift	33.58	NM_002520.6	c.860_861insCTGC	p.Trp288CysfsTer12		
405	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	34.31	NM_002520.6	c.859_860insTCTG	p.Trp288CysfsTer12		
407	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	42.89	NM_002520.6	c.859_860insTCTG	p.Trp288CysfsTer12		
421	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	38.5	NM_002520	c.860_863dupTCTG	p.Trp288CysfsTer12		
423	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	38.43	NM_002520.6	c.859_860insTCTG	p.Trp288CysfsTer12		
432	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	30.11	NM_002520	c.860_863dupTCTG	p.Trp288CysfsTer12		
441	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	27.05	NM_002520	c.860_863dupTCTG	p.Trp288CysfsTer12		
455	*de novo*	NPM1	5	170837557	G>G/GGCTC	insertion	frameshift	15.2	NM_002520	c.873_874insGCTC	Trp290Fs*10		
477	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	33.85	NM_002520	c.860_863dupTCTG	p.Trp288CysfsTer12		
507	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	23.03	NM_002520.6	c.859_860insTCTG	p.Trp288CysfsTer12		
511	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	25.38	NM_002520.6	c.859_860insTCTG	p.Trp288CysfsTer12		
527	*de novo*	NPM1	5	170837545	C>C/CTGCA	insertion	frameshift	42.35	NM_002520.6	c.861_862insTGCA	p.Trp288CysfsTer12		
531	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	45.27	NM_002520.6	c.859_860insTCTG	p.Trp288CysfsTer12		
548	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	39.7	NM_002520.6	c.859_860insTCTG	p.Trp288CysfsTer12		
552	*de novo*	NPM1	5	170837545	C>C/CTGCA	insertion	frameshift	40.56	NM_002520	c.863_864insCATG	p.Trp288CysfsTer12		
582	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	40.64	NM_002520.6	c.859_860insTCTG	p.Trp288CysfsTer12		
583	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	35.47	NM_002520.6	c.859_860insTCTG	p.Trp288CysfsTer12		
588	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	17.82	NM_002520	c.860_863dupTCTG	p.Trp288CysfsTer12		
616	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	14.61	NM_002520	c.860_863dupTCTG	p.Trp288CysfsTer12		
659	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	43.21	NM_002520.6	c.859_860insTCTG	p.Trp288CysfsTer12		
666	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	34.68	NM_002520.6	c.859_860insTCTG	p.Trp288CysfsTer12		
667	*de novo*	NPM1	5	170837545	C>C/CTGCA	insertion	frameshift	41.58	NM_002520.6	c.861_862insTGCA	p.Trp288CysfsTer12		
683	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	41.19	NM_002520.6	c.859_860insTCTG	p.Trp288CysfsTer12		
684	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	38	NM_002520.6	c.859_860insTCTG	p.Trp288CysfsTer12		
693	*de novo*	NPM1	5	170837545	C>C/CTGCA	insertion	frameshift	46.14	NM_002520.6	c.861_862insTGCA	p.Trp288CysfsTer12		
703	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	44.45	NM_002520.6	c.859_860insTCTG	p.Trp288CysfsTer12		
716	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	43.04	NM_002520.6	c.859_860insTCTG	p.Trp288CysfsTer12		
733	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	43.7	NM_002520.6	c.859_860insTCTG	p.Trp288CysfsTer12		
761	*de novo*	NPM1	5	170837545	C>C/CTGCA	insertion	frameshift	40.35	NM_002520.6	c.861_862insTGCA	p.Trp288CysfsTer12		
766	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	35.76	NM_002520.6	c.859_860insTCTG	p.Trp288CysfsTer12		
790	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	33.87	NM_002520.6	c.859_860insTCTG	p.Trp288CysfsTer12		
803	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	42.47	NM_002520.6	c.859_860insTCTG	p.Trp288CysfsTer12		

UPN	AML category	Gene	Chr	Coordinate (Hg19)	Variant	Type	Consequence	VAF	Transcript	c.DNA	protein	Sift	PolyPhen
824	*de novo*	NPM1	5	170837545	C>C/CTGCA	insertion	frameshift	16.4	NM_002520	c.863_864insCATG	p.Trp288CysfsTer12		
830	*de novo*	NPM1	5	170837545	C>C/CTGCA	insertion	frameshift	40.53	NM_002520.6	c.861_862insTGCA	p.Trp288CysfsTer12		
838	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	38.41	NM_002520.6	c.859_860insTCTG	p.Trp288CysfsTer12		
847	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	37.2	NM_002520.6	c.859_860insTCTG	p.Trp288CysfsTer12		
852	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	41.89	NM_002520.6	c.859_860insTCTG	p.Trp288CysfsTer12		
853	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	40.64	NM_002520.6	c.859_860insTCTG	p.Trp288CysfsTer12		
854	*de novo*	NPM1	5	170837544	T>T/TCTGC	insertion	frameshift	38.16	NM_002520.6	c.860_861insCTGC	p.Trp288CysfsTer12		
865	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	23.62	NM_002520	c.860_863dupTCTG	p.Trp288CysfsTer12		
867	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	28.58	NM_002520.6	c.859_860insTCTG	p.Trp288CysfsTer12		
877	*de novo*	NPM1	5	170837547	G>G/GTTAC	insertion	frameshift	42.71	NM_002520.6	c.863_864insTTAC	p.Trp288CysfsTer12		
919	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	45.4	NM_002520.6	c.859_860insTCTG	p.Trp288CysfsTer12		
931	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	41.44	NM_002520.6	c.859_860insTCTG	p.Trp288CysfsTer12		
981	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	41.22	NM_002520.6	c.859_860insTCTG	p.Trp288CysfsTer12		
1025	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	46.72	NM_002520.6	c.859_860insTCTG	p.Trp288CysfsTer12		
1042	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	40.2	NM_002520	c.860_863dupTCTG	p.Trp288CysfsTer12		
1095	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	18.54	NM_002520	c.860_863dupTCTG	p.Trp288CysfsTer12		
1100	*de novo*	NPM1	5	170837548	G>G/C	snv	missense	34.62	NM_002520.6	c.864G>C	p.Trp288Cys	deleterious (0)	probably damaging (0.88)
1140	*de novo*	NPM1	5	170837544	T>T/TCTGC	insertion	frameshift	41.76	NM_002520.6	c.860_861insCTGC	p.Trp288CysfsTer12		
1147	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	23.8	NM_002520	c.860_863dupTCTG	p.Trp288CysfsTer12		
1168	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	36.08	NM_002520.6	c.859_860insTCTG	p.Trp288CysfsTer12		
1199	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	43.61	NM_002520.6	c.859_860insTCTG	p.Trp288CysfsTer12		
1219	*de novo*	NPM1	5	170837545	C>C/CTGCA	insertion	frameshift	43.08	NM_002520.6	c.861_862insTGCA	p.Trp288CysfsTer12		
1222	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	16.18	NM_002520	c.860_863dupTCTG	p.Trp288CysfsTer12		
1236	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	37.53	NM_002520.6	c.859_860insTCTG	p.Trp288CysfsTer12		
1285	*de novo*	NPM1	5	170837545	C>C/CTGCT	insertion	frameshift	13.34	NM_002520.6	c.861_862insTGCT	p.Trp288CysfsTer12		
1290	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	23.43	NM_002520	c.860_863dupTCTG	p.Trp288CysfsTer12		
1298	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	35.82	NM_002520.6	c.859_860insTCTG	p.Trp288CysfsTer12		
1314	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	38.39	NM_002520.6	c.859_860insTCTG	p.Trp288CysfsTer12		
1338	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	42	NM_002520.6	c.859_860insTCTG	p.Trp288CysfsTer12		
1351	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	41.92	NM_002520.6	c.859_860insTCTG	p.Trp288CysfsTer12		
1364	*de novo*	NPM1	5	170837545	C>C/CTGCT	insertion	frameshift	39.9	NM_002520.6	c.861_862insTGCT	p.Trp288CysfsTer12		
1370	*de novo*	NPM1	5	170837545	C>C/CTGCA	insertion	frameshift	23.33	NM_002520	c.863_864insCATG	p.Trp288CysfsTer12		
1376	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	40	NM_002520.6	c.859_860insTCTG	p.Trp288CysfsTer12		
1379	*de novo*	NPM1	5	170837543	C>C/CTCTG	insertion	frameshift	42.3	NM_002520.6	c.859_860insTCTG	p.Trp288CysfsTer12		
6	CS-AML	NRAS	1	115258747	C>C/T	snv	missense	12.37	NM_002524.4	c.35G>A	p.Gly12Asp	deleterious (0)	possibly damaging (0.459)
61	CS-AML	NRAS	1	115256530	G>G/T	snv	missense	55.83	NM_002524.4	c.181C>A	p.Gln61Lys	deleterious (0.01)	possibly damaging (0.751)
102	CS-AML	NRAS	1	115258744	C>C/T	snv	missense	20.01	NM_002524.4	c.38G>A	p.Gly13Asp	deleterious (0.03)	possibly damaging (0.394)
108	*de novo*	NRAS	1	115258744	C>C/T	snv	missense	25.09	NM_002524.4	c.38G>A	p.Gly13Asp	deleterious (0.03)	possibly damaging (0.394)
185	*de novo*	NRAS	1	115258744	C>C/T	snv	missense	31.21	NM_002524.4	c.38G>A	p.Gly13Asp	deleterious (0.03)	possibly damaging (0.394)

UPN	AML category	Gene	Chr	Coordinate (Hg19)	Variant	Type	Consequence	VAF	Transcript	c.DNA	protein	Sift	PolyPhen
274	*de novo*	NRAS	1	115258747	C>C/T	snv	missense	12.78	NM_002524.4	c.35G>A	p.Gly12Asp	deleterious (0)	possibly damaging (0.459)
320	*de novo*	NRAS	1	115258748	C>C/T	snv	missense	40.03	NM_002524.4	c.34G>A	p.Gly12Ser	deleterious (0.03)	possibly damaging (0.475)
404	*de novo*	NRAS	1	115258744	C>C/T	snv	missense	13.35	NM_002524.4	c.38G>A	p.Gly13Asp	deleterious (0.03)	possibly damaging (0.394)
447	sAML	NRAS	1	115258744	C>C/T	snv	missense	22.48	NM_002524	c.38G>A	p.Gly13Asp	tolerated (0.971)	possibly damaging (0.383)
447	sAML	NRAS	1	115258747	C>C/G	snv	missense	12.68	NM_002524	c.35G>C	p.Gly12Ala	tolerated (0.968)	possibly damaging (0.589)
470	CS-AML	NRAS	1	115258747	C>C/A	snv	missense	23.08	NM_002524.4	c.35G>T	p.Gly12Val	deleterious (0)	possibly damaging (0.58)
505	CS-AML	NRAS	1	115258744	C>C/T	snv	missense	38.67	NM_002524.4	c.38G>A	p.Gly13Asp	deleterious (0.03)	possibly damaging (0.394)
548	*de novo*	NRAS	1	115256477	G>G/T	snv	missense	33.48	NM_002524.4	c.234C>A	p.Phe78Leu	deleterious (0)	possibly damaging (0.784)
830	*de novo*	NRAS	1	115258747	C>C/T	snv	missense	38.48	NM_002524.4	c.35G>A	p.Gly12Asp	deleterious (0)	possibly damaging (0.459)
931	*de novo*	NRAS	1	115258744	C>C/T	snv	missense	22.8	NM_002524.4	c.38G>A	p.Gly13Asp	deleterious (0.03)	possibly damaging (0.394)
1140	*de novo*	NRAS	1	115258747	C>C/T	snv	missense	21.47	NM_002524.4	c.35G>A	p.Gly12Asp	deleterious (0)	possibly damaging (0.459)
241	*de novo*	PHF6	X	133527636	C>C/T	snv	stop gained	49.49	NM_032458.2	c.346C>T	p.Arg116Ter		
378	CS-AML	PHF6	X	133551270	T>T/G	snv	missense	40.47	NM_032458.2	c.906T>G	p.His302Gln	deleterious (0.01)	possibly damaging (0.816)
423	*de novo*	PHF6	X	133551239	G>G/A	snv	missense	60.97	NM_032458.2	c.875G>A	p.Cys292Tyr	deleterious (0)	probably damaging (0.995)
633	CS-AML	PHF6	X	133511739	T>T/TA	insertion	frameshift	41.23	NM_032458.2	c.92_93insA	p.Leu32ThrfsTer4		
830	*de novo*	PHF6	X	133551333	G>G/A	snv	splicing	69.21	NM_032458.2	c.968+1G>A	p.?		
13	CS-AML	PTPN11	12	112926852	C>C/T	snv	missense	39.78	NM_002834.3	c.1472C>T	p.Pro491Leu	tolerated (0.19)	probably damaging (0.894)
64	*de novo*	PTPN11	12	112926910	G>G/C	snv	missense	22.89	NM_002834.3	c.1530G>C	p.Gln510His	deleterious (0)	probably damaging (0.99)
91	*de novo*	PTPN11	12	112888261	G>G/C	snv	missense	53.16	NM_002834.3	c.277G>C	p.Gly93Arg	deleterious (0)	probably damaging (0.993)
110	*de novo*	PTPN11	12	112888156	A>A/T	snv	missense	23.06	NM_002834.3	c.172A>T	p.Asn58Tyr	deleterious (0.01)	probably damaging (0.91)
182	*de novo*	PTPN11	12	112926888	G>G/C	snv	missense	41.01	NM_002834.3	c.1508-1509GG>CT	p.Gly503Ala	deleterious (0)	probably damaging (0.993)
213	*de novo*	PTPN11	12	112888210	G>G/A	snv	missense	9.95	NM_002834.3	c.226G>A	p.Glu76Lys	deleterious (0.02)	probably damaging (0.915)
455	*de novo*	PTPN11	12	112888198	G>G/A	snv	missense	23.66	NM_002834	c.214G>A	p.Ala72Thr	tolerated (0.998)	probably damaging (0.956)
477	*de novo*	PTPN11	12	112888165	G>G/A	snv	missense	35.57	NM_002834	c.181G>A	p.Asp61Asn	tolerated (0.999)	probably damaging (0.986)
616	*de novo*	PTPN11	12	112926248	G>G/A	snv	missense	15.23	NM_002834	c.1381G>A	p.Ala461Thr	tolerated (0.999)	probably damaging (0.999)
684	*de novo*	PTPN11	12	112926872	C>C/T	snv	missense	16.23	NM_002834.3	c.1492C>T	p.Arg498Trp	deleterious (0.02)	possibly damaging (0.899)
693	*de novo*	PTPN11	12	112888162	G>G/C	snv	missense	17.48	NM_002834.3	c.178G>C	p.Gly60Arg	deleterious (0)	probably damaging (0.999)

UPN	AML category	Gene	Chr	Coordinate (Hg19)	Variant	Type	Consequence	VAF	Transcript	c.DNA	protein	Sift	PolyPhen
847	*de novo*	PTPN11	12	112888163	G>G/C	snv	missense	19.37	NM_002834.3	c.179G>C	p.Gly60Ala	deleterious (0.01)	probably damaging (0.997)
852	*de novo*	PTPN11	12	112888165	G>G/T	snv	missense	37.73	NM_002834.3	c.181G>T	p.Asp61Tyr	deleterious (0)	probably damaging (0.998)
1152	*de novo*	PTPN11	12	112888190	A>A/T	snv	missense	13.05	NM_002834.3	c.206A>T	p.Glu69Val	deleterious (0)	possibly damaging (0.705)
1152	*de novo*	PTPN11	12	112888165	G>G/C	snv	missense	11.17	NM_002834.3	c.181G>C	p.Asp61His	deleterious (0)	probably damaging (0.996)
1166	CS-AML	PTPN11	12	112888163	G>G/C	snv	missense	17.06	NM_002834.3	c.179G>C	p.Gly60Ala	deleterious (0.01)	probably damaging (0.997)
1219	*de novo*	PTPN11	12	112926885	C>C/T	snv	missense	44.68	NM_002834.3	c.1505C>T	p.Ser502Leu	deleterious (0)	probably damaging (1)
1376	*de novo*	PTPN11	12	112888211	A>A/G	snv	missense	33.11	NM_002834.3	c.227A>G	p.Glu76Gly	deleterious (0)	possibly damaging (0.767)
320	*de novo*	RAD21	8	117866660	CACTGTCAACAATT>CACTGTCAACAATT/C	deletion	frameshift	39.4	NM_006265.2	c.972_984delAATTGTTGACAGT	p.Ile325SerfsTer32		
548	*de novo*	RAD21	8	117870612	T>T/TA	insertion	frameshift	49.2	NM_006265.2	c.459dupT	p.Ile154TyrfsTer6		
548	*de novo*	RAD21	8	117869584	C>C/T	snv	missense	11.98	NM_006265.2	c.610G>A	p.Glu204Lys	tolerated (0.35)	benign (0.052)
1236	*de novo*	RAD21	8	117869505	C>C/T	snv	splicing	37.9	NM_006265.2	c.688+1G>A	p.?		
3	CS-AML	RUNX1	21	36259312	G>G/A	snv	missense	50.81	NM_001754.4	c.179C>T	p.Ala60Val	tolerated (0.2)	benign (0.003)
6	CS-AML	RUNX1	21	36259373	A>A/AGCGGCGG	insertion	frameshift	23.36	NM_001754.4	c.111_117dupCCGCCGC	p.Phe40ProfsTer100		
61	CS-AML	RUNX1	21	36171756	G>G/GT	insertion	frameshift	42.87	NM_001754.4	c.808dupA	p.Thr270AsnfsTer330		
62	CS-AML	RUNX1	21	36231783	G>G/A	snv	nonsense	45.01	NM_001754	c.601C>T	p.Arg201*		
62	CS-AML	RUNX1	21	36252920	T>T/TAGCATTTCTCAGCTC	insertion	inframe	39	NM_001754	c.427_441dupGAGCTGAGAAATGCT	p.Glu143_Ala147dup		
82	sAML	RUNX1	21	36259166	T>T/C	snv	missense	49.85	NM_001754.4	c.325A>G	p.Asn109Asp	deleterious (0)	probably damaging (0.987)
82	sAML	RUNX1	21	36231792	C>C/T	snv	missense	39.3	NM_001754.4	c.592G>A	p.Asp198Asn	deleterious (0)	possibly damaging (0.216)
102	CS-AML	RUNX1	21	36231782	C>C/T	snv	missense	14.36	NM_001754.4	c.602G>A	p.Arg201Gln	deleterious (0)	benign (0.135)
131	CS-AML	RUNX1	21	36164707	G>G/GC	insertion	frameshift	38.55	NM_001754.4	c.1167dupG	p.Gln390AlafsTer210		
139	CS-AML	RUNX1	21	36206705	A>A/G	snv	splicing	25.44	NM_001754.4	c.805+2T>C	p.?		
139	CS-AML	RUNX1	21	36252869	C>C/CGGGTTAGGTT	insertion	frameshift	15.66	NM_001754.4	c.492_493insAACCTAACCC	p.Gly165AsnfsTer51		
226	sAML	RUNX1	21	36259229	C>C/A	snv	stop gained	43.33	NM_001754.4	c.262G>T	p.Glu88Ter		
250	CS-AML	RUNX1	21	36164903	T>T/TGCCGCTGCAGGGC	insertion	frameshift	30.97	NM_001754.4	c.968-9_971dupGCCCTGCAGCGGC	p.Asp326CysfsTer278		
258	CS-AML	RUNX1	21	36171600	G>G/GA	insertion	frameshift	18.84	NM_001754.4	c.964dupT	p.Ser322PhefsTer278		
265	CS-AML	RUNX1	21	36231782	C>C/T	snv	missense	46.39	NM_001754.4	c.602G>A	p.Arg201Gln	deleterious (0)	benign (0.135)
265	CS-AML	RUNX1	21	36252940	G>G/A	snv	missense	42	NM_001754.4	c.422C>T	p.Ser141Leu	deleterious (0.03)	probably damaging (0.994)
361	CS-AML	RUNX1	21	36259172	G>G/A	snv	missense	39.84	NM_001754.4	c.319C>T	p.Arg107Cys	deleterious (0)	probably damaging (0.998)
369	sAML	RUNX1	21	36252933	CTCAGCCGAGTAGTTTTCATCA>CTCAGCCGAGTAGTTTTCATCA/C	deletion	missense	13.91	NM_001754.4	c.408_428delTGATGAAAACTACTCGGCTGA	p.Asn136_Glu143delinsLys		
378	CS-AML	RUNX1	21	36231805	GATGGCTCTGTGGTAGGT>GATGGCTCTGTGGTAGGT/G	deletion	frameshift	63.13	NM_001754.4	c.562_578delACCTACCACAGAGCCAT	p.Thr188GlnfsTer19		

UPN	AML category	Gene	Chr	Coordinate (Hg19)	Variant	Type	Consequence	VAF	Transcript	c.DNA	protein	Sift	PolyPhen
378	CS-AML	RUNX1	21	36252875	A>A/C	snv	missense	44.72	NM_001754.4	c.487T>G	p.Phe163Val	deleterious (0)	probably damaging (0.995)
387	CS-AML	RUNX1	21	36259245	C>C/CA	insertion	frameshift	44.55	NM_001754.4	c.245dupT	p.Ala83GlyfsTer55		
470	CS-AML	RUNX1	21	36252910	A>A/C	snv	missense	35.38	NM_001754.4	c.452T>G	p.Met151Arg	deleterious (0)	probably damaging (0.999)
505	CS-AML	RUNX1	21	36259335	CA>CA/C	deletion	frameshift	49.6	NM_001754.4	c.155delT	p.Met52ArgfsTer20		
505	CS-AML	RUNX1	21	36252865	C>C/A	snv	missense	47.41	NM_001754.4	c.497G>T	p.Arg166Leu	deleterious (0.01)	probably damaging (0.992)
508	*de novo*	RUNX1	21	36206833	C>C/A	snv	stop gained	32.14	NM_001754.4	c.679G>T	p.Glu227Ter		
605	CS-AML	RUNX1	21	36231855	TG>TG/T	deletion	frameshift	79.19	NM_001754.4	c.528delC	p.Ile177SerfsTer34		
733	*de novo*	RUNX1	21	36164479	T>T/C	snv	missense	62.11	NM_001754.4	c.1396A>G	p.Met466Val	deleterious (0.02)	probably damaging (0.922)
846	CS-AML	RUNX1	21	36171622	CAG>CAG/C	deletion	frameshift	11.85	NM_001754.4	c.941_942delCT	p.Ser314CysfsTer285		
940	CS-AML	RUNX1	21	36231773	C>C/T	snv	missense	78.5	NM_001754	c.611G>A	p.Arg204Gln	tolerated (0.999)	probably damaging (0.994)
997	CS-AML	RUNX1	21	36259327	G>G/GA	insertion	frameshift	27.37	NM_001754.4	c.163_164insT	p.Ala55ValfsTer83		
997	CS-AML	RUNX1	21	36259328	C>C/G	snv	missense	27.91	NM_001754.4	c.163G>C	p.Ala55Pro	tolerated (0.29)	benign (0.016)
997	CS-AML	RUNX1	21	36259329	C>C/G	snv	missense	27.64	NM_001754.4	c.162G>C	p.Glu54Asp	tolerated (0.45)	probably damaging (0.989)
1110	sAML	RUNX1	21	36252866	G>G/C	snv	missense	26.3	NM_001754.4	c.496C>G	p.Arg166Gly	deleterious (0)	probably damaging (0.996)
1201	sAML	RUNX1	21	36164611	C>C/A	snv	nonsense	65.22	NM_001754	c.1264G>T	p.Glu422*		
1270	CS-AML	RUNX1	21	36252865	C>C/A	snv	missense	68.23	NM_001754.4	c.497G>T	p.Arg166Leu	deleterious (0.01)	probably damaging (0.992)
1343	sAML	RUNX1	21	36171612	G>G/GA	insertion	frameshift	28.77	NM_001754.4	c.952dupT	p.Ser318PhefsTer282		
149	CS-AML	SETBP1	18	42531907	G>G/A	snv	missense	42.06	NM_015559	c.2602G>A	p.Asp868Asn	tolerated (1)	probably damaging (0.998)
1095	*de novo*	SETBP1	18	42531194	C>C/T	snv	missense	44.87	NM_015559	c.1889C>T	p.Ala630Val	tolerated (0.895)	benign (0.017)
62	CS-AML	SF3B1	2	198267705	C>C/T	snv	missense	47.23	NM_012433	c.1774G>A	p.Glu592Lys	tolerated (0.999)	probably damaging (0.999)
82	sAML	SF3B1	2	198266834	T>T/C	snv	missense	31.47	NM_012433.2	c.2098A>G	p.Lys700Glu	deleterious (0.01)	probably damaging (0.993)
102	CS-AML	SF3B1	2	198267480	T>T/C	snv	missense	38.63	NM_012433.2	c.1877A>G	p.Asn626Ser	deleterious (0)	probably damaging (0.975)
369	sAML	SF3B1	2	198266834	T>T/C	snv	missense	43.5	NM_012433.2	c.2098A>G	p.Lys700Glu	deleterious (0.01)	probably damaging (0.993)
505	CS-AML	SF3B1	2	198266834	T>T/C	snv	missense	47.79	NM_012433.2	c.2098A>G	p.Lys700Glu	deleterious (0.01)	probably damaging (0.993)
518	CS-AML	SF3B1	2	198267360	T>T/A	snv	missense	31.79	NM_012433.2	c.1997A>T	p.Lys666Met	deleterious (0)	probably damaging (0.997)
519	CS-AML	SF3B1	2	198267359	C>C/G	snv	missense	49.79	NM_012433.2	c.1998G>C	p.Lys666Asn	deleterious (0)	probably damaging (0.993)
519	CS-AML	SF3B1	2	198266803	G>G/GC	insertion	frameshift	11.67	NM_012433.2	c.2128dupG	p.Ala710GlyfsTer5		
1043	CS-AML	SF3B1	2	198266834	T>T/C	snv	missense	34.48	NM_012433	c.2098A>G	p.Lys700Glu	tolerated (0.996)	probably damaging (0.999)
1275	CS-AML	SF3B1	2	198267359	C>C/A	snv	missense	35.42	NM_012433.2	c.1998G>T	p.Lys666Asn	deleterious (0)	probably damaging (0.993)
13	CS-AML	SMC1A	X	53441941	C>C/T	snv	missense	85.22	NM_006306.2	c.287G>A	p.Arg96His	deleterious (0)	probably damaging (1)

UPN	AML category	Gene	Chr	Coordinate (Hg19)	Variant	Type	Consequence	VAF	Transcript	c.DNA	protein	Sift	PolyPhen
91	*de novo*	SMC3	10	112343991	G>G/T	snv	missense	33.12	NM_005445.3	c.1142G>T	p.Arg381Leu	deleterious (0.01)	probably damaging (0.999)
847	*de novo*	SMC3	10	112343991	G>G/A	snv	missense	20.75	NM_005445.3	c.1142G>A	p.Arg381Gln	deleterious (0.02)	probably damaging (0.973)
931	*de novo*	SMC3	10	112356174	G>G/C	snv	missense	24.34	NM_005445.3	c.1982G>C	p.Arg661Pro	deleterious (0.01)	probably damaging (0.996)
46	sAML	SRSF2	17	74732935	CGGCGGCTGTGGTGTGAGTCCGGGG>CGGCGGCTGTGGTGTGAGTCCGGGG/C	deletion	inframe deletion	56.78	NM_001195427.1	c.284_307delCCCCGGACTCACACCACAGCCGCC	p.Pro95_Arg103del		
126	CS-AML	SRSF2	17	74732935	CGGCGGCTGTGGTGTGAGTCCGGGG>CGGCGGCTGTGGTGTGAGTCCGGGG/C	deletion	inframe deletion	72.04	NM_001195427.1	c.284_307delCCCCGGACTCACACCACAGCCGCC	p.Pro95_Arg103del		
131	CS-AML	SRSF2	17	74732959	G>G/C	snv	missense	48.48	NM_001195427.1	c.284C>G	p.Pro95Arg	deleterious (0.02)	possibly damaging (0.668)
149	CS-AML	SRSF2	17	74732959	G>G/T	snv	missense	39.53	NM_003016	c.284C>A	p.Pro95His	tolerated (0.999)	probably damaging (0.984)
226	sAML	SRSF2	17	74732959	G>G/C	snv	missense	25.75	NM_001195427.1	c.284C>G	p.Pro95Arg	deleterious (0.02)	possibly damaging (0.668)
242	CS-AML	SRSF2	17	74732959	G>G/T	snv	missense	51.12	NM_001195427.1	c.284C>A	p.Pro95His	deleterious (0.01)	probably damaging (0.979)
246	*de novo*	SRSF2	17	74732959	G>G/C	snv	missense	43.95	NM_001195427.1	c.284C>G	p.Pro95Arg	deleterious (0.02)	possibly damaging (0.668)
470	CS-AML	SRSF2	17	74733073	A>A/T	snv	missense	32.07	NM_001195427.1	c.170T>A	p.Phe57Tyr	deleterious (0.05)	probably damaging (0.914)
605	CS-AML	SRSF2	17	74732959	G>G/C	snv	missense	51.99	NM_001195427.1	c.284C>G	p.Pro95Arg	deleterious (0.02)	possibly damaging (0.668)
753	CS-AML	SRSF2	17	74733191	C>C/A	snv	missense	67.43	NM_001195427.1	c.52G>T	p.Val18Leu	deleterious (0.01)	probably damaging (0.857)
1201	sAML	SRSF2	17	74732959	G>G/T	snv	missense	40.19	NM_003016	c.284C>A	p.Pro95His	tolerated (0.999)	probably damaging (0.984)
1298	*de novo*	SRSF2	17	74732959	G>G/C	snv	missense	44.04	NM_001195427.1	c.284C>G	p.Pro95Arg	deleterious (0.02)	possibly damaging (0.668)
1324	sAML	SRSF2	17	74732960	G>G/C	snv	missense	47.66	NM_001195427.1	c.283C>G	p.Pro95Ala	tolerated (0.22)	possibly damaging (0.621)
226	sAML	STAG2	X	123220488	C>C/CT	insertion	frameshift	88.25	NM_001042749.1	c.3145_3146insT	p.Ala1050SerfsTer4		
349	CS-AML	STAG2	X	123179171	T>T/A	snv	stop gained	46.61	NM_001042749.1	c.620T>A	p.Leu207Ter		
380	CS-AML	STAG2	X	123229304	G>G/C	snv	splicing	94.08	NM_001042749.1	c.3783+5G>C	p.?		
387	CS-AML	STAG2	X	123220440	C>C/T	snv	stop gained	92.28	NM_001042749.1	c.3097C>T	p.Arg1033Ter		
413	CS-AML	STAG2	X	123184970	G>G/A	snv	splicing	97.32	NM_001042749.1	c.1018-1G>A	p.?		
470	CS-AML	STAG2	X	123176470	G>G/GA	insertion	frameshift	96.15	NM_001042749.1	c.437_438insA	p.Met148AsnfsTer3		
470	CS-AML	STAG2	X	123197850	AGAT>AGAT/A	deletion	inframe deletion	13.28	NM_001042749.1	c.1975_1977delGAT	p.Asp659del		
633	CS-AML	STAG2	X	123176438	G>G/C	snv	missense	61.02	NM_001042749.1	c.405G>C	p.Met135Ile	deleterious (0.05)	probably damaging (0.992)
877	*de novo*	STAG2	X	123224469	C>C/T	snv	stop gained	40	NM_001042749.1	c.3322C>T	p.Gln1108Ter		
1110	sAML	STAG2	X	123190063	G>G/A	snv	missense	21.75	NM_001042749.1	c.1282G>A	p.Ala428Thr	deleterious (0)	probably damaging (0.994)
1337	CS-AML	STAG2	X	123197716	C>C/T	snv	stop gained	10.01	NM_001042749.1	c.1840C>T	p.Arg614Ter		
1337	CS-AML	STAG2	X	123202497	T>T/TA	insertion	frameshift	10	NM_001042749.1	c.2349_2350insA	p.Glu785GlyfsTer9		
28	*de novo*	TET2	4	106197481	C>C/CT	insertion	frameshift	51.05	NM_001127208.2	c.5814_5815insT	p.Tyr1939LeufsTer17		

UPN	AML category	Gene	Chr	Coordinate (Hg19)	Variant	Type	Consequence	VAF	Transcript	c.DNA	protein	Sift	PolyPhen
46	sAML	TET2	4	106164778	C>C/T	snv	stop gained	94.04	NM_001127208.2	c.3646C>T	p.Arg1216Ter		
61	CS-AML	TET2	4	106157740	AG>AG/A	deletion	frameshift	45.6	NM_001127208.2	c.2642delG	p.Arg881SerfsTer40		
149	CS-AML	TET2	4	106157512	G>G/T	snv	nonsense	44.48	NM_017628	c.2413G>T	p.Gly805*		
149	CS-AML	TET2	4	106190781	CAG>CAG/C	deletion	frameshift	44.3	NM_001127208	c.4062_4063delAG	p.Arg1354Serfs*46		
196	*de novo*	TET2	4	106196421	CT>CT/C	deletion	frameshift	41.97	NM_001127208.2	c.4755delT	p.Ser1586GlnfsTer10		
222	sAML	TET2	4	106157527	C>C/T	snv	stop gained	52.11	NM_001127208.2	c.2428C>T	p.Gln810Ter		
222	sAML	TET2	4	106190799	TC>TC/T	deletion	frameshift	31.47	NM_001127208.2	c.4078delC	p.Leu1360TrpfsTer3		
246	*de novo*	TET2	4	106158322	GA>GA/G	deletion	frameshift	46.55	NM_001127208.2	c.3224delA	p.Asp1075ValfsTer7		
246	*de novo*	TET2	4	106157982	A>A/AG	insertion	frameshift	44.54	NM_001127208.2	c.2883_2884insG	p.Gln962AlafsTer10		
275	*de novo*	TET2	4	106164787	C>C/A	snv	missense	41.87	NM_001127208	c.3655C>A	p.His1219Asn	tolerated (1)	probably damaging (0.999)
377	*de novo*	TET2	4	106182957	T>T/TAAAGGGTCCTAGAGGGTG	insertion	nonsense	19.2	NM_001127208	c.3997_3998insAAGGGTCCTAGAGGGTGA	p.Leu1332_Met1333ins6		
387	CS-AML	TET2	4	106163986	T>T/A	snv	splicing	45.24	NM_001127208.2	c.3501-5T>A	p.?		
402	*de novo*	TET2	4	106190854	T>T/C	snv	missense	42.59	NM_001127208.2	c.4132T>C	p.Cys1378Arg	deleterious (0)	probably damaging (0.994)
441	*de novo*	TET2	4	106197285	T>T/C	snv	missense	45.42	NM_001127208	c.5618T>C	p.Ile1873Thr	tolerated (1)	probably damaging (0.999)
441	*de novo*	TET2	4	106164082	TGGG>TGGG/TGG	deletion	splicing	44.41	NM_001127208	c.3594+1delG	p.?		
447	sAML	TET2	4	106196306	C>C/T	snv	nonsense	38.74	NM_001127208	c.4639C>T	p.Gln1547*		
447	sAML	TET2	4	106196402	TA>TA/T	deletion	frameshift	34.6	NM_001127208	c.4736delA	p.Tyr1579Phefs*17		
659	*de novo*	TET2	4	106164068	G>G/A	snv	missense	48.37	NM_001127208.2	c.3578G>A	p.Cys1193Tyr	deleterious (0)	probably damaging (1)
659	*de novo*	TET2	4	106196558	TA>TA/T	deletion	frameshift	49.56	NM_001127208.2	c.4892delA	p.Tyr1631PhefsTer64		
683	*de novo*	TET2	4	106196778	AT>AT/A	deletion	frameshift	48.91	NM_001127208.2	c.5112delT	p.Asp1704GlufsTer15		
684	*de novo*	TET2	4	106180880	G>G/A	snv	missense	26.98	NM_001127208.2	c.3908G>A	p.Ser1303Asn	deleterious (0.01)	probably damaging (0.998)
716	*de novo*	TET2	4	106197360	C>C/T	snv	missense	52.4	NM_001127208.2	c.5693C>T	p.Ser1898Phe	deleterious (0)	probably damaging (0.999)
716	*de novo*	TET2	4	106157614	C>C/CA	insertion	frameshift	44.18	NM_001127208.2	c.2515_2516insA	p.His839GlnfsTer7		
761	*de novo*	TET2	4	106164778	C>C/T	snv	stop gained	42.7	NM_001127208.2	c.3646C>T	p.Arg1216Ter		
786	CS-AML	TET2	4	106180870	T>T/A	snv	missense	79.64	NM_001127208.2	c.3898T>A	p.Phe1300Ile	deleterious (0.02)	probably damaging (0.993)
803	*de novo*	TET2	4	106196213	C>C/T	snv	stop gained	92.41	NM_001127208.2	c.4546C>T	p.Arg1516Ter		
865	*de novo*	TET2	4	106158470	T>T/TA	insertion	frameshift	45.18	NM_017628	c.3371_3372insA	p.Lys1125Glnfs*5		
865	*de novo*	TET2	4	106190882	A>A/G	snv	missense	39.32	NM_001127208	c.4160A>G	p.Asn1387Ser	tolerated (0.998)	probably damaging (0.999)
940	CS-AML	TET2	4	106196829	T>T/G	snv	missense	49.5	NM_001127208	c.5162T>G	p.Leu1721Trp	tolerated (0.997)	possibly damaging (0.754)
1025	*de novo*	TET2	4	106180823	CCTT>CCTT/C	deletion	inframe deletion	33.73	NM_001127208.2	c.3852_3854delCTT	p.Phe1285del		
1100	*de novo*	TET2	4	106156048	C>C/T	snv	stop gained	36.14	NM_001127208.2	c.949C>T	p.Gln317Ter		
1168	*de novo*	TET2	4	106156234	GCTTA>GCTTA/G	deletion	frameshift	39.95	NM_001127208.2	c.1136_1139delCTTA	p.Tyr380SerfsTer46		
1290	*de novo*	TET2	4	106158349	C>C/T	snv	nonsense	49.43	NM_017628	c.3250C>T	p.Gln1084*		
1290	*de novo*	TET2	4	106156589	CTGTTCCATTG>CTGTTCCATTG/C	insertion	frameshift	20.58	NM_017628	c.1496_1505delCATTGTGTTC	p.Pro499Leufs*31		
1298	*de novo*	TET2	4	106164016	A>A/T	snv	missense	47.52	NM_001127208.2	c.3526A>T	p.Arg1176Trp	deleterious (0)	probably damaging (1)

UPN	AML category	Gene	Chr	Coordinate (Hg19)	Variant	Type	Consequence	VAF	Transcript	c.DNA	protein	Sift	PolyPhen
1298	*de novo*	TET2	4	106164079	AAGT>AAGT/A	deletion	missense	43.06	NM_001127208.2	c.3590_3592delAGT	p.Lys1197_Trp1198delinsArg		
1324	sAML	TET2	4	106193748	C>C/T	snv	stop gained	94.64	NM_001127208.2	c.4210C>T	p.Arg1404Ter		
1351	*de novo*	TET2	4	106164936	G>G/GT	insertion	splicing	46.95	NM_001127208.2	c.3803+1_3803+2insT	p.?		
1364	*de novo*	TET2	4	106182995	A>A/G	snv	missense	47.14	NM_001127208.2	c.4034A>G	p.Tyr1345Cys	deleterious (0)	probably damaging (1)
1364	*de novo*	TET2	4	106162565	T>T/G	snv	missense	43.33	NM_001127208.2	c.3479T>G	p.Ile1160Ser	deleterious (0)	probably damaging (0.993)
35	*de novo*	TP53	17	7577094	G>G/A	snv	missense	11.69	NM_000546.5	c.844C>T	p.Arg282Trp	deleterious (0)	probably damaging (0.999)
35	*de novo*	TP53	17	7578235	T>T/C	snv	missense	9.16	NM_000546.5	c.614A>G	p.Tyr205Cys	deleterious (0)	probably damaging (0.993)
6	CS-AML	U2AF1	21	44524456	G>G/A	snv	missense	44.16	NM_001025203.1	c.101C>T	p.Ser34Phe	deleterious (0)	probably damaging (0.998)
102	CS-AML	U2AF1	21	44514777	T>T/C	snv	missense	40.26	NM_001025203.1	c.470A>G	p.Gln157Arg	deleterious (0)	probably damaging (0.993)
132	sAML	U2AF1	21	44524456	G>G/A	snv	missense	9.7	NM_006758	c.101C>T	p.Ser34Phe	tolerated (1)	probably damaging (1)
166	CS-AML	U2AF1	21	44524456	G>G/A	snv	missense	46	NM_001025203.1	c.101C>T	p.Ser34Phe	deleterious (0)	probably damaging (0.998)
262	CS-AML	U2AF1	21	44524456	G>G/A	snv	missense	37.04	NM_001025203.1	c.101C>T	p.Ser34Phe	deleterious (0)	probably damaging (0.998)
447	sAML	U2AF1	21	44524456	G>G/A	snv	missense	39.41	NM_006758	c.101C>T	p.Ser34Phe	tolerated (1)	probably damaging (1)
786	CS-AML	U2AF1	21	44524456	G>G/A	snv	missense	39.62	NM_001025203.1	c.101C>T	p.Ser34Phe	deleterious (0)	probably damaging (0.998)
825	CS-AML	U2AF1	21	44514780	C>C/T	snv	missense	47.53	NM_001025203.1	c.467G>A	p.Arg156His	deleterious (0.05)	possibly damaging (0.607)
868	CS-AML	U2AF1	21	44514780	C>C/T	snv	missense	30.86	NM_001025203.1	c.467G>A	p.Arg156His	deleterious (0.05)	possibly damaging (0.607)
94	*de novo*	WT1	11	32413565	C>C/A	snv	missense	33.95	NM_024426	c.1385G>T	p.Arg462Leu	tolerated (1)	probably damaging (0.999)
222	sAML	WT1	11	32417941	C>C/CA	insertion	frameshift	34.73	NM_024426.4	c.1110dupT	p.Val371CysfsTer14		
222	sAML	WT1	11	32417911	A>A/AC	insertion	frameshift	34.27	NM_024426.4	c.1140dupG	p.Ser381ValfsTer4		
361	CS-AML	WT1	11	32417911	A>A/AC	insertion	frameshift	42.45	NM_024426.4	c.1140dupG	p.Ser381ValfsTer4		
362	*de novo*	WT1	11	32417914	G>G/C	snv	missense	48.58	NM_024426.4	c.1138C>G	p.Arg380Gly	deleterious (0.02)	probably damaging (0.946)
362	*de novo*	WT1	11	32413573	C>C/CTTAG	insertion	frameshift	35.23	NM_024426.4	c.1376_1377insCTAA	p.Lys459AsnfsTer2		
406	*de novo*	WT1	11	32413565	C>C/T	snv	missense	19.87	NM_024426.4	c.1385G>A	p.Arg462Gln	deleterious (0.03)	probably damaging (0.998)
623	CS-AML	WT1	11	32417907	G>G/GCCGA	insertion	frameshift	8.5	NM_024426	c.1141_1144dupTCGG	p.Ala382Valfs*4		
924	*de novo*	WT1	11	32417941	C>C/CA	insertion	frameshift	21.28	NM_024426.4	c.1110dupT	p.Val371CysfsTer14		
1036	*de novo*	WT1	11	32417913	C>C/CT	insertion	frameshift	18.26	NM_024426.4	c.1138_1139insA+1139C>A	p.Arg380GlnfsTer5		
1166	CS-AML	WT1	11	32417942	A>A/AG	insertion	frameshift	19.8	NM_024426.4	c.1109_1110insC	p.Val371CysfsTer14		
1343	sAML	WT1	11	32417941	C>C/CA	insertion	frameshift	24.89	NM_024426.4	c.1110dupT	p.Val371CysfsTer14		
1201	sAML	ZRSR2	X	15836766	GT>GT/GTT	insertion	splicing	69.15	NM_005089	c.827+2dupT	p.?		
1343	sAML	ZRSR2	X	15841230	C>C/CAGCCGG	insertion	inframe insertion	41.66	NM_005089.3	c.1314_1315insAGCCGG	p.Gly438_Ser439insSerArg		

Supplemental Table 3. Univariate analysis for CR by AML category on the whole patients' cohort.

All patients	**CS-AML (N=55)**		**sAML (N=100)**		***de novo* AML (N=258)**	
Characteristics	**HR (95% CI)**	***P***	**HR (95% CI)**	***P***	**HR (95% CI)**	***P***
Age [years]						
≥60	2.22 (0.46-16.27)	0.36	0.15 (0.05-0.38)	0.06	0.45 (0.2-1.01)	0.0001
ECOG PS						
2-3	0.28 (0.04-2.32)	0.19	0.39 (0.08-1.67)	0.21	0.13 (0.05-0.36)	0.0001
WBC count [x109/L]						
≥50	0.71 (0.14-5.39)	0.70	0.54 (0.15-1.92)	0.34	0.31 (0.11-0.79)	0.01
Induction arm						
ICE	0.81 (0.17-3.79)	0.78	0.68 (0.3-1.51)	0.35	1.14 (0.44-2.96)	0.78
Genetics*						
normal karyotype	0.98 (0.05-7.05)	0.98	1.66 (0.5-6.53)	0.42	0.66 (0.21-1.8)	0.44
t(8;21)	-		-		1.93 (0.37-35.59)	0.53
inv(16)/t(16;16)	-		-		3.12 (0.61-57)	0.28
complex karyotype	-		0.55 (0.23-1.25)	0.16	-	
chromosome 7 abnormalities	-		1.66 (0.5-6.53)	0.42	-	
chromosome 5 abnormalities	-		1.04 (0.17-8.21)	0.96	-	
RUNX1-RUNX1T1	-		-		2.14 (0.41-39.36)	0.47
CBFB-MYH11	-		-		3.4 (0.67-62.06)	0.24
biallelic *CEBPa*	-		-		2 (0.38-37.03)	0.51
NPM1	-		-		0.53 (0.17-1.43)	0.23
FLT3-ITD	>99.99 (0-NA)	0.99	1.42 (0.26-10.59)	0.69	0.39 (0.15-1.02)	0.05
KMT2A-PTD	2.87 (0.59-21.02)	0.22	2.37 (0.29-49.13)	0.46	-	
ASXL1	0.67 (0.12-5.25)	0.66	0.06 (0-0.89)	0.07	-	
BCOR	>99.99 (0-NA)	1.00	-		-	
RUNX1	0.55 (0.1-2.8)	0.47	1.33 (0.1-17.65)	0.82	-	
SF3B1	>99.99 (0-NA)	0.99	-		-	
STAG2	1.07 (0.14-22.47)	0.95	-		-	
U2AF1	1.12 (0.15-23.23)	0.92	-		-	
SRSF2	0.16 (0.02-1.02)	0.05	0.06 (0-0.89)	0.07	-	

Abbreviations: ICE, idarubicin, cytarabine and etoposide; HR, hazard ratio; CI, confidence interval; NA, not applicable.

*For this analysis, only chromatin-spliceosome mutations and abnormalities included in the ELN risk stratification occurring in at least 3 patients in each AML category were considered.

Supplemental Table 4. Multivariable analysis for CR, OS and DFS on 55 CS-AML patients.

CS-AML patients	**Complete remission**		**Overall survival**		**Disease free survival**	
Characteristics	**HR (95% CI)**	***P***	**HR (95% CI)**	***P***	**HR (95% CI)**	***P***
Age [years]						
≥60	5.73 (0.09-2224.02)	0.46	1.17 (0.34-4.00)	0.80	3.88 (0.92-16.38)	0.07
ECOG PS						
2-3	0.03 (0-32.95)	0.31	42.76 (3.22-566.79)	0.004	23.26 (0.87-622.92)	0.06
WBC count [x109/L]						
≥50	3.51 (0.02-20080.4)	0.66	0.18 (0.03-1.22)	0.08	0.06 (0.01-0.58)	0.01
Induction arm						
ICE	0.39 (0.01-7.11)	0.55	0.56 (0.21-1.48)	0.24	0.23 (0.05-0.98)	0.05
Gene mutations						
KMT2A-PTD	3.13 (0.14-115.45)	0.46	0.64 (0.21-2.00)	0.45	0.99 (0.28-3.48)	0.98
FLT3-ITD	>99.99 (0-NA)	1.00	2.21 (0.36-13.4)	0.39	3.4 (0.49-23.45)	0.21
ASXL1	5.97 (0.12-3620.83)	0.45	0.61 (0.18-2.06)	0.43	1.38 (0.36-5.21)	0.64
BCOR	>99.99 (0-NA)	1.00	0.78 (0.12-5.11)	0.79	0.73 (0.09-5.75)	0.76
RUNX1	0.09 (0-1.75)	0.14	3.55 (1.28-9.87)	0.01	3.13 (1.1-8.95)	0.03
SF3B1	>99.99 (0-NA)	1.00	0.36 (0.06-1.99)	0.24	0.2 (0.02-1.62)	0.13
SRSF2	0.16 (0-10.43)	0.42	1.50 (0.34-6.56)	0.59	0.4 (0.06-2.75)	0.35
STAG2	0.3 (0-20.86)	0.56	0.79 (0.14-4.40)	0.78	0.37 (0.05-2.97)	0.35
U2AF1	0.4 (0-38.95)	0.66	6.87 (1.71-27.55)	0.006	16.46 (3.14-86.31)	0.0009
Abbreviations: ICE, idarubicin, cytarabine and etoposide; HR, hazard ratio; CI, confidence interval.						

Supplemental Table 5. Multivariable analysis for CR, OS and DFS on the whole patients' cohort, excluding *RUNX1* and or *U2AF1*-mutated patients.

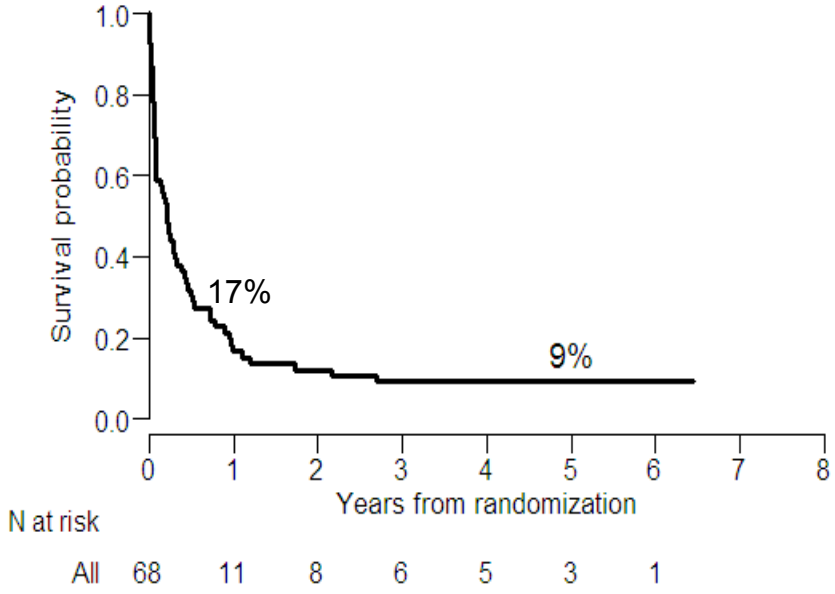
All patients	**Complete remission**		**Overall survival**		**Disease free survival**	
Characteristics	**HR (95% CI)**	***P***	**HR (95% CI)**	***P***	**HR (95% CI)**	***P***
Age [years]						
≥60	0.3 (0.16-0.57)	0.0003	1.69 (1.24-2.32)	0.0010	1.19 (0.83-1.71)	0.3448
ECOG PS						
2-3	0.24 (0.1-0.59)	0.0015	2.37 (1.57-3.58)	<0.0001	1.58 (0.94-2.64)	0.0838
WBC count [x109/L]						
≥50	0.4 (0.18-0.88)	0.0224	1.82 (1.31-2.54)	0.0004	1.76 (1.23-2.5)	0.0018
Induction arm						
ICE	0.88 (0.46-1.67)	0.6871	1.24 (0.92-1.65)	0.1525	1.29 (0.95-1.76)	0.1060
AML category						
sAML	0.09 (0.04-0.18)	<0.0001	3.82 (2.74-5.33)	<0.0001	2.72 (1.85-3.98)	<0.0001
CS-AML	0.85 (0.23-4.19)	0.8184	1.83 (1.07-3.14)	0.0281	1.94 (1.15-3.26)	0.0126
Abbreviations: ICE, idarubicin, cytarabine and etoposide; HR, hazard ratio; CI, confidence interval.						

Supplemental Table 6. Demographic and clinical characteristics of an independent, single-center cohort (N=50) treated at ASST Ospedale Papa Giovanni XXIII between 2012 and 2020.

Clinical characteristics	**CS-AML N=8**	***P****	**sAML N=20**	***P***	***de novo* AML N=22**	***P****
Age [years], median (range)	61.1 (29.4-75)	0.0564	70 (48.5-77.9)	0.0011	55.5 (25.5-79.2)	0.6960
<60, n(%)	3 (37.5)	0.3107	3 (15)	0.0076	12 (54.5)	0.6817
≥60, n(%)	5 (62.5)		17 (85)		10 (45.5)	
Gender, n (%)		1.0000		0.2410		0.3742
M	7 (87.5)		16 (80)		14 (63.6)	
F	1 (12.5)		4 (20)		8 (36.4)	
ECOG PS, n (%)		0.2808		0.4454		0.5448
0-1	8 (100)		15 (75)		19 (86.4)	
2-3	-		5 (25)		3 (13.6)	
Hepatomegaly, n (%)	1 (12.5)	0.6399	5 (25)	0.4454	3 (13.6)	1.0000
Splenomegaly, n (%)	2 (25)	1.0000	7 (35)	0.8605	6 (27.3)	1.0000
Extramedullary involvement, n (%)	1 (12.5)	0.4974	1 (5)	1.0000	2 (9)	1.0000
Hemoglobin [g/dL], median (range)	8.6 (4.8-12.9)	0.6902	9.2 (6.3-13.7)	0.6283	9.8 (4.6-13.2)	0.8144
WBC count [x10^9/L], median (range)	9.6 (0.4-118)	0.3955	3.4 (0.9-51.8)	0.0281	11.9 (1.1-175.5)	0.7652
Platelets, median (range)	75 (23-321)	0.7749	78 (8-187)	0.6472	56 (4-203)	0.4118
BM blast cells (%), median (range)	90 (30-95)	0.0024	30 (20-80)	0.0141	65 (10-92)	0.0369
AML with multilineage dysplasia, n (%)	2 (25)	0.0957	13 (65)	0.0006	3 (13.6)	0.5894
Induction treatment, n (%)		0.0882		0.0003		0.4690
Intensive chemotherapy	7 (87.5)		9 (45)		21 (95.5)^	
HMAs	1 (12.5)		11 (55)§		1 (4.5)	

Abbreviations: CS-AML, chromatin-spliceosome acute myeloid leukemia; sAML, secondary acute myeloid leukemia; *de novo* AML, *de novo* acute myeloid leukemia; ECOG PS, Eastern Cooperative Oncology Group Performance Status; WBC, white blood cell count; BM, bone marrow; HMAs, hypomethylating agents. *P*-values refer to: *CS-AML vs sAML; **sAML vs *de novo* AML; *** *de novo* AML vs CS-AML. Hepatomegaly was defined as lower liver edge >2 cm from costal margin. Splenomegaly was defined as spleen >1 cm from costal margin, confirmed by ultrasound scan with longitudinal axis >12 cm. Extramedullary AML was defined as AML presenting with central nervous system involvement or mass lesions. ^3 *de novo* AML patients also received FLT3 inhibitors. §1 sAML patients received venetoclax with HMAs.

Supplemental Figure 1. Kaplan-Meier analysis of OS on 68 patients not achieving CR after 1 or 2 induction cycles.



Supplemental Figure 2. Kaplan-Meier analysis of OS on 50 consecutive patients of the single-center cohort. CS-AML vs *de novo* AML, *P*=0.13; sAML vs *de novo* AML, *P*=0.004; CS-AML vs sAML, *P*=0.46.