

Patient	Category	Nucleotide change	Amino acid change	Comment
1316	double	357del	D69fsX159	N-terminal stop
		1106_1107insACT	S319delinsRL	In-frame insertion in bZIP
2169	double	213del	P22fsX159	N-terminal stop
		1066_1067insGCAACGTGGACAAGCAGC	V308_E309insDKQRNV	In-frame insertion in bZIP
2192	double	ins396GG	L81fsX160	N-terminal stop
		1060_1062dup	K304dup	In-frame duplication in bZIP
2218	double	1062_1094dup (homozygous)	Q305_L315dup	In-frame duplication in bZIP
2230	double	381del	F57fsX159	N-terminal stop
		1076_1078dup	E309_T310insK	In-frame insertion in bZIP
2234	double	392_395dup	F82fsX108	N-terminal stop
		1084_1089dup, 1090G>A	V314delinsQKM	In-frame insertion in bZIP
2240	double	332del	S161fsX159	N-terminal stop
		1076_1078dup	E309_T310insK	In-frame insertion in bZIP
2242	double	252_261del	P34fsX156	N-terminal stop
		1064_1066dup	Q305dup	In-frame duplication in bZIP
2253	double	213del	P22fsX159	N-terminal stop
		1051_1052insGACAAGGCCAAGCAGCGCAACGTGGAGACGCAGCAC	T310_Q311insQHHKAKQRNVET	In-frame insertion in bZIP
2273	double	472_473insT	D107fsX169	N-terminal stop
		1062_1079dup	R306_Q311dup	In-frame duplication in bZIP
2545	double	474C>G	P108X	N-terminal stop
		813del	L220fsX317	Frameshift between TAD2 and bZIP, stop in bZIP
2748	double	424_425insA	K92fsX107	N-terminal stop
		1057_1058insTTG	K302_A303insV	In-frame insertion in bZIP
2753	double	302_317del	P51fsX154	N-terminal stop
		1085_1087dup	K313dup	In-frame duplication in bZIP
3101	double	362_363insCC (homozygous)	A71fsX160	N-terminal stop
3117	double	397del	F82fsX159	N-terminal stop
		1087_1089dup	K313dup	In-frame duplication in bZIP
3327	double	1104_1115del (homozygous)	S319_D322del	In-frame deletion in bZIP

4336	double	218_219insC	P23fsX107	N-terminal stop
		1016G>C	R289P	Substitution in bZIP
5352	double	311del	G53fsX159	N-terminal stop
		679_691dup	P180fsX324	Frameshift between TAD1 and TAD2, stop in bZIP
5362	double	349_359del	A66fsX103	N-terminal stop
		1033G>C	A295P	Substitution in bZIP
5364	double	376_377insG	D75fsX107	N-terminal stop
		486_522dup	A124fsX181	Frameshift, stop in TAD2
6376	double	1091_1092insTGCTGGAGCTGCAGCGCAACGTGGAGACGCAGCAGAAGG (homozygous)	Q305_L317dup	In-frame duplication in bZIP
6735	double	377_389del	D75fsX155	N-terminal stop
		1084_1086dup	Q312dup	In-frame duplication in bZIP
6975	double	397C>T	Q83X	N-terminal stop
		1073_1075dup	V308dup	In-frame duplication in bZIP
7127	double	437del	V95fsX159	N-terminal stop
		1087_1089dup	K313dup	In-frame duplication in bZIP
7142	double	354delCG	I68fsX106	N-terminal stop
		1087_1089dup	K313dup	In-frame duplication in bZIP
7148	double	248_249del	G32fsX106	N-terminal stop
		1072_1073insGTGGAGACGCAGCACCTAAAATCG	Q311_Q312insHLKSVETQ	In-frame insertion in bZIP
7149	double	382del	F77fsX159	N-terminal stop
		1087_1089dup	K313dup	In-frame duplication in bZIP
7406	double	406_412del	R86fsX157	N-terminal stop
		1084_1086dup	Q312dup	In-frame duplication in bZIP
2176	single	218_219insC	P23fsX107	N-terminal stop
2188	single	852_858dup	P239fsX322	Frameshift between TAD2 and bZIP, stop in bZIP
2194	single	468_469insAACC	D105fsX108	N-terminal stop
2237	single	1188_1189insAGCGCAACGTGGAGACGCAGCAGAAGGTGCTGGAGCTGACCAGTGACAA TGACCGCTGCGCAAGCGGGTGGAAACAGCTGAGCCGCGAACTGGACACGCTGCGGGGC ATCTTCCGCCAGCTGCCA	P346fsX359	Frameshift and stop in bZIP
3096	single	1114_1134del	D322_V328del	In-frame deletion in bZIP

4341	single	309_311delinsTT	G53fsX159	N-terminal stop
6247	single	564_565insTA	Y138fsX160	N-terminal stop
6362	single	648_649insG	E166fsX169	N-terminal stop
6462	single	445_446insCCAA	T98fsX108	N-terminal stop
7075	single	218_219insC	P22fsX107	N-terminal stop
7185	single	1029_1030insGGACCC	N293delinsKDP	In-frame insertion in bZIP
7302	single	505del	A118fsX159	N-terminal stop
7324	single	1075_1203dup	V351fsX402	Frameshift and stop in bZIP
2183#	not included	722-736dup	H191_H195dup	Duplication in TAD2
5359#	not included	575G>C	R142T	Substitution in TAD2

bZIP indicates basic leucine zipper region; TAD1, first transactivation domain; and, TAD2, second transactivation domain. Column “category” indicates whether a case was considered double or single *CEBPA* mutant AML in gene expression profiling and survival analyses. Nucleotide numbering is according to NCBI Entrez accession no. XM_009180.3, in which the major translational start codon starts at nucleotide position 151. All mutations are heterozygous, unless “homozygous” is specifically indicated. The locations of functional domains are derived from Mueller and Pabst.¹

* AML case not included in gene expression profiling and survival analyses because data not available.

AML case considered to be non-mutant in gene expression profiling and survival analysis; see main text.

REFERENCE

1. Mueller BU, Pabst T. C/EBPalpha and the pathophysiology of acute myeloid leukemia. *Curr Opin Hematol.* 2006;13:7-14.