

Table 2Genetic findings of the patients with *CEBPA* mutations.

Patient	Category	Nucleotide changes	Amino acid changes	Comments
4	double	218_219insC	H24fsX107	Produces N-terminal stop codon
		1129_1130insATGTGGAGACGCAGCAGAAAGGTGCTGGAGCTGACCAGTGAC AATGACCGCCTGCGCAAGC	K326_R327insHVETQQKVLELTSD NDRLRK	In-frame insertion in bZIP
6	double	200_218delinsCT	S17fsX101	Produces N-terminal stop codon
		1087_1089dup	K313dup	In-frame duplication in bZIP
7	double	368_369insA	F73fsX107	Produces N-terminal stop codon
		1080_1082del	Q312del	In-frame deletion in bZIP
13	double	303_316del	L52fsX102	Produces N-terminal stop codon
		1062_1063insTTG	K304_Q305insL	In-frame insertion in bZIP
19	double	215_225del	P23fsX103	Produces N-terminal stop codon
		1101_1102insCAGCGCAACGTGGAGACGCAGCAGAAAGGTGCTGGAGCTG	L317_T318insQRNVETQQKVLEL	In-frame insertion in bZIP
22	double	213del	P23fsX159	Produces N-terminal stop codon
		1064_1129dup	K326_R327insQRNVETQQKVLELT SDNDRLRK	In-frame insertion in bZIP
27	double	324_328dup	S61fsX161	Produces N-terminal stop codon
		1062_1063insTTG	K304_Q305insL	In-frame insertion in bZIP
39	double	213del	P23fsX159	Produces N-terminal stop codon
		1081-1086dup	Q311_Q312dup	In-frame duplication in bZIP
47	double	397del	Q83fsX159	Produces N-terminal stop codon
		1101_1102insCAGCGCAACGTGGAGACGCAGCAGAAAGGTGCTGGAGCTG	L317_T318insQRNVETQQKVLEL	In-frame insertion in bZIP
49	double	297_304del	E50fsX104	Produces N-terminal stop codon
		758del	P204fsX317	Frameshift between TAD2 and bZIP; produces stop codon in bZIP
35	single	1087_1089dup	K313dup	In-frame duplication in bZIP

bZIP: basic leucine zipper region, TAD2: second transactivation domain.

Nucleotide numbering was performed according to NCBI Entrez accession no. XM_009180.3, in which the major translational start codon starts at nucleotide position 151. The locations of functional domains are derived from Mueller and Pabst.1