Number Individuals	Familial (F)/ De novo (D)	GATA2 Protein (NP_116027.2)	GATA2 cDNA (NM_032638.5)	Mutation Effect	ACMG/AMP Classification (Curated)	ACMG/AMP criteria	Overall Phenotype	Chromosomal Abnormalities	Somatic Gene Mutations
1	N/A	p.(Glu6Alafs*178)	c.17_18del	FS	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A*	-7	
1	F	p.(Gln20*)	c.58C>T	NS	Pathogenic	PVS1, PS4_Supporting, PM2	*T-ALL, HA, ID*	45,XX,dic(21;22)(p11.2;p11.2)	
7	F	p.(Gly28Alafs*52) (in cis p.(His26Pro))	c.77A>C/c.83delG	FS	Pathogenic	PVS1, PS4_Supporting, PM2, PP1_Moderate	*M/A, ID, L, HA*	-7	
3	F	p.(Glu44*)	c.130G>T	NS	Pathogenic	PVS1, PS4_Supporting, PM2	*L, HA, ID*		
1	F	p.(Tyr59*) (in cis p.?)	c.177C>G (in cis c.[140T>G;142T>C; 145T>C])	NS	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A, ID*	+8	
1	N/A	p.(Ser54*)	c.161C>A	NS	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A*	-7	
1	D	p.(Arg69Leufs*115)	c.206_208delGCGinsT	FS	Pathogenic	PVS1, PS2, PS4_Supporting, PM2	*M/A*		
1	N/A	p.(Val70Leufs*114)	c.207_208delCG	FS	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A*	der(1;7) add	
1	D	p.?	c.222_229+6del14ins21	INDEL	Pathogenic	PVS1, PS2, PS4_Supporting, PM2	*M/A*	+8	
2	F	p.?	c.229+13_229+14insGCCins203_229+13	INDEL	Likely Pathogenic	PS4_Supporting, PM2	*M/A, ID*	+8	
3	F	p.(Arg78Profs*107)	c.232dup [c.230-1_230insC]	FS	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A, L, HA*	-7	
1	N/A	p.(Gly82Argfs*103) [G81fs*]	c.243delinsGC	FS	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A, ID, HA*	NK	
1	N/A	p.(Arg86Profs*98) [C85fs*]	c.257_258del	FS	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A, ID*	-7	
1	N/A	p.(Arg86Alafs*33)	c.256del	FS	Pathogenic	PVS1, PS4_Supporting, PM2	*ID, M/A, HA*		
1	N/A	p.(Gly101Alafs*18) [G101Afs*16]	c.302del	FS	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A, CMML, ID, HA*	del(11)(q13q23), -7, +8	ASXL1
3	N/A	p.(Ala103Glnfs*16)	c.303del	FS	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A*	-7, der(1;7)(q10;p10), der(1;7) add	
1	N/A	p.(Ala103Glnfs*16)	c.306del	FS	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A*	-7, add	
5	F	p.(Leu105Profs*15)	c.312_313dup	FS	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A, ID, L, HA*	-7	
1	N/A	p.(Ser106Cysfs*78)	c.317_318del	FS	Pathogenic	PVS1, PS4_Supporting, PM2	*ID*	NK	
2	F	p.(Ala107Cysfs*78) [S106fs]	c.318dup [c.318_319insT]	FS	Pathogenic	PVS1, PS4_Supporting, PM2	*ID, L*		
15	N/A	p.(Thr117=)	c.351C>G	SPL	Pathogenic	PS3_Very Strong, PS4, PM2, PP1_Strong, PP3	*M/A, ID, L, HA, NS*	-7, +8, Chr1 translocation	FLT3 SETBP1
1	N/A	p.(Val118Glyfs*100) [V618fs]	c.353del	FS	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A*	-7	
1	N/A	p.(Gly136Argfs*49)	c.404dup	FS	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A, ID*		ASXL1
1	N/A	p.(Ser139Cysfs*78)	c.414_417del	FS	Pathogenic	PVS1, PS4_Supporting, PM2	*ID, M/A, HA, L*		
1	N/A	p.(Ser139Cysfs*45)	c.416_417del	FS	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A*	-7	
1	N/A	p.(Val140Cysfs*45) [V140Cfs*44]	c.417dup	FS	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A, ID, HA*	NK	ASXL1
2	N/A	p.(Tyr141*)	c.423C>A	NS	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A, ID*	-7	TERC
1	F	p.(Gly146Valfs*72)	c.437del	FS	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A, ID*	NK	
1	N/A	p.(Glu180*)	c.538G>T	NS	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A, ID*	NK	
1	N/A	p.(Thr188Hisfs*14)	c.561dup	FS	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A*	NK	
1	D	p.(Ala194Serfs*8)	c.579dup	FS	Pathogenic	PVS1, PS2, PS4_Supporting, PM2	*L, M/A*	-7	
1	N/A	p.(Ala198Glyfs*20)	c.593del	FS	Pathogenic	PVS1, PS4_Supporting, PM2	*HA, ID*		
1	F	p.(Gly199Leufs*22)	c.586_593dup	FS	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A, ID, HA*	-9q	ASXL1
5	F	p.(Gly200Valfs*18) [G199fs*]	c.599del [c.594delG]	FS	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A, ID, L*	-7	
11	F	p.(Ser201*) [G200fs]	c.599dup [c.599_600insG]	NS	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A, ID, HA, L*	+8, -7	GATA2

1	N/A	p.(Arg204*)	c.610C>T	NS	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A, ID, HA*	46,XX,der(9)t(1;9)(q12;q12), r(9)(q12q?34)[15]/46,XX [9]	
1	N/A	p.(Val211Argfs*72)	c.627_630dup	FS	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A*	-7	
1	N/A	p.(Glu224*)	c.670G > T	NS	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A, ID, HA*	NK	
1	N/A	p.(Leu229Cysfs*5)	c.685del	FS	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A*	-7	
1	N/A	p.(Gly237Alafs*89) [M236Ifs325*]	c.710del [c.708delC]	FS	Pathogenic	PVS1, PS4_Supporting, PM2	*HA, L, ID*		
2	N/A	p.(Ile246Hisfs*36) [P245fs*]	c.735dup [c.735_736insC]	FS	Pathogenic	PVS1, PS4_Supporting, PM2	*ID, M/A, HA*		
1	F	p.(Tyr260Cysfs*25) [Y260fs*24, D259fs*]	c.769_778dup	FS	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A, ID, HA*	-7	
1	N/A	p.(Phe265Glufs*58)	c.793_802del	FS	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A*		
3	N/A	p.(Gly268*)	c.802G>T	NS	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A, ID, L*	-13q -7 +8	STAG2, MLL, NRAS, TP53, WT1
2	N/A	p.(Gly273Thrfs*8)	c.817_818del [c.814_815del]	FS	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A*	-7 +8	
1	N/A	p.(Pro274Thrfs*8) [G273fs*]	c.818dupG [c.819insG]	FS	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A*	Other	
1	F	p.(Ala286Val)	c.857C>T	MS	Likely pathogenic	PS3, PS4_Supporting, PM2, PP3	*CMML*		
1	N/A	p.(Ser290*)	c.869C>A	NS	Pathogenic	PVS1, PS4_Supporting, PM2	*HA/ID*		
3	F	p.(Cys298Leufs*86)	c.892dup	FS	Pathogenic	PVS1, PS4_Supporting, PM2	*ID, M/A*	Chr1-7 translocation +8	GATA2 RUNX1 ASXL1 ATRX BRCA2 GPRC5A IDH2, NRAS STAG2
2	F	p.(Trp306Alafs*77)	c.915_916del	FS	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A, ID, HA*	-7	
1	N/A	p.(Trp306*)	c.917G>A	NS	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A, HA, L, ID*	-7	
1	N/A	p.(Thr311Argfs*71)	c.932_937delinsG	FS	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A*	-7	
1	F	p.(His313Tyr)	c.937C>T	MS	Likely Pathogenic	PS4_Supporting, PM1, PM2, PP3	*M/A, ID*	-5, -7, +8, add10, -12, -18, -21	
1	F	p.(Tyr314Cysfs*66)	c.941_951del	FS	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A, ID*	-7	
1	N/A	p.(Leu315Pro)	c.944T>C	MS	Likely Pathogenic	PS4_Supporting, PM1, PM2, PP3	*ID*		
1	N/A	p.(Ala318Thrfs*12)	c.941_951dup	FS	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A, ID, HA*	-6	ASXL1
1	F	p.(Ala318Thr)	c.952G>A	MS	Pathogenic	PS3, PS4_Supporting, PM1, PM2, PP3	*M/A, ID, HA*	+1,der(1:7)(q10:p10), +8	
1	F	p.(Cys319Serfs*5)	c.956_962del	FS	Pathogenic	PVS1, PS4_Supporting, PM2	*ID, HA*		
1	N/A	p.(His323Glnfs*61)	c.968dup	FS	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A*	-7	
12	F	p.(Arg330*)	c.988C>T	NS	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A, ID, L, HA, JMML*	-7, +8, +1	STAG2 EZH2 GATA1 HECW2 KRAS GATA2
1	N/A	p.(Leu332Thrfs*53)	c.989_992dup [c.992_993insGACC]	FS	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A, HA, L*	-7, +8	
1	N/A	p.(Leu332Glnfs*60)	c.970_994dup	FS	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A*	-7	
6	N/A	p.(Arg337*)	c.1009C>T	NS	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A, ID, L, HA*	-7	ASXL1
1	N/A	p.?	c.1017+2T>G	SPL	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A, ID*	-7	SETBP1
30	F	p.? INT enhancer (ETS site)	c.1017+572C>T	REG	Pathogenic	PS3, PS4, PM1, PM2, PP1_Strong	*M/A, ID, L, HA, CMML, ALL, B-ALL*	-7 +8 +1 der(Y)t(Y;1)(q11.23;q21) der(1;7)(q10;p10)	ASXL1 TET2 CEBPA
2	F	p.? INT enhancer (del ETS site)	c.1017+513_1017+540del (c.1017+512del28)	REG	Likely Pathogenic	PS3_Supporting, PS4_Supporting, PM1, PM2	*M/A, ID, HA*		
1	N/A	p.?	c.1017+1delG	SPL	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A*	-7	

2	N/A	p.?	c.1018-10_1037del	SPL	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A*	-7	
1	N/A	p.?	c.1018-11_1027del	SPL	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A*	-7	
2	F	p.?	c.1018-2A>G	SPL	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A, L*	-7	ASXL1 SETBP1
1	N/A	p.?	c.1018-2A>T	SPL	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A*	-7	
1	N/A	p.? [S340Gfs*99]	c.1017+2T>G	SPL	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A, ID, HA*		
1	D	p.? [S340Afs*49]	c.1017+2T>C	SPL	Pathogenic	PVS1, PS2, PS4_Supporting, PM2	*M/A, HA, L, ID*		
2	F	p.(Ser340Lysfs*40)	c.1018_1028del	FS	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A, ID*	NK	
2	N/A	p.? [S340-N381del]	c.1018-1 G>A	DEL	Likely Pathogenic	PVS1_Strong, PS4_Supporting, PM2	*M/A,HA, ID*	der(22)t(1;22)(q12;p13)/der(15)t(1;15)(q12;p13)	ASXL1
1	N/A	p.? [S340-N381del]	c.1018-1G>T	DEL	Likely Pathogenic	PVS1_Strong, PS4_Supporting, PM2	*HA,ID*		
1	N/A	p.(Ser340Trpfs*47)	c.1019del	FS	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A*	-7	
2	N/A	p.(Ala342del)I [341delA]	c.1024_1026del [c.1021_1023]	DEL	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A*	-7	
1	N/A	p.(Ala341Aspfs*53)	c.1019_1020insCG ACTGGGAGGGC AAGGCAG	FS	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A, HA, L, ID*		
1	N/A	p.(Ala341Profs*46)	c.1021del	FS	Pathogenic	PVS1, PS4_Supporting, PM2	*HA, ID, L*		
2	N/A	p.? [A341Rfs*38]	c.1018-3_1031del	SPL	Pathogenic	PVS1, PS4_Supporting, PM2	*ID, L, HA, HL*		
1	N/A	p.(Ala341Profs*45)	c.1021_1024del [c.1019_1022del]	FS	Pathogenic	PVS1, PS4_Supporting, PM2	*L, M/A, HL*		
1	N/A	p.(Ala342Argfs*42)	c.1023dup	FS	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A, ID*	NK	
1	N/A	p.(Arg343Profs*42) [A342Gfs*41]	c.1025_1026insG CCG	FS	Pathogenic	PVS1, PS4_Supporting, PM2	*ID, HA*		
2	F	p.(Ala342Profs*45)	c.1023del	FS	Pathogenic	PVS1, PS4_Supporting, PM2	*ID, L*	NK	
3	N/A	p.(Arg344Glyfs*43)	c.1020_1029dup	FS	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A, ID, L, HA*	NK	
2	F	p.(Arg344Glnfs*41)	c.1023_1026dup	FS	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A, ID, HA*	-7	
1	D	p.(Arg344Lysfs*37)	c.1031_1049del	FS	Pathogenic	PVS1, PS2, PS4_Supporting, PM2	*M/A*	NK	
1	N/A	p.(R344Kfs*40)	N/A	FS	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A*	NK	
1	N/A	p.(Gly346Serfs*40)	c.1035_1036ins TCTGGCC	FS	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A, ID, HA*		
1	D	p.(Thr347Argfs*38)	c.1035_1038dup	FS	Pathogenic	PVS1, PS2, PS4_Supporting, PM2	*M/A*	-7	
1	N/A	p.(Thr347Argfs*42)	c.1023_1038dup (c.1038_1039insC GCCAGAAGAGC CGGC)	FS	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A*	-7	
2	F	p.(T347fs)	16bp tandem dup	FS	Pathogenic	PVS1, PS4_Supporting, PM2	*ID, HA*		
1	N/A	p.(Cys348Valfs*39)	c.1041del	FS	Pathogenic	PVS1, PS4_Supporting, PM2	*ID, M/A, HA*	-7	
1	D	p.(Cys349Phe) [C348F]	c.1046G>T	MS	Likely Pathogenic	PS2, PS4_Supporting, PM1, PM2, PM5, PP3	*M/A*	-7	
1	D	p.(Cys349Arg)	c.1045T>C	MS	Pathogenic	PS2, PS4_Supporting, PM1, PM2, PM5, PP3	*M/A, ID*	46,XY,der(3)tdic(1;3)(p11;p25)	ASXL1, TET2 U2AF1
1	N/A	p.(Cys349Gly)	c.1045T>G	MS	Likely Pathogenic	PS4_Supporting, PM1, PM2, PM5, PP3	*ID, M/A,L*		
1	N/A	p.(Asn351Ser)	c.1052A>G	MS	Likely Pathogenic	PS4_Supporting, PM1, PM2, PP3	*M/A*	-7	
1	F	p.(Cys352Arg)	c.1054T>C	MS	Likely Pathogenic	PS4_Supporting, PM1, PM2, PM5, PP3	*M/A*		
1	N/A	p.(Cys352Gly)	c.1054T>G	MS	Likely Pathogenic	PS4_Supporting, PM1, PM2, PM5, PP33	*M/A*	-7	
1	N/A	p.(Cys352Phe)	c.1055G>T	MS	Likely Pathogenic	PS4_Supporting, PM1, PM2, PM5, PP3	*M/A*	der(1;7)(q10;p10),+1	
2	F	p.(Thr354Pro)	c.1060A>C	MS	Likely Pathogenic	PS4_Moderate, PM1, PM2, PM5, PP3	*M/A, ID, L, HA*	NK	

52	F	p.(Thr354Met)	c.1061C>T	MS	Pathogenic	PS3, PS4, PM1, PM2, PM5, PP1_Strong, PP3	*M/A, ID, L, HA*	-7, +8, +21, -5q, 1q abnormality, isochromosome 17, F100 t(1q:7p) +8 replaced by monodicentric 6	biCEBPA ASXL1 DNMT3A NPM1 PTPN11 WAC
1	F	p.(Thr354Arg)	c.1061C>G	MS	Likely Pathogenic	PS4_Supporting, PM1, PM2, PM5, PP3	*M/A, ID*	46,XX [20] / 92,XXXX [2]	
2	F	p.(Thr358del) [355delT]	c.1065_1067del [c.1063_1065delACA]	DEL	Pathogenic	PS3, PS4_Moderate, PM2, PM4	*M/A*	-7 +8	
1	N/A	p.(Thr358del)	c.1072_1074del	DEL	Pathogenic	PS3, PS4_Moderate, PM2, PM4	*ID*		
1	N/A	p.(Thr356_Asn365del)	c.1066_1095del	DEL	Likely Pathogenic	PS4_Supporting, PM2, PM4_Strong	*M/A*	-7	
2	N/A	p.(Thr357Ala)	c.1069A>G	MS	Likely Pathogenic	PS4_Supporting, PM1, PM2, PM5, PP3	*M/A*	der(1;7), add +8	
1	N/A	p.(Thr357Ile)	c.1070C>T	MS	Likely Pathogenic	PS4_Supporting, PM1, PM2, PM5, PP3	*M/A, ID*	NK	
1	F	p.(Thr358Asn), p.(Leu359Val) (in-cis)	c.1073C>A c.1075T>G	MS	Pathogenic/ VUS	PS3, PS4_Supporting, PM1, PM2, PP3/ PM1, PM2, PM5, PP3, BP2	*M/A, HA, ID*		
1	F	p.(Leu359Val)	c.1076T>C	MS	Likely Pathogenic	PS4_Supporting, PM1, PM2, PP3	*M/A, ID, L*	46,XX,del(5)(q2?3q 3?3) [18] / 46,XX	
3	F	p.(Trp360Arg)	c.1078T>A	MS	Likely Pathogenic	PS4_Moderate, PM1, PM2, PM5, PP3	*ID, L, HA*		
1	F	p.(Trp360Leu)	c.1079G>T	MS	Likely Pathogenic	PS4_Supporting, PM1, PM2, PM5, PP3	*M/A, HA, ID*	-7	
2	F	p.(Arg361Gly)	c.1081C>G	MS	Likely Pathogenic	PS4_Moderate, PM1, PM2, PM5, PP3	*M/A, ID, L*		
7	N/A	p.(Arg361Cys)	c.1081C>T	MS	Likely Pathogenic	PS4, PM1, PM5, PP3	*MA, ID, L, HA*	-7	
1	D	p.(Arg361Leu)	c.1082G>T	MS	Pathogenic	PS2, PS3, PS4_Supporting, PM1, PM2, PM5, PP3	*L, HA, ID*		
8	F	p.(Arg361His)	c.1082 G>A	MS	Pathogenic	PS4, PM1, PM2, PM5, PP3	*M/A, NS, ID*	-7, +8	BCOR
1	N/A	p.(Arg362_Asn365 del) [R361del4]	c.1084_1095del [c.1083_1094del12]	DEL	Likely Pathogenic	PS4_Supporting, PM2, PM4_Strong	*M/A, ID, HA*	-7, +21, +8	ASXL1
16	N/A	p.(Arg362*)	c.1084C>T	NS	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A, ID, L, HA*	-7, add +8 -20q	
2	F	p.(Arg362Pro)	c.1085G>C	MS	Likely Pathogenic	PS4_Supporting, PM1, PM2, PP3	*M/A, ID, L*	-7 +8	
1	N/A	p.(Asp367Thrfs*20)	c.1099del	FS	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A, HA, ID*		
3	F	p.(Asp367Glyfs*17)	c.1099dup [c.1099insG]	FS	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A, ID, HA*	NK	ASXL1
1	N/A	p.(Pro368Argfs*15)	c.1103_1104del	FS	Pathogenic	PVS1, PS4_Supporting, PM2	*NS*		
1	N/A	p.(Cys370Trp)	c.1110C>G	MS	Likely Pathogenic	PS4_Supporting, PM1, PM2, PP3	*M/A*	-7	
4	N/A	p.(Asn371Lys)	c.1113C>A	MS	Pathogenic	PS1, PS4, PM1, PM2, PP3	*MA, L, ID*	-7 +8	
2	N/A	p.(Asn371Lys)	c.1113C>G	MS	Pathogenic	PS1, PS4_Moderate, PM1, PM2, PP3	*M/A, HA,ID*	-7, +mar	ASXL1
1	N/A	p.(Asn371Lysfs*16)	c.1113del	FS	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A*	-7, +8	
6	N/A	p.(Ala372Thr)	c.1114G > A	MS	Likely Pathogenic	PS4, PM1, PM2, PP3	*M/A, ID, L*	-7 , +15, +20 t(11;19), +8	DNMT3A
3	F	p.(Cys373_Tyr377del) [C373del5]	c.1117_1131del [c.1116_1130del15]	DEL	Likely Pathogenic	PS4_Supporting, PM2, PM4_Strong*	*M/A, ID*	-7	
1	N/A	p.(Cys373Tyr)	c.1118G>A	MS	Likely Pathogenic	PS4_Supporting, PM1, PM2, PM5, PP3	*M/A, ID, L*	+1 -15	
1	N/A	p.(Cys373Arg)	c.1117T>C	MS	Likely Pathogenic	PS3_Supporting, PS4_Supporting, PM1, PM2, PM5, PP3	*L, M/A, ID*	-7	
1	N/A	p.(Leu375Val), p.?	c.1123C>G in-cis with 355 bp of GATA2 locus (part of last intron & exon) that has been duplicated and	MS, INS	Likely Pathogenic	PM1, PM2, PM5, PP3	*M/A*	NK	None

			inserted into the last exon						
3	N/A	p.(Leu375Phe)	c.1123C>T	MS	Likely Pathogenic	PS4_Moderate, PM1, PM2, PM5, PP3	*M/A, ID, L, HA*	-7, +8, +20	NRAS
2	D	p.(Leu375Profs*12)	c.1124del	FS	Pathogenic	PVS1, PS2, PS4_Supporting, PM2	*M/A*	-7	
1	D	p.(Tyr376*)	c.1128C>G	NS	Pathogenic	PVS1, PS2, PS4_Supporting, PM2	*M/A*	NK	
1	N/A	p.(Tyr377Asp)	c.1129T>G	MS	Likely Pathogenic	PS4_Supporting, PM1, PM2, PP3	*ID, HA*		
1	N/A	p.(Lys378*)	c.1132A>T	NS	Pathogenic	PVS1, PS4_Supporting, PM2	*HA, ID*	hyperdiploidy (>80 chr), +8	
1	N/A	p.(Asn381Metfs*6)	c.1142del	FS	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A, ID*	NK	
1	D	p.? [S340_N381del, V382Gfs*23 or N381_V382ins41]	c.1143+2T>A	SPL	Pathogenic	PVS1, PS2, PS4_Supporting, PM2	*M/A, ID, HA*	+8	
3	D	p.? [N381fs*20]	c.1143+5G>A	SPL	Likely Pathogenic	PS3, PS4_Moderate, PM2, PM6_Supporting	*ID, M/A*		
1	N/A	p.? [N381fs*]	c.1143+200_1198 del	DEL	Pathogenic	PVS1, PS4_Supporting, PM2	*HA, ID, L*	NK	ASXL1
3	F	p.(Pro385Gln)	c.1154C>A	MS	Likely Pathogenic	PS4_Supporting, PM1, PM2, PP3	*M/A, ID, L, T-ALL*	-7	
1	N/A	p.(Thr387Asn)	c.1160C>A	MS	Likely Pathogenic	PS4_Supporting, PM1, PM2, PP3	*M/A*	+8	SETBP1 ASXL1 RUNX1
2	F	p.(Met388Val)	c.1162A>G	MS	Likely Pathogenic	PS4_Supporting, PM1, PM2, PM5, PP3	*M/A, HA, ID*	-7 +8	
5	F	p.(Met388Thr)	c.1163T>C	MS	Likely Pathogenic	PS4_Supporting, PM1, PM2, PM5, PP3	*ID, HA*		
1	N/A	p.(Lys390Glu)	c.1168A>G	MS	Likely Pathogenic	PS4_Moderate, PM1, PM2, PP3	*M/A*	der(1;7) add	
1	N/A	p.(Glu391Glyfs*85)	c.1172_1175del	FS	Pathogenic	PVS1, PS4_Supporting, PM2	*ID, HA*		
12	F	p.(Arg396Trp)	c.1186C>T	MS	Pathogenic	PS4, PM1, PM2, PM5, PP3	*M/A, ID, L, HA*	+8, +mar, -7, -21	
23	N/A	p.(Arg396Gln)	c.1187G>A	MS	Pathogenic	PS2, PS3, PS4, PM1, PM2, PM5, PP1_Moderate, PP3	*M/A, ID, L, HA*	+8, -7, +11, der(1;16), add	STAG2 BCOR FANCA ASXL1
3	F	p.(Arg396Leu)	c.1187G>T	MS	Likely Pathogenic	PS3_Supporting, PS4_Supporting, PM1, PM2, PM5, PP1_Supporting, PP3	*M/A, HA, ID*		
22	F	p.(Arg398Trp)	c.1192C>T	MS	Pathogenic	PS3, PS4, PM1, PM2, PM5, PP1_Moderate, PP3	*M/A, ID, HA, CMML, JMML, L*	+1, -7, +8, -X	ASXL1 TP53, MLL ASXL1
4	F	p.(Arg398Gln)	c.1193G>A	MS	Likely Pathogenic	PS4, PM1, PM5, PP3	*M/A, ID*	-7, +8	
1	N/A	p.(Lys406Serfs*77)	c.1200_1216dup [c.1216_1217ins17]	FS	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A*	-7	
1	N/A	p. (Phe428Leufs*108)	c.1281dup [c.1281_1282insC]	FS	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A*		NRAS BOD1L CDH23 SETBP1 SF3A1 SF3B1
1	N/A	p.(His442Glnfs*95)	c.1322_1325dup	FS	Likely pathogenic	PVS1_Moderate, PS4_Supporting, PM2	*M/A*		CEBPA WT1
8	N/A	p.(Ser447Arg)	c.1339A>C	MS	Pathogenic	PS1, PS4, PM2, PP1_Moderate, PP3	*M/A, ID, HA*	+8	ASXL1
2	N/A	p.(Ser447Arg)	c.1341C>A	MS	Likely pathogenic	PS1, PS4_Moderate, PM2	*M/A*	-7	
1	N/A	Deletion (whole protein)		DEL	Pathogenic	PVS1, PS4_Supporting, PM2	*L, M/A, ID, DF, NS*	+21	
1	N/A	Deletion (whole protein)		DEL	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A, ID, DF, NS, HA*		
1	N/A	Deletion (whole protein)		DEL	Pathogenic	PVS1, PS4_Supporting, PM2	*HA, ID*		
1	N/A	Deletion (whole protein)		DEL	Pathogenic	PVS1, PS4_Supporting, PM2	*ID, HA*		
2	N/A	Deletion (whole protein)	c.1-?_1443+?del	DEL	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A, ID*	NK	
1	N/A	Deletion (whole protein)		DEL	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A, ID, DF, NS, HA*	-7	
1	N/A	Deletion (whole protein)		DEL	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A, HA, ID, L*	-7, +8	

1	N/A	Deletion (whole protein)		DEL	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A, HA, ID*		
1	N/A	Deletion (whole protein)		DEL	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A*	-7	
1	N/A	del3q21		DEL	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A, ID, NS*	del3q21, -7	
1	N/A	Deletion		DEL	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A, HA, ID*	45,XY,-7[7]/ 46,XY,-7,+mar[10]/ 46,XY[8]	
1	N/A	Deletion		DEL	Pathogenic	PVS1, PS4_Supporting, PM2	*HA, ID*		
1	F	Delete ATG start codon		DEL	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A*	t(2;12)(p21;p13)	CEBPA NPM1 STAG2 NRAS
3	F	p.? (M1del290)	c.-45-155_871+527del	DEL	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A, ID, L, HA*	NK	
1	N/A	p.?		DEL	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A, HA*		
1	F	p.?		DEL	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A*	-7	
1	F	del ZF2 & C-terminus	c.1018-?	DEL	Pathogenic	PVS1, PS4_Supporting, PM2	*M/A, ID*		
1	N/A	del ZF2 & C-terminus	c.1018-?_1443+? del	DEL	Pathogenic	PVS1, PS4_Supporting, PM2	*ID, M/A*		