Somatic variant profiling in chronic phase pediatric chronic myeloid leukemia

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Table S1. Panel including 148 leukemia-associated genes / gene regions

Gene	Exon
ABL1	all
ADGRV1 (GPR98)	all
ANKRD26	all with 5´UTR
ARID1A	all
ASXL1	13, 14
ASXL2	all
ATM	all
ATRX	all
BCL2	all
BCOR	all
BCORL1	all
BIRC3	6-9
BRAF	11 - 16
BRCC3	all, without exon 3
BTK	14 - 16
CALR	9
CARD11	4 - 10
CBL	8,9
CCND1	1
CCND3	5
CD58	1 - 3
CD79A	all
CD79B	all
CDH23	all
CDKN2A	all
CEBPA	all
CHD2	all
CREBBP	all
CSF3R	all
CSNK1A1	all
CTCF	all
CUX1	all, without exon 1 und 24
CXCR4	all
DDX3X	all
DDX41	all
DDX54	all
DHX29	all
DIS3	all
DNM2	all
DNMT3A	all
EGR2	2
EP300	all
ETNK1	5, 6, 7, 8, 9
ETV6	all
EZH2	all
FANCL	all
FAS	all
FAT4	all

FBXW7	8 - 12
FLT3	all, without exon 1
FOXO1	all
FOXP1	16 - 18
GATA1	2,3
GATA2	3, 4, 5, 6, 7
GNA13	all
GNAS	1, 8, 9
GNB1	all
HRAS	all
ID3	all
IDH1	all
IDH2	4
IGLL5	1
IKBKB	7
IKZF1	all
IKZF3	5
IL2RG	all
IL7R	5 + 6
IRF4	all
JAK1	14 -17
JAK2	12, 14, 16, 20, 21, 23
JAK3	11 - 16, 19, 20, 21
KDM5A	all
KDM6A	all
KIT	1-3, 8-13, 16-18, 20-21
KLF2	all
KLHL6	all
KMT2D	all
KRAS	2, 3, 4, 5
LRP1B	all
LUC7L2	all
MAP2K1	2, 3
MECOM	all
MEF2B	all
MPL	10
MYC	all
MYD88	all
NF1	all
NFKBIE	1
NOTCH1	26 - 34
NOTCH2	26 - 34
NPM1	12
NRAS	2,3
NSD2 (WHSC1)	20, 21
PAX5	all
PHF6	all
PIGA	all
PLCG2	19, 20, 24
POT1	all

PPM1D	all
PRDM1	all
PRPF8	all
PTEN	5, 7
PTPN1	all with 5'UTR
PTPN11	3, 4, 7, 8, 12,13
RAD21	all
RB1	all
RHOA	all
RPS15	all
RUNX1	all
SAMD9	all
SAMD9L	
SAMHD1	all
SETBP1	6
SETD2	all
SF1	all, without exon 1
SF3A1	all
SF3B1	13 - 16
SGK1	all
SH2B3	2
SMARCA4	all
SMC1A	all
SMC3	all
SOCS1	all
SOS1	all
SRSF2	all
STAG1	all
STAG2	all
STAT3	20 + 21
STAT5B	15 - 18
STAT6	9 - 14
SUZ12	14 - 16
TBL1XR1	all
TCF3	17 - 19
TENT5C (FAM46C)	all
TET2	all
TLR2	all
TNFAIP3	all
TNFRSF14	all
TP53	all
TRAF3	all
U2AF1	all
U2AF2	all
UBR5	58
WT1	all
XPO1	12 - 18
ZBTB7A	all, without exon 2
ZMYM3	all
ZRSR2	all

Table S2. Classification and detailed information concerning pathogenic variants found in n=14 individuals

[a]UPN	Gene	Variant	[b]VAF% (depth)	In-Silico Data	Database research	[c]Variant origin	Classification (according to ACMG (Richards et al., 2015))
1	*ASXL1*	c.2644C>T p.(Gln882*)	5 (2068)	no further information	no further information	Somatic	Likely pathogenic (PVS1_strong, PM2)
2	*ASXL1*	c.2404G>T p.(Glu802*)	5 (2286)	no further information	no further information	somatic	likely pathogenic (PVS1_strong, PM2)
3	*ASXL1*	c.3824C>G p.(Ser1275*)	40 (2255)	no further information	COSMIC: COSM1291094	somatic	Likely pathogenic (PVS1_strong, PM2)
4	*ASXL1*	c.4127dup p.(Pro1377Serfs*3)	31 (2036)	no further information	ClinVar: VCV000521420.5 (Submitter: Invitae, Ambry Genetics è pathogenic)	somatic	Likely pathogenic (PVS1_strong, PM2)
5	*ASXL1*	c.1900_1922del p.(Glu635Argfs*15)	5 (1204)	no further information	COSMIC: COSM36165	somatic	Likely pathogenic (PVS1_strong, PM2, PP5_moderate)
	ASXL1	c.2077C>T p.(Arg693*)	5 (1728)	no further information	COSMIC: COSM51388	somatic	Likely pathogenic (PVS1_strong, PM2, PP5_moderate)
6	*ASXL1*	c.2242C>T p.(Gln748*)	40 (1687)	no further information	COSMIC: COSM307766	Somatic	Likely pathogenic (PVS1_strong, PM2, PP5)
7	*ASXL2*	c.2782dup p.(Thr928Asnfs*18)	11 (1000)	no further information	no further information	Somatic	Likely pathogenic (PVS1_strong, PM2)
8	*BCOR*	c.1005dup p.(Ser336Leufs*45)	8 (1624)	no further information	no further information	Somatic	Likely pathogenic (PVS1, PM2)
9	*GATA2*	c.1160_1165del p.(Thr387_Met388del)	5 (1910)	no further information	ClinVar: VCV001315540.1 (Submitter: GeneDx -> VUS); COSMIC: COSM5944129	Somatic	Likely pathogenic (PM1, PM2, PM4)
10	*IKZF1*	c.826_827del p.(Ser276Leufs*212)	34 (2466)	no further information	no further information		Likely pathogenic (PVS1_strong, PM2)
11	*KDM6A*	c.3334_3338dup p.(Ala1115Tyr*7)	5 (841)	no further information	no further information	somatic	Likely pathogenic (PVS1, PM2)
12	*KMT2D*	c.14710C>T p.(Arg4904*)	14 (930)	no further information	ClinVar: VCV000094176.11 (Submitter: Invitae, GeneDx è pathogenic)	Somatic	Pathogenic (PVS1, PM2, PP5)
13	*KMT2D*	c.5007del p.(Val1670Serfs*52)	5 (793)	no further information	COSMIC: COSM6979957	Somatic	Likely pathogenic (PVS1, PM2)
14	*TET2*	c.4075C>T p.(Arg1359Cys)	8 (1135)	REVEL: 0,23	COSMIC: COSM41649	somatic	Likely pathogenic (PM1, PM2, PP5_moderate)

[a]UPN = unique patient number; [b]VAF = variant allele frequency; [c]Variant origin assessed in MR2 or better