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Integrated genomic analysis to reduce chromosomal analysis for the diagnosis of pediatric hematologic malignancies: addressing the shortage of cytogenetic technologists

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Contributions: DL, MML and YZ conceived and designed the study, analyzed the data, and wrote the initial version of the manuscript. All authors (DL, SKT, GW, SRR, KMB, HN, LW, DMW, BT, LFS, EM, VP, ML, MEP, STH, MML, YZ)) interpreted the results and reviewed and contributed to the final version of the manuscript.

Chromosomal analysis (CA) has been part of the standard care for patients with hematological malignancies since Dr. Nowell and colleague discovered the Philadelphia chromosome in chronic myelogenous leukemia in 1961¹. CA examines cancer-associated numerical and structural abnormalities at the single-cell level and has played a significant role in leukemia diagnosis, risk stratification, and treatment selection². However, the resolution of CA in detecting chromosomal rearrangements and copy number variations (CNVs) is limited. Newer molecular technologies with much higher resolution and scalability, such as fluorescence *in situ* hybridization (FISH), chromosome microarray (CMA), and next generation sequencing (NGS), have been developed and are now widely implemented to detect genomic aberrations in cancer (herein collectively referred to as integrated genomic analysis in this study)³⁻⁷. CA requires both the knowledge of cancer and recognition of related aberration patterns. It usually takes one to two years of post-baccalaureate training to become a cytogenetic technologist and several more years to be an experienced cancer cytogenetic technologist. As large numbers of cytogenetic technologists retire, we are seeing a nationwide shortage of cytogenetic technologists. The situation is getting worse since pandemic as less people were entering the cytogenetic specialty⁸⁻¹⁰. Given this workforce challenge, we retrospectively reviewed 201 pediatric hematologic malignancy cases at our institution that underwent CA, FISH, CMA, and NGS tests to evaluate if integrated genomic analyses can redefine the need for conventional cytogenetics without impacting the clinical care of patients.

This study was performed in accordance with the ethical standards detailed in the Declaration of Helsinki and under the oversight of the Institutional Review Board of Children's Hospital of Philadelphia. Genetic testing results and patient records of 201 consecutive children, adolescents, and young adults with leukemia or lymphoma enrolled in the Children's Oncology Group (COG) clinical trials were reviewed (**Supplementary Table 1**). In addition to CA and targeted FISH assays required for COG participation, we performed CMA and NGS analyses in our CLIA-certified clinical laboratory. The CMA utilized the Illumina genome-wide SNP array (Illumina, San Diego, CA). Our customized comprehensive hematologic malignancy NGS panel (COHEM) include the DNA panel that interrogates 118 cancer genes known to be associated with hematologic malignancies for single nucleotide variants (SNVs), small insertions and deletions (indels), and CNVs, and an RNA panel that targets 117 cancer genes and over 700 exons for known and novel fusions by using the Anchored Multiplex PCR technology (ArcherDX, Boulder, CO)^{11,12}. The identified variants were classified according to established guidelines¹³.

The demographics of the cohort are detailed in **Figure 1A and 1B**. The median patient age was seven years (range 1 to 24), and 113/201 (56.2%) were male. The most common diagnosis was B-lymphoblastic leukemia/lymphoma (B-ALL/LBL; n=135, 67.2%), followed by acute myeloid leukemia (AML)/myeloid sarcoma (n=40, 19.9%), T-lymphoblastic leukemia/lymphoma (T-ALL/LBL; n=21, 10.4%), chronic myeloid leukemia (CML; n=3, 1.5%), mixed phenotype acute leukemia (MPAL; n = 1, 0.05%), and acute undifferentiated leukemia (AUL, n = 1, 0.05%) (**Figure 1C**). All patients underwent CA and FISH testing, however, 22 patients (10.9%) had insufficient cell growth *in vitro*, preventing informative CA (**Figure 2**). The majority of cases

underwent successful COHEM (200/201, 99.5%) and CMA (184/201, 91.5%) testing (**Supplementary Table 1**).

At least one clinically significant variant was detected in each case (**Supplementary Table 1**). Among 179 patients with CA results, 26 (14.5%) showed a normal karyotype (**Supplementary Table 1; Figure 2**). However, clinically significant genomic aberrations, including translocations, CNVs and fusions, were identified in 17/26 (65.4%) cases via FISH, CMA, and/or COHEM testing, likely reflecting the growth advantage of normal cells over tumor cells in culture or limited CA resolution.

Among the remaining 153 cases, CA revealed an additional finding with defined clinical significance but without impacting risk stratification or therapy (referred to as category 1) that was not detected by other methodologies in only one patient with T-ALL (0.7%; **Figure 2; Supplementary Table 1**, case #12). In this case, karyotyping identified a balanced translocation between chromosomes 11 and 14 with possible breakpoints at 11p1?3 and 14q11.2 (46,XY,t(11;14)(p1?3;q11.2)) in 12 out of 20 metaphase cells. This finding was confirmed by metaphase FISH using a break-apart probe set for TCR Alpha/Delta (TRA/D) at 14q11.2 (**Supplementary Table 1**). Rearrangements involving *TRA* or *TRD*, which encode the T-cell receptor α and δ chains, respectively, have been found in 5-10% of T-ALL cases^{14,15}. We have now included this FISH probe set in our T-ALL FISH panel to ensure the detection of these important fusions.

In 45 cases (29.4%, 45/153), CA revealed additional findings, mostly providing chromosome structural aberration information to confirm or augment results of FISH, CMA, and COHEM testing, although such data are not currently used for diagnosis, prognosis, or treatment selection (referred to as Category 2; **Figure 2; Supplementary Table 1**). These results included non-recurrent complex low-level subclonal structural variations (n=1), derivative chromosomes (n=21), isochromosomes (n=7), dicentric chromosomes (n=2), balanced translocations (n=8), three-way translocations (n=6), an inversion (n=1), an insertion (n=1), a gain of whole chromosome (n=1), complex rearrangements involving multiple chromosomes (n=4), and (near) tetraploid genomes (n=2) (**Figure 2; Supplementary Table 1**). In most cases with a derivative chromosome, isochromosome, dicentric chromosome, balanced translocation, or three-way translocation, results from concurrent FISH, CMA, and/or COHEM analyses also suggested such abnormalities (**Supplementary Table 1**). For example, CMA identified loss of 8p23.3p11.21, 8p11.21, and 12p13.33p11.22 in patient #19, consistent with chromosome analysis showing a dicentric chromosome 45,XY,dic(8;12)(p11.21;p11.22) (**Supplementary Table 1**). Similarly, loss of 7p together with gain of 7q identified by CMA in patient #110 was consistent with an isochromosome i(7)(q10) on karyotype (**Supplementary Table 1**). In patient #56, a *PICALM::MLL10* fusion identified by COHEM testing, clarified the finding of a balanced translocation between chromosomes 10 and 11 at possible breakpoints of 10p12 and 11q14.2, respectively. In patient #60, a derivative chromosome 16 [der(16)t(X;16)(p11.2;p13.1)] with a loss of 16pter->16p13.1 and a duplication of Xpter->Xp11.2 structurally resembled a ~44.7 Mb gain of chromosome Xp22.33p11.3 and a ~12.9 Mb loss of 16p13.3p13.12 identified by CMA.

These results emphasize the importance of cell-based assay CA in characterizing balanced and certain unbalanced chromosomal aberrations when suspected by FISH, CMA, and NGS assay data. Although none of these structural abnormalities identified in the 45 cases had defined clinical significance, CA facilitated improved knowledge of these genetic alterations and could potentially lead to discovery of new disease-defining aberrations, especially in rare leukemia subtypes.

Our collective data suggest that CA was not necessary for cancer genetic diagnostics in more than two-thirds of our pediatric leukemia/lymphoma cohort, which could allow cytogenetic technologists to focus critical efforts on a smaller number of CA cases and expedite FISH assays. Based on the results of this study, our laboratory implemented a new policy in which we do not routinely run CA for hematologic malignancy cases unless these data are required for enrollment on a specific clinical trial; however, efficient chromosomal preparations are performed for all cases, so that CA can be subsequently added if indicated by results of FISH, CMA, and/or NGS testing. We continue to perform CMA on all leukemia and lymphoma cases, and TCA/D FISH is added when T-ALL is suspected or confirmed. In the first six months after implementing the new CA policy in April 2024, we have reduced our average CA volume by 43% which resulted in reduced average turnaround time (TAT) for CA from 20 days to 11 days (**Figure 2**) for cases in which CA is required. Importantly, this reduction of CA volume has also led to decreased average TAT for cancer FISH analysis from 6.8 days to 4.8 days.

In summary, our collaborative study demonstrates that while FISH, CMA, and NGS testing cover a broad spectrum of genomic alterations and are scalable, CA remains a valuable approach to identify structural configuration of chromosomes at a single-cell level and cannot be completely replaced in clinical laboratories for pediatric hematological malignancies at this time. Nonetheless, integrated genomic analysis can effectively reduce the CA volume for a large percentage of routine cases and may, in turn, help to mitigate the current shortage of cytogenetic technologists without compromising patient care in pediatric hematological malignancies. Importantly, we recommend proactive chromosomal preparations for all patients with triage to CA testing for a subset of cases as indicated by other test results. Ongoing developments in machine learning and artificial intelligence-based automatic karyotyping systems are expected to provide a better solution for comprehensive chromosomal analysis in hematological malignancies in the future.

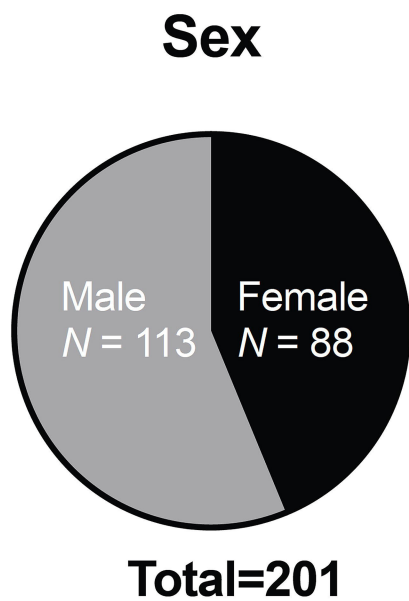
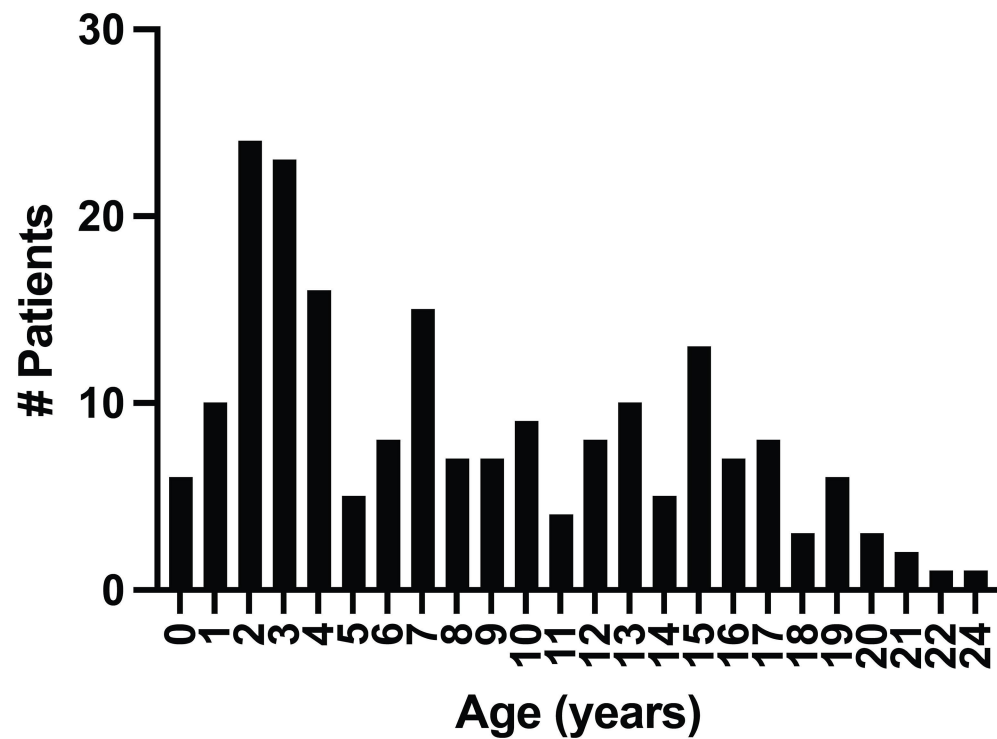
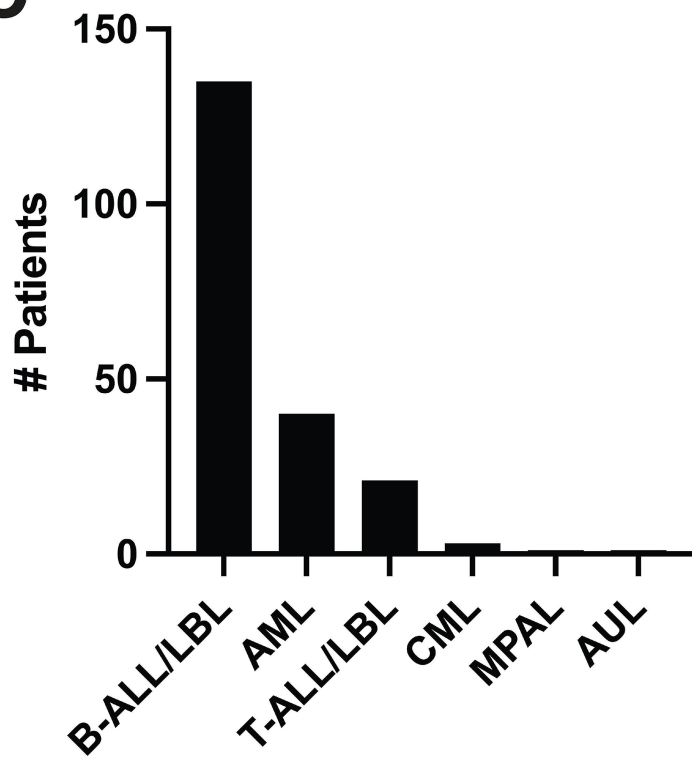
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Figure Legends

Figure 1. Overview of study cohort. (A) Sex and (B) Age (at sampling) characteristics of children, adolescents, and young adults in the study cohort (n=201); (C) Number and type of leukemia and lymphoma diagnoses included in study cohort.

Figure 2. Study workflow

A**B****C**

Total 201 patients
analyzed 2019-2024

22 patients with insufficient
cell growth

179 patients with
karyotyping

26 patients with normal
chromosomes

153 patients with
abnormal karyotype
results

karyotyping revealed an
additional finding **with defined
clinical significance** in one
patient with T-ALL (0.7%)
Category 1

karyotyping revealed additional
findings **without defined
clinical significance** in 45
patients (29.4%)
Category 2

karyotyping revealed **no
additional findings** in 107
patients (69.9%)
Category 3

Solution:

Add TCR Alpha/Delta
FISH probe set in
FISH test; if positive,
may consider adding
karyotyping

May consider adding karyotyping
when suggested structural
chromosomal abnormalities by other
methods are of clinical significance

Not applicable

Table S1. Genetic testing results and patient demographics of 201 consecutive children, adolescents, and young adults with hematological malignancies

Patient #	Sex	Age at ordering	Order date	Indication	Karyotype	FISH Results	Concordance CNV results between Cyto and Array	Additional clinical significant findings from array compared to karyotype	Concordance between NGS and Cyto	Additional significant findings in NGS compared to karyotype	Findings unique from cytogenetics
1	M	3	3/26/2024	B-ALL	46,XY,(12;21)(p13;q22)[5].ish t(12;21)(RUNX1+,ETV6-;RUNX1+,ETV6+)[46,XY[14]	nuc ish(CEP4,CEP10)x2[200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2A)x2[200] nuc ish(ETV6x2,RUNX1x3)[ETV6 con RUNX1x1][186/200] nuc ish(CRLF2x2)[200]	None	partial losses of 4q, 9q involving PAX5, and 12q involving BTG1	ETV6:RUNX1	Loss of exons 2 through 6 of the PAX5 (NM_016734.2) gene on chromosome 9p DOT1L (NM_032482.3), c.964-2A>G (p.?) KRAS (NM_033860.4), c.350>A (p.Gly12Asp) NRAS (NM_002524.5), c.38G>A (p.Gly13Asp)	NA (normal karyotype*)
2	M	13	2/9/2024	B-ALL	No growth	nuc ish(CEP4,CEP10)x2[200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2A)x2[200] nuc ish(ETV6,RUNX1)x2[200] nuc ish(CRLF2x2)[200] nuc ish(ABL1x2)[200] nuc ish(PDGFRBx2)[200]	N/A, Cyto no growth	segmental chromosome losses involving chromosome arms 2p, 8q, 9p, 12p, and 17q, as well as copy number gain of 3p26.3p13	N/A, Cyto no growth	PAX5:FOXP1; FLT3 (NM_004119.3), c.2028C>A (p.Asn676Lys); gain of partial 3p (including MYD88 and SETD2), loss of partial 9p (including JAK2, partial PAX5, and biallelic loss of CDKN2A/B), and loss of partial 17q (NF1, SUZ12, IKZF3)	NA (no growth)
3	M	8	2/7/2024	B-ALL	57~58,XY,+X,+4,+5,+6,del(6)(q13),der(6)(t(2;6)(q11.2;q13),+8,+10,+12,+14,+del(14)(q11.2),+17,+21,+21[cp15]/46,XY[5]	nuc ish(CEP4,CEP10)x3[198/200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2A)x2[200] nuc ish(ETV6x3,RUNX1x4)[186/200] nuc ish(ETV6x2,RUNX1x3)[180/200] nuc ish(CEP8,MYC,IGH)x3[198/200] nuc ish(CRLF2x3)[196/200]	a hyperdiploid genome with gains of whole chromosomes 4, 5, 8, 10, 12, 21, and X, and multiple segmental copy number variants and copy number neutral loss of heterozygosity (cnLOH) involving chromosomes 6, 14, and 17	multiple segmental copy number variants and loss of heterozygosity, including homozygous loss of CDKN2A/B on chromosome 9p and cnLOH chromosome 11.	A high hyperdiploid genome with gains of whole chromosomes 4, 5, 8, 12, 21, and X.	homozygous loss of the CDKN2A/B genes on chromosome 9p as well as copy neutral loss of heterozygosity (cnLOH) across chromosome 11. KRAS (NM_033860.4), c.35G>T (p.Gly12Val) NRAS (NM_002524.5), c.183A>T (p.Gln61His)	None (Category 3)
4	F	7	1/25/2024	B-ALL	No growth	nuc ish(CEP4,CEP10)x3[182/200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2A)x2[200] nuc ish(ETV6x2,RUNX1x4)[180/200] nuc ish(CEP8x2,MYC,x2,IGHx3)[180/200] nuc ish(CRLF2x2)[200]	N/A, Cyto no growth	three copies of chromosomes 4, 6, 14, 17, and 18 and four copies of chromosome 21, consistent with a high hyperdiploid genome	N/A, Cyto no growth	A hyperdiploid genome with multiple chromosome gains including gains of chromosomes 4, 6, 10, 14, 17, 18, and 21; copy neutral loss of heterozygosity across multiple chromosomes, a complex CNV involving cnLOH with partial loss of 19p. NRAS (NM_002524.5), c.182A>T (p.Gln61Leu)	NA (no growth)
5	F	5	1/19/2024	B-ALL	52,XX,+X,+4,+9,+14,+21,+21[cp14]/46,XX[6]	nuc ish(CEP4x3,CEP10x3)[190/200] nuc ish(ABL1x3,BCR)x2[164/200] nuc ish(KMT2A)x2[200] nuc ish(ETV6x2,RUNX1x4)[194/200] nuc ish(CEP8x2,MYC,x2,IGHx3)[60/200] nuc ish(CRLF2x3)[190/200]	trisomy for chromosomes 4, 9, and X and tetrasomy for chromosome 21 (with two extra copies), consistent with a high hyperdiploid genome (51 chromosomes total)	copy neutral loss of heterozygosity (cnLOH) of 12q24.11q24.33, gain of 14q11.2q23.1, gain with LOH of 14q23.1q32.33, and loss of partial CREBBP gene.	gain of whole chromosomes 4, 9, 21 (with two extra copies), and X, consistent with a high hyperdiploid	P2RY8:~CRLF2: loss of partial 16p (involving partial CREBBP gene), and complex rearrangement of chromosome 14 (~3x gain of TINF2 followed by cnLOH of BCL118); CREBBP (NM_004380.3), c.5783dup (p.Pro1929Alafs*37) NRAS (NM_002524.5), c.38G>A (p.Gly13Asp) SETD2 (NM_014159.7), c.7490_7491insGCC (p.Arg2498Profs*7) SETD2 (NM_014159.7), c.6973C>T (p.Gln2325*)	None (Category 3)
6	M	9	1/10/2024	AML	46,XY,t(15;17)(q24.1;q21.2)[9]/46,XY[11]	nuc ish(PML,RARA)x3[PML con RARAx2][120/200] nuc ish(PMLx2,RARAx3)[PML con RARAx1][60/200]	None, arr(X,Y)x1,(1-22)x2	None arr(X,Y)x1,(1-22)x2	PML:RARA	None (Category 3)	
7	F	0	1/8/2024	B-ALL	No growth	nuc ish(CEP4,CEP10)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(3'KMT2Ax1,5'KMT2Ax2)[3'KMT2A con 5'KMT2Ax1][82/200] nuc ish(ETV6,RUNX1)x2[200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(CRLF2x2)[200]	N/A, Cyto no growth	a ~492kb loss involving KMT2A on chromosome 11q23.3 (KMT2A exons 9-36)	N/A, Cyto no growth	KMT2A:MLLT1 and loss of partial chromosome 11q involving part of the KMT2A gene	NA (no growth)
8	M	10	3/15/2024	B-ALL	46~48,XY,7del(4)(p172),add(7)(q34),add(12)(p12.1),del(12)(p12.2),p13.31,+18,+del(22)(q13.2),+mar[cp9]/46,XY[11]	nuc ish(CEP4,CEP10)x2[200] nuc ish(ABL1x2,BCR)x3[54/200] nuc ish(KMT2A)x2[200] nuc ish(ETV6x1,RUNX1x2)[186/200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(CRLF2x2)[200] nuc ish(ABL1x2)[200] nuc ish(ABL2x2)[200] nuc ish(PDGFRBx2)[200]	partial losses of 12p involving ETV6, gain of chromosome 18	loss of 8q, 16p, 17p/q and 20q, cnLOH of partial 9p	loss of partial 12p (containing ETV6)	EZH2 (NM_004456.5), c.2187dup (p.Asp730*) RAD21 (NM_006265.3), c.1432C>T (p.Arg478*) loss of partial 8q (RAD21) and partial 17q (NF1 and SUZ12)	complex low level subclonal structural variations (Category 2)
9	F	12	3/13/2024	AML	46,XX[20]	nuc ish(RUNX1T1,RUNX1)x2[200] nuc ish(KMT2A)x2[200] nuc ish(NUP98x2)[200] nuc ish(MYH11x3,CBFBx2)[50/200]	None, normal karyotype	loss of 9q21.12 and gain of 16p13.11	None, normal karyotype	CEBPA (NM_004364.5), c.68_78del (p.Pro23Glnfs*81) CEBPA (NM_004364.5), c.937_939del (p.Lys313del) CSF3R (NM_00760.4), c.1919C>A (p.Thr640Asn) WT1 (NM_024426.6), c.1151_1158dup (p.Ala387Tyrfs*70)	NA (normal karyotype)
10	M	15	2/20/2024	T-ALL	51,XY,+X,+8,+9,+10,der(16)dup(16)(p11.2),p13.3[del(16)(p13.3pter),t(17)(q10),+19][12]/46,XY[8]	nuc ish(RANBP1,7,X)X2[200] nuc ish(ABL1x1,BCR)x2[176/200] nuc ish(KMT2A)x2[200] nuc ish(ETV6,RUNX1)x2[200] nuc ish(CEP8x3,MYC,x3,IGHx2)[170/200] nuc ish(CRLF2x3)[174/200]	gain of whole chromosomes 8, 10, 19, and X, gain of 9p24.3q11 (~43Mb), 9q12q34.11 (~67Mb), 9q34.13q34.3 (~7Mb), 16p13.3p11.2 (~25Mb), and loss of 9q34.11q34.13 (~2Mb including the ABL1 gene), and 16p13.3 (~7Mb)	cnLOH of 17p (including the TP53 gene), a mosaic loss of 18p (18p11.3p11.1, 15Mb)	gains of whole chromosomes 8, 10, 19, and X, as well as multiple segmental gains and complex involving chromosomes 9, 16, and 17	SET:~NUP214 fusion JAK3 (NM_000215.4), c.1718C>T (p.Ala573Val) JAK3 (NM_000215.4), c.1533G>A (p.Met511Ile) JAK3 (NM_000215.4), c.2021T>C (p.Val674Ala) JAK3 (NM_000215.4), c.1688_1696del (p.Lys563_Cys565del) PHF6 (NM_032458.3), c.730-1G>A (p.?) TP53 (NM_000546.6), c.54_55del (p.Ser20Argfs*8) WT1 (NM_024426.6), c.1124_1125insATGGCCGACG (p.Val376Trpfs*17)	a derivative chromosome and an isochromosome (Category 2)

11	F	9	2/20/2024	B-ALL	47,XX,7del(2)(p11.2),+7,+10,add(12)(p12.2),t(12;21)(p13;q22),-13(cp4).ish add(12)(ETV6-),t(12;21)(RUNX1+,ETV6-);RUNX1+,ETV6+;/84-90,idem(cp11)/46,XX[5]	nuc ish(CEP4x2,CEP10x3)[22/200] (CEP4,CEP10)x4[72/200] (CEP4x4,CEP10x6)[100/200] nuc ish(ABL1x4,BCRv4)[80/200] (ABL1x4,BCRv5)[94/200] nuc ish(KMT2Ax3)[74/200] (KMT2Ax4)[86/200] nuc ish(ETV6x1,RUNX1x3)[ETV6 con RUNX1x1][20/200] (ETV6x2,RUNX1x4)[ETV6 con RUNX1x2][92/200] (ETV6x3,RUNX1x6)[ETV6 con RUNX1x3][84/200] nuc ish(CEP8,MYC,IGH)x4[162/200] nuc ish(CRLF2x4)[170/200]	gains of whole chromosomes 7, 9, 10, and 18 and losses of chromosome 13, several segmental losses leading to loss of CDKN2A/B and ETV6	losses on 4p12 (~393Kb) and 15q14q15.3 (~4.2Mb)	ETV6:RUNX1; gain of whole chromosomes 2, 7, 10, 16, 18, 20, 21, and 22, loss of whole chromosome 13, partial loss of 9p and 12p	None	None (Category 3)
12	M	11	2/20/2024	T-ALL	46,XY,t(11;14)(p13;q11.2)[12].ish t(11;14)(3'tcra/d+;5'tcra/d+;3'tcra/d)/46,XY[8]	nuc ish(RANBP17,TLX3)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6,RUNX1)x2[200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(CRLF2x2)[200]	None	an ~82 kilobase loss involving the STIL gene and part of the TAL1 gene, homozygous loss of the CDKN2A and CDKN2B genes on chromosome 9.	None	STIL:TAL1; loss of partial chromosome 1p including the upstream enhancer region of the TAL1 gene	Yes (Category 1)
13	F	17	2/7/2024	AML	46,XX,t(8;21)(q22;q22)[4]/47,idem, dup(17)(q21.31q24.1),+der(21)(8;21)(13).ish +der(21)(RUNX1+,RUNX1T1+);46,XX[3]	nuc ish(RUNX1T1,RUNX1x3)(RUNX1T1 con RUNX1x2)[88/200] (RUNX1T1,RUNX1)x4(RUNX1T1 con RUNX1x3)[96/200] nuc ish(KMT2Ax2)[200] nuc ish(NUP98x2)[200] nuc ish(MYH11,CFB8)x2[200]	copy number gain involving chromosomes 8q, 17q, and 21q.	None	RUNX1:RUNX1T1; gain of partial 8q (including RAD21), partial 17q (including CD79B), and partial 21q (including partial RUNX1)	RAD21 (NM_006265.3), c.1188dup (p.Val397Cysfs*6)	None (Category 3)
14	M	17	2/5/2024	T-ALL	46,XY,del(q11q22.31)[14]/46,XY[4]	nuc ish(RANBP17,TLX3)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6,RUNX1)x2[200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(CRLF2x2)[200] nuc ish(CEP4,CEP10)x2[200]	a 57Mb heterozygous loss of 6q12q22.31	segmental losses on chromosome 9p involving homozygous loss of CDKN2A/B and heterozygous loss of multiple genes including MLLT3 and JAK2, and a 38Mb gain of 13q22.2q34.	loss of partial 9p including homozygous loss of CDKN2A/B	NOTCH1 (NM_017617.5), c.4793_4795delinsCCT (p.Arg1598_Val1599delinsProLeu) NOTCH1 (NM_017617.5), c.4742_4743insCCTTTTAGTCTCGTCC (p.Pro1581_Pro1582insSerPheSerGlyPro) NOTCH1 (NM_017617.5), c.4745_4746insCCC (p.Pro1582dup) TAL1 (NM_003189.5), c.7404dup (p.?)	None (Category 3)
15	F	15	1/30/2024	B-ALL	58-59,XX,+x,+x,+1,+4,+6,+7,+10,+14,+14,+17,+18,+21,+21[cp3]/46,XX[17]	nuc ish(CEP4,CEP10)x3[100/200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6x2,RUNX1x4)[96/200] nuc ish(CEP8x2,MYCx2,IGHx4)[100/200] nuc ish(CRLF2x4)[94/200] nuc ish(ABL1x2)[200] nuc ish(ABL2x3)[20/200] nuc ish(PDGFRBx2)[200]	three copies of chromosomes 4, 6, 7, 10, 17, and 18, and four copies of chromosomes 14, 21, and X	None	whole chromosome gains (a total of ~58 chromosomes), including an estimated three copies of chromosomes 4, 6, 7, 10, 17, and 18, and four copies of chromosomes 14, 21, and X.	KMT2D (NM_003482.4), c.5269C>T (p.Arg1757*) PTPN11 (NM_002834.5), c.179G>T (p.Gly60Val)	None (Category 3)
16	M	3	1/5/2024	T-ALL	46,XY,del(9)(p21.2pter)[8]/46,XY[12]	nuc ish(RANBP17,TLX3)x2[200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6,RUNX1)x2[200] nuc ish(CRLF2x2)[200]	complex deletions on chromosome 9p involving loss of multiple genes including loss of a copy of JAK2 and MLLT3 and homozygous loss of CDKN2A/B	a ~246 kb gain involving the MYB gene on chromosome 6q23.3, a low-level mosaic deletion on 10q23.2-23.31 including PTEN, and loss of partial chromosome Y.	gain involving MYB on chromosome 6, loss involving JAK2 and homozygous loss of CDKN2A/B on chromosome 9, and loss involving PTEN on chromosome 10; SNV in ETS1	ETS1 (NM_005238.4), c.645_649dup (p.Asp217Glyfs*41)	None (Category 3)
17	M	15	1/8/2024	B-ALL	No growth	nuc ish(CEP4,CEP10)x3[50/200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6x2,RUNX1x4)[46/200] (ETV6x2,RUNX1x3)[8/200] nuc ish(CEP8,MYC,IGH)x3[28/200] (CEP8x2,MYCx2,IGHx3)[20/200] nuc ish(CRLF2x3)[44/200] nuc ish(ABL1x2)[200] nuc ish(ABL2x3)[22/200] nuc ish(PDGFRBx2)[200]	N/A, Cyto no growth	three copies of whole chromosomes 4, 6, 8, 10, 14, 17 and 18, four copies of chromosomes 21, and two copies of chromosome X (gain one extra copy)	N/A, Cyto no growth	a high hyperdiploid genome; subclonal P2R8:CRLF2; FLT3 (NM_004119.3), c.2039C>T (p.Ala680Val) FLT3 (NM_004119.3), c.2508_2510del (p.Ile836del)	NA (no growth)
18	M	13	10/24/2023	B-ALL	46,XY,del(9)(p13)[17]/46,idem,t(2;3)(p13;q32)[3]	nuc ish(CEP4,CEP10)x2[200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6,RUNX1)x2[200] nuc ish(CRLF2x2)[200] nuc ish(ABL1x2)[200] nuc ish(ABL2x2)[200] nuc ish(PDGFRBx2)[200]	segmental losses on chromosome 9p including homozygous loss of 9p21.3 involving CDKN2A/B	None	loss of chromosome 9p including homozygous loss of CDKN2A/B	loss of exons 2-11 of LEF1 on chromosome 4q NRAS (NM_002524.5), c.35G>C (p.Gly12Ala) PAX5 (NM_016734.3), c.1013-1G>T, (p.?)	None (Category 3)
19	M	4	12/28/2023	B-ALL	45,XY,del(8;12)(p11.21,p11.22),t(12;21)(p13;q22)[9].ish t(12;21)(RUNX1+,ETV6-);RUNX1+,ETV6+;/46,idem,+7(21)(q10)[6].ish t(21)(RUNX1+);46,XY[5]	nuc ish(CEP4,CEP10)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6x1,RUNX1x3)[ETV6 con RUNX1x1][100/200] (ETV6x1,RUNX1x4)[ETV6 con RUNX1x1][62/200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(3'CRLF2x2,5'CRLF2x1)(3'CRLF2 con 5'CRLF2x1)[164/200]	gain of whole chromosome 21, loss of 8p23.3p11.21, 8p11.21, 12p13.33p11.22	loss of 4q31.23, 5q31.3, 5q33.3, Xp22.33, Yp11.2, and criLOH of 14q11.2q32.33	ETV6:RUNX1; gain of chromosome 21, partial chromosome 12p	Loss of partial chromosome 5q, copy neutral LOH of chromosome 14q	a dicentric chromosome and an isochromosome (Category 2)
20	M	8	11/13/2023	B-ALL	46,XY,t(12;21)(p13;q22),add(22)(q13)[13].ish t(12;21)(RUNX1+,ETV6-);RUNX1+,ETV6+;/46,XY[7]	nuc ish(CEP4,CEP10)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6x1,RUNX1x3)[ETV6 con RUNX1x1][200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(CRLF2x2)[200] nuc ish(ABL1x2)[200] nuc ish(ABL2x2)[200] nuc ish(PDGFRBx2)[200]	None	gain of whole chromosome 10 and partial gain involving chromosome Xq, losses on chromosome 4, 6, 7, 12, 14, 20, and 22	ETV6:RUNX1	loss 4q, loss partial 12p, gain partial Xq KRAS (NM_033360.4), c.576G>C (p.Leu19Phe) KRAS (NM_033360.4), c.355G>C (p.Asp119His) NSD2 (NM_132335.4), c.2395G>A (p.Glu1099Lys) UBA2 (NM_005499.3), c.212C>G (p.Ser71*)	None (Category 3)

21	F	16	8/5/2023	B-ALL	56,XX,+X,+4,+10,+14,+14,+17,+18,+18,+21,+21[2]/57,idem,+X[13]/46,XX[5]	nuc ish(CEP4,CEP10)x3[198/200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6x2,RUNX1x4)[196/200] nuc ish(CEP8x2,MYCx2,IGHx4)[196/200] nuc ish(CRLF2x3)[30/200] nuc ish(CRLF2x3)[100/200] nuc ish(CRLF2x3)[68/200] nuc ish(ABL1x2)[200] nuc ish(ABL2x2)[200] nuc ish(PDGF8x2)[200]	gains of whole chromosomes X, 4, 10, 17, 18, and 21. trisomies (three copies) X, 10, and 17, and tetrasomies (four copies) X, 18, and 21.	None	gain of one or two copies of whole chromosomes 4, 10, 14, 17, 18, 21 and X (~57 chromosomes)	CREBBP (NM_004380.3), c.3836+1G>A (p.?) NRAS (NM_002524.5), c.38G>T (p.Gly13Val)	None (Category 3)
22	M	2	8/3/2023	AML	46,XY,t(9;11)[p21;q23][17]	nuc ish(KMT2Ax2)[5'KMT2A sep 3'KMT2Ax1][42/62]	None, arr(X,Y)x1,(1-22)x2	None, arr(X,Y)x1,(1-22)x2	KMT2A:MLL3	PTPN11 (NM_002834.4), c.226G>A (p.Glu76Val) FLT3 (NM_004119.3), c.1323_1328dup (p.Leu442_Ala443dup)	None (Category 3)
23	F	2	8/3/2023	B-ALL	47,XX,+21[11].ish del(X)(p22.33p22.33)[3'CRLF2+5'CR LF2-]/47,idem,del(1)(q32),del(3)(p13p22)/6/46,XX[3]	nuc ish(CEP4,CEP10)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6x2,RUNX1x3)[186/200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(3'CRLF2x2,5' CRLF2x1)[3' CRLF2 con 5' CRLF2x1][190/200]	a 319 kb loss within Xp22.33 with the proximal breakpoint upstream of the exon 1 of the CRLF2 gene and the distal breakpoint in the intron 1 of the P2RY8 gene. loss of 1q32.1q44 in 25% of cells and a ~33 Mb gain of 21q11.1q22.3 consistent with gain of chromosome 21, a deletion within 3p22.1p13	None	P2RY8:CRLF2; loss of partial 1q, partial 3q; gain 21	IKZF1 (NM_006060.6), c.331C>T, p.Arg111*	None (Category 3)
24	M	16	7/22/2023	AML	46,XY[20]	nuc ish(RUNX1T1,RUNX1)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(NUP98x2)[200] nuc ish(MYH11,CBF8)x2[200]	None, normal karyotype	cnLOH of 19q11q13.43	None, normal karyotype	cnLOH of 19q CEBPA (NM_004364.5), c.934_936dup, (p.Gln312dup) FLT3 (NM_004119.3), c.2505T>G, (p.Asp835Glu)	NA (normal karyotype)
25	M	4	7/20/2023	B-ALL	47,XY,+21c[20]	nuc ish(CEP4,CEP10)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6x2,RUNX1x3)[200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(3' CRLF2x3,5' CRLF2x1)[3' CRLF2 con 5' CRLF2x1][160/200]	Trisomy 21	gain of whole chromosome 5 and chromosome Xp22.33q28 and segmental losses of partial IKZF1 on chromosome 7p12.2, CDKN2B on 9p21.3, multiple genes on 14q32.33, partial CREBBP on 16p13.3, multiple genes on 19q13.12, and multiple genes on Xp22.33.	gain 21 germline	P2RY8:CRLF2; gains of 5, 7, 9, 16, X in tumor PAX5 (NM_016734.3), c.963dup, (p.Ala322Argfs*19)	None (Category 3)
26	F	4	7/17/2023	B-ALL	46,XX[20]	nuc ish(PBX1x3,TCF3x2)[60/200]	None, normal karyotype	a high hyperdiploid genome with gains of whole chromosomes X, 4, 5, 6, 8, 10, 11, 12, 14, 16, 17, 18, and 21, as well as two segmental aberrations.	None, normal karyotype	gain of whole chromosomes 4, 5, 6, 8, 10, 11, 12, 14, 16, 17, 18, 21 and X, gain of partial 1q	NA (normal karyotype likely due to growth advantage of normal cells)
27	F	13	10/23/2023	AML	47,XX,+6[20].ish t(5;11)(q35;p15)[3'NUP98+,5'NUP98-,3'NUP98-,5'NUP98+]	nuc ish(RUNX1T1,RUNX1)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(NUP98x2)[5'NUP98 sep 3'NUP98x1][194/200] nuc ish(MYH11,CBF8)x2[200]	gain of whole chromosome 6	None	NUP98:NSD1; gain of whole chromosome 6	FLT3 (NM_004119.3), c.1779_1832dup, (p.Asp593_Leu610dup)	None (Category 3)
28	F	1	10/12/2023	AML	48,XX,+8,+21c[5]/47,XX,+21c[16]	nuc ish(RUNX1T1x2,RUNX1x3)[166/200]/(RUNX1T1 x3,RUNX1x3)[34/200] nuc ish(NUP98x2)[200] nuc ish(KMT2Ax2)[200] nuc ish(MYH11,CBF8)x2[200]	gain of whole chromosome 21	None	Germline Trisomy 21	GATA1 (NM_002049.4), c.140_174del, (p.Ser47Cysfs*9) STAG2 (NM_006603.5), c.575delinsTC, (p.Tyr192Phefs*2)	a gain of whole chromosome (Category 2)
29	M	3	9/27/2023	B-ALL	64,XY,+X,+4,+5,+6,+8,+8,+10,+11,+12,+14,+17,+18,+18,+21,+21,+21,+22,+22[9]/46,XY[11]	nuc ish(CEP4,CEP10)x3[198/200] nuc ish(CEP8x4,MYCx4,IGHx3)[197/200] nuc ish(ABL1x2,BCR4x4)[198/200] nuc ish(KMT2Ax3)[195/200] nuc ish(ETV6x3,RUNX1x5)[197/200] nuc ish(CRLF2x3)[200]	gains of whole chromosomes 4, 5, 6, 8, 10, 11, 12, 14, 17, 18, 21, 22, and X.	a loss of partial 9p (involving the CDKN2A/B genes) and a relative loss of partial 12p (involving the ETV6 gene)	gain of whole chromosomes 4-6, 8(4x), 10 12, 14, 17, 18(4x), 21(5x), 22(4x) and X(2x) (~64 chromosomes)	loss of CDKN2A/B genes and relative loss of partial ETV6 gene on chromosome 12	None (Category 3)
30	F	1	9/24/2023	AML	48,XX,+8,+21c[8]/47,XX,+21c[14]	nuc ish(RH103072/RH67219,TERC,RH11834/RH123089)x2[RH103072/RH67219 sep TERC con RH11834/RH123089x1][31/200] nuc ish(RUNX1T1,RUNX1)x3[38/200]/(RUNX1T1x2,RUNX1x3)[162/200] nuc ish(NUP98x3)[25/200] nuc ish(KMT2Ax3)[29/200] nuc ish(MYH11,CBF8)x2[200]	non-mosaic gain of chromosome 21 and a low level mosaic gain of chromosome 8 in 5-10%	None	Germline Trisomy 21	GATA1 (NM_002049.4), c.191_194del (p.Arg64Thrfs*72) F2H2 (NM_00456.5), c.427_447del (p.Gly143_Leu149del) RAD21 (NM_006265.3), c.1322-1G>A (p.?)	None (Category 3)
31	M	3	8/23/2023	B-ALL	48,XY,+X,+21c[7].ish del(X)(p22.33)x2[3' CRLF2+ ,5' CRLF2-]/47,XY,+21c[13]	nuc ish(CEP4,CEP10)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6x2,RUNX1x3)[200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(3' CRLF2x3,5' CRLF2x1)[3' CRLF2 con 5' CRLF2x1][190/200]	gain of chromosome X with loss of 319 kilobases within Xp22.33, resulting in P2RY8:CRLF2	loss of 17p and gain of 17q	P2RY8:CRLF2; and gain of whole chromosome X	loss of 17p; gain of 17q JAK2 (NM_004972.4), c.2044_2047delinsGACC, (p.Ile682Asp) JAK2 (NM_004972.4), c.2047A>G, (p.Arg683Gly) JAK2 (NM_004972.4), c.2624C>A, (p.Thr875Asn)	None (Category 3)
32	M	3	8/19/2023	B-ALL	46,XY,del(9)(p271),t(12;21)[p13;q22],add(12)(p12)[13].ish t(12;21)(RUNX1+,ETV6- :RUNX1+,ETV6+),add(12)(ETV6-)/46,idem,- add(12),der(12)t(12;21)(RUNX1+,ETV6-)/5/46,XY[2]	nuc ish(CEP4,CEP10)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax1)[20/200] nuc ish(ETV6x1,RUNX1x3)[ETV6 con RUNX1x1][74/200] nuc ish(ETV6x2,RUNX1x4)[ETV6 con RUNX1x1][20/200] nuc ish(ETV6x2,RUNX1x4)[ETV6 con RUNX1x1][2/200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(CRLF2x2)[200]	gain of partial chromosome 21 encompassing RUNX1 gene, the losses involving CDKN2A/B and PAX5 on chromosome 9, and loss of ETV6 on chromosome 12	None	ETV6:RUNX1; P2RY8:CRLF2; loss of partial 9p, complex of partial 12p, gain of 721q	None	None (Category 3)

33	M	14	5/28/2023	B-ALL	46,XY,der(16)de(16)(p13.3)dup(16)(p11.2)dup(16)(q11.2q21),de(20)(q11.2q13.3)[9]ish t(X;14)(p22;q32)/t(Y;14)(p11.3;q32)(5' CRLF2+,3' CRLF2+,IGH-;3)IGH+,5'IGH-,3' CRLF2+,5' CRLF2-/46,XY[10]	nuc ish(CEP4,CEP10)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6,RUNX1)x2[200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(3' CRLF2x3,5' CRLF2x2)[3' CRLF2 con 5' CRLF2x2][186/200] nuc ish(ABL1x2)[200] nuc ish(ABL2x2)[200] nuc ish(PDGFRBx2)[200] nuc ish (3'IGHx2,5'IGHx1)[3'IGH con 5'IGHx1][188/200]	gain Xp22.33, loss of 34q32.33 and 20q11.21q13.33, loss and gains of 16	loss of 5q33.3, 7p12.2, 9p21.2,	gain of partial Xp, loss of 16p	losses of partial 5q and partial 7p JAK2 (NM_004972.4), c.2047A>G, (p.Arg683Gly) CRLF2 overexpression	None (Category 3)
34	F	12	5/19/2023	T-ALL	46,XX,de(1)(p36.12p36.33),?der(1)del(1)(7p12p31)del(1)(p36.12p36.33),del(3)(q13.11q21.2),del(6)(q14.2q22.1),del(9)(p24.1p24.3),del(11)(p12p14.3),del(12)(p13.1p12.3),der(13)del(13)(q12.3q14.11)del(13)(q21.2q21.33),?del(20)(q13.31q13.2)[cp13]/46,XX[7]	nuc ish(RANP17,TLX3)x2(200) nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6,RUNX1)x2[200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(CRLF2x2)[200]	~19.2 Mb loss of 1p36.33p36.12, a 22.6 Mb loss of 3q13.11q21.2, a 4.4 Mb loss of 9p24.3p24.1, a ~708 kb loss of 20q12.1, a ~4.5 Mb loss of 20q13.13q13.2, and multiple segmental losses on chromosome 12, including a ~1.5 Mb loss of 12p13.1p12.3, a 3.7 Mb loss of 12p12.3, a ~4.0 Mb loss of 12q11q12, and a ~764 kb loss of 12q24.3, which were detected in 75-80% of cells. Additionally, several low-level mosaic CNVs including a 32.9 Mb loss of 6q14.2q22.1, a ~8.6 Mb loss of 13q12.3q14.11, and a ~10 Mb loss of 13q21.2q21.33 were detected	a 14.4 Mb loss of 11p14.3p12 in >90% of cells that encompassed multiple genes including WT1	losses of partial 9p and partial 11p	NOTCH1::NOTCH1 ETV6 (NM_001987.5), c.287_311delinsATCTACGGAG, (p.Leu96_Tyr104delinsHisLeuArgSer) EZH2 (NM_004456.5), c.2069G>A, (p.Arg690His) EZH2 (NM_004456.5), c.3G>C, (p.Met17) FBXW7 (NM_033632.3), c.2065C>T, (p.Arg689Trp) NOTCH1 (NM_017617.5), c.7400C>A, (p.Ser2467*) PTEN (NM_000314.8), c.7167T>A, (p.Met239Lys)	None (Category 3)
35	F	1	5/4/2023	B-ALL	46,XX,t(5;7)(q23;q36),der(9)t(9;15)(p13.2;q24.1),der(15)t(9;15)(p13.2;q24.1)del(9)(p21.3p22.1)[10],ish der(9)(PML+),der(15)(PML+),del(X)(p22.33)[3' CRLF2+,5' CRLF2-/46,XX[10]	nuc ish(CEP4,CEP10)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6,RUNX1)x2[200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(3' CRLF2x2,5' CRLF2x1)[3' CRLF2 con 5' CRLF2x1][140/200] nuc ish(ABL1x2)[200] nuc ish(ABL2x2)[200] nuc ish(PDGFRBx2)[200]	a 307 kb loss of Xp22.33 in approximately 50% of cells, which brings exon 1 of the CRLF2 gene to the promoter of P2RY8 gene leading to the P2RY8::CRLF2 fusion; There were also segmental losses on 5q and 9p, all estimated to occur in approximately 65% of cells. There was a mosaic homozygous loss of MLLT3 and CDKN2A/B genes in the background of a larger heterozygous deletion of 6.6 Mb at 9p22.1-p21.3.	None	P2RY8::CRLF2; PAX5::PML; loss of partial 9p	KRAS (NM_004985.5), c.35G>C (p.Gly12Ala)	None (Category 3)
36	F	2	5/2/2023	B-ALL	No growth	nuc ish(CEP4,CEP10)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax1)[56/200] nuc ish(ETV6x3,RUNX1x4)[ETV6 con RUNX1x2][52/200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(CRLF2x2)[200]	N/A, Cyto no growth	a 36.7 Mb terminal loss on chromosomes 11q22.1q25, a 11.2 Mb terminal region on 12p13.33p13.2 (with the breakpoint involving the ETV6 gene), a 21.6 Mb gain on 21q11.2q22.12 (with the distal breakpoint involving the RUNX1 gene), a 253 kb cNLOH on 12q21.33 (involving the BTG1 gene), and cNLOH involving the short arm of chromosome 9	N/A, Cyto no growth	ETV6::RUNX1; cNLOH of 9p	NA (no growth)
37	F	19	7/2/2023	B-ALL	46,XX,t(X;14)(p22.33;q32)[7],ish t(X;14)(IGH+;IGH+;46,XX)[12]	nuc ish(CEP4,CEP10)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6,RUNX1)x2[200] nuc ish(CEP8x2,MYCx2,IGHx3)[184/200] nuc ish(CRLF2x2)[5' CRLF2 sep 3' CRLF2x1][186/200] nuc ish(ABL1x2)[200] nuc ish(ABL2x2)[200] nuc ish(PDGFRBx2)[200]	complex losses of 14q32.33 involving IGH	a 63kb loss of 5q33.3 (63kb) involving the partial EBF1 gene, a 37kb loss of 7p12.2 involving the partial IKZF1 gene, and a 88kb loss of 13q14.2 involving the partial RB1 gene	None	partial losses of 5q, 7p, 10q, 13q CRLF2 (NM_022148.4), c.700_701insGCC (p.Ile233_Leu234insCys) IKZF1 (NM_006060.6), c.422G>A (p.Gly141Glu)	None (Category 3)
38	M	13	6/30/2023	B-ALL	46,XY,t(1;19)(q23;p13.3),dup(5)(q11.2q31.1)[19]	nuc ish(PBX1,TCF3)x3[PBX1 con TCF3x2][198/200]	a 1.4Mb gain of 5q11.2 and 79Mb gain of 5q11.2q31.1, a 669kb loss on chromosome Xp21.1	None	N/A, NGS not performed	N/A, NGS not performed	None (Category 3)
39	F	4	6/29/2023	B-ALL	No growth	nuc ish(CEP4,CEP10)x3[31/100] (CEP4x4,CEP10x3)[37/100] nuc ish(ABL1,BCR)x2[100] nuc ish(KMT2Ax2)[100] nuc ish(ETV6x2,RUNX1x4)[65/100] nuc ish(CEP8x2,MYCx2,IGHx3)[65/100] nuc ish(CRLF2x4)[67/100]	N/A, Cyto no growth	gains of multiple whole chromosomes, including chromosomes X, 4, 6, 10, 14, 17, and 21	N/A, Cyto no growth	somatic copy number gain of one or two extra chromosomes X, 4, 6, 10, 14, 17 and 2 KRAS (NM_004985.5), c.436G>A, (p.Ala146Thr) NRAS (NM_002524.5), c.64C>A (p.Gln221Lys)	NA (no growth)
40	F	3	6/27/2023	B-ALL	46,XX,t(12;21)(p13;q22),del(12)(p12.2)q13.31[1]ish t(12;21)(RUNX1+;ETV6-RUNX1+;ETV6+)[47,XX,idem,+10(5)/48,idem,+10,+der(12)(12;21)[1],ish +der(21)(RUNX1+;ETV6+)[49,idem,+10,+der(12)(12;21),+der(21)(12;21)[1],ish +der(12)(RUNX1+;ETV6+),+der(21)(RUNX1+;ETV6+)[46,XX[12]	nuc ish(CEP4x2,CEP10x3)[194/200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6x1,RUNX1x3)[ETV6 con RUNX1x1][172/200] (ETV6x2,RUNX1x4)[ETV6 con RUNX1x2][14/200] (ETV6x2,RUNX1x5)[ETV6 con RUNX1x2][10/200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(CRLF2x2)[200]	an 18.7Mb loss of 12p13.33p12.2 (contain	a 482kb loss of 3p14.2 (involving FHT1), a 234kb loss of 9p13.2 (involving partial PAX5), a 331kb loss of 12q21.33 (involving partial BTG1), and a 1.1Mb loss of 16p13.3.	ETV6::RUNX1; partial loss of 12p	partial loss of 9p	None (Category 3)
41	F	6	6/22/2023	B-ALL	46,XX,der(21)dup(21)(q21q22)[4],ish t(21)(RUNX1amp)/47,idem,+x[4]/46,XX[17]	nuc ish(CEP4,CEP10)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6x2,RUNX1amp)[102/200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(CRLF2x3)[96/200]	complex alteration on chromosome 21, including chromothripsis impacting chromosome 21q11.1q22.11 and amplification of region 21q22.11q22.3 encompassing the RUNX1 gene.	None	at least 5 copies of partial 21q involving the RUNX1 gene, gain of whole chromosome X	loss of partial chromosome 9p involving the CDKN2A gene CDKN2A (NM_000077.5), c.164del, (p.Gly55Alafs*91) KRAS (NM_004985.5), c.436G>A, (p.Ala146Thr) NRAS (NM_002524.5), c.183A>T, (p.Gln61His) SH2B3 (NM_005475.3), c.924delinsGCTCCGGAGG, (p.Gly309Leufs*19)	None (Category 3)

42	M	10	6/22/2023	B-ALL	46,XY[20]	nuc ish(RUNX1T1,RUNX1)x2[200] nuc ish(KMT2A)2[200] nuc ish(MYH11,CBFB)x2[200] nuc ish(NUP98)x2[200]	None, normal karyotype	loss of whole chromosomes 2, 3, 4, 7, 12, 13, 15, 16, and 17 at approximately 5-10% mosaic level, suggesting a low-hypodiploid tumor genome (37 chromosomes total).	None, normal karyotype	TP53 (NM_000546.6), c.818G>A (p.Arg273His) germline mosaic	NA (normal karyotype likely due to growth advantage of normal cells)
43	M	11	4/4/2023	B-ALL	47,XY,t(12;21)(p13;q22),+21c[12].ish t(12;21)(RUNX1+,ETV6+;RUNX1+,ETV6+)/47,XY,+21c[8]	nuc ish(CEP4,CEP10)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2A)2[200] nuc ish(ETV6x2,RUNX1x4)[ETV6 con RUNX1x1][198/200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(CRLF2x2)[200]	germline gain of 21	None	ETV6:RUNX1; germline gain of 21	partial loss of 9p	None (Category 3)
44	M	4	3/31/2023	B-ALL	46,XY,der(12)t(12;21)(p13;q22),ider(21)t(12;21)(p13;q22)[6].ish der(12)(RUNX1+,ETV6-),ider(21)(ETV6+,RUNX1+);47,ide m,+der(21)t(12;21)(p13;q22)add(21)(p11)[5].ish der(21)RUNX1+,ETV6+/46,XY[14]	nuc ish(CEP4,CEP10)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2A)2[200] nuc ish(ETV6x3,RUNX1x4)[ETV6 con RUNX1x2][42/200] [ETV6x4,RUNX1x5][ETV6 con RUNX1x3][80/200] [ETV6x5,RUNX1x6][ETV6 con RUNX1x4][66/200] nuc ish(CEP8x2,MYC,x3,IGHx2)[20/200] nuc ish(CRLF2x2)[200]	gain (4 copies or more) involving ETV6 on chromosome 12 and gain (4 copies or more) of RUNX1 on chromosome 21	a 166 kilobase (kb) loss on 7q34 in 85-90% of cells, a 52 Megabase (Mb) gain of chromosome 8q21.3q23.3 in less than 5% of cells, two ~2 Mb losses involving 9q in 90-95% of cells, a 626 kb loss of 14q11.2 in 95 to 100% of cells, several segmental losses on chromosome 19	ETV6:RUNX1; gains of multiple exons within both ETV6 and RUNX1	partial loss of 19p and 19q	None (Category 3)
45	F	18	3/21/2023	AML	46,XX,t(11;17)(q23;q21.3;p13.3)19p13.3,ider,+X,+5,+15,+18,+der(19)t(11;17)(q23;q21.3)19p13.3,TCF3+,LSI1+,RARA+,3'KMT2A+,TCF3+,LSI19p13.3+,LSI19q13.3+][11]	nuc ish(KMT2A)2[500]	low-level mosaic gains of whole chromosomes X, 5, 15, 18, 20, and 21	segmental gains of chromosomes 17q21.33q25.3 and 19p13.3q13.43.	KMT2A:MLL1	None	a three-way translocation (Category 2)
46	F	5	3/7/2023	B-ALL	46,XX[20]	nuc ish(ABL1,BCR)x3[ABL1 con BCRx2][120/200]	None, normal karyotype	gains of whole chromosomes 8 and 14, cnLOH involving partial chromosome 16p, and segmental losses including loss of CDKN2A/B and PAX5 on chromosome 9p.	None, normal karyotype	BCR:ABL1 (p210) USH2A (NM_206933.2), c.4133_4134dup (p.Asn1379Serfs*54)	NA (normal karyotype likely due to growth advantage of normal cells)
47	F	7	3/1/2023	B-ALL	47,XX,+21c[20]	nuc ish(CEP4,CEP10)x2[200] nuc ish(CEP8,MYC,IGH)x2[199/200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2A)2[200] nuc ish(ETV6x2,RUNX1x3)[200] nuc ish(3'CRLF2x2,5'CRLF2x1)5'CRLF2 con 3'CRLF2x1[180/200]	germline trisomy 21	somatic loss of EBF1 on Sq33.3, and a ~320 kb mosaic deletion within the pseudoautosomal region on Xp22.33, which is predicted to result in the P2RY8:CRLF2 fusion	germline trisomy 21.	P2RY8:CRLF2 JAK2 (NM_004972.4), c.2047A>G (p.Arg683Gly)	None (Category 3)
48	M	7	2/22/2023	B-ALL	51,XY,+X,+9,+14,+21,+21[8]/46,XY[7]	nuc ish(CEP4,CEP10)x2[200] nuc ish(ABL1x3,BCR)2[104/200] nuc ish(KMT2A)2[200] nuc ish(ETV6x2,RUNX1x4)[162/200] nuc ish(CEP8x2,MYC,x2,IGHx3)[104/200] nuc ish(CRLF2x3)[102/200]	trisomic gains of chromosomes X, 9, and 14, tetrasomic gain of chromosome 21	cnLOH in chromosomes 4, 6, 11, 18, and 20 with varying mosaic levels, segmental cnLOH and segmental CNV in chromosomes 13 and 17.	gains of whole chromosomes X, 9, 14, and 21	NRAS (NM_002524.5), c.35G>T (p.Gly12Val) CREBBP (NM_004380.3), c.4336C>T (p.Arg1446Cys)	None (Category 3)
49	M	15	2/22/2023	CML	46,XY,t(9;22)(q34;q11.2)[20]	nuc ish(ABL1,BCR)x2[500]	None, arr(X,Y)x1,(1-22)x2	None, arr(X,Y)x1,(1-22)x2	BCR:ABL1	GATA2 (NM_032638.5), c.1096G>T (p.Gly366Trp) PPH6 (NM_032458.3), c.385C>T (p.Arg129*) RUNX1 (NM_001754.5), c.496C>T (p.Arg166*)	None (Category 3)
50	M	4	2/20/2023	B-ALL	46,XY,del(6)(q21q23.2),t(12;21)(p13;q22)[11].ish t(12;21)(RUNX1+,ETV6+;RUNX1+,ETV6+)/47,ider,+X[8]/46,XY[1]	nuc ish(CEP4,CEP10)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2A)2[200] nuc ish(ETV6x2,RUNX1x3)[ETV6 con RUNX1x1][198/200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(CRLF2x3)[20/200]	loss of partial 6q and gain of a copy of X-chromosome.	loss of 9p	ETV6:RUNX1; gain of chromosome X.	losses of partial 4p (NSD2 exons 2-4), 9p (PAX5 exons 2-6), and 10q (PTEN exons 2-5)	None (Category 3)
51	M	2	2/3/2023	B-ALL	46,XY,[9](q10),del(13)(q13.3q33.2)der(19)t(19)(q23.3q13.3)[7].ish [9](ABL1+),der(19)(PXB1+,TCF3+)[46,XY[13]	nuc ish(CEP4,CEP10)x2[200] nuc ish(ABL1x3,BCR)2[182/200] nuc ish(KMT2A)2[200] nuc ish(ETV6,RUNX1)x2[200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(CRLF2x2)[200] nuc ish(ABL1x3)[184/200] nuc ish(ABL2x3)[188/200] nuc ish(PDGFRb)2[200] nuc ish(PBX1x3,TCF3x2)[PBX1 con TCF3x1][190/200]	losses of 9p24.3p13.1 [involving CDKN2A/B], 13q13.3q33.2 and 19p13.3, and gains of 1q23.3q44 and 9q21.11q34.3	None	TCF3:PBX1; gain of chromosome 1q and loss of partial 19p including exons 17-19 of the TCF3 gene, partial 13q loss	None	a derivative chromosome and an isochromosome (Category 2)
52	M	15	1/31/2023	B-ALL	45,XY,der(7;9)(p11.2;p13.1)[3]/46,XY[17]	nuc ish(CEP4,CEP10)x2[200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2A)2[200] nuc ish(ETV6,RUNX1)x2[200] nuc ish(CRLF2x2)[200] nuc ish(ABL1x2)[200] nuc ish(ABL2x2)[200] nuc ish(PDGFRb)2[200]	a ~57 Mb loss of 7p22.3p11.2, a ~39 Mb loss of 9p24.3p13.1	None	loss on chromosome 7p involving IKZF1 and loss on chromosome 9p involving CDKN2A/B and PAX5.	KRAS (NM_033366.3), c.34G>A (p.Gly12Ser) KDM6A (NM_021140.3), c.4193_4198delinsGGCCCTCCG (p.Ser1398Tprfs*142) CDKN2A (NM_000077.4), c. c.387C>G (p.Tyr129*)	a derivative chromosome (Category 2)
53	F	3	1/25/2023	T-ALL	47,XX,+8[11]/46,XX[14]	nuc ish(RANBP1,TXN3)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2A)2[200] nuc ish(ETV6,RUNX1)x2[200] nuc ish(CEP8x3,MYC,x3,IGHx2)[168/200] nuc ish(CRLF2x2)[200]	gain of chromosome 8	deletion on chromosome 1p33, complex losses involving chromosome 9p including homozygous loss of CDKN2A/B, loss involving PTEN on chromosome 10q23.31, and losses involving T-cell receptor loci on chromosome 14q11.2.	gain of whole chromosome 8	STIL:;TAL1 loss of partial 9p involving homozygous loss of CDKN2A/B, and loss of partial PTEN on chromosome 10q NOTCH1 (NM_017617.4), c.7398_7399insAGGGGGG (p.Ser2467Argfs*13) PTEN (NM_000314.6), c.715_729del (p.Met239_Phe243del)	None (Category 3)

54	M	10	1/17/2023	T-ALL	46,XY,der(7)del(7)(p21.2pter)del(7)(q33q36.3),del(12)(p11.3)[8]/46,XY[12]	nuc ish(RANBP17,TX3x2)[200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2A2)[200] nuc ish(ETV6x1,RUNX1x2)[125/200] nuc ish(CRLF2x2)[200]	losses of 7p22.3p21.2 (16.0Mb), 7q33q36.3 (18.7Mb), and 12p13.33p11.23 (16.7Mb).	a copy neutral loss of heterozygosity (cnLOH) of 1p36.33p13.3	loss of partial 7q (including BRAF, EZH2, and KMT2C) and partial 12p (including ETV6, ETNK1, and KRAS).	cnLOH of partial 1p (including CSF3R, MPL, TAL1, and JAK1) EZH2 (NM_004456.4), c.73C>T (p.Arg25*) EZH2 (NM_004456.4), c.865_866insAATAAGGT (p.Cys289*) JAK1 (NM_002227.3), c.1954T>C (p.Tyr652His) RAD21 (NM_006265.2), c.1247_1250delins15 (p.Phe416*)	None (Category 3)
55	M	13	1/12/2023	B-ALL	46,XY,(9)(q10),der(21)dup(21)(q11.2q22)del(21)(q22)[20].ish (9)(ABL1+),der(21)(RUNX1amp)	nuc ish(CEP4,CEP10)x2[200] nuc ish(ABL1x3,BCR2x2)[190/200] nuc ish(KMT2A2)[200] nuc ish(ETV6x2,RUNX1amp)[190/200] nuc ish(CEP8x2,MYC3,IGHx2)[192/200] nuc ish(CRLF2x2)[200] nuc ish(ABL1x3)[188/200] nuc ish(ABL2x2)[200] nuc ish(PDGFRBx2)[200]	an intrachromosomal amplification of partial 21q along with multiple segmental gains of 21q and loss of the terminal part of 21q; loss of 9p and gain of 9q	multiple segmental chromosome gains and losses involving chromosomes 1p, 3p, 4q, 11q, 17q	gains of partial chromosome 21q (including RUNX1, ERG, and UZF1), loss of 9p and gain of 9q	P7FN11 (NM_002834.4), c.182A>T (p.Asp61Val)	an isochromosome (Category 2)
56	M	16	12/22/2022	AML	45,XY,t(10;11)(p12;q14.2),del(11)(p12p14.3),add(12)(p12.1),-18,der(21)t(7;21)(p11.2)(p16)/46,XY[3]	nuc ish(RUNX1T1,RUNX1)x2[200] nuc ish(KMT2A2x2)[200] nuc ish(MYH11,CBF8)x2[200] nuc ish(NUP98x2)[200]	loss of 11p14.3p12 (12.4Mb), 12p13.33p12.1 (25.7Mb), 17q11.2 (1.4Mb), and 18q22.3q23 (7.2Mb)	None	PICALM:MLLT10; loss of partial 11p (including WT1), 12p (including ETV6), and partial 17q (including NF1 and SUZ12)	ASXL1 (NM_015338.5), c.1660_1661del (p.Ser554Cysfs*10) PHF6 (NM_032458.2), c.955C>T (p.Arg319*)	a derivative chromosome (Category 2)
57	F	12	12/5/2022	B-ALL	46,XX,t(9;22)(q34;q11.2),del(20)(q13.1)[7]/46,XX[13]	nuc ish(CEP4,CEP10)x2[200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(ABL1,BCR)x3(ABL1 con BCRx2)[152/200]/(ABL1,BCR)x4(ABL1 con BCRx3)[28/200] nuc ish(KMT2A2x2)[200] nuc ish(ETV6,RUNX1)x2[200] nuc ish(CRLF2x2)[200] nuc ish(ABL1x2)[5'ABL1 sep 3'ABL1x1][146/200]/(5'ABL1x2,3'ABL1x3)(5'ABL1 con 3'ABL1x1)[30/200] nuc ish(ABL2x2)[200] nuc ish(PDGFRBx2)[200]	loss involving 20q; loss of 9p21.1 (170 kb) involving loss of MTP and homozygous loss of CDKN2A, gain of 9q34.12q34.3 (7.3Mb)	None	BCR:ABL1; loss partial 20q; loss of partial 9p, gain of partial 9q	loss of partial 7p	None (Category 3)
58	M	12	12/5/2022	AML	46,XY,t(9;11)(p21.3;q23)[10]/47,der(+8)(7)/47,der(+6,del(10)(p173),der(12)(1;12)(q10;p11.2)[3]	nuc ish(RUNX1T1,RUNX1)x2[500] nuc ish(KMT2A2)[500]	gain of whole chromosome 8	None	KMT2A:MLT3, low-level mosaic gain of chromosome 8	ASXL1 (NM_015338.5), c.1934dup (p.Gly646Trpfs*12) FLT3 (NM_004119.2), c.2508_2510del (p.Leu836del)	a derivative chromosome and a balanced translocation (Category 2)
59	F	19	11/29/2022	B-ALL	No growth	nuc ish(CEP4,CEP10)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2A1)[184/200] nuc ish(ETV6x2,RUNX1x3-amp)[180/200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(CRLF2x2)[200] nuc ish(ABL1x2)[200] nuc ish(ABL2x2)[200] nuc ish(PDGFRBx2)[200]	N/A, Cyto no growth	a complex copy number pattern involving 21q consistent with intrachromosomal amplification of chromosome 21 (IAMP21). Additional segmental alterations, including loss of PAX5, ETV6, and RB1. Copy neutral loss of heterozygosity (cnLOH) was identified across 9p24.3p21.3 and involved CDKN2A/B and JAK2	N/A, Cyto no growth	at least four copies of RUNX1 on chromosome 21q. Loss of heterozygosity involving CDKN2A/B and JAK2 and loss of PAX5 on chromosome 9p, loss of ETV6 on chromosome 12, and homozygous loss of RB1 on chromosome 13.	NA (no growth)
60	F	13	11/29/2022	B-ALL	47,XX,der(16)t(X;16)(p11.2;p13.1),-21c[7]/47,XX,+21c[13]	nuc ish(CEP4,CEP10)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2A2)[200] nuc ish(ETV6x2,RUNX1x3)[200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(CRLF2x3)[172/200]	a ~44.7 Mb gain of chromosome Xp22.33p11.3 with a relative loss of Xp22.33, a ~12.9 Mb loss of 16p13.3p13.12, gain of whole chromosome 21 was noted in 100% of cells	a ~39 Mb cnLOH on chromosome 9p24.3p13.1 with loss of CDKN2A, and a ~70.6 Mb region of cnLOH across 12q14.2. This alteration includes loss of heterozygosity of SH2B3 and homozygous deletion of BTG1.	P2RV8:CRLF2, biallelic loss of SH2B3, a very low level FLT3 internal tandem duplication, and multiple copy number variants, including the known germline trisomy 21; gain of partial Xp, loss of CDKN2A on chromosome 9p	JAK2 (NM_004972.3), c.2047A>G (p.Arg683Gly) JAK2 (NM_004972.3), c.2049A>T (p.Arg683Ser) copy-neutral loss of heterozygosity involving SH2B3 on chromosome 12q.	a derivative chromosome (Category 2)
61	F	13	9/16/2022	AML	45,X(X[6]/46,XX[14])	nuc ish(RUNX1T1,RUNX1)x2[200] nuc ish(KMT2A2)[200] nuc ish(NUP98x2)[200] nuc ish(MYH11,CBF8)x2[200]	loss of chromosome X in a small percentage of cells	three segmental losses involving the long arm of chromosome 4, and again of partial RUNX1 on chromosome 21 in ~70% of cells.	None	a gain of partial (exons 2-6) RUNX1 gene and low-level mosaic cnLOH of chromosome X	None (Category 3)
62	M	6	8/17/2022	AML	46,XY,t(11;17)(q23;q21)[19].ish t(11;17)(5'KMT2A+,3'KMT2A-;5'KMT2A+,3'KMT2A+)/46,XY[1]	nuc ish(RUNX1T1,RUNX1)x2[200] nuc ish(CEP4,CEP10)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(ETV6,RUNX1)x2[200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(CRLF2x2)[200]	gain of 11q23.3 involving KMT2A and gain of 17q12 involving MLLT6	None	KMT2A:MLLT6; Copy number analysis identified gain of partial 11q involving KMT2A (exon 1-8)	None	None (Category 3)
63	F	3	7/8/2022	B-ALL	46,XX[20]	nuc ish(CEP4,CEP10)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2A2)[200] nuc ish(ETV6,RUNX1)x2[200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(CRLF2x2)[200]	None, normal karyotype	a segmental loss of 5p13.3	None, normal karyotype	KRAS (NM_033360.3), c.386G>A (p.Gly13Asp) NRAS (NM_002524.4), c.176C>A (p.Ala59Asp) NRAS (NM_002524.4), c.35G>T (p.Gly12Val) NRAS (NM_002524.4), c.386G>A (p.Gly13Asp)	NA (normal karyotype)
64	F	9	11/23/2022	AML	46,XX,t(8;21)(q22;q22)[2]/46,der(11)(p11.2p15.1)[5]/46,XX[13]	nuc ish(KMT2A2)[200] nuc ish(NUP98x2)[200] nuc ish(MYH11,CBF8)x2[200]	loss of 11p15.1p11.12	None	RUNX1:RUNX1T1; loss of 11p	ASXL1 (NM_015338.5), c.1934dup (p.Gly646Trpfs*12) NRAS (NM_002524.4), c.182A>G (p.Gln61Arg)	None (Category 3)
65	M	16	11/14/2022	AML	46,XY,der(9)t(9;10)(p13;p12)ins(9;11)(p13;q23.3q23.3),der(10)t(9;10)ins(9;11),der(11)t(9;11)(p17),ish der(9)(3'KMT2A+),der(10)(5'KMT2A+),der(11)(5'KMT2A+,3'KMT2A-)/47,der(+8)[3]	nuc ish(RUNX1T1x3,RUNX1x2)[60/200] nuc ish(3'KMT2Ax3,3'KMT2Ax2)(5'KMT2A con 3'KMT2Ax1)[190/200] nuc ish(NUP98x2)[200] nuc ish(MYH11,CBF8)x2[200]	segmental losses impacting MLLT10 and KMT2A in the majority of cells. These segmental losses are likely related to the possible three-way rearrangement involving chromosomes 9, 10, and 11	gain of chromosome 8 in 15% of cells	KMT2A:MLLT10 and partial loss of 11q, gain of whole chromosome 8	SETD2 (NM_014159.6), c.4792C>T (p.Arg1598*) SETD2 (NM_014159.6), c.7535_7540delinsG (p.Leu2512Argfs*6) KRAS (NM_033360.3), c.356G>A (p.Gly12Asp)	a complex rearrangement involving multiple chromosomes (Category 2)
66	M	2	11/11/2022	B-ALL	53,XY,+x,+4,+6,+14,+17,+21,+21[4]/46,XY[16]	nuc ish(CEP4x3,CEP10)x2[130/200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2A2)[200] nuc ish(ETV6x2,RUNX1x4)[124/200] nuc ish(CEP8x2,MYC3,IGHx3)[124/200] nuc ish(CRLF2x3)[128/200]	a high hyperdiploid tumor genome with gains of multiple whole chromosomes, including chromosomes X, 4, 6, 14, 17, and 21.	None	a high hyperdiploid tumor genome with gains of multiple whole chromosomes, including chromosomes X, 4, 6, 14, 17, and 21.	CREBBP (NM_004380.2), c.4275C>A (p.Asn1425Lys) KRAS (NM_033360.3), c.346G>T (p.Gly12Cys) KRAS (NM_033360.3), c.356G>A (p.Gly12Asp) KRAS (NM_033360.3), c.356G>T (p.Gly12Val) NRAS (NM_002524.4), c.356G>A (p.Gly12Asp)	None (Category 3)
67	M	8	10/24/2022	Mixed acute leukemia, T/myeloid	46,XY[20]	nuc ish(RANBP17,TX3)x2[200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2A2)[200] nuc ish(ETV6,RUNX1)x2[200] nuc ish(CRLF2x2)[200]	None, arr(X,Y)x1,(1-22)x2 and normal karyotype	None, arr(X,Y)x1,(1-22)x2 and normal karyotype	None, normal karyotype	FLT3 (NM_004119.2), c.1837+40_1837+41ins93 (p.7) RUNX1 (NM_001754.4), c.415_418delinsGTGGGA (p.Asn139_Tyr140delinsCysGlyAsn) RUNX1 (NM_001754.4), c.908C>G (p.Ser303*) WT1 (NM_024426.5), c.1122_1125delinsCC (p.Val376Alafs*13)	NA (normal karyotype)

68	F	5	10/10/2022	B-ALL	61,XX,+X,+X,+4,+5,+6,+8,del(9)(p21.22),+10,+11,+12,+14,+14,del(14)(q24.2qter),+17,+21,+21,+22[9]/46,XX[9]	nuc ish(CEP8,CEP10)x3[183/200] nuc ish(CEP8,MYC,IGH)x3[179/200] nuc ish(ABL1x2,BCR)x3[181/200] nuc ish(KMT2Ax3)[176/200] nuc ish(ETV6x3,RUNX1x4)[180/200] nuc ish(CRLF2x2)[175/200] nuc ish(KMT2Ax2)[5'KMT2A sep 3'KMT2Ax1][126/200]	gain of an extra copy of whole chromosomes 4, 5, 6, 8, 10, 11, 12, and 22, tetrasomy of chromosome 21; loss of 9p and gain of Xp, Xq, and 14q	copy-neutral loss of heterozygosity of whole chromosomes 7, 16, and 20; gain of 17p and 19q	gain of whole chromosomes X, 4, 5, 6, 8, 10, 11, 12, 14, 17, 22; loss of 9p and gain of 14q	KRAS (NM_033360.3), c.356G>A (p.Gly12Asp) CREBBP (NM_004380.2), c.4337G>A (p.Arg1446His)	None (Category 3)
69	M	0	5/27/2021	AML	46,XY,der(4)(qter->q31::11q27.2->11q21::11q23->11qter),der(10)(qter->q31::11q27.2->11q23::10p12->10qter),der(11)(11pter->11q21::10p12->10qter),add(20)(q13.3)[6],ish der(4)(3'KMT2A+),der(10)(5'KMT2A+),der(11)(5'KMT2A-,3'KMT2A-)/46,XY[14]	nuc ish(RANBP17,TLX3)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6,RUNX1)x2[200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(CRLF2x2)[200]	None, array not performed	None, array not performed	KMT2A:MLL10	None	a three-way translocation (Category 2)
70	M	15	5/29/2021	T-ALL	46,XY,del(6)(q14.1q22)[4]/46,XY[16]	nuc ish(RANBP17,TLX3)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6,RUNX1)x2[200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(CRLF2x2)[200]	loss of 6q14 (4.1Mb of size), loss of 6q14.1q21 (31.6Mb), loss of 6q21q22.31 (11.4Mb)	copy neutral loss of heterozygosity (cnLOH) of 6q22.31q27 (46.1Mb), loss of 14q11.2 (499.5Kb) and cnLOH of 9p24.3p13.3 (33.6Mb) with the exception that 9p21.3 (741.4Kb), involving CDKN2A, MTP, and CDKN2B appeared to be homozygous loss.	NOTCH1:NOTCH1	USP7 (NM_003470.2), c.2571_2572dup (p.Glu858Valfs*5) NOTCH1 (NM_017617.4), c.7475C>A (p.Ser2492*) PTEN (NM_000314.6), c.642del (p.Gln214Hisfs*7)	None (Category 3)
71	M	12	6/12/2021	T-ALL	46,XY,del(9)(q34q34)[7],ish del(9)(ABL1-)/46,del,del(9)(q21q31),7del(12)(p13)[7],nuc ish(ETV6x1)[10/200]/46,XY[6]	nuc ish(RANBP17,TLX3)x2[200] nuc ish(ABL1x1,BCR)x2[170/200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6x1,RUNX1x2)[10/200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(CRLF2x2)[200]	a 4.9 Mb deletion on 9q21.11q31.1, a 2.5 Mb deletion on 9q34.11q34.13	a 1.2 Mb gain on 16p13.11, and a 93.8 Mb segment of cnLOH on 12q12q24.	SET:NUP214; loss of 9q	NRAS (NM_002524.4), c.356G>A (p.Gly12Asp) UZAF1 (NM_006758.2), c.104G>T (p.Arg35Leu) WT1 (NM_024426.5), c.1158_1159ins17 (p.Ala387*) NOTCH1 (NM_017617.4), c.7171C>T (p.Gln2391*) SH2B3 (NM_005475.2), c.1383C>G (p.Tyr461*) PHF6 (NM_032458.2), c.99_100delins13 (p.Ser34Phefs*51) cnLOH of 12q	None (Category 3)
72	F	6	6/12/2021	B-ALL	61*52,XX,+X,+2,del(2)(q22qter),+4,+5,+7,del(7)(q32q36),+8,+8,+10,+11,(12)(q10),+14,+17,+18,+21,+21,+21,+22[cp18]/46,XX[2]	nuc ish(CEP4,CEP10)x3[190/200] nuc ish(ABL1x2,BCR)x3[188/200] nuc ish(KMT2Ax3)[190/200] nuc ish(ETV6x2,RUNX1x4)[116/200] nuc ish(ETV6x2,RUNX1x3)[10/200] nuc ish(ETV6x2,RUNX1x5)[10/200] nuc ish(CEP8x4,MYCx4,IGHx3)[188/200] nuc ish(CRLF2x3)[192/200]	mosaic gains of one extra copy of chromosomes 4, 5, 7, 10, 11, 14, 17, 18, 22, and X and two extra copies of chromosomes 8 and 21, a 148.4 Mb gain in 2p25.3q22.3, a 26.4 Mb gain in 6p25p22.2	a 144.3 Mb LOH on 6p22.2q27, and copy neutral LOH (cnLOH) of chromosomes 9 and 13	a high hyperdiploid genome, chromosomes 4, 5, 7, 8, 10, 11, 14, 17, 18, 21, 22, and X, and copy neutral LOH (cnLOH) of chromosomes 6, 9, and 13.	KRAS (NM_033360.3), c.386G>A (p.Gly13Asp)	None (Category 3)
73	F	3	6/17/2021	B-ALL	46,XX,del(12)(p12),t(12;21)(p13;q22),del(13)(q12)[15],ish del(12)(ETV6-),t(12;21)(ETV6-RUNX1+),ETV6+,RUNX1+]/46,XX[5]	nuc ish(CEP4,CEP10)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6x1,RUNX1x3)[ETV6 con RUNX1x1][190/200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(CRLF2x2)[200]	a 16.1 Mb loss on 12p13.3p12.3 involving multiple genes including ETV6, an 8.1 Mb gain on 12p12.3p12.1 and a 3.2 Mb loss on 12p12.1p11.23 on the short arm of chromosome 12, an 83.1 Mb loss on 13q12.3q34 involving the majority of chromosome 13 including RBL1.	several small CNVs including a loss on 2p11.2, 6p22.2, 6q25.3 and 12q21.33, a 6.5 Mb gain on 13q12.12q12.3	ETV6:RUNX1; complex CNV on 12p and 13p	None	None (Category 3)
74	M	15	6/22/2021	B-ALL	No growth	nuc ish(ABL1x3)[143/200] nuc ish(ABL2x2)[200] nuc ish(PDGFRBx2)[200] nuc ish(CEP4,CEP10)x2[200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(ABL1x3,BCR)x2[140/200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6,RUNX1)x2[200] nuc ish(CRLF2x2)[200] nuc ish(ABL1x2)[200] nuc ish(PDGFRBx2)[200] nuc ish(CEP4,CEP10)x2[200] nuc ish(CEP8x2,MYCx2,IGHx3)[138/200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6,RUNX1)x2[200] nuc ish(CRLF2x2)[5'CRLF2 sep 3'CRLF2x1][69/70] nuc ish(IGHx2)[5'IGH sep 3'IGHx1][192/200]	N/A, Cyto no growth	losses on chromosome 2p, 6q, and 9p and gain of chromosome 9q	N/A, Cyto no growth	loss of chromosome 9p and gain of chromosome 9q PAX5 (NM_016734.2), c.547G>T (p.Gly183Cys) KRAS (NM_033360.3), c.406G>T (p.Val14Leu)	NA (no growth)
75	F	4	7/2/2021	B-ALL	No growth	nuc ish(CRLF2x2)[200] nuc ish(ABL1x2)[200] nuc ish(PDGFRBx2)[200] nuc ish(CEP4,CEP10)x2[200] nuc ish(CEP8x2,MYCx2,IGHx3)[138/200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6,RUNX1)x2[200] nuc ish(CRLF2x2)[5'CRLF2 sep 3'CRLF2x1][69/70] nuc ish(IGHx2)[5'IGH sep 3'IGHx1][192/200]	N/A, Cyto no growth	loss of partial IKZF1, loss of multiple genes on 9p including homozygous loss of CDKN2A, and alterations on chromosome 10.	N/A, Cyto no growth	loss of 7p, 9p and 10q CRLF2 (NM_022148.3.2), c.695T>G (p.Phe232Cys)	NA (no growth)
76	M	8	7/5/2021	T-ALL	46,XY,del(6)(q12q15)[20]	nuc ish(RANBP17,TLX3)x3[21/200]/(RANBP17,TLX3 x4)[30/200]/(RANBP17,TLX3)x2[149/200] nuc ish(CEP8,MYC,IGH)x3[30/200]/(CEP8,MYC,IGH)x4[20/200]/(CEP8,MYC,IGH)x2[150/200] nuc ish(ABL1,BCR)x3[24/200]/(ABL1,BCR)x4[19/200]/(ABL1,BCR)x2[157/200] nuc ish(KMT2Ax3)[25/200]/(KMT2Ax4)[20/200]/(KMT2Ax2)[155/200] nuc ish(ETV6,RUNX1)x3[18/200]/(ETV6,RUNX1)x4[20/200]/(ETV6,RUNX1)x3[162/200] nuc ish(CRLF2x3)[20/200]/(CRLF2x2)[180/200]	~24.1 Mb loss of 6q12q15	multiple mosaic copy number alterations including a ~58.9 kb loss on 1p33 including STIL, ~481.8 kb homozygous loss of 7q34 including TCRB gene, ~29.7 Mb loss of 9p24.3p21.1 including homozygous loss of CDKN2A/B, gain of ~44.2 Mb on the q arm of chromosome 13 (13q21.33q34), and a ~269 kb loss of 14q11.2 including TCRA/D genes	None	STIL:TAL1; loss of partial chromosome 1p resulting in the gene fusion; loss of partial 9p	None (Category 3)

77	M	6	7/14/2021	B-ALL	48,XY,-7,+10,der(12)t(X;12)(p11.3;p12.1),-20,+21,+21+21[17],ish(X;12)(CRF2+,ETV6)/46,XY[2]	nuc ish(CEP4x2,CEP10x3)[192/200] nuc ish(ABL1x3,BCR2x2)[32/200] nuc ish(KMT2A2x2)[200] nuc ish(ETV6x1,RUNX1x5)[124/200] nuc ish(ETV6x2,RUNX1x5)[54/200] nuc ish(ETV6x1,RUNX1x3-4)[20/200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(CRLF2x3)[199/200]	gain of Xp22.33p13.3 (~42.9 Mb in size); loss of chromosome 7; gain of chromosome 10; loss of 12p13.33p12.1 (including ETV6 and ~24 Mb in size); complex segmental alterations on chromosome 20; and gain (~5 copies) of 21q11.2q22.2 (~25.4 Mb in size)	chromosome 9 LOH; loss or LOH of 13q13.1q34 (including RB1 and ~82.5 Mb in size);	gain (~5 copies) of partial 21q involving RUNX1 and ERG; gain of partial Xp, loss of chromosome 7, gain of chromosome 10, loss of ETV6 on chromosome 12p, and loss of partial 20q.	NF1 (NM_001042492.2), c.7549C>T (p.Arg2517*) LOH of chromosomes 9, loss or LOH of chromosome 13 including RB1	None (Category 3)
78	M	17	7/16/2021	B-ALL	46,XY,del(20)(q12q13.3)[8]/46,XY[12]	nuc ish(CEP4,CEP10)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2A2x2)[200] nuc ish(ETV6,RUNX1)x2[200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(CRLF2x2)[200] nuc ish(ABL1x2)[200] nuc ish(ABL2x2)[200] nuc ish(PDGFR8x2)[200]	loss of 20q12q13.2 (17.5Mb of size) at a mosaic level of 25-30%.	copy neutral loss of heterozygosity (cnLOH) of 9p24.3p13.3 (33.9Mb of size) involving the CDKN2A gene	None	P2RY8::CRLF2 JAK2 (NM_004972.3), c.2043_2046delinsGGGGAG (p.Ile682delinsGlyGlu) JAK2 (NM_004972.3), c.2043delinsCAGCCGCC (p.Leu681_ile682insArgProPro)	None (Category 3)
79	M	7	7/25/2021	B-ALL	46,XY,tins(9;22)(q34;q11.2q11.2),dup(21)(q21q22)[14],ish fms(9;22)(ABL1+,BCR+;ABL1-,BCR+),dup(21)(RUNX1 amp)/46,XY[6]	nuc ish(CEP4,CEP10)x2[200] nuc ish(ABL1x2,BCR2x3)[ABL1 con BCRx1][84/200] nuc ish(KMT2A2x2)[200] nuc ish(ETV6x2,RUNX1amp)[196/200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(3'CRLF2x2,5'CRLF2x1)(3'CRLF2 con 5'CRLF2x1)[8/200]	an 859kb loss of 9q34.11q34.12 (including exon 1 of the ABL1 gene), a complex gain pattern involving 20.0Mb of 21q21.1q22.2 (including both RUNX1 and ERG), and a 310kb loss of 22q11.23 (including all but the first exon of the BCR gene).	a 16.6kb loss of 12q24.12 (including exon 2 of the SH2B3 gene)	iAMP21, BCR::ABL1, P2RY8::CRLF2, loss of partial 9q	SH2B3 (NM_005475.2), c.1174C>T (p.Arg327Trp) partial 12q	None (Category 3)
80	F	2	8/7/2021	B-ALL	57,XX,+X,+X,+4,+6,+9,+10,+14,+17,+21,+21,+21[10]/46,XX[10]	nuc ish(CEP4,CEP10)x3[198/200] nuc ish(ABL1x3,BCR2x2)[84/200] nuc ish(KMT2A2x2)[200] nuc ish(ETV6x2,RUNX1x5)[126/200] nuc ish(ETV6x1,RUNX1x5)[72/200] nuc ish(CEP8x2,MYC2,IGHx3)[196/200] nuc ish(CRLF2x4)[196/200]	gain of multiple whole chromosomes leading to trisomy 4, 6, 9, 10, 14, 17, tetrasomy X, and 4 copies of chromosome 21 with up to 75% mosaic level, consistent with a high hyperdiploid tumor genome.	loss of 12p13.2p13.1 (1.5Mb) involving ETV6 at 20% mosaic level and very low level (<10%) mosaic allelic imbalance for the entire chromosome 15.	Hyperdiploid genome with copy number gain of chromosomes 4, 6, 9, 10, 14, 17, 21, and X. Most of these chromosomes gained one extra copy while chromosome 21 gained three extra copies and the X chromosome was detected with two extra copies.	NRAS (NM_002524.4), c.182A>G, p.Gln61Arg PTPN11 (NM_002834.4), c.1504T>C, p.Ser502Pro PTPN11 (NM_002834.4), c.182A>T, p.Asp61Val	None (Category 3)
81	F	2	8/9/2021	AML	48,XX,+8,+9,der(9)(9;11)(p21;q23)t(9;11)(p13;p21)x2,der(11)(8;11)(p21;q23)[19],ish der(9)(5'KMT2A-3'KMT2A+),der(11)(5'KMT2A+3'KMT2A-)	nuc ish(5'KMT2A2,3'KMT2A2x4)[5'KMT2A con 3'KMT2A2x4][146/200]	gain of chromosome 8, 9p13.3q34.3 (106.7Mb of size) and 11q23.3q25 (16.5Mb), loss of 9p21.3p13.3 (13.9Mb) and 11q23.3 (22.7kb)	cnLOH of 11q23.2q23.3 (4.5Mb)	KMT2A-MLLT3. Gain of whole chromosome 8, complex CNV partial 9p, gain of 9q, complex CNV partial 11q	NRAS (NM_002524.4), c.182A>G, p.Gln61Arg TET2 (NM_001127208.2), c.4594C>T (p.Gln1532*)	None (Category 3)
82	F	15	8/16/2021	B-ALL	46,XX[20]	nuc ish(ABL1x2)[200] nuc ish(ABL2x2)[200] nuc ish(PDGFR8x2)[200] nuc ish(CEP4x3,CEP10x3)[185/200] nuc ish(CEP8x2,MYC2,IGHx4)[199/200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2A2x3)[191/200] nuc ish(ETV6x2,RUNX1x4)[193/200] nuc ish(CRLF2x3)[199/200]	None, normal karyotype	gain of one or two extra copies of chromosomes X, 4, 6, 10, 11, 14, 18, and 21, gain of 5p15.33q15.5 (95.6Mb), cnLOH of 5q15.5q35.2 (79.4Mb), gain of 5q35.2q35.3 (2.2Mb), gain of 5q35.3 (2.8Mb), loss of 7q22.1 (608.4Mb), cnLOH of 12p13.33p11.23 (27.2Mb), gain of 12p11.23q24.33 (106.4Mb), gain of 17q11.2q25.3 (55.1Mb)	None, normal karyotype	a hyperdiploid genome with copy number gains of whole chromosomes X, 4, 6, 10, 11, 14, 18, and 21, with multiple segmental chromosome aberrations involving 5p, 5q, 12p, 12q, and 17q. Most of whole chromosomes gains resulted in three copies while chromosome 14, 18 and 21 show four copies.	NA (normal karyotype likely due to growth advantage of normal cells)
83	M	3	9/11/2021	T-ALL	46,XY,del(13)(q21q22)[7],ish t(5;14)(q35;q32)(RANBP17+,TLX3+,GH+;IGH-,RANBP17-,TLX3+)[2]/46,XY[8]	nuc ish(RANBP17x2,TLX3x3)(RANBP17 con TLX3x2)[200/200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2A2x2)[200] nuc ish(ETV6,RUNX1)x2[200] nuc ish(CRLF2x2)[200]	loss of ~20.7 Mb on 13q14.2q21.33 involving multiple genes including RB1 and CYSLTR2	loss of 306 kb on 1q31.3q32.1 involving the majority of the PTPRR gene, a complex copy number variant on 9p21.3 and around ~2.5 Mb in size, involving homozygous loss of CDKN2A/B; and loss of ~54 kb involving part of the CTCF gene on 16q22.1.	loss of partial 13q including RB1	homozygous loss of CDKN2A/B on chromosome 9p and loss of exons 3-7 of CTCF on 16q DNM2 (NM_001005360.2), c.2437_2438insTT (p.Pro813Leufs*103) IL7R (NM_002185.4), c.732_733ins15 (p.Thr244_ile245insAlaTrpCysSerArg) PHF6 (NM_032458.2), c.346C>T (p.Arg116*)	None (Category 3)
84	M	2	9/22/2021	B-ALL	56,XY,+X,+4,+6,+7,+14,+17,+18,-21,+21[15]/46,XY[4]	nuc ish(CEP4x4,CEP10x2)[184/200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2A2x2)[200] nuc ish(ETV6x2,RUNX1x4)[170/200] nuc ish(ETV6x2,RUNX1x3)[20/200] nuc ish(CEP8x2,MYC2,IGHx3)[188/200] nuc ish(CRLF2x3)[190/200]	multiple whole chromosomes, including an extra copy of chromosome X, trisomy for chromosomes 6, 7, 14, 16, 17, and 18, and tetrasomy for chromosomes 4 and 21.	a segmental chromosome gain of 16p13.11, a segmental loss involving two copies of 7q34 containing TCRB, and cnLOH of whole chromosomes 2, 8, 15, and 20 were also detected	a hyperdiploid genome with copy number gain of chromosomes X, 4, 6, 7, 14, 17, 18, and 21	cnLOH impacting chromosomes 2, 8, 15, and 20 CREBBP (NM_004380.2), c.4885_4890+1delinsCCCC (p.?) KRAS (NM_033360.3), c.38G>A (p.Gly13Asp)	None (Category 3)
85	M	4	9/30/2021	B-ALL	57-59,XY,+X,+Y,add(3)(p26),+4,+5,+dd(5)(p15),+6,+8,+9,+10,+14,+18,+21,+21,+mar[cp4]/46,XY[56]	nuc ish(CEP4,CEP10)x3[53/200] nuc ish(CEP8,MYC,IGH)x3[45/200] nuc ish(ABL1x3,BCR2x2)[62/200] nuc ish(KMT2A2x2)[200] nuc ish(ETV6x2,RUNX1x4)[54/200] nuc ish(CRLF2x2)[200]	trisomy 4, 5, 6, 8, 9, 14, and 18, and tetrasomy 21.	multiple alterations on chromosome 10 including gain of 10p (~4 copies) and partial 10q (3 copies), as well as copy neutral loss of heterozygosity (cnLOH) of the terminal 10q	gain of chromosome 21	None	None (Category 3)
86	F	2	10/15/2021	B-ALL	No growth	nuc ish(CEP4,CEP10)x2[200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2A2x2)[200] nuc ish(ETV6x1,RUNX3)[ETV6 con RUNX1x1][199/200] nuc ish(CRLF2x2)[200] nuc ish(PDGFR8x2)[200] nuc ish(ABL1x2)[200] nuc ish(ABL2x2)[200]	N/A, Cyto no growth	~93 Kb, ~3.7 Mb, and ~250 Kb losses at 9p13.2, 12p13.2p12.3, and 12q21.33q22, respectively	N/A, Cyto no growth	ETV6::RUNX1 KDM6A (NM_021140.3), c.1397C>G (p.Ser466*)	NA (no growth)
87	F	11	10/18/2021	B-ALL	46,XX[19]	nuc ish(CEP4,CEP10)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2A2x2)[200] nuc ish(ETV6,RUNX1)x2[200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(CRLF2x2)[200] nuc ish(ABL1x2)[200] nuc ish(ABL2x2)[200] nuc ish(PDGFR8x2)[200]	None, normal karyotype	~115.5 kb loss on 7p12.2 involving part of the IKZF1 gene and ~57.4 kb loss on 4q12 including FIP1L1	None, normal karyotype	IGH::EPOR with EPOR overexpression	NA (normal karyotype likely due to the limited resolution)

88	M	2		10/22/2021	T-ALL	46,XY,del(6)(q12q16),del(9)(p12pter)[7]/93,idemx2,+1,7(17)(q10)[1]/46,XY[12]	nuc ish[RANBP17,TLX3]x4[12/200] nuc ish[ABL1,BCR]x4[10/200] nuc ish[KMT2Ax2]x8[200] nuc ish[ETV6,RUNX1]x4[14/200] nuc ish[CEP8,MYC,IGH]x4[10/200] nuc ish[CRLF2x2]x4[14/200]	loss of the short arm of chromosome 9 (80% mosaic), ~34.7 Mb loss of 6q12q16.1 (80% mosaic)	a segmental loss of 7q34 containing TCRB, and a ~12.4 Mb copy-neutral loss of heterozygosity in 12q24.31q24.33	loss of partial 9p	loss of partial 4q RPL10 (NM_006013.4), c.292C>A (p.Arg98Ser) NOTCH1 (NM_017617.4), c.4799T>C (p.Leu1600Pro) FBXW7 (NM_033632.3), c.1393C>T (p.Arg465Cys)	None (Category 3)
89	M	9		10/24/2021	B-ALL	46,XY[20]	nuc ish[CEP4,CEP10]x2[200] nuc ish[CEP8x2,MYCx2,IGHx3][53/200] nuc ish[ABL1,BCR]x2[200] nuc ish[KMT2Ax2]x2[200] nuc ish[ETV6,RUNX1]x2[200] nuc ish[CRLF2x2]x5[CRLF2 sep 3'CRLF2x1]60[200]	None, normal karyotype	trisomy of chromosome 14 (~75% mosaic), a homozygous loss of TCRB in 7q34, and a ~45.5 Mb copy-neutral loss of heterozygosity involving 6q12q21 (~75% mosaic). Additionally, loss of two copies of the TCRB loci on chromosomes 7q34 in a very high percentage of cells was noted	None, normal karyotype	gain of chromosome 14 NRAS (NM_002524.4), c.35G>A (p.Gly12Asp)	NA (normal karyotype likely due to growth advantage of normal cells)
90	F	1		10/26/2021	B-ALL	46,XX,del(9)(p13p24)[12]/46,XX[8]	nuc ish[CEP4,CEP10]x2[200] nuc ish[CEP8,MYC,IGH]x2[200] nuc ish[ABL1,BCR]x2[200] nuc ish[KMT2Ax2]x2[200] nuc ish[ETV6,RUNX1]x2[200] nuc ish[CRLF2x2]x2[200] nuc ish[ABL1x2]x2[200] nuc ish[ABL2x2]x2[200] nuc ish[PDGFRb2]x2[200]	~30.9 Mb loss from the short arm of chromosome 9	an interstitial loss of ~812 Kb in14q11.2 (80% mosaic) harboring the TCRA/D genes.	loss on chromosome 9p involving CDKN2A/B and PAX5	PAX5:CBAF2T3 NRAS (NM_002524.4), c.38G>A (p.Gly13Asp)	None (Category 3)
91	F	6		11/10/2021	B-ALL	46,XX,7der(6)t(6:8)(q14;q21)-8,der(11)t(8:11)(q21;q14),t(12:21)(p13;q22),+der(21)t(12:21)(11),ish 7der(6)(MYC+),der(11)(MYC+),t(12:21)(RUNX1+,ETV6-);RUNX1+,ETV6+,der(21)(RUNX1+,ETV6+),46,XX[9]	nuc ish[CEP4,CEP10]x2[200] nuc ish[CEP8x1,MYCx3,IGHx2][162/200] nuc ish[ABL1,BCR]x2[200] nuc ish[KMT2Ax1][174/200] nuc ish[ETV6x3,RUNX4][ETV6 con RUNX1x2][157/200] nuc ish[CRLF2x1][155/200]	a terminal 87.5Mb loss of 6q14.1q27 in 80% of cells, a 33.9Mb loss of 8p23.12 in 80% of cells, an 880kb gain (4 copies) of 8p12, an 18.5Mb loss of 8q11.1q12.3, a terminal 66.5Mb gain of 8q21.12q24.3, a 2.73Mb gain of 11q13.1q13.2 in 60% of cells, a 2.31Mb loss of 11q13.3 in 80% of cells, a 11.2Mb gain (3+ copies) of 11q13.3q14.1 in approximately 50% of cells, a 54.0Mb loss of 11q14.1q25 in 80% of cells, an 11.8Mb gain of 12p13.33p13.2 including the first five exons of the ETV6 gene in 85% of cells, and a 21.7Mb gain of 21q11.2	a 264kb loss of 5q33.3 (including partial EBF1) in 80% of cells, a 74kb loss of exons 2-3 of the ABL1 gene (NM_001178116.2) located on 10p12.2,	ETV6:RUNX1; gain of partial 12p (including partial ETV6) and gain of partial 21q (including partial RUNX1).	None	two derivative chromosomes (Category 2)
92	F	16		11/18/2021	T-ALL	46,XX,t(5;13)(p15.3;q21),del(5)(q21),7inv(7)(p15q11.2),7(17)(p15;p13),dup15(q24q26),del(17)(p13)[14]/46,XX[5]	nuc ish[RANBP17,TLX3]x1[128/200] nuc ish[ABL1,BCR]x2[200] nuc ish[KMT2Ax2]x2[200] nuc ish[ETV6,RUNX1]x2[200] nuc ish[CEP8,MYC,IGH]x2[200] nuc ish[CRLF2x2]x2[200]	a ~72.8 Mb loss of 5q21.3q35.3 in ~55% of cells, an 8.9 Mb loss of 17p13.3p13.1 in ~60% of cells, and a 27.8 Mb gain of 15q24.1q26.3 in ~55% of cells	None	IQGAP2:TSLP; loss of 5q and 17q and gain of 15q	TP53 (NM_000546.5) c.733G>A (p.Gly245Ser) and c.602T>A (p.Leu201*) FBXW7 (NM_033632.3) c.1394G>A (p.Arg465His) KRAS (NM_033360.3) c.35G>A (p.Gly12Asp) PRPF40B (NM_01031698.2) c.1694_1695ins15, p.Leu565_Phe566insMetThrProArgGly ETV6 (NM_001987.4) c.1396delinsCC, p.Arg399Profs*27 NOTCH1 (NM_017617.4) c.4721T>C, p.Leu1574Pro SLU12 (NM_015355.3) c.601_602insCAAAAGC (p.Cys201Serfs*12)	None (Category 3)
93	F	2		11/25/2021	B-ALL	45,XX,del(9;20)(p13.2;q11.2),del(14)(q21q31)[4]/46,XX[16]	nuc ish[CEP4,CEP10]x2[200] nuc ish[ABL1,BCR]x2[200] nuc ish[KMT2Ax2]x2[200] nuc ish[ETV6,RUNX1]x2[200] nuc ish[CEP8,MYC,IGH]x2[200] nuc ish[CRLF2x2]x2[200]	~36.8 Mb loss of 9p24.3p13.2 in 75% of cells. Additional loss of ~33 Kb on the other homologous chromosome 9 resulted in homozygous loss of partial CDKN2A/B. Additionally, a ~4.6 Mb loss was noted from 14q21.1q21.2, a ~33 Mb loss was detected from 14q21.2 to q31.1, and a ~31.8 Mb loss was detected from 20q11.21 to q13.33	None	loss of 9p and 20q	P2RY8:CRLF2 PAX5:NOL4 JAK1 (NM_002227.3), c.2108G>T (p.Ser703Ile) KRAS (NM_033360.3), c.436G>A (p.Ala146Thr) PAX5 (NM_016734.2), c.777>G (p.Val26Gly)	a dicentric chromosome (Category 2)
94	F	2		12/11/2021	B-ALL	53,XX,+x,+6,+10,+14,+15,+21,+21[6]/54,idem,+17[8]/46,XX[6]	nuc ish[CEP4x2,CEP10x3][166/200] nuc ish[ABL1,BCR]x2[200] nuc ish[KMT2Ax2]x2[200] nuc ish[ETV6x2,RUNX1x4][144/200] nuc ish[ETV6x2,RUNX1x5][20/200] nuc ish[CEP8x2,MYCx2,IGHx3][170/200] nuc ish[CRLF2x3][164/200]	trisomy of chromosomes X, 6, 10, 14, 15, and 17, and tetrasomy of chromosome 21	None	gain of whole chromosomes X, 6, 10, 14, 15, 17, and 21. All whole chromosome gains resulted in 3 copies except chromosome 21 which had 4 copies.	KRAS (NM_033360.3), c.35G>A (p.Gly12Asp) KRAS (NM_033360.3), c.183A>C (p.Gln61His) NRAS (NM_002524.4), c.182A>T (p.Gln61Leu)	None (Category 3)
95	M	2		12/17/2021	B-ALL	46,XY,t(9;17)(p27;q21),t(10;11.16)(q11.2;q17.2;q21)[12],ish t(10;11.16)(CBFB+;NUP98+;MYH11+)[46,XY[8]	nuc ish[ABL1x2]x2[200] nuc ish[ABL2x2]x2[200] nuc ish[PDGFRb2]x2[200] nuc ish[CEP4,CEP10]x2[200] nuc ish[CEP8,MYC,IGH]x2[200] nuc ish[ABL1,BCR]x2[200] nuc ish[KMT2Ax2]x2[200] nuc ish[ETV6,RUNX1]x2[200] nuc ish[CRLF2x2]x2[200]	None	complex CNVs of partial 9p include a 486kb loss of 9p21.3 involving CDKN2A (NM_00077.5) and CDKN2B (NM_004936) with homozygous loss of CDKN2B and a 21.6kb gain of 9p13.2 including partial PAX5 gene (NM_016734.3); Additionally, a 524kb loss of 7p21.1, a 2.42Mb loss of 8q12.1q12.2, a 287kb loss of 11q12.1, a 1.3Mb loss of 16q21, and a 2.1Mb loss of 17q12q21.2 were detected in about 80% of cells.	None	Complex CNV of 9p and loss of 17q	a balanced translocation and a three-way translocation (Category 2)
96	M	16		12/18/2021	AML	46,XY,t(3;21)(q21;q22)[13]/46,XY[7]	nuc ish[RUNX1T1,RUNX1]x2[200] nuc ish[KMT2Ax2]x2[200] nuc ish[MYH11,CBFB]x2[200] nuc ish[MYH11,CBFB]x2[200] nuc ish[ABL1,BCR]x2[200] nuc ish[NUP98x2]x2[200]	None	a loss of 15q15.1q15.2 (1.67 Mb) in 40% of cells.	GATA2:ERG	SETD2 (NM_014159.6), c.4715+2T>A (p.?) SF3B1 (NM_012433.3), c.2111T>A (p.Ile704Asn)	None (Category 3)
97	F	2		12/26/2021	B-ALL	46,XX,7del(9)(p13),t(12:21)(p12;q22),der(12)t(12:21)(p13;q22)[10],ish t(12:21)(RUNX1+,ETV6-);RUNX1+,ETV6+,der(12)(RUNX1+,ETV6-),46,XX[10]	nuc ish[CEP4,CEP10]x2[200] nuc ish[ABL1,BCR]x2[200] nuc ish[KMT2Ax2]x2[200] nuc ish[ETV6x1,RUNX1x3][ETV6 con RUNX1x1][96/200] nuc ish[ETV6x1,RUNX1x4][ETV6 con RUNX1x1][100/200] nuc ish[CEP8,MYC,IGH]x2[200] nuc ish[CRLF2x2]x2[200]	~32.8 Mb loss of 8p23.3p12 in 45% of cells, a ~22.9 Mb loss of 9p24.3p21.3, a ~5 Mb loss of 9p21.2p21.1, a ~3.4 Mb loss of 9p13.3p13.2, and a ~975 kb loss of 9p13.2p13.1; a ~384 kb loss of 9p13.2	loss of heterozygosity of ~3.7 Mb spanning from 9p21.3p21.2, ~2.1 Mb impacting 9p21.1p13.3, and ~3.6 kb involving 9p13.1	ETV6:RUNX1; loss of partial 12p (including partial ETV6) and 9p; gain of 21q	None	a derivative chromosome (Category 2)

98	M	1	12/27/2021	B-ALL	54~56,XY,+X,+Y,+6,+8,+10,+14,+17,18,+21,+21[cp12]/46,XY[9]	nuc ish(CEP4x2,CEP10x3)[192/200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6x2,RUNX1x4)[186/200] nuc ish(CEP8,MYC,IGH)x3[188/200] nuc ish(CRLF2x3)[30/200] nuc ish(CRLF2x3)[160/200]	two copies of chromosomes X and Y, three copies of chromosomes 6, 8, 10, 14, 17, and 18, and four copies of chromosome 21	None	whole chromosome gains of X, Y, 6, 8, 10, 14, 17, 18, 21	None	None (Category 3)
99	M	15	12/31/2021	B-ALL	No growth	nuc ish(CEP4,CEP10)x2[200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6,RUNX1)x2[200] nuc ish(CRLF2x2)[200] nuc ish(ABL1x2)[200] nuc ish(ABL2x2)[200] nuc ish(PDGFRBx2)[200]	N/A, Cyto no growth	cnLOH impacting a ~80.1 Mb region spanning across 2p25.3p11.2 in 15% of cells, as well as several segmental losses impacting 9p21.3p21.1	N/A, Cyto no growth	P2RY8::CRLF2 PAX5 (NM_016734.2), c.1176A>G (p.*392Trpext*111)	NA (no growth)
100	F	4	1/19/2021	B-ALL	No growth	nuc ish(CEP4,CEP10)x2[200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6x2,RUNX1amp)[178/200] nuc ish(CRLF2x2)[200]	N/A, Cyto no growth	a copy gain of whole chromosome 21	N/A, Cyto no growth	a CNV pattern consistent with atypical iAMP21 KRAS (NM_004985.3), c.467T>C (p.Phe156Ser)	NA (no growth)
101	M	3	1/19/2021	B-ALL	No growth	nuc ish(CEP4,CEP10)x2[200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6x1,RUNX1x2)[ETV6 con RUNX1x1][199/200] nuc ish(CRLF2x2)[200]	N/A, Cyto no growth	a 103.5 kb loss on 9p13.2 involving part of the PAX5 gene in ~35% of cells as well as a 33.6 Mb loss on 12p13.33p11.1 involving multiple genes including ETV6 in ~75% of cells.	N/A, Cyto no growth	ETV6 (NM_001987.4) exon 5 - RUNX1 (NM_001754.4) exon 3 Loss on 9p (partial deletion of the PAX5 gene) and loss on chromosome 12p.	NA (no growth)
102	M	2	10/24/2022	AML	47,XY,der(10)t(10;11)(p12;q23),der(11)t(10;11)(p12;q23)del(11)(q22.3q23.3)+der(17)t(17)(p13p11.2)del(17)(q11.2q21.1)[9]ish t(10;11)(p12;q23)del(11)(q22.3q23.3)+der(17)t(17)(p13p11.2)del(17)(q11.2q21.1)[9]ish	nuc ish(RUNX1T1,RUNX1)x2[200] nuc ish(NUP98x2)[200] nuc ish(MYH11,CBFβ)x2[200] nuc ish(KMT2Ax2)[200]ish t(10;11)(p12;q23)del(11)(q22.3q23.3)+der(17)t(17)(p13p11.2)del(17)(q11.2q21.1)[9]ish	None, array not performed	None, array not performed	KMT2A::MLLT10	ASXL1 (NM_015338.6), c.1934dup, (p.Gly646Trpfs*12) KRAS (NM_004985.5), c.36_38dup, (p.Gly13dup) loss of partial 3p including exons 2-21 of the SETD2 gene (NM_014159.6) and gain of partial 17q	None (Category 3)
103	M	8	1/7/2022	B-ALL	47,XY,+21[7]/46,XY[13]	nuc ish(CEP4,CEP10)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6x2,RUNX1x3)[110/200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(CRLF2x2)[200]	gain of whole chromosome 21	None	gain of whole chromosome 21	PAX5 (NM_016734.2), c.113G>A (p.Arg38His) CREBBP (NM_004380.2), c.5509A>T (p.Lys1837*) IKZF1 (NM_006060.5), c.475A>T (p.Asn159Tyr)	None (Category 3)
104	F	19	1/8/2022	B-ALL	46,XX[20]	nuc ish(CEP4,CEP10)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6,RUNX1)x2[200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(CRLF2x2)[200] nuc ish(ABL1x2)[200] nuc ish(ABL2x2)[200] nuc ish(PDGFRBx2)[200]	None, normal karyotype	a small gain of chromosome 22q13.2 involving partial gene of EP300 and loss of chromosome4q11.2 involving the T-cell receptor (TCR) region.	None, normal karyotype	NRAS (NM_002524.4), c.182A>C (p.Gln61Pro) partial chromosome 22q gain involving part of the EP300 gene	NA (normal karyotype)
105	M	3	1/12/2022	B-ALL	No growth	nuc ish(CEP4x3,CEP10x2)[144/200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6x2,RUNX1x3)[146/200] nuc ish(CEP8,MYC,IGH)x3[150/200] nuc ish(CRLF2x3)[146/200]	N/A, Cyto no growth	a high hyperdiploid tumor genome with gains of whole chromosomes X, 4, 6, 8, 14, 17, and 21. In addition, copy neutral loss of heterozygosity (cnLOH) of ~36.3 Mb ranging from 9p24.3p13.2 was detected. Within this region of cnLOH, there is a 148 kb homozygous loss involving CDKN2A and CDKN2B. Finally, an intragenic deletion impacting CREBBP was noted on 16p13.3.	N/A, Cyto no growth	a high hyperdiploid tumor genome with gains of whole chromosomes X, 4, 6, 8, 14, 17, and 21. KRAS (NM_033360.3), c.35G>A (p.Gly12Asp) homozygous (two copy) loss of CDKN2A/B on 9p and loss of partial CREBBP on 16p	NA (no growth)
106	F	13	2/7/2022	B-ALL	49,X,-X,+1,t(1;19)(q23;p13.3),der(1)t(1;10)(p33;q24),+6,+8,+8,de(9)[p21p22]15,ish t(1;19)(PBX1+,TCF3+,PBX1+,TCF3+),46,XX[5]	nuc ish(CEP4,CEP10)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6,RUNX1)x2[200] nuc ish(CEP8,MYC,IGH)x2[164/200] nuc ish(CRLF2x1)[120/200] nuc ish(PBX1x4,TCF3x3)(PBX1 con TCF3x2)[162/200]	loss of X; gain of 1p33q44 (200.0Mb), 6, 8	loss of 5q33.3 (95kb), 9p22.3p21.1 (14.5Mb, including CDKN2A/28) and 9p13.2 (261.1kb, including PAX5), gain of 10q24.2q26.3 (35.7Mb), as well as copy-neutral loss of heterozygosity (cnLOH) of 17q11.2q12 (8.1Mb) and 17q21.31q25.3 (37.1Mb).	TCF3::PBX1; gain of chromosomes partial 1p, 1q, 6, 8 and partial 10q, loss of partial 9p and X,	cnLOH of 17q TP53 (NM_000546.5), c.844C>T (p.Arg282Trp) TP53 (NM_000546.5), c.845G>C (p.Arg282Pro)	a derivative chromosome (Category 2)
107	F	15	2/10/2022	AML	46,XX,t(6;12)(p21;p13)[12]/46,t(6;12)13(p21;p13;q17)[10]/46,XX,der(7)(7;7)(p15;p15)or del(7)(p15)[5]	nuc ish(RUNX1T1,RUNX1)x2[500]	None, array not performed	None, array not performed	ETV6::TAF8	WT1 (NM_024426.5), c.1425_1428delinsTTGTTT (p.Arg76delinsCysPhe) WT1 (NM_024426.5), c.1146_1153dup (p.Arg385Leufs*77) NRAS (NM_002524.4), c.35G>A (p.Gly12Asp)	None (Category 3)
108	F	13	2/18/2022	B-ALL	No growth	nuc ish(CEP4x1,CEP10x2)[120/200] nuc ish(ABL1,BCR)x2[80/200] nuc ish(ABL1,BCR)x1[40/200] nuc ish(KMT2Ax2)[198/200] nuc ish(ETV6x1,RUNX1x2)[116/200] nuc ish(CEP8,MYC,IGH)x2[198/200] nuc ish(CRLF2x2)[200]	N/A, Cyto no growth	loss of whole chromosomes X, 2, 3, 4, 7, 9, 10, 12, 13, 15, 16, 17, and 20, indicating a low-hypodiploidy. In addition, multiple segmental losses were detected on chromosomes 5, 10, and 22.	N/A, Cyto no growth	loss of whole chromosomes X, 2, 3, 4, 7, 9, 10, 12, 13, 15, 16, 17, and 22 RB1 (NM_000321.2), c.192_193insGCCCT (p.Lys65Alafs*14) TP53 (NM_000546.5), c.659A>C (p.Tyr220Ser)	NA (no growth)
109	M	14	2/18/2022	B-ALL	46,XY,del(9)(p21.3p22.1)[10]/46,XY[10]	nuc ish(CEP4,CEP10)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6,RUNX1)x2[200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(CRLF2x2)[200] nuc ish(ABL1x2)[200] nuc ish(ABL2x2)[200] nuc ish(PDGFRBx2)[200]	a complex alteration on chromosome 9p with regions of copy neutral loss of heterozygosity (as listed in the table), as well as loss of CDKN2A/B	None	One region of copy number loss and loss of heterozygosity (LOH) with potential clinical significance (Tier 2) on chromosome 9p and including loss of CDKN2A/B	KRAS (NM_033360.3), c.179G>A (p.Gly60Asp) ATRX (NM_000489.4), c.3957del (p.Val1320Serfs*26)	None (Category 3)

110	M	4	2/19/2022	AML	47,XY,(7)(q10),+21c,+21(13)/47,XY,+21c[7]	nuc ish(RUNX1T1x2,RUNX1x3)[102/200] (RUNX1T1x2,RUNX1x4)[98/200] nuc ish(KMT2Ax2)[200] nuc ish(NUP98x2)[200] nuc ish(MYH11,CBFβ)x2[200]	loss of 7p, gain of 7q and chromosome 21	loss of heterozygosity of partial chromosome 9p	Trisomy 21, loss of 7p, gain of 7q	GATA1 (NM_002049.3), c.151_186delInsT (p.Ser51Leufs*5) EPOR (NM_000121.3), c.1316G>A (p.Trp439*) KMT2A (NM_005933.3), c.6478C>T (p.Arg2160*)	an isochromosome (Category 2)
111	F	2	3/9/2022	AML	47,XX,+21c[19]	nuc ish(RUNX1T1x2,RUNX1x3)[200] nuc ish(KMT2Ax2)[200] nuc ish(NUP98x2)[200] nuc ish(MYH11,CBFβ)x2[200]	non-mosaic gain of chromosome 21	a 46.5 Mb gain of 2p25.3p21; a 5.7 Mb gain of 5q21.2q21.3; loss of whole chromosome 9 except for a 1.4 Mb gain of 9p22.2p22.1, and a 34.1 Mb region of copy neutral loss of heterozygosity across 11p15.5p13	Gain of chromosome 21	GATA1 (NM_002049.4), c.-13_144dup (p.Ala49Profs*43) CTCF (NM_006565.3), c.610dup (p.Thr204Asnfs*26) STAG2 (NM_006603.4), c.1197T>G>A (p.p.7) TERT (NM_198253.2), c.-124C>T (p.7) MPL (NM_005373.2), c.1544G>T (p.Trp515Leu) gain of partial 2p, loss of whole 9, c.NL0H11p	None (Category 3)
112	F	3	3/17/2022	B-ALL	No growth	nuc ish(CEP4,CEP10)x3[110/200] nuc ish(ABL1,BCR)x3[110/200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6x2,RUNX1x4)[102/200] nuc ish(CEP8x3,MYCx3,IGHx4)[114/200] nuc ish(CRLF2x3)[112/200]	N/A, Cyto no growth	multiple whole chromosome gains at a mosaic level up to 50%, including chromosomes X, 4, 8, 9, 14, 17, 18, 21 and 22; in addition, copy-neutral loss of heterozygosity of chromosome 12 as well as multiple segmental rearrangements involving 10p and 10q were detected	N/A, Cyto no growth	multiple whole chromosome gains of chromosomes X, 4, 8, 9, 12, 14, 17, 18, 21 and 22; gain of 10p and partial 10q ASXL2 (NM_018263.5), c.1206_1210del (p.Lys402Asnfs*4)	NA (no growth)
113	M	3	3/21/2022	B-ALL	46,XY,del(12)(p13p13),t(12;21)(p13;q22)[19],ish del(12)(ETV6-),t(12;21)(RUNX1+ETV6-;RUNX1+ETV6+)/46,XY[1]	nuc ish(CEP4,CEP10)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6x1,RUNX1x3)[ETV6 con RUNX1x1][194/200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(CRLF2x2)[200] nuc ish(ABL1x2)[200] nuc ish(ABL2x2)[200] nuc ish(PDGFRβx2)[200]	loss of 12p13.2p13.1 (involving ETV6)	losses of 3p26.1 and 18q21.2	ETV6:RUNX1; Loss of 12p	UBA2 (NM_005499.2), c.157G>T (p.Asp53Tyr) loss of 4p	None (Category 3)
114	M	3	3/22/2022	B-ALL	55,XY,+x,+y,+5,+6,+8,+14,+17,+18,+21[8]/56,idem,+7[2]/46,XY[15]	nuc ish(CEP4,CEP10)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6x2,RUNX1x3)[116/200] nuc ish(CEP8,MYC,IGH)x3[118/200] nuc ish(CRLF2x4)[120/200]	gains of whole chromosomes X, Y, 5, 6, 7, 8, 14, 17, 18 and 21.	copy neutral loss of heterozygosity (cnLOH) of 9p24.3p21.3 (21.8Mb) and 9p21.3 (15.8Mb) involving PAX5 as well as loss of 9p21.3 (174.9kb) involving CDKN2A were detected	multiple whole chromosome gains, including chromosomes X, Y, 5, 6, 7, 8, 9, 14, 17, 18, and 2	KRAS (NM_033360.3), c.346G>T (p.Gly12Cys) KRAS (NM_033360.3), c.35G>C (p.Gly12Ala) complex CNV of partial 9p	None (Category 3)
115	F	5	4/5/2022	B-ALL	46,XX,7(1;6)(p3?;q27?),t(12;21)(p13;q22),del(12)(p13)[5],ish t(12;21)(RUNX1+ETV6-;46,XX,del(7)(q22q36),del(9)(p13p22;RUNX1+ETV6+),del(12)(ETV6-)/46,XX[25]	nuc ish(CEP4,CEP10)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6x1,RUNX1x3)[ETV6 con RUNX1x1][28/200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(CRLF2x2)[200]	None, array(X,1-22)x2	None, array(X,1-22)x2	ETV6:RUNX1	None	a balanced translocation (Category 2)
116	M	6	5/4/2022	B-ALL	46,XY[25]	nuc ish(CEP4,CEP10)x3[130/200] nuc ish(ABL1x3,BCR)x2[130/200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6x3,RUNX1x4)[128/200] nuc ish(CEP8,MYC,IGH)x3[132/200] nuc ish(CRLF2x4)[134/200]	None, normal karyotype	gains of whole chromosomes 4, 5, 6, 8, 9, 10, 12, 14, 17, 18, 21 (4 copies), X, and Y. Additionally, a 71Mb gain of 16p12.3 to q24.3 was detected.	None, normal karyotype	multiple whole chromosome gains including two copies of chromosomes X and Y, three copies of chromosomes 4, 6, 8-10, 12, 14, 17, and 18, as well as four copies of chromosome 21; gain of 16q ETV6 (NM_001987.4), c.416_417del (p.Ser139Tyrfs*14)	NA (normal karyotype likely due to growth advantage of normal cells)
117	F	15	5/4/2022	AML	46,XX,der(5)(pter->5q22::5q34->75qter),der(10)(11qter->11q23::10p12->10qter),der(11)(11pter->11q23::?10p12->?10pter::11q23->11q14::5q34->5q22::18q21.1->?18qter),der(11)(11pter->11q14::11q23->11qter),der(18)(18pter->18q21.1->?14)[47],idem,+i(1)(q10)[4]/46,XX[1],ish der(5)(D5S23/D5S721+,CSF1R-),der(10)(11qter+,3'KMT2A+),der(11)(NUP98+,5'KMT2A+,KMT2A+,CSF1R+,BCL2+,D5S23/D5S721-,11qter-),der(11)(NUP98+,KMT2A-,11qter-),der(18)(BCL2-)	nuc ish(RUNX1T1,RUNX1x2)[200] nuc ish(KMT2Ax2)[5'KMT2A sep 3'KMT2Ax1][194/200] nuc ish(NUP98x2)[200] nuc ish(MYH11,CBFβ)x2[200]	a gain of 1q, large whole chromosome arm gain, several segmental alterations were detected, including a 2,084 kb loss of 5q22.2q22.3, a 219 kb loss of 5q22.3, a 1,934 kb loss of 10p12.31 involving MLLT10, a 261 kb loss of 10p12.2, a 371 kb loss of 11q22.2, a 75 kb loss of 11q23.3 involving KMT2A, a 361 kb loss of 11q24.3, a 588 kb loss of 18q11.2, and two separate segmental losses on 18q12.1	None	KMT2A:MLLT10, gain of 1q and loss of partial 11q involving part of KMT2A	NRAS (NM_002524.4), c.386G>A (p.Gly13Asp) KRAS (NM_033360.3), c.386G>A (p.Gly13Asp)	a complex rearrangement involving multiple chromosomes (Category 2)
118	M	11	5/5/2022	B-ALL	46,XY,del(6)(q15q722),del(12)(p12p13)[8]/46,XY[12]	nuc ish(CEP4,CEP10)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6x1,RUNX1x2)[72/200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(CRLF2x2)[200] nuc ish(ABL1x2)[200] nuc ish(ABL2x2)[200] nuc ish(PDGFRβx2)[200]	segmental chromosome losses involving chromosomes 6q, and 12p including ETV6 gene	segmental chromosome losses involving chromosomes 1p, 5q, 13q, and 17q	loss of 12p	IKZF1 (NM_006060.5), c.970_985del (p.Ala324Argfs*86) losses of 5q, 13q, and 17q	None (Category 3)
119	M	12	5/17/2022	AML	46,XY,t(9;11)(p21;q23)[20],ish t(9;11)(5'KMT2A-;3'KMT2A+;5'KMT2A+,3'KMT2A-)	nuc ish(RUNX1T1,RUNX1x2)[200] nuc ish(KMT2Ax2)[3'KMT2A sep 5'KMT2Ax1][198/200] nuc ish(NUP98x2)[200] nuc ish(MYH11,CBFβ)x2[200] nuc ish(NUP98x2)[200]	None, arr(X,Y)x1,(1-22)x2	None, arr(X,Y)x1,(1-22)x2	KMT2A:MLLT3	None	None (Category 3)
120	M	14	5/21/2022	AML	46,XY[20]	nuc ish(RUNX1T1,RUNX1x2)[200] nuc ish(KMT2Ax2)[200] nuc ish(MYH11,CBFβ)x2[200] nuc ish(NUP98x2)[200]	None, arr(X,Y)x1,(1-22)x2 and normal karyotype	None, arr(X,Y)x1,(1-22)x2	None, normal karyotype	CEBPA (NM_004364.4), c.68dup (p.His244Iaf*84) CEBPA (NM_004364.4), c.924_936del (p.Gln312del) GATA2 (NM_001145661.1), c.1085G>A (p.Arg362Gln) PHF6 (NM_032458.2), c.27dup (p.Gly10Argfs*12) CSF3R (NM_00760.3), c.1853C>T (p.Trp618Ile)	NA (normal karyotype)

121	F	1	6/14/2022	AML	46,XX,t(9;11)(p21,q23)[18].ish t(9;11)(3'KMT2A+,5'KMT2A-;5'KMT2A+,3'KMT2A-)/46,XX[2]	nuc ish(RUNX1T1,RUNX1x2)[200] nuc ish(KMT2Ax2)[84/200] nuc ish(NUP98x2)[200] nuc ish(MYH11,CBFβ)[200]	None, array(X,1-22)x2	None, array(X,1-22)x2	KMT2A::MLLT3	None	None (Category 3)
122	F	17	10/10/2022	B-ALL	45,XX,-7,7del(12)(p12pter),der(21)dup(21)q21.2del(21)(q21)ish der(21)(RUNX1amp),del(X)(p22.33)3'CRF2+,5'CRF2-;/46,XX[13]	nuc ish(CEP4,CEP10)x2[200] nuc ish(CEP8x2,MYCx2,IGHx3)[10/200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6x2,RUNX1x4)[199/200] nuc ish(ETV6x1,RUNX1x3)[ETV6 con RUNX1x1][198/200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(CRF2x2)[200/200]	at least five copies of a 23.2 Mb region encompassing RUNX1 on 21q21.2, loss of whole chromosome 7	copy neutral loss of heterozygosity across 12p13.3p12.3 encompassing ETV6	intrachromosomal amplification of 21 (IAMP21), F2RY8::CRF2	JAK1 (NM_002227.3), c.1972G>T (p.Val658Phe) JAK2 (NM_004972.3), c.2617G>A (p.Asp873Asn) NF1 (NM_001042492.2), c.2033dup (p.Ile679Aspfs*21) IKZF1 (NM_006605.5), c.556G>C (p.Asp186His) losses of 3p, 5q, and 17q	None (Category 3)
123	F	22	12/19/2022	AML	88-90,XXX[cp3]/46,XX[17]	nuc ish(RUNX1T1,RUNX1x4)[64/200] nuc ish(KMT2Ax4)[59/200] nuc ish(MYH11,CBFβ)x4[60/200] nuc ish(NUP98x4)[61/200]	None, array(X,1-22)x2	None, array(X,1-22)x2	None	USHA2 (NM_206933.2), c.8168_8171dup (p.Ala2725Thrfs*33) SRSF2 (NM_003016.4), c.284_307del (p.Pro95_Arg102del)	a near tetraploid genome (Category 2)
124	M	4	12/19/2022	B-ALL	No growth	nuc ish(CEP4,CEP10)x2[200] nuc ish(CEP8x2,MYCx2,IGHx4)[188/200] nuc ish(ABL1x3,BCRxx2)[199/200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6x2,RUNX1x4)[199/200] nuc ish(CRF2x3)[200/200]	N/A, Cyto no growth	gain of one extra copy of chromosomes X, 9, and 14, and gain of two extra copies of chromosome 21 (tetrasomy)	N/A, Cyto no growth	gain of one extra copy of whole chromosomes 9, 14, and X and gain of two extra copies of whole chromosome 21	NA (no growth)
125	M	3	12/20/19	B-ALL	46,XY[20]	nuc ish(CEP4,CEP10)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6x2,RUNX1x4)[199/200] nuc ish(CRF2x3)[200/200]	None, normal karyotype	a 347Kb heterozygous loss within 5p33.3 involving EBF1 (apparently non-mosaic), an 18Mb duplication of the short arm of chromosome 10 involving GATA3 (~50% mosaicism), and a complicated pattern of copy number loss involving the short arm of chromosome 12	None, normal karyotype	ETV6::RUNX1 CTCF (NM_006565.3), c.855_856insGTGGCCG (p.Lys286Valfs*7) 3 CNVs	NA (normal karyotype likely due to the limited resolution)
126	M	7	12/16/19	B-ALL	51,XY,+x,+4,+14,+21,+21[11]/51,ide m,der(1)7dup(1)(q732q741)7del(1)(q74)[4]/46,XY[5].arr[GRCh37] 1q32.1q41[200]509197_2207625241 x3[0.15],1q41q44[220801974_249238992]x1[0.15]	nuc ish(CEP4x3,CEP10x2)[194/200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(CEP8x2,RUNX1x4)[196/200] nuc ish(CEP8x2,MYCx2,IGHx3)[190/200] nuc ish(CRF2x3)[194/200]	trisomy of chromosomes X, 4, 14 (~75% mosaicism), tetrasomy of chromosome 21 (~75% mosaicism), ~20Mb gain of chromosome 1q32.1q41, ~28Mb loss of chromosome 1q41q44 (~15% mosaic)	~114Kb loss of chromosome 9p13.2 involving PAX5 (~95%)	gain of chromosomes 4 (3 copies), 14 (3 copies), 21 (4 copies), X (2 copies), and loss of partial 9p (including PAX5 exons 7-8)	TET2 (NM_001127208.2), c.1379C>T (p.Ser460Phe), FLT3 (NM_004119.2), c.1768_1776delinsCCGAGTGGG (p.Phe590_Val592delinsProSerGly), CREBBP (NM_004380.2), c.4303G>A (p.Asp1435Asn),	None (Category 3)
127	F	3	12/3/19	B-ALL	47,XX,del(9)(p21p22),-13,de[20](q11.2),+21,+21[16]/46,XX[4]	nuc ish(ETV6x2,RUNX1x4)[177/200]	copy number gains of whole chromosome 21, segmental rearrangements involving chromosomes 9p and 20q	loss of whole chromosome X, as well as multiple segmental rearrangements involving chromosomes 5q, 6q, 8p, 13q, 16q, 20p	losses of 9p and 20q, gain of 21q	loss of whole chromosome X, complex 13q	None (Category 3)
128	F	1	11/22/19	B-ALL	55,XX,+x,+6,+9,+14,+15,+17,+18,+21,+21[8]/46,XX[12]	nuc ish(CEP4,CEP10)x2[200] nuc ish(ABL1x3,BCRxx2)[148/200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6x2,RUNX1x4)[175/200] nuc ish(CEP8x2,MYCx2,IGHx3)[162/200] nuc ish(CRF2x3)[130/200]	None, array not performed	None, array not performed	high hyperdiploidy	PTPN11 (NM_002834.4), c.1508G>T (p.Gly503Val)	None (Category 3)
129	M	2	11/12/19	B-ALL	46,XY,7del(9)(p22),t(12;21)(p13;q22)ish t(12;21)(RUNX1+,ETV6-;RUNX1+,ETV6+)/46,XY[8]	nuc ish(ETV6x2,RUNX1x3)[ETV6 con RUNX1x1][124/200] [ETV6x2,RUNX1x4][ETV6 con RUNX1x1][60/200]	loss of 9p13.2 (including exons 1-7 of PAX5)	loss of 2q37.1q37.2, 4q31.21, 7p12.2 (including exons 1-2 of IKZF1), 10q24.1, 11q23.3 (including POU2AF1, SDHD, PAFAH1B2, ZBTB16), and gain of whole chromosome 21	ETV6::RUNX1, loss of 9p	loss of 7p, gain for whole chromosome 21	None (Category 3)
130	F	21	16/1/19	T-ALL	46,XX[20]	nuc ish(CEP4,CEP10)x2[200] nuc ish(ABL1 amp,BCRxx2)[86/200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6,RUNX1)x2[200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(RANBP17,TLX3)x2[200]	None, normal karyotype	The main CNVs are an amplification involving a 479kb region on 9q34.12q34.13 including both ABL1 and NUP214 genes, overlapping deletions on 9p21.3 resulting in biallelic loss of CDKN2A/B, a 58kb loss on 1p33 including the STIL gene. Additionally, multiple small deletions were also observed on many other chromosomes resulting in heterozygous loss of WT1 and biallelic loss of RB1.	None, normal karyotype	NUP214::ABL1, STIL::TAL1, homozygous loss of partial 9p (including CDKN2A/B) and partial 13q (including RB1), NOTCH1 (NM_017617.4), c.4744_4746del p.Pro1582del	NA (normal karyotype)
131	M	7	10/16/19	B-ALL	51,XY,+x,+4,+8,+14,+21[12]/52,ide m,+mar[8]	nuc ish(CEP4x3,CEP10x2)[190/200] nuc ish(ABL1x3,BCRxx2)[22/200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6x2,RUNX1x3)[185/200] nuc ish(CEP8,MYC,IGH)x3[195/200] nuc ish(CRF2x3)[188/200]	None, array not performed	None, array not performed	hyperdiploid	SETD2 (NM_014159.6), c.3911_3912insCCGG (p.Tyr1305Argfs*9) KRAS (NM_03360.3), c.38G>A (p.Gly13Asp) KRAS (NM_03360.3), c.35G>A (p.Gly12Asp) Two copy-neutral loss of heterozygosity	None (Category 3)
132	M	2	10/5/19	B-ALL	57,XY,+x,+4,+der(4)t(1;4)(q27;q27)1,6,+8,+10,+14,+18,+18,+21,+21[10]/46,XY[10]	nuc ish(CEP4x4,CEP10x3)[192/200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6x2,RUNX1x4)[174/200] [ETV6x2,RUNX1x3][16/200] nuc ish(CEP8,MYC,IGH)x3[190/200] nuc ish(CRF2x3)[186/200]	None, array not performed	None, array not performed	hyperdiploid	FLT3 (NM_004119.2), c.1727T>C (p.Leu576Pro)	a derivative chromosome (Category 2)
133	M	3	8/28/19	B-ALL	55,XY,+x,+4,+6,+8,+10,del(12)(p13)1,+14,+17,+18,+21[1]/56,idem,+21[1]/46,XY[18]	nuc ish(CEP4,CEP10)x3[153/200]/(CEP4x3,CEP10x2)[13/200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6x1,RUNX1x4)[155/200]/(ETV6x1,RUNX1x3)[23/200] nuc ish(CEP8,MYC,IGH)x3[153/200]/(CEP8x3,MYCx3,IGHx2)[24/200] nuc ish(CRF2x3)[187/200]	None, array not performed	None, array not performed	gain of chromosomes 4, 6, 8, 10, 14, 17, 18, and X and tetrasomy 21, loss of partial 12p including the ETV6 gene	None	None (Category 3)
134	M	0	7/20/19	AML	47,XY,der(16)t(11;16)(q13;q24),add(21)(p11.2),+21c[12].ish der(16)(CBFB+,16qtel+,KMT2A+),add(21)(RUNX1+)/47,XY,+21c[8]	nuc ish(RUNX1T1x2,RUNX1x3)[200] nuc ish(KMT2Ax3)[36/200]	None, array not performed	None, array not performed	Trisomy 21	GATA1 (NM_002049.3), c.3G>A (p.?)	a derivative chromosome (Category 2)

135	F	3	7/15/19	B-ALL	46,X,-X,del(6)(q715q21),+10,t(12;21)(p13;q22),del(12)(p13)[18].ish t(12;21)(RUNX1+,ETV6-);RUNX1+,ETV6+,de(12)(ETV6-)/46,XX[1]	nuc ish(CEP4x2,CEP10x3)[164/200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6x1,RUNX1x3)[ETV6 con RUNX1x1][196/200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(CRLF2x2)[194/200] nuc ish(CEP4,CEP10)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6x1,RUNX3)[ETV6 con RUNX1x1][200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(CRLF2x2)[200]	None, array not performed	None, array not performed	ETV6:RUNX1, loss of partial 12p (involving the ETV6 gene), gain of whole chromosome 10 and loss of whole chromosome X.	None	None (Category 3)
136	M	1	7/5/19	B-ALL	No growth	nuc ish(CEP4,CEP10)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6x1,RUNX3)[ETV6 con RUNX1x1][200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(CRLF2x2)[200]	None, array not performed	None, array not performed	N/A, Cyto no growth	ETV6:RUNX1, loss of 9p and loss of 12p	NA (no growth)
137	M	2	6/26/19	B-ALL	46,XY[19]	nuc ish(CEP4x3,CEP10x3)[110/200]/(CEP4x3,CEP10x2)[66/200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax3)[170/200] nuc ish(ETV6x3,RUNX1x4)[120/200]/(ETV6x3,RUNX1x5)[72/200] nuc ish(CEP8,MYC,IGHx3)[164/200] nuc ish(CRLF2x3)[187/200]	None, array not performed	None, array not performed	None, normal karyotype	KMT2D (NM_003482.3), c.11147dup (p.Leu3716Phefs*296) an extra copy of chromosome X, a complex CNV on chromosome 1 including gain of 1q and copy neutral loss of heterozygosity on 1p, trisomies 4, 6, 8, 10, 11, 12, 14, 17, 18 and tetrasomy chromosome 21.	NA (normal karyotype likely due to growth advantage of normal cells)
138	M	12	6/17/19	AUL	46,XY,der(6)t(6;11;17)(6pter->6q26::17q25->17qter),der(11)t(6;11;17)(11pter->11q23::6q27->6qter),der(11)t(11;17)(11pter->11q22::17p11.2->17pter),der(17)t(6;11;11;17)(11qter->11q22::17p11.2->17q25::6q26->6q27::11q23->11qter)[20].ish der(11)(5'KMT2A+,3'KMT2A-),der(11)(KMT2A-),der(17)(5'KMT2Ax1,3'KMT2Ax2)(5'KMT2A con 3'KMT2Ax1)	nuc ish(KMT2Ax2)(5'KMT2A sep 3'KMT2Ax3)[168/200] nuc ish(KMT2Ax3)(5'KMT2A con 3'KMT2Ax2)[22/200] nuc ish(KMT2Ax3,3'KMT2Ax2)(5'KMT2A con 3'KMT2Ax2)[10/200] nuc ish(CEP4,CEP10)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(ETV6,RUNX1)x2[200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(CRLF2x2)[200]	The changes on the chromosome 11 included a 10.53Mb loss or LOH of 11q22.32q23.3 (low mosaicism) and a 19.86Mb gain of 11q23.3q25 (20-25% mosaicism). Chromosomal abnormalities involving the chromosome 17 include a 34.46Mb loss or c.nLOH of 17p13.3q12 (5-10% mosaicism), a 1.38Mb loss of 17q11.2 (65-75% mosaicism), and a 46.24Mb gain of 17q12q25.3 (20-25% mosaicism). A 314kb deletion on 6q26 was also identified. Additional findings include a 9.59Mb c.nLOH on 2q37.1 (5-10% mosaicism) and a 229kb gain of 12p13.33 involving KDM5A.	None	KMT2A:MLLT4, loss of partial 17q including the NF1 gene.	FLT3 (NM_004119.2), c.1798_1799insGGATCCCGATTTCAGAGAATGAATG (p.Tyr599_Asp600insGlyIleProAspPheArgIuTyGluTyr) SUZ12 (NM_015355.3), c.1786dup (p.Trp596Asnfs*6) ASXL1 (NM_015338.3), c.1934dupG (p.Gly646Trpfs*12) RUNX1 (NM_001754.4), c.940_950delinsCCA (p.Ser14Prof*283) BCORL1 (NM_021946.4), c.3496dup (p.Ala1166Glyfs*50) PHF6 (NM_032458.2), c.826delinsGGCGCT (p.Lys276Glyfs*5)	a derivative chromosome (Category 2)
139	M	2	6/3/19	B-ALL	No growth	nuc ish(CEP4,CEP10)x2[194/200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6x1,RUNX1x2)[ETV6 con RUNX1x1][160/200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(CRLF2x2)[193/200]	None, array not performed	None, array not performed	N/A, Cyto no growth	ETV6:RUNX1, KRAS (NM_033360.3), c.64C>G (p.Gln22Glu) partial loss of chromosome 12p including ETV6	NA (no growth)
140	F	2	6/6/19	B-ALL	46,XX,7ins(9;22)(q34;q13q11.2)[19].ish ins(9;22)(ABL1+,BCR+;ABL1-,BCR-)[5]/46,XX[1]	nuc ish(CEP4,CEP10)x2[200] nuc ish(ABL1,BCR)x2[ABL1 con BCRx1][156/200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6,RUNX1)x2[200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(CRLF2x2)[200]	None, array not performed	None, array not performed	BCR:ABL1	None	an insertion (Category 2)
141	M	7	6/6/19	B-ALL	46-47,XY,der(1)(X:1)(q22;p36),+10,del(12)(p13),t(12;21)(p13;q22)[cp 19].ish del(12)(ETV6-),t(12;21)(RUNX1+,ETV6-);RUNX1+,ETV6+/46,XY[1]	nuc ish(CEP4x2,CEP10x3)[50/200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6x2,RUNX1x3)[ETV6 con RUNX1x1][138/200] nuc ish(ETV6x1,RUNX1x3)[ETV6 con RUNX1x1][60/200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(CRLF2x2)[200]	None, array not performed	None, array not performed	ETV6:RUNX1; partial loss of 12p and partial gain of Xq	None	a derivative chromosome (Category 2)
142	F	9	1/6/2021	B-ALL	No growth	nuc ish(CEP4,CEP10)x2[200] nuc ish(CEP8x2,MYCx2,IGHx3)[116/200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6x1,RUNX1x4)[ETV6 con RUNX1x1][199/200] nuc ish(CRLF2x2)[200]	N/A, Cyto no growth	multiple copy number variations (CNVs) including loss of partial 12p involving ETV6, partial 12q involving BTG1, and partial 5p, and gain of partial Xq, trisomy 14, and trisomy 21.	N/A, Cyto no growth	ETV6:RUNX1; a small percentage of supporting reads suggested a P2RY8:CRLF2 fusion gene as well; multiple cytogenetics alterations in the bone marrow specimen including loss of ETV6 on chromosome 12p and gain of chromosome 21.	NA (no growth)
143	M	17	1/6/2021	B-ALL	64-65,XY,+x,+3,+4,+5,+6,+7,+8,+9,+10,+11,+12,+14,+15,+16,+17,+18,+21,+21,+21[cp13]/46,XY[7]	nuc ish(CEP4,CEP10)x3[194/200] nuc ish(CEP8x3,MYCx3,IGHx4)[192/200] nuc ish(ABL1x3,BCR)x2[158/200] nuc ish(KMT2Ax3)[192/200] nuc ish(ETV6x3,RUNX5)[199/200] nuc ish(CRLF2x3)[150/200]/(CRLF2x4)[45/200] nuc ish(ABL1x3)[153/200] nuc ish(ABL2x3)[140/200] nuc ish(PDGFBRx3)[152/200]	a high hyperdiploid genome with multiple whole chromosome gains including an extra copy of chromosome X, three copies of chromosomes 3, 4, 5, 6, 7, 9, 8, 10, 11, 12, 16, 17, and 18; four copies of chromosome 14; and greater than four copies of chromosome 21.	c.nLOH of chromosome 13 and segmental alterations impacting the long arm of chromosome 1	a high hyperdiploid genome with multiple chromosome gains, including two copies of chromosome X, three copies of chromosomes 3, 4, 5, 6, 7, 8, 10, 11, 12, 16, 17, and 18, four copies of chromosome 14, and amplification (greater than four copies) of chromosome 21	copy-neutral LOH of chromosome 13 and mosaic gain of 1q and chromosome 9 were observed. CBL (NM_005188.3), c.1096-1G>C (p.?)	None (Category 3)
144	M	7	2/11/21	B-ALL	46,XY,t(7;14)(p14;q12),t(10;12;21)(q11.2;p13;q22),del(12)(p13)[13].ish t(10;12;21)(CEP10+,RUNX1+;ETV6-;RUNX1+,ETV6+),del(12)(ETV6-)/46,XY[7]	nuc ish(CEP4,CEP10)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6x1,RUNX1x3)[ETV6 con RUNX1x1][196/200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(CRLF2x2)[200]	loss of partial chromosome 12p involving ETV6	gain of Xq28 (1.71Mb) and loss of multiple partial chromosomes including 4q28.3 (252.59Kb), 5q31.3 (350.03Kb), 7p14.3p14.2 (975.60Kb), 12p13.3p13.1 (3.55Mb), 12p13.1p12.3 (3.20Mb), 12p12.3 (646.91Kb), 12q21.33 (259.51Kb), 14q12 (2.03Mb), and 19q13.32q13.33 (2.62Mb), with varying mosaic levels ranged from 70% to 80%	ETV6:RUNX1; loss of partial chromosome 12p involving ETV6	gain of partial Xq involving RPL10	a balanced translocation and a three-way translocation (Category 2)

145	M	7	1/28/21	B-ALL	46,XY,(4;12;21)(p16;p13;q22),der(4)t(4;12;q26;q15)[3],ish t(4;12;21)(RUNX1+;ETV6+;RUNX1+;ETV6+),arr[GRCCh37]4q26q35.2(117830724_190915650)x3,6q15q27(92996311_170919470)x1/47,idem,+mar[11]/46,XY[16]	nuc ish(CEP4,CEP10)x2[200] nuc ish(ABL1x1,BCR2x2)[146/200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6x2,RUNX1x3)[ETV6 con RUNX1x1][142/200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(CRLF2x2)[200]	a 73.1Mb gain of chromosome 4q26q35.2	multiple losses including a 6.4Mb loss of chromosome 3p22.1p21.31 (including the SETD2 gene), a 77.9Mb loss of chromosome 6q15q27, a 5.0Mb loss of chromosome 9q34.11q34.2 (including ABL1 and TSC1), a 231kb loss of 12q21.33 (including part of the BTG1 gene), and a 135kb loss of 21q22.12 (involving the RUNX1 gene).	ETV6:RUNX1; gain of partial 4q	NGS analysis performed on the DNA from this bone marrow specimen identified multiple Tier 2 copy number variations including gain of partial 4q (including FBXW7) and losses of partial 3p (including SETD2), 6q, partial 9q (including ABL1), and exons 6-8 of the ETV6 gene on 12p.	a derivative chromosome and a three-way translocation (Category 2)
146	M	15	1/26/21	AML	45,X,-Y,(8;17;21)(q22;p13;q22)[18],ish t(8;17;21)(RUNX1T1+;RUNX1+;RUNX1T1+;RUNX1+)/46,XY[1]	nuc ish(RUNX1T1,RUNX1x3)(RUNX1T1 con RUNX1x1)[140/200] nuc ish(KMT2Ax2)[200] nuc ish(NUP98x2)[200] nuc ish(MYH11,CBF8)x2[200]	Loss of Y	an ~11.15 Mb cnlOH region spanning from 19p13.3p13.2 and encompassing multiple genes	RUNX1:RUNX1T1	Five sequence variants with potential clinical significance (Tier 2): CSF3R (NM_000760.3), c.1735A>T (p.Asn579Tyr) SETD2 (NM_014159.6), c.4715+1G>C (p.?) CREBBP (NM_004380.2), c.4298_4299insCC (p.Asp1435Trpfs*25) WT1 (NM_024426.5), c.1389_1390insTCTCTCTTG (p.Arg463_Lys464insSerSerLeu) WT1 (NM_024426.5), c.377>A (p.Cys13Ser) Two copy number variation(s) and/or loss of heterozygosity (LOH) with potential clinical significance (Tier 2): loss of chromosome Y and copy neutral loss of heterozygosity on chromosome 19p involving multiple genes.	a three-way translocation (Category 2)
147	M	0	5/18/21	AML	46,XY,inv(11)(p15q22)[6],ish inv(11)(p15)(3'NUP98+)(q22)(5'NUP98+)/46,XY[14]	nuc ish(KMT2Ax2)[200] nuc ish(ETV6,RUNX1x2)[200] nuc ish(RUNX1T1,RUNX1x2)[200] nuc ish(NUP98x2)[5'NUP98 sep 3'NUP98x1][130/200] nuc ish(MYH11,CBF8)x2[200]	None, arr(X,Y)x1,(1-22)x2	None, arr(X,Y)x1,(1-22)x2	NUP98:DDX10	NRAS (NM_002524.4), c.183A>T (p.Gln61His)	None (Category 3)
148	M	4	5/13/21	T-ALL	46,XY[20]	nuc ish(RANBP17,TLX3)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6,RUNX1x2)[200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(CRLF2x2)[200]	None, normal karyotype	a 74 kb deletion on 1p33 responsible for the STIL-TAL1 fusion detected. A 127 kb loss of CDKN2A/B (including homozygous loss of CDKN2A) on chromosome 9p21.3 was also detected.	None, normal karyotype	STIL: TAL1; loss of partial 1p impacting upstream of TAL1; Six sequence variant(s) with potential clinical significance (Tier 2): NOTCH1 (NM_017617.4), c.7395del (p.Thr2466Argfs*11) NOTCH1 (NM_017617.4), c.7358_7370delinsGGACTCTCTTG (p.Val2453Glyfs*24) NOTCH1 (NM_017617.4), c.7502_7510del (p.Gln2501_Val2504delinsLeu) PTEN (NM_000314.6), c.542_552delins13 (p.Leu181Profs*19) PTEN (NM_000314.6), c.435_438delinsGGG (p.Phe145Leufs*8) USP7 (NM_003470.2), c.765delinsTGG (p.Val256Glyfs*4); Two copy number variations with potential clinical significance (Tier 2): loss of LEF1 on 4q and loss of CDKN2A/B on 9p	NA (normal karyotype likely due to the limited resolution)
149	F	20	4/28/21	B-ALL	hypodiploid/doubling 34+2n->X,-X,-2,-3,-4,-5,-7,-9,-13,-15,-16,-17,-20[11]/63*64,idemx2,-1,-6,-11,-12,-13(cp8)/46,XX[6]	nuc ish(CEP4x1,CEP10x2)[122/200] (CEP4x2,CEP10x4)[8/200] nuc ish(ABL1x1,BCR2x2)[132/200] (ABL1x2,BCR2x4)[16/200] nuc ish(KMT2Ax3)[10/200] nuc ish(ETV6x3,RUNX1x4)[12/200] nuc ish(CEP8,MYC,IGH)x4[10/200] nuc ish(CRLF2x1)[130/200] nuc ish(ABL1x1)[130/200] nuc ish(ABL2x4)[10/200] nuc ish(PDGFRBx1)[128/200]	a low hypodiploid clone (34 chromosomes) which might undergo endoreduplication and double the number of chromosomes	relative loss (apparently two copies with mosaic loss of heterozygosity) of whole chromosomes X, 2, 3, 4, 5, 7, 9, 13, 15, 16, 17, and 20, and relative gain of whole chromosomes 1, 6, 8, 10, 11, 12, 14, 18, and 21), which are likely consistent with four copies with heterozygosity in the chromosomes retained in the diploid state. In addition, a ~37 kb homozygous loss involving the CDKN2A/B genes on 9p21.3 was observed.	low hypodiploid	TP53 (NM_000546.5), c.839G>A (p.Arg280Lys); homozygous loss of part of the CDKN2A/B and RB1 genes;	None (Category 3)
150	F	2	4/19/21	B-ALL	46,XX[20]	nuc ish(CEP4,CEP10)x3[165/200]/(CEP4x3,CEP10x2)[25/200] nuc ish(CEP8,MYC,IGH)x3[200] nuc ish(ABL1x3,BCR2x2)[183/200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6x2,RUNX1x4)[186/200] nuc ish(CRLF2x2)[200]	None, normal karyotype	mosaic gains of one extra copy of chromosomes 4, 6, 8, 9, 10, 14, 17, and 18 and two extra copies of chromosomes 21, consistent with a high hyperdiploid tumor genome. In addition, a mosaic 83.4 Mb gain in 1q21.1q24.12 and copy number neutral LOH (cnLOH) of chromosome X were also detected.	None, normal karyotype	a high hyperdiploid genome with multiple whole chromosome gains, including chromosomes 4, 6, 8, 9, 10, 14, 17, 18, and 21; FLT3 (NM_004119.2), c.2516A>G (p.Asp839Gly);	NA (normal karyotype likely due to growth advantage of normal cells)
151	M	20	4/19/21	AML	46,XY,inv(16)(p13.1q22)[20]	nuc ish(RUNX1T1,RUNX1x2)[200] nuc ish(KMT2Ax2)[200] nuc ish(NUP98x2)[200] nuc ish(MYH11,CBF8)x3(MYH11 con CBF8x2)[200]	None	a 1.54 Mb mosaic loss on 17q11.2 involving the NF1 and SUZ12 genes in approximately 90% of cells.	CBF8:MYH11	NF1 (NM_001042492.2), c.2033dup (p.Ile679Aspfs*21); loss of partial 17q including the NF1 and SUZ12 genes.	None (Category 3)
152	M	18	4/15/21	B-ALL	55,XY,+X,+Y,+5,+6,+10,+10,+21,+21,-22[4]/46,XY[14]	nuc ish(CEP4x2,CEP10x4)[170/200] nuc ish(ABL1x2,BCR2x3)[174/200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6x2,RUNX1x4)[180/200] (ETV6x2,RUNX1x3)[10/200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(CRLF2x4)[172/200]	a high hyperdiploid tumor genome. With mosaic gains of multiple chromosomes including one extra copy of chromosomes X, Y, 5, 6, and 22, and two extra copies of chromosomes 10 and 21	None	A high hyperdiploid genome with gains of one extra copy of chromosomes 5, 6, 22, X and Y, and two extra copies of chromosomes 10 and 21	FLT3 (NM_004119.2), c.1988A>C (p.Lys663Thr);	None (Category 3)
153	M	10	4/9/21	CML	46,XY,t(9;22)(q34;q11.2)[20]	nuc ish(ABL1,BCR)x3(ABL1 con BCRx2)[198/200]	None, Array not performed	None, Array not performed	BCR:ABL1 (p210)	None	None (Category 3)
154	F	4	4/9/21	AML	47,XX,del(2)(p16p22),t(3;19)(p21;p13.3),del(9)(q21q31),del(12)(q24),+21[20]	nuc ish(RUNX1T1x2,RUNX1x3)[150/200] nuc ish(KMT2Ax2)[200] nuc ish(NUP98x2)[200] nuc ish(MYH11,CBF8)x2[200]	a mosaic gain of whole chromosome 21; mosaic segmental losses on chromosomes 2p, 3p, 3q, 9q, and 12q	mosaic segmental losses on chromosomes 5p, 7q, and 19p in ~60% of cells	Losses of partial 2p, partial 9q and partial 12q; gain of whole chromosome 21.	ETV6:EP300; multiple segmental copy number loss involving additional partial chromosomes	a balanced translocation (Category 2)

155	F	1	3/29/21	B-ALL	46,X,der(X)(X1)(Xpter->Xp11.2::Xq21.73->Xp11.2::Xq21.73->Xq24::11q23.3->11q23.3::11p13->11pter),der(11)(X1)(11qter->11q23.3::11p13->11q23.3::Xq24->Xqter)[20],ish der(X)(3'KMT2A+),der(11)(5'KMT2A+,3'KMT2A+)	nuc ish(RUNX1T1,RUNX1)x2[200] nuc ish(3'KMT2Ax3,5'KMT2Ax2)[3'KMT2A con 5'KMT2Ax1][194/200] nuc ish(NUP98x2)[200] nuc ish(MYH11,CBFB)x2[200]	an 88.9kb loss within 11q23.3, which includes exons 9-36 of the KMT2A gene (NM_001197104.1)	None	KMT2A:SEPT6; loss of partial 11q involving exons 9-36 of KMT2A	PTPN11 (NM_002834.4), c.218C>T (p.Thr73Ile);	None (Category 3)
156	M	16	4/6/21	T-ALL	46,XY,del(6)(q14q21),del(9)(p13p21),7inv(11)(q14q23),del(12)(p12p13),del(16)(q12q23)[2],ish ?inv(11)(5'KMT2A+,3'KMT2A+)[47,sl +8(14)/46,sd1,der(5)(7)(5,20)[20pter->20qter::5q11.1->5qter],-20(7)/46,XY[2]	nuc ish(RANBP17,TLX3)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[5'KMT2A sep 3'KMT2Ax1][182/200] nuc ish(ETV6x1,RUNX1x2)[184/200] nuc ish(CEP8x2,MYCx3,IGHx2)[190/200] nuc ish(CRLF2x2)[200]	a 12.2Mb loss of chromosome 2q36.1q37.1, a 33.0Mb loss of chromosome 6q14.1q21, an 18.0Mb loss of chromosome 9p21.3p13.1 (including the CDKN2A/28 and PAX5 genes) with an apparent homozygous 1.64Mb deletion within this region encompassing the CDKN2A/28 genes, a 9.27Mb loss of 12p13.31 (including ETV6), a 26.6Mb loss of 16q12.2q23.2. Additionally, a gain of chromosome 8 was identified in approximately 75-80% of cells.	a 229kb loss within 11p15.1, a 1.14Mb loss of chromosome 11q14.1q14.2, and a 1.28Mb loss of 17q21.31 in approximately 95% of cells. A 2.8Mb loss of chromosome 3q25.3q25.33 and a 4.1Mb loss of chromosome 3q27.2 were identified. A 3.06Mb loss of 16p13.3 and a 1.19Mb loss of 16p12.3p12.2 were identified	KMT2A:PRPF19; Significant CNV findings include gain of whole chromosome 8, and partial 3q (including BCL6), partial 9p (with homozygous loss of CDKN2A/28), partial 12p (including ETV6), and partial 16q.	EP300 (NM_001429.3), c.3671+1G>A (p.?). losses of partial 3q (including BCL6, partial 11p, and partial 16p (including CREBBP).	an inversion (Category 2)
157	F	7	2/15/21	B-ALL	57,XX,+X,+4,+5,+6,+10,der(11)(1:12)[q21,p13],+14,+17,+18,+18,+21,+21[11],ish der(12)(ETV6+)[46,XX[7]	nuc ish(CEP4,CEP10x3)[194/200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6x2,RUNX1x4)[184/200] [ETV6x2,RUNX1x3][14/200] nuc ish(CEP8x2,MYCx2,IGHx3)[196/200] nuc ish(CRLF2x3)[198/200]	a high hyperdiploid tumor genome with gain of an extra copy of chromosome X, three copies of chromosomes 4, 5, 6, 10, 14, and 17, and four copies of chromosomes 18 and 21. Gain of chromosome 1q	cnLOH of whole chromosomes 2 and 22 were also detected. Most of these findings have a mosaic level of 90-95%.	a high hyperdiploid tumor genome with multiple whole chromosome gains, resulting in three copies of chromosomes 4, 5, 6, 10, 14, 17, and X and four copies of chromosomes 18 and 21. A segmental gain of chromosome 1q was also noted.	copy neutral loss of heterozygosity on chromosomes 2 and 22; NRAS (NM_002524.4), c.386G>A (p.Gly13Asp)	a derivative chromosome (Category 2)
158	M	3	2/15/21	B-ALL	59,+62,XY,+X,+Y,der(2)(1:2)[q21;q37],+2,+4,der(5)del(5)(q21q34)[1;5][q21;q34],+5,+6,+8,+10,+11,+12,+14,+17,+18,+21,+21,+72[cp14]/46,XY[6]	nuc ish(CEP4,CEP10x3)[190/200] nuc ish(ABL1x2,BCR)x3[40/200] nuc ish(KMT2Ax3)[140/200] nuc ish(ETV6,RUNX1x3)[128/200] [ETV6x3,RUNX1x4][68/200] nuc ish(CEP8,MYC,IGH)x3[194/200] nuc ish(CRLF2x4)[154/200] [CRLF2x3][40/200]	a high hyperdiploid tumor genome with gains of a copy of whole chromosomes X, Y, 2, 4, 6, 8, 10, 11, 12, 14, 17, 18 and 21	copy-neutral loss of heterozygosity (cnLOH) of 22q11.21q13 and several copy number gains and losses of partial chromosomes.	a high hyperdiploid genome with multiple whole chromosome gains (including chromosomes 2, 4, 6, 8, 10, 11, 12, 14, 17, 18, 21, and X)	copy-neutral loss of heterozygosity (cnLOH) of chromosome 22, and several segmental chromosome rearrangements;	two derivative chromosomes (Category 2)
159	M	17	2/12/21	B-ALL	No growth	nuc ish(CEP4x3,CEP10x2)[180/200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6x2,RUNX1x3)[176/200] nuc ish(CEP8x2,MYCx2,IGHx3)[180/200] nuc ish(CRLF2x3)[158/200] [CRLF2x4][20/200] nuc ish(ABL1x2)[200] nuc ish(ABL2x2)[200] nuc ish(PDGFRBx2)[200]	N/A, Cyto no growth	a hyperdiploid genomic profile (~52 chromosomes). The whole chromosome gains resulted in three copies of chromosomes 4, 6, 17, 18, and 21 and two copies of chromosome X. In addition, loss of the CDKN2A/B genes on 9p21.3 is noted.	N/A, Cyto no growth	a hyperdiploid genomic with multiple chromosome gains, including three copies of chromosomes 4, 6, 17, 18, 21 and one extra copy of chromosome X, consistent; KRAS (NM_033360.3), c.35G>T (p.Gly12Val), PAX5 (NM_016734.2), c.461_474delinsTCTCCC (p.Ser154Phefs*86), PTPN11 (NM_002834.4), c.215C>T (p.Ala72Val)	NA (no growth)
160	F	15	2/12/21	B-ALL	46,XX[20]	nuc ish(CEP4,CEP10x2)[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6,RUNX1)x2[200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(CRLF2x2)[200]	None, normal karyotype	deletions of 7q34 (~394.79kb), 9p21.3p21.3 (~30kb) involving CDKN2A/CDKN2B genes, 12p13.3p13.1 (~14.40Mb) involving multiple genes, a duplication of 12p13.1p13.3 (~565kb), and a cnLOH of 13q12.11q34 (~115.10Mb)	None, normal karyotype	loss of partial 12p (including ETV6) and copy-neutral loss of heterozygosity (cnLOH) of 13q, KRAS (NM_033360.3), c.35G>A (p.Aspl19Asn), R81 (NM_000321.2), c.260_264+7delinsAGG (p.Val87Glyfs*22)	NA (normal karyotype likely due to the limited resolution)
161	M	14	12/30/20	B-ALL	46,XY[19]	nuc ish(RUNX1T1,RUNX1)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(NUP98x2)[200] nuc ish(MYH11,CBFB)x2[200] nuc ish(PML,RARA)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(EVI1x2)[200]	None, normal karyotype	a 1.36Mb non-mosaic gain within chromosome 9q28 and a 1.42Mb mosaic gain within 7q11.23 detected in ~50% of cells.	None, normal karyotype	FLT3 (NM_004119.2), c.1743_1922dup (p.Val641_Ala642ins90), KRAS (NM_033360.3), c.35G>T (p.Gly12Val), RAD21 (NM_006265.2), c.439_440del (p.Glu147Argfs*12)	NA (normal karyotype)
162	M	9	12/16/20	B-ALL	45,-48,X,-Y,del(11)(q17q),t(12;21)(p13;q22),+16,+18,+21[cp16],ish t(12;21)(RUNX1+,ETV6-;RUNX1+,ETV6+)[46,XY[5]	nuc ish(CEP4x2,CEP10x3)[40/200] [CEP4,CEP10x4][10/200] nuc ish(ABL1,BCR)x4[14/200] nuc ish(KMT2Ax1)[40/200] (KMT2Ax3)[10/200] (KMT2Ax4)[8/200] nuc ish(ETV6x2,RUNX1x3)[ETV6 con RUNX1x1][38/200] [ETV6x2,RUNX1x4][ETV6 con RUNX1x1][18/200] [ETV6x2,RUNX1x5][ETV6 con RUNX1x1][14/200] [ETV6x4,RUNX1x6][ETV6 con RUNX1x2][10/200] nuc ish(CEP8x2,MYCx3,IGHx2)[22/200] (CEP8,MYC,IGH)x4[10/200] nuc ish(CRLF2x1)[10/200] [CRLF2x3][10/200]	whole chromosome loss of chromosome Y and whole chromosome gains of chromosomes 10, 16, 18, 20, and 22. The complex alterations on chromosome 12 (including loss of part of the ETV6 gene) and chromosome 21 (including gain of the RUNX1 gene) are consistent with a translocation involving chromosomes 12p and 21q, resulting in the ETV6-RUNX1 fusion	whole chromosome gains and loss at varying degrees of mosaicism, indicating clonal heterogeneity. Pertinent alterations include a ~126 kb loss of 3q26.32 involving the TBL1XR1 gene, a ~258 kb loss involving part of PAX5 on 9p13.2, several segmental losses on chromosome 12 including a ~285 kb loss involving ETV6 on 12p13.2, a ~149 kb loss involving KRAS on 12p12.1, and a ~260 kb loss involving BTG1 on 12q21.33, complex gains of chromosome 21 impacting RUNX1 gene,	ETV6:RUNX1; loss of chromosome Y, and gains of whole chromosomes 16, 20, 22, and partial 21q.	Multiple copy number variants (CNVs) including gain of 6q, loss of partial 11q, segmental losses on 12p (impacting ETV6 and KRAS), loss of partial RB1 on 13q,	None (Category 3)
163	F	7	12/15/20	B-ALL	55,XX,+X,+4,+der(7-9)(q10;q10),+8,-10,+14,+18,+21,+21[8]/56,ider(7-9)(q10;q10),+7,+9[2]/46,XX[15]	nuc ish(CEP4,CEP10x3)[168/200] nuc ish(ABL1x3,BCR)x2[160/200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6x2,RUNX1x4)[170/200] nuc ish(IGHx3)[164/200] nuc ish(CRLF2x3)[170/200]	a high hyperdiploid genome with multiple chromosome gains including trisomy (three copies) for chromosomes X, 4, 8, 10, 14, and 18 in 40% of cells as well as tetrasomy (four copies) of chromosome 21. Gains of chromosomes 7 and 9.	a ~6.3 Mb loss from 13q14.2 to 13q14.3 and a deletion of exons 18-27 of RB1 on 13q14.2 within the 6.3 Mb 13q deletion. A likely constitutional finding, a region of homozygosity on 11p encompassing the WT1 gene, was also noted.	High hyperdiploidy genome including three copies of chromosomes 4, 7, 8, 9, 10, 14, 18, and X and four copies of chromosome 21	loss of RB1 located on the q arm of chromosome 13. DOTT1 (NM_032482.2), c.1005+1G>A (p.?). NRAS (NM_002524.4), c.386G>A (p.Gly13Asp), KRAS (NM_033360.3), c.376>T (p.Gly13Cys)	a derivative chromosome (Category 2)

164	M	7	12/9/20	T-ALL	46,XY[20]	nuc ish(RANBP17x2,TLX3x3)[RANBP17 con TLX3x2][140/200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6,RUNX1)x2[200] nuc ish(IGHx2)[200] nuc ish(CRLF2x2)[200]	None, normal karyotype	multiple segmental chromosomal deletions including a 184.4Kb loss of Xq26.2, a 491.8Kb loss of 7q34 with a partial homozygous loss, a 2.0Mb loss of 9p21.3 with homozygous loss involving CDKN2A/CDKN2B, a 75.4Mb cNLOH of chromosome 9q, a 92.3Kb homozygous loss of 13q14.2 involving partial RB1, a 507.2Kb loss of 14q11.2 involving TCF7L1/TCF7L2, a 1.1Mb loss of 16q12.2, a 327.7Kb loss of 16q22.1, and a 255.4Kb loss of 20q11.22.	None, normal karyotype	loss of partial 9p (including CDKN2A/2B), partial 13q (involving RB1), and partial 14q (including PHF6). Additionally, loss of 16q, gain of partial 10p, and low-level mosaic copy-neutral LOH of 9q (including NOTCH1) were observed.	NA (normal karyotype likely due to the limited resolution)
165	F	0	11/18/20	B-ALL	46,XX,t(4;11)(q21;q23)[19]/46,XX[1]	nuc ish(KMT2Ax2)[5*KMT2A sep 3*KMT2Ax1][194/200] nuc ish(CEP4,CEP10)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(ETV6,RUNX1)x2[200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(CRLF2x2)[200]	None, arr(1-22,X)x2	None, arr(1-22,X)x2	KMT2A:AFF1	None	None (Category 3)
166	F	18	11/16/20	AML	51,XX,+4,+8,+10,+17,add(17)(p11.2),+19(2)/46,XX[17]	nuc ish(RUNX1x3,RUNX1x2)[170/200] nuc ish(KMT2Ax2)[200] nuc ish(NUP98x2)[200] nuc ish(MYH11,CBF8)x2[200]	gains of whole chromosomes 4, 8, and 19 and a ~42.4 Mb tetrasomy (4 copies) of 10p15.3q11.2, a ~92.8 Mb gain on 10q11.21q26.3, a ~59.3 Mb gain on 17p11.2q25.3.	cNLOH of a 48.8 Mb region on 5q31.1q35.3, cNLOH of whole chromosome 7, and a 21.6 Mb region on 17p13.3p11.2. Finally, a 176 kb intragenic loss of RUNX1 on 21q22.12 was detected	gains of whole chromosomes 4, 8, 10, and 19, and a gain of 17q, indicating a hyperdiploid genome with ~50 chromosomes	In addition, copy neutral loss of heterozygosity was identified for the whole chromosomes 7 and 12, 17p and partial 5q. Loss of exons 4-7 of RUNX1: ASXL1 (NM_015338.5), c.1934dup (p.Gly646Trpfs*12), TP53 (NM_00046.5), c.718A>G (p.Ser240Gly), SUZ21 (NM_015355.3), c.506-1G>A (p.), IKZF1 (NM_006060.5), c.479T>C (p.Leu160Pro)	None (Category 3)
167	M	13	11/10/20	B-ALL	46,XY,t(4;11)(q21;q23)[11]/46,XY[9]	nuc ish(CEP4,CEP10)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[5*KMT2A sep 3*KMT2Ax1][199/200] nuc ish(ETV6,RUNX1)x2[200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(CRLF2x2)[200] nuc ish(ABL1x2)[200] nuc ish(ABL2x2)[200] nuc ish(PDGFR8x2)[200]	None, arr(X,Y)x1,(1-22)x2	None, arr(X,Y)x1,(1-22)x2	KMT2A:AFF1	None	None (Category 3)
168	F	8	11/3/20	B-ALL	46,XX,der(1)(1;7)(p37;q37),der(7)del(7)(p12p14)(1;7),der(21)del(21)(q11.2q21)dup(21)(q21q22)[16],ish der(21)(RUNX1 amp),del(X)(3' CRLF2+,5' CRLF2-)/46,XX[4]	nuc ish(CEP4,CEP10)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6x2,RUNX1amp)[198/200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(3' CRLF2x2,5' CRLF2x1)[3' CRLF2 con 5' CRLF2x1][196/200]	a 314.8 kb loss of Xp22.33, a 4.5 Mb loss of 1p32.3p32.2, 518 kb loss of 4q22.1, 9.3 Mb loss of 7p14.1p12.1 involving IKZF1, loss of 1.85 Mb on 7q36.1 involving EHZ2, 10.8 Mb loss on 21q11.2q21.2, and gain of 15.7 Mb of 21q21.2q22.2 involving RUNX1.	161.8 kb loss of 4q31.3 involving FBXW7, 7 regions of copy neutral loss of heterozygosity on 9p (37.4 Mb on 9p24.3p13.2 and 1.7 Mb on 9p13.2p13.1) involving JAK2, CDKN2A/B and PAX5, 175.5 kb loss on 12q21.31 involving BTG1, 4.1 Mb loss of the terminus of the short arm of chromosome 19 (19p13.3),	a CNV pattern consistent with IAMP21; P2RY8:CRLF2,	loss of partial 4q involving exons 3-12 of the FBXW7 gene, loss of partial 7p including IKZF1, loss of partial 7q including EHZ2, copy neutral loss of heterozygosity of 9p involving JAK2 gene, CDKN2A/B, and PAX5, loss of partial 12q including SH2B3, and loss of partial 19p. JAK2 (NM_004972.3), c.2047A>G (p.Arg683Gly) JAK2 (NM_004972.3), c.2049A>T (p.Arg683Ser) JAK2 (NM_004972.3), c.2624C>A (p.Trp875Asn) JAK2 (NM_004972.3), c.1812_1815delins22 (p.Ser605delinsArgTrpAlaGluGlyGlyLeu)	a complex rearrangement involving multiple chromosomes (Category 2)
169	M	21	10/30/20	AML	46,XY[20]	nuc ish(RUNX1T1,RUNX1x2)[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(NUP98x2)[132/200]/(NUP98x2)[5*NUP98 sep 3*NUP98x1][68/200] nuc ish(MYH11,CBF8)x2[200]	None, normal karyotype	a 222.5 kb gain of Xp22.33 involving the CRLF2 gene, copy neutral loss of heterozygosity (cNLOH) impacting 50.3 Mb on the short arm of chromosome 6 (6p25.3p12.3), and a 1.4 Mb loss on 17q11.2 involving the NF1 and SUZ12 genes (~85 mosaicism).	None, normal karyotype	loss of partial 17q (involving NF1 and SUZ12), and gain of partial Xp (involving CRLF2) NPM1 (NM_002520.6), c.860_863dup (p.Trp288Cysfs*12) NF1 (NM_001042492.2), c.4431-1G>C (p.), NF1 (NM_001042492.2), c.2033dup (p.Ile679Aspfs*21)	NA (normal karyotype likely due to the limited resolution)
170	F	3	1/31/20	B-ALL	56,XX,+x,+4,+6,+der(1;9)(q10;p10),+10,+14,+17,+18,+21,+21[11]/46,XX[8]	nuc ish(CEP4,CEP10)x3[192/200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6x2,RUNX1x4)[140/200] nuc ish(ETV6x2,RUNX1x3)[54/200] nuc ish(CEP8x2,MYCx2,IGHx3)[190/200] nuc ish(CRLF2x3)[190/200]	high-hyperdiploid genome trisomy of chromosome X, 4, 6, 10, 14, 17 and 18 (~75% mosaicism); tetrasomy of chromosome 21, a gain of long arm of chromosome 1 (~75% mosaicism) and a gain of short arm of chromosome 9 (~75% mosaicism); The mosaic gain of 1q and 9p is consistent with the observation of der(9)(1;9)	None	a high hyperdiploid genome with multiple chromosome gains, including gains of one extra copy of whole chromosomes X, 4, 6, 10, 14, 17, 18, and two additional copies of chromosome 21; gain of chromosome 1q and 9p	cNLOH of 9q; FLT3 (NM_004119.2), c.2508_2510del (p.Ile836del)	a derivative chromosome (Category 2)
171	M	8	10/15/20	B-ALL	46-47,XY,add(2)(q37),7+10,del(12)(p17),add(12)(p13),7-20,der(21)(12;21)(p13;q22)[cp20],sh del(12)(ETV6-),add(12)(ETV6-),der(21)(RUNX1+,ETV6+)	nuc ish(CEP4x2,CEP10x3)[152/200] nuc ish(CEP4,CEP10)x2[200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6x1,RUNX1x2)[ETV6 con RUNX1x1][120/200]/(ETV6x1,RUNX1x3)[ETV6 con RUNX1x1][80/200] nuc ish(CRLF2x2)[200]	complex alterations on 12p involving ETV6. A 135.5 Mb gain of multiple genes on 2q12.2q37.3 in 15% of cells, gain of whole chromosome 10 in 50-55% of cells, a 11.2 Mb loss on 20p13p12.2 in 95% of cells.	complex alterations on 9p. Homozygous loss of 278 kb on Xp21.1, a 357 kb loss involving FHIT on 3p14.2, a 1.2 Mb gain impacting the FAT1 gene on 4q35.1q35.2, a 58.7 Mb loss on 18q11.2q23 in 25% of cells	ETV6:RUNX1; gain of chromosomes 10; homozygous loss of exons 6-8 of ETV6 and gain of ETKN1, exons 2-4 of KRAS were observed on 12p.	loss of partial 9p and whole chromosome 18, and gain of partial 17q, and copy neutral loss of heterozygosity (cNLOH) of 11q.	None (Category 3)
172	M	1	10/10/20	T-ALL	46,XY,del(9)(p21p21)[5].rsa 9p21.3(CDKN2A,CDKN2B)x0 mos hnz/46,XY[15]	nuc ish(RANBP17,TLX3)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6,RUNX1)x2[200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(CRLF2x2)[200]	a mosaic homozygous loss involving the CDKN2A/B genes	a ~395 kb loss of the PRSS1 gene on 7q34, a 29 Mb complex alteration involving copy neutral loss of 9p24.3p21.1, ~1.2 Mb loss of the NFKB1A gene on 14q13.2q13.3, a 93 kb loss on 17q11.2 involving the NF1 gene and a ~1.4 Mb gain of PAR1 region involving CRLF2 on Xp22.3/1p11.3.	a complex CNV with copy neutral (cn) LOH of JAK2 and homozygous loss of CDKN2A/B;	loss of partial 17q involving exons 16-36 of NF1; and gain of partial Xp involving CRLF2; PTEN (NM_000314.6), c.702_712del (p.Glu235Hisfs*4), PTEN (NM_000314.6), c.699_700insGGTCCCTC (p.Arg234Glyfs*11), PTEN (NM_000314.6), c.697_700delins14 (p.Arg233Trfs*13), RUNX1 (NM_001754.4), c.946del (p.Glu316Asnfs*12), PHF6 (NM_032458.2), c.635G>A (p.Cys211Tyr), NOTCH1 (NM_017617.4), c.7327_7328insCTAGCCC (p.Val2443Alafs*2), MYB (NM_005375.3), c.1108_1128del (p.Ser370_Ile376del)	None (Category 3)

173	M	2	9/28/20	B-ALL	46,XY,der(6)(pter->6q12::6p21->pter),t(9)(q10),der(19)(t11;19)(q23;p13.3)[9].ish i46,XY,t(7;14)(p14;q12),t(10;12)(q11;q9)(ABL1+),der(19)(PBX1+TCF3+) 46,XY[11]	nuc ish(CEP4,CEP10)x2[200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(ABL1x3,BCRx2)[162/200] nuc ish(KMT2A2)[200] nuc ish(ETV6,RUNX1)x2[200] nuc ish(CRLF2x2)[200] nuc ish(ABL1x3)[172/200] nuc ish(ABL1x3)[170/200] nuc ish(PBX1x3,TCF3x2)[PBX1 con TCF3x1][170/200] nuc ish(PDGF8x2)[200]	an 84.5Mb terminal gain of 1q23.3q44 (involving PBX1 in the breakpoint), a 44.6Mb terminal gain of 6p25.3p21.1, a 106.6Mb terminal loss of 6q12q27, a 39.1Mb terminal loss of 9p24.3p13.1 (containing CDKN2A/2B and PAX5), a 75.4Mb terminal gain of 9q12q34.3, and a 1.35Mb terminal loss of 19p13.3 (involving TCF3 in the breakpoint).	a 634kb interstitial loss of 6q12 and an 86.5Mb terminal loss of 13q12.2q34 (including RB1 and BRCA2).	TCF3::PBX1; gain of 1q, 6p, and 9q and losses of 6q, 9p, and partial 19p involving the ELANE gene.	Loss of whole chromosome 13	a derivative chromosome and an isochromosome (Category 2)
174	F	0	12/8/20	B-ALL	92<4n>,XXXX[6]/46,XX[19]	nuc ish(CEP4,CEP10)x2[200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2A2)[5xKMT2A sep 3xKMT2A1][15/200](KMT2A4)[30/200] nuc ish(ETV6,RUNX1)x2[200] nuc ish(CRLF2x2)[200]	None, array not performed	None, array not performed	None	KMT2A::MLLT10	a tetraploid genome (Category 2)
175	M	19	9/28/20	AML	46,XY,t(8;21)(q22;q22)[19]/46,XY[1]	nuc ish(RUNX1T1,RUNX1x3)[RUNX1T1 con RUNX1x3][196/200] nuc ish(KMT2A2)[200] nuc ish(NUP98x2)[200] nuc ish(CBFBx2)[200] nuc ish(MYH11,CBFB)x2[200]	None, arr(X,Y)x1,(1-22)x2	None, arr(X,Y)x1,(1-22)x2	RUNX1::RUNX1T1	KIT (NM_000222.2), c.2466T>G (p.Asn822Lys), RAD21 (NM_006265.2), c.1641delinsGGT (p.Asp548Valfs*65) ASXL1 (NM_015338.5), c.1900_1922del (p.Glu635Argfs*15)	None (Category 3)
176	F	2	9/20/20	B-ALL	59,XX,+X,+X,+5,+6,+10,+10,+der(11)t(11;12)(q14;q11),+14,+17,+18,+21,+21,+22[6]/46,XX[14]	nuc ish(CEP4x2,CEP10x4)[168/200] nuc ish(ABL1x2,BCR3x3)[152/200] (ABL1,BCR)x3[18/200] nuc ish(KMT2A2)[200] nuc ish(ETV6x2,RUNX1x4)[134/200] (ETV6x1,RUNX1x4)[18/200] (ETV6x2,RUNX1x4)[20/200] nuc ish(CEP8x2,MYC2,IGHx2)[170/200] nuc ish(CRLF2x4)[166/200]	a high hyperdiploid genome with multiple chromosome gains. The main findings include trisomy 5, 6, 14, 17, 18, and 22, and tetrasomy X, 10, and 21; Segmental copy number variations (CNVs), including a ~78.3 Mb gain from 11p15.5q14.1, a ~1.88 Mb gain of 12p11.23 involving PPF1BP1, and a ~95.3 Mb gain of 12q12q24.33 in 50% of cells.	copy neutral loss of heterozygosity (cn-LOH) of chromosome 19. A ~984 kb loss of 12p13.2 involving ETV6 and P41212 in 20% of cells.	a high hyperdiploid genome with multiple chromosome gains (as listed in the table), including three copies of chromosomes 5, 6, 10, 14, 17, 18, and 22 and four copies of chromosomes X, 10, and 21. Segmental copy number variants include gain of 11p, gain of partial 11q involving the SF1 gene, and gain of 12q.	Chromosome 19 copy neutral loss of heterozygosity was also identified. Loss of partial 12p involving ETV6,	None (Category 3)
177	M	19	8/24/20	B-ALL	47,XY,+X[20]	ish(CEP4,CEP10)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2A2)[200] nuc ish(ETV6,RUNX1)x2[200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(CRLF2x3)[144/200] nuc ish(ABL1x2)[200] nuc ish(ABL2x2)[200] nuc ish(PDGF8x2)[200]	gain of whole chromosome X	a 34.4Mb cnLOH of 6p25.3p21.31, and a total 842.8kb (at minimum) loss of 9p21.3 involving the CDKN2A/B genes with partial homozygous loss of CDKN2A.	gain of whole chromosome X	loss of partial 9p involving the CDKN2A/2B genes (apparently homozygous loss of part of CDKN2A, SNP microarray is pending and should confirm), and cnLOH of partial 6p involving TPMT, MSH2 (NM_000251.2), c.2087C>T (p.Pro696Leu) PAX5 (NM_016734.2), c.1129C>A (p.Arg377*) NRAS (NM_002524.4), c.386G>A (p.Gly134Asp)	None (Category 3)
178	F	9	8/19/20	B-ALL	46,XX,del(7)(q22q36),del(9)(p13p22),der(21)dup(21)(q22)[14].ish der(21)(RUNX1amp)/46,XX[6]	nuc ish(CEP4,CEP10)x2[200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2A2)[200] nuc ish(ETV6x2,RUNX1x1,RUNX1 amp)[200] nuc ish(CRLF2x2)[200]	a 2.8Mb duplication of 21q21.2q21.3, an 18.5Mb duplication of 21q21.3q22.3 encompassing RUNX1, and a 148kb terminal deletion within 21q22.3. This CNV pattern is consistent with IAMP21; a 48.6Mb deletion on 7q22.1q36.1, a 19.1Mb deletion on 9p22.1p13.1 including CDKN2A/2B and PAX5	a 516kb deletion on 13q14.2 involving the first two exons of the RB1 gene, a 3.78Mb deletion on 13q14.2q14.3 including the last 10 exons of the RB1 gene, a 49kb deletion on 14q23.2 which encompasses the HIF1A gene, and a 519kb deletion on 6q22.31.	an amplification of chromosome 21q; loss of partial 7q (involving RELN, BRAF, and EZH2); loss of partial 9p (involving CDKN2A/2B, and PAX5) is observed.	loss of partial 13q (involving NUDT15, and partial RB1)	None (Category 3)
179	M	14	1/3/20	CML	46,XY,t(9;22)(q34;q11.2)[17]	nuc ish(ABL1x3,BCR3x3)[ABLI con BCRx2][186/200] nuc ish(KMT2A2)[200] nuc ish(CBFBx2)[200]	None, arr(X,Y)x1,(1-22)x2	None	BCR::ABL1	None	None (Category 3)
180	F	20	8/15/20	B-ALL	46,XX,del(2)(p16pter),del(9)(p13p22),er,t(9;14)(p17q;q32)[19]/46,XX[6]	nuc ish(CEP4,CEP10)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2A2)[200] nuc ish(ETV6,RUNX1)x2[200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(CRLF2x2)[200] nuc ish(ABL1x2)[200] nuc ish(ABL2x2)[200] nuc ish(PDGF8x2)[200]	a 55.05 Mb loss at chromosome 2p25.3p16.1 and loss of an entire 9p (~55.8 Mb) in approximately 15% of cells.	None	loss of 2p and loss of 9p. Chromosome 2p contains the DNMT3A, ASXL2, MSH2, and MSH6 genes, and 9p contains the IAK2, CDKN2A/2B, and PAX5 genes.	ETV6 (NM_001987.4), c.391dup (p.Ser131Phefs*23) KMT2D (NM_003482.3), c.2713G>T (p.Glu950*)	a balanced translocation (Category 2)
181	F	19	8/14/20	AML	No growth	nuc ish(CEP4,CEP10)x2[200] nuc ish(ABL1,BCR)x3[BCR con ABL1x2][128/200] nuc ish(KMT2A2)[200] nuc ish(ETV6,RUNX1)x2[200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(CRLF2x2)[199/200]	N/A, Cyto no growth	a gain of whole chromosome 5 and multiple small focal deletions. The latter include an intragenic ~51kb loss of the IKZF1 gene on chromosome 7p12.2, an intragenic ~91kb loss of the PAX5 gene on chromosome 9p13.2, a ~261kb loss involving BTG1 gene and a ~135kb loss involving almost the entire RB1 gene on chromosome 13q14.2. In addition, a ~649kb loss on 14q11.2 involving T-cell receptor genes alpha and delta (TCRA/D) and a ~376kb loss on 14q32.33 involving IGHG gene was also detected.	N/A, Cyto no growth	gain of 5, loss of partial 7p (including partial IKZF1), loss of partial 9p (including partial PAX5), and loss of partial 13q (including partial RB1).	NA (no growth)
182	F	6	8/7/20	B-ALL	46,XX,t(12;21)(p13;q22)[19].ish t(12;21)(RUNX1+,ETV6-);RUNX1+,ETV6-/ 46,XX[1]	nuc ish(CEP4,CEP10)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2A2)[200] nuc ish(ETV6x2,RUNX1x3)[ETV6 con RUNX1x1][196/200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(CRLF2x1)[14/200]	a ~142kb loss spanning exon 2 of the ETV6 gene on chromosome 12p13.2	a ~138kb loss on chromosome 7q34 involving T-cell receptor beta (TCRB), and a complex copy number alteration involving T-cell receptor alpha/delta (TCRA/D) on chromosome 14q11.2 (including heterozygous loss of a ~202kb region and an adjacent homozygous deletion of a ~367kb)	ETV6::RUNX1; a heterozygous loss of exon 2 of the ETV6 gene	NSD2 (NM_133335.3), c.3295G>A (p.Glu1099Lys) UBA2 (NM_005499.2), c.359-1G>T (p.?)	NA (normal karyotype*)

183	F	10	8/1/20	AML	46,XX[19]	nuc ish(RUNX1T1,RUNX1)x2[200] nuc ish(PML,RARA)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(CBFb2)[200] nuc ish(NUP98x2)[200]	None, array not performed and normal karyotype	None, array not performed	None, normal karyotype	IDH2 (NM_002168.3), c.419G>A (p.Arg140Gln), NRAS (NM_002524.4), c.35G>A (p.Gly12Asp)	NA (normal karyotype)
184	M	6	7/13/20	B-ALL	46,XY[20]	nuc ish(CEP4x3,CEP10x2)[105/200]/(CEP4x3,CEP10x3)[75/200] nuc ish(CEP8x2,MYCx2,IGHx4)[160/200] nuc ish(ABL1x3,BCRx2)[137/200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6x2,RUNX1x4)[174/200]/(ETV6x2,RUNX1x3)[25/200] nuc ish(CRLF2x2)[200]	None, normal karyotype	gains of one extra copy of chromosomes X, 4, 5, 8, 10, and 17, gain of one extra copy of chromosomes 9 and mosaic gain of two extra copies of chromosomes 14, 18 and 21. Additionally, segmental mosaic chromosome rearrangements were observed, including 99.3Mb gain of 1q21.2q44, 95.2Mb gain of 7q11.21q36.3, 100.0Mb gain of 11p15.5q22.1, 34.7Mb copy-neutral loss of heterozygosity (cnLOH) of 11q22.1q25, 31.1Kb loss of 12p13.2 (containing partial ETV6), 331.5Kb loss of 12q21.33, and complex chromosome 16 rearrangement	None, normal karyotype	a high hyperdiploid genome with multiple chromosome gains including gains of one extra copy of whole chromosomes X, 4-6, 9-10, 17 and two extra copies of chromosome (tetrasomy) 14, 18, and 21. Additionally, gain of 1q, 7q, 11p, 16p, loss of partial 12p (involving exons 4-5 of ETV6), copy-neutral loss of heterozygosity (cnLOH) of 16q, and complex 11q rearrangement (gain of SF1, EED, and cnLOH of KMT2A, CBL, ETS1) were observed. P2RY8::CRF2 gene fusion; PTPN11 (NM_002834.4), c.1508G>C (p.Gly503Ala)	NA (normal karyotype likely due to growth advantage of normal cells)
185	F	4	7/10/20	B-ALL	56~58,XX,-X,+4,+5,+6,+der(9)del(9)(p13)del(9)(q22),+10,+14,+16,+17,+18,+18,+21(cp10)/46,XX[13]	nuc ish(CEP4,CEP10)x3[180/200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6x2,RUNX1x3)[184/200] nuc ish(CEP8x2,MYCx2,IGHx3)[182/200] nuc ish(CRLF2x3)[182/200]	a high-hyperdiploid tumor genome (~56-58 chromosomes). Findings include gain (3 copies) of chromosomes X, 4, 5, 6, 10, 14, 16, 21, (4 copies) of chromosome 18, (3 copies) of chromosome 9p13.2q22	a complex genomic alteration pattern on chromosome 17, consistent with the gain (3 copies) of whole chromosome 17 with intermittent 2 copy regions, and a likely loss of heterozygosity on 17q21.2q21.31 involving STAT5B and STAT3 genes.	a high hyperdiploid genome with multiple chromosome gains (as listed in the table) including gains of one extra copy of whole chromosomes X, 4-6, 9, 10, 14, 16, 21 and two extra copies of chromosome 18 (tetrasomy 18).	Additionally, gain of 17p and partial 17q was identified.	None (Category 3)
186	M	4	7/7/20	B-ALL	54,XY,+X,dup(6)(p22pter),+8,+10,+14,+17,+18,+21,+21(3)/55,sj,+4,-6,+dup(6)(p22pter)[12]/56,sdl1,-9[7]/46,XY[3]	nuc ish(CEP4,CEP10)x3[180/200] nuc ish(ABL1x3,BCRx2)[76/200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6x2,RUNX1x4)[158/200] nuc ish(ETV6x2,RUNX1x3)[24/200] nuc ish(CEP8,MYC,IGH)x3[184/200] nuc ish(CRLF2x3)[182/200]	a high-hyperdiploid tumor genome (~56 chromosomes). Findings include gains of whole chromosomes X (2 copies), 4, 8, 10, 14, 17, 18 (3 copies), and 21 (4 copies) in ~60% of cells and gain of chromosome 9 (3 copies) in 45% of cells.	a complex segmental copy number alteration pattern (gains and losses) in conjunction with LOH on 6p25.3p22.1 (~70% mosaicism), a deletion on 3p22.3 involving ARPP21 gene, and a deletion on 12p13.2 involving ETV6 gene.	a high hyperdiploid genome with multiple chromosome gains (as listed in the table), including gains of one extra copy of whole chromosomes X, 4, 8, 9, 10, 14, 17, 18, and two extra copies of chromosome 21 (tetrasomy 21)	gain with loss of heterozygosity of partial 6p (including TPMT), and loss of partial 12p (including partial ETV6).	None (Category 3)
187	M	7	6/26/20	B-ALL	46,XY,del(13)(q27)[10]/46,XY[10]	nuc ish(CEP4,CEP10)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6,RUNX1)x2[200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(CRLF2x2)[200] nuc ish(ABL1x2)[200] nuc ish(ABL2x2)[200] nuc ish(PDGFRb2x2)[200]	a complex mosaic segmental loss on chromosome 13q13.1q22.2 (~85% mosaicism) including BRCA2 gene.	a 28.06 Mb mosaic copy neutral loss of heterozygosity (cnLOH) on chromosome 9p24.3p21.1. In addition, a low level mosaic allelic imbalance was detected on the terminal 15 Mb region of 6p25.3p23 chromosome	None	cnLOH of partial 9p (containing JAK2 and CDKN2A/B, E2H2 (NM_004456.4), c.409_410insGT (p.Glu137Glyfs*4), ATRX (NM_000489.4), c.4749_4752del (p.Lys1583Asnfs*72))	None (Category 3)
188	M	7	1/14/21	T-ALL	45,X,-Y,del(11)(q714)[17].ish t(5;14)(q35.1;q32)[RANBP17+,TLX+,GH+]/IGH-,RANBP17-,TLX3+/46,XY[3]	nuc ish(RANBP17x2,TLX3x3)[RANBP17 con TLX3x2][194/200] nuc ish(KMT2Ax2)[194/200] nuc ish(CRLF2x1)[190/200] nuc ish(ABL1,BCR)x2[200] nuc ish(ETV6,RUNX1)x2[200] nuc ish(CEP8,MYC,IGH)x2[200]	loss of whole chromosome Y, and 36.2 Mb loss on 11q14.1q23.1.	a 1.4Mb loss on 9p21.3 including homozygous loss of the CDKN2A and CDKN2B genes. Two additional losses were observed in a small percentage of cells: loss of 2.8 Mb on 10q11.22q11.23 and loss of 5.4 Mb on 13q14.13 including the RB1 gene.	loss of partial 11q, and loss of CRLF2.	homozygous loss of CDKN2A/B on chromosome 9p, USH2A (NM_206933.2), c.2072G>T (p.Cys691Phe) PHF6 (NM_032458.2), c.904del (p.His302Ilefs*49) NOTCH1 (NM_017617.4), c.4793G>C (p.Arg1598Pro) NOTCH1 (NM_017617.4), c.4754T>C (p.Leu1585Pro) FBXW7 (NM_033632.3), c.726G>A (p.Gln242Gln) FBXW7 (NM_033632.3), c.1669G>A (p.Gly557Arg)	None (Category 3)
189	F	24	1/21/20	B-ALL	47,XX,del(11)(p11.12p14.3),+21c[19]/47,XX,+21c[1]	nuc ish(CEP4,CEP10)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6x2,RUNX1x3)[200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(CRLF2x2)[200]	apparently non-mosaic gain of chromosome 21, a 28Mb deletion of 11p14.3p11.12	a 33Mb copy neutral cnLOH of 9p24.3p13.3, a 63Kb homozygous deletion of 9p21.3 involving CDKN2A/B, a deletion of the short arm of chromosome 17 (17p13.3p11.2), a partial 17p as well as the entire 17q (17p11.2q25.3 and a gain of chromosome 21 (apparently non-mosaic). A 341Kb deletion of Xp21.1 and a 523Kb deletion of 13q21.31	trisomy 21, partial loss of 11p involving the WT1 gene.	homozygous loss of partial 9p involving CDKN2A/B, IKZF1 (NM_006060.5), c.550C>T (p.Arg184Trp) PAX5 (NM_016734.2), c.963dup (p.Ala322Argfs*19) KMT2D (NM_003482.3), c.15546_15550delinsTACTCCCCCCCC (p.Leu5183Thrfs*18) PTPN11 (NM_002834.4), c.215C>T (p.Ala72Val)	None (Category 3)
190	M	4	5/12/20	B-ALL	53,XY,+X,+6,+10,+14,+18,+21,+21(5)/46,XY[15]	nuc ish(CEP4x2,CEP10x3)[190/200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6x2,RUNX1x3)[176/200] nuc ish(ETV6x2,RUNX1x3)[20/200] nuc ish(CEP8x2,MYCx2,IGHx3)[190/200] nuc ish(CRLF2x3)[192/200]	a high-hyperdiploid tumor genome (~53 chromosomes). Findings include gains (3 copies) of whole chromosomes 6, 10, 14, and 18, gain (4 copies) of chromosome 21, and gain (2 copies) of the X chromosome.	a mosaic 299kb loss of 13q14.11 was identified, which includes the entire FOXO1 gene.	a high hyperdiploid genome with gains of one extra copy of whole chromosomes X, 6, 10, 14, 18 and two additional copies of chromosome 21	KRAS (NM_033360.3), c.35G>C (p.Gly12Ala)	None (Category 3)
191	M	10	5/12/20	B-ALL	46,der(X)(q28->q21::p22.33->q28),Y,del(12)(p13.1p13.2),t(12;21)(p13;q21)[6].ish del(12)(ETV6-),t(12;21)(ETV6-RUNX1+ETV6-RUNX1+)/46,idem.,t(7;12)(p22;p13)[3].ish t(7;12)(RUNX1+ETV6-RUNX1-,ETV6)/46,XY[11]	nuc ish(CEP4,CEP10)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6x1,RUNX1x3)[ETV6 con RUNX1x1][148/200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(CRLF2x1)[140/200] nuc ish(ABL1x2)[200] nuc ish(ABL2x2)[200] nuc ish(PDGFRb2x2)[200]	a 1.9 Mb loss of chromosome 12p13.2p13.1 (involving the ETV6 gene), a 2.8 Mb loss of Xp22.33 (including the CRLF2 gene) as well as a 71.6 Mb gain of Xq21.1q28	None	ETV6::RUNX1, loss of partial 12p (including ETV6), loss of partial Xp (including CRLF2), and gain of partial Xq (including STAG5, BCORL1, PHF6, and RPL10).	TINF2 (NM_001099274.1), c.297+3_297+4insGCCCCCCG (p.?)	a derivative chromosome (Category 2)
192	M	5	5/5/20	B-ALL	45,XY,der(6)(pter->q17::21q21-21q22::12p13->12pter),der(12)(6qter->6q25::6q16->6q17::12p172->12ater),der(12)(11:22)(p11.2::q11.1),der(13;15)(q10;q10),der(21)(21pter->21q21::12p172->12p13::21q22->21q22::21q22->21qter),22[16].ish der(6)(ETV6+RUNX1-),der(12)(11;22)(ETV6-BCR+),der(21)(dimRUNX1)x2/46,XY[4]	nuc ish(CEP4,CEP10)x2[200] nuc ish(ABL1,BCR)x2[200] nuc ish(KMT2Ax2)[200] nuc ish(ETV6x1,RUNX1x4)[ETV6 con RUNX1x1][196/200] nuc ish(CEP8,MYC,IGH)x2[200] nuc ish(CRLF2x2)[200]	a ~ 22.4 Mb loss of 12p13.3p12.1 (including ETV6), a ~6.1 Mb loss of 12p12.1p11.22, A 23.1 Mb gain of 21q11.1q22.11 and a ~11.7 Mb gain of 21q22.1q22.3 (including RUNX1).	a 775 kb loss of 2p16.1 (including the FANCL gene), a ~49.8 Mb loss of 6q16.2q25.1 and a ~228.9 kb loss of 13q12.11 involving the ZMYM2 and ZMYM5 genes.	ETV6::RUNX1, Loss of partial 12p involving ETV6 and partial gain of 21q, loss of partial 6q involving the MYB gene	NRAS (NM_002524.4), c.35G>A (p.Gly12Asp).	a complex rearrangement involving multiple chromosomes (Category 2)

193	F	17	5/5/20	T-ALL	45,X,t(X;12)(p10;q10),t(1;2)(p35;p21),del(5)(q23q33),del(7)(q31),del(12)(p12),-13,t(14;22)(q11.2;p11.2),der(17)t(13;17)(q11;p11.2)[cp14]/46,XX[6]	nuc ish[RANBP17,TLX3]x4[26/200] nuc ish[ABL1,BCR]x4[15/200] (ABL1x3,BCR)4[20/200] (ABL1x2,BCR)4[15/200] nuc ish[KMT2A]x4[20/200] nuc ish[ETV6x1,RUNX1x2][96/200] [ETV6x2,RUNX1x4][24/200] nuc ish[CEP8,MYC,IGH]x4[68/200] nuc ish[CRLF2x1][24/200]	a 2.8 Mb loss of 5q23.1q23.2, a 23.6 Mb loss of 5q23.2q33.1 (including the CSF1R gene), a 24.3 Mb loss of 12p13.3p12.1 (including the ETV6 gene), and a 18.0 Mb loss of 17p13.3p11.2 (including the TP53 gene). a 32.7 Mb low level mosaic loss of 7q31.11q36.3	a 40.3 Mb copy-neutral loss of heterozygosity (cnLOH) of 17q21.2q25.3	loss of partial 5q including CSF1R, loss of 12p including ETV6, and loss of 17p including TP53.	NOTCH1 (NM_017617.4), c.7327_7333del (p.Val2443Hisfs*32), SUZ12 (NM_015355.3), c.1109_1110insTCCTATT (p.Ala371Profs*39), RUNX1 (NM_001754.4), c.396_397insCCGGG (p.Met133Profs*14)	a derivative chromosome and two balanced translocations (Category 2)
194	M	10	5/4/20	B-ALL	47,XY,(9)(q10),del(12)(p12),+21c[3]/47,XY,der(9)(qter?q10:q10?q32,d,e)(12)(p12),+21c[3]/47,XY,+21c[1]	nuc ish[CEP4,CEP10]x2[200] nuc ish[ABL1x3,BCR]x2[96/200] nuc ish[KMT2A]x2[200] nuc ish[ETV6 amp,RUNX1x3][34/200]/[ETV6x3-4,RUNX1x3][8/200]/[ETV6x2,RUNX3][158/200] nuc ish[CEP8,MYC,IGH]x2[200] nuc ish[CRLF2x2][200]	complex alterations on chromosomes 9 and 12 and apparently non-mosaic gain of chromosome 21	None	complex alterations on chromosomes 9 and 12 and apparently non-mosaic gain of chromosome 21	None	an isochromosome (Category 2)
195	M	3	4/19/20	B-ALL	57,XY,+x,+4,+6,+10,+14,+14,+17,+18,+21,+21,+22[11]/46,XY[8].ish der(?)[CEP8,-MYC+]	nuc ish[CEP4x3,CEP10x2][175/200] nuc ish[ABL1x2,BCR]x3[185/200] nuc ish[KMT2A]x2[200] nuc ish[ETV6x2,RUNX1x4][170/200] nuc ish[CEP8x2,MYC]x3,IGHx4[170/200] nuc ish[CRLF2x3][177/200]	a high hyperdiploid tumor genome (~57 chromosomes) with multiple mosaic gains, involving three copies of chromosomes X, 4, 6, 10, 17, 18, and 22 and four copies of chromosomes 14 and 21.	low level mosaic complicated CNVs on chromosome 8.	a high hyperdiploid genome with multiple chromosome gains, including gains of one extra copy of whole chromosomes X, 4, 6, 10, 17, 18, 22 and two additional copies of chromosome 14 and 21	gain of partial 8q and copy-neutral loss-of-heterozygosity of chromosome 9	None (Category 3)
196	M	17	3/2/20	AML	46,XY,t(1;3)(q12;q21),t(8;21)(q22;q22)[19]	nuc ish[RUNX1T1,RUNX1]x3[RUNX1T1 con RUNX1x2][200]	None	a 19.65Mb mosaic loss of chromosome 2p23.3p21 including EML4 and ALK, a 57.03Mb mosaic cnLOH of chromosome 11q14.1qter including CBL and KMT2A, a 495.2kb mosaic loss of chromosome 6p24.1p23 and a 244.6kb mosaic loss of chromosome 15q24.1 containing PML	RUNX1:RUNX1T1	cnLOH of partial 11q (contains the CBL gene); CBL (NM_005188.3), c.1228-2A>G (p.?), TP53 (NM_000546.5), c.818G>A (p.Arg273His)	a balanced translocation (Category 2)
197	F	10	1/7/20	B-ALL	47,XX,+21[9]/46,XX[11]	nuc ish[CEP4,CEP10]x2[200] nuc ish[ABL1,BCR]x2[200] nuc ish[KMT2A]x2[200] nuc ish[ETV6x2,RUNX1x3][162/200] nuc ish[CEP8,MYC,IGH]x2[200] nuc ish[CRLF2x2][200] nuc ish[ABL1x2][200] nuc ish[ABL2x2][200] nuc ish[PDGFRb]x2[200]	a gain of chromosome 21 (~60% mosaicism).	a 424kb gain (~50% mosaicism) and a 357kb loss (~25% mosaicism) both involving DMD gene in Xp21.1, a 1.2Mb deletion of 16p11.2 (~65% mosaicism).	gain of whole chromosome 21	IKZF1 (NM_006060.5), c.475A>T (p.Asn159Tyr)	None (Category 3)
198	F	3	1/7/20	B-ALL	48,XX,+x,+21c[1]/47,XX,+21c[19]	nuc ish[CEP4,CEP10]x2[200] nuc ish[ABL1,BCR]x2[200] nuc ish[KMT2A]x2[200] nuc ish[ETV6x2,RUNX1x3][200] nuc ish[CEP8,MYC,IGH]x2[200] nuc ish[5'CRLF2x1.3'CRLF2x3][5'CRLF2 con 3'CRLF2x1][105/200]	a gain of chromosomes X (~70% mosaicism) and 21 (apparently non-mosaic).	a 314kb loss in Xp22.33 involving P2RY8, a 2.3Mb loss of 2p11.2 (~70% mosaicism), a 26Mb copy-neutral loss of heterozygosity (cnLOH) affecting the short arm of chromosome 6 (~70% mosaicism),	Trisomy 21, gain of whole chromosome X	P2RY8::CRLF2, JAK2 (NM_004972.3), c.2049A>T (p.Arg683Ser), IKZF1 (NM_006060.5), c.560_563delinsAGGGGGATAG (p.Ala187_Leu188delinsGluGlyAspSer),	None (Category 3)
199	M	10	2/28/20	AML	45,X,-Y,t(8;21)(q22;q22)[20]	nuc ish[RUNX1T1x3,RUNX1x3][RUNX1T1 con RUNX1x2][196/200]	a mosaic loss of the whole Y chromosome detected in approximately 75% of cells.	None	RUNX1:RUNX1T1, Loss of chromosome Y.	FLT3 (NM_004119.2) c.1791_1883dup (p.?), RAD21 (NM_006265.2) c.2T>C (p.?), JAK3 (NM_000215.3) c.1970G>A (p.Arg657Gln)	None (Category 3)
200	F	7	2/28/20	T-ALL	46,XX,t(10;11)(p12;q14),del(12)(p13pter)[10].ish del(12)[ETV6-]/46,XX[10]	nuc ish[RANBP17,TLX3]x2[200] nuc ish[ABL1,BCR]x2[200] nuc ish[KMT2A]x2[200] nuc ish[ETV6x1,RUNX1x2][156/200] nuc ish[CEP8,MYC,IGH]x2[200] nuc ish[CRLF2x2][200]	a 13.4Mb mosaic loss of chromosome 12p13.3p13.1 which involves the ETV6 gene.	three mosaic regions of cnLOH were identified, including a ~36Mb region on 9p24.3p13.3 including almost the whole short arm of chromosome 9, a 1.1Mb region on 11p15.5, and a 40.9Mb region on 17q21.2q25.3.	PICALM::MLLT10. Heterozygous loss of partial 12p involving ETV6	cnLOH of partial 9p involving CDKN2A/B and cnLOH of partial 17q	None (Category 3)
201	F	12	2/28/20	AML	46,XX,t(15;17)(q24.1;q21.2)[9]/46,XX[11]	nuc ish[PML,RARA]X3[PML con RARAx2][60/200]	None	a 1.32Mb non-mosaic gain on chromosome 1q21.1q21.2 containing multiple genes, including GIAS, GJA8 and BCL9	PML::RARA	None	None (Category 3)

* The cyptic t(12;21)(p13;q22) was detected by metaphase FISH analysis, so that the two individuals were consider normal chromosomal analysis.