Supplementary Information

Supplementary Methods

Patient selection

We studied 87 cases with a blood smear available at diagnosis. The cases were first identified locally on the basis of a routine cell morphology screen. In blinded review by three independent cytologist (LB, CS, and KM), cases were only included if all three reviewers agreed with the diagnosis of B-PLL. A fourth blinded review of ten representative cases (five cases of B-PLL: BPLL_24, BPLL_26, BPLL_28, BPLL_33, BPLL_34; and 5 excluded cases) validated our approach. The four cytologists agreed on all five cases of B-PLL and on one excluded case (N°45). For the excluded cases N°76 and N°80, only two of the four cytologists diagnosed B-PLL; for the excluded cases N°49 and N°67, only one of the four cytologists diagnosed B-PLL (see Supplemental Table S1). A diagnosis of MCL was ruled out by karyotyping (K) and FISH assays: no *CCND1* rearrangements or other infrequent translocations involving *CCND2* and *CCND3* were observed. Unfortunately, bone marrow biopsies were not available for CCND1 or SOX11 immunostaining.

FISH

Standard FISH was performed on interphase nuclei and metaphases in all 34 cases. The specific probes were *ATM* (11q22), *TP53* (17p13), IGH/CCND3 (Cytocell, Cambridge, UK), D13S319 (13q14), centromere of chromosome 12 (Metasystems, Altlussheim, Germany), *MYC* (8q24) (DAKO, Santa Clara, CA and/or Zytovision, Bremerhaven, Germany), *MYCN* (2p24) (Abbott Molecular, Des Plaines, IL), *CCND1* (11q13) (DAKO). A break-apart probe for detecting *CCND2* (12p13) rearrangements was built using the home-grown bacterial artificial chromosome clones RP11-578L13 and RP11-388F6. A deletion probe targeting *TNFRSF10A-D* genes (8p21) was built with clones RP11-599A17 and RP11-692J4. Clones were selected using the University of California Santa Cruz Genome Bioinformatic database (NCBI38/hg38 build) and obtained from Genoscope (Evry, France). Extraction, labeling, and hybridization procedures were performed as described previously¹.

Cell sorting

The samples used for WES, targeted deep sequencing and RNA-Seq were obtained from fresh or cryopreserved mononuclear cells, when available. The CD19+ (and CD5+, IGL or IGK when appropriate) B-cells and CD3+ T-cells were sorted as described previously². The purity of the cell fractions was assessed by flow cytometry, and was always greater than 96%. DNA and RNA were extracted from sorted cell fractions using the All Prep DNA/RNA kit (Qiagen, Courtaboeuf, France), according to the manufacturer's recommendations.

Whole Exome Sequencing

DNA extracted from sorted CD19+ tumor cells (and CD5+, IGL or IGK when appropriate) and nontumor (CD3+) cells was used for exome capture with the SureSelect V5 Mb All Exon Kit (Agilent Technologies, Les Ulis, France) following the standard protocols. Paired-end sequencing (2 x 100 bp) was performed using HiSeq2000 sequencing instruments (Illumina, San Diego, CA). The mean coverage in the targeted regions was 106X (Supplemental Table S8). Reads were mapped to the reference genome hg19 using the Burrows–Wheeler Aligner (BWA) alignment tool version 0.7.10. PCR duplicates were removed using Picard Tools - MarkDuplicates (1.119). Local realignment around indels and base quality score recalibration were performed using GATK 3.2 (Genome Analysis ToolKit). Reads with a mapping quality score < 30 were ignored. SNVs and indels were called with VarScan2 somatic 2.3.7. The null hypothesis of equal allele frequencies between tumor and reference was tested using the two-tailed Fisher exact test. The variants were adopted as candidate mutations when P value was <0.01 and allele frequency was <0.1 in the reference sample. Variants were annotated with Annovar. We excluded synonymous single nucleotide variants (SNVs), variants located in intergenic, intronic, untranslated regions and non-coding RNA regions, and removed variants with mapping ambiguities. Mutations were searched in Catalogue of Somatic Mutations in Cancer database (http://cancer.sanger.ac.uk/cosmic/). The effect of the mutation was predicted by SIFT (http://sift.jcvi.org/) and PolyPhen2 (http://genetics.bwh.harvard.edu/pph2/) algorithms. Somatic copy number variations (CNV) were identified with Control-FREEC (v9.1).

Targeted deep sequencing

Primers flanking exons containing candidate somatic variants were designed using Primer3 (http://frodo.wi.mit.edu/primer3/). Short fragments of 100 to 200 bp were PCR-amplified from genomic DNA of sorted fractions and were subsequently pooled for library construction. PCR products were end-repaired, extended with an 'A' base on the 3' end, ligated with indexed pairedend adaptors (NEXTflex, Bioo Scientific) using the Bravo Platform (Agilent) and amplified by PCR for 4 cycles. Amplicon libraries were sequenced in an Illumina MiSeq flow cell using the onboard cluster method, as paired-end sequencing (2x150 bp reads) (Illumina, San Diego, CA). The mean coverage 2021X. was Quality of reads was evaluated using FastQC 0.11.2. (http://www.bioinformatics.bbsrc.ac.uk/projects/fastqc/). Reads were mapped to the reference genome hg19 using the Burrows-Wheeler Aligner (BWA) alignment tool version 0.7.10. Local realignment around indels and base quality score recalibration were performed using GATK 3.2 (Genome Analysis ToolKit). SNVs and indels were called with VarScan2 somatic 2.3.7.

Clonal organization

Evaluation of the clonal or subclonal nature of chromosomal abnormalities was performed by comparing the percentage of abnormal cells in interphasic FISH analyses (when available) with the percentage of monotypic (IGL or IGK) CD19+ cells defined by immunophenotyping. We considered an abnormality to be clonal when the difference between the two percentages did not exceed 20%, and subclonal when the difference was greater. For mutations detected in purified tumor cell fractions, we used the variant allele frequency (VAF) to distinguish between clonal and subclonal mutations with a threshold of 40% (or 80%, when combined with copy loss) to take account of the potential contamination by non-tumor cells.

RNA Sequencing and data analysis

RNA-Seq libraries were prepared using the SureSelect Automated Strand Specific RNA Library Preparation Kit as per manufacturer's instructions (Agilent technologies) and subjected to paired-end (101 bp) sequencing on HiSeq2000 (Illumina). Quality of reads was evaluated using FastQC 0.11.2. Sequences were filtered with Trimommatic and alignment was performed with Tophat2 version 2.0.14 and Bowtie1 version 1.0.0. The filtered reads were aligned to a reference genome hg19. In average, 88.95% of reads were aligned and counted with HTSeq (v0.5.4p5).

Differential expression analysis: raw counts of reads per Human GENCODE transcripts (GRCh37 release 24)³ were quantified with Salmon version 0.8.2⁴. These transcripts level raw counts have been aggregated to obtain gene level raw counts. Differential expression analysis was performed using DESeq2 package version 1.6.3⁵ with R statistical software version 3.1.2. Differentially expressed genes have been selected with an adjusted p-value of 0.01.

Gene Set Enrichment Analysis (GSEA)⁶ was performed using the GSEAPreranked tool (v3.0). Prior to conducting gene set enrichment analysis, differential expression analysis has been conducted using DESeq2 taking into account the batch effect observed in RNA-seq of B-PLL, CLL and pMCL cells. Only genes with an adjusted p-value lower than 5% have been considered. These genes have been ranked by log2 fold-change and captured in an RNK-formatted file. GSEAPreranked tool has been launched with the following option: 'scoring_scheme classic' using hallmark gene (H) and curated gene (C2) sets available from MSigDB (v6.2)⁷.

Functional analysis of the differential gene list has been explored for the GO gene sets with the Investigate application (http://software.broadinstitute.org/gsea/msigdb) to compute the statistical score of the overlaps between gene list and GO gene sets.

Fusion transcripts detection: sequences were aligned with Tophat2 version 2.0.14⁸ and Bowtie1 version 1.0.0 to a reference genome hg19. In average, 88.95% of reads were aligned. The potential

fusion transcripts were evaluated using Tophat2 –fusion-search option. A minimum of 10 supporting (spanning) reads was used as a threshold for fusion transcripts and the sequence around donor and acceptor sites of potential chimeric reads was manually evaluated to discard potential false positives.

In vitro cell viability and programmed cell death (PCD) assays

The viability of primary B-PLL cells was assessed with the ATP-based CellTiter-Glo 2.0 assay (Promega, Madison, WI). After pretreatment (or not) with the BET proteins inhibitors (iBET) OTX015 or JQ1 (500 nM), B cells were seeded onto 96-well plates (at a density 50×10^3 cells per well) and exposed for 48h to increasing concentrations of fludarabine, ibrutinib, idelalisib or venetoclax. We used drugs concentrations as previously described⁹. The OTX015 and JQ1 concentrations of 500 nM were the lowest with a significant *in vitro* effect on B-PLL cell viability (50% loss of viability; Supp Fig S5A and data not shown). The cells were then incubated for 15 minutes with 50 µl of CellTiter-Glo[®] reagent. Next, the luminescence was measured in an Infinite M1000 Pro plate reader (TECAN), using an integration time of 500 ms. Viability was determined by normalizing luminescence units against a DMSO control (the solvent used for drug dilution). Alternatively, PCD was measured by flow cytometry. Briefly, 2×10^6 B-PLL cells/ml were treated for 48h with ibrutinib (7.5 µM), idelalisib (50 µM), venetoclax (10 nM), or OTX015 (500 nM), and PCD was measured by annexin-V-APC (0.1 µg/ml; BD Biosciences) and propidium iodide (PI, 0.5 µg/ml; Sigma) co-labeling. All drugs were purchased from Selleckchem (Houston, TX). All these analyses were performed on stored cells resulting in spontaneous apoptosis rates of 10-15%.

Supplementary References

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Supplementary Tables

Table S1. Results of the morphologic review

		Reviewe	er 1	Review	/er 2	Reviewe	er 3	Level of	Reviewe	er 4	Level of
ID	ID article	% of prolymphocytes	Diagnosis	% of prolymphocytes	Diagnosis	% of prolymphocytes	Diagnosis	agreement between the first 3 reviewers	% of prolymphocytes	Diagnosis	agreement between the 4 reviewers
1	BPLL_1	68	B-PLL	80	B-PLL	73	B-PLL	B-PLL: 3/3			
2	BPLL_2	57	B-PLL	60	B-PLL	80	B-PLL	B-PLL: 3/3			
3	BPLL_3	71	B-PLL	72	B-PLL	56	B-PLL	B-PLL: 3/3			
4	BPLL_4	90	B-PLL	92	B-PLL	97	B-PLL	B-PLL: 3/3			
5	BPLL_5	90	B-PLL	94	B-PLL	89	B-PLL	B-PLL: 3/3			
6	BPLL_6	74	B-PLL	90	B-PLL	88	B-PLL	B-PLL: 3/3			
7	BPLL_7	74	B-PLL	80	B-PLL	76	B-PLL	B-PLL: 3/3			
8	BPLL_8	76	B-PLL	64	B-PLL	67	B-PLL	B-PLL: 3/3			
9	BPLL_9	70	B-PLL	85	B-PLL	93	B-PLL	B-PLL: 3/3			
10	BPLL 10	95	B-PLL	97	B-PLL	88	B-PLL	B-PLL: 3/3			
11	BPLL_11	83	B-PLL	73	B-PLL	79	B-PLL	B-PLL: 3/3			
12	BPLL_12	63	B-PLL	60	B-PLL	58	B-PLL	B-PLL: 3/3			
13	BPLL_13	90	B-PLL	93	B-PLL	96	B-PLL	B-PLL: 3/3			
14	BPLL_14	85	B-PLL	68	B-PLL	77	B-PLL	B-PLL: 3/3			
15	BPLL_15	82	B-PLL	73	B-PLL	55	B-PLL	B-PLL: 3/3			
16	BPLL_16	70	B-PLL	92	B-PLL	95	B-PLL	B-PLL: 3/3			
17	BPLL_17	70	B-PLL	66	B-PLL	67	B-PLL	B-PLL: 3/3			
18	BPLL_18	95	B-PLL	89	B-PLL	93	B-PLL	B-PLL: 3/3			
19	BPLL_19	97	B-PLL	98	B-PLL	93	B-PLL	B-PLL: 3/3			
20	BPLL_20	70	B-PLL	84	B-PLL	75	B-PLL	B-PLL: 3/3			
21	BPLL_21	75	B-PLL	96	B-PLL	93	B-PLL	B-PLL: 3/3			
22	BPLL_22	80	B-PLL	93	B-PLL	92	B-PLL	B-PLL: 3/3			
23	BPLL_23	80	B-PLL	81	B-PLL	72	B-PLL	B-PLL: 3/3			
24	BPLL_24	80	B-PLL	91	B-PLL	68	B-PLL	B-PLL: 3/3	62	B-PLL	B-PLL: 4/4
25	BPLL_25	68	B-PLL	66	B-PLL	63	B-PLL	B-PLL: 3/3			
26	BPLL_26	90	B-PLL	95	B-PLL	96	B-PLL	B-PLL: 3/3	92	B-PLL	B-PLL: 4/4
27	BPLL_27	100	B-PLL	100	B-PLL	99	B-PLL	B-PLL: 3/3			
28	BPLL_28	84	B-PLL	86	B-PLL	70	B-PLL	B-PLL: 3/3	78	B-PLL	B-PLL: 4/4
29	BPLL_29	79	B-PLL	74	B-PLL	75	B-PLL	B-PLL: 3/3			
30	BPLL_30	56	B-PLL	65	B-PLL	63	B-PLL	B-PLL: 3/3			
31	BPLL_31	80	B-PLL	85	B-PLL	83	B-PLL	B-PLL: 3/3			
32	BPLL_32	71	B-PLL	83	B-PLL	71	B-PLL	B-PLL: 3/3			
33	BPLL_33	71	B-PLL	78	B-PLL	90	B-PLL	B-PLL: 3/3	80	B-PLL	B-PLL: 4/4
34	BPLL_34	75	B-PLL	72	B-PLL	79	B-PLL	B-PLL: 3/3	75	B-PLL	B-PLL: 4/4
35	excluded (MCL)	57	B-PLL	64	B-PLL	55	B-PLL	B-PLL: 3/3			
35	excluded (MCL)	61	B-PLL	57	B-PLL	58	B-PLL	B-PLL: 3/3			
3/	excluded (MCL)	88	B-PLL	/8	B-PLL	/5	B-PLL	B-PLL: 3/3			
38	excluded (MCL)	/5	B-PLL Other	/9	B-PLL	80	B-PLL	B-PLL: 3/3			
39	excluded	<10	Other	<10	MZL ?	<10	MZL ?	B-PLL: 0/3			
40	excluded	<10	Other	23	MZL ?	<10	Other	B-PLL: 0/3			
41	excluded	46	Other	13	Other	13	Other	B-PLL: 0/3			
42	excluded	<10	MZL ?	<10	MZL ?	<10	Other	B-PLL: 0/3			
43	excluded	<10	Other	<10	Uther	<10	Other	B-PLL: U/3			
44	excluded	53	Other	66	B-PLL	53	Other	B-PLL: 1/3			

		Reviewe	er 1	Review	ver 2	Reviewe	r 3	Level of	Reviewe	er 4	Level of
ID	ID article	% of prolymphocytes	Diagnosis	% of prolymphocytes	Diagnosis	% of prolymphocytes	Diagnosis	agreement between the first 3 reviewers	% of prolymphocytes	Diagnosis	agreement between the 4 reviewers
45	excluded	<10	Other	<10	MZL	<10	MZL	B-PLL: 0/3	40	other	B-PLL: 0/4
46	excluded	48	HCL_V ?	<10	MZL	<10	MZL	B-PLL: 0/3			
47	excluded	50	Other	<10	MZL	<10	MZL	B-PLL: 0/3			
48	excluded	35	Other	25	Other	45	Other	B-PLL: 0/3			
49	excluded	35	Other	64	B-PLL	23	Other	B-PLL: 1/3	45	other	B-PLL: 1/4
50	excluded	8	Other	64 atypical	Other	73 atypical	Other	B-PLL: 0/3			
51	excluded	50	Other	<10	MZL	47	MZL?	B-PLL: 0/3			
52	excluded	46	Other	31	Other	24	Other	B-PLL: 0/3			
53	excluded	15	Other	<10	atypical LPD	9	Other	B-PLL: 0/3			
54	excluded	65 atypical	Other	48 atypical	Other	49 atypical	Other	B-PLL: 0/3			
55	excluded	38	Other	47	Other	<10	Other	B-PLL: 0/3			
56	excluded	<10	Other	36	Other	17	Other	B-PLL: 0/3			
57	excluded	36	Other	51	Other	42	Other	B-PLL: 0/3			
58	excluded	30	Other	30	MCL?	24	Other	B-PLL: 0/3			
59	excluded	35	Other	14	MZL	25	MZL	B-PLL: 0/3			
60	excluded	50	Other	39	Other	33	Other	B-PLL: 0/3			
61	excluded	45	Other	31	Other	43	Other	B-PLL: 0/3			
62	excluded	28	Other	<10	Other	5	Other	B-PLL: 0/3			
63	excluded	30	Other	<10	Other	<10	MZL?	B-PLL: 0/3			
64	excluded	5	Other	<10	Other	<10	Other	B-PLL: 0/3			
65	excluded	67 atypical	Other	20	Other	18	Other	B-PLL: 0/3			
66	excluded	22	Other	0	Other	<10	Other	B-PLL: 0/3			
67	excluded	75	B-PLL	49	MZL	42	MZL	B-PLL: 1/3	40	MZL	B-PLL: 1/4
68	excluded	25	Other	15	Other	<10	Other	B-PLL: 0/3			
69	excluded	30	Other	41	Other	13	Other	B-PLL: 0/3			
70	excluded	35	Other	39	MCL?	29	Other	B-PLL: 0/3			
71	excluded	50	Other	35	Other	15	Other	B-PLL: 0/3			
/2	excluded	29	Other	3/	Other	19	Other	B-PLL: 0/3			
/3	excluded	<10	Other	/	Other	<10	Other	B-PLL: 0/3			
74	excluded	30	Other	1/	Other	25	Other	B-PLL: 0/3			
75	excluded	20	Other	/	Other	<10	Other	B-PLL: 0/3	<i>co</i>	0.011	0.011.0/4
/6	excluded	//	B-PLL Other	52	Other	52	Other	B-PLL: 1/3	60	B-PLL	B-PLL: 2/4
70	excluded	<10	Other	<10	Other	<10	Other	B-PLL: 0/3			
/8	excluded	6/	B-PLL Other	4/	MZL?	49	Other	B-PLL: 1/3			
79	excluded	50	Other	32	Other	20	Uther	B-PLL: U/3			
80	excluded	/0	B-PLL	50	MZL?	52	MZL?	B-PLL: 1/3	68	B-PLL	B-PLL: 2/4
81	excluded	25	Other	33	MZL?	12	Other	B-PLL: 0/3			
82	excluded	45	Other	50	Other	32	Other	B-PLL: 0/3			
83	excluded	15	Other	34	Other	<10	Other	B-PLL: 0/3			
84	excluded	40	Other	25	MZL?	38	Other	B-PLL: 0/3			
85	excluded	15	Other	7	Other	10	Other	B-PLL: 0/3			
86	excluded	28	Other	20	Other	<10	Other	B-PLL: 0/3			
87	excluded	32	Other	18	Other	<10	Other	B-PLL: 0/3			

Other: not B-PLL

Atypical: lymphocyte with nucleolus but not a typical prolymphocyte

MZL: marginal zone lymphoma; MCL: mantle cell lymphoma, LPD: lymphoproliferative disorder

Reviewer 4 conducted a blinded morphological review of 10 cases in a second round

Table S2. Immunophenotyping results for the patients with B-PLL

ID Patient	% CD5+ / CD19+	CD5 expression	% CD23+ / CD19+	CD23 expression	Matutes' score
BPLL_1	85	+	24	-	2
BPLL_2	4	-	96	+	1
BPLL_3	99	+	32	+	3
BPLL_4	99	+	44	+	3
BPLL_5	98	+	10	-	1
BPLL_6	70	+	85	+	3
BPLL_7	0	-	0	-	0
BPLL_8	76	+	21	-	2
BPLL_9	0	-	0	-	0
BPLL_10	97	+	5	-	1
BPLL_11	75	+	30	+	2
BPLL_12	0	-	0	-	1
BPLL_13	2	-	62	+	1
BPLL_14	10	-	11	-	1
BPLL_15	74	+	0	-	1
BPLL_16	63	+	8	-	2
BPLL_17	na	-	na	-	0
BPLL_18	0	-	0	-	0
BPLL_19	na	-	na	-	0
BPLL_20	53	+	2	-	2
BPLL_21	3	-	6	-	na
BPLL_22	na	+	na	-	1
BPLL_23	69	+	12	-	3
BPLL_24	32	+	10	-	1
BPLL_25	3	-	25	-	1
BPLL_26	na	+	na	-	na
BPLL_27	0	-	5	-	0
BPLL_28	0	-	78	+	2
BPLL_29	93	+	34	+	2
BPLL_30	97	+	17	-	3
BPLL_31	45	+	99	+	2
BPLL_32	7	-	98	+	1
BPLL_33	96	+	100	+	2
BPLL_34	0	-	7	-	0

A cut-off of 30% of CD19+ cells was used to define CD5 and CD23 positivity na: not available

Table S3. Karyotype and FISH results for patients with B-PLL

ID Patient	Karyotype (according to ISCN 2016)	MYC (0: normal, 1:translocation, 2: gain) *	17p deletion (TP53) *	Trisomy 18/18q	Trisomy 3/3q	13q14 deletion *	Trisomy 12 *	8p deletion (TNFRSF10) *	11q deletion (ATM) *
BPLL_1	46,XY,del(13)(q12q14)[8]/46,XY[31]	0	0	0	0	1 (64%)	0	0	0
BPLL_2	49,XY,+i(3)(q10),t(4;8)(q21;q24),del(9)(q21q32),+12,+18[20]	1 (67%)	0	1	1	0	1 (41%)	0	0
BPLL_3	44,XY,add(7)(q31),-9,del(14)(q24),- 15,del(17)(p13),add(21)(p11)[3]/44,s1,t(5;10)(q?21;p?11)[8]/44~46,sdl1,- del(14)(q24),der(14)del(14)(q24)add(14)(p10),+1~2mar[cp7]	0	1 (89%)	0	0	0	0	1 (5%)	0
BPLL_4	45,X,-Y,der(8)t(1;8)(q11;p21),t(<u>8</u> ;22)(q24;q11)[6]/45,X,- Y,der(8)t(1;8)(q11;p21)t(8;22)(q24;q11),t(<u>8</u> ;22)(q24;q11)[15]	1 (74%)	0	0	0	0	0	1 (83%)	0
BPLL_5	46,XY,add(1)(q2?5),?inv(2)(p24q14) or der(2) or dic(2.?),add(3)(p21),add(7)(p22),add(10)(q22),add(11)(q13),del(13)(q13q14),add or del(17)(p11),der(18)?t(11;18)(q13;q21)[10]/47,s1,+add(3)(p21)[2]	2 (3 copies, 5%)	1 (93%)	1**	1	1 (93%)	0	0	0
BPLL_6	46-47,XY,+3[10],ins(8)(q?22;?)[8],-9[8],+18[4],+mar1[3],+mar2[2][cp10]	2 (3 copies, 81%)	1 (18%)	1	1	1 (32%)	0	0	0
BPLL_7	46,XY,t(8;22)(q24;q11)[13]/46,XY[1]	1 (not quantified)	0	0	0	0	0	0	0
BPLL_8	48,XY,t(8;22)(q24;q11),+12,+18[15]	1 (89%)	0	1	0	0	1 (78%)	0	0
BPLL_9	46,XX,t(8;14)(q24;q32),del(10)(q23q25)[20]/45,idem,-15,der(16)t(15;16)(q1?4;p13)[8]/46,XX[2]	1 (80%)	0	0	0	0	0	0	0
BPLL_10	45,XX,t(2;8)(p11;q24),del(3)(p12),+add(3)(q11),der(8)t(8;9)(p12;q?21),-9,-17,der(19)t(9;19)(?p;q13)[20]	1 (94%)	1 (96 %)	0	0	1 (9%)	0	1 (89%)	0
BPLL_11	48,XX,t(8;14)(q24;q32),+12,del(13)(q13q22),+18[9]/47,sl,- X[8]/47,sdl1,t(1;6)(q43;q11),t(11; <u>14</u>)(q13;q32)[3]	1 (90%)	0	1	0	1 (80%)	1 (82%)	0	0
BPLL_12	49,XX,add(1)(q11),der(2)add(2)(p1?2)add(2)(q2?3),- 8,+der(12)add(12)(p1?2)add(12)(q?11),del(13)(q14q21),add(14)(q32),?add(20)(q13),+mar1,+mar2x2[cp6]	0	0	0	0	1 (56%)	1 (56%)	ND	0
BPLL_13	46-47,X,-Y,+3,t(8;14)(q24;q32),+18[cp12]/46,XY[3]	1 (80%)	0	1	1	0	0	0	0
BPLL_14	44,XX,del(4)(q11),del(6)(q14q24),-7,add(9)(p24),-10,-17,add(20)(q13),- 21,+2mar[24]/44,XX,idem,add(3)(q25)[2]	0	1 (79%)	0	0	1 (12%)	0	0	1 (85%)
BPLL_15	46,XX,der(4)?t(4;5)(q32;q15~21),-5,add(6)(q12),del(17)(p11),-19,- 20,+3mar[8]/46,XX,t(5;8)(p15;q11)[4]/46,XX[3]	0	1 (75%)	0	0	0	0	0	0
BPLL_16	not done	1 (88%)	1 (89%)	ND	ND	1 (90%)	0	ND	0
BPLL_17	46,XY,t(8;22)(q24;q11),del(10)(q22q26)[17]	1 (94%)	0	0	0	0	0	0	0
BPLL_18	47,XY,t(8;14;19)(q24;q32;p1?3),+12[15]	1 (94%)	0	0	0	0	1 (82%)	0	0
BPLL_19	48,XX,+4,t(8;14)(q24;q32),+12[17]	1 (78%)	0	0	0	0	1 (82%)	0	0
BPLL_20	47,XY,+15,add(17)(p12)[7]/46,SI,- Y[4]/46,XY,add(8)(q32)[5]/46,XY,der(12)t(8;12)(p13;q21),add(17)(p12)[2]/46,XY[3]	2 (3 copies, 20%)	1 (90%)	0	0	0	0	1 (6%)	0
BPLL_21	48,XY,add(4)(p12),t(8;14)(q24;q32),del(11)(p13p15),+12,t(13;16)(q14;p13),del(17)(p11p13),- 21,+2mar[9]/46,XY[1]	1 (74%)	0	0	0	0	1 (75%)	0	0
BPLL_22	46,X,add(X)(p21),der(8)add(8)(p11)t(8;22)(q24;q11),der(22)t(8;22)(q24;q11)[20]	1 (92%)	0	1**	0	0	0	1	0
BPLL_23	48,XY,t(8;14)(q24;q32),add(16)(q24),+der(18)t(3;18)(q?11;q?)x2[4]/46,XY[22]	1 (61%)	0	1	1	0	0	0	0
BPLL_24	46,XX,del(6)(q14q24),i(17)(q10)[11]/46,XX,t(7;7;14)(q11;q3?3;q32),del(17)(p12)[8]/46,XX[1]	0	1 (92%)	0	0	0	0	0	0
BPLL_25	40,X,add(X)(q21),t(1;8)(p10;p10),-5,- 6,add(7)(q31),der(9)t(3;9)(q13;q13),der(10)t(9;10)(q13;p12),add(12)(q14),der(13;22)(q10;q10)del(13)(q13 q21),-15,add(15)(p11),-16,tr(16),-17,add(17)(p13),- 21,add(21)(p11),add(22)(q13),+1~2mar[16]/80,s1x2[4]/46,XX[9]	2 (4 copies, 42%; 8 copies, 5%)	1 (62%)	0	1	0	0	0	0

ID Patient	Karyotype (according to ISCN 2016)	MYC (0: normal, 1:translocation, 2: gain) *	17p deletion (TP53) *	Trisomy 18/18q	Trisomy 3/3q	13q14 deletion *	Trisomy 12 *	8p deletion (TNFRSF10) *	11q deletion (ATM) *
BPLL_26	48,XY,t(8;14)(q24;q32),+der(18)x2[4]/48,sl,del(3)(p11p22)[15]	1 (92%)	0	1	0	0	0	0	0
BPLL_27	46,XY,t(8;14)(q24;q32)[20]	1 (78%)	0	0	0	0	0	0	0
BPLL_28	44~46,XY,+Y,der(2)t(2;8)(p11;?),der(3)t(3;6)(p11;?),-6,der(7)t(7;14)(p22;q11),- 8,der(12)t(12;15)(p12;?),der(14)t(8;14)(q22;q32),der(15)t(15;17)(p11;q23)x2,der(16)t(15;16)(q23;q22),+2~ 3mar[cp20].	0	1 (51%)	0	0	1 (27%)	0	ND	0
BPLL_29	47,XX,t(8;14)(q24;q32),+mar[19]	1 (78%)	0	0	0	0	0	0	0
BPLL_30	46,XX,t(8;22)(q24;q11)[2]/46,XX[34]	1 (52%)	1 (70%)	0	0	0	0	1 (6%)	0
BPLL_31	48,XY,+3,t(8;14)(q24;q32),+mar1[12]/49,sl,+mar2[2]	1 (not quantified)	0	0	1	0	0	0	0
BPLL_32	49,XY,+3,der(7)t(7;8)(p21;?)del(7)(q33q35),r(8),+12,+18[13]/46,XY[7]	2 (amplification, 79%)	0	1	1	0	1 (74%)	1 (80%)	0
BPLL_33	46,XX,t(8;14)(q24;q32),der(10)t(1;10)(q11;p14)[16]/46,XX[7]	1 (86%)	0	0	0	1 (25%)	0	0	0
BPLL_34	45,XY,t(1;10)(p2?1;q25),t(3;11)(q23;p15),der(9)t(9;17)(q?22;q11),der(14)t(14;17)(p11;q?),- 17[16]/45,XY,der(9)t(9;17),t(10;20)(q24;p13),t(12;21)(q11;q22),der(14)t(14;17),-17[3]/46,XY[1]	0	1 <mark>(</mark> 87%)	0	0	0	0	0	0

0: absence; 1: presence; tri: trisomy; ND: not determined

* % of cells bearing the abnormality determined by interphase FISH analysis

** determined by WES and confirmed by FISH

Table S4. Recurrent copy-number variations from WES coverage data in 16 patients with B-PLL. Theminimally altered regions are cited with regard to the hg19 reference genome.

				Number of	
Losses	Start	End	Length	cases	Candidate genes
17p13	1629307	7677922	6048616	6	TP53
8p21	21924203	23432383	1508181	4	TNFRSF10A/B/C/D, EGR3
5q32-q33.1	149755481	149827491	72011	3	TCOF1, RPS14, CD74
9q21	71002296	79999625	8997329	3	no candidate genes
Xq28	152613014	153881710	1268697	2	no candidate genes
9q34	134379493	138927656	4548164	2	NOTCH1
13q14	45517397	53422012	7904616	2	RB1, MIR15A, MIR16-1
14q24	68758421	69263126	504706	2	ZFP36L1, RAD51B
19p13	1592411	2823326	1230916	2	no candidate genes

				Number of	
Gains	Start	End	Length	cases	Candidate genes
18q21.32-q23	56702915	77960906	21257991	6	BCL2
chromosome 3	361267	197762956	197401689	4	no candidate genes
chromosome 12	175960	133808310	133632350	4	no candidate genes
17q24	63923550	67243919	3320369	4	no candidate genes
1q31.3-q42	197063118	225140263	28077146	3	no candidate genes
8q24	127569322	128753330	1184009	3	МҮС
4q27-q35.2	121828503	190947668	69119165	2	no candidate genes

patient ID	IGHV		% homology	Unmutated/ minimally
BPLL_8	V1-2	95.5	significantly mutated	
BPLL_28	V1-2	100	unmutated	x
BPLL_30	V3-21	96.9	significantly mutated	
BPLL_21	V3-21	95.8	significantly mutated	
BPLL_29	V3-21	88.6	significantly mutated	
BPLL_6	V3-23	96.9	significantly mutated	
BPLL_22	V3-23	96.9	significantly mutated	
BPLL_33	V3-23	94.9	significantly mutated	
BPLL_13	V3-23	94.1	significantly mutated	
BPLL_10	V3-30	96.6	significantly mutated	
BPLL_5	V3-7	100	unmutated	x
BPLL_4	V3-74	97.2	minimally mutated	x
BPLL_1	V3-74	94.1	significantly mutated	
BPLL_32	V4-34	95. <mark>8</mark>	significantly mutated	
BPLL_34	V4-34	100	unmutated	х
BPLL_24	V4-39	95.5	significantly mutated	
BPLL_18	V4-39	88.3	significantly mutated	
BPLL_19	V4-4	96.2	significantly mutated	
BPLL_27	V4-4	93.3	significantly mutated	

Table S5. IGHV sequencing data for patients with B-PLL

Table S6. List of variants detected by WES in 16 patients with B-PLL. A total of 309 non-synonymous somatic mutations were identified in 287 genes. We validated 299 mutations by using targeted deep resequencing and/or RNA-Seq.

	Patient								nbBases	nbBas	VAF	nbBases	nbBase	VAF	Somatic_p-	Confirme	Confirmation	Sift	Polyphe
PRL1 Direction Dir	ID	Gene	Mutation Type	Mutation	#Chr	Position	BaseRef	BaseCalled	RefNorm	esAltN	Norm	RefTumo	s	Tumor	value	d (y/n)	method	score	n score
PFUL: IMAX Description Description <thdescription< th=""> Description <thdescript< td=""><td>BPLL_1</td><td>ZBTB40</td><td>missense</td><td>ZBTB40:NM_014870:exon2:c.T557G:p.M186R.</td><td>chr1</td><td>22816998</td><td>8 T</td><td>G</td><td>197</td><td>2</td><td>1.01</td><td>70</td><td>68</td><td>49.28</td><td>1.45E-29</td><td>У</td><td>MiSeq+RNA-Seq</td><td>NA</td><td>0.899</td></thdescript<></thdescription<>	BPLL_1	ZBTB40	missense	ZBTB40:NM_014870:exon2:c.T557G:p.M186R.	chr1	22816998	8 T	G	197	2	1.01	70	68	49.28	1.45E-29	У	MiSeq+RNA-Seq	NA	0.899
UPUL: UPUL: <th< td=""><td>BPLL_1</td><td>NFASC</td><td>missense</td><td>NFASC:NM_001005388:exon25:c.C2837T:p.T946l.</td><td>chr1</td><td>2.05E+08</td><td>) C</td><td>Т</td><td>234</td><td>1</td><td>0.43</td><td>84</td><td>74</td><td>46.84</td><td>4.98E-34</td><td>9</td><td>MiSeq</td><td>NA</td><td>NA</td></th<>	BPLL_1	NFASC	missense	NFASC:NM_001005388:exon25:c.C2837T:p.T946l.	chr1	2.05E+08) C	Т	234	1	0.43	84	74	46.84	4.98E-34	9	MiSeq	NA	NA
DELL (IPPRIM meane LEPTIMIN (1993) and a Grad public (1993) mean Grad public (1993) mean Max	BPLL_1	LBRTM4	missense	LRRTM4:NM_024993:exon3:c.C577T;p.L193F.	chr2	77746418	G	A	325	0	0	113	108	48.87	4.67E-52	ÿ	MiSeq	NA	NA
DFL1 IPO28 meane INFERM (Selde) and a Transformation of the second s	BPLL_1	LRRTM1	missense	LRRTM1:NM_178839:exon2:c.G160A:p.A54T.	chr2	80530785	i c	Т	129	0	0	50	35	41.18	4.79E-17	ÿ	MiSeq	NA	0
PHL 5 SIMP means SIMP (matrix) SIMP (matrix) dot SIMP (matrix) SIMP (matrix	BPLL 1	MYD88	missense	MYD88:NM_002468:exon5:c.T794C:p.L265P	chr3	3818264	1 T	С	264	0	0	88	77	46.67	8.37E-39	ý	MiSea+RNA-Sea	NA	0.999
IPPLI_1 IPPLI IPPLI IPPLI IPPLI <t< td=""><td>BPLL 1</td><td>SLMAP</td><td>missense</td><td>SLMAP:NM_007159:exon2:c.G341A:p.R114Q.</td><td>chr3</td><td>57817252</td><td>G</td><td>A</td><td>391</td><td>16</td><td>3.93</td><td>187</td><td>158</td><td>45.8</td><td>1.20E-45</td><td>ý</td><td>MiSea+RNA-Sea</td><td>NA</td><td>0.993</td></t<>	BPLL 1	SLMAP	missense	SLMAP:NM_007159:exon2:c.G341A:p.R114Q.	chr3	57817252	G	A	391	16	3.93	187	158	45.8	1.20E-45	ý	MiSea+RNA-Sea	NA	0.993
EPUL IPSO/T Instantingtong FOOT All 1985 conto 15, 105 accord 12, 2000	BPLL 1	SPICE1	missense	SPICE1:NM 144718:exon2:c.G10T:p.V4F.	chr3	1.13E+08	C	A	182	2	1.09	94	53	36.05	2.41E-19	ú	MiSea+BNA-Sea	NA	0.427
IPPL_1 IPSN/T IPSN/E IPSN/E<	BPLL 1	FBXW7	frameshift insertion	EBXW7:NM_018315;exop8;o_1156_1157;psCCGATGCGGT;p_C386fs	chr4	153E+08	-	ACCECATCEE	249	0	0	86	47	35.34	5 17E-25	ú	MiSea+BNA-Sea	NA	1
IPPL_1 IPPL matterse IPPL MULTIPET meanTis CostSAL predict other State State CostS	BPLL 1	FBXW7	missense	FBXW7:NM_018315;exon8;c.C1153G;p.B385G.	chr4	1.53E+08	G	С	257	2	0.77	85	56	39.72	6.99E-27	ú	MiSea+BNA-Sea	NA	0.06
IPPL_L IPPE messees IPPE MULTING Procession (1997) 3:08 india HM0085 C A 77 0 0 23 25 25 10 105 105 105 <	BPLL 1	LIFB	missense	LIFR:NM_001127671;exon17;c.G2435A;p.B812Q.	chr5	38485983	Ē	Ť	273	ō	0	111	69	38.33	1.44E-32	ú	MiSea	0.47	0.081
BPLL 192P1 essence State 192 193 49.0 106-0 y Messa N Ma BPLL 106 nearces VCDM y=000 (mass) 0.001 (BPLI 1	NEIB	missense	NEIB:NM_001190738; exop8:c G1307T:p S436	chr9	14120455	i c	Å	72	0	i n	23	25	52.08	7 42E-13	ú	MiSea	NA	0.063
BPLL LUE1 respons 30V LOBEND 00757 month CONTER (25%) oright 10 Oright 30 State 30 S	BPLL 1	SVEP1	missense	SVEP1/NM_153366; exop38; c A79581; p H26531	chr9	1 13E+08	T	A	322	3	0.92	131	35	42.04	4 00E-39	ú	MiSeq	NA	NA
BPLL Victor memory Victor Victor <td>BPLL 1</td> <td>LDB1</td> <td>stopgain SNV</td> <td>LDB1/NM_001113407;evop11;o_G1057T;p_E353X</td> <td>chr10</td> <td>1.04E+08</td> <td>Ċ</td> <td>A</td> <td>261</td> <td>2</td> <td>0.76</td> <td>88</td> <td>70</td> <td>44.3</td> <td>126E-32</td> <td>ú</td> <td>MiSea+BNA-Sea</td> <td>NA</td> <td>0.73539</td>	BPLL 1	LDB1	stopgain SNV	LDB1/NM_001113407;evop11;o_G1057T;p_E353X	chr10	1.04E+08	Ċ	A	261	2	0.76	88	70	44.3	126E-32	ú	MiSea+BNA-Sea	NA	0.73539
UPLL NUS memore MUSH Model Mo	BPLL 1	WDB11	missense	VDB11/NM_018117;exop24:o_T2936G:p_E979C	chr10	123E+08	T	G	122	1 ñ	0.10	55	63	53.39	3 18E-25	u u	MiSea+BNA-Sea	0.11	0.84
BPLL IBP Insertific meterine IPERPL Signal oth eth MA MA BPLL 1910 Insertific meterine Signal Color Signal Signal Color Signal Sig	BPLL 1	MUC6	missense	MUC6:NM_005961;evop31;c.45057G;p.N16865	chr11	1017744	ί τ	ř	1983	204	933	1304	198	13.18	1.51E-04	<u> </u>	BNA-Sea	0.48	MA
BPLL IPIC meanse SPCAMP_202049 cond defits G A 266 I 0.77 77 71 62.28 1. Messa 1. BPLL IPINE MESS	BPLL 1	BB1	frameshift insertion	BB1NM_000321;exop7:c 707_708insAG:p K236fs	chr13	48934252	-	46	71	2	2.74	8	31	79.49	6.41E-18	2	MiSea+BNA-Sea	NA NA	NA
BPIL 1 IPTP490 misses DTMP114 (L922) associal Child (L923)	BPLL 1	ISHC4	missense	SHC4-NM_203349;evon8:o_C1174T;p_B392\/	ohr15	49148218	6	A	268	1	0.37	97	71	42.26	193E-33	2	MiSeq Miseq		1
UPPLL4 LAPPIO missees CAPUTION MASTER Org Org State State Org Or	BDLL 1	DITENIMS	missense	DITDNM3/NM_031220-aven3/a C1421/a 0489	ohr17	6428760		~	138	4	2.82	47	56	54.37	8.91E-22	<u> </u>	MiSeq	0.05	0.956
UPPLL4 IF/SEC Part of the second of a ACRESS and the second of ACRESS and the ACRESS and ACRESS and the ACRESS and ACRESS and the ACRESS and AC	BDLL 4	CADN12	missense	CADN12.NM_144E7E	ohin	20974042		T	294		167	120	129	49.12	2.965-44	<u> </u>	MiGee	0.00	N0.000
UPD-L UNL Distance UNL NU COST is and 2, CADDA DUME choice Choice Cost Cost <td>BDLL 4</td> <td>CAFINIS CESE1</td> <td>missense</td> <td>CAPINIS.INF_144313.exect5.c. A2099C.c. K700E</td> <td>ohr2</td> <td>1995+09</td> <td></td> <td></td> <td>125</td> <td>5</td> <td>1.01</td> <td>50</td> <td>120 E0</td> <td>40.12</td> <td>4.355-17</td> <td></td> <td>MiSee+DNA-See</td> <td>N0.00</td> <td></td>	BDLL 4	CAFINIS CESE1	missense	CAPINIS.INF_144313.exect5.c. A2099C.c. K700E	ohr2	1995+09			125	5	1.01	50	120 E0	40.12	4.355-17		MiSee+DNA-See	N0.00	
Implicit	BDLL 4		missense	ULLINM 000EE1	- chi2	10100207			274		4.30	100	70	40.01	4.00L-11	<u> </u>	MiSerrinke-Seq		0.333
BPLL 4 INCH INSCREPT IA COD 2 164 57 40 1124 1012-5 9 165st 10A BPLL 4 IVI INSCREPT IVI INSCREPT IVI	BDLL_4		missense	VIII:NP_000001:ex012:0.0400A:p.01446.	chr3	47027252		<u> </u>	274	<u> </u>	0.30	97	64	20.31	1.046-01	<u> </u>	MiSeq+RNA-Seq		0.700
UNIT Discrime Discrime <thdiscrime< th=""> Discrime <thd< td=""><td>DPLL_4</td><td></td><td>missense</td><td>NDEAL2:NP_UID175:exon14:0.01340A:p.0000K.</td><td>chr3</td><td>47037253</td><td></td><td><u> </u></td><td>120</td><td></td><td>1.04</td><td>57</td><td>40</td><td>33.75</td><td>4.00E-23</td><td><u>y</u></td><td>MiSeq</td><td></td><td>NA NA</td></thd<></thdiscrime<>	DPLL_4		missense	NDEAL2:NP_UID175:exon14:0.01340A:p.0000K.	chr3	47037253		<u> </u>	120		1.04	57	40	33.75	4.00E-23	<u>y</u>	MiSeq		NA NA
UPH La Lish Control Missee Control Control A for any and a control of the contro	DPLL_4		splice site	INA LANVIANA 000000 A COMA LIONA	chro	1.000+00		<u><u></u></u>	120	4	1.04	51	40	41.24	1.046-14	9	Imioeq Mic		
UPTLE Missense PMORPHING MOSS Standard, Algology, PM, Colum Orb 1 Colum D <thd< th=""> <thd< th=""> <thd< th=""> <thd< th=""></thd<></thd<></thd<></thd<>	DPLL_4		missense	ADUCADIO AMA COACOE - 7 COACA - MONEAU	chr4	1405-00			103	0	4.52	33	10	44.04	0.11E-20	9	Mice d	0.13	0.53503
BPILL INADIC INSTRUCT Construct Constr	BPLL_4	ARRIGAPIU	missense		chr4	1.43E+00			34	1	1.05	51	34	35.73	3.4 IE-11	У	Mic VDNA C		0.97
BPLL UNAL/CompUNITY Second Control LinkAL, product One District L Bis L L L Bis L L	BPLL_4		missense	NSD I:NP_U22455:exon20:c.A60856:p.12023A.	chr5	1.77E+U8	<u> </u>	6	236		0.42	121	93	43.46	6.58E-35	<u>у</u>	MiSeq+RIVA-Seq		0.997
BPLLA Interest Interest <t< td=""><td>BPLL_4</td><td>JARIDZ</td><td>missense</td><td>[JARID2:NP_004973:exon7:0.117420:p.V581A.</td><td>chrb</td><td>15497198</td><td></td><td>L.</td><td>163</td><td></td><td>1.21</td><td>44</td><td>29</td><td>39.73</td><td>3.52E-15</td><td><u>y</u></td><td>Mibeq+RNA-beq</td><td>0.95</td><td>0.02</td></t<>	BPLL_4	JARIDZ	missense	[JARID2:NP_004973:exon7:0.117420:p.V581A.	chrb	15497198		L.	163		1.21	44	29	39.73	3.52E-15	<u>y</u>	Mibeq+RNA-beq	0.95	0.02
BPLL4 FULX Imagenetics FULX BPL4 FLA S M3 Z T S< S< S< <th< td=""><td>BPLL_4</td><td>KHURBSZ</td><td>missense</td><td>KHURBS2:NM_152688:exon6:c.U7751:p.P2535.</td><td>chr6</td><td>62604575</td><td><u> </u></td><td>A</td><td>102</td><td>2</td><td>1.92</td><td>30</td><td>28</td><td>48.28</td><td>3.83E-13</td><td>У</td><td>MiSeq+RNA-Seq</td><td>NA NA</td><td>0.979</td></th<>	BPLL_4	KHURBSZ	missense	KHURBS2:NM_152688:exon6:c.U7751:p.P2535.	chr6	62604575	<u> </u>	A	102	2	1.92	30	28	48.28	3.83E-13	У	MiSeq+RNA-Seq	NA NA	0.979
IPPL_4 IPAX4 splese IVA or/r I 127-138 C G C 2/5 B 3 C 3 B 8/7 I 288-30 y MBseq NA NA BPLL_4 IVCC missence ICLOST missence CDCDST/ML BBCL A NA D 32 BPLL_4 CDCDST missence CDCDST/ML BBCL A NB A NA D 32 BPLL_4 CDCDST missence CDCDST/ML BBCL C L43 O O TT BSO 4132 L382/7 MBseq NA D 40 D 32 BPLL_4 TPHCE Missence CASIT TSS C T TSS A BS 9 S2 A D 33 S3	BPLL_4	IND	missense	MD:NM_001164635;exon3;c.A473G;p.N1605.	chrb	1.51E+08	<u>A</u>	6	103	2	1.9	36	34	48.57	1.93E-14	9	MiSeq	NA NA	0.926
BPLL4 ICC missense IPMC-MUQUAB/ second_c AF30cp, V180L chr6 1.22±V8 G C 27b 5 T12 881 4.22±V8 Mage NA USe BPLL4 GCD057 missense DCD057/M1 MSe AT A NA C NA O O A C A A A A A A A A A A A A A	BPLL_4	PAX4	splice site		chr/	1.27E+U8		6	217	5	2.25	83	79	48.77	7.69E-30	У	MiSeq		NA
LBPLL4 LOLDS // missense DULUS ///PUR 3F845xeenon2 c AFSIGUL pN494F1 ohr 33/H480/LA A C H3 U 0 (1 500 41.32 11.82-01 y Ph3eq NA Out BPLL4 [IFL4] [IFL24] IFL24	BPLL_4	IMYC .	missense	MYC:NM_UU2467:exon2:c.G478C:p.V16UL.	chr8	1.29E+08		<u> </u>	216	6	2.7	15	112	88.19	4.32E-66	У	MiSeq+RNA-Seq	NA NA	0.92
BPLL4 BIG4 missense BICs4MU_017638:eon.2 cABSE.p.Y152. ohr11 ThE+U8 I A N8 4 2.57 b85 5.2 44.07 8.2.28-25 y MSseq NA 0.397 BPLL4 TIPHOE missense TIPHOEMU_013381eson.1 c4851p.1162. ohr11 118+08 A G 196 4 2.83 65 45.33 66.83 6.68.83 6.68.23 6.82.2 WSseq NA 0.937 BPLL4 TIPHOE missense TIPHOEMU_013381eson.1 c4851p.1162. ohr11 175763743 C T 198 4 198 32 48.17 30.228.2 WSseq NA 0.493 BPLL4 EPN3 missense FIN3MU_032821esonA2.66653Ap.R2200. ohr16 7077722 C T 53.35 5 137 256 80 23.2 18.84 19.84 10.84 10.83 BPLL4 EPN3MU_03208 CMT0 C353313 AT C 25.2 8 30.80 </td <td>BPLL_4</td> <td>CCDC67</td> <td>missense</td> <td>CCDC67:NM_181645:exon12:c.A1330C:p.N464H.</td> <td>chr11</td> <td>93141460</td> <td><u>A</u></td> <td>Ľ</td> <td>143</td> <td>U</td> <td>0</td> <td>1</td> <td>50</td> <td>41.32</td> <td>1.13E-20</td> <td>У</td> <td>MiSeq</td> <td>NA</td> <td>NA</td>	BPLL_4	CCDC67	missense	CCDC67:NM_181645:exon12:c.A1330C:p.N464H.	chr11	93141460	<u>A</u>	Ľ	143	U	0	1	50	41.32	1.13E-20	У	MiSeq	NA	NA
BPLL4 UPK2 missense UPK2/LWP/LUBE RUMP/LUBE RUMP/LUB	BPLL_4	BTG4	missense	BTG4:NM_017589:exon2:c.A1581:p.K53I	chr11	1.11E+08		<u>A</u>	146	4	2.67	66	52	44.07	8.21E-18	У	MiSeq	NA NA	0.997
BPLL4 I HHUE missense Mi	BPLL_4	UPK2	missense	UPK2:NM_UU6760:exon4:c.A356G:p.Y119U.	chr11	1.19E+08	A	G	196	4	2	83	69	45.39	2.26E-25	У	MiSeq	NA NA	0.997
BPLL4 TBCTD4 missense TBCTD4/M_UNB32 exeron2.c GB07Ap,E203K. ohr13 T5398 55 C T TB8 4 198 93 52 48,17 3.02E-30 y MSeq NA 0.98 BPLL4 FR01N-MU033221eron2.c G6850Ap,P22200. ohr18 1019717 C T 559 5 137 266 80 2312 115E-21 y MSeq NA 0.948 BPLL4 ZNF208 missense FM01N/MU0753 second-ac AS0Gp Ar/DR ohr18 2119717 C 252 8 3.08 105 102 49.28 2.37E-34 y MSeq=NA NA 0.93 BPLL4 ZBSP2 frameshift deletion ZBSP2/MU007398 seconds of TL 572detp.191.9104. ohr 393313 AT - 36 3.03 2 87 3.78E-47 y MSeq+PNA-Seq NA 0.42 BPLL5 K/FIB missense R/FIB/MU007338 seconds of TL 572detp.191.91194. ohr 1035710 G A 10.25 102	BPLL_4	TRHUE	missense	TRHUE:NM_U13381:exon1:c.U4851:p.1162I.	chr12	72667043			119	1	0.83	17	30	63.83	6.61E-20	У	MiSeq	0.13	0.548
BPLL_4 HYDIN missense HYDIN Missense HYDIN Missense K Missense NA NA BPLL_4 ZVF208 missense BCDR.ML.00123385.exon10:c.A4376G.p.NI\$55. chvX 39321444 T C 556 3 5.08 5 33 88.64 2.58E-13 y MissertNa-Seq NA 0.32 BPLL_5 K/FIB missense BCDR.NL.00123385.exon10:c.A4376G.p.NI\$55. chvX 39321444 T C 56 3 5.08 5 33 88.64 2.58E-13 y MissertNa-Seq NA 0.35 BPLL_5 K/FIB missense FLNB <m.00101631716:roxo14:c305510.0267m.ot 0.43<="" td=""> GC T<</m.00101631716:roxo14:c305510.0267m.ot>	BPLL_4	TBC1D4	missense	TBC1D4:NM_014832:exon2:c.G607A:p.E203K.	chr13	75936635	C	T	198	4	1.98	99	92	48.17	3.02E-30	У	MiSeq	NA NA	0.98
BPLL_4 FBN3 missense FBN3.MM_032447.exon18.c.C339G:p.P739R chr19 8191617 G C 172 5 2.82 16 14 46.67 5.00E-10 y MSeq NA 0.93 BPLL_4 2MF208 Image control A37002 C 257327 T C 252 8 3.08 105 102 43.28 2.377-34 y MSeq NA NA BPLL_4 2RSP2 framewhit deletion 2RSP2.MM_0070389 exon2e.c571572d driv C 255 3 3.95 3.97.75 3.95E-47 y MSeq NA 0.92 BPLL5 KP1B missense BCCR.MU_0010389 exon2e.c53867.p. P1194. chr.1 1035710 G A 182 0 0 75 3.27.1 1.27E-17 y MSeq NA 0.93 BPLL5 USD1 missense FLM8IM_001004317.exon12.c.03367.p. C10305.0 C T 181<0 0 223 23 50 18864	BPLL_4	HYDIN	missense	HYDIN:NM_032821:exon42:c.G6659A:p.R2220Q.	chr16	70977722	C	T	359	5	1.37	266	80	23.12	1.16E-21	У	MiSeq	NA	0.44481
BPLL_4 ZMF208 missense ZMF208 missense CMF208 Missense CMF208 Missense NA NA BPLL_4 ZMF208 frameshift deletion ZFSF2 Missense SUB1 XMF208 NA NA BPLL_5 KIF1B missense BCOR:NM_0007153:exon06.57, ST7574.02 ohx S3924444 T C 56 3 508 5 39 88.64 256E-13 y Miseq:PNA-Seq NA 0.92 BPLL_5 KIF1B missense KIF1B/MI_015074.exon35.0.G1873Ap.D627N1 ohr1 10357110 G A 182 0 0 72 35 32,71 127E-17 y Miseq NA 0.98 BPLL5 FUND missense FUND Ch17 10057110 G A 182 0 0 72 35 30.1 1048-42 y Miseq NA 0.95 BPLL5 USDT missense FUNDV01752:exon12.0.G1367.0.G13934p.C6440.0 ohr7<	BPLL_4	FBN3	missense	FBN3:NM_032447:exon18:c.02396G:p.P799R.	chr19	8191617	G	C	172	5	2.82	16	14	46.67	5.80E-10	У	MiSeq	NA	0.99
BPLL_4 ZRSR2 Itameshit deletion ZRSR2/LVM_C005085 exon0.c. S/T_572det; p131del chr/X TS833813 AT - 36 3 3.03 2 87 97.75 3.192-47 y MSeq+RNA-Seq NA BPLL_5 BCDR missense CTMM_0012385 exon0.c. S7L_572det; p.014535. chr/X 33921444 T C 55 3 5.08 5 33 8.84 2.58E-13 y MSeq+RNA-Seq NA 0.92 BPLL_5 ILS missense FLNB.ML_0105074:exon13.c.C33671.p. P113L. chr/A 76703885 C T 160 0 0 25 0.2 3.864 2.58E-13 y MSeq+NA-Seq NA 0.935 BPLL_5 USOT missense FLNB missense FLNB MSeq NA 0.955 BPLL_5 USOT missense PIPSX2.ML_015216:exon13.c.01474G.p.5432A. chr/s 102E-06 T G 195 1 0.51 2.5 58.33 10.055 58.33 1	BPLL_4	ZNF208	missense	ZNF208:NM_007153:exon4:c.A509G:p.K170R.	chr19	22157327	' T	С	252	8	3.08	105	102	49.28	2.37E-34	У	MiSeq	NA	NA
BPLL_4 BCOR missense BCORN_M_001723385:exon10: c.443762:p.11453. ohx 33321444 T C 56 3 508 5 33 88.64 2.56E-19 y MiSequen3eq NA 0.32 BPLL_5 KiFIB missense FLMB: M0.0107373Ap.D627N. ohr1 033710 G A 182 0 0 72 35 31.71 1.27E-77 y MiSeq MA-Seq NA 0.98 BPLL_5 IVIS01 missense FLMB: M0.00104317:exon21c.C33561:p.P1119L. ohr3 55103043 C T 160 0 755 102 39.63 187E-26 y MiSeq MA-Seq NA 0.93 BPLL_5 DISO1 missense PIPISK2. missense PIPISK2. MiSeq MA-Seq NA 0.021 BPLL_5 ZIMF425 missense RTM_000100518:exon13.0: T1474G:p.5492A ohr5 1.02E+08 T G 135 1 0.51 2.55 35.33 1016:e-25 y MiSeqHNA-Seq	BPLL_4	ZRSR2	frameshift deletion	ZRSR2:NM_005089:exon8:c.571_572del:p.191_191del.	chrX	15833813	AT AT	-	96	3	3.03	2	87	97.75	3.19E-47	У	MiSeq+RNA-Seq		NA
BPLL_S KIF18 missense KIF18:NM_00106374e.uon350.c.51873Ap.D627N. oh1 10357110 G A 182 0 0 72 35 32,71 1.27E-17 y MiSeq NA 0.38 BPLL_S FLNB missense FLNB missense V NA 0.355 0 0 175 100 0 0 155 102 33.68 187E-26 y MiSeq NA 0.355 BPLL_S DISO1 missense PIPISK2.NM_015216.exon13.o.T1474G:p.5492A oh14 76703985 G T 181 0 0 25 35 58.33 108E-21 y MiSeq NA 0.031 BPLL_S PIPISK2 missense RP14M_000269 exon4: c.G132Ap, V570M oh6 55538168 G A 358 1 0.49 130 140 51.86E-21 y MiSeq NA 0.331 BPLL_S RP1 missense RP14M_000269 exon6.c.G1726Ap, V570M oh6 <t< td=""><td>BPLL_4</td><td>BCOR</td><td>missense</td><td>BCDR:NM_001123385:exon10:c.A4376G:p.N1459S.</td><td>chrX</td><td>39921444</td><td>T</td><td>C</td><td>56</td><td>3</td><td>5.08</td><td>5</td><td>39</td><td>88.64</td><td>2.56E-19</td><td>У</td><td>MiSeq+RNA-Seq</td><td>NA</td><td>0.92</td></t<>	BPLL_4	BCOR	missense	BCDR:NM_001123385:exon10:c.A4376G:p.N1459S.	chrX	39921444	T	C	56	3	5.08	5	39	88.64	2.56E-19	У	MiSeq+RNA-Seq	NA	0.92
BPLL_S FLMB missense FLME:MU_001164377:exon21c.C33561_p.DF11SL ohr3 58103043 C T 160 0 0 155 102 33.63 1.87E-26 y Miseq=NANA 0.555 BPLL_S USD1 missense PPIPSX2.MU_015216:exon13:o.T1474Gp.5432A ohr5 10.22±08 T G 135 1 0.51 25 35 58.33 101E-25 y Miseq=NA-Seq NA 0.031 BPLL_S ZNF425 missense ZNF425.MU_010561:exon4:c.G133/h;p.G644D. ohr5 10.22±08 T G 135 1 0.49 130 140 5185 1.94E-42 y Miseq NA 0.033 BPLL_S RP1 missense RP1:MU_006263:exon4:c.G133/h; D644D. ohr3 55538168 G A 358 1 0.28 38 43 33.33 2.51E-28 y Miseq NA 0.334 BPLL_S GDF6 missense GDF6:MU_000100557:exon14:c.03304E/p.D1028/s. ohr8	BPLL_5	KIF1B	missense	KIF1B:NM_015074:exon19:c.G1879A:p.D627N.	chr1	10357110) G	A	182	0	0	72	35	32.71	1.27E-17	У	MiSeq	NA	0.98
BPLL_S USU1 missense PIPSK2.NM_015216:exon13:o.T1474G:p.S492A ohr4 //6/03985 L 181 U U 2.3 S0 1.862-21 y Missenthance NA NA BPLL_S PIPSK2.NM_015216:exon13:o.T1474G:p.S492A ohr5 1022+08 T G 195 1 0.51 225 35 58.33 101E-22 y Missenthance NA 0.021 BPLL_S ZMF425 missense RP14M2.50(0.00263):exon4:c.G1321A:p.U578M ohr7 1.49E+08 C T 203 1 0.49 130 140 5185 1.94E+42 y Misseq NA 0.031 BPLL_S GDF6 missense RP14M3:030Tp.K110N ohr8 55538168 G A 358 1 0.49 303.33 2.51E-28 y Misseq NA 0.332 BPLL_S KIF27 frameshift deletion KIF27:ML017576:exon14:c.3283deKp102028is. ohr9 86474138 C -<	BPLL_5	FLNB	missense	FLNB:NM_001164317:exon21:c.C3356T:p.P1119L.	chr3	58109043		<u> </u>	160	0		155	102	39.69	1.87E-26	У	MiSeq	NA NA	0.955
BPLL_S PHPSKZ missense PHPSKZMU_DISZID:exon13:o: 114 (45:p342A chris 102:E408 1 0.5 15 1 0.51 25 35 58.33 101:E25 y Missen2 N/A 0.001 BPLL_S ZMF425 missense ZMF425 Missen3e CMF425 Missen3e N/A 0.011 0.031 BPLL_S RP1 missense RP1 missense RP1 Missen3e CMF25 y Missen3e N/A 0.031 BPLL_S GDF6 missense GDF6.NM_001001575:exon14:c.3083delAp_D1026is. chr8 97172591 C A 319 9 2.74 85 104 55.03 5.80E-45 y Misseq NA 0.334 BPLL_S KIF27 frameshit deletion KIF27.NM_017576:exon14:c.3083delAp_D1026is. chr9 138E+08 C - 414 0 0 80 67 45.56 3.68E-46 y Misseq NA 0.39 0 0 67	BPLL_5	0501	missense		chr4	76703985	4 5	<u> </u>	181	U U		29	29	50	1.66E-21	У	MiSeq+RNA-Seq	NA	
BPLL_S ZMP42s missense ZMP42s/LML_00100/Bb1exon/t.c.C1931A; LD44U. chr/r 149-149 T30 T40 S105 T482-42 y MRsq NA U.034 BPLL_S GDF6 missense GDF6.ML_001001557:exon1c.G3301;p.K110N. chr/r 149-158 C A 358 1 0.49 530 140 510s T482-42 y MRsq NA U.033 BPLL_S GDF6 missense GDF6.ML_001001557:exon1c.G3301;p.K110N. chr/l 43538 C A 319 9 2.74 85 104 55.03 5.80E-45 y MRsq NA 0.334 BPLL_S KIF27 frameshit deletion KIF27:ML_0070576:exon16:c.03034e/kp.D1028/s. chr/l 118E+08 C - 414 0 0 80 67 45.58 3.68E-46 y MSeq NA NA BPLL_S NUP214 missense NUP214/NL_005050:exon18: c.2495A;p.Q819K. chr/l 113E+08 C A 330 <td>BPLL_5</td> <td>PPIP5K2</td> <td>missense</td> <td>PPIP5K2:NM_U15216:exon13:c. I 1474G:p. 5452A.</td> <td>chr5</td> <td>1.02E+08</td> <td></td> <td>6</td> <td>195</td> <td>1</td> <td>0.51</td> <td>25</td> <td>35</td> <td>58.33</td> <td>1.01E-25</td> <td>9</td> <td>MiSeq+RNA-Seq</td> <td>NA</td> <td>0.021</td>	BPLL_5	PPIP5K2	missense	PPIP5K2:NM_U15216:exon13:c. I 1474G:p. 5452A.	chr5	1.02E+08		6	195	1	0.51	25	35	58.33	1.01E-25	9	MiSeq+RNA-Seq	NA	0.021
BPLL_S HP1 missense HP1:NM_U0b2b3:exon8.col72bAp, b's10M. ch/8 555/3168 G A 358 1 0.28 38 43 33.33 2 51E-28 y Miseq NA 0.332 BPLL_S GDF6 missense GDF6/M_00100575:exon16: c.30301;p,K110N. oh/8 97172531 C A 399 9 2.74 85 104 55.05 y Miseq NA 0.332 BPLL_S KIF27 frameshift deletion KIF27.ML_017576:exon14: c.3083delAp,D1028fs. oh/9 86474138 C - 414 0 0 80 677 45.58 9.68E-46 y Miseq NA 0.332 BPLL_S T/// T missense T/// T1002180:exon8.c.226950;p,B00P oh/9 134E+08 C A 330 2 0.6 40 16 28.75 558E-13 y Miseq NA 0.332 BPLL_S MSP3 missense T/// T4187300. Oh/9 134E+08 C	BPLL_5	ZNF425	missense	ZNF425:NM_UU1001661:exon4:c.G1931A:p.G644U.	chr/	1.49E+U8			203	1	0.49	130	140	51.85	1.94E-42	У	MiSeq		0.031
BPLL_S GDF6 missense GDF5:MP_00700557:exon1b::G3301p; X10N, oh/6 97172591 C A 319 9 2.74 85 104 55.03 5.80E-45 y MiSeq NA 0.332 BPLL_S K/F27 frameshind delation (KF27/M0_01757):exon1b::0.3301p; X10N, oh/6 8474738 C - 414 0 0 80 67 45.56 3.68E-46 y MiSeq NA 0.332 BPLL_S INC missense TNC: MM_002160:exon1b::0.528950; P.B300P oh/9 118E+08 C G 98 2.2 102 58 36.25 183E-12 y MiSeq NA 0.339 BPLL_S IMSEr3 missense NUP214 MID00085:exon1b:: 0.2495A; p.Q819K oh/9 138E+08 C A 330 2 0.6 40 16 2.857 5.58E-13 y MiSeq+RNA-Seq 0.06 0.423 BPLL_S IMMSF3 missense IMMSF3NM_0001202.0:won5.c24917,9130W oh/11 13855270 <td>BPLL_5</td> <td>RP1</td> <td>missense</td> <td>RP1:NM_006269:exon4:c.G1726A:p.V576M.</td> <td>chr8</td> <td>55538168</td> <td>G</td> <td>A</td> <td>358</td> <td>1</td> <td>0.28</td> <td>98</td> <td>49</td> <td>33.33</td> <td>2.51E-28</td> <td>У</td> <td>MiSeq</td> <td>NA NA</td> <td>0.334</td>	BPLL_5	RP1	missense	RP1:NM_006269:exon4:c.G1726A:p.V576M.	chr8	55538168	G	A	358	1	0.28	98	49	33.33	2.51E-28	У	MiSeq	NA NA	0.334
BPLL_S KIF27 frameshit deletion KIF27.NM_007576:exon14:o.3083delAp.D1028is. ohn 88474138 C - 414 0 0 800 67 45.58 3.68E-46 y Miseq NA NA BPLL_S NUP missense TVC missense TVC/M_00210:exon18: 0.22950; P300P ohn 118E+08 C G 98 2 102 58 36.25 183E-12 y Miseq+RNA-Seq NA 0.08 BPLL_S NUP214 missense NUP214:NM_005085:exon18: C.2455A.p.Q819K. ohn 1.13E+08 C A 330 2 0.6 40 16 28.57 5.58E-13 y Miseq+RNA-Seq NA 0.08 BPLL_S TMS9F3 missense TMS9F3/MU_20123:exon2.c.01035; D.037H. ohn 13955270 C T 153 0 0 117 110 48.46 115E-31 y Miseq 0.06 0.423 BPLL_S OR8H2 missense OR8H2:NM_0010520:exon5.c.C494	BPLL_5	GDF6	missense	GDF6:NM_001001557:exon1:c.G3301:p.K110N.	chr8	9717259		A	319	9	2.74	85	104	55.03	5.80E-45	У	MiSeq	NA	0.932
BPLL_S INC missense INC:MM_UUZ/bU:evonito.i22935C,p.R300P. ohig 118E+08 C G 38 2 2 102 58 36.25 183E-12 y MiSeq NA 0.393 BPLL_S NUP214 missense INUP214 missense NUP214 MiSeq NA 0.433 0.6 40 16 28.57 558E-13 y MiSeq+RNA-Seq NA 0.030 BPLL_S IM3SF3 missense TM3SF3.ML_020123.evon2.c.G103C;p.D37H. chr10 98336580 C G 252 1 0.4 56 15 21.13 4.85E-10 y MiSeq+RNA-Seq 0.06 0.423 BPLL_S INAV2 missense INAV2.IMI_0011108:evon7.c.C12817; p.R430W. chr11 19355270 C T 153 0 0 114 170 16 47.85 2.40E-45 y MiSeq 0.40 380 BPLL_S AHNAK missense AHNAK/MUD01520:evon5.c.C4394T;p.P169L chr11 <t< td=""><td>BPLL_5</td><td>KIF27</td><td>frameshift deletion</td><td>KIFZ7:NM_017576:exon14:c.3083delA:p.D1028fs.</td><td> chr9</td><td>86474138</td><td></td><td>-</td><td>414</td><td></td><td></td><td>80</td><td>67</td><td>45.58</td><td>9.68E-46</td><td>У</td><td>MiSeq</td><td></td><td>NA</td></t<>	BPLL_5	KIF27	frameshift deletion	KIFZ7:NM_017576:exon14:c.3083delA:p.D1028fs.	chr9	86474138		-	414			80	67	45.58	9.68E-46	У	MiSeq		NA
BPLL_5 NUP214: missense NUP214: MISeq+RNA-Seq NA 0.08 BPLL_5 TM35F3 missense TM35F3: MISeq+RNA-Seq 0.61 252 1 0.4 56 15 21.3 4.85E-10 y Miseq+RNA-Seq NA 0.08 0.423 BPLL_5 NAV2 missense NAV2.NM_0001108:exon7.c: C1030:p.p.037H. chr10 98336580 C G 252 1 0.4 56 15 21.3 4.85E-10 y Miseq+RNA-Seq NA 0.08 0.838 BPLL_5 NAV2 missense NAV2.NM_00105200:exon1.c: C2817.p. FM30V. chr11 55872802 C T 260 3 1.14 170 156 47.85 2.40E-45 y Miseq 0.001 BPLL_5 AHNAK missense AHNAK.MI_0010520:exon5.c: C49391;p.F1665L chr	BPLL_5	INC	missense	TNU:NM_002160:exon8:c.G2699C:p.R900P.	chr9	1.18E+08		G	98	2	2	102	58	36.25	1.83E-12	У	MiSeq	NA	0.999
BPLL_S IMSSF3 missense IMSSF3/ML_020123.exon2.c.G103C.p.D37H. ohr10 98336580 C G 252 1 0.4 56 15 21.13 4.85E-10 y Miseq+RN-Seq 0.06 0.423 BPLL_S NAV2 missense NAV2.2 missense NAV2.2 Miseq+RN-Seq 0.06 0.423 BPLL_S OR8H2 Miseq+RN-Seq 0.411 13955270 C T 153 0 0 117 110 4.846 115E-31 y Miseq 0.13 0.001 BPLL_S OR8H2.NM_00105200.exon1b.c.264Tp.1735M ohr11 55872602 C T 260 3 114 170 156 47.85 2.40E-45 y Miseq 0.01 BPLL_S AHNAK missense AHNAK.VM_0010520.exon5.c.C4394Tp.P165L ohr11 6226695 G A 141 1 0.7 53 54 50.47 2.32E-23 y Miseq NA 0.6011 BPLL_S	BPLL_5	NUP214	missense	NUP214:NM_005085:exon18:c.C2455A:p.Q819K.	chr9	1.34E+08		A	330	2	0.6	40	16	28.57	5.58E-13	У	MiSeq+RNA-Seq	NA	0.08
BPLL_S INAV2 missense INAV2.NM_00111108:exon7.a.C12891;p.R430V. ohr11 19355270 C T T53 0 0 117 110 48.46 115E-31 y MSeq NA 0.380 BPLL_S DR8H2 missense DR8H2.MV_001005200: exon5.c C2847; p.155M. chr11 5527202 C T 260 3 114 170 156 47.65 y MSeq NA 0.801 BPLL_S ARNAK missense AHNAK.MM_001520: exon5.c C49347; p.1665L. chr11 62296895 G A 141 1 0.7 53 54 50.47 2.32E-23 y MSeq NA 0.60018 BPLL_S ARHCEF12 missense ARHCEF12.MV_001196685; exon14.c C1991; p.R400C chr11 1.2E+08 C T 428 0 0 102 35.44 2.90E-36 y MSeqH2Ne_q NA 1.854 BPLL_S FGF6 missense FGF6/NV_0.20398: exon25.c C44371; p.T50M chr14 1.2E+08	BPLL_5	TM9SF3	missense	TM9SF3:NM_020123:exon2:c.G109C:p.D37H.	chr10	98336580	U C	G	252	1	0.4	56	15	21.13	4.85E-10	У	MiSeq+RNA-Seq	0.06	0.423
BPLL_5 OR8H2_M missense OR8H2_MM_001005200_exon1b_C284T;p_135M. ohr11 55872002 C T 260 3 1.14 170 156 47.85 2.40E-45 y MSeq 0.13 0.001 BPLL_5 AHNAK missense AHNAK_M0_001620_exon1b_C.C4931T;p_1566L ohr11 6226685 G A 141 1 0.7 53 54 50.47 2.32E-23 y MSeq NA 0.6011 BPLL_5 AHNAK missense AHNAK_M0_001620_exon1b_C.C1931T;p_1600C ohr11 12E+08 C T 428 0 0 102 56 35.44 2.30E-36 y MSeq NA 0.6311 BPLL_5 FGF6 missense EGF6.NVL_020396.exon1b_C.C1431T;p_1750M. ohr12 4553300 G A 158 1 0.63 130 100 43.48 7.7E-27 y MSeq NA 0.354	BPLL_5	NAV2	missense	NAV2:NM_001111018:exon7:c.C1288T:p.R430W.	chr11	19955270	<u>) C</u>	Т	153	0	0	117	110	48.46	1.15E-31	У	MiSeq	NA	0.988
BPLL_5 IAHUAK: Missense IAHUAK:MM_001620:exon5:c.C43947:p.71665L ohr1 62:26695 G A 141 1 0.7 53 54 50.47 2.32E-23 y Miseq NA 0.6011 BPLL_5 ARHGEF12 missense AAHGEF12:MM_00198665:exon14:c.C11987:p.R400C. chr11 1.2E+08 C T 428 0 102 56 35.44 2.30E-36 y Miseq <na< td=""> NA 1.6011 BPLL_5 FGF6 Misense FGF6:NM_020396:exon2:c:C4397:p.1750M. chr12 453300 G A 158 1 0.63 130 100 43.48 7.37E-27 y Miseq NA 0.354</na<>	BPLL_5	OR8H2	missense	OR8H2:NM_001005200:exon1:c.C284T:p.T95M.	chr11	55872802	2 C	Т	260	3	1.14	170	156	47.85	2.40E-45	У	MiSeq	0.13	0.001
BPLL_5 ARHGEF12 missense ARHGEF12:NM_001198665:exon14:c.C1198T:p.R400C. chr11 1.2E+08 C T 428 0 0 102 56 35.44 2.90E-36 y Misseq+RNA-Seq NA 1 BPLL_5 FGF6 missense FGF6/NM_020996:exon2:c.C449T:p.T150M. chr12 4553300 G A 158 1 0.63 130 100 43.48 7.9TE-27 y Misseq NA 0.954	BPLL_5	AHNAK	missense	AHNAK:NM_001620:exon5:c.C4994T:p.P1665L.	chr11	62296895	G	A	141	1	0.7	53	54	50.47	2.32E-23	У	MiSeq	NA	0.60018
BPLL_5 [FGF6 missense FGF6:NM_020398:exon2:o.C449T:p.T150M. chr12 4553300 G A 158 1 0.63 130 100 43.48 7.97E-27 y MiSeq NA 0.954	BPLL_5	ARHGEF12	missense	ARHGEF12:NM_001198665:exon14:c.C1198T:p.R400C.	chr11	1.2E+08) <u>C</u>	Т	428	0	0	102	56	35.44	2.90E-36	У	MiSeq+RNA-Seq	NA	1
	BPLL_5	FGF6	missense	FGF6:NM_020996:exon2:c.C449T:p.T150M.	chr12	4553300) G	A	158	1	0.63	130	100	43.48	7.97E-27	У	MiSeq	NA	0.954

Patient					1			nbBases	nbBase	VAF	nbBases	nbBase	VAF	Somatic p-	Confirme	Confirmation	Sift	Polyphe
n la	Gene	Mutation Type	Mutation	#Chr	Position	BaseBef	BaseCalled	BefNorm	sAltNor	Norma	BefTumor	5	Tumor	value	d (who)	method	score	n score
BPLL 5	ALXIP	missense	MLXIP:NM_014938-evop1-c_G268C-p_E90Q	chr12	123E+08	G	C C	218	1	0.46	151	83	35.47	139E-26		MiSea	NA	NA
BPLL 5	VITSE	missense	INTS6-NM_012141;even14:o_01838G/n_H613B	ohr13	51948824	T	ř	53	t i	0.40	0	12	100	2 48E-13	<u> </u>	MiSeq+PNA-Seq	NA	0.999
	100	missense	NM_000E46. avera E T20EC	las17	7570224	т Т	č	1/9	L i	0.67	- ů	67	100	9.425-56	y	MiSeq+DNA_Seq	NO.	0.000
BPII 5		missense	TANE2:NM 025185; even 25: c C5596T; c B1866W	ohr17	61498939	 	⊢ ¥	173	+	114	213	121	36.23	9 19E-24	<u>y</u>	MiSea		
BDLL 5 G	STDN/	stongain SNV	STDN//NM_001039877.augo5.c C727T.a 02/2V	ohr19	47236306	i c	ò	230	2	1.17	1/13	91	39.99	2.94E_29	<u> </u>	MiSeq	0.06	0.639902
	ND	stopganitoriv	TND/MM_002295	- child	1755.00		Ť	100		1.23	193	65	20.00	1.525-19	<u>y</u>	MiGen	0.00	0.055002
DPLL_0		missense		Chri	1.132+00		T	100	3	0.00	104	104	30.40	1.00E-10	<u>y</u>	Mice	0.23	0.303
BPLL_D		missense	NYAP2:NP_02069:exon4:c.U7041:p.A235V.	chrz	2.26E+UC			263		0.30	001	124	43.37	0.30E-42	У	MiDeq	0.07	
BPLL_6	111088	missense	PYYU88:NP_UU2468:exon5:c.1734U;p.L265P	chr3	3818264		L	200		5.21	102	203	66.56	2.25E-50	У	Piceq	NA	0.999
BPLL_6 P	ACSL1	frameshift deletion	AUSL1:NM_UU1995:exon18:c.1666_1681del:p.556_561del.	chr4	1.86E+08	AIGIGUI	-	353	1	0.28	254	108	29.83	5.85E-35	У	MiSeq	NA	NA
BPLL_6 E	SLC6A19	missense	SLC6A19:NM_001003841:exon6:c.G815A:p.G272D.	chr5	1214108	I G	A	162	2	1.22	77	95	55.23	8.06E-33	У	MiSeq	NA	0.998
BPLL_6 F	PDE4D	missense	PDE4D:NM_001197218:exon1:c.G194T:p.R65M.	chr5	59064142	2 C	A	259	2	0.77	138	98	41.53	6.33E-35	У	MiSeq	0.06	NA
BPLL_6 F	RIN1	missense	RIN1:NM_004292:exon3:c.A292C:p.T98P.	chr11	66103323	T	G	148	8	5.13	144	31	17.71	2.60E-04	У	MiSeq	NA	0.025
BPLL_6 K	(LRF2	missense	KLRF2:NM_001190765:exon5:c.C462G:p.H154Q.	chr12	10046123	C (G	314	5	1.57	147	144	49.48	6.93E-50	У	MiSeq	NA	NA
BPLL_6 S	6LITRK5	missense	SLITRK5:NM_015567:exon2:c.A1101C:p.K367N.	chr13	88328744	A	С	301	7	2.27	174	178	50.57	4.98E-51	У	MiSeq	NA	0.968
BPLL_6 K	(IF23	missense	KIF23:NM_004856:exon12:c.A1259T:p.Y420F	chr15	69728097	Α Ι	Т	301	1	0.33	272	34	11.11	5.24E-10	У	MiSeq	0.1	0.488
BPLL_6 T	'P53	missense	TP53:NM_000546:exon8:c.G845C.R282P	chr17	7577093	C	G	135	2	1.46	78	84	51.85	7.41E-26	ý	MiSea	NA	1
BPLL 6 K	(BTAP9-9	missense	KBTAP9-9:NM_030975:exon1:c.C482A:p.S161Y.	chr17	39412115	C	A	342	0	0	177	22	11.06	1.29E-10	ú	MiSea	0.08	NA
BPLI 8	CACHD1	missense	CACHD1:NM_020925;exop14;c A1909G;p N637D	chr1	65129488	Ā	G	192	1 ñ	1 ñ	41	33	44.59	7.47E-22	ń		0.14	0.889
BPLL 8	POLIB2B	missense	POLB2B:NM_000938:exop7:c_T832C:p_E278L	chr4	57865879	T	Ē	213	1 ī	0.47	63	49	43.75	123E-25	 u	MiSea	NA	0.999
BPLL 8	AM160A1	missense	EAM160A1/NM_001109977.evon11.c G2296A/p A7667	chr4	153E±09	Ġ	Ā	166		0.41	56	28	33 33	3 15E-14	<u>×</u>	MiSeq	1	NΔ
		finisserise faanse als faarlasiaa	TERMINE 0012E1997	-1	1.332100			299	2	0.0	124	70	20.00	2 525-22	y	MiGee		NO
		mameshin deletion	THEIN, NP_001231301; exon 13:0; 1001deld; p. A33015; DCLAC1, NM_014739; except2; e_C20111; e_D071C	chio	1.246+00		-	200		0.00	249	70	20.01	1.022-33	<u>y</u>	MiGer-		0.992
DPLL_0		missense	DOLAF INFLUIGTSTEXON IZIC.UZUTTI: D. ROTIU.	chro	1.37E+00		<u> </u>	001	<u> </u>	0.15	243	01	23.30	1.03E-30	9	Mioeq		0.332
DPLL_0 F	1L	missense	PC:NM_000040716:exon10:c.C22356:p.D765E.	chrii	00010323			62	<u> </u>	0	20	61	42.00	2.10E-00	У	Piecim	0.1	0.001
BPLL_8 E	SCL /A	missense	BUL 7A:IVM_UZU333:exon1:c.G86A:p.HZ3H.	chr12	1.22E+08	6	A	121	1	0.82	15	44	74.58	5.87E-28	n		NA	0.985
BPLL_8 F	7	missense	F7:NM_000131:exon8:c.C752T:p.A251V.	chr13	1.14E+08	C	Ť	224	0	0	50	44	46.81	6.34E-28	n		NA	0.994
BPLL_8 F	PPM1A	missense	PPM1A:NM_021003:exon2:c.G625A:p.G209S.	chr14	60750046	G	A	172	1	0.58	34	39	53.42	6.95E-24	У	MiSeq	0.28	0.57173
BPLL_8 F	RNFT1	missense	RNFT1:NM_016125:exon2:c.C358T:p.H120Y.	chr17	58040344	G	A	169	2	1.17	44	46	51.11	1.87E-23	У	MiSeq	NA	0.03
BPLL_8)OK5	missense	DOK5:NM_018431:exon3:c.A183C:p.E61D.	chr20	53205030	A	С	325	2	0.61	64	54	45.76	1.11E-33	У	MiSeq	0.15	0.384
BPLL_8 E	3COR	missense	BCOR:NM_001123385:exon4:c.T2771G:p.F924C.	chrX	39931828	A	С	101	2	1.94	0	39	100	6.15E-33	У	MiSeq	NA	0.997
BPLL_10	4AGI1	missense	MAGI1:NM_001033057:exon23:c.G3874A:p.G1292R.	chr3	65342568	C	Т	227	2	0.87	68	75	52.45	1.33E-35	У	MiSeq	NA	0.024
BPLL_10	1RPS30	missense	MRPS30:NM_016640:exon5:c.G1234A:p.G412S.	chr5	44815218	G	A	117	0	0	52	76	59.38	5.92E-29	ý	MiSea+RNA-Sea	NA	0.74
BPLL 10 T	BAPPC13/C5Or	missense	C5orf44:NM_024941:exon3:c.G215T:p.G72V.	chr5	64931870	G	Т	151	4	2.58	94	98	51.04	9.36E-27	ú	BNA-Sea	0.07	NA
BPLL 10 H	ILA-DBB5	missense	HLA-DBB5:NM_002125;exop2;c T286A;p E96L	chr6	32489766	A	T	73	10	12.05	52	31	37.35	128E-04	ú	BNA-Seq	0.9	0
BPLL 10 F	SCLAF1	stopgain SNV	BCLAE1/NM_001077440;evop4;c_GB324;p_W211X	chr6	1.37E+08	<u>с</u>	Ť	660	2	0.3	328	122	27.11	5.36E-50	u	MiSea	NA	0.73327
BPLI 10	00L11 12	missense	POM121 12:NM 182595:even1:e G4888:e P163H	ohr7	53103852		Å	108	1 5	0.0	57	36	38.71	9.77E-15	- / ·	MiSeq	0.14	0.737
BDLL 10	4901	misserise splice site	NA	ohr8	16035500	T T		99	2	198	51	62	54.87	7.88E-20	<u>y</u>	MiSeq MiSeq	N0.14	NO
BDLL 10	DM17	spilce site	DDM17.NM_02290E	-1-10	645429			151		2.00	60	62	44.72	4 10E 10	<u>y</u>	MiCala DNA Cala	NIO NIO	0.000
DFLL_10F		missense	DADDC2.NM_0000701	- L-12	010420		<u>,</u>	570	<u> </u>	0.40	205		10.1	4. IUL-10	<u>y</u>	DNA C	NIA NIA	0.000
DPLL_10 F	ADPUS	missense		chria	25671172		A .	572	53	0.40	205	63	10.1	1.06E-05	У	RIVA-Seq		0.331
BPLL_10/2	FHX2	missense	2FHX2:NI//_U33400:exon2:c.G1301A:p.G4340.	chr 14	24003234			73	<u> </u>	1.25	50	42	45.65	3.33E-13	У	MiDeq		NA 0.100
BPLL_10	3362	missense	G5G2:NIP_031365:exon1:c.A1345G:p.1443A.	chr17	3628574	A	<u> </u>	204	1 5	2.39	3	108	97.3	8.59E-75	У	IVIIDeq	NA	0.108
BHLL_10	P53	missense	TP53:NM_000546:exon7:c.C743A:p.A248D	chr17	7577538		T	84		1.18	0	42	100	5.65E-33	У	MiSeq+RNA-Seq	NA	1
BPLL_10 F	ISAD1	missense	RSAD1:NM_018346:exon7:c.G1090A:p.V364M.	chr17	48561104	G	A	69	2	2.82	39	30	43.48	2.11E-09	У	MiSeq+RNA-Seq	0.05	0.208
BPLL_10	C19orf44	missense	C19orf44:NM_032207:exon2:c.C25T:p.R9C.	chr19	16611628) C	T	55	6	9.84	40	40	50	1.99E-07	У	RNA-Seq	NA	0.006
BPLL_10 Z	'FX	frameshift deletion	ZFX:NM_001178086:exon6:c.663_682del:p.221_228del	chrX	24228425	AGAAGA	-	485	3	0.61	139	93	40.09	2.72E-48	У	MiSeq+RNA-Seq	NA	NA
BPLL_13	1TOR	missense	MTOR:NM_004958:exon30:c.T4358C:p.L1453P.	chr1	11217320	A	G	357	2	0.56	169	149	46.86	2.02E-56	У	MiSeq	NA	0.992
BPLL_13	CDC28B	missense	CCDC28B:NM_024296:exon4:c.A503T:p.N168I.	chr1	32669958	A	Т	116	0	0	56	46	45.1	6.09E-19	ÿ	MiSeq+RNA-Seq	NA	0.996
BPLL_13 10	QCA1	missense	IQCA1:NM_024726:exon2:c.G190A:p.V64I.	chr2	2.37E+08	С	Т	215	2	0.92	79	76	49.03	1.44E-32	ý	MiSeq	0.07	NA
BPLL 13	DSEC1	stopgain SNV	IQSEC1:NM_001134382;exon3;c.C402A;p.Y134X.	chr3	12978114	G	T	323	1	0.31	326	167	33.87	2.18E-41	ú	MiSea+BNA-Sea	0.09	0.735421
BPLL 13	1YD88	missense	MYD88:NM_002468; exop3; c C656G; p S219C	chr3	38182032	Ē	Ġ	183	i i		173	121	34.75	1.69E-31	v v	MiSea+BNA-Sea	NA	0.955
BPLI 13	P63	missense	TP63/NM_001114980;evon8;c T1013C;p I338T	chr3	19F±09	T		234		043	176	95	35.06	8.03E-29	<u> </u>	MiSea	NA	0.725
BDL 12	012541	missense	COL 2541-NM 198721 even 20 o C1090T o B364W	ohrð	1 1E+09		<u>د</u>	87		0.40	55	16	22.54	9.74E-07	<u> </u>	MiSeq	NA	0.999
BDLL 19	ICUC2 INT	missense	DCUC23M (19/12/29)	chi4	1 5552.00		<u> </u>	249			1/1	10/	42.34	3.14E-07 1.9EE-29	2	MiGee	0.11	0.000
PDLL 12		missense	DUNU2.10/_01/033(eX0023(C/C f0130(p. V2007)). CUCV140.NM_001100000		1.550+00			240		0.20	221	222	42.43 E0.11	1.30E-30 2.40E-74	<u>y</u>	MiC	0.10	0.013
DPLL 13	SUCTIAS	missense	COULT MOTIVING OUT 100002(EXOND)C, 10000(D,F,02000	Chr4	1.576+08	+	6	304	+	0.20	221	222	30.11	3.40E-74	У	mibeq Mic	0.10	0.333
BPLL_13	PAIS1	missense	3PA (3):NIM_145026;exon5;c.G461A;p.G161B.	chrb	44329636		A	137		0.72	69	64	48.12	2.44E-23	У	Ploeq	NA	0.995
BPLL_13	JUL9A1	missense	CUL9A1:NM_UU1851:exon28:c.A1826G:p.Q609R	chr6	70961869	T		150	0	0	56	62	52.54	4.02E-28	У	MiSeq	0.32	0.463
BPLL_13	CSMD3	missense	CSMD3:NM_198123:exon18:c.T2955G:p.F985L	chr8	1.14E+08	A	C	195	1	0.51	93	85	47.75	2.02E-32	У	MiSeq	NA	0.999
BPLL_13 G	GLIS3	missense	GLIS3:NM_152629:exon3:c.C542A:p.A181D	chr9	411847	G	T	229	1	0.43	103	119	53.6	2.75E-45	У	MiSeq	NA	0.754

Patient	2	T	u		_			nbBases	nbBase	VAF	nbBases	nbBase	VAF	Somatic_p-	Confirme	Confirmation	Sift	Polyphe
	Gene	Mutation Type	Mutation	#Uhr	Position	BaseRe	BaseCalled	RetNorm	sAltNor	Norma	Reflumor	S	lumor	value	dlymj	method	score	n score
BPLL_13	PCSK5	missense	PCSK5:NM_001190482:exon30:c.G4008C:p.E1336D.	chr9	78936542	6		223	U	U U	157	61	27.98	1.54E-21	У	MiSeq	0.39	NA
BPLL_13	OR5R1	missense	0R5R1:NM_001004744:exon1:c.G59A:p.R20Q.	chr11	56185650	C	T	321	1	0.31	181	148	44.98	6.61E-52	У	MiSeq	0.23	0.081
BPLL_13	C14orf93	missense	C14orf93:NM_001130708:exon3:c.A724G:p.N242D	chr14	23465351	T	C	204	8	3.77	122	88	41.9	6.20E-23	У	MiSeq+RNA-Seq	0.09	0.168
BPLL_13		missense	CDKN3:NM_001130851:exon4:c.C190T:p.H64Y	chr14	54878318	C	T	229	3	1.29	105	108	50.7	9.28E-39	У	MiSeq	NA	1
BPLL_13	AK7	missense	AK7:NM_152327:exon16:c.C1856T:p.A619V.	chr14	96949438	С	T	196	2	1.01	126	100	44.25	1.42E-30	У	MiSeq	0.09	0.721
BPLL_13	CHD2	frameshift insertion	CHD2:NM_001271:exon28:c.3485_3486insG:p.L1162fs.	chr15	93536118	-	G	74	0	0	51	19	27.14	2.62E-07	У	MiSeq	NA	1
BPLL_13	NFAT5	missense	NFAT5:NM_173215:exon13:c.A3502G:p.N1168D	chr16	69727512	A	G	82	0	0	38	41	51.9	1.51E-16	У	MiSeq	NA	0.474
BPLL_13	CTAGE1	missense	CTAGE1:NM_172241:exon1:o.T2215C:p.F739L.	chr18	19995560	A	G	256	0	0	355	53	12.99	1.52E-12	У	MiSeq	NA	NA
BPLL_13	ITGB1BP3	missense	ITGB1BP3:NM_170678:exon8:c.C599T:p.P200L.	chr19	3942177	C	Т	182	2	1.09	103	89	46.35	3.31E-29	y	MiSeq	NA	0.988
BPLL_13	CYP4F12	missense	CYP4F12:NM_023944:exon7:c.C814T:p.R272W.	chr19	15794469	С	T	403	2	0.49	213	229	51.81	2.48E-78	ý	MiSeq	NA	0.771
BPLL 13	HAUS5	stopgain SNV	HAUS5:NM_015302:exon13:c.C1171T:p.Q391X.	chr19	36109943	С	Т	42	0	0	14	13	48.15	5.19E-07	ý	RNA-Sea	0.96	0.64092
BPLL 13	PAX1	missense	PAX1:NM_006192;exon2;c.C573G;p.I191M.	chr20	21687362	С	G	353	1	0.28	218	197	47.47	6.68E-64	ý	MiSea	0.33	0.926
BPLL 13	BCOR	frameshift insertion	BCOR:NM_001123385;exon14;c.4942_4943insA;p.P1648fs	chrX	39913173	-	T	70	2	2.78	2	67	97.1	3.31E-35	ú	MiSea+BNA-Sea	NA	NA
BPLI 18	PCNXL2	missense	PCNXI 2:NM_014801;exop2;c C284G;p P95B	obr1	2.33E+08	G	C C	117	1 n	0	158	129	44.95	8 10E-25	ú	MiSea+BNA-Sea	NA	NA
BPLL 18	TPO	missense	TPD:NM_001206744;evop12;c G20294;p V677I	chr2	1499783	G	Ă	143	2	138	72	65	47.45	134E-22	<u> </u>	MiSea	NA	0.562
BDII 18		ctongoin SNV	CYCR4/NM_003467;over2;o C1013C;o S338Y	ohr2	137E+08	G	<u> </u>	73	<u> </u>	0	77	82	5157	2 79E-18	2	MiSog+DNA-Sog	0.06	0.002
BDII 19		stopganiorv	COBL 11NM_014900;even5;e T718C;e E240V	ohr2	1665+08	0	L č	30			42	41	49.4	7.71E-08	<u> </u>	MiSeg+DNA-Seg	N0	0.10200
DFLL_10	TTUA	missense	TTUL 4.NM_014640.cvcc.9vc_C2107Avc_A702T	ohr2	2.25+09	- C		192	2	102	42	125	43.4	2.665-22	<u> </u>	MiSea+DNA-Sea	0.26	0.000
BDI 1 10		missense	CCDC99/NM_017785.cupp8/a_C9057		1895.09		Ť.	210	6	1.03	210	212	44.1	3.000-33	<u> </u>	MiSee	N/A	0.001
DPLL_10		missense	ULDU33:NM_UT7703:exono:C.U3U31:p. 1302M.	chr5	1.63E+00			210		0	210	212	43.3	3.04E-43	<u>y</u>		NA NA	0.344
BPLL_18	VAROZ	missense	VAR52:NM_UUTI67733:exon4:c.Go3A:p.R20H	chrb	3066364	6	<u> </u>	12	L U	<u> </u>	31	52	62.65	3.32E-20	<u>y</u>	IRNA-Seq	NA	
BPLL_18	FIBIN	missense	FIBIN:NM_203371:exon1:c.A583G:p.R195G.	chr11	27016656	A	6	117	0	U	59	46	43.81	1.38E-18	У	MiSeq	NA	0.996
BPLL_18	CLECLI	missense	ULEUL1:NM_1/2004:exon2:c.A440G:p.H147R.	chr12	9875286	نا ا	C	69	U	U	93	46	33.09	4.24E-10	У	MiSeq+RNA-Seq	0.1	0.263
BPLL_18	WBP4	frameshift deletion	WBP4:NM_007187:exon5:c.356delG:p.R119fs.	chr13	41642790	A	-	89	0	0	80	55	40.74	2.87E-15	У	MiSeq	NA	NA NA
BPLL_18	GPHN	missense	GPHN:NM_001024218:exon22:c.C2110T:p.R704C	chr14	67647553	С	T	215	0	0	127	83	39.52	1.31E-30	У	MiSeq+RNA-Seq	NA	0.986
BPLL_18	MUC16	missense	MUC16:NM_024690:exon3:c.G28442A:p.S9481N.	chr19	9059004	C	T	111	0	0	73	82	52.9	1.86E-25	У	MiSeq	NA	NA
BPLL_18	IGLL5	missense	IGLL5:NM_001178126:exon1:c.C133T:p.P45S.	chr22	23230366	C	T	49	0	0	13	16	55.17	3.95E-09	У	MiSeq+RNA-Seq	0.37	NA NA
BPLL_19	RAVER2	frameshift deletion	RAVER2:NM_018211:exon10:c.1735_1753del:p.579_585del.	chr1	65278514	TTATCAT	i -	244	0	0	101	35	25.74	9.96E-18	У	MiSeq	NA	NA
BPLL_19	NR113	missense	NR1I3:NM_001077482:exon4:c.G283T;p.A95S.	chr1	1.61E+08	С	A	475	8	1.66	63	62	49.6	2.13E-40	ÿ	MiSeq	0.11	0.011
BPLL 19	CXCR4	frameshift deletion	CXCR4:NM_003467;exon2;c.960_961del;p.320_321del,p.Ser325Ginf;	s chr2	1.37E+08	CA	-	93	0	0	43	12	21.82	3.01E-06	Ú.	MiSea	NA	NA
BPLL 19	LGI2	missense	LGI2:NM_018176:exon6:c.C613G:p.L205V.	chr4	25019653	G	С	385	0	Ō	190	113	37.29	3.35E-47	ú	MiSea	0.46	0.003
BPLI 19	LITP15	missense	UTP15:NM_032175:evon6:o_T553C:p_Y185H	ohr5	72866416	T	Č	98	ň	ň	65	33	33.67	4 51E-12	ú	MiSea	NA	0.999
BPLL 19	CUNT1	missense	CLINTI:NM_014666;evop1;c.C226;p.Riddrill	chr5	157E+08	Ġ	ř	102	1 ñ	1 n	61	43	4135	7.26E-16	<u> </u>	MiSea	NΔ	<u>Ν</u> Δ
BDII 19	HOVAS	missense	HEXA5:NM_019102:even1:e_C2020:p.160	ohr7	27183025		т	132			74	37	33.33	5 18E-15	<u> </u>	MiSeq	NO	0.474
DFLL_13	LINCED	missense	LINCED.NM 000072	-le-9	21103023			60	<u> </u>		20	16	44.44	1.105.00	<u> </u>	Micee		0.414
DPLL_IJ		missense	UNUGU:NM_0010E9;	chro	33313011	6	<u> </u>	45			20	10 E0	50.0E	1. IUE-00	<u>y</u>	Mice -	NA 1	0.30
DPLL_13	551K5	missense	DDI NM 000705 00 000007 M007U	chrib	1123310	6	<u> </u>	40	<u> </u>		53	50	52.25	5.20E-12	<u>y</u>	Imibed Imio	0.00	
BPLL_19	PPL	missense	PPL:NPL002705;exon22;c.U30201;p.A1007V.	chr lb	4935636	6	<u> </u>	1297		5.6	52	53	50.48	1.97E-32	У	Initiaed	0.23	0.001
BPLL_19	RAB37	missense	RAB37:NM_001163330:exon3:c.01781:p.H60Y	chr1/	72739310	U U		94	<u> </u>	U U	52	41	44.09	1.18E-15	У	MiSeq	NA	0.999
BPLL_19	CCDC137	missense	CCDC137:NM_199287:exon3:c.C381G:p.H127Q.	chr17	79637367	C	G	2789	63	2.21	39	35	47.3	1.89E-34	У	MiSeq	0.64	0.008
BPLL_19	HSBP1L1	missense	HSBP1L1:NM_001136180:exon3:c.T128A:p.M43K.	chr18	77728098	T	A	86		0	41	25	37.88	3.74E-11	У	MiSeq	NA	NA NA
BPLL_19	FTHL17	missense	FTHL17:NM_031894:exon1:c.G451A:p.G151S.	chrX	31089620	C	T T	325	5	1.52	128	111	46.44	2.08E-43	У	MiSeq	NA	0.108
BPLL_19	GPKOW	missense	GPKOW:NM_015698:exon7:c.C947A:p.T316N.	chrX	48972644	G	Т	1159	0	0	98	103	51.24	1.56E-98	У	MiSeq	NA	0.125
BPLL_22	ARHGEF11	frameshift insertion	ARHGEF11:NM_198236:exon30:c.2963_2964insGG:p.A988fs	chr1	1.57E+08	-	CC	58	0	0	35	29	45.31	1.44E-10	У	MiSeq	NA	NA
BPLL_22	TGFBRAP1	missense	TGFBRAP1:NM_004257:exon12:c.T2459C:p.l820T	chr2	1.06E+08	A	G	235	0	0	132	142	51.82	4.25E-49	У	MiSeq	0.08	0
BPLL_22	SF3B1	missense	SF3B1:NM_012433:exon15:c.A2098G:p.K700E.	chr2	1.98E+08	Т	С	141	0	0	69	82	54.3	1.04E-30	У	MiSeq	NA	0.999
BPLL_22	GLB1	missense	GLB1:NM_001135602:exon6:c.G538A:p.G180R	chr3	33093274	С	Т	143	0	0	81	63	43.75	2.14E-23	y y	MiSeq	NA	1
BPLL 22	MITE	missense	MITF:NM_198158;exon2;c.T71C;p.I24T	chr3	69987010	T	С	145	3	2.03	99	90	47.62	2.47E-24	ý	MiSeq	NA	0.996
BPLL 22	PCDHB4	missense	PCDHB4:NM_018938:exon1:c.C76T:p.R26C.	chr5	1.41E+08	Ċ	Ť	142	2	1.39	80	93	53.76	8.25E-29	ý	MiSea	0.15	0
BPLL 22	HMMB	splice site	NA	ohr5	1.63E+08	Ā	T	74		1 0	48	37	43.53	7.60E-13	ú	MiSea	NA	NA NA
3PLI 22	TCP11	missense	TCP11-NM_001093728-evop7-c_C895T-p_L299E	chr6	35088283	G	i à	176	t õ	t ñ	103	93	47.45	1.31E-32	L í	MiSea	0.12	0 137
3PH 22	PKHD1	missense	PKHD1/NM 170724 avon 39 o 46335G o V2112C	ohrE	51776752	T	- 7 - 1	55		t õ	35	29	45.31	3.27E-10	<u> </u>	MiSea	NA NA	0.101
2011 22	SVNE1	missense	CVNE1.NM_032071;evenE2;e C77994;e C2E979	ohrE	1535,09		T T	100			- 33	79	49.01	2.23E-21	<u> </u>	MiSee	0.09	0.000
DFLL_22		missense	ADCA19.NM_152701		1.550+00		<u> </u>	200		0.40	0.0	110	40.43	4.305-21	2	Mic	0.00	0.002
DPLL_22		missense			48311575		H	206		0.48	115	112	43.34	4.33E-38	<u> </u>	Impeq	NA NA	INA 0.100
SPLL_22		missense	LUBL:NP_UI5138:exon10:c.G2041A:p.A6811.	chr/	51096752			168		0.59	104	11/	52.94	7.05E-36	У		NA	0.108
SPLL_22	PCLO	missense	PCLU:NM_033026:exon3:c.C2326T:p.P7765	chr7	82764540	G	A	215		0	121	150	55.35	3.61E-50	У	MiSeq	NA	NA
BPLL_22	IMAC	missense	MYC:NM_002467:exon3:c.T1167G:p.F389L.	chr8	1.29E+08	T	G	242	1	0.41	151	126	45.49	6.50E-41	У	MiSeq	NA	0.76114
RDII 22	CACNA2D4	missense	CACNA2D4:NM 172364:exon1:c.A103C:p.I35L	chr12	2027537	1 T	G	106	0	0	54	43	44.33	3.02E-17	v	MiSea	0.14	NA NA

Patient	1		-		1		pbBases	pbBase	VAF	pbBases	pbBase	VAF	Somatic n	Confirme	Confirmation	Sife	Poluphe
	Mutation Tune	Mutation	#Chr	Position	BacoBof	BacoCalled	RefNorm	sAltNor	Norma	BefTumor	iibbase e	Tumor	uslue	d (who)	method	SCOTO	n score
BDL 22 CD4	missonso	CD4:NM_000616:even5:e_C5114:e_V171M	obr12	6924062	G	A	109		0	51	62	54.87	6.05E=24	a (yiii)	MiSea	0.28	0
BPLL 22 KIAA1551/C12orf	frameshift deletion	C12or(35:NM_018169;evop4;c 2959_2962del:p 987_988del	chr12	32136848	1 444C	-	173	1 ñ	1 ñ	120	84	41 18	172E-27	2	MiSeq	NA NA	NA
BPLI 22 SETD1B	frameshift insertion	SETD1B:NM_015048:even5:c_1635_1636ipsT:p_545fs	ohr12	122E+08	-	т	46	1 ñ	l ő	47	42	47.10	2 74E-10	2	MiSeq	NA	NA
BPLI 22 CBEBBP	frameshift insertion	CBEBBP:NM_004380;evop7;o_1640_1641ipsT;p_S547fs	chr16	3831241	-	Å	79	t õ	ň	36	45	55.56	9.67E-18	u u	MiSeq	NA	NA
BPLI 22 PBKCB2	missense	PBKCB:NM_212535;exon1:c_687T:n_K29N	chr16	23847583	G	T	78	2	25	36	21	36.84	9.21E-08	u v	MiSea	NA	0.998
BPLI 22 PBKCB1	missense	PBKCB:NM_212535;exop3;c A1054G;p S352G	chr16	24135291		Ġ	155	4	2.52	114	94	45.19	159E-23	ú	MiSea	NA	0.926
BPLL 22 SPATA20	missense	SPATA20:NM_022827;evop10:c_A1138G:p_1380V	chr17	48627936	A	Ğ	113	i i	0	65	42	39.25	4.06E-16	ú	MiSea	0.2	0.119
BPLI 22 CBLN2	missense	CBLN2:NM_182511;evop3;o C3311;p B111C	chr18	70209065	G	Ā	75	t õ	ň	48	44	47.83	8.09E-15	u u	MiSeq	NA	0.984
BPLI 24 PPP1B21	missense	PPP1B21/NM_152994;exop10:c_G935A;p_B312H	chr2	48698263	G	A	103	3	2.83	65	34	34.34	9.98E-10	u v	MiSea	NA	0.999
BPLL 24 ZDBF2	missense	ZDBF2:NM_020923;exon5:c.C2507T;p.P836L	chr2	2.07E+08	C C	T	185	2	1.07	118	85	41.87	2.26E-26	ń		NA	NA
BPLL 24 STAC	missense	STAC:NM_003149:exop2:o.G188A:p.B63Q	chr3	36484932	G	Å	196	Ō	0	93	76	44.97	3.06E-31	u	MiSea	NA	0.202
BPLI 24 MBPS27	missense	MBPS27:NM_015084;exon10:c_G853A;n_A285T	chr5	71519662	Ē	T	114	1	0.87	83	27	24.55	106E-08	ú	BNA-Seq	0.58	0
BPLL 24 WBNIP1	missense	WBNIP1:NM_020135:exon2:c.A959C:p.K320T	chr6	2769061	A	Ċ	53	Ó	0	21	29	58	1.99E-12	ú	MiSea+BNA-Sea	0.1	0.712
BPLL 24 OPBK1	missense	OPBK1:NM_000912:exon4:c.G853A:p.V285I	chr8	54142147	Ċ	T	223	3	1.33	123	92	42.79	2.07E-30	ú	MiSea	0.83	0
BPLL 24 KIAA0020	missense	KIAA0020:NM_014878:exon18:c.C1816T:p.L606F.	chr9	2804462	Ĝ	Å	145	Ō	0	93	73	43.98	8.19E-25	ú	MiSea+RNA-Sea	NA	0.96
BPLL 24 ANP32B	onframeshift deletic	ANP32B:NM_006401:exon6:c.653_655del:p.218_219del	chr9	1.01E+08	TGA	-	90	Ō	Ō	68	22	24.44	5.53E-08	ú	MiSea+BNA-Sea	NA	NA
BPLL 24 ZNF33A	missense	ZNF33A:NM_006954:exon5:c. T943A:p.C315S	chr10	38343995	Т	Α	196	Ö	Ō	138	80	36.7	1.06E-26	ú	MiSea+BNA-Sea	0.41	0.152
BPLL 24 C11orf74	missense	C11orf74:NM_138787:exon3:c.C262T:p.R88C.	chr11	36654959	Ċ	T	113	2	1.74	66	65	49.62	6.04E-20	ý	MiSea	NA	0.987
BPLL 24 SYT12	missense	SYT12:NM_001177880:exon3:c.G103A:p.A35T	chr11	66802184	G	A	110	1	0.9	40	43	51.81	1.02E-18	ú	MiSea	NA	0.157
BPLL 24 BIRC3	frameshift insertion	BIRC3:NM_182962;exon10:c.1643_1644insG:p.L548fs	chr11	1.02E+08	- 1	G	116	Ó	0	98	16	14.04	7.50E-06	ý	MiSea	NA	NA
BPLL 24 MLL2/KMT2D	frameshift deletion	MLL2:NM_003482:exon19:c.4884delT:p.G1628fs.	chr12	49438606	C	-	112	1	0.88	82	38	31.67	8.67E-12	ú	MiSea+RNA-Sea	NA	NA
BPLL_24 PPM1H	missense	PPM1H:NM_020700:exon5:c.G941C:p.R314P.	chr12	63131295	С	G	69	0	0	67	19	22.09	5.08E-06	ý	MiSeq	NA	NA
BPLL_24 AHNAK2	missense	AHNAK2:NM_138420:exon7:o.A5465G:p.K1822R.	chr14	1.05E+08	T	C	129	6	4.44	130	34	20.73	1.82E-05	ý	RNA-Seq	0.12	NA
BPLL_24 PMM2	missense	PMM2:NM_000303:exon6:c.A462T:p.R154S.	chr16	8905509	A	Т	91	0	0	64	32	33.33	2.61E-11	ý	MiSeq+RNA-Seq	NA	1
BPLL_24 NFATC3	missense	NFATC3:NM_173163:exon5:c.G1688T:p.R563I.	chr16	68200832	G	Т	248	0	0	261	58	18.18	2.65E-16	ÿ	MiSeq+RNA-Seq	NA	1
BPLL_24 TP53	missense	TP53:NM_000546:exon5:c.G394C:p.V132L	chr17	7578536	T	G	75	0	0	30	7	18.92	2.84E-04	ý	MiSeq	NA	0.991
BPLL_24 TP53	frameshift deletion	TP53:NM_000546:exon5:c.384delT:p.P128fs	chr17	7578546	G	-	73	0	0	6	30	83.33	3.20E-21	ÿ	MiSeq+RNA-Seq	NA	NA
BPLL_24 GRB7	missense	GRB7:NM_005310:exon5:c.G523A:p.G175R	chr17	37899492	G	A	239	1	0.42	281	67	19.25	9.66E-16	y	MiSeq	NA	0.849
BPLL_24 PITPNC1	missense	PITPNC1:NM_012417:exon9:c.G850A:p.A284T.	chr17	65688855	G	Α	283	2	0.7	307	172	35.91	2.81E-38	ÿ	MiSeq+RNA-Seq	0.06	NA
BPLL_24 ZNF416	missense	ZNF416:NM_017879:exon4:c.C889G:p.P297A.	chr19	58084383	G	С	226	0	0	142	104	42.28	5.52E-36	У	MiSeq	NA	0.939
BPLL_24 PTPRA	missense	PTPRA:NM_080840:exon13:c.A1040G:p.D347G	chr20	3001980	A	G	183	1	0.54	154	57	27.01	1.60E-16	У	MiSeq+RNA-Seq	NA	0.049
BPLL_24 RBL1	missense	RBL1:NM_002895:exon19:c.A2641T:p.S881C	chr20	35646763	T	A	208	0	0	170	26	13.27	2.73E-09	У	MiSeq	NA	0.99
BPLL_24 PHACTR3	missense	PHACTR3:NM_001199505:exon5:c.G547T:p.A183S	chr20	58342255	G	Т	112	1	0.88	103	30	22.56	2.96E-08	У	MiSeq	0.19	0.002
BPLL_24 SON1	frameshift deletion	SON:NM_032195:exon3:c.3742delG:p.V1248fs	chr21	34925279	I T	-	205	0	0	174	47	21.27	2.87E-15	У	MiSeq	NA	NA
BPLL_24 SON2	missense	SON:NM_032195:exon3:c.C3745G:p.P1249A	chr21	34925282	C (G	209	0	0	174	46	20.91	3.50E-15	У	MiSeq	NA	0.943
BPLL_24 TLR7	missense	TLR7:NM_016562:exon3:c.C917T:p.P306L.	chrX	12904544	C	Т	308	5	1.6	347	92	20.96	5.78E-18	У	MiSeq	NA	0.244
BPLL_24 MAGEB16	stopgain SNV	MAGEB16:NM_001099921:exon2:c.G679T:p.E227X.	chrX	35820992	G	T	155	1	0.64	130	45	25.71	4.14E-13	У	MiSeq	0.1	0.58003
BPLL_24 EFNB1	missense	EFNB1:NM_004429:exon3:c.T491C:p.V164A.	chrX	68059591	I T	С	93	0	0	97	40	29.2	6.50E-11	У	MiSeq	NA	0.997
BPLL_27 SYPL2	onframeshift deletic	SYPL2:NM_001040709:exon6:c.723_752del:p.241_251del.	chr1	1.1E+08	ICAGGAC	-	447	2	0.45	64	62	49.21	7.00E-45	У	MiSeq	NA	NA
BPLL_27 PPP1R1C	missense	PPP1R1C:NM_001080545:exon3:c.G151T:p.D51Y.	chr2	1.83E+08	G	T	46	0	0	43	27	38.57	9.65E-08	У	MiSeq	NA	NA
BPLL_27 GHRL	missense	GHRL:NM_001134941:exon4:c.G307A:p.D103N	chr3	10328412	C (T	274	0	0	117	75	39.06	3.62E-34	n		NA	0.847
BPLL_27 SETD2	missense	SETD2:NM_014159:exon6:c.T4730C:p.L1577P.	chr3	47147596	A	G	105	2	1.87	65	49	42.98	6.57E-15	У	MiSeq+RNA-Seq	NA	1
BPLL_27 RNF180	missense	RNF180:NM_001113561:exon3:c.A161G:p.Q54R	chr5	63507917	A	G	146	1	0.68	76	73	48.99	6.43E-26	n		NA	0.017
BPLL_27 BTN2A1	missense	BTN2A1:NM_078476:exon3:c.G247C:p.E83Q	chr6	26459873	G	C	189	1	0.53	104	84	44.68	9.75E-30	У	RNA-Seq	NA	0.988
BPLL_27 ACAT2	missense	ACAT2:NM_005891:exon4:c.G373T:p.A125S.	chr6	1.6E+08	G	Ť	258	3	1.15	204	100	32.89	4.31E-27	n		0.32	0.229
BPLL_27 LIMK1	missense	LIMK1:NM_002314:exon6:c.C617T:p.P206L	chr7	73520213	C	T	136	0	0	76	90	54.22	8.12E-31	У	MiSeq+RNA-Seq	0.09	0.02
BPLL_27 EZH2	missense	EZH2:NM_152998:exon19:c.A2102G:p.E701G	chr7	1.49E+08	T	C	175	1	0.57	72	54	42.86	2.84E-23	У	MiSeq+RNA-Seq	NA	0.999
BPLL_27 KBKB	missense	IKBKB:NM_UU1190720:exon6:c.A506T:p.K169M	chr8	42163895	A	T	145		0	96	39	28.89	1.59E-14	У	MiSeq+RNA-Seq	NA	0.973
BPLL_27 SCAI	missense	SUAI:NM_173690:exon15:c.C1321T:p.P441S	chr9	1.28E+08	G	A	209	3	1.42	106	117	52.47	1.09E-38	У	MiSeq+RNA-Seq	NA	0.999
BPLL_27 FAUS3	missense	FAU53:NM_U21727:exon5:c.G703A:p.V235M.		61646028			226	<u> </u>		100	95	48.72	1.36E-39	У	MiSeq+RNA-Seq	NA	0.977
BPLL_27 LM07	missense	LMU /:NM_U15842:exon11:c.G1855A:p.D619N	chr13	/6395659	G	<u>A</u>	337	7	2.03	140	109	43.78	9.01E-40	У	MiSeq	NA	0.98
BPLL_27 CHD8	missense	UHU8:NM_020920:exon38:c.A6517C;p.S2173R	chr14	21854164	T	G	261	4	1.51	104	85	44.97	2.05E-33	2	MiSeq+RNA-Seq	NA	
BPLL_27 BBM25	Irrameshift deletion	[RBM25:NM_U21233:exon11:c.1194_1195del;p.398_399del.	chr14	73572606		-	103	2	1.9	55	28	33.73	1.05E-09	<u>у</u>	RIVA-Seq	NA	I NA
BPLL_27 BYB3	missense	HYH3:NM_UU1243995:exon88:c.1124790;p.L4160P	chr15	34130675			143			89	76	46.06	6.04E-26	У	MiSeq	NA	NA NA
BPLL_27 CSK	missense	USK:NM_UU4383:exon11:o.G953A:p.R318H	chr15	75094101	G	A	140		0	101	67	39.88	1.16E-21	У	MiSeq+RNA-Seq	NA	
BPLL_27 SREBF1	I trameshift deletion	SREBF1:NM_001005291:exon3:c.283delG:p.095fs	chr17	17723734	G	-	202	3	1.46	64	73	53.28	5.15E-32	У	MiSeq	NA	I NA

Patient				1	1	-		nhBases	nhBas	VAF	nhBases	nhBase	VAF	Somatic n-	Confirme	Confirmation	Sift	Polynhe
In	Gene	Mutation Type	Mutation	#Chr	Position	BaseBef	BaseCalled	BefNorm	esAltN	Norm	BefTumo	s	Tumor	value	d (who)	method	score	n score
BPLI 27	МАРКА	missense	MAPK4/NM_002747;evop2;c G1224;p B410		48190450	G	A	491	3	0.61	163	142	46.56	3 56E-66		MiSea	0.61	0.06
BPLL 27		missense	IZUMO4/NM_001039846;evop9:c G5934:p B198K	chr19	2099013	G	<u> </u>	93	1 ñ	0.01	66	46	41.07	3 93E-15	2	MiSeg+BNA-Seg	0.01	0.00
BPLL 27		missense	ICAM1:NM_000201:evon6:o_C1361T:p_T454I	chr19	10395639	t č	T	98	1 õ	1 ñ	43	38	46.91	1.63E-16	2	MiSeq+BNA-Seq	NA NA	0.00
BPLL 27	EMB2	missense	EMB2:NM_152919:evop13:o_G13094:p_V437M		14866540	ř	Ť	306	1 õ	1 ñ	133	91	40.62	170E-40	2	nioeq ninin oeq	MA	0.305
BPLI 30	CSMD2	missense	CSMD2-NM_052896;even65:e_G98087;e_G3270W	chr1	33990638	ř	Å	165	2	12	59	50	45.87	7 79E-22		MiSea	MA	0.648
BDLL 30	00M02	missense	ADAM30.NM_021794;auap1;a_C13964;a_L466L	ohrl	1.2E+08	č	<u> </u>	100	1	0.77	63	90	52.27	7.00E-25	<u> </u>	MiSeq	NO	0.040
BDLL 30	NEASC	missense	NEASC-NM_001005388-auge18:a C1930A-p.2400.	ohri	2 05E+08	- C	ò	125	1	0.11	64	58	47.54	1.00E-20	<u> </u>	MiSeq	NO	0.000
BDLL 30	MSHR	missense	MSH8:NM_000179-avaptic_C71Trp_S24L	ohr?	48010443	r c		302	2	0.14	1/19	130	46.59	4.30E-22	<u> </u>	MiSeg MiSeg_DNA_Seg	0.22	0.021
PDLL_30		missense	DES.NM_001927	orii2	2.25,09	L č		142		4.7	F9	64	F2 02	E E E E 20	<u>y</u>	Mideq+HMM-deq	0.22	0.000
BPLL_30	DEO RETIDO	missense Generalisis in eestine	EETD2.NM_0144E9	chr2	47102490		cT	192	1	9.1	0/	45	24.00	1.105 14	<u> </u>	Mices Into Con	NA	0.43725
BPLL_30	JETUZ TETO	frameshirtinsertion	TET2.NM_001127200	chr3	1.065+00	TCCCCTC		120 EC	+	0.13	64	40	24.00	2 525 00	<u> </u>	MiSe	NA	
BPLL_30	ICIZ	trameshint deletion	IL CCT,NM_001121200.exon3:0.4173_4102det.p. 1333_1334det.	chr4	EE227402			22			15	10	54.02	2.33E-00	<u> </u>	MiGent DNA Gen	0.2	
BPLL_30	LOGT	missense	M/DT.NM_000130301;exon 10:0.12302(3;p.3700A	chro	1275,09		2	72		2.67	50	40	34.33	4.00E-00	<u>y</u>	MiSeq+NNA-Seq	0.5	0.300
DPLL_30	PIT U1	missense	PTT UT:NM_U00 F30:eX0h3:C.A1204C;D.E4200	chro	1.37E+00	<u> </u>	<u> </u>	13	4	2.07	100	40	44.44	3.40E-11	9	Miced	NA NA	0.405
DPLL_30	F200	missense	F2D0:N#_00104013;eX004;C.ATF3C;p.L333F	chro	1.046+00	<u> </u>		201	4	1.35	132	71	44.3	2.43E-23	9	Mic	1NA 0.00	0.000
DPLL_30		missense	CULZ (A I:NIM_U32000: exon IU:C.UZ23 (I:p.P (40L.	chr3	1.17E+00			140		0.00	04		45.01	4.30E-24	у	I'lloeg Mrc	0.03	0.004
BPLL_30	ADLIMI	missense	ADLIMI:NM_UUZ313:exon4:c.Ub3U1:p.PZ17L	chriu	1. IDE+00	5	<u> </u>	100		0	37	31	24.22	0.07E-10	У	I'lloeg Mrc	NA NA	0.004
BPLL_30	URSTET	missense	UR51E1:NM_152430:exon2:c.1186:p.N6K.	chr11	4673774		6	37	1	2.63	18	18	50	1.65E-06	У	MiSeq	NA	0.264
BPLL_30	SURLI	missense	SURL1:NM003105:exon25:c.U35141:p.R1172U.	chr11	1.21E+08			214	2	0.93	120	79	33.7	3.36E-27	У		0.11	0.999
BPLL_30	IMPU	missense	IMPU:NM_003276;exon4;c.010251;p.P342L.	chr12	98927060			93	0	0	54	64	54.24	1.57E-21	у	MiSeq+RNA-Seq	0.08	
BPLL_30	BNIP2	missense	BNIP2:NM_UU433U:exon6:c.A886G:p.M296V.	chr15	59964888		<u> </u>	100	3	2.91	68	29	29.9	7.00E-08	у	MiSeq+RNA-Seq	NA	0.904
BPLL_30	CHD2	frameshift deletion	CHD2:NM_001271:exon33:c.4160_4178del:p.1387_1393del.	chr15	93545429	AAAAAAA	-	84	1	1.18	45	40	47.06	5.70E-14	у	MiSeq+RNA-Seq		<u> </u>
BPLL_30	TP53	missense	TP53:NM_000546:exon8:c.T824A:p.L275Q	chr17	7577114	C	Ť	92	1	1.08	2	46	95.83	1.55E-33	у	MiSeq+RNA-Seq	NA	
BPLL_30	JUP	missense	JUP:NM_021991:exon11:c.C1853T:p.A618V	chr17	39913957	G	A	109	0	0	42	53	55.79	4.78E-23	У	MiSeq+RNA-Seq	NA	0.838
BPLL_30	ZNF134	missense	ZNF134:NM_003435:exon3:c.A803G:p.Y268C.	chr19	58132290	A	G	105	1	0.94	53	59	52.68	1.16E-20	У	MiSeq+RNA-Seq	0.18	0.987
BPLL_32	TP73	missense	TP73:NM_001204190:exon3:c.G466A:p.E156K	chr1	3638768	G	A	91	0	0	27	23	46	6.89E-13	У	MiSeq	NA	0.096
BPLL_32	NOTCH2	stopgain SNV	NOTCH2:NM_024408:exon34:c.A6397T:p.K2133X.	chr1	1.2E+08	Т	A	193	0	0	68	82	54.67	1.13E-37	У	MiSeq+RNA-Seq	NA	0.73513
BPLL_32	MYD88	missense	MYD88:NM_002468:exon4:c.T695C:p.M232T	chr3	38182259	T	С	147	0	0	68	141	67.46	3.91E-47	У	MiSeq+RNA-Seq	NA	1
BPLL_32	DROSHA	missense	DROSHA:NM_013235:exon25:c.T3140C:p.L1047S	chr5	31431688	A	G	222	0	0	126	121	48.99	1.32E-42	У	MiSeq+RNA-Seq	NA	NA
BPLL_32	TTC1	onframeshift deletio	TTC1:NM_003314:exon8:c.864_866del:p.288_289del.	chr5	1.59E+08	AAT	-	182	1	0.55	96	50	34.25	2.80E-19	У	MiSeq+RNA-Seq	NA	NA
BPLL_32	DNAH11	missense	DNAH11:V973A	chr7	21639655	T	С	250	0	0	109	116	51.56	1.28E-47	У	MiSeq	NA	NA
BPLL_32	TNFRSF10C	onframeshift deletio	TNFRSF10C:NM_003841:exon5:c.641_685del:p.214_229del.	chr8	22974405	GCCCCAG	-	31	0	0	2	37	94.87	7.39E-18	n		NA	NA
BPLL_32	PLEC	missense	PLEC:NM_201383:exon14:c.C1670T:p.A557V	chr8	1.45E+08	G	A	243	1	0.41	98	69	41.32	1.51E-30	У	RNA-Seq	NA	0.66782
BPLL_32	PARD3	missense	PARD3:NM_001184787:exon8:c.G998A:p.R333Q	chr10	34673075	С	T	111	0	0	55	52	48.6	1.83E-20	У	MiSeq	NA	0.999
BPLL_32	ARAP1	missense	ARAP1:NM_001040118:exon14:c.G1992C:p	chr11	72415197	С	G	158	0	0	45	45	50	1.55E-24	У	MiSeq	0.12	0.05
BPLL_32	PDZD3	missense	PDZD3:NM_001168468:exon3:c.G100A:p.E34K	chr11	1.19E+08	G	A	219	2	0.9	102	91	47.15	4.56E-34	У	MiSeq	NA	0.629
BPLL_32	PUS7L	missense	PUS7L:NM_001098615:exon7:c.C1489T:p.R497C	chr12	44130420	G	A	106	2	1.85	119	48	28.74	4.98E-10	У	RNA-Seq	NA	1
BPLL_32	TENC1	missense	TENC1:NM_170754:exon18:c.C1924T:p.R642C	chr12	53453349	С	T	227	0	0	160	80	33.33	3.25E-27	У	MiSeq	NA	0.999
BPLL_32	NCKAP1L	missense	NCKAP1L:NM_001184976:exon6:c.C422T:p.T141	chr12	54903518	С	T	109	1	0.91	106	50	32.05	1.35E-12	У	MiSeq+RNA-Seq	0.41	0.28
BPLL_32	N4BP2L2	missense	N4BP2L2:NM_033111: exon 7: c. A1449T: p. K483N.	chr13	33017225	T	A	117	0	0	38	44	53.66	1.10E-21	У	RNA-Seq	NA	NA
BPLL_32	ATP10A	missense	ATP10A:NM_024490:exon10:c.G2164A:p.V722M.	chr15	25959001	С	Т	314	0	0	128	117	47.76	1.54E-51	У	MiSeq	NA	0.971
BPLL_32	UBR1	missense	UBR1:NM_174916:exon22:c.T2408G:p.V803G.	chr15	43319998	A	С	118	0	0	55	51	48.11	5.96E-21	y	MiSeq+RNA-Seq	NA	0.992
BPLL_32	BRCA1	missense	BRCA1:NM_007294:exon10:c.C2317T:p.P773S	chr17	41245231	G	A	339	9	2.59	150	137	47.74	8.99E-46	y	MiSeq	0.06	0.296
BPLL_32	KIF2B	missense	KIF2B:NM_032559:exon1:c.C547A:p.P183T.	chr17	51900941	С	A	404	3	0.74	184	127	40.84	9.46E-50	ý	MiSeq	NA	0.099
BPLL_32	ACTG1	missense	ACTG1:NM_001199954:exon2:c.C56A:p.A19D	chr17	79479325	G	Т	277	0	0	111	112	50.22	5.24E-49	v	MiSea+RNA-Sea	NA	0.443
BPLL_32	ANKRD12	missense	ANKRD12:NM_015208:exon9:c.T3054A:p.D1018E	chr18	9256319	Т	A	58	0	0	71	24	25.26	3.01E-06	ý	RNA-Seq	NA	0.99
BPLL 32	MATK	missense	MATK:NM_139354;exon7;c.C571G;p.L191V	chr19	3781653	G	С	166	2	1.19	67	49	42.24	3.07E-20	v	MiSea	0.46	0.001
BPLL 32	GPR112	missense	GPR112:NM_153834;exon21;c.C8444A;p.A2815E.	chrX	1.35E+08	Ē	Ā	43	Ō	0	0	44	100	7.62E-26	ý	MiSea	NA	0.754
BPLL 33	DPYD	stopgain SNV	DPYD:NM_000110:exon11:c.A1252T:p.K418X.	chr1	98039403	T	A	529	1	0.19	99	94	48.7	5.72E-62	ý	MiSea	0.56	0.66176
BPLL 33	LBIF1	missense	LBF1:NM_018372;exon4:c.A1957G;p.I653V	chr1	1.11E+08	Ť	Ċ	212	i i	0	51	41	44.57	2.16E-25	ú	MiSea+BNA-Sea	1	0
BPLL 33	SMG5	missense	SMG5:NM_015327;exon18:o.G2636A;p.B879H	chr1	1.56E+08	Ċ	Ť	177	Ť	Ť	63	34	35.05	5.37E-18	u v	MiSea+BNA-Sea	NA	0.976
BPLI 33	L 18L 1	missense	II 1BL1:NM_016232:exon11:o_G1328A:p_B443Q	chr2	103E+08	Ğ	Å	228	3	13	22	26	54.17	2.68E-20	, v	MiSea	NA	
BPLI 33	SE3B1	missense	SE3B1:NM_012433;evon15;c A2098G;n K700E	chr2	1.98E+08	Ť	i ii	158	t ĭ	0.63	25	30	54.55	2.28E-20	, v	MiSeg+BNA-Seg	NA	0.999
BPLI 33	AFF4	missense	AFE4:NM_014423:evon3:o_C4577:p_B153C	ohr5	132E+08	Ġ	Ă	205	t i	0.00	22	21	48.84	1.22E-17	- <u>y</u>	MiSeg+BNA-Seg	NA	1
BPLI 33	MUC17	missense	MUC17-NM_001040105;evop3;o C5138T;p T1713	ohr7	1.022+00	t č	T	432	t ó	0.45	61	42	40.78	2.61E-34	<u>y</u>	MiSea	NA	0 25442
BPLI 33	MET	missense	MET-NM_000245;even2:e_G911T;n_B304	ohr7	1.16E+08	- ă	Η τ΄	433		0.23	80	71	47.02	2.01E-04	×	MiSea	NA	0.999
BPLI 33	MYC	missense	MYC:NM_002467;evon2:o.C482T;p.S161	chr8	129E+08	t č	t †	529	1 2	0.20	44	44	50	4.33E-40	<u> </u>	MiSeg+BNA-Sec	NA	0.983
		IIIISSEIISE	1110.18 COOL 101. COOL 10. COOL 1. D. O 10 IE.	1 0000	1 1.200700		1 1	1 020	1 6	1 0.00	1 77	1 77	1 00	1 7.000 70		proceeding and only	1 1307	, 0.000

Patient								nbBases	nbBas	VAF	nbBases	nbBase	VAF	Somatic_p-	Confirme	Confirmation	Sift	Polyphe
D	Gene	Mutation Type	Mutation	#Chr	Position	BaseRef	BaseCalled	RefNorm	esAltN	Norm	RefTumo	s	Tumor	value	d (y/n)	method	score	n score
BPLL_33	COL5A1	missense	COL5A1:NM_000093:exon65:c.C5293T:p.R1765C.	chr9	1.38E+08	С	T	260	6	2.26	63	49	43.75	1.07E-23	у	MiSeq	NA	0.77514
BPLL_33	DDB1	missense	DDB1:NM_001923:exon21:c.C2660T:p.T887M.	chr11	61076456	G	A	79	0	0	16	16	50	7.37E-11	ÿ	RNA-Seq	NA	0.031
BPLL_33	FAT3	missense	FAT3:NM_001008781:exon6:c.G4277A:p.R1426K.	chr11	92507288	G	A	325	1	0.31	86	62	41.89	4.34E-35	у	MiSeq	1	NA
BPLL_33	NCOR1	stopgain SNV	NCOR1:NM_001190440:exon36:c.C5459G:p.S1820X	chr17	15965185	G	С	132	1	0.75	29	28	49.12	1.40E-16	У	MiSeq+RNA-Seq	0.08	0.73546
BPLL_33	SMG8	missense	SMG8:NM_018149:exon3:c.T2665A:p.L889I.	chr17	57290849	T	A	202	1	0.49	38	31	44.93	1.77E-20	у	MiSeq+RNA-Seq	NA	0.963
BPLL_33	SLC39A6	missense	SLC39A6:NM_012319:exon2:c.G329A:p.R110H.	chr18	33706642	С	Т	313	1	0.32	75	19	20.21	2.74E-12	У	RNA-Seq	0.2	0.39595
BPLL_33	BCOR	missense	BCOR:NM_001123385:exon10:c.A4376G:p.N1459S.	chrX	39921444	Т	C	147	1	0.68	28	24	46.15	1.41E-15	у	MiSeq+RNA-Seq	NA	0.92
BPLL_34	UBR3	missense	UBR3:NM_172070:exon23:c.A3468G:p.I1156M.	chr2	1.71E+08	A	G	463	12	2.53	209	200	48.9	1.20E-65	у	MiSeq+RNA-Seq	NA	0.82
BPLL_34	PGM2	missense	PGM2:NM_018290:exon6:c.T554G:p.1185S.	chr4	37841716	T	G	245	3	1.21	226	29	11.37	9.38E-07	у	RNA-Seq	NA	0.336
BPLL_34	ZSWIM6	missense	ZSWIM6:NM_020928:exon8:c.C1915G:p.L639V.	chr5	60825956	С	G	97	6	5.83	44	46	51.11	3.44E-13	у	RNA-Seq	0.11	NA
BPLL_34	TINAG	missense	TINAG:NM_014464:exon7:c.G1024A:p.V342I.	chr6	54214638	G	A	130	3	2.26	78	92	54.12	5.95E-26	у	MiSeq	0.4	0
BPLL_34	FAM120B	missense	FAM120B:NM_032448:exon2:c.T1213A:p.S405T.	chr6	1.71E+08	T	A	282	1	0.35	324	38	10.5	2.23E-09	У	RNA-Seq	NA	0
BPLL_34	OCM	missense	OCM:NM_001097622:exon2:c.T89C:p.F30S.	chr7	5922151	T	C	252	4	1.56	126	126	50	5.34E-42	У	MiSeq	NA	1
BPLL_34	PGAM2	ionframeshift deletio	PGAM2:NM_000290:exon1:c.314_316del:p.105_106del.	chr7	44104813	GGC	-	133	6	4.32	75	69	47.92	1.96E-18	у	RNA-Seq	NA	NA
BPLL_34	MLLT3	ionframeshift deletio	MLLT3:NM_004529:exon5:c.502_504del:p.168_168del.	chr9	20414340	CTA	-	105	0	0	60	42	41.18	5.41E-16	n		NA	NA
BPLL_34	DDIT4	missense	DDIT4:NM_019058:exon3:c.C271T:p.H91Y.	chr10	74034518	С	Т	226	9	3.83	130	137	51.31	5.22E-36	у	MiSeq+RNA-Seq	0.29	0.653
BPLL_34	DOCK1	missense	DDCK1:NM_001380:exon10:c.G910A:p.V304M.	chr10	1.29E+08	G	A	162	0	0	71	71	50	1.20E-29	у	MiSeq	NA	NA
BPLL_34	KRT3	missense	KRT3:NM_057088:exon1:c.G251A:p.R84Q.	chr12	53189576	С	T	141	8	5.37	62	77	55.4	2.79E-22	у	MiSeq	NA	NA
BPLL_34	PIWIL1	missense	PIWIL1:NM_004764:exon16:c.C1921T:p.R641W	chr12	1.31E+08	С	T	148	3	1.99	77	51	39.84	4.82E-17	у	MiSeq	NA	0.985
BPLL_34	TMEM63C	missense	TMEM63C:NM_020431:exon8:c.G515A:p.R172Q.	chr14	77699816	G	A	63	4	5.97	41	33	44.59	7.04E-08	у	RNA-Seq	NA	NA
BPLL_34	CILP	missense	CILP:NM_003613:exon9:c.C2729G:p.A910G.	chr15	65489895	G	С	227	6	2.58	123	140	53.23	6.60E-41	у	MiSeq	0.48	0
BPLL_34	TP53	missense	TP53:NM_000546:exon7:c.T707G:p.V236G	chr17	7577574	T	C	224	4	1.75	0	99	100	7.66E-80	у	MiSeq+RNA-Seq	NA	0.999
BPLL_34	PYCR1	missense	PYCR1:NM_153824:exon4:o.G508A:p.V170I	chr17	79892834	С	T	55	0	0	39	51	56.67	9.84E-15	у	MiSeq+RNA-Seq	0.1	0.014
BPLL_34	LAMA1	missense	LAMA1:NM_005559:exon50:c.G7079A:p.R2360H.	chr18	6965403	С	T	492	11	2.19	256	203	44.23	3.78E-63	у	MiSeq	NA	0
BPLL_34	ADNP2	missense	ADNP2:NM_014913:exon4:o.C3145T:p.R1049C.	chr18	77896441	С	T	105	2	1.87	61	27	30.68	6.09E-09	у	MiSeq	NA	0.994
BPLL_34	SPTBN4	missense	SPTBN4:NM_020971:exon14:c.C1852A:p.R618S.	chr19	41018548	C	A	52	0	0	19	16	45.71	3.43E-08	у	MiSeq	NA	1
BPLL_34	LTN1	missense	LTN1:NM_015565:exon23:c.A4269T:p.E1423D.	chr21	30316078	T	A	279	7	2.45	127	110	46.41	8.10E-37	у	MiSeq+RNA-Seq	0.6	0.008

Patient ID	ChrA	Gene A	Band	ChrB	Gene B	Band	Number of spanning reads	Strand orientation (forward:f / reverse:r)	Confirmation by RT-PCR	Corresponding abnormalities by karyotype
BPLL_32	chr13:114514709	FAM70B exon 8	13q34	chr13:114536552	GAS6 intron 2	13q34	161	rr	ND	
BPLL_34	chr10:104161003	NFKB2 exon 19	10q24.32	chr10:105232917	CALHM3 3'UTR exon 3	10q24.33	676	fr	ND	
BPLL_27	chr14:106213663	IGHG1	14q32.33	chr8:128750494	MYC intron 1	8q24.21	20	rf	ND	t(8;14)(q24;q32)
BPLL_30	chr8: 127794677	PVT1 intron 1	8q24.21	chr22:22792944	IGLV4-69*01	22q11.22	11	ff	yes	t(8;22)(q24;q11)
BPLL_4	chr8:128078031	PVT1 intron 5	8q24.21	chr22:22888886	IGLL5 intron 1	22q11.22	38	ff	yes	t(8;22)(q24;q11)
BPLL_30	chr10:93558648	TNKS2 intron 1	10q23.32	chr14:78177178	SLIRP intron 1	14q24.3	18	ff	ND	
BPLL_5	chr3:75832457	ZNF717 exon 2	3p12.3	chr18:56686740	no gene	18q21.32	16	rf	ND	der(18), add(3)(p22)

 Table S7. Fusions detected by RNA-Seq. The coordinates refer to the hg19 reference genome.

ND: not done

Table S8. Quality control data for WES.

					% Targeted exome	% Targeted exome
					covered by at	covered by at least
Patient ID	Fraction	Total reads	%Unmapped	Mean depth	least 1 read	10 reads
BPLL 1	CD19+/CD5+	1.48E+08	0.12137	106 x	91.62	86.179
BPLL_1	CD3+	1.82E+08	0.032739	146 x	91.671	88.545
BPLL_4	CD19+	1.45E+08	0.041366	95 x	87.118	80.568
BPLL_4	CD3+	2.01E+08	0.069637	109 x	89.508	84.935
BPLL_5	CD19+	1.54E+08	0.5213	94 x	89.387	81.924
BPLL_5	CD3+	2.23E+08	0.50716	119 x	91.843	87.29
BPLL_6	CD19+	1.65E+08	0.043966	129 x	91.641	87.596
BPLL_6	CD3+	1.80E+08	0.067951	119 x	91.672	87.647
BPLL_8	CD19+	1.02E+08	0.022356	54 x	91.552	80.11
BPLL_8	CD3+	1.59E+08	0.02019	105 x	91.855	87.253
BPLL_10	CD19+/CD5+	2.86E+08	0.031002	107 x	91.578	89.399
BPLL_10	CD3+	1.59E+08	0.029828	121 x	91.54	88.282
BPLL_13	CD19+	1.53E+08	0.043803	122 x	91.514	86.481
BPLL_13	CD3+	1.79E+08	0.037851	130 x	91.692	88.968
BPLL_18	CD19+/IGK+	1.84E+08	0.022249	98 x	91.829	87.161
BPLL_18	CD3+	9.57E+07	0.021904	69 x	91.728	83.383
BPLL_19	CD19+	1.22E+08	0.021803	86 x	91.71	85.768
BPLL_19	CD3+	1.95E+08	0.042149	131 x	91.705	86.44
BPLL_22	CD19+	1.74E+08	0.2666	116 x	88.184	80.881
BPLL_22	CD3+	1.58E+08	0.46315	92 x	88.921	81.941
BPLL_24	CD19+/IGK+	1.76E+08	0.042913	117 x	86.75	79.931
BPLL_24	CD3+	1.71E+08	0.060721	103 x	87.321	81.548
BPLL_27	CD19+	1.61E+08	0.040653	127 x	91.645	88.275
BPLL_27	CD3+	1.88E+08	0.041159	143 x	91.574	86.851
BPLL_30	CD19+/CD5+	1.12E+08	0.019185	89 x	91.463	85.493
BPLL_30	CD3+	1.06E+08	0.021597	88 x	91.396	84.335
BPLL_32	CD19+	1.30E+08	0.04556	97 x	91.578	86.2
BPLL_32	CD3+	1.43E+08	0.038769	108 x	91.615	87.074
BPLL_33	CD19+/CD5+	1.12E+08	0.0185	51 x	91.319	78.561
BPLL_33	CD3+	1.87E+08	0.020833	129 x	91.837	89.375
BPLL_34	CD19+/IGK+	1.35E+08	0.016767	94 x	91.781	85.962
BPLL_34	CD3+	1.58E+08	0.01649	99 x	91.833	86.756

Supplementary Figures



Figure S1. Somatic variants in coding regions identified by WES in 16 patients with B-PLL. A. Numbers and types of somatic mutations identified in each patient. The median number of somatic mutation was 20 per patient (range: 12-30). B. Distribution of the 309 somatic mutations identified in the 16 patients with B-PLL. The most frequent alterations were somatic missense mutations (n=266, 86%) and insertions/deletions (n=29, 10%). C. Distribution of base changes, with transitions in purple and transversions in blue.



Figure S2. Genes with recurrent somatic mutations, in patients with B-PLL.

% of aberrations



Figure S3. Clonal and subclonal aberrations. Histograms represent the percentage of the aberrations classified as clonal (black) or subclonal (grey). The n corresponds to the number of patients with the genomic abnormality. For example, the translocation t(MYC) was observed in 19 cases with available data (see the Supplementary Methods above). The aberration was clonal in 17 of the 19 cases (89.5%).



Figure S4. Overall survival in patients with B-PLL, according to the presence (median [95%CI]: 66.5 [11.1-66.5] or absence (median [95%CI]: 125.7 [57.5-132.1] of del17p.



Figure S5. (A) Mean of cell viability assessed by ATP-based CellTiter-Glo 2.0 kit in B-PLL cells from 3 patients (B_PLL8, BPLL_13 and BPLL_18) exposed to increasing doses of OTX015. (B) Cell death was quantified in primary B-PLL cells from patient BPLL_8 (with t(*MYC*)) with or without pretreatment with JQ1 (500 nM) and and exposure for 48h to ibrutinib (7.5 μ M), idelalisib (50 μ M), or venetoclax (10 nM). The percentages refer to annexin-V-positive or annexin-V-/PI-positive cells. (C) Primary B-PLL cells from patient BPLL_34 (with del17p but no *MYC* activation) were treated and analyzed as in (B). Bars represent the mean ± SEM.

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