Three novel fusion transcripts of the *paired box 5* gene in B-cell precursor acute lymphoblastic leukemia

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Supplementary Information, including Supplementary Methods, References, Figures and Tables.

Supplementary Patients and Methods

Patient cohort. Among pediatric patients enrolled in the AIEOP-BFM ALL2000 protocol (for whom conventional or molecular cytogenetic analyses were available), 19 BCP-ALL presented visible abnormalities on the 9p chromosomal region, and were, therefore, considered for this study. In addition, an adult BCP-ALL patient was analyzed, as having a 9p abnormality in addition to the t(9;22) translocation. The main patients' features are reported in Supplementary Table S1.

FISH Analysis. It was performed on bone marrow metaphases from archival methanol:acetic acid-fixed chromosome suspensions, as previously described.[1] BAC and fosmid clones were derived from the University of California Santa Cruz (UCSC) database (release of February 2009, GRCh37/hg19) and previously tested on normal human metaphases (Supplementary Tables S2-S3). Briefly, chromosome preparations from bone marrow cells were hybridized in situ with 1µg of each Bacterial Artificial Chromosome (BAC)/fosmid probe labelled by nick translation. Hybridization was performed at 37°C in 2X SSC, 50% (vol/vol) formamide, 10% (wt/vol) dextran sulfate, 5µg COT1 DNA (Bethesda Research Laboratories, Gaithersburg, MD, USA), and 3µg sonicated salmon sperm DNA in a volume of 10 µL. Post-hybridization washings were performed at 60°C in 0.1X SSC (three times). In co-hybridization experiments, the probes were directly labeled with Fluorescein, Cy3 and Cy5 or indirectly labeled with biotin–dUTP and subsequently detected by diethylaminocoumarin (DEAC)-conjugated streptavidin. Chromosomes were identified by DAPI staining. Digital images were obtained using a Leica DMRXA epifluorescence microscope equipped with a cooled CCD camera (Princeton Instruments, Boston, MA). Cy3 (red; New England Nuclear, Boston, MA, USA), fluorescein (green; Fermentas Life Sciences, Milan, IT), Cy5 (IR; New England Nuclear, Boston, MA, USA) and DAPI (blue) fluorescence signals, which were detected using specific filters, were recorded separately as gray-scale images. Pseudocoloring and merging of images were performed with Adobe Photoshop software. Pt.5 To better clarify the complex rearrangement involving chromosomes 7 and 9 in PT19, a number of eighteen BAC clones, specific for 7p11.2, 7p14.1, 7p15.2, 7p15.3, 7p22.3, respectively, were pooled together and used as a single probe in FISH experiments.

RNA Isolation. Total RNA was extracted by the standard guanidinium thiocyanate-phenol-chloroform extraction method.[2]

Rapid Amplification of cDNA Ends (RACE) Analyses. 3' RACE PCR and 5' RACE PCR were performed using commercial assays (3' RACE or 5' RACE System for Rapid Amplification of cDNA Ends, Life Technologies, Carlsbad, California, USA), according to the manufacturer's instructions. A primer specific for *PAX5* exon1A was used in combination with the commercial UAP primer (3' RACE); first strand cDNA synthesis was primed using a gene-specific antisense oligonucleotide on the *PAX5*-partner gene sequence (5' RACE, see Supplementary Table S4). Gene specific primers were designed (Supplementary Table S4).
Both 3' and 5' RACE products were cloned into a T&A cloning vector (RBC Bioscience Corporation, New Taipei City, Taiwan), and selected clones were directly sequenced, in both directions and using vector-specific M13 oligonucleotides, by the Applied Biosystems ABI-3130 Genetic Analyzer instrument (Life Technologies). Alignment was carried out using the Basic Local Alignment Search Tool database (BLAST, www.blast.ncbi.nlm.nih.gov).

RT-PCR. One µg of total RNA was reversely transcribed with Superscript II reverse transcriptase (Life Technologies). RT-PCR was performed using a *PAX5* forward primer and the reverse primer

of the specific partner gene, to amplify the breakpoint region (Supplementary Table S4). All the RT-PCR reactions were performed in the following conditions: one step (2' at 94°C), thirty-five cycles of amplification (30 s at 94°C, 30 s at 60°C, 60 s at 72°C), using Platinum Taq DNA Polymerase (Life Technologies).

Characterization of *CHFR* expression in normal tissues.
To investigate the *CHFR* tissue-specific expression pattern, we tested cDNA from multiple tissues (bladder, brain, cervix, esophagus, heart, kidney, liver, lung, ovary, placenta, prostate, skeletal muscle, small intestine, spleen, testes, thymus, thyroid, and trachea) (First Choice Total RNA Survey Panel, Catalog No. AM6000, Ambion, Milan, Italy), together with cDNA from fetal brain RNA, peripheral blood, and bone marrow (Clontech, Saint-Germain-en-Laye, France). cDNA was obtained by reverse transcription using the QuantiTect Reverse Transcription Kit (Qiagen) and amplified by the Applied Biosystems Real-Time PCR System 7300 in the presence of 1X Platinum®SYBR Green qPCR SuperMix-UDG with ROX (Life Technologies), and 300nM of each sense and antisense primer. All measurements were performed at least in triplicate.
The PCR conditions were as it follows: 2 min at 50°C, 10 min at 95°C, followed by 40 cycles of 15 s at 95°C and 1 min at 60°C for all the primer pairs used. Melting curves were acquired and analyzed to control the specificity. Two primer pairs, corresponding to the *CHFR* wild-type and novel transcript isoform, were designed to amplify products of 65 and 133 bp, respectively. Primer sequences are reported in the Supplementary Table S4.
The gene expression level was calculated using the relative quantification approach based on the ΔΔCt method.[3] In all the experiments, *GAPDH* gene was used as reference, and normal bone marrow as calibrator.

RNA-Seq Analysis Total RNA was checked for integrity and purity by capillary electrophoresis (Agilent Bioanalyser 2100, Agilent Technologies, Santa Clara, CA, USA), requiring a RNA Integrity Number of 8 or more. 2µg RNA was used to prepare a double-stranded cDNA library for high-throughput sequencing by Truseq RNA kit (Illumina, San Diego, CA, USA). 6pM of the library was loaded in one lane of an Illumina flowcell, subjected to cluster bridge amplification in a cBot station and paired-end sequenced on a GAIIx instrument with a 2×76 protocol (Illumina pipeline version 1.6). After sequencing run completion, clusters were converted into reads by using Illumina's proprietary software OLB (Offline BaseCaller, v.1.9.0). Raw reads were mapped against the University of California Santa Clara (UCSC) human genome sequence, release 19 (hg19) by using BWA (v. 0.5.9, allowing up to 3 mismatches in the alignment and a trimming of bases having a phred quality below 15).[4] Not perfectly mapped or trimmed read pairs (i.e.: those having at least one unmapped mate, more than 7 trimmed bases, or more than 1000 nt between the two mates) were re-aligned against hg19 and RefSeq gene models (http://www.ncbi.nlm.nih.gov/refseq/) by TopHat (v. 1.3.1, allowing up to 3 mismatches).[5] Reads having at least one unmapped pair, or characterized by a distance between the two mates above 1000 nucleotides were used as an input to FusionMap software (v. 6.2.1.41, using Human B37 and RefSeq as reference genome and transcriptome model, respectively)[6] to search for genomic rearrangements.
Candidate events derived from FusionMap were further filtered as follows:
a) We kept only those events having 3 or more non-clonal supporting reads
b) We excluded events eventually localized in repeated regions of the genome and specifically:
-all the records in FusionMap output sharing one of the breakpoints (even though they had a different mate);
-candidates having the whole junction region (i.e. 60 nt across the junction) completely matched anywhere in the reference genome (in order to compensate for transcripts whose structure was not considered in FusionMap). This was assessed by means of a Blat[7] alignment against the hg19 reference;

-candidates having either of the two halves (i.e. length=30 nt) of the junction matching in multiple locations of the genome according to their Blat alignment.
c) We required a coverage of at least 10 reads in the regions flanking the rearrangement, in order to exclude those regions not sufficiently covered by sequencing reads to have a reliable characterization. Sequencing coverage was calculated by using the utility samtools (v. 0.1.17, command mpileup); [8]
d) We filtered out the events having an incidence of the reads supporting the fusion event below 10% of the total reads on either side.

Copy Number Variation analysis. Five diagnostic samples (patients 5, 7, 10, 15 and 19), as well as their corresponding remission specimens, were genotyped by the Affymetrix Cytogenetics Whole Genome 2.7M Array (Affymetrix, Santa Clara, CA), while patient 22 has been analyzed by Genome-Wide Human SNP Array 6.0 (Affymetrix). The Affymetrix® Cytogenetics Whole Genome 2.7M and 6.0 Arrays provide dense coverage across the entire genome due to high number of markers, including 400,000 SNPs and 2.3 millions of non-polymorphic copy-number markers. Therefore, it enables to detect small aberrations (gains/losses) and copy number neutral loss of heterozygosity (LOH) regions.
Briefly, 100ng of genomic DNA were amplified by an overnight whole-genome amplification reaction and purified by magnetic beads according to the manufacturer's instructions. The samples were then fragmented to generate small (<300 bp) products which were subsequently biotin-labeled, denatured and loaded into the arrays. After hybridization, the chips were washed, stained (streptavidin-PE), and scanned using the Gene Chip® Scanner 3000. Affymetrix GeneChip Command Console™ (AGCC) v3.1 generated .CEL files, which were analyzed by Chromosome Analysis Suite software (Affymetrix).
A minimum number of 20 markers and a minimal length of 100 Kb and 50 Kb, respectively were considered for defining amplified and deleted regions. Strechtes of Loss Of Heterozygosity were annotated if covering a minimal region of 2000 Kb.

Supplementary References

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Supplementary Figure Legend.

Supplementary Figure S1. FISH on deleted cases. Results obtained on 14 cases with probes identifying the deleted 9p segment including *PAX5*. BAC probes, shown below each panel, are coloured according to their corresponding signals in the merged images. The white arrows indicate the deleted chromosome 9 [del(9)].

Supplementary Figure S2. *CHFR* novel RNA isoform *CH-029, exons 19-20* and *CH-001.* The analyses have been performed in patient 5, cell lines, and normal tissues. **A-** Contig map of BAC and fosmid clones used in FISH experiments to identify the breakpoint region in 12q24.33 chromosome band in patient 5. The fosmid in red, containing *POLE*, showed a splitting signal on both der(9) and der(12); the BAC in green was retained on der(12), while the clones in blue were completely translocated on der(9). Alternative transcripts of *CHFR* are reported according to the UCSC Genome Browser (GRCh37/hg19). Colored dots correspond to primers, listed in the black right panel, used in RT-PCR and/or RT-qPCR experiments **B-** The new transcript *CH-029* (*left panel*), as well as the newly identified *exons 19-20* (*right panel*), are expressed both in patient 5 (lane 1) and in TOM1 BCP-ALL control cell line (lane 3). **C**- *CH-001* (*left panel*), as well as the wild type *PAX5* (*right panel*), are expressed in patient 5 (lane 1) and in the TOM1 cells (lane 3). **D**- RT-PCR screening of *CH-029* expression in 293T (kidney fibroblasts, lane 3) and several haematological cell lines [TOM1, and REH (lanes 1 and 4, respectively; B-ALL); HL-60, and THP1 (lanes 2 and 7, respectively; AML); U937 (lane 5, histiocytic lymphoma); K562 (lane 6, CML). **E-** Tissue expression pattern of *CH-001* (dark gray, *upper panel*) and *CH-029* (light gray, *bottom panel*). Quantitative data are shown in the table. **F-** Pseudocolor merged image, and raw images corresponding to FISH performed by co-hybridizing probes specifically designed for *PAX5*, *POLE*, and *CHFR* and labeled with different fluorochromes. FISH results confirm the map position of the breakpoint on chromosome 12 as within *POLE*. No extra signal for the probe specific for *CHFR* was observed on der(9). Consequently, being aware that *CHFR* shows an opposite transcriptional orientation to *PAX5* and that a simple t(9;12) translocation could not generate an in-frame fusion, we hypothesize that a cryptic rearrangement involving *CHFR* (i.e. not detectable at the level of FISH resolution) should have necessarily occurred prior or at the time of the translocation.

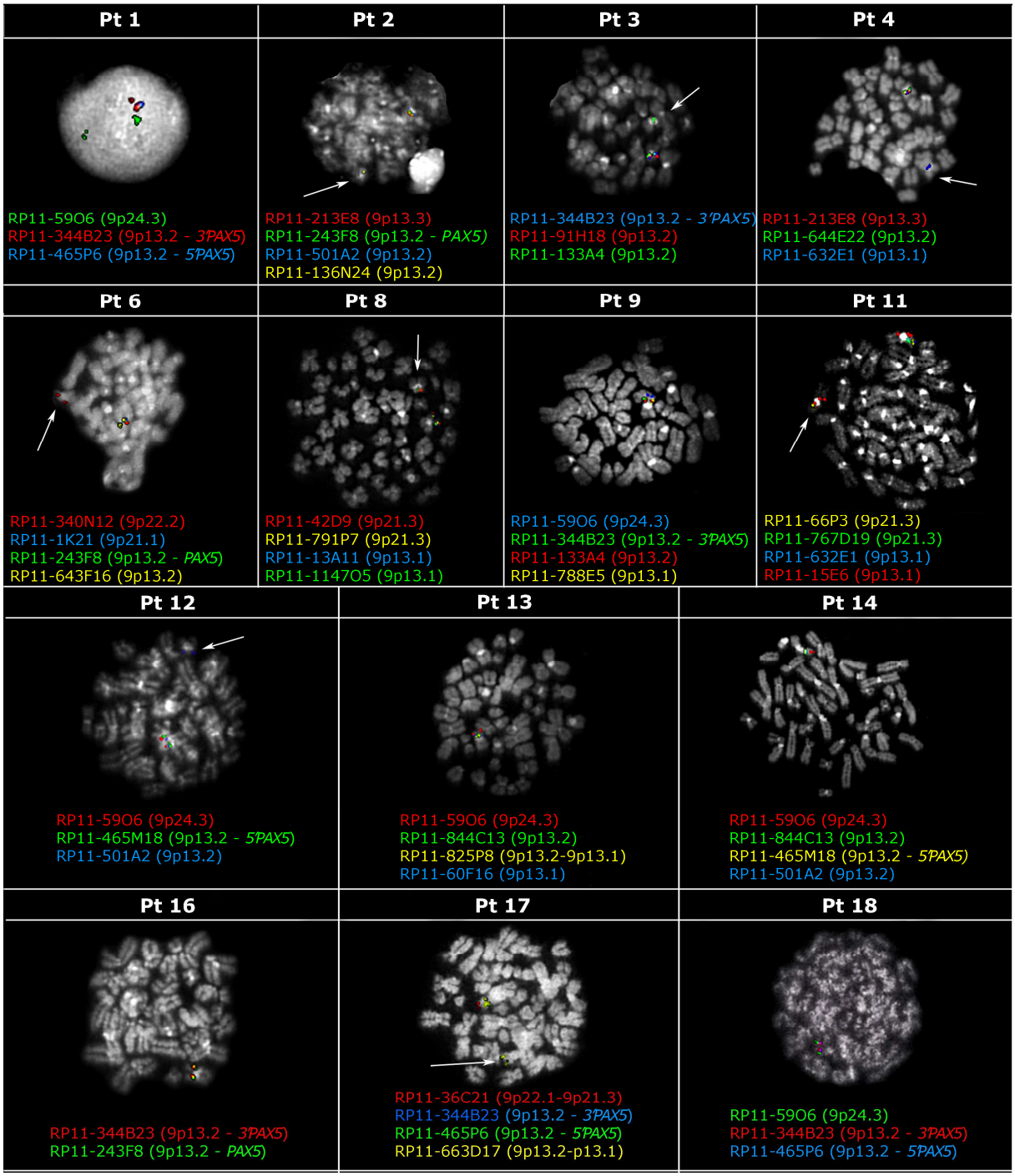
Supplementary Figure S3. *POM121C/PAX5* fusion gene by RNA-Seq analysis. Mapping of reads on hg19 reference sequence (from UCSC genome browser) is shown in the upper part of the plot. Sequencing reads supporting the fusion are shown as mapping across the ad-hoc built reference of the fusion transcript. The blue track displays the depth of coverage across the junction.

Supplementary Figure S4. FISH results obtained in patient 19. A- FISH pseudocolor merged image, and raw images corresponding to each probe labeled with a different fluorochrome. RP11-1054F23 (mapping at the 3' end of POM121C) displayed splitting signals on both dic(7;9) and der(9)t(?;7;9) chromosomes, apart from a signal on the normal chromosome 7. This result indicates that there are two copies of the *PAX5/POM121C* fusion gene, since RP11-1054F23 co-localizes with the PAX5 probes on both dic(7;9) and der(9) (see also Figure 1A). Conversely, RP11-93G19 was used in a pool of probes together with all the BACs listed in the Supplementary Table S3 (and indicated by a double asterisk) and mapping on the short arm of chromosome 7. This pool of probes [behaving as a Partial Chromosome Painting (PCP) of 7p] displayed signal on the normal chromosome 7, on the dic(7;9) and on a marker chromosome [that we indicated as der(?)t(?;7)], possibly resulting from a further rearrangement (translocation) of a second copy of the dic(7;9) with a chromosome of unknown origin (indicated as chr?). **B**- The results of each informative hybridization are illustrated by coloured circles corresponding to every single used probe.

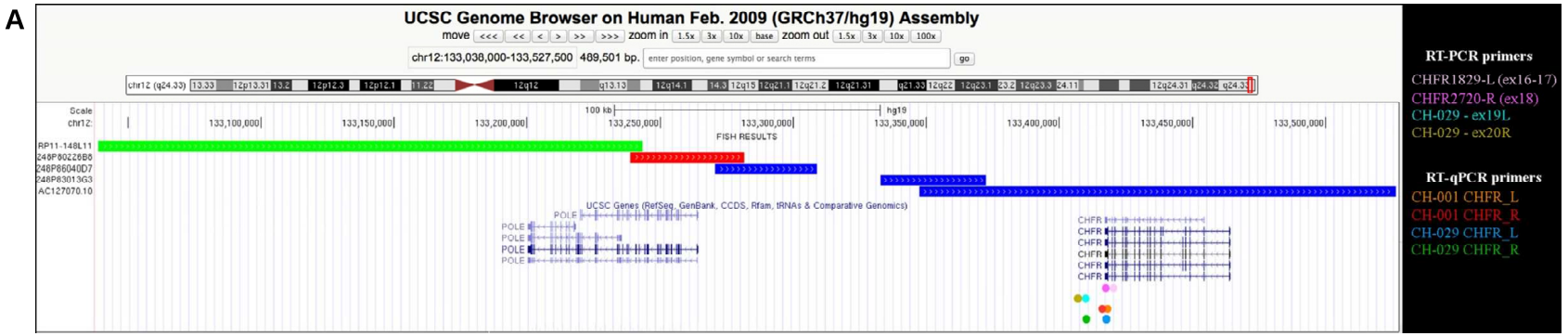
Chromosome 7q and 9 are indicated by purple and green areas, respectively. White areas correspond to unknown chromosome material.

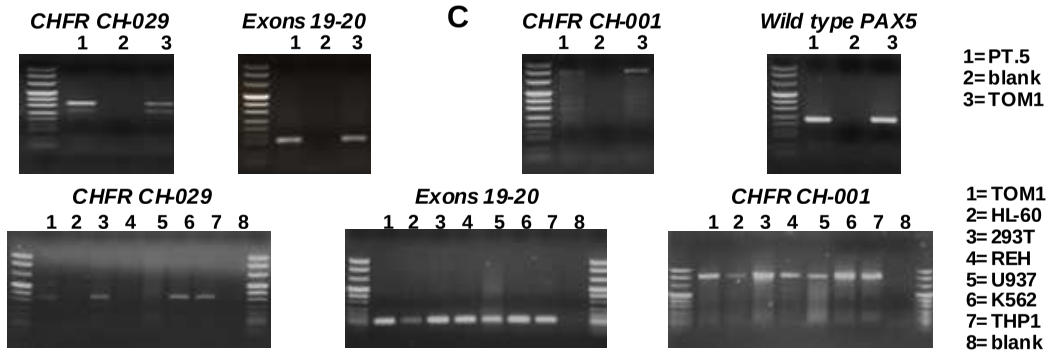
Supplementary Figure S5. *PAX5/MLLT3* fusion gene. A- Schematic representation of the 9p13 deletion, leading to the genesis of the *PAX5*/*MLLT3* fusion gene**. B-** Partial chromatograms showing the fusion junctions at both genomic (*left*) and RNA (*right*) level between *PAX5* and *MLLT3.* **C-** Copy Number Variation analysis performed in patient 20.

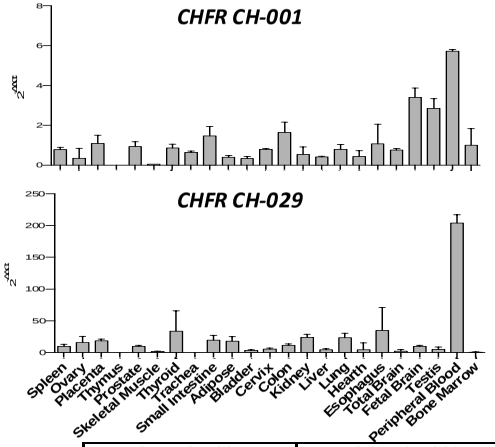
Supplementary Figure S1

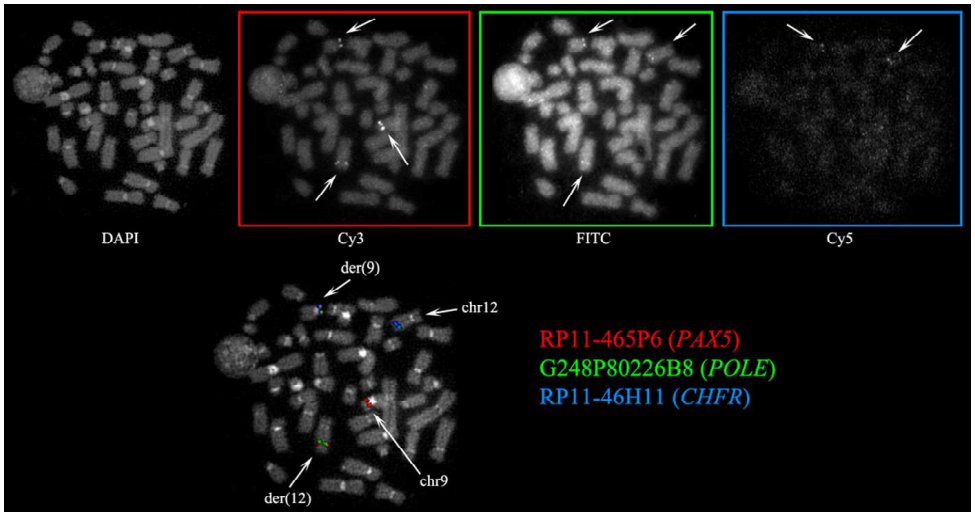


Supplementary Figure S2



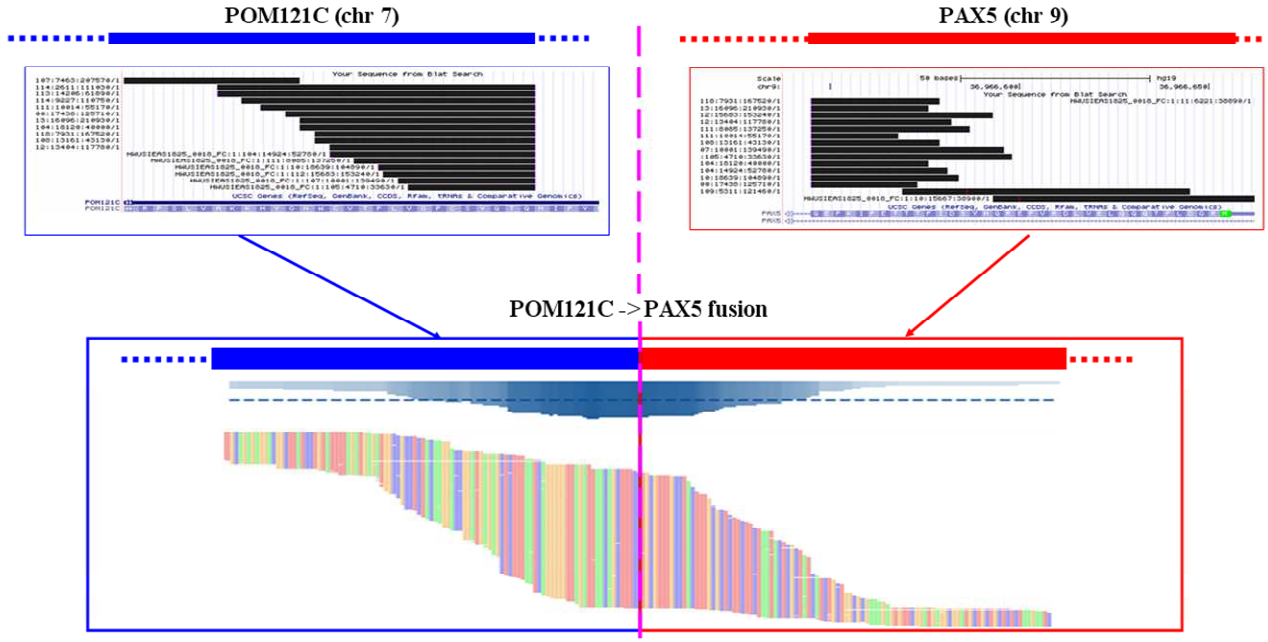






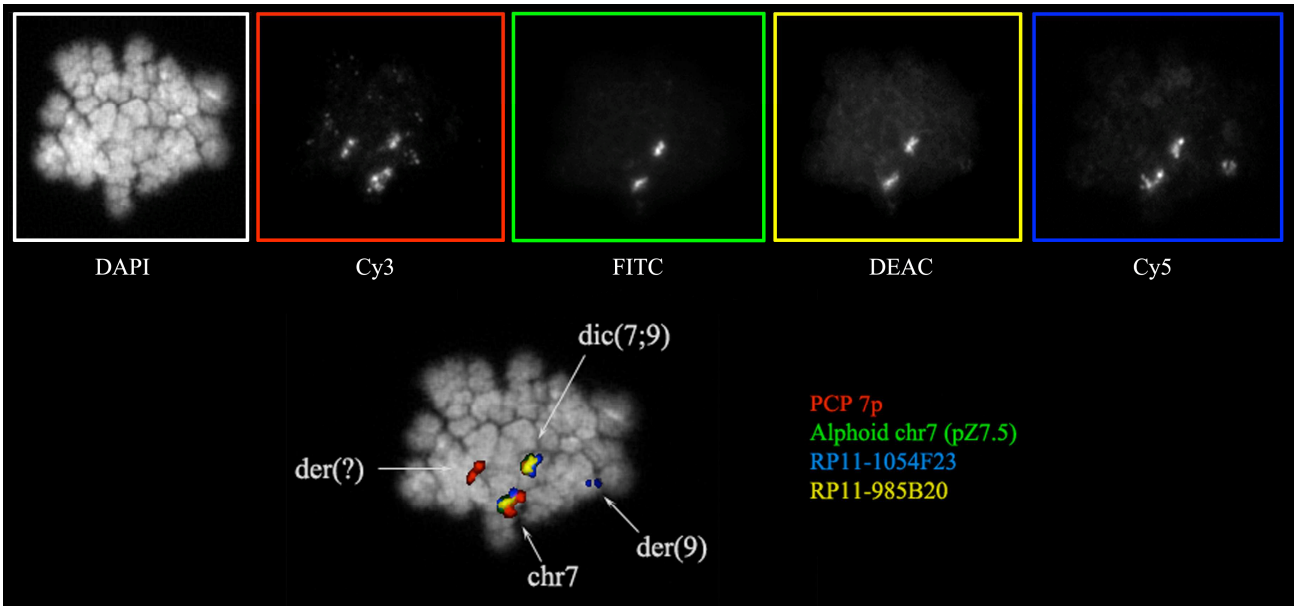
Samples	CHFR CH-001			CHFR CH-029		
	$2^{-\Delta\Delta Ct}$	Range $2^{-(\Delta\Delta Ct \pm SD)}$		$2^{-\Delta\Delta Ct}$	Range $2^{-(\Delta\Delta Ct \pm SD)}$	
Spleen	0.79	0.68	0.91	9.71	7.26	13.00
Ovary	0.35	0.14	0.86	16.04	10.04	25.62
Placenta	1.10	0.81	1.50	18.74	16.20	21.69
Thymus	0.00	0.00	0.00	0.00	0.00	0.00
Prostate	0.94	0.75	1.18	10.08	8.92	11.39
Skeletal Muscle	0.05	0.05	0.06	2.04	1.94	2.14
Thyroid	0.88	0.73	1.05	34.14	17.70	65.86
Trachea	0.64	0.57	0.72	0.00	0.00	0.00
Small Intestine	1.47	1.11	1.95	19.52	13.88	27.44
Adipose	0.40	0.32	0.49	18.04	12.98	25.08
Bladder	0.34	0.27	0.44	3.64	2.92	4.54
Cervix	0.80	0.77	0.83	5.64	4.19	7.59
Colon	1.65	1.25	2.17	11.75	10.06	13.73
Kidney	0.54	0.32	0.92	23.86	19.87	28.66
Liver	0.42	0.40	0.45	4.73	3.50	6.39
Lung	0.81	0.64	1.04	23.59	18.26	30.48
Hearth	0.43	0.25	0.73	4.81	1.49	15.50
Esophagus	1.07	0.55	2.06	35.02	17.18	71.38
Total Brain	0.76	0.68	0.84	2.17	0.87	5.41
Fetal Brain	3.41	3.01	3.87	9.83	8.04	12.01
Testis	2.86	2.45	3.34	5.13	2.93	9.00
Peripheral Blood	5.72	5.64	5.81	204.13	192.26	216.73
Bone Marrow	1.00	0.54	1.84	1.00	0.64	1.57

Supplementary Figure S3

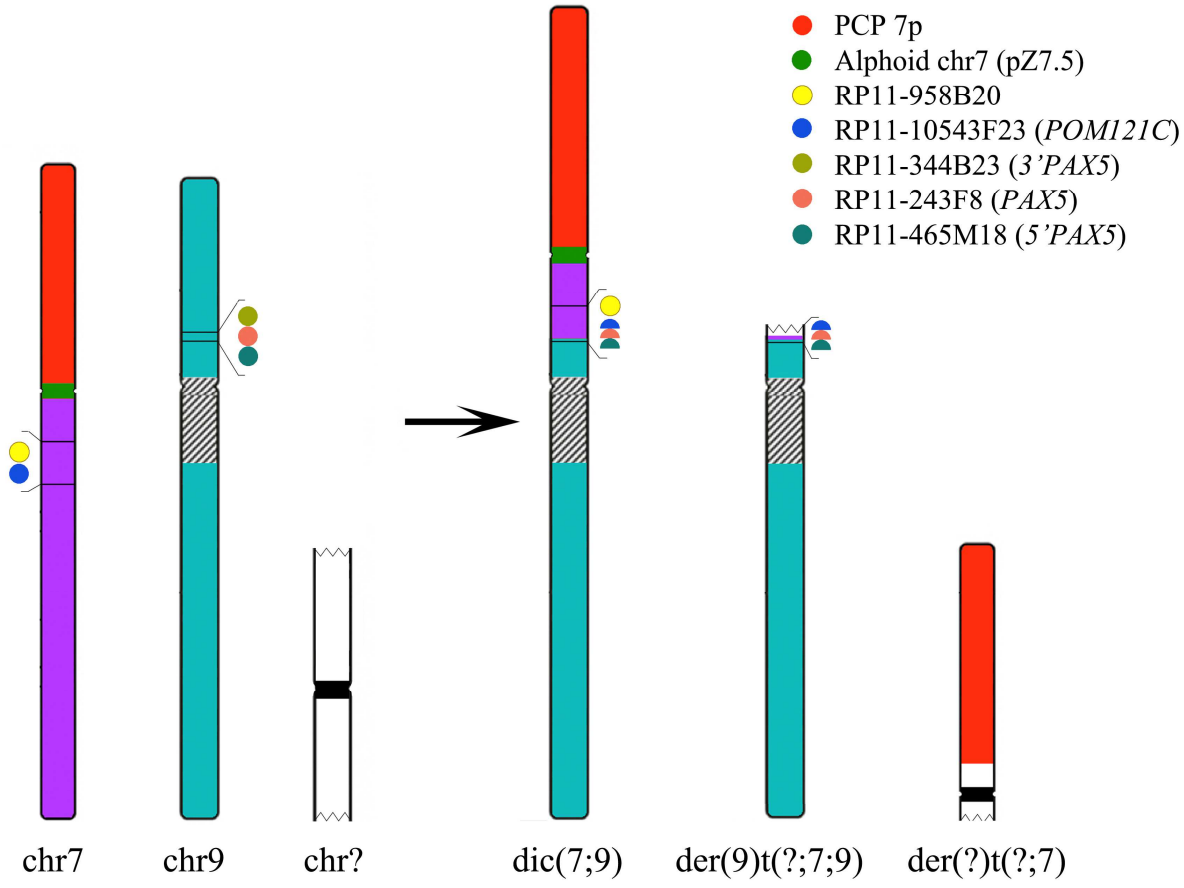


Supplementary Figure S4

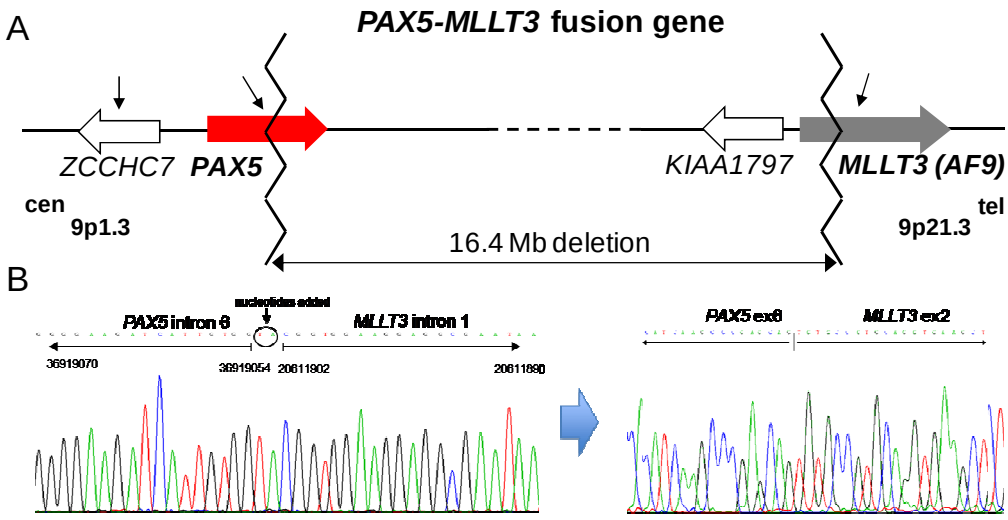
A



B



Supplementary Figure S5



C

Chr	Type	From	To	Size (kbp)	Marker Count	Genes	sno/miRNA
1	Loss	p31.1	p31.1	41	45	-----	-----
2	Loss	p11.2	p11.2	393	91	-----	-----
2	Loss	p22.3	p22.3	36	24	-----	-----
3	Loss	p21.31	p21.31	47	42	-----	-----
4	Gain	p15.1	p15.1	51	44	-----	-----
4	Loss	q13.2	q13.2	114	63	UGT2B17	-----
5	Gain	p15.2	p15.2	72	21	-----	-----
6	Loss	q16.3	q16.3	24	45	-----	-----
6	Loss	q14.1	q14.1	58	102	-----	-----
6	Loss	q12	q12	40	57	-----	-----
7	Loss	p12.2	p12.2	42	29	**IKZF1**	-----
8	Gain	p11.23	p11.22	152	58	ADAM3A, ADAM3A, ADAM3A, ADAM5P	-----
8	Loss	p23.1	p23.1	130	40	FAM86B2, DEFB109P1, FAM66A	-----
8	Loss	p23.1	p23.1	633	144	DEFB106A, DEFB106B, SPAG11B, SPAG11B, DEFB104B, DEFB105A, DEFB103A, SPAG11B, SPAG11B, DEFB107B, ...	-----
9	Loss	p13.2	p21.3	16209	11234	**from PAX5 to MLLT3**	hsa-mir-873, hsa-mir-876, hsa-mir-31, hsa-mir-491,
11	Loss	q11	q11	78	61	OR4C6, OR4P4, OR4S2	-----
14	Loss	q32.33	q32.33	757	323	LOC100133469, KIAA0125, ADAM6	-----
15	Loss	q21.1	q21.1	21	20	-----	-----
15	Loss	q11.2	q11.2	414	80	-----	-----
15	Loss	q11.2	q11.2	958	244	LOC646214, POTEB, OR4M2, LOC650137, OR4N4, CXADRP2, LOC727924, BCL8	hsa-mir-1268
15	Loss	q14	Q14	155	79	GOLGA8B, GOLGA8B, GOLGA8A	hsa-mir-1233
16	Gain	p11.2	p11.1	258	150	LOC283914, LOC283914, LOC146481	-----
18	Loss	q21.1	q21.1	25	34	SMAD2, SMAD2, SMAD2	-----
X	Gain	q21.31	q21.31	250	69	-----	-----
X	Gain	q21.32	q21.32	62	36	-----	-----
X	Gain	q21.31	q21.31	530	111	-----	-----
X	LOH	q11.1	Q28	92253	21562	CT47A5, CUL4B, DACH2, CT47A6, MAGEC3, TSPAN6, CXorf39, CXorf48, GLRA4, SLITRK2, ...	hsa-mir-1321, hsa-mir-514-3, hsa-mir-652, hsa-mir-892b, hsa-mir-325, hsa-mir-513b, hsa-mir-507, ...
X	LOH	p22.33	p11.1	55036	14761	RGN, TRAPPC2, GAGE12E, MAGED4, CLCN4, MAGEH1, UXT, HSD17B10, KDM6A, PIR, ...	hsa-mir-222, hsa-mir-660, hsa-mir-651, ACA12, hsa-mir-502, U107, ...

Supplementary Table S1. Clinical and cytogenetic features of patients carrying a PAX5 alteration.

Pt	Gender	Age	WBC count	Immunophenotype	Karyotype	*PAX5* Status (FISH)
1	F	1	5,500	cALL	46,XX,t(2;11)(q10;p10),inv(7)(p15q11),del(9)(p22)[4]/46,XX[13]	**deleted**
2	M	5	71,200	cALL	45,XY,del(6)(q?),der(9)t(9;?)(p13;?),-13,der(19)t(19;?)(p or q;?)[6]/46,XY[2]	**deleted**
3	M	5	42,060	cALL	45,XY,dic(9;20)(p13;q11)[15]/46,idem,+20[3]/46,XY[1]	**deleted**
4	M	7	65,490	cALL	46,XY,dic(9;17)(p13;p11),der(12)t(Y;12)(q1?;p1?),+ mar [9]	**deleted**
5	M	4	19,800	cALL	46,XY,t(9;12)(p13;q24.3)[5]	**translocated-*PAX5/CHFR***
6	M	4	10,800	cALL	46,XY,del(9)(p?)[2]/46,XY[7]	**deleted**
7	F	1	370,000	cALL	46,XX,der(7)t(7;11)(q11.2;q12),der(9)t(7;9)(q11.2;p13),-11,+mar[10]/46,XX[2]	**translocated-*PAX5/AUTS2***
8	M	15	13,240	pre-pre-B-ALL	47,XY,+8,del(9)(p?)[6]	**deleted**
9	F	9	29,000	cALL	46,XX,del(9)(p12)[5]/46,XX[5]	**deleted**
10	M	3	1,240	pre-B-ALL	45,XY,-7,der(9)t(7;9)(q22;p22)[3]/46,XY[8]	**translocated-*PAX5/AUTS2***
11	F	8	109,180	cALL – biclonal B	na	**deleted**
12	M	1	81,370	pre-B ALL	na	**deleted**
13	M	4	17,690	cALL	46,XY,dic(9;20)(p11;q11),+21[9]/47,idem,+22[4]	**deleted**
14	M	15	50,690	cALL	na	**deleted**
15	M	10	124,000	cALL	45,XY,dic(9;12)(p13;p13)[6]/46,XY[3]	**translocated-*PAX5/SOX5***
16	M	4	19,300	pre-pre-B / cALL	46-49,XY,+X,del(9)(p?) or dic(9;20),+21,+21[cp7]/46,XY[2]	**deleted**
17	M	1	123,000	cALL	46,XY,t(9;10)(q33;q23),del(9)(p12p21)[11]/46,XY[1]	**deleted**
18	M	13	147,000	pre-preB/cALL	46,XY,r(5),del(9)(p12),der(10),der(11)t(11;13)(q23;q12)t(13;22)(q14;q1?),der(13)t(11;13),add(17)(p1?),der(22)t(13;22)[4]	**deleted**
19	M	1	8,040	cALL	46,XY,t(2;6)(q33;q13),dic(7;9)(q11;p13)?inv(7)(p12p22),+der(9)[11]/47,idem,+mar[2]/46,XY[4]	**translocated-*PAX5/POM121C***
20	M	57	75,000	ALL	46,XY,t(9;22)(q34;q11)[4]/46,XX[6]	**deleted-*PAX5/MLLT3***

M=male; F=female; cALL=common Acute Lymphoblastic Leukemia; WBC count= number of white blood cells/µl; na=not available.

Supplementary Table S2. BAC used for FISH analysis to characterize the *PAX5* deleted cases.

SUPPLEMENTARY TABLE S2. BACs used for FISH analysis to characterize *PAX5* deleted cases.					PT															
GENE	BAC NAME	Acc. No.	CHR. BAND	MAP POSITION (hg19)	1	2	3	4	6	8	9	10	11	12	13	14	16	17	18
DOCK8	RP11-59O6	/	9p24.3	chr9:198,713-383,816	9; del(9)	9	9	9	9; del(9)	9; del(9)	9	9	9; del(9)	9	9	9	9	9	9
	RP11-10G22	/	9p24.3	chr9:1,533,223-1,709,628	9	-	-	-	-	-	-	-	-	-	-	-	-	9	-
PTPRD	RP11-338L20	/	9p24.1-9p23	chr9:8,981,678-9,142,717	-	-	-	-	-	9; del(9)	-	-	9; del(9)	-	-	-	-	-	-
	RP11-298E19	/	9p22.3	chr9:14,310,338-14,474,348	9	-	-	-	-	-	-	-	-	-	-	-	-	9	-
CNTLN	RP11-340N12	/	9p22.2	chr9:17,146,369-17,308,494	9	-	-	-	9; del(9)	-	-	-	-	-	-	-	-	9	-
NFIB	RP11-296P7	/	9p22.1	chr9:19,026,890-19,028,127	-	-	-	-	-	9; del(9)	-	-	9; del(9)	-	-	-	-	-	-
DENND4C	RP11-101L23	/	9p22.1	chr9:19,142,752-19,339,389	9	-	-	-	-	-	-	-	-	-	-	-	-	9	-
ACER2; SLC24A2	RP11-361B3	/	9p22.1	chr9:19,390,694-19,576,389	-	-	-	-	-	9; del(9)	-	-	-	-	-	-	-	-	-
	RP11-36C21	/	9p22.1-9p21.3	chr9:19,818,493-19,989,520	9	-	-	-	-	9; del(9)	-	-	-	-	-	-	-	9	-
	RP11-410F21	/	9p21.3	chr9:20,143,896-20,316,463	-	-	-	-	9; del(9)	-	-	-	-	-	-	-	-	-	-
MLLT3	RP11-1100C5	/	9p21.3	chr9:20,422,666-20,591,558	-	-	-	-	-	9; del(9)*	-	-	9; del(9)	-	-	-	-	-	-
MLLT3; FOCAD	RP11-42D9	/	9p21.3	chr9:20,570,274-20,746,915	-	-	-	-	-	9; del(9)*	-	-	9; del(9)	-	-	-	-	-	-
FOCAD	RP11-791P7	/	9p21.3	chr9:20,663,654-20,851,416	-	-	-	-	-	9	-	-	-	-	-	-	-	-	-
FOCAD	RP11-66P3	/	9p21.3	chr9:20,784,480-20,945,583	-	-	-	-	-	9	-	-	9; del(9)	-	-	-	-	-	-
FOCAD; PTPLAD2	RP11-767D19	/	9p21.3	chr9:20,917,109-21,086,861	-	-	-	-	9	9	-	-	9	-	-	-	-	9	-
	RP11-354P17	/	9p21.3	chr9:21,343,280-21,536,271	-	-	-	-	-	9	-	-	9	-	-	-	-	-	-
MTAP; CDKN2A	RP11-615P15	/	9p21.3	chr9:21,764,403-21,981,385	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
DMRTA1	RP11-624G3	/	9p21.3	chr9:22,304,545-22,471,940	-	-	-	-	-	-	-	-	9	-	-	-	-	-	-
	RP11-12N11	/	9p21.3	chr9:23,436,448-23,597,889	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
ELAVL2	RP11-110I5	/	9p21.3	chr9:23,688,378-23,854,636	-	-	-	9	-	-	-	-	-	-	-	-	-	-	-
ELAVL2	RP11-31K16	AL161628.9	9p21.2	chr9:23,722,809-23,833,246	-	-	-	-	-	-	-	-	9	-	-	-	-	-	-
	RP11-477G9	AL353753.6	9p21.2	chr9:25,920,470-26,099,334	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
	RP11-48L13	AL354676.10	9p21.1	chr9:29,577,130-29,649,069	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
TAF1L; TMEM215	RP11-1K21	/	9p21.1	chr9:32,629,173-32,804,393	-	-	-	-	9	9	-	-	-	-	-	-	-	-	-
	RP11-573M23	AL356494.13	9p13.3	chr9:34,335,596-34,417,345	-	-	-	-	-	9	-	-	9	-	-	-	-	-	-
	RP11-145E14	/	9p13.3	chr9:35,574,708-35,746,923	-	-	-	-	-	9	-	-	9	-	-	-	-	-	-
	RP11-213E8	/	9p13.3	chr9:35,751,414-35,902,246	-	9	9	9	-	9	-	-	-	-	-	-	-	-	-
RECK	RP11-626F21	/	9p13.3	chr9:36,020,455-36,184,946	-	9	9	9	-	9	-	-	9	-	-	-	-	-	-
RNF38	RP11-844C13	/	9p13.2	chr9:36,331,767-36,515,625	-	-	-	-	-	-	-	9	9	9	9	9	-	-	-
	RP11-450B8	/	9p13.2	chr9:36,509,103-36,709,458	-	9	9	9	-	9	-	9	-	9	-	-	-	-	-
3' PAX5	RP11-344B23	/	9p13.2	chr9:36,774,163-36,949,832	9	9	9	9	-	9	9	9	-	-	-	-	9	9	9
PAX5	RP11-243F8	/	9p13.2	chr9:36,844,935-37,033,204	9	9	9	9	9	9	9	9; del(9)*	-	-	-	-	9	9; del(9)*	-
5' PAX5	RP11-465P6	/	9p13.2	chr9:36,909,192-37,075,371	9	9	9	9	-	9	9	9; del(9)*	-	-	-	-	-	9; del(9)*	9
5' PAX5	RP11-465M18	/	9p13.2	chr9:36,975,063-37,209,364	-	-	9	-	-	-	9	9; del(9)	9	9	9	9	-	9; del(9)	-
5' PAX5	RP11-652D9	/	9p13.2	chr9:36,986,566-37,189,379	-	9	9	9	-	9	9	9; del(9)	-	9	-	-	-	-	-
ZCCHC7	RP11-91H18	/	9p13.2	chr9:37,138,096-37,318,047	-	-	9	-	-	-	-	9; del(9)	-	9	-	-	-	-	9
ZCCHC7	RP11-133A4	/	9p13.2	chr9:37,221,544-37,396,251	-	-	9; del(9)*	-	-	-	9	9; del(9)	-	9; del(9)*	-	-	-	-	-
	RP11-501A2	/	9p13.2	chr9:37,392,581-37,596,054	-	9	9; del(9)	9	-	9	9	9; del(9)	9	9; del(9)	9	9	-	-	-
	RP11-643F16	/	9p13.2	chr9:37,628,199-37,777,278	-	-	-	-	9	-	9	-	-	-	-	9	-	9; del(9)	-
	RP11-644E22	/	9p13.2	chr9:37,777,285-37,950,151	-	9	9; del(9)	9	-	9	9	-	-	-	-	9	-	-	-
SHB	RP11-666G2	/	9p13.2	chr9:37,945,191-38,138,311	-	-	-	-	-	-	-	-	9	-	9	9; del(9)	-	-	-
	RP11-136N24	/	9p13.2	chr9:38,090,149-38,279,495	-	9; del(9)	-	9	-	-	-	-	-	-	-	-	-	-	-
	RP11-663D17	/	9p13.2-9p13.1	chr9:38,267,084-38,441,277	-	-	-	-	9	-	9	-	-	-	-	-	-	-	9
	RP11-825P8	/	9p13.2-9p13.1	chr9:38,291,105-38,491,094	-	9; del(9)	-	9	-	-	-	-	9	-	9	9; del(9)	-	9; del(9)	-
	RP11-1050I3	/	9p13.1	chr9:38,444,223-38,618,809	-	-	-	9; del(9)*	-	-	9	-	9	-	-	-	-	-	-
	RP11-632E1	/	9p13.1	chr9:38,512,027-38,684,880	-	9; del(9)	-	9; del(9)	-	9	9	-	9	-	-	-	-	-	-
	RP11-788E5	/	9p13.1	chr9:38,568,002-38,733,846	-	9; del(9)	9; del(9)	9; del(9)	-	9	9	-	-	-	-	-	-	-	-
	RP11-15E6	/	9p13.1	chr9:38,733,908-38,890,388	-	-	-	-	-	-	-	-	-	-	9	9; del(9)	-	-	-
	RP11-622N4	/	9p13.1	chr9:38,797,151-38,978,531	-	-	9; del(9)	-	-	9	-	-	9; del(9)	-	-	-	-	-	-
	RP11-13A11	/	9p13.1	chr9:38,798,813-38,973,416	-	-	-	-	-	9	-	-	-	-	9	9; del(9)	-	-	-
	RP11-1147O5	/	9p13.1	chr9:38,890,456-39,046,992	-	-	-	-	-	9; del(9)*	-	-	9	-	9	-	-	-	-
	RP11-1150N16	/	9p13.1	chr9:38,995,367-39,128,143	-	-	-	-	-	9; del(9)	-	-	9	-	9	-	-	-	-
	RP11-60F16	/	9p13.1	chr9:38,978,534-39,150,161	-	-	-	-	-	9; del(9)	-	-	9	-	9	-	-	-	-

Symbols: / BACs mapped by end sequencing; - not analysed; * clones showing a fainter signal intensity on the deleted chromosome 9 [del(9)] than on the normal homolog

deleted
partially deleted
undeleted

Supplementary Table S3. BAC used for FISH analysis to characterize the *PAX5* translocated cases.

GENE	BAC NAME	Acc. No.	CHR. BAND	MAP POSITION (hg19)	FISH RESULTS				
					Pt 5	Pt 7	Pt 10	Pt 15	Pt 19
	RP11-59O6	/	9p24.3	chr9:198,713-383,816	9, der(12)	-	-	9	-
	RP11-450B8	/	9p13.2	chr9:36,509,103-36,709,458	-	-	-	9	-
3' PAX5	RP11-344B23	/	9p13.2	chr9:36,774,163-36,949,832	9, der(12)	9, der(7)*	-	9	9
PAX5	RP11-243F8	/	9p13.2	chr9:36,844,935-37,033,204	9, der(9)*, der(12)	9, der(9), der(7)*	-	9, dic(9;12)	9, dic(7;9), der(9)
5' PAX5	RP11-465P6	/	9p13.2	chr9:36,909,192-37,075,371	9, der(9), der(12)	9, der(9)	-	9, dic(9;12)	-
5' PAX5	RP11-465M18	/	9p13.2	chr9:36,975,063-37,209,364	9, der(9), der(12)*	-	-	-	9, dic(7;9), der(9)
	RP11-133A4	/	9p13.2	chr9:37,221,544-37,396,251	-	-	9, der(9)	-	-
	RP11-713A20**	/	7p22.3	chr7:13,709-203,581	-	-	-	-	
	RP11-1133D5**	/	7p22.3	chr7:1,135,418-1,297,910	-	-	-	-	
	RP11-414M15**	/	7p22.3	chr7:2,072,278-2,274,856	-	-	-	-	
	RP11-964D13**	/	7p22.2	chr7:2,862,624-3,048,184	-	-	-	-	
	RP11-3F6**	/	7p22.2	chr7:3,540,892-3,709,123	-	-	-	-	
	RP11-135H12**	/	7p22.2	chr7:3,640,957-3,800,970	-	-	-	-	
	RP11-117I8**	/	7p22.2	chr7:3,692,526-3,867,016	-	-	-	-	
	RP11-42B7**	/	7p22.2	chr7:4,159,942-4,322,618	-	-	-	-	
	RP11-93G19**	/	7p22.1	chr7:5,552,812-5,706,061	-	-	-	-	7, dic(7;9), der(?)
	RP11-269M4**	/	7p15.3	chr7:20,910,403-21,076,176	-	-	-	-	
	RP11-465M2**	/	7p15.3	chr7:21,253,297-21,448,738	-	-	-	-	
	RP11-1132K14**	/	7p15.2	chr7:27,146,581-27,303,174	-	-	-	-	
	RP11-147A4**	/	7p14.1	chr7:37,337,348-37,517,010	-	-	-	-	
	RP11-107G20**	/	7p14.1	chr7:38,539,793-38,694,006	-	-	-	-	
	RP11-177B1**	/	7p14.1	chr7:41,277,099-41,446,653	-	-	-	-	
	RP11-781C22**	/	7p11.2	chr7:54,987,970-55,164,028	-	-	-	-	
	RP11-433C12**	/	7p11.2	chr7:55,082,305-55,259,309	-	-	-	-	
	RP11-771A19**	/	7p11.2	chr7:55,153,402-55,342,655	-	-	-	-	
	pZ7.5				-	-	-	-	7, dic(7;9)
	RP11-958B20	/	7q11.23	chr7:74,564,923-74,912,389	-	-	-	-	7, dic(7;9)
POM121C	RP11-1054F23	/	7q11.23	chr7:74,924,365-75,079,589	-	-	-	-	7, dic(7;9), der(9)*
AUTS2	RP11-409E13	/	7q11.22	chr7:69,113,431-69,267,822	-	7, der(7)	-	-	-
AUTS2	RP11-168B15	/	7q11.22	chr7:69,470,146-69,653,704	-	7, der(7)	-	-	-
AUTS2	RP11-666K18	/	7q11.22	chr7:70,037,637-70,232,635	-	7, der(7), der(9)*	-	-	-
3' AUTS2	RP11-485M20	/	7q11.22	chr7:70,197,568-70,373,244	-	7, der(7), der(9)	-	-	-
	RP11-867L5	/	7q36.3	chr7:158,840,095-159,032,247	-	-	7, der(9)	-	-
	RP11-636N1	/	12p13.33	chr12:1,896,751-2,073,841	-	-	-	12	-
	RP11-639O1	/	12p13.2	chr12:11,737,815-11,916,241	-	-	-	12	-
	RP11-418C2	/	12p13.2	chr12:11,916,329-12,086,277	-	-	-	12	-
	RP11-502N13	AC008114.25	12p13.1	chr12:14,632,639-14,757,140	-	-	-	12	-
	RP11-1018J8	/	12p12.3	chr12:15,158,390-15,370,563	-	-	-	12	-
	RP11-489N6	/	12p12.3	chr12:16,193,015-16,279,962	-	-	-	12	-
	RP11-69C13	/	12p12.3	chr12:16,748,391-16,895,630	-	-	-	12	-
	RP11-871F6	/	12p12.3	chr12:17,532,546-17,749,334	-	-	-	12	-
	RP11-1144H2	/	12p12.3	chr12:19,415,511-19,596,445	-	-	-	12	-
	RP11-3F5	/	12p12.3	chr12:19,509,245-19,698,514	-	-	-	12	-
	RP11-352D19	/	12p12.3	chr12:19,596,473-19,774,537	-	-	-	12	-
	RP11-145J1	/	12p12.3	chr12:19,654,367-19,806,117	-	-	-	12	-
	RP11-412N19	/	12p12.3	chr12:19,713,852-19,857,192	-	-	-	12	-
	RP11-1006K22	/	12p12.3	chr12:19,819,709-20,011,494	-	-	-	12	-
	RP11-138F13	/	12p12.2	chr12:20,029,484-20,186,763	-	-	-	12	-
	RP11-12D15	/	12p12.1	chr12:22,319,120-22,478,292	-	-	-	12	-
	RP11-136L21	/	12p12.1	chr12:22,589,365-22,771,823	-	-	-	12	-
	RP11-449P1	/	12p12.1	chr12:22,786,727-22,949,390	-	-	-	12	-
	RP11-347F21	/	12p12.1	chr12:23,052,014-23,215,093	-	-	-	12	-
	RP11-1082A20	/	12p12.1	chr12:23,287,922-23,485,440	-	-	-	12	-
	RP11-760E23	/	12p12.1	chr12:23,497,977-23,680,073	-	-	-	12	-
	G248P80439E6	WI2-650J12	12p12.1	chr12:23,634,673-23,674,964	-	-	-	12	-
	G248P88992C6	WI2-2755E11	12p12.1	chr12:23,655,369-23,694,733	-	-	-	12	-
3' SOX5	RP11-98J14	/	12p12.1	chr12:23,671,226-23,822,046	-	-	-	12, dic(9;12)	-
SOX5	RP11-1152G11	/	12p12.1	chr12:23,803,421-23,946,124	-	-	-	12, dic(9;12)	-
SOX5	G248P82399C2	WI2-1644F4	12p12.1	chr12:23,929,057-23,970,777	-	-	-	12, dic(9;12)	-
SOX5	RP11-672O12	/	12p12.1	chr12:23,946,131-24,126,509	-	-	-	12	-
SOX5	RP11-162F10	/	12p12.1	chr12:24,012,390-24,167,969	-	-	-	12	-
SOX5	RP11-681A17	AC090675.10	12p12.1	chr12:24,132,898-24,304,940	-	-	-	12	-
SOX5	G248P84907B3	WI2-1187D6	12p12.1	chr12:24,285,277-24,327,630	-	-	-	12	-
SOX5	RP11-628A8	/	12p12.1	chr12:24,304,948-24,469,455	-	-	-	12, dic(9;12)	-
SOX5	RP11-77H23	/	12p12.1	chr12:24,459,186-24,647,483	-	-	-	12, dic(9;12)	-
5' SOX5	RP11-958I6	/	12p12.1	chr12:24,647,516-24,831,923	-	-	-	12, dic(9;12)	-
	RP11-485K18	AC024939.24	12p11.22	chr12:28,518,058-28,576,560	-	-	-	12, dic(9;12)	-
	RP11-313F23	AC140847.4	12p11.1	chr12:34,267,708-34,400,580	-	-	-	12, dic(9;12)	-
	RP11-450O16	/	12q24.12-q24.13	chr12:112,119,304-112,311,122	12, der(12)	-	-	-	-
	RP11-1128D1	/	12q24.13	chr12:112,799,929-112,950,786	12, der(12)	-	-	-	-
	RP11-136L23	/	12q24.23	chr12:118,479,911-118,640,443	12, der(12)	-	-	-	-
	RP11-80G18	/	12q24.23	chr12:120,013,477-120,190,144	12, der(12)	-	-	-	-
	RP11-1081C6	/	12q24.31	chr12:121,535,680-121,718,399	12, der(12)	-	-	-	-
	RP11-90B6	/	12q24.31	chr12:123,224,657-123,387,601	12, der(12)	-	-	-	-
	RP11-161A18	/	12q24.31	chr12:125,294,205-125,465,944	12, der(12)	-	-	-	-
	RP11-886H4	/	12q24.32	chr12:128,965,212-129,117,533	12, der(12)	-	-	-	-
	RP11-60M19	/	12q24.32	chr12:129,946,465-130,123,216	12, der(12)	-	-	-	-
	RP11-72E15	/	12q24.33	chr12:130,907,263-131,047,343	12, der(12)	-	-	-	-
	RP11-825F9	/	12q24.33	chr12:131,117,968-131,301,835	12, der(12)	-	-	-	-
	RP11-270L16	/	12q24.33	chr12:132,069,545-132,238,137	12, der(12)	-	-	-	-
	RP11-357I22	/	12q24.33	chr12:132,856,378-133,065,041	12, der(12)	-	-	-	-
POLE	RP11-148L11	/	12q24.33	chr12:133,038,915-133,243,361	12, der(12)	-	-	-	-
POLE	G248P80226B8	WI2-612C16	12q24.33	chr12:133,238,813-133,281,615	12, der(12), der(9)	-	-	-	-
	G248P86040D7	WI2-1588G13	12q24.33	chr12:133,270,749-133,308,916	12, der(9)	-	-	-	-
	G248P83013G3	WI2-1090N5	12q24.33	chr12:133,332,740-133,372,365	12, der(9)	-	-	-	-
CHFR	RP11-46H11	AC127070.10	12q24.33	chr12:133,347,391-133,526,403	12, der(9)	-	-	-	-
	RP11-637F20	/	12q24.33	chr12:133,465,282-133,613,019	12, der(9)	-	-	-	-

/ BACs mapped by end sequencing; - not analyzed; * clones showing a significantly fainter signal intensity on the derivative chromosome than on the normal homolog; ** BACs pooled together and used as a single probe in FISH experiments

splitting signals
translocated or deleted
retained

Supplementary Table S4. Primers used in RT-PCR in RT-qPCR analyses.

Primer RT-PCR	**Sequence**
PAX5-A (ex1)	ATGGATTTAGAGAAAAATTATCCGACT
PAX5-B (ex3)	GTGCCTAGCGTCAGTTCCAT
PAX5-R (ex5)	CTTGCGCTTGTTGGTGTCGG
UAP (RACE kit)	CUACUACUACUAGGCCACGCGTCGACTAGTAC
AUAP (RACE kit)	GGCCACGCGTCGACTAGTAC
AAP (RACE kit)	GGCCACGCGTCGACTAGTACGGGIIGGGIIGGGIIG
AUTS2-A (ex6)	GATGCCTTCGTCTCTCTTGC
AUTS2-B (ex7)	ACCACAGGCTCAAAGATTGG
AUTS2-C (ex7)	TACTCAGAGGTGGGGGTTGA
AUTS2-D (ex8)	TGAGTCTTCGCTGGAGTGC
SOX5ex5 - primer A	TTGTTCTTGTTGCTGCTTGG
SOX5ex7 - primer B	GGAGGGAATACGGGAATCAT
POM121Cex6R	CACCAGGGGCTCAAATGCTGA
POM121Cex13R	GCAGGCAGGGTAAAGGTAAATG
CHFR1829-L (exons 16-17)	CCGAGTTGCCAGTGGCCGTAA
CHFR2720-R (exon 18)	GGGCCGCGGTCACTCACAC
Sequence CH-029 - primer exon19L	GCACGCAGAATAGTGCTGGGCT
Sequence CH-029 - primer exon20R	ACGTTTCCTCGCTGCACGATAC
MLLT3 (ex5)	GTTCCTTGAAGGCCATCTTAGGAAC
RT-PCR product length	
PAX5-A + PAX5-R =594bp	
PAX5-B + PAX5-R =210bp	
PAX5-A + AUTS2-A = 609bp	
PAX5-A + AUTS2-B = 795bp	
PAX5-A + AUTS2-C = 929bp	
PAX5-A + AUTS2-D = 1161bp	
CHFR1829L + CHFR2720R =912bp	
CHFR1829L + primer XR =340bp	
Sequence CH-029: exon19L + exon20R=115bp	
PAX5-A + SOX5ex5 = 953bp	
PAX5-A + SOX5ex7 = 1066bp	
PAX5-B + SOX5ex5 = 569bp	
PAX5-B + SOX5ex7 = 682bp	
PAX5-A + POM121Cex6R = 759bp	
PAX5-B + MLLT3ex5 = 1078 bp	
Primer RQ-PCR	**Sequence**
Wild-Type_transcript_CH-001 CHFR_L	GTTTTCACAGCCCCCTGAG
Wild-Type_transcript_CH-001 CHFR_R	AGAAGAGTCACCCCAGAGCA
Novel_transcript_CH-029 CHFR_L	TCAAAAACTAAGCATCCAGAGG
Novel_transcript_CH-029 CHFR_R	GCGAGCCCAGCACTATTCT
GAPDH left	TGCACCACCAACTGCTTAGC
GAPDH right	GGCATGGACTGTGGTCATGAG

Supplementary Table S5. Copy Number Variation analysis of the *PAX5* rearranged cases.

Chr	Type	From	To	Start	End	Genes	sno/miRNA
PT5 PAX5/CHFR							
5	LOH	q23.3	q31.1	128181233	131358999	*SLC27A6, ISOC1, ADAMTS19, CHSY3, HINT1, LYRM7, CDC42SE2, RAPGEF6, FNIP1, ACSL6*	-----
7	Mosaicism/ Loss	p22.3	p14.1	0	38372423	Deletion from TARP included (in homozygosis) towards the telomere (about 38MB deletion)	*hsa-mir-339, hsa-mir-589, hsa-mir-1302-6, hsa-mir-3146, hsa-mir-1183, HBII-336, hsa-mir-148a, hsa-mir-196b, hsa-mir-550-1, hsa-mir-550-2, hsa-mir-548n*
7	Mosaicism/ Gain	q11.2	q34	71137981	142398377	From *WBSCR17* to *MTRNR2L6*	*hsa-mir-4284, hsa-mir-590, hsa-mir-1285-1, hsa-mir-653, hsa-mir-489, hsa-mir-591, hsa-mir-25, hsa-mir-93, hsa-mir-106b, hsa-mir-4285, hsa-mir-5480, hsa-mir-592, hsa-mir-593, hsa-mir-129-1, hsa-mir-182, hsa-mir-96, hsa-mir-183, hsa-mir-335, hsa-mir-29a, hsa-mir-29b-1, hsa-mir-490*
7	Mosaicism/ Loss	q34	q36.3	142398377	159118443	PRSS2, PRSS1, TRY6 towards the telomere (about 16MB deletion)	*hsa-mir-548f-4, hsa-mir-1975, hsa-mir-671, hsa-mir-153-2, hsa-mir-595*
14	Loss	q11.2	q11.2	22851730	22957488	*TCRA*	
PT7 PAX5/AUTS2							
8	Gain	q24.21	q24.21	130444596	130675632	-----	-----
9	Loss	p24.3	p13.2	209111	36807359	**From pter to *PAX5*. *CDKN2A* homozygously deleted.**	*hsa-mir-101-2, hsa-mir-3152, U92, hsa-mir-491, hsa-mir-31, hsa-mir-876, hsa-mir-873, SNORD121B, SNORD121A*
14	Loss	q11.2	q11.2	22892460	22980283	*TCRA*	-----
PT10 PAX5/AUTS2							
3	Loss	q26.32	q26.32	176682276	177009190	*TBL1XR1*	-----
7	Loss	p22.3	q11.22	0	69674010	**from pter to AUTS2 (IKZF1 included) (from 53288671 to 66696241 CN=2)**	hsa-mir-589, hsa-mir-1302-6, hsa-mir-3146, hsa-mir-1183, HBII-336, hsa-mir-148a, hsa-mir-196b, hsa-mir-550-1, hsa-mir-550-2, hsa-mir-548n, hsa-mir-1200, ACA9, ACA5, ACA5c, ACA5b
9	Loss	p24.3	p13.2	209111	36950390	**From *pter* to *PAX5* (*CDKN2A* and *MLLT3* included)**	hsa-mir-101-2, hsa-mir-3152, U92, hsa-mir-491, hsa-mir-31, hsa-mir-876, hsa-mir-873, SNORD121B, SNORD121A
14	Loss	q11.2	q11.2	22910936	22980283	***TCRA***	-----
PT15 PAX5/SOX5							
7	Loss	p14.1	p14.1	38305834	38407409	*TARP*	-----
8	Loss	q12.1	q12.1	60037057	60512064	From 60037057 to 60249036 (homozygous deletion)	-----
9	Loss	p24.3	p13.2	209111	36970472	**From *pter* to *PAX5*. From *MLLT3* to *CDKN2B-AS* (19.932.623-22.046.390) and *MOBKL2B*: homozygous deletion.**	*hsa-mir-101-2, hsa-mir-3152, U92, hsa-mir-491, hsa-mir-31, hsa-mir-876, hsa-mir-873, SNORD121B, SNORD121A*
12	Loss	p13.33	p12.1	192042	24324950	**From *FAM138D* to *SOX5* (TEL deletion included).** Except copy number=2 in region p12.1, from 23.702.993 to 23.957.694)	*hsa-mir-613, hsa-mir-614*
14	Loss	q11.2	q11.2	22873308	22957488	*TCRA, TCRD,TCRVD2,TRA@*	-----
PT19 PAX5/POM121C							
2	Loss	q14.3	q14.3	122641895	127139214	*CNTNAP5*	-----
2	Loss	q21.1	q21.1	131351694	136629632	from *CFC1* to *MCM6*	hsa-mir-663b, hsa-mir-128-1
2	Loss	q34	q36.3	211278282	230759606	from *LANCL1* to *TRIP12*	*hsa-mir-548f-2, hsa-mir-26b, hsa-mir-375, hsa-mir-3131, hsa-mir-153-1, hsa-mir-3132, hsa-mir-4268*
6	Loss	q22.1	q22.1	114998398	115659458	-----	-----
7	Loss	p21.2	p21.2	14570721	15336102	*DGKB, TMEM195*	-----
7	Loss	p14.1	p14.1	38288583	38372423	*TARP*	-----
7	Loss	q11.23	q22.1	75073337	159118443	**From *POM121C* to qter**	ACA14a, hsa-mir-1285-1, hsa-mir-653, hsa-mir-489, hsa-mir-591, hsa-mir-25, hsa-mir-93, hsa-mir-106b, hsa-mir-4285, hsa-mir-548o, hsa-mir-592, hsa-mir-593, hsa-mir-129-1, hsa-mir-182, hsa-mir-96, hsa-mir-183, hsa-mir-335, hsa-mir-29a, hsa-mir-29b-1, hsa-mir-490, hsa-mir-548f-4, hsa-mir-1975, hsa-mir-671
9	Loss	p24.3	p13.2	209111	36962346	**From *PAX5* to pter. Homozygously deleted at: *CDKN2A, CDKN2B, CDKN2B-AS, DMRTA1, ELAVL2, TUSC1*. Homozygously deleted from 26409648 to 28005524, no genes.**	hsa-mir-101-2, hsa-mir-3152, U92, hsa-mir-491, hsa-mir-31, hsa-mir-876, hsa-mir-873, SNORD121B, SNORD121A
9	Gain	p13.2	q32	36977143	115815714	From *PAX5* to *ZFP37*	hsa-mir-1299, hsa-mir-204, hsa-mir-7-1, hsa-mir-4289, hsa-mir-3153, hsa-mir-4290, SNORA84, hsa-mir-4291, hsa-let-7a-1, hsa-let-7f-1, hsa-let-7d, hsa-mir-2278, hsa-mir-23b, hsa-mir-27b, hsa-mir-3074, hsa-mir-24-1, hsa-mir-1302-8, hsa-mir-32
14	Loss	q11.2	q11.2	22587678	22978470	*TCRA*	-----

Grey = specific IG and/or TCR somatic rearrangements

Bold = Translocation breakpoint

LOH = Copy number neutral Loss Of Heterozygosity