Supporting Information

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HCC1954 HCC1954Blood

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Fig. 52. Chimeric cDNA validation. The order of the chimeras is as the same as in Table 1, also as listed below. For each chimera, the left lane1 is HCC1954, right lane is HCC1954 blood. L stands for ladder in base pairs. Positive control used MRE11A wild-type cDNA fragment. 1, t(5;8)(q35.3;q24.21). 2, t(5;8)(p15.33;q24.21); 3, t(5;8)(q23.1;q23.1); 4, t(4;11)(q32;q21); 5, t(9;18)(p24.1;q12.2); 6, t(8;2)(q24.12;q22.1); 7, intronic inversion between 8q24.12 and 8q24.22.







Fig. 54. Genomic PCR-walking to amplify the t(4;11)(q32;q21) break junction. Forward primer: 1, TCT TCG CTT TAG CCA GAA AT (in MREA11exon11); 2', GAA GTA GAA CCA TTA CAT TTG TC (in intergenic region of chr4). Reverse primers (in intergenic region of chr4): 2, GAC AAA TGT AAT GGT TCT ACT TC; 3, CAA GGT AAT TTA CAG ATT CAA TG; 4, AGT GAC AAG AAA GAT TTC CAG ATG; 5, AGG AAA GAG CTT GTG AGT GG; 6, TTC CAT TGC ATT TCT GAC AAC TGC; 7, TCT TGC ACA TTC CCA GAG ACT; 8, TCT TGC ACA TTC CCA GAG ACT. Control lane is from Takara kit, a 17,000-bp fragment. Interchromosomal break of t(4;11)(q32;q31) was speculated to occur in intron 11 of MRE11A and an intergenic region on chromosome 4. A forward primer in MRE11A exon11 was paired up with serials of reverse primers in the intergenic region on chromosome 4 to amplify genomic break junction fragments by long-range PCR. We were able to amplify a series of genomic DNA fragments from the break junction specifically in HCC1954. The shortest fragment amplified in LR-PCR, which was \approx 9 to 10 kb by primer pair (1+4), was subsequently shotgun sequenced with the 454 GS FLX technology. The fine assembly and mapping of this 9-kb fragment was resolved as described in Fig. 4.

Table S1. Chimeric cDNA identified in 454 reads and validated by RT-PCR

Proposed				
rearrangements	Chromosomes locations	Genes affected	Effect on coding	Uniqueness
	I	nter-chromosome		
intragenic to intergenic	t(5;8)(q35.3;q24.21)	NSD1	Truncation	HCC1954 only
intragenic to intragenic	t(5;8)(p15.33;q24.21)	CLPTM1L and PVT1*	Truncation	HCC1954 only
intragenic to intergenic	t(5;8)(q23.1;q23.1)	EIF3E*	Truncation	HCC1954 only
intragenic to intergenic	t(4;11)(q32;q21)	MRE11A	Truncation	HCC1954 only
intragenic to intragenic	t(9;18)(p24.1;q12.2)	PDCD1LG2 and C18orf10	Chimeric protein	HCC1954 only
intragenic to intragenic	t(8;12)(p11.21;q12)	SLC20A2 and DBX2*	Truncation	HCC1954 only
Intragenic to intergenic	t(8;2)(q24.12;q22.1)	SAMD12	Chimeric protein	HCC1954 only
		Intrachromosome		
Tandem duplication	5q14.1	JMY	Extension	HCC1954 and HCC1954BL
Tandem duplication	6q24.2	UTRN	Truncation	HCC1954 and HCC1954BL
Inversion	8q24.12: 8q24.22	SAMD12 and PHF20L1	Truncation	HCC1954 only
Tandem duplication	17q12	LCRG1	Extension	HCC1954 and HCC1954BL
Inversion	21q21.3: 21q22.11	ZNF294 and TIAM1*	Truncation	HCC1954 and HCC1954BL
Translocation	6q26	MAP3K4	Truncation	HCC1954 and HCC1954BL
Tandem duplication	7p15	MPP6	Extension	HCC1954 and HCC1954BL
Translocation	21q22.11: 21q22.12	IFNGR2 and RUNX1	Truncation	HCC1954 and HCC1954BL
Translocation	7q21–22	AKAP9 (Yotiao)	Truncation	HCC1954 and HCC1954BL
Translocation	8q24.12: 8q24.21	SAMD12 and PVT1	Truncation	HCC1954 and HCC1954BL
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* Chimeric events identified by additional screening algorithm (see paper text).

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Table S2. Coverage of 454 reads at the break junctions of chimeric and wild-type transcripts

Genes involved in the chimeras	No. 454 reads traverse the wild-type junction for the first gene involved	No. 454 reads traverse the wild-type junction for the second gene involved	No. 454 reads traverse the chimeric junction
NSD1	5	N/A	10
PVT1 and CLPTM1L	4	1	2
EIF3E	14	N/A	27
MRE11A	6	N/A	4
PDCD1LG2 and C18orf10	0	4	1
SLC20A2 and DBX2	1	0	3
SAMD12	21	N/A	6
PHF20L1 and SAMD12	11	N/A	10

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