Recurrent loss of the Y chromosome and homozygous deletions within the pseudoautosomal region 1: association with male predominance in mantle cell lymphoma

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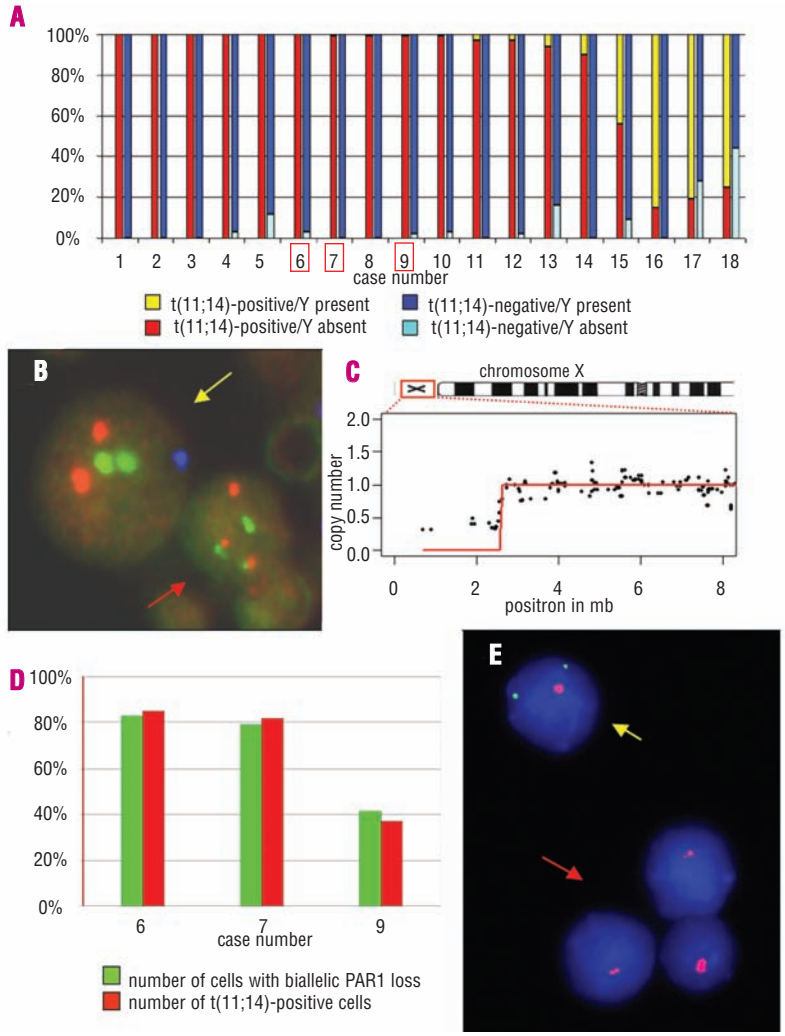

Supplementary Figure 1. (A) Percentages of cells with chromosome Y loss in both the t(11;14)(q13;q32)-postive MCL tumor cells (left column) and the t(11;14)(q13;q32)-negative normal cells (right column) of 18 analyzed by FISH primary MCL with chromosome Y loss. In each case, 100 tumor cells and 100 normal cells were evaluated by FISH. MCL showing loss in Xp22.33 are marked by a red box. (B) FISH on interphases of MCL 9 using the LSI IGH/CCND1 dual probe labeled in red/green (Abbott/Vysis) in combination with a probe for Yq12 labeled in blue (CEP Y, spectrum aqua, Abbott/Vysis). Colocalization of red and green signals indicates a MCL nucleus (red arrow) harboring a translocation t(11;14)(q13;q32). A blue signal indicating the presence of chromosome Y was only detectable in the nuclei of t(11;14)-negative cells (yellow arrow). (C) GeneChip copy number data of MCL 6 showing a homozygous deletion in the pseudoautosomal region 1 (PAR1) in Xp22.33. Copy number analysis was performed using the CNAG program (v2.0). Black dots represent copy number estimators of the SNP probes smoothed by a median filter of size n=3. The red line marks the result of the segmentation process. The profile of genomic imbalances is shown from Xp telomere (left) to 8Mb (right) with a value of 1 indicating the presence of one X chromosome. The tumor cell content of this MCL was 85%.

(D) Percentages of cells with biallelic PAR1 loss (green) compared with percentages of t(11;14)-positive cells (red) in the 3 MCL showing homozygous PAR1 deletion. Percentages for both alterations were determined in separate hybridization experiments evaluating 100 cells each. (E) FISH on interphases of MCL 7 using a locus-specific probe for the PAR1. The PAC clone RP4-674K6 targeting the *CSF2RA* locus in Xp22.33 was labeled in spectrum green and two pooled BAC clones RP13-156P1 and RP11-296N8 targeting a control locus in Xq28 were labeled in spectrum orange. In cells with PAR1 deletion, no green signal was detectable (red arrow). The cell marked by a yellow arrow showed two green signals indicating the presence of both X-chromosomal and Y-chromosomal PAR1. False color display using the ISIS software version 5.1.9 (Metasystems, Altlussheim, Germany).

Supplementary Table 1. **Genes mapping into PAR1 and BAC/PAC clones used for FISH validation.**

Gene symbol	*Gene name*	*Physical position [bp] * at chromosomes X and Y*	*BAC/PAC clones*
PLCXD1	phosphatidylinositol-specific phospholipase C	132,992-160,020	
GTPBP6	GTP binding protein like	161,426-170,887	
PPP2R3B	protein phosphatase 2, regulatory subunit B	264,970-272,590	
SHOX	short stature homeobox	555,079-590,146	RP11-800K15
ATRX	α thalassemia/mental retardation syndrome X-linked	772,383-982,187	
PGAM4	phosphoglycerate mutase family 3	1,324,940-1,326,617	
CSF2RA	colony stimulating factor 2 receptor, α	1,355,173-1,396,262	RP4-674K6
TAF9B	transcription associated factor 9B	1,383,007-1,393,032	
IL3RA	interleukin 3 receptor, α precursor	1,499,188-1,545,408	
SLC25A6	solute carrier family 25, member A6	1,548,872-1,554,821	
ASMTL	acetylserotonin O-methyltransferase-like	1,565,861-1,615,583	
P2RY8	G-protein coupled purinergic receptor P2Y8	1,625,295-1,699,827	
CRLF2	cytokine receptor-like factor 2	1,672,024-1,687,565	RP11-475E20
SFRS17A	splicing factor, arginine/serine-rich 17A	1,754,313-1,765,236	
ASMT	acetylserotonin O-methyltransferase	1,758,175-1,805,801	
ZBED1	Ac-like transposable element	2,397,816-2,412,369	

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