

SUPPLEMENTARY MATERIAL 2

Chimeric transcripts resulting from complex duplications in chromosome Xq28

Human Genetics

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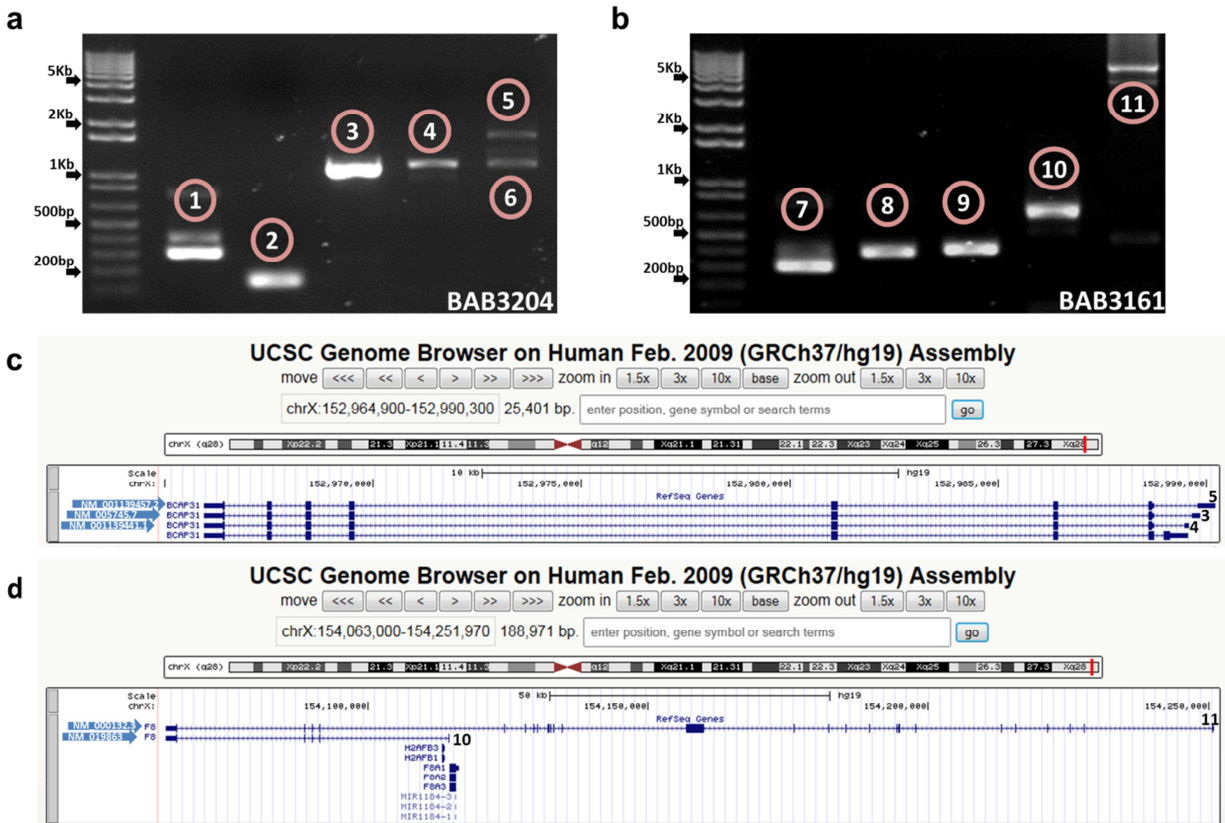
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Supplementary Fig. 1 Chimeric transcripts from patients with Xq28 duplication. Expression of the predicted chimeric genes in the LCLs derived from patients BAB3204 and BAB3161 were confirmed using RT-PCR. (a) RT-PCR results from patient BAB3204. The bands in the agarose gel represent: 1- Size: 305 bp represents RT-PCR positive control (*KDMC4*); 2- Size: 137 bp represents RT-PCR band obtained using primers that target exons flanking genomic breakpoint junction. Bands 3-5 correspond to RT-PCR of three *BCAP31* fusion transcripts: 3- NM_005745.7, 1078 bp; 4- NM_001139441.1, 1112 bp; 5- NM_001139457.2, 1619 bp. 6- Size: 1463 bp represents an unexpected alternative splicing transcript for *BCAP31*. (b) RT-PCR results from patient BAB3161. The bands in the agarose gel represent: 7- Size: 305 bp represents RT-PCR positive control (*KDMC4*); 8- Size: 386 bp represents RT-PCR band obtained using primers that target exons flanking genomic breakpoint junction of *F8* transcript II (short, NM_019863.2); 9- Size: 395 bp represents RT-PCR band obtained using primers that target exons flanking genomic breakpoint junction of transcript I (long, NM_000132.3); 10- Size: 721 bp represents *F8* fusion transcript II; 11- Size: 7695 bp represents *F8* fusion transcript I. Splicing variant transcripts in the RefSeq database for *BCAP31* (c) and *F8* genes (d). Numbers shown on the right correspond to the RT-PCR bands obtained from BAB3204 (a) and BAB3161 (b)