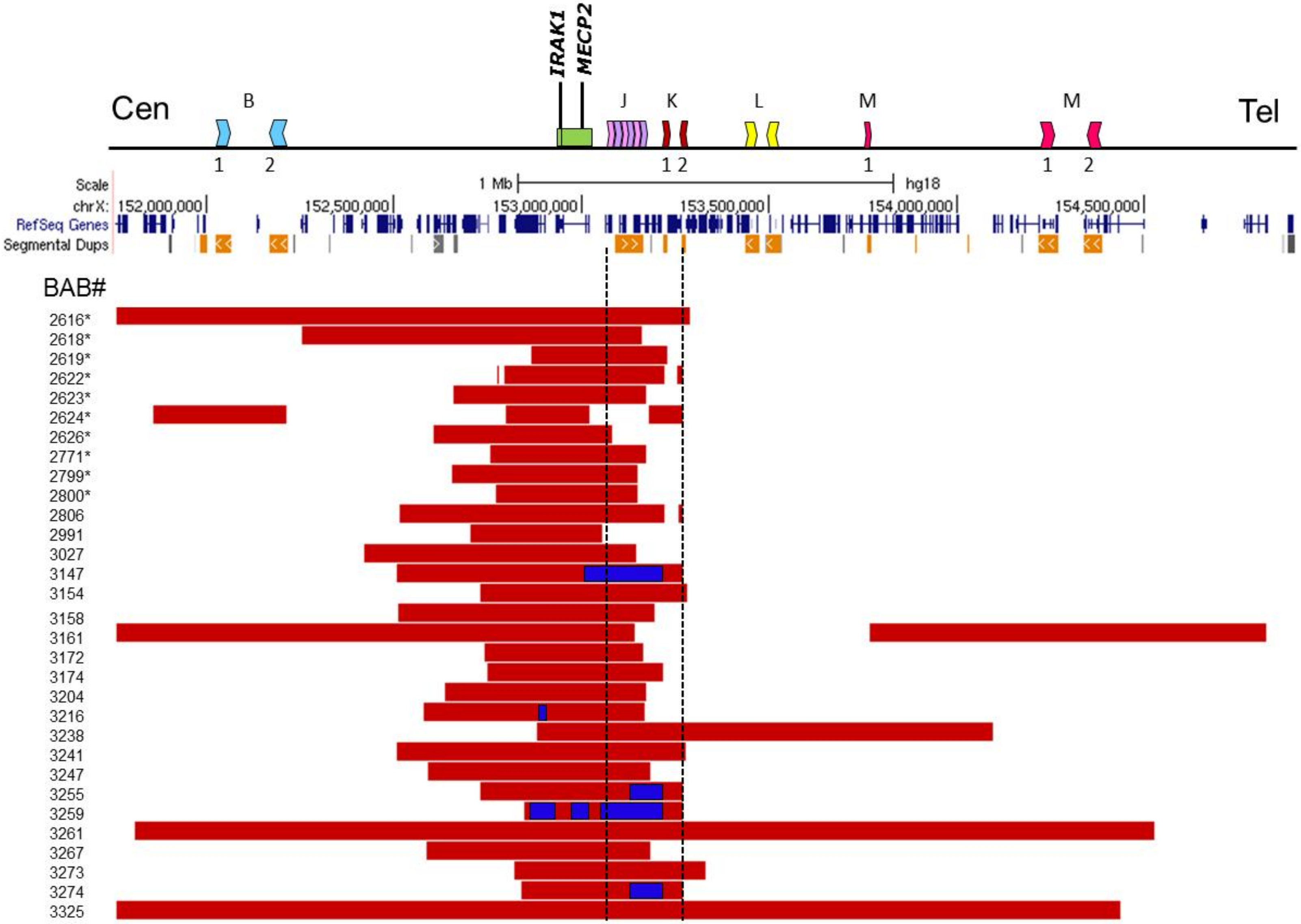
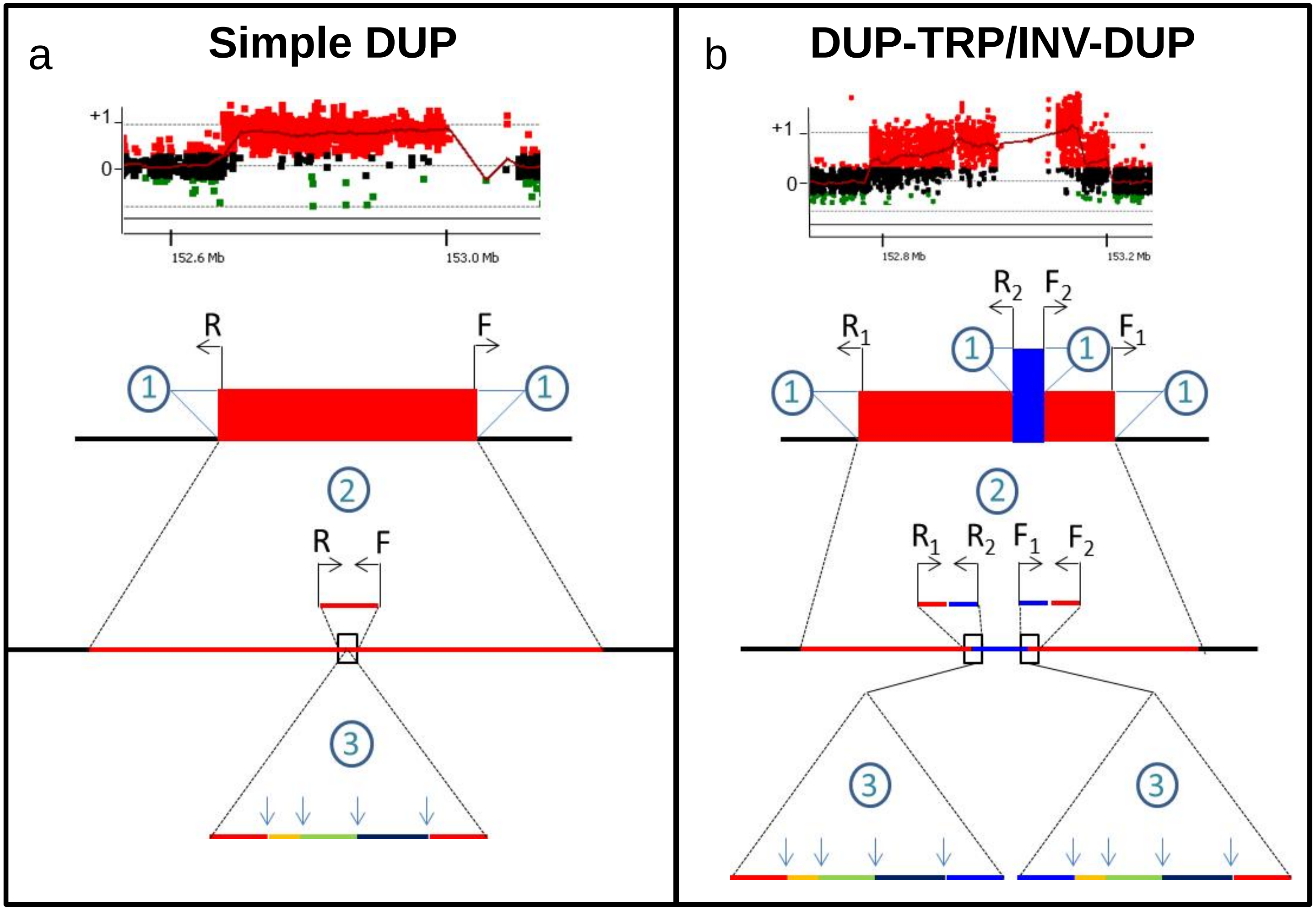
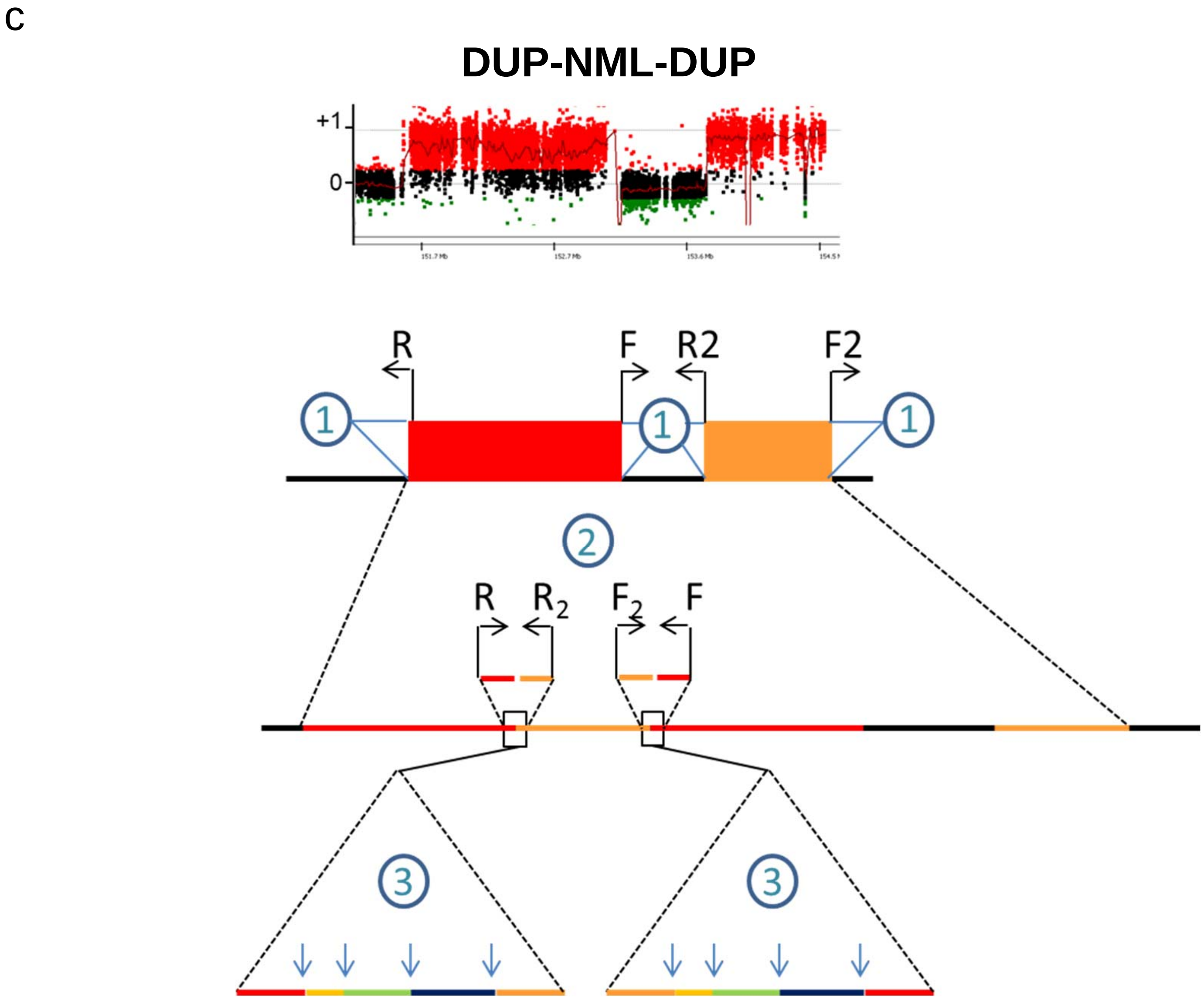
Supplementary Figure 1: Graphic representation of the duplicated region at Xq28 in each one of the 31 samples as revealed by aCGH. Duplications are represented in red and triplications in blue. Top: Genomic region harboring the alterations involving *MECP2*. UCSC genes and segmental duplication tracks are shown underneath the position track which is relative to the NCBI Build 36 for the X chromosome.
The smallest region of overlap (SRO), delimited by black dashed lines, includes two genes, *IRAK1* and *MECP2* that are shown in green boxes. Selected segmental duplications or low copy-repeats (LCRs) are shown as color-matched arrows that reflect their orientation in respect to their LCR pair. Asterisks indicate patients for whom aCGH data was previously published[1].



Supplementary Figure 2: Graphical general representation of the strategy used to obtain breakpoint junctions in patients with (a) an apparent simple duplication, (b) DUP-TRP/INV-DUP rearrangement, (c) DUP-NML-DUP rearrangement. F and R: outward facing primers; #1: copy-number transition state obtained by high-resolution aCGH; #2: breakpoint junction product obtained by long-range PCR using outward facing primer pairs; #3: template switching events, indicated by blue arrows, consist of short segments inserted or deleted at the breakpoint junctions. They can be obtained by sequencing long-range PCR products #2. DUP: duplication; DUP-TRP/INV-DUP: duplication-inverted triplication-duplication; DUP-NML-DUP: duplication-normal copy number-duplication.

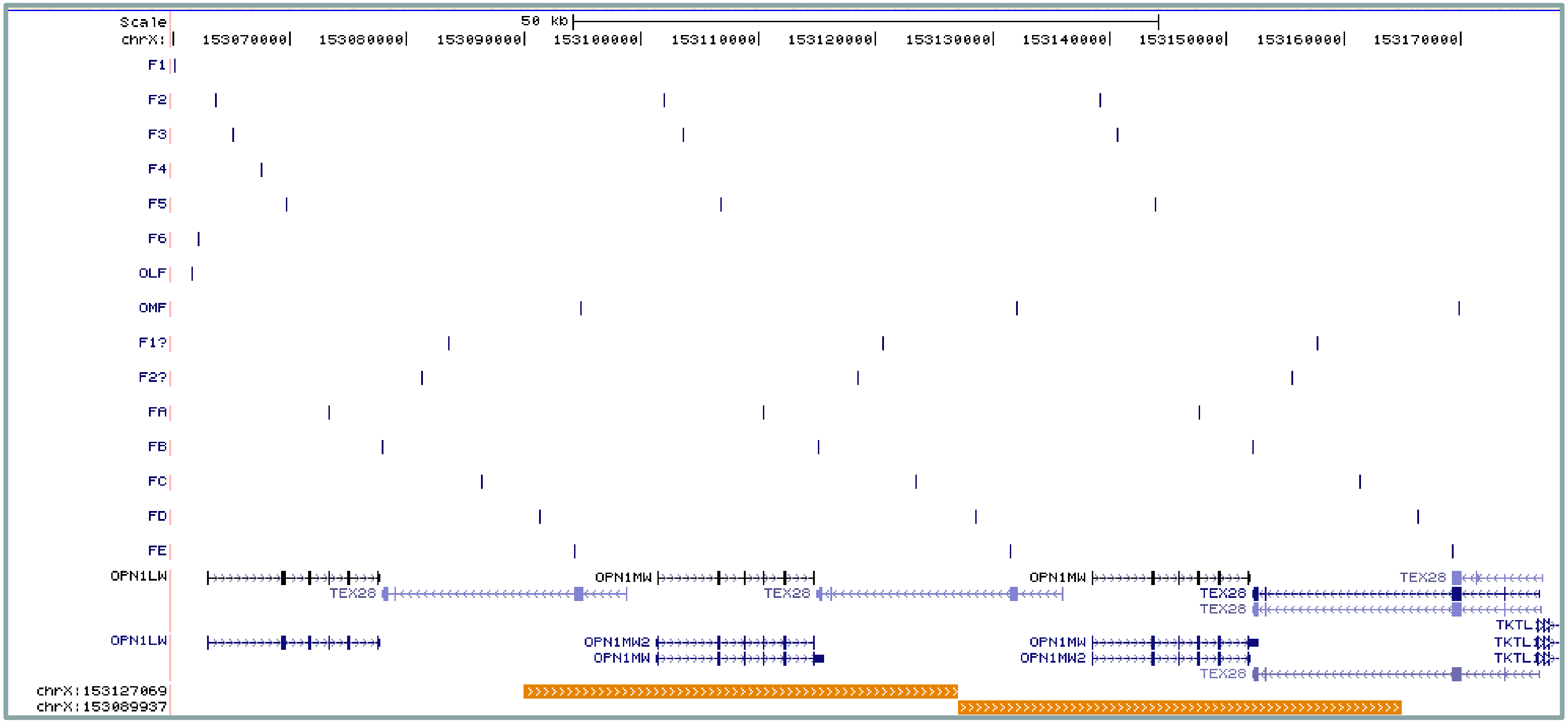




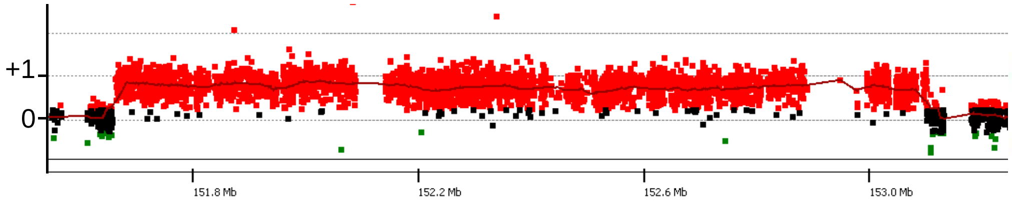
C

DUP-NML-DUP

Supplementary Figure 3: Relative location of primers spanning LCRJ (Opsin panel). UCSC genes and segmental duplication tracks are shown underneath the position track which is relative to the NCBI Build 36 for the X chromosome.



Supplementary Figure 4: Rearrangements without evidence for multiple template-switches. For each patient aCGH result along with the breakpoint junction sequences obtained by long-range PCR and Sanger sequencing are shown. Breakpoint junction sequence is aligned to the proximal and distal genomic references and color-matched. Strand of alignment (+ or -) is indicated in parenthesis. Microhomology at the breakpoint is indicated by black bold underlined letters.



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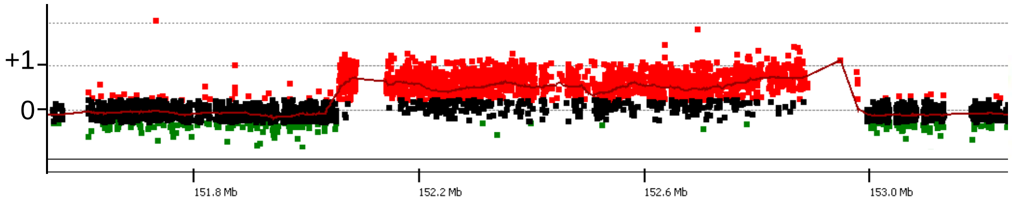
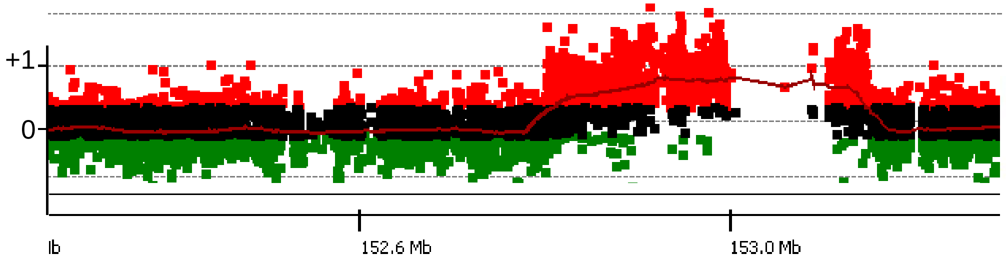
BAB2616	GCCCAGGCCTACCTTGCTCTGCCA**GTG**TCTATTTGATTCTTCTCTCTTTTCTTC
R1_intron_*NSDHL* (+)	ATATCTCCTATATCATTTTTTATT**GTG**TCTATTTGATTCTTCTCTCTTTTCTTC



Fb_intron_*TEX28C* (+)	GTGGCCAGGATGGATGAGCTCACTCT**GA**GCAAATGTGCCAAAGCAAGGGGCCCTG
BAB2618	GTGGCCAGGATGGATGAGCTCACTCT**GA**CTTATTATAACATTTTATTATATAAAA
R3_intergenic (+)	AGTGAAAGGAACCAAACACAAAAGAA**GA**CTTATTATAACATTTTATTATATAAAA



F4_LCR_K1 (+)	GCCTGGACGGGGACTGTGTCCTCCCT**CCCC**GGGCTGCCCGGCGCCCTCGCTCAGGGAGTG
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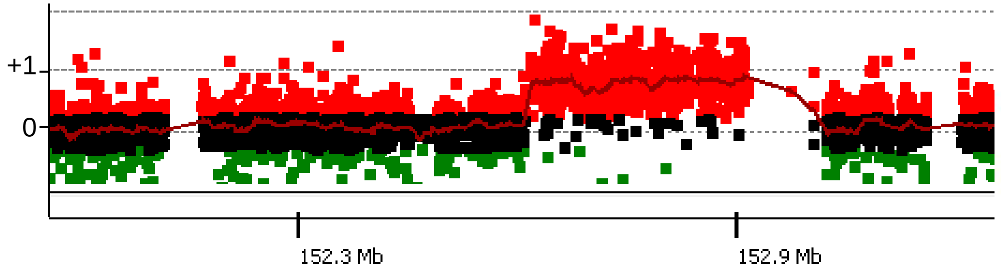

*Alu*Sc

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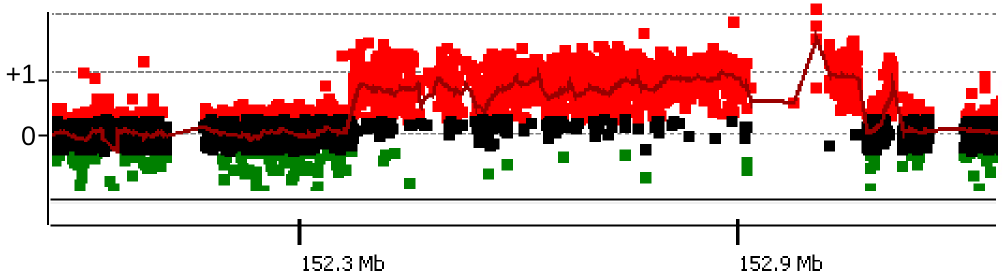
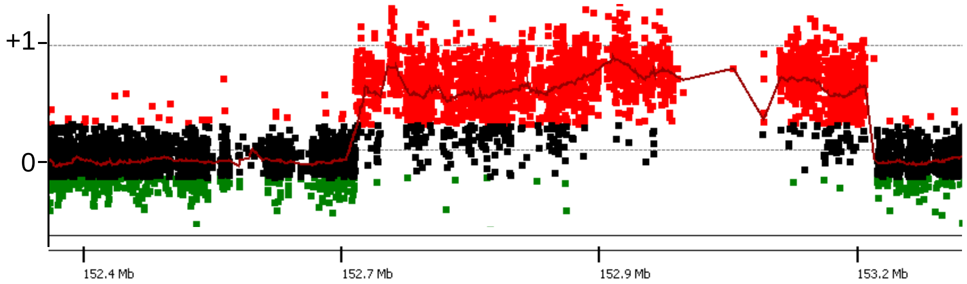
R1_intergenic (+)	CTACTCaGGAGGCTGAGGCAGGAGAATt**GCTTGAACCCGGGAGG**caGAaGTTGCAGTGaG

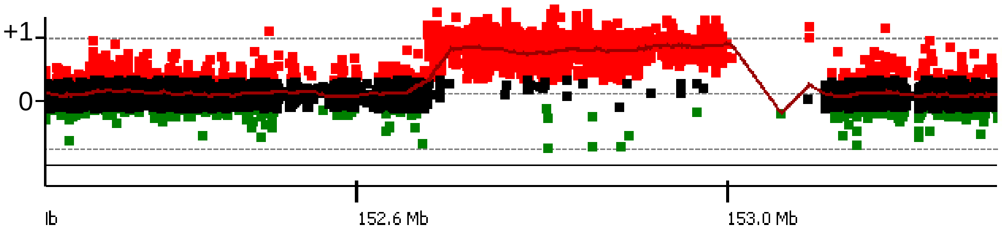
*Alu*Sg



R3_intergenic (-)	GCAGCCTCAAACTCCCGGGCTTAAACGATCCT**CC**CATCTGTCTCCTGAGCAGCTGAGACT
BAB2806	GCAGCCTCAAACTCCCGGGCTTAAACGATCCT**CC**TGCACCACCTGCACACTGCCTTGGGG
R2_intron_ *FAM58A* (+)	GAGCGTGGGGCAACACTGTCTACACAGATGGC**CC**TGCACCACCTGCACACTGCCTTGGGG



F2_intergenic (+)	CCAGCTAATTTTTGTATTTTTAGTAGAGACGGGGTTTCACCATGTTGGCCAGGCTGGT
BAB3154	CCAGCTAATTTTTGTATTTTTAGTAGAGAGG-CGACTCACAGCTCAGAGAGGACGTAC
R1_intron_*PDZD4* (+)	TCTGGAGGCAGGGCCCGGGGCCCTCCTGAGGGCGACTCACAGCTCAGAGAGGACGTAC
	*



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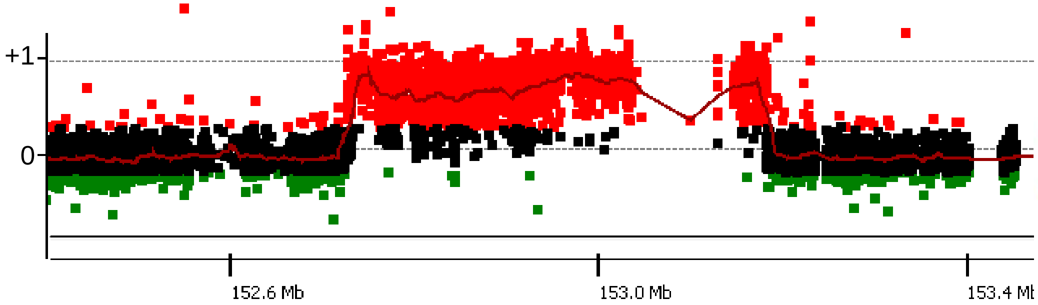

F_intergenic (+)	GGTCAGTTCCTAAATAAGGAAAGAGCTTTTATCCAAACCAGAAAGAGTGAGCCATGGC
BAB3174	GGTCAGTTCCTAAATAAGGAAAGAGCTTTGTGGATTGTAGATGGAAGGCGCTGCAGAT
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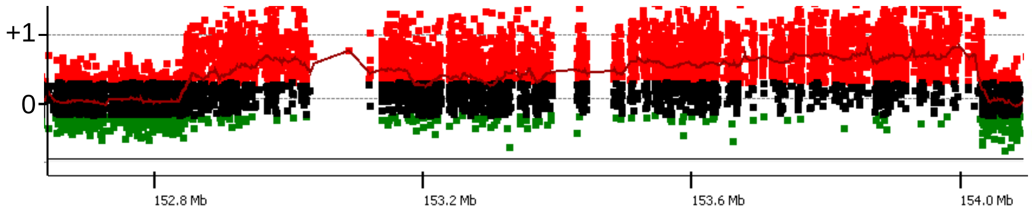

F_intergenic (+)	ATCTGTCTCTCATTCAGACGCAGTCCAAGTAGAGGTCCCAAAGCAAAAAGATTCAGAT
BAB3238	ATCTGTCTCTCATTCAGACGCAGTCCAAGTAGAGTGGTACCCGCTGGGACACTCAGCA
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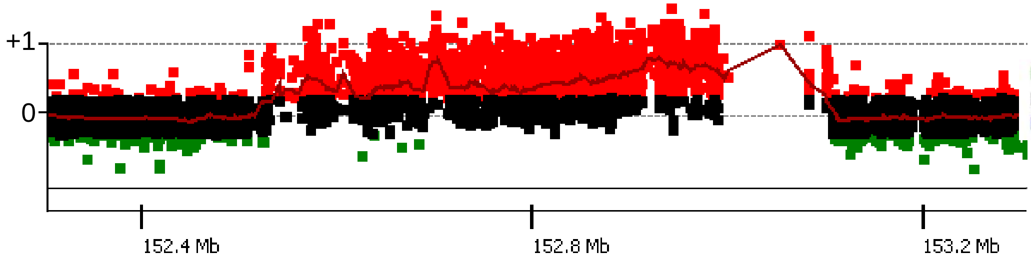

F2_intron_*TKTL1* (+)	CGAGATCAGCCTGGGCAAGATGGCAATCCCCTGTGCAGAATGTTACAAAAATTAGCTGG
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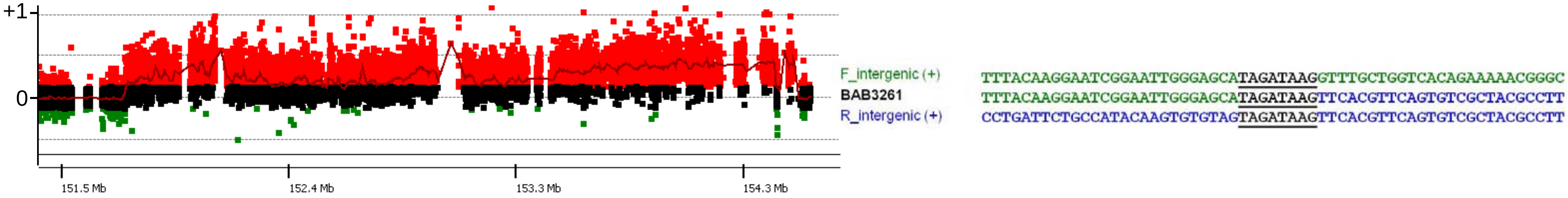
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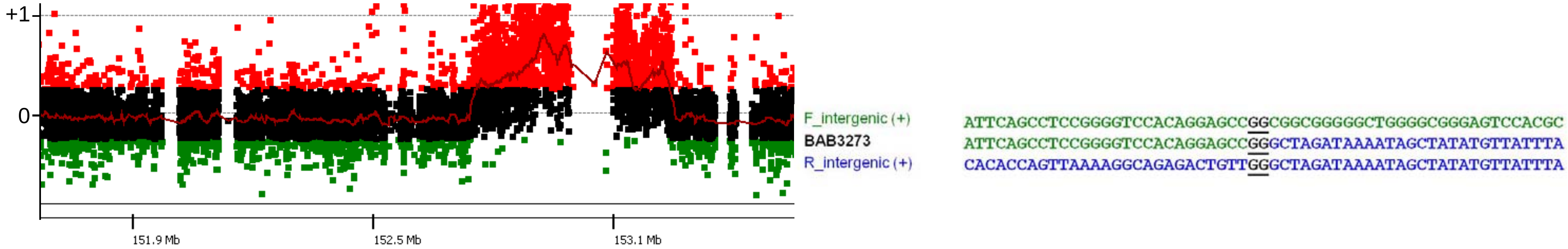
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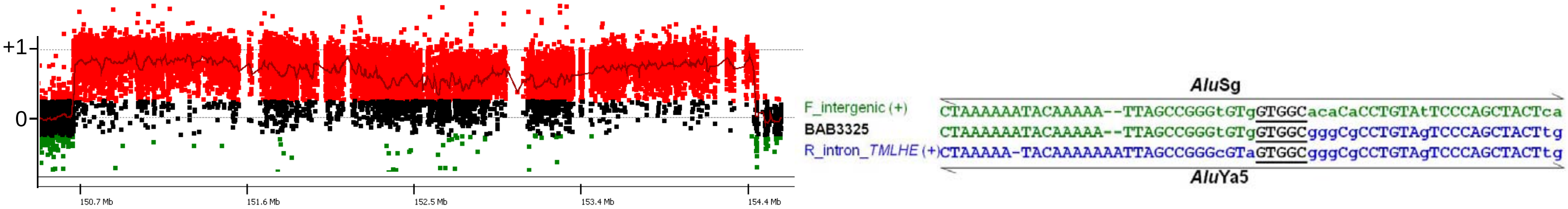
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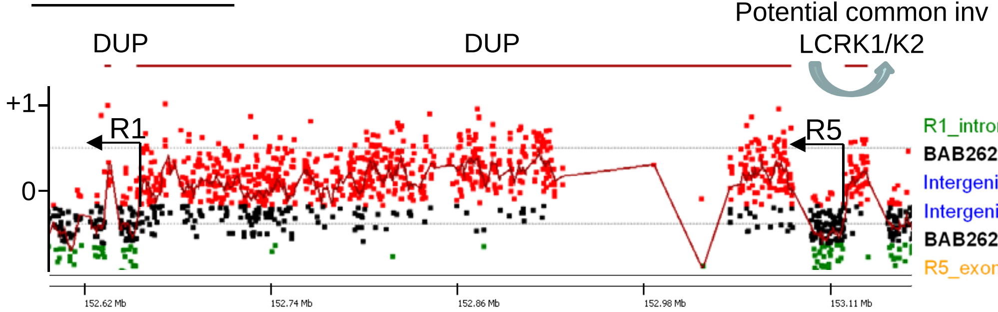
CTAAAAA-TACAAAAAAATTAGCCGGGcGTaGTGGCgggCgCCTGTAgTCCCAGCTACTtg
AluYa5

Supplementary Figure 5: Rearrangements with evidence for multiple template-switches. For each patient aCGH result along with the breakpoint junction sequences obtained by long-range PCR and Sanger sequencing are shown. For selected samples there is also a graphic representation of the genomic structure for the reference genome (top) and for the surmised genomic structure (bottom), showing predicted order, origins, and relative orientations of duplicated sequences. Breakpoint junction sequence is aligned to the proximal and distal genomic references and color-matched. Strand of alignment (+ or -) is indicated in parenthesis. Microhomology at the breakpoint is indicated by black bold underlined letters. Arrows show orientation of DNA sequence relative to the positive strand; filled arrows with circled numbers below represent a template switch that resulted in insertion or deletion of segments. Last arrow signifies resumption of replication on the original template which produced the CGR identified by aCGH. The series of events shown could also have occurred in the reverse order.

BAB2622



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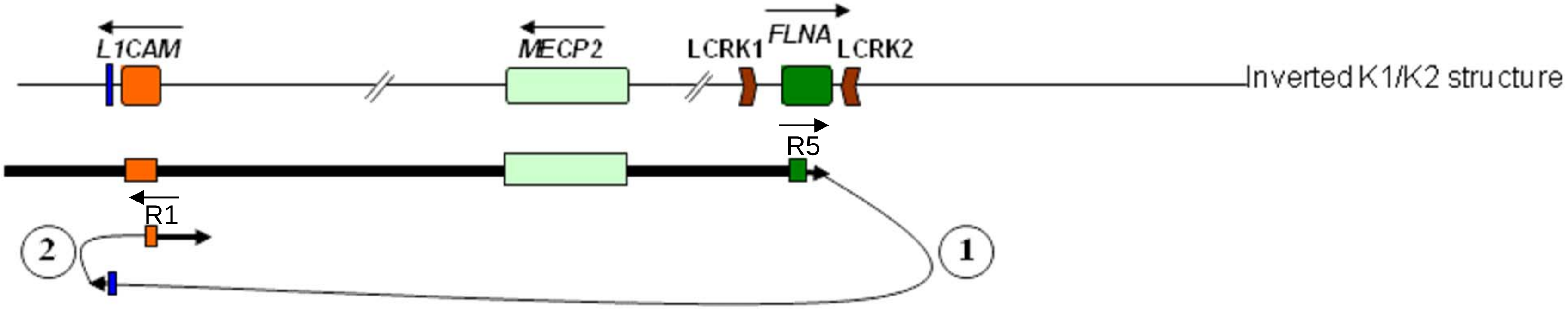
Intergenic (-)		TTTCTTTCGACAGAATCTCCTCTGTTGCCCAGACTGGAGTGCAGTGGGACAGTCATG
Intergenic (-)	.//.	TTAAAAAACACAAGGCTTAAAAAGTGCCAATATGGCTGGGCGCAGTGGCTCACGCC
BAB2622	.//.	TTAAAAAACACAAGGCTTAAAAAGTGCCAATATGGCTGGGACATGGCGGTGCCCAG
R5_exon_*L1CAM* (+)		CAGCGCCTCGCACCCTCGGCCATGAGCCGGATCTCATGGGACATGGCGGTGCCCAG



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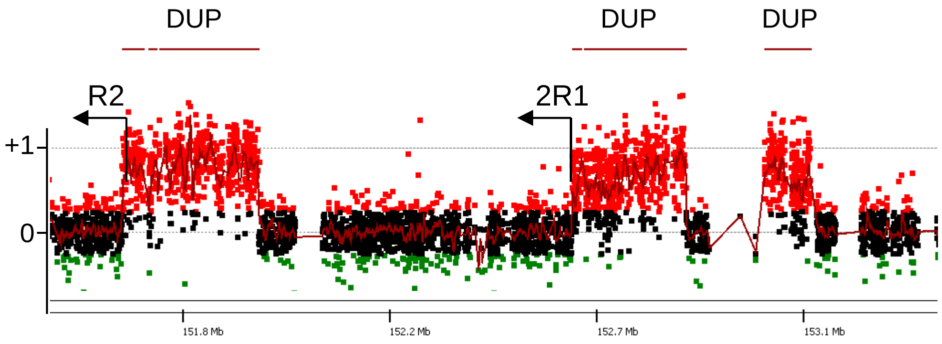
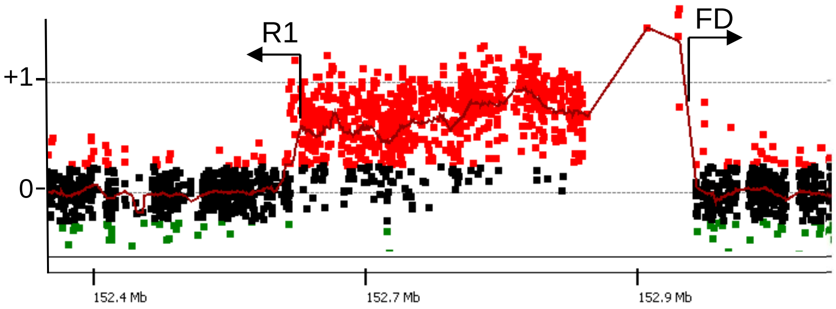


*Alu*Sg

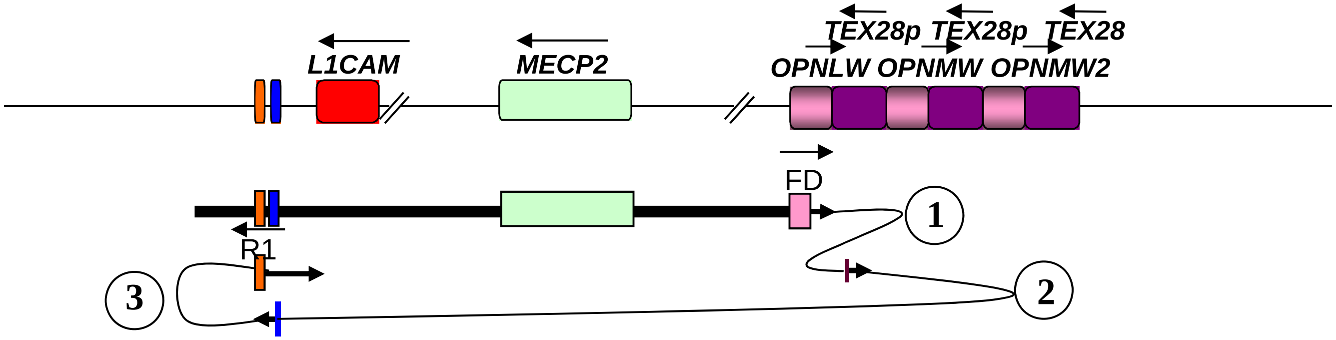
2R1_intergenic (-)	TCtGCCTCCTGgGTTCAaGtgATTcTCCTGCCTCAGCCTCCtgAGTAGCTGGGAtTAC
BAB2624	TCtGCCTCCTGgGTTCAaGtgATTcTCCTGCCTCAGCCTCCcaAGTAGCTGGGAcTAC
R2_intron_ *ZNF185* (+)	TCcGCCTCCTGaGTTCAtGccATTgTCCTGCCTCAGCCTCCcaAGTAGCTGGGAcTAC

*Alu*Y

BAB2800



FD_ intron_ *Opsin* (+)	CCCATCTCAGGAGAATTTTTGCAAACGCATAAAATGAGACCCATAGGATTACAAAGGC
BAB2800	CCCATCTCAGGAGAATTTTTGCATTTAGCACATGTGCCTTGGAGGAAAGGGGAGCACT
Intron_*TEX28* (+)	TTAAGGTGAACAATTCAGCGGCATTTAGCACATGTGGTGTTGTGCAACCACCATGGCT
Intergenic (-)	TTCTCATACAGACTGGTGCTACTTATTCTGCATTTGCCTTGGAAAGCTCTTGCTCTTG
R1_Intergenic (+)	GCATGGCAGGTCTCTCGCCAATAGGTTTACAGAAGAGTCAGGAGGAAAGGGGAGCACT



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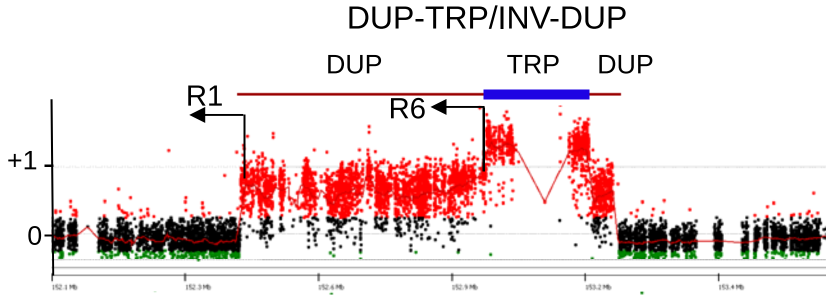
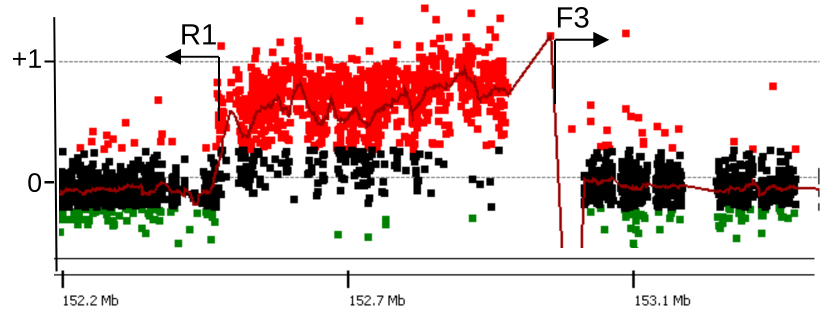


R6_intron_*MECP2* (-)	TAGCCCACTTAGTCAGTTGTGTCTAAGCCACTTGATGAACCAGTCAATCCAGTCTTCA
BAB3147	TAGCCCACTTAGTCAGTTGTGTCTAAGCTGTTTCTGGCTTGCAATTCTGTCACCTCAG
R1_intergenic (+)	GTTCCAGAAGAAATATAGGCACGGAGGCTGTTTCTGGCTTGCAATTCTGTCACCTCAG

BAB2799



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3'UTR_TEX28 (–)       CAGCCTGGTGAAATGGATATAGACGCCCACCTGCCTCACACTTCTGTTTTGCCAGGAT
                                         cis
3'UTR_TEX28 (–)       ATGTAACCACCAGCCTGGTGAAATGGATATAGACGCCCACCTGCCTCACACTTCTGTT
Intron_Opsin (-)      GTGCGGCAGCGTGTGCAAGAAAGAGACCTGGAATGCAAACACGCTATGCGTTCACTGA
                                                trans
Intron_Opsin (+)      AACGCATAGCGTGTTTGCATTCCAGGTCTCTTTCTTGCACACGCTGCCGCACCACGCC

Intron_Opsin (+)      CCCCACCTTTCAGAGGCTGCTTGGGTCATAGATCCACCTGGGCCTACAGAGCACATGT
BAB2799               CCCCACCTTTCAGAGGCTGCTgccaaccCTGGAAGATTCTCAGCTATTAGCTTTTCAG
R_Intron_ ABCD1 (+)   CTTTGAATATTTGAGTTTTTCAATCATTCTGGAAGATTCTCAGCTATTAGCTTTTCAG
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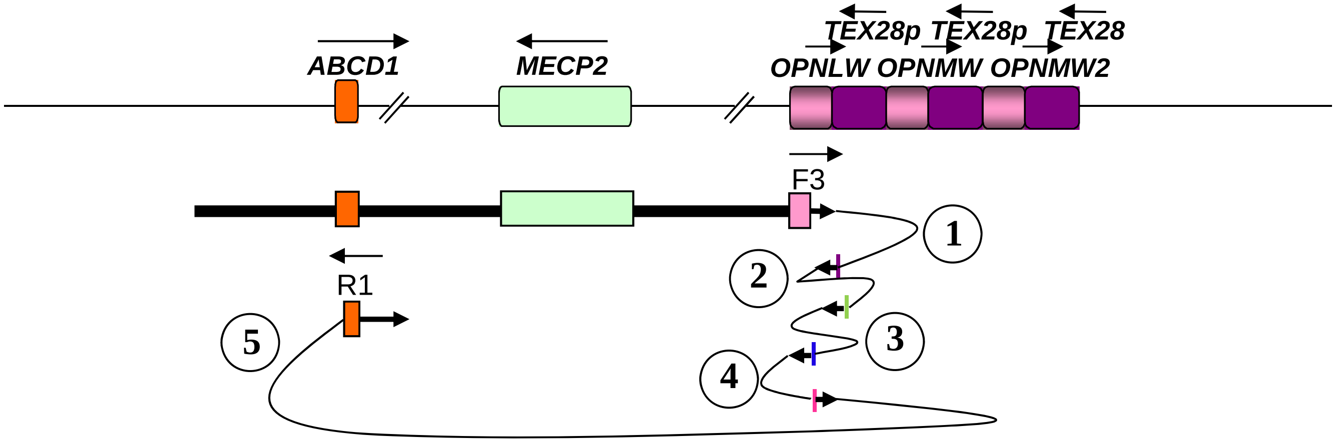

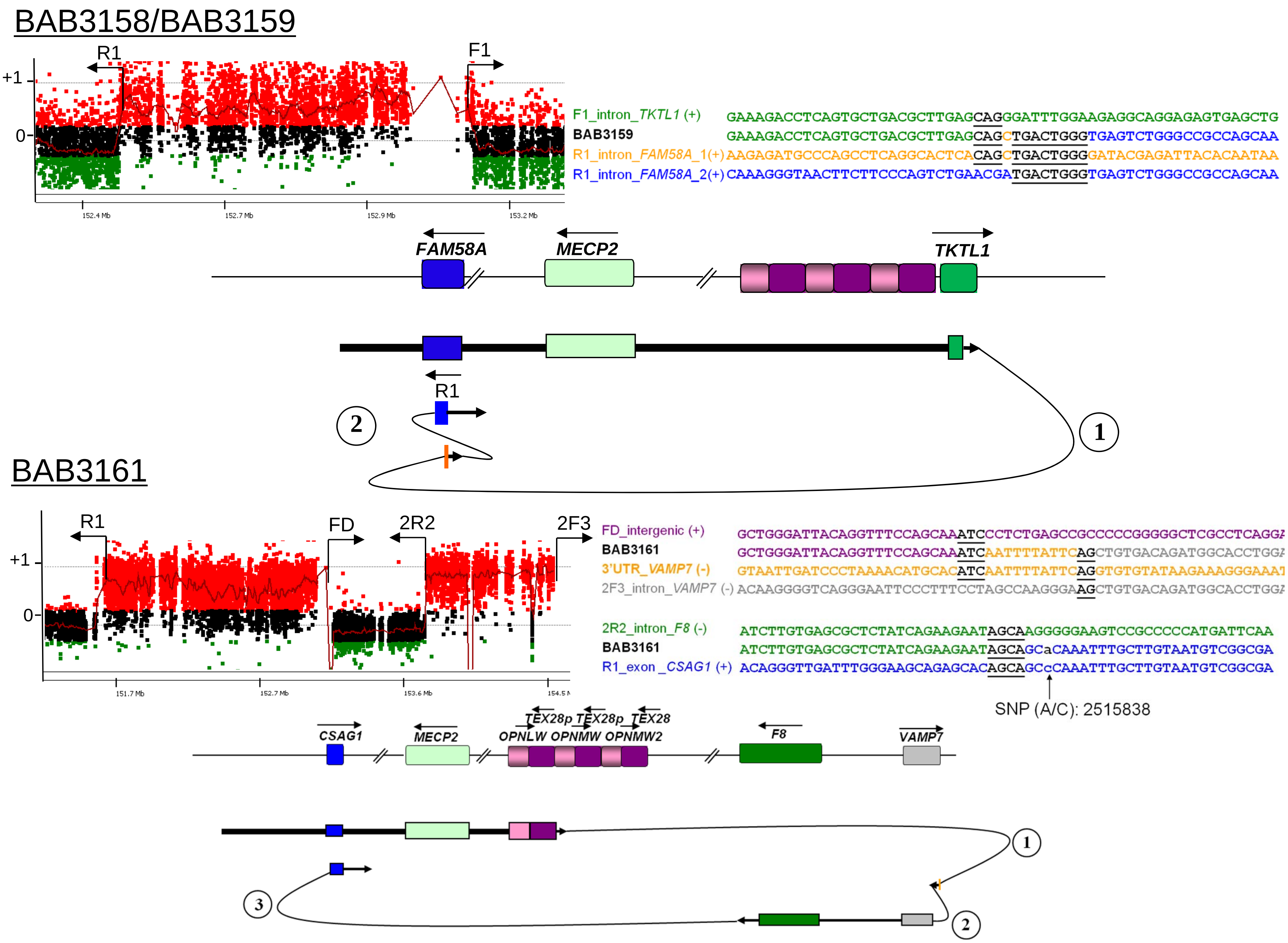
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R1_intron_FAM58A_1(+) AAGAGATGCCCAGCCTCAGGCACTCACAGCTGACTGGGGATACGAGATTACACAATAA
R1_intron_FAM58A_2(+) CAAAGGGTAACTTCTTCCCAGTCTGAACGATGACTGGGTGAGTCTGGGCCGCCAGCAA
FAM58A
MECP2
TKTL1
R1
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3'UTR_VAMP7 (-) GTAATTGATCCCTAAAACATGCACATCAATTTTATTCAGGTGTGTATAAGAAAGGAAA
2F3_intron_VAMP7 (-) ACAAGGGGTCAGGAATTCCCTTTCCTAGCCAAGGGAAGCTGTGACAGATGGCACCTGG
2R2_intron_F8 (-) ATCTTGTGAGCGCTCTATCAGAAGAATAGCAAGGGGGAAGTCCGCCCCCATGATTCAA
BAB3161 ATCTTGTGAGCGCTCTATCAGAAGAATAGCAGCaCAAATTTGCTTGTAATGTCGGCGA
R1_exon _CSAG1 (+) ACAGGGTTGATTTGGGAAGCAGAGCACAGCAGCcCAAATTTGCTTGTAATGTCGGCGA
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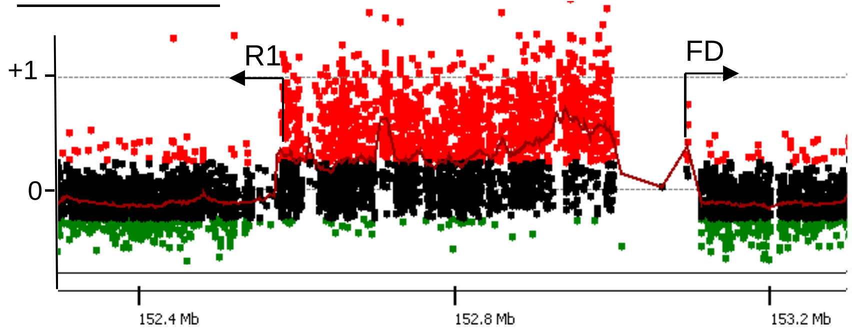
BAB3204



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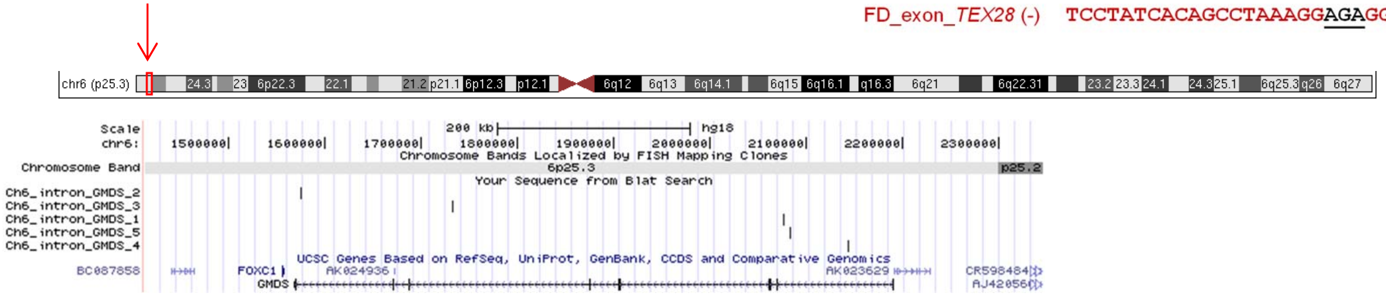
BAB3204	TGAGGATTTGCAGCACAGAATTCTCTCCCCCAAAACACACTCATTGCCACACGTGCTGT
Ch6_intron_*GMDS* (+)	TGGTCTACCATGGCACAGAATTCTCTCCCCCAAAACACACTCAGTTTCAGTACATTGCA
Ch6_intron_*GMDS* (-)	AGTGTCCAGTGGTGGTTTTATTCCAAGTCTACACCGTCGCTGATTGCCACACGGCAGCC
exon_*TEX28* (-)	CTTACAGCAGGGGACGTGCTTAGAGACAGAGGATGTGGCCCAGCAACAAAACCTGCTGT

exon_*TEX28* (-)	TGCAGAAGGTAAAGGCAGAGCTGGAAGAAGCCAAGAGGTTCCACATCAGCCTCCAGGAG
BAB3204	TGCAGAAGGTAAAGGCAGAGCGGCTAGTGGGAGGTGGCATGGGAGAGTGTGGAGAAGGC
Ch6_intron_*GMDS* (+)	TCCGCCTGGTGCTGGAGGTGCGGCTAGTGGGAGGTGGCATGGGAGAGTGTGGAGAAGGC

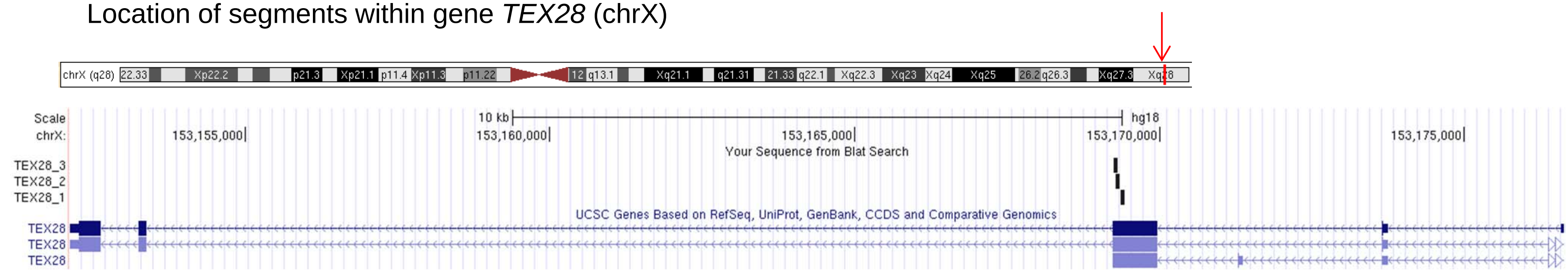
Ch6_intron_*GMDS* (+)	AACCCGAGACAGGCAGACTGGGAGCCCAGACCTGCCACTGCCCACCGTGCAGCCTTGGC
BAB3204	AACCCGAGACAGGCAGACTGACATTTTAGAAAAGAGGCTGATGTGGAACCTCTTTGGTT
Ch6_intron_*GMDS* (-)	AAGAGTCTTAGCATACTTTGACATTTTAGAAAAGAAAAACAGAATTGAAGGGGACACAA
exon_*TEX28* (-)	AGACCTCTCCTTTAGGCTGTGATAGGACTCCTGGAGGCTGATGTGGAACCTCTTGGCTT
Ch6_intron_*GMDS* (+)	AAGTACTGAATACTTGGGCAAGTGCCCCTTTTTGTTTCAACAAAAGATCATCTTTGGTT

Ch6_intron_*GMDS* (+)	GGAGTAAATATTATGCTATAGAAATCTGTGTAAAACTCGTCCAGGACTCCACCAACAAGT
BAB3204	GGAGTAAATATTATGCTATAGAGGTCTCTGACTGACCTGCAGCTGTTGCTGGAGTCCCTT
FD_exon_*TEX28* (-)	TCCTATCACAGCCTAAAGGAGAGGTCTCTGACTGACCTGCAGCTGTTGCTGGAGTCCCTT

Location of segments within gene *GMDS* (chr6)

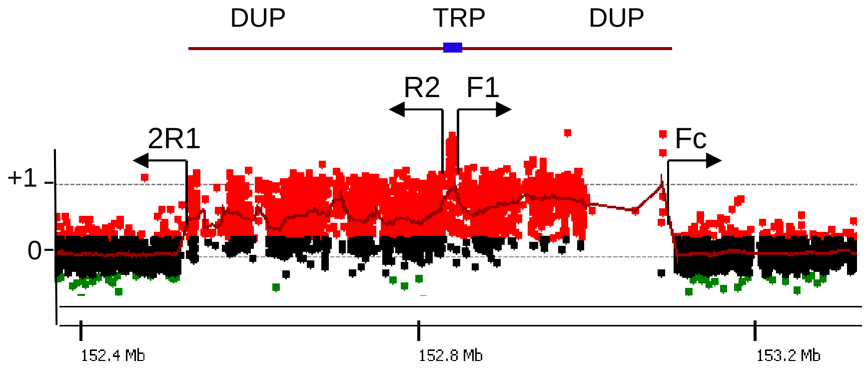


Location of segments within gene *TEX28* (chrX)

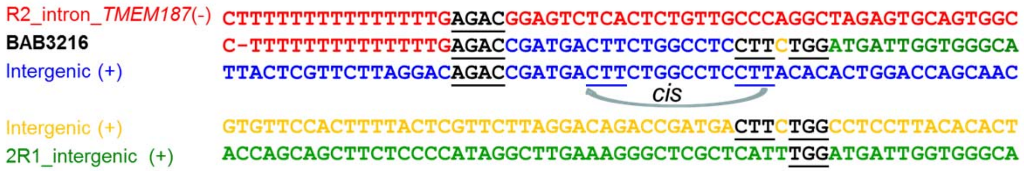


BAB3216

DUP-TRP/INV-DUP



Breakpoint junction R2 + 2R1



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Intergenic (+)        TTACTCGTTCTTAGGACAGACCGATGACTTCTGGCCTCCTTACACACTGGACCAGCAAC
                                                         cis
Intergenic (+)        GTGTTCCACTTTTACTCGTTCTTAGGACAGACCGATGACTTCTGGCCTCCTTACACACT
2R1_intergenic  (+)   ACCAGCAGCTTCTCCCCATAGGCTTGAAAGGGCTCGCTCATTTGGATGATTGGTGGGCA
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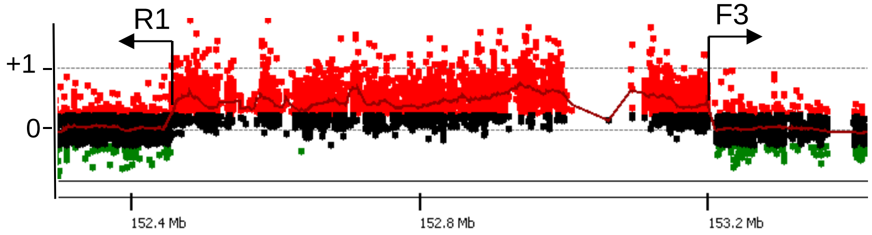
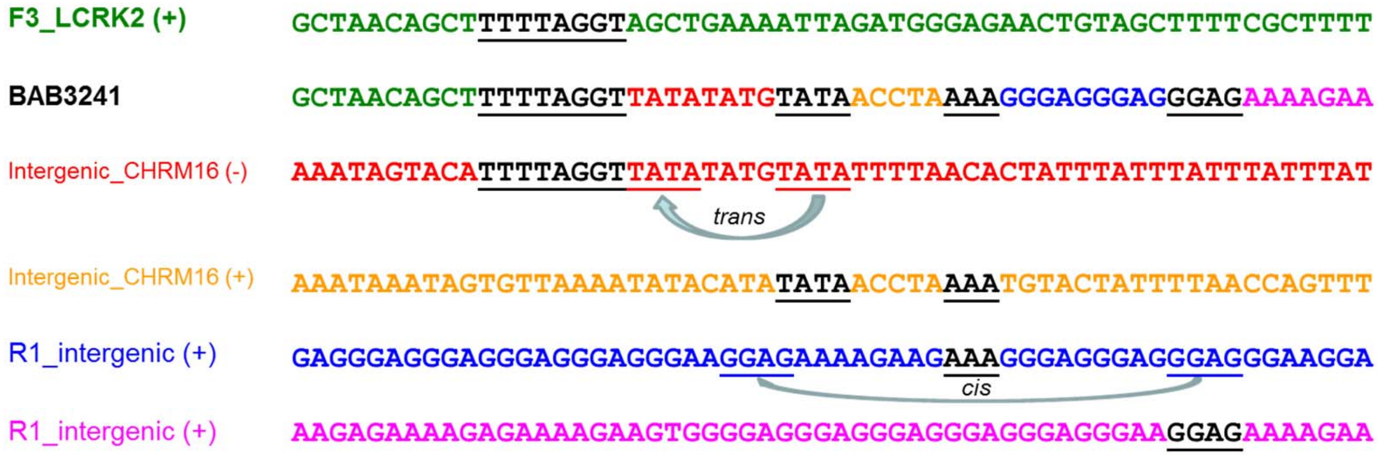
Breakpoint junction F1 + Fc: ~10 kb band ; sequencing of this entire product was not achieved due to the high frequency of repetitive sequences within it.

BAB3241





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F3_LCRK2 (+)             GCTAACAGCTTTTTAGGTAGCTGAAAATTAGATGGGAGAACTGTAGCTTTTCGCTTTT

BAB3241                  GCTAACAGCTTTTTAGGTTATATATGTATAACCTAAAAGGGAGGGAGGGAGAAAAGAA

Intergenic_CHRM16 (-)    AAATAGTACATTTTAGGTTATATATGTATATTTTAACACTATTTATTTATTTATTTAT
                                              trans

Intergenic_CHRM16 (+)    AAATAAATAGTGTTAAAATATACATATATAACCTAAAATGTACTATTTTAACCAGTTT

R1_intergenic (+)        GAGGGAGGGAGGGAGGGAGGGAAGGAGAAAAGAAGAAAGGGAGGGAGGGAGGGAAGGA
                                                        cis

R1_intergenic (+)        AAGAGAAAAGAGAAAAGAAGTGGGGAGGGAGGGAGGGAGGGAGGGAAGGAGAAAAGAA
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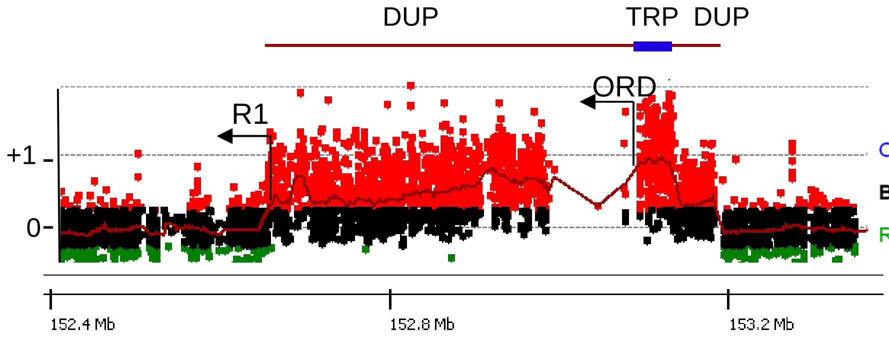
BAB3255

DUP-TRP/INV-DUP



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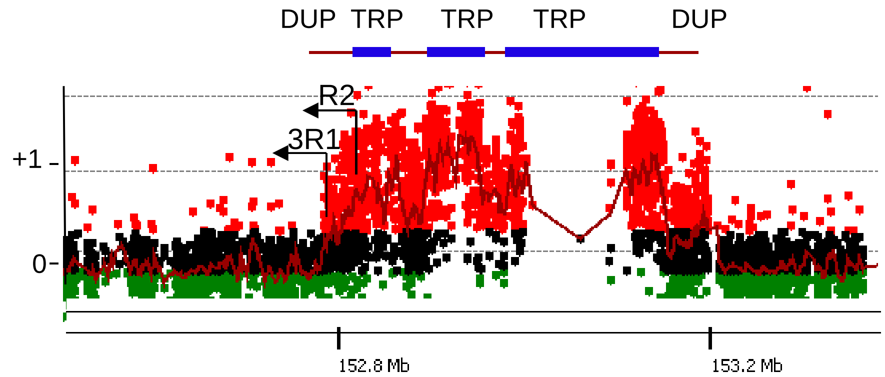
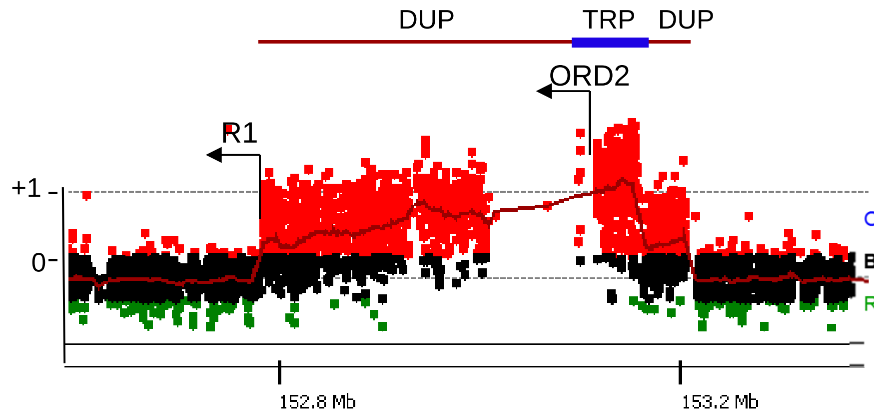
ORD_intron_ TEX28 (-)   GAGTTTTGGGGGGACATGTACCCCATAGCAGTATGCTTAACTTTTTAAGAAAGAGG
BAB3255                 GAGTTTTGGGGGGACATGTACCTGCTGGAGCCCGGCCCAAGGCTAGAGGCCTGGAG
R1_intron_PDZD4 (+)     GGGGGTAGATACAACGGGGCGCCTGCTGGAGCCCGGCCCAAGGCTAGAGGCCTGGAG
```

BAB3259



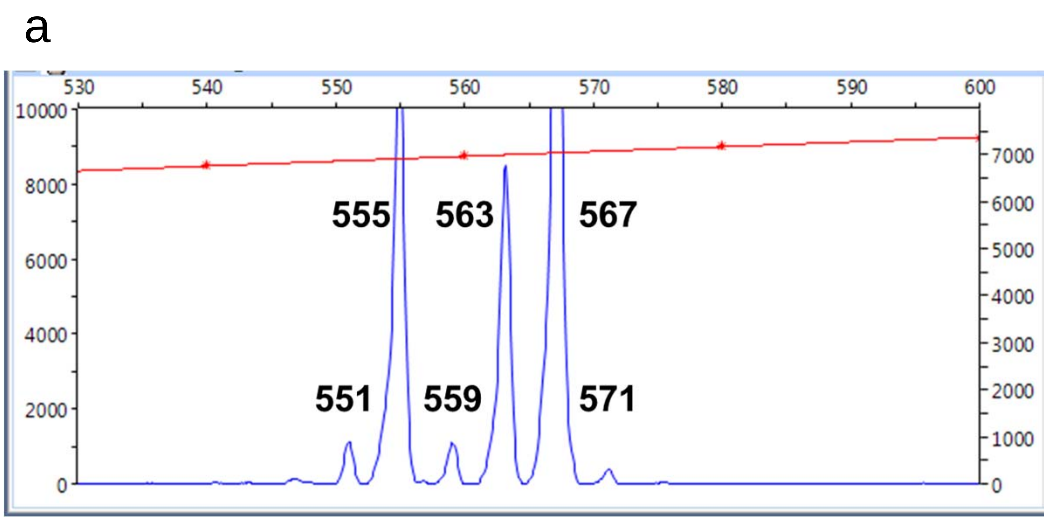
```
R2_intron_ RENPB (-)     AGAAGTGAGGATTACATCTTCCTAGGCCCGGGCTGGACGCGGTGGCTCGTGCCTGT
BAB3259                  AGAAGTGAGGATTACATCTTCCTAGCCATGCAGAGATCCAGGAGCACTGTTTACCT
Intergenic (-)           TCGAGGGAGGGAGGGCAGGAGGGAGCCATGCAGAGATCCAGGAGGGCATCACAGGA
intron_MECP2 (-)         GTTATTGTTGGCCTTAGAATATGTCCCACTACAGGTTTGCAGAGCACTGTAGTCAA
intron_MECP2 (+)         ATATATATATATATATATATATATATATACACACACACACAAAACAAAGTTTACCT
intron_ MECP2 (+)        TTTTAACTATTTCTAAGTGTACAATGAATGAACTTTTTTTTTTTTTTTTTGAGAC
BAB3259                  TTTTAACTATTTCTAAGTGTACAATGctcgtttgttGTGCTGAAGCTCCAGGGACT
3R1_intron_ARHGAP4 (+)   GCTCTGCTCCCCCTCAGTGCTAAAGGGAGGCAAAAGGTGCTGAAGCTCCAGGGACT
```

BAB3274/BAB3275

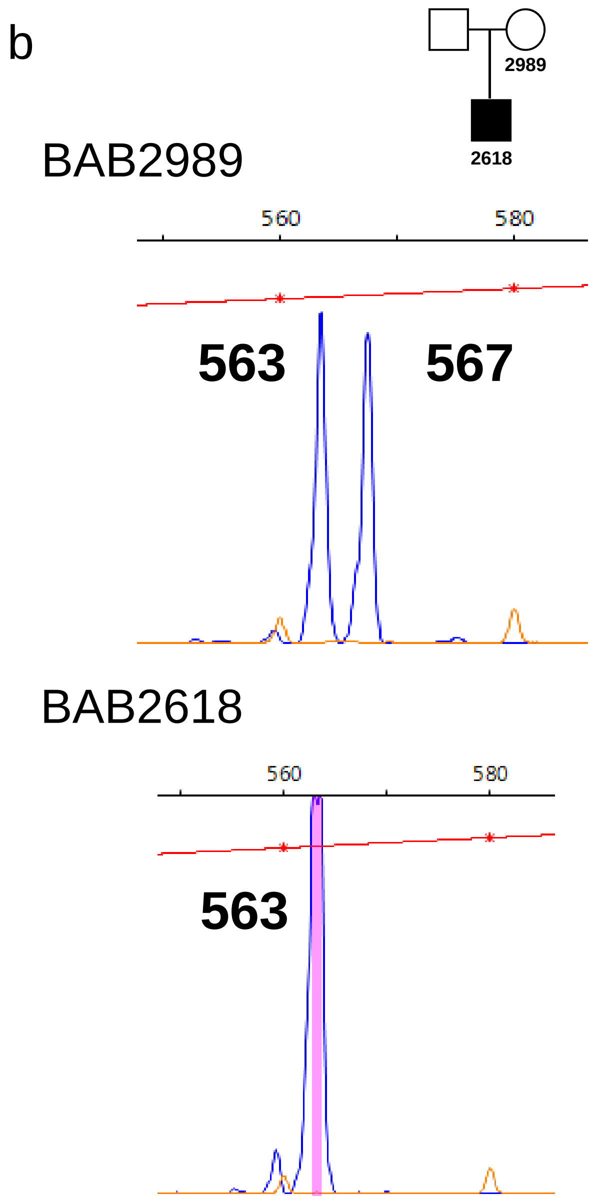


```
OR2_intron_ TEX28 (-)     GCAGTGAGTCATGATTGCACCACTGCACTCCATTCTGGGTGACAGAGCAAGACCCT
BAB3274/BAB3275           GCAGTGAGTCA---------CACAA-----CAAAACAAAGAATACAGACATTTTCA
R1_intron_ARHGAP4 (+)     GCCCTGTTTCAAAAACAAAACACAACAAAACAAAACAAAGAATACAGACATTTTCA
```

Supplementary Figure 6: Microsatellite Xq28_4 (see Methods for further information). This microsatellite consists of a tetranucleotide repeat with two different sequence unit variation (GATG and GATA). (a) Genotype result of a pool of DNA consisted of 29 females. It shows that at least six alleles are present in the population in the following order and relative frequency: 551 bp (2%), 555 bp (30%), 559 bp (2%), 563 bp (20%), 567 bp (45%), 571 (1%) as calculated by the proportional height of each peak. (b) BAB2618 carries only one allele (563) inherited from mother in both copies of his Xq28 duplicated segments. As this was a *de novo* duplication, it supports that an intrachromosomal event took place in the maternal germline or early in the post-zygotic cell divisions.



Allele (bp)	Relative frequency (%)
551	2
555	30
559	2
563	20
567	45
571	1



Supplementary Table 1: Sequence and location of the Opsin panel primers and microsatellite Xq28_4 primers (NCBI36)

Primers	**Location**	**Sequence 5' => 3'**
F1	chrX 153060169 153060192	AGTCAAGGGGTCCCAGCTGAAGCA
F2	chrX 153063565 153063588	CCACTTGGCTTTTGGCTGCACACC
	chrX 153101975 153101998	
	chrX 153139093 153139116	
F3	chrX 153065123 153065146	CACAGGTCTTCTTACCCTGAGGGT
	chrX 153103534 153103557	
	chrX 153140652 153140675	
F4	chrX 153067484 153067508	GTGTGCTGGACACTCTGTGAGGATG
F5	chrX 153069618 153069639	GGTAAGCCAGTCGGGGCCCAGG
	chrX 153106749 153106770	
	chrX 153143867 153143888	
F6	chrX 153062088 153062112	AACTGAGGCGAGGCTACGGAGTTGG
OLF	chrX 153061648 153061667	GGGCCCTGGCAAGTCTGTGG
OMF	chrX 153094819 153094842	GCTCACAGTGTTGCCATCCCGACT
	chrX 153131939 153131962	
	chrX 153169744 153169767	
F1b	chrX 153083451 153083471	ATGGAGGAGAAGTCGGGGAAA
	chrX 153120582 153120602	
	chrX 153157704 153157724	
F2b	chrX 153081253 153081276	TATTCCATTTGTCTCTGCCCCAGG
	chrX 153118384 153118407	
	chrX 153155502 153155525	
FA	chrX 153073308 153073332	GGGTGCAGTCTTACATGATTGTCCT
	chrX 153110438 153110462	
	chrX 153147556 153147580	
FB	chrX 153077917 153077941	TGGTTACATAATCGGCAAGCAAGAG
	chrX 153115047 153115071	
	chrX 153152165 153152189	
FC	chrX 153086336 153086360	TCCAGGACTGCGAGAGAATAACTTT
	chrX 153123465 153123489	
	chrX 153161273 153161297	
FD	chrX 153091353 153091377	CGGCTGCTGACCACGATAAGACCTA
	chrX 153128479 153128503	
	chrX 153166286 153166310	
FE	chrX 153094297 153094320	GCCTCACCTACACTTCTCCTCCTG
	chrX 153131417 153131440	
	chrX 153169222 153169245	
Xq28F4	chrX 153019757 153019780	GGATTGAGATGATCCTGGCTAAAA
Xq28R4	chrX 153020294 153020316	AGATGACAAGTGTGCACAACCAT

Supplementary Table 2: Detailed features for 76 long and short distance template-switching events in 31 patients with *MECP2* duplication.

* no sequencing data

** Possibly inverted haplotype

in red: dup visible on aCGH; # in blue: trp visible on aCGH

Y: Yes; NA: not available; NT: not tested;TS: Template Switching; ref: reference; jct: junction; Del: deletion; Ins: insertion

Patient BAB #	Long distance TS	Short distance TS	Template distance in the ref strand (bp)	Direction of misalignment	Strand	Microhomology	Insertion/deletion Size (bp)	Distance break from previous jct	Original copy tested?
2616	1	-	1,527,425	Backward	+	GTG	1,527,425		
2618	1	-	906,032	Backward	+	GA	906,032		
2619	1	-	363,833	Backward	+	CCCC	363,833		
2622	1	-	474,757	Backward**	-	0	5,347		
	2	-	20,296	Forward	+	TGGG	459,808		
2623	1	-	600,086	Backward	+	TGAG	600,086		
		Del	5	Forward	+	CC	7	1: 3 bp	Y; not present
2624	1	-	941,461	Backward	-	TCCTGCCTCAGCCTCC (*Alu* Sg-*Alu* Y)	263,885		
	2*	-	NA	NA	NA	NA	220,961		
	3*	-	NA	NA	NA	NA	92,092		
2626/2628	1	-	1,674	NA (LCR)	-	CCA + ins aaag	55		Y; not present
	2	-	472,892	Backward	+	GCTG	472,892		
2771	1	-	338,801	Backward	+	GCTTGAACCCGGGAGG (*Alu* Sc-*Alu* Sg)	413,726		
2799	1	-	5,955	NA (LCR)	-	AT	3		
		Ins	5	Forward	-	A	11	1: 2 bp	NA
	2	-	5,869	NA (LCR)	-	GC	9		
		Ins	24	Forward	+	ACACGCT	32	2: 2 bp	NA
	3	-	421,907	Backward	+	CT + ins gccaacc	421,762		
2800	1	-	13,120	NA (LCR)	+	GCA	11		NT
	2		314,234	Backward	-	TG	7		
		Ins	136	Backward	+	GGA	301,242	2: 4 bp	
2806	1	-	746,699	Backward**	+	CC	746,699		
2991	1	-	26,931	Backward	+	GC	80		
		Del	14	Forward	+	TG	16	1: 20 bp	Y; not present
	2	-	327,001	Backward	+	CC	353,836		
3027	1	-	783	NA (LCR)	-	CTA	3		
		Ins	5	Backward	+	CGT	27	1: 0 bp	NT
	2	-	643,893	Backward	+	TTG	37		
	3	-	4,575	Forward	+	CCC	649,139		
3147	1*	-	~37,616	Backward	-	Homology LCRs K	~210,491		
	2	-	502,977	Backward	+	GC	762,410		
3154	1	-	551,462	Backward	+	GA	551,462		
3158/3159	1	-	679,274	Backward	+	CAG	9		Y; not present
	2	-	494 bp	Backward	+	TGACTGGG	679,759		
3161	1	-	1,725,353	Forward	-	ATC	12		Y; not present
	2	-	7,019	Backward	-	AG	1,055,894		
	3	-	2,108,425	Backward	+	AGCA	1,445,994		
3172	1	-	349,416	Backward	+	GGCAG	349,416		
3174	1	-	465,348	Backward	+	T	465,348		

Supplementary Table 2 (continued)

Patient BAB #	Long distance TS	Short distance TS	Template distance in the ref strand (bp)	Direction of misalignment	Strand	Microhomology	Insertion/deletion Size (bp)	Distance break from previous jct	Original copy tested?
3204	1	-	ChrX-Chr6 (*GMDS*)	Chr 6	+	G	29		Y; not present
	2	-	501,093 kb (*GMDS*)	Chr 6- Backward	-	A	10		
	3	-	Chr6-ChrX (*TEX28*)	Chr X	-	No	25		
	4	-	ChrX-Chr6 (*GMDS*)	Chr 6	+	GC	57		
	5	-	410,481 bp (*GMDS*)	Chr 6- Forward	-	G	12		
	6	-	Chr6-ChrX (*TEX28*)	Chr X	-	GA	15		
	7	-	ChrX-Chr6 (*GMDS*)	Chr 6	+	TCTT	24		
	8	-	Chr6-ChrX (*TEX28*)	Chr X	-	AGA	462,096		
3216	1*	-	~177,396	Backward	-	NA	~13,807		
	2	-	325,992	Backward	+	AGAC	17		
		Ins	8	Backward	+	CTT	4	2: 17 bp	Y; not present
	3	-	1,919	Forward	+	TGG	515,261		
3238	1	-	1,214,087	Backward	+	G	1,214,087		
3241	1	-	ChrX-Chr16	Chr 16	-	TTTTAGGT	12		
		Ins	4	Chr 16	+	TATA	8	1: 8 bp	Y; not present
	2		Chr16-ChrX	Chr X	+	AAA	13		
		Ins	20	Backward	+	GGAG	770,145	3: 9 bp	Y; not present
3247	1	-	590,628	Backward	+	GCAGA	590,628		
3255	1*	-	~37,616	Backward	-	Homology LCRs K	~130,436		
	2	-	360,231	Backward	+	CC	539,604		
3259	1*	-	~37,616	Backward	-	Homology LCRs K	158,983		Y; not present
	2*	-	NA	NA		NA	59,446		
	3*	-	NA	NA		NA	62,313		
	4	-	196,709	Forward	-	AG	19		
	5	-	103,229	Backward	-	GAG	6		
	6	-	1,201	Forward	+	GT	32		
	7	-	16,861	Backward	+	G + ins ctcgtttgtt	422,095		
3261	1	-	2,718,836	Backward	+	TAGATAAG	2,718,836		
3267/3268		Del	14	Forward	+	GCT	17	2: 34 bp (upstream)	Y; not present
	1	-	596,325	Backward	+	T	596,325		
		Del	2	Forward	+	AT	4	2: 14 bp	Y; not present
3273	1	-	508,717	Backward	+	GG	508,717		
3274/3275	1*	-	~37,616	Backward	-	Homology LCRs K	125,752		
		Del	0	Forward	+	0	9	2: 0 bp (upstream)	
	2	-	255,329	Backward	+	CAC	430,023		
		Del	NA	Forward	+	AA	5	2: 2 bp	Y; not present
3325	1	-	3,753,569	Backward	+	GTGGC (*Alu* Sg-*Alu* Ya5)	3,753,569		

References

1. Carvalho, C.M. *et al.* Complex rearrangements in patients with duplications of *MECP2* can occur by fork stalling and template switching. *Hum Mol Genet* **18**, 2188-203 (2009).