

Supplemental information

**Inverted triplications formed by iterative
template switches generate structural variant
diversity at genomic disorder *loci***

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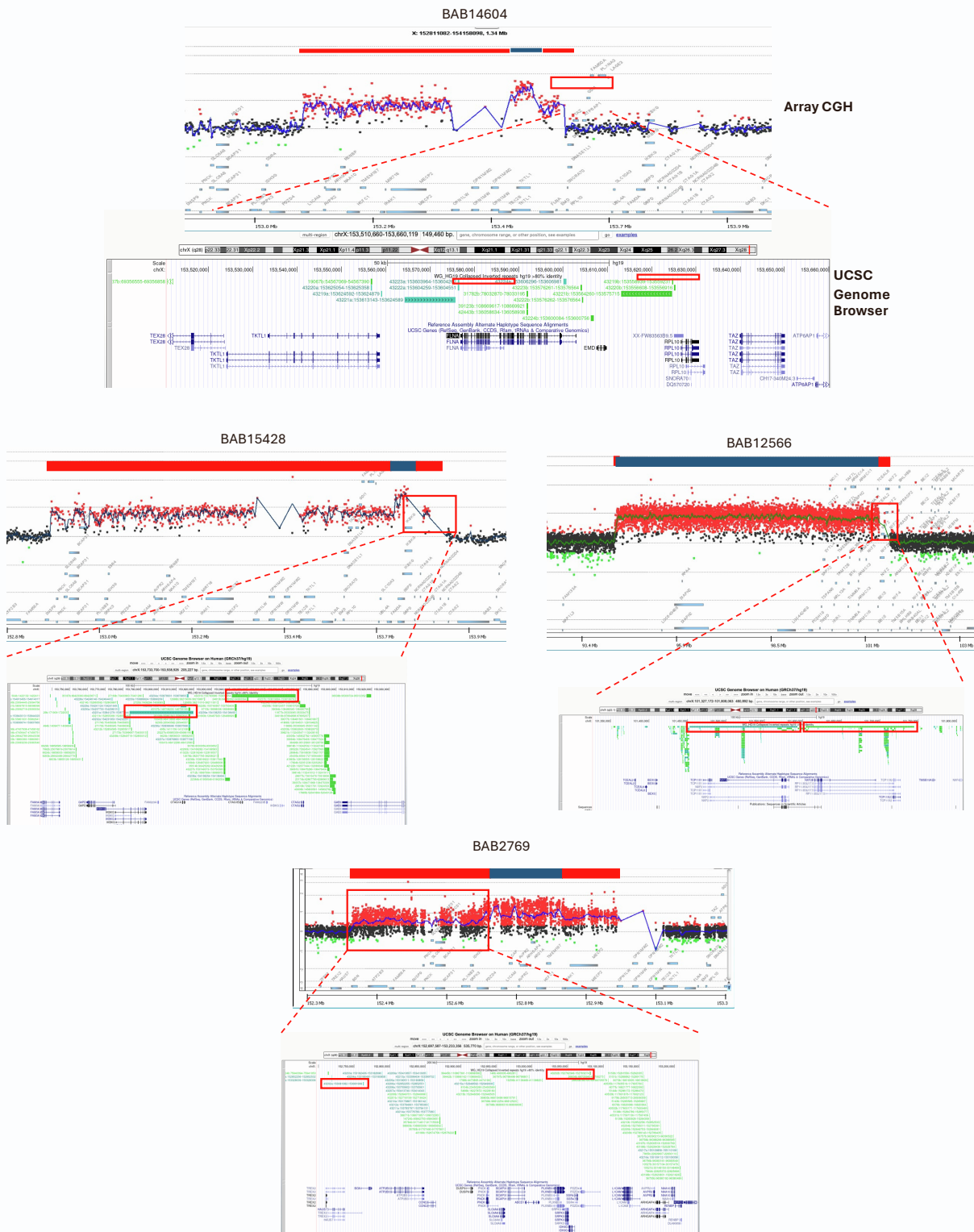


Figure S1: Four pairs of inverted LCRs generating DUP-TRP/INV-DUP events, related to Figure 1. (43202a/43202a; 43221a/43221b (K1/K2); 43231a/43231b (L1/L2)) shown within the *MECP2* locus with the fourth pair being located at Xq22.1 (37696a/ 37696b). The corresponding location in the UCSC genome browser is shown.

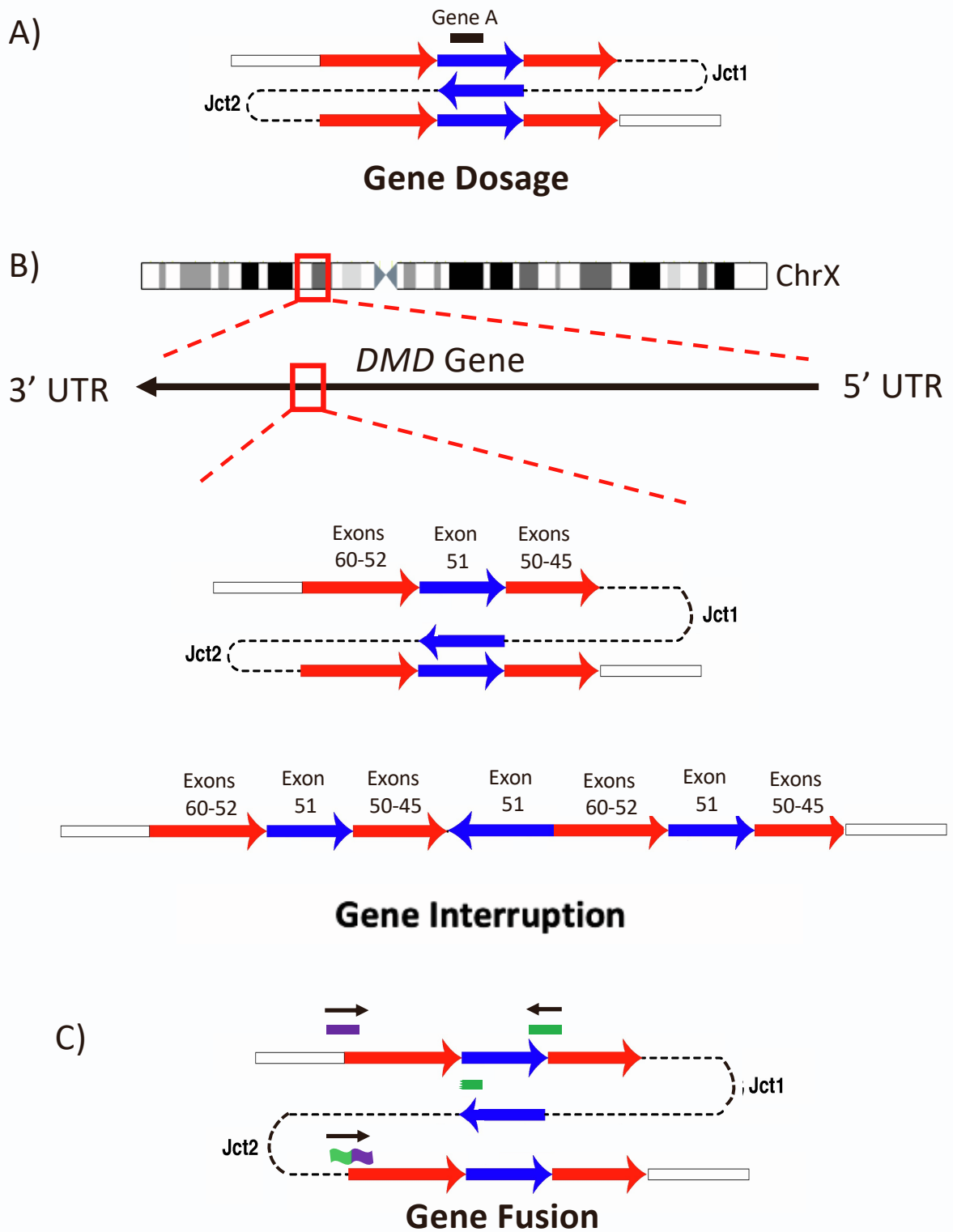
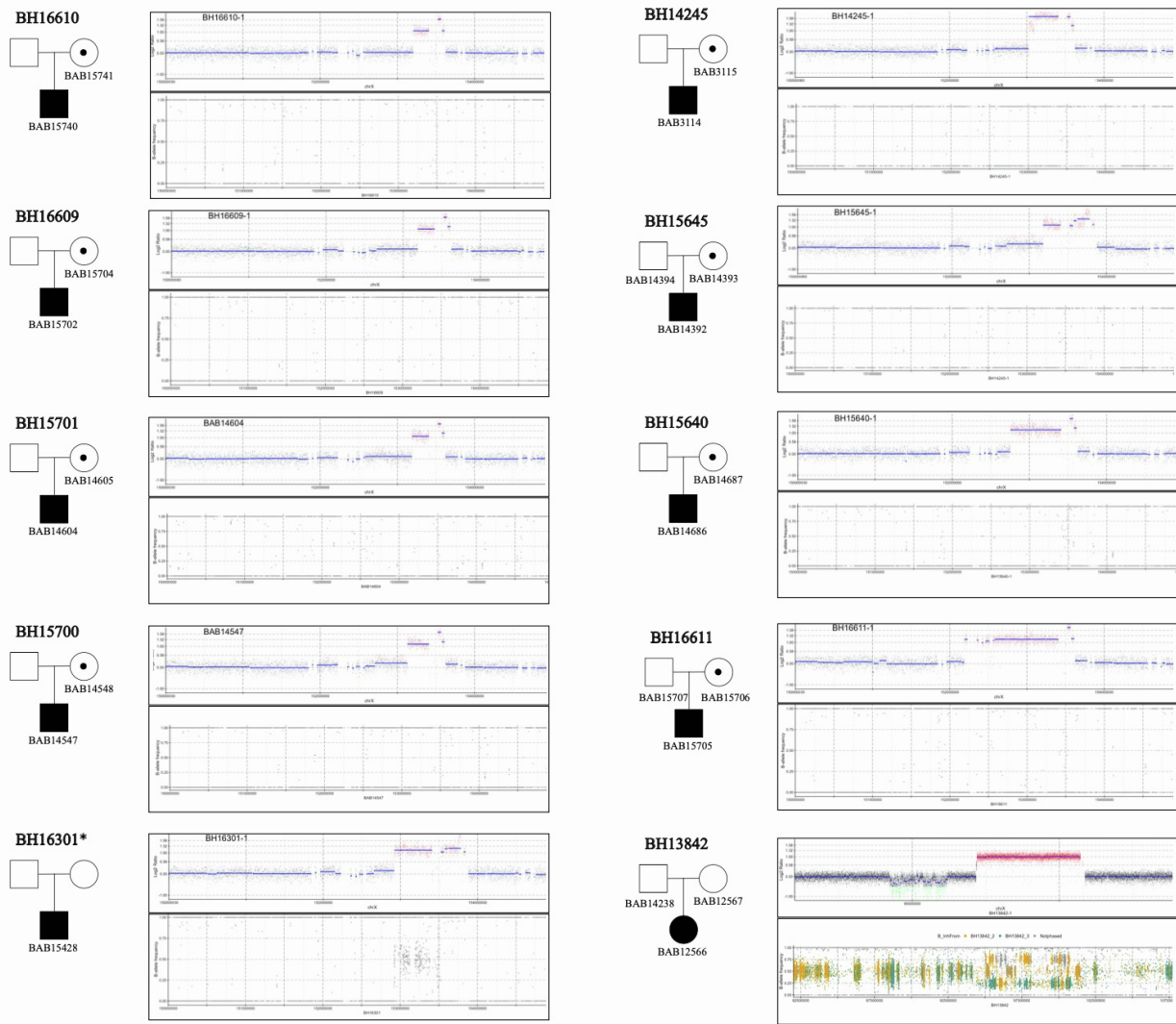


Figure S2: Possible pathogenic effects of a DUP-TRP/INV-DUP event, related to Figure 1. Events include gene dosage (A) gene interruption (B) as characterized by a disruption within the *DMD* gene²⁴ or gene fusion (C) events.



*Parental samples not available - inheritance unknown

Figure S3: VizCNV plots, Related to Table 1. Showing each of the 10 individuals where short-read WGS was performed showing both the Log₂ ratio as well as the B-allele frequency.

BABNumber	Duplication Start	Duplication End	DUP1 Size	Triplication Start	Triplication End	TRP Size	Duplication Start	Duplication End	DUP2 Size	Total Size
BAB15789	152934679	153473892	539213	153505538	153562778	57240	153562880	153623000	60120	688321
BAB15740	153206000	153504574	298574	153505538	153576037	70499	153576037	153623000	46963	417000
BAB15705	152191064	153420198	1229134	153505485	153565901	60416	153576037	153623000	46963	1431936
BAB15702	153190100	153504574	314474	153505538	153576037	70499	153576037	153623000	46963	432900
BAB15428	152922014	153751734	829720	153752634	153783184	30550	153820217	153836222	16005	914208
BAB15420	153000860	153352116	351256	153352787	153564086	211299	153576037	153623000	46963	622140
BAB15418	153055212	153504574	449362	153523215	153576037	52822	153576037	153623000	46963	567788
BAB14686	152754991	153420198	665207	153505538	153576037	70499	153576037	153623000	46963	868009
BAB14604	153189231	153504574	315343	153505538	153565901	60363	153576037	153623000	46963	433769
BAB14547	153130888	153504574	373686	153505538	153576037	70499	153576037	153623000	46963	492112
BAB14392	153180328	153723986	543658	153723986	153783184	59198	153783184	153877318	94134	696990
BAB12566	94346142	94393007	46865	94395899	101440936	7045037	101452290	101745566	293276	7399424
BAB3274	153183739	153504574	320835	153505538	153576037	70499	153576037	153623000	46963	439261
BAB3255	153074186	153374088	299902	153505538	153576037	70499	153576037	153623000	46963	548814
BAB3216	152930997	153246726	315729	153246726	153254972	8246	153254972	153505637	250665	574640
BAB3147	152850937	153353716	502779	153353716	153576037	222321	153576037	153623000	46963	772063
BAB3114	153027282	153097982	70700	153097982	153576037	478055	153576037	153623000	46963	595718
BAB2805	153015998	153108170	92172	153108170	153576037	467867	153576037	153623000	46963	607002
BAB2801	152976275	153286528	310253	152938984	153576037	637053	153576037	153623000	46963	646725
BAB2797	153027282	153045830	18548	153045830	153576037	530207	153576037	153623000	46963	595718
BAB2796	153116401	153390414	274013	153390414	153576037	185623	153576037	153623000	46963	506599
BAB2772	153167337	153504574	337237	153505538	153576037	70499	153576037	153623000	46963	455663
BAB2769	152763385	153084246	320861	153084246	153276174	191928	152929368	153504559	575191	741174
BAB2727	153183448	153505538	322090	153505538	153576037	70499	153576037	153623000	46963	439552

Table S1: Genomic location and size of each genomic fragment in the DUP-TRP/INV-DUP structure for each individual within the cohort, related to Table 1.
All coordinates are based on array positions in Hg19 genome reference.

Inverted Repeat (IR) Pair	Location IR1 (Hg19)	Size IR1 (bp)	Location IR2 (Hg19)	Size IR2 (bp)	Similarity	Distance (bp)
43202a/43202b	chrX:152762349-152763274	926	chrX:153081083-153081999	917	98.12	317810
43221a/43221b (K1/K2)	chrX:153564261-153575715	11455	chrX:153613144-153624589	11446	99.23	37614
43231a/43231b(L1/L2)	chrX:153783687-153819654	35968	chrX:153841277-153877244	35968	99.92	21624
37696a/37696b	chrX:101452532-101593093	140562	chrX:101603859-101744479	140621	99.89	10767

Table S2: LCR summary table including the genomic location (Hg19) as well as nucleotide size (bp), distance between the inverted pair and similarity score (%) each of the 4 pairs identified in this cohort, related to Figure 1

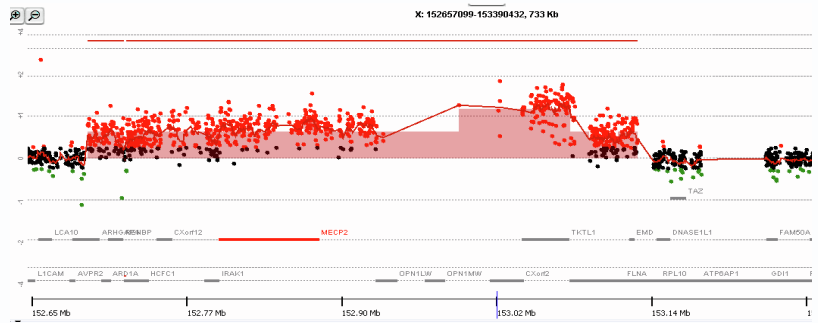
Patient Identifier	Internal Identifier	ArrayCGH	Short-Read WGS	Oxford Nanopore	HiFi Sequencing	Optical Genome Mapping	Strand-Seq
BAB15740	BH16610_1	X	X	N/A	N/A	X	N/A
BAB15705	BH16611_1	X	X	N/A	N/A	X	N/A
BAB15702	BH16609_1	X	X	N/A	N/A	X	N/A
BAB15789	N/A	X	N/A	X	N/A	X	N/A
BAB15428	BH16301_1	X	X	N/A	N/A	X	N/A
BAB15418	BH16300_1	X	N/A	N/A	X	X	N/A
BAB15421*	BH16299_2	X	N/A	N/A	X	X	N/A
BAB12566	BH13842_1	X	X	X	X	X	N/A
BAB14686	BH15640_1	X	X	N/A	X	X	N/A
BAB14604	BH15701_1	X	X	X	X	X	N/A
BAB14547	BH15700_1	X	X	X	X	X	X
BAB14392	BH15645_1	X	X	N/A	X	N/A	N/A
BAB3274	BH16112_1	X	N/A	X	X	X	N/A
BAB3255	BH16108_1	X	N/A	N/A	X	X	N/A
BAB3216	N/A	X	N/A	N/A	N/A	N/A	N/A
BAB3147	BH16111_1	X	N/A	N/A	X	X	N/A
BAB2805	N/A	X	N/A	N/A	N/A	N/A	N/A
BAB2801	BH15649_1	X	N/A	N/A	X	X	N/A
BAB2797	N/A	X	N/A	N/A	N/A	N/A	N/A
BAB2796	BH16110_1	X	N/A	N/A	X	X	N/A
BAB3114	BH14245_1	X	X	X	X	X	X
BAB2772	N/A	X	N/A	N/A	N/A	X	N/A
BAB2769	N/A	X	N/A	N/A	N/A	X	N/A
BAB2727	BH16106_1	X	N/A	N/A	X	N/A	N/A

Table S3: Sequencing methodology that was performed on each sample within this cohort, related to Table 1

Data S1: ArrayCGH showing the *MECP2* critical region, nucleotide-level resolution of junction 2 as well as resolved haplotype and OGM data for the region (where applicable) for each sample, related to Table 1.

BAB2727

Breakpoint Junction 2:

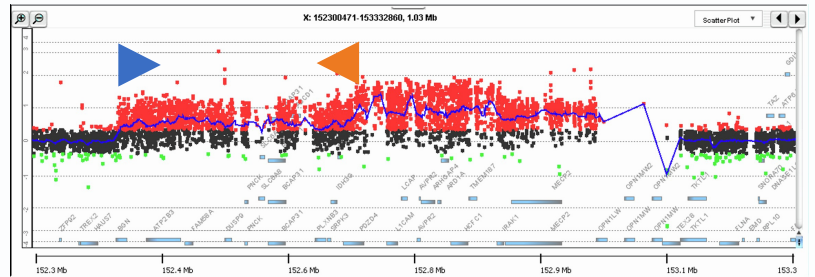


Distal_reference_TEX28B CTGTTTCCTCCGTCAGACTTTTCCTCCAAGTTGTGGGCCATGATTAGGCTCTAGCTCAATTTGCCAAA
 BAB2727 CTGTTTCCTCCGTCAGACTTTTCCTCCAAGTTGTGGGCCAATGGCTCACACACAAATGTGTTTCAGGCC
 Distal_reference_TEX28B (74 bp further ahead) CTCAGAAATGACCAAGAATATACTTTTCTGGCTGGCCCAATGGCTCACAC---CTGTAATCCAGC
 Proximal_reference_intron_ARHGAP4TTTGACCGGCCGGTCCCTCCTTTGTGGTGTCTGTGGGTGTAACACCCCAATGTGTTTCAGGCC
LINE-1 LINE-1

Haplotype Structure: *Not Available*

Adapted from Carvalho *et al.* ¹⁶

BAB2769

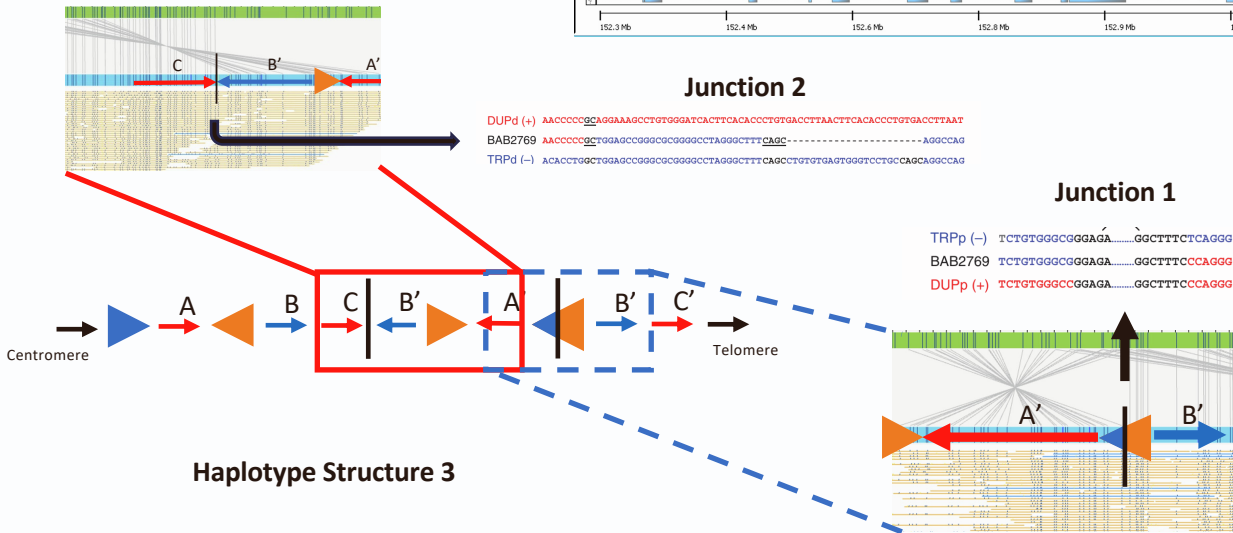


Junction 2

DUPd (+) AACCCCGCAGGAAAGCCTGTGGGATCACTTCACACCCCTGTGACCTTAACCTTCACACCCCTGTGACCTTAAT
 BAB2769 AACCCCGCTGGAGCCGGGCGCGGGCCCTAGGGCTTTCAGC-----AGGCCAG
 TRPd (-) ACACCTGGCTGGAGCCGGGCGCGGGCCCTAGGGCTTTCAGCCTGTGTGATGGGTCTCTGCAGCAGGCCAG

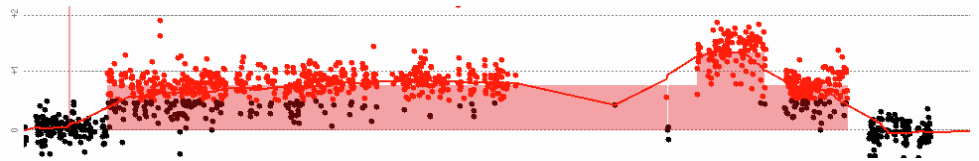
Junction 1

TRPd (-) TCTGTGGGCGGGAGÁ.....GGCTTTCACAGGG
 BAB2769 TCTGTGGGCGGGAGA.....GGCTTCCCAAGG
 DUPp (+) TCTGTGGGCGGGAGA.....GGCTTCCCAAGG



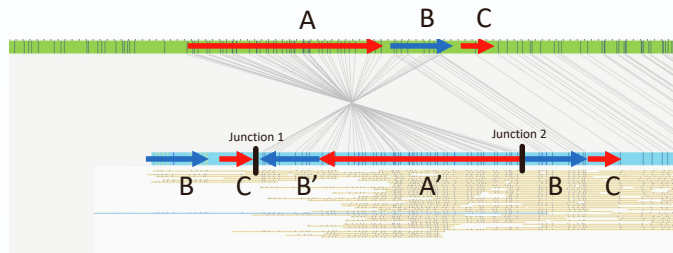
Adapted from Carvalho et al. ⁵

BAB2772

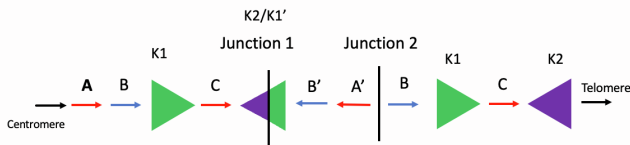


Breakpoint Junction 2:

TRPp (-) GCCAGCCTGGTCTCAA
 jct2 BAB2772 GCCAGCCTGGGGGAGGG
 DUPp (+) GAGGTGTTGGGGGAGGG

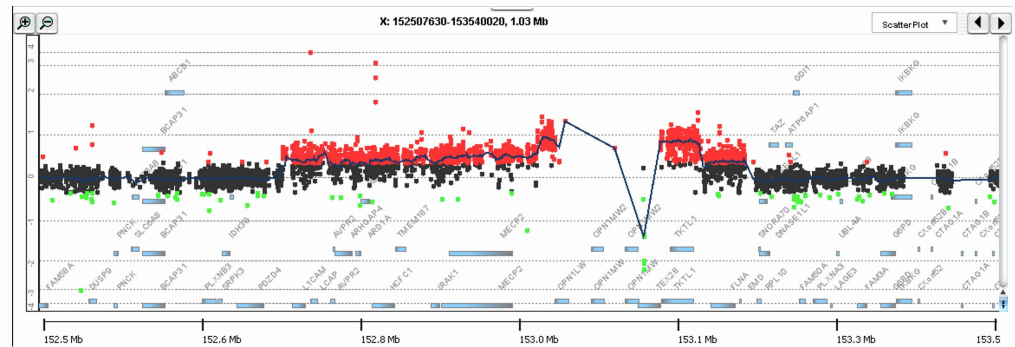


Haplotype Structure 3:



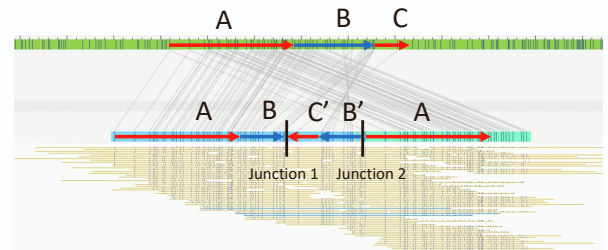
Adapted from Carvalho et al. ⁵

BAB2796

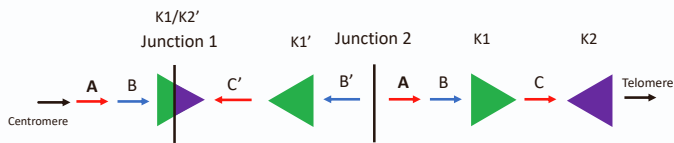


Breakpoint Junction 2:

TRPp (-) TCTCTTG--CCTCAGC
 jct2 BAB2796/BAB2980 TCTCTTGAA**T**TATAAC
 DUPp (+) CTAATTC**T**TTATAAC

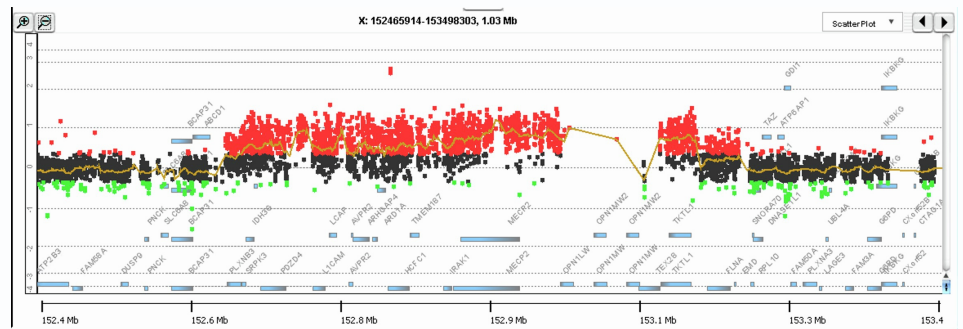


Haplotype Structure 2



Adapted from Carvalho et al. ⁵

BAB2797



Breakpoint Junction 2:

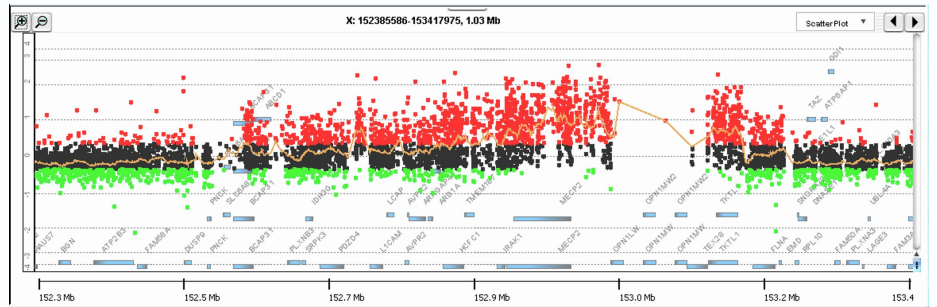
TRPp (-) GCACAGGGCAGGATG
jct2 BAB2797 GCACAGGATGTTTT
DUPp (+) AAATCCATTGTTTT

Haplotype Structure: *Not Available*

Adapted from Carvalho et al. ⁵

BAB2801

Breakpoint Junction 2:



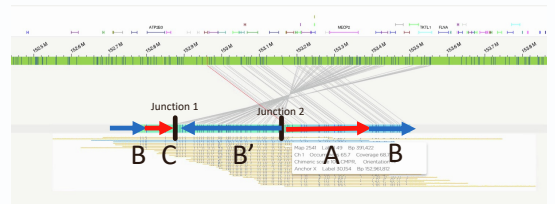
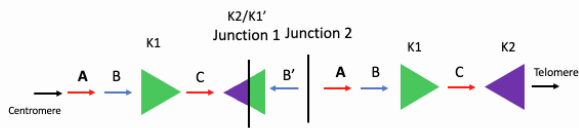
chrX:152955672

chrX:152955637 (+) TACTGTAGCCCCTCTTCAACACACTCAA**CCC****A****CCC****CCTCAAGACTCCACCTGGGGCCTGAGTCAGTGGCC**
 Junction 2 **ATAGTTTCACTGTGTTCCACGCATTCAT**CCC****T****CCC****CCTCAAGACTCCACCTGGGGCCTGAGTCAGTGGCC****

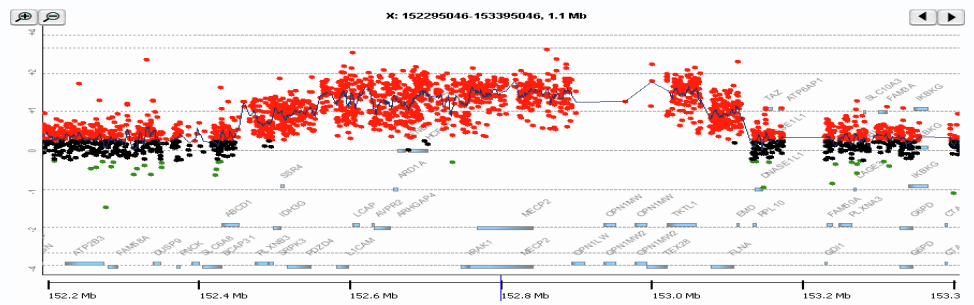
chrX:153198729 (-) **ATAGTTTCACTGTGTTCCACGCATTCAT**CCC****T****CCC****TCTCCTTAACCCGTGGCAACCACTGATTTTTTACT****

chrX:153198702

Haplotype Structure 4



BAB2805



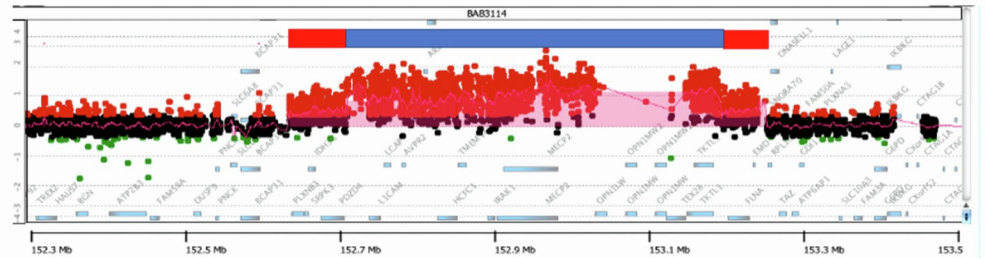
Breakpoint Junction 2:

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TRPp (-) AACAGACAAAAAAGGCCGGGC
jct2 BAB2805 AACAGACAAAAAAGATG-AAA
DUPp (+) ATAAAAATAAAAAATATGGAAA
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Haplotype Structure: *Not Available*

Adapted from Carvalho et al. ⁵

BAB3114

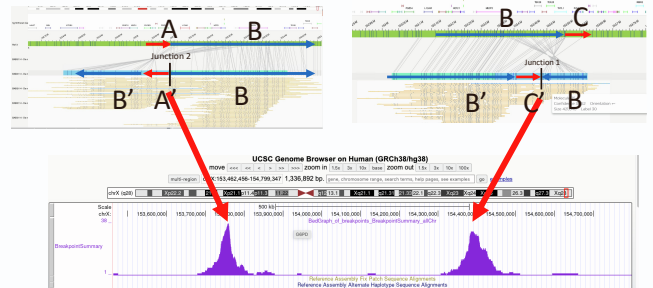
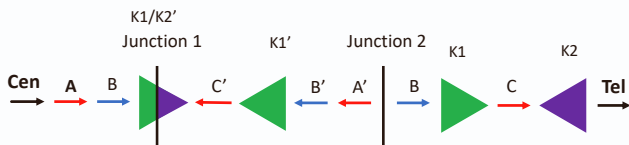


Breakpoint Junction 2:

chrX:153024486

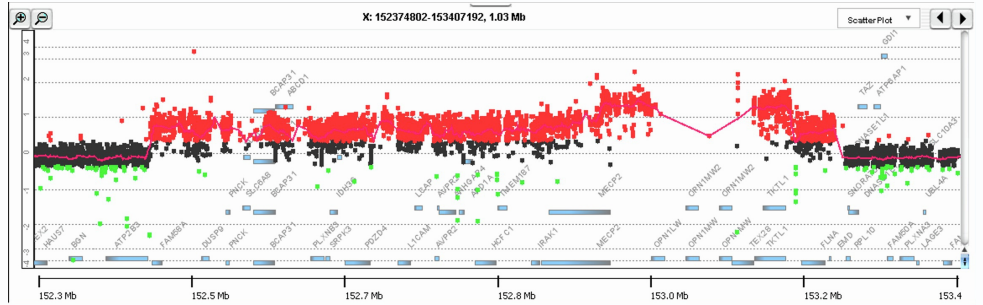
chrX:153024456 (+) AACAAATGGTATAGGGGAACTTGATATCCACATGCAAAAGAATAAAGTTGGAGGCTGGGCATGGTG
 Junction 2 CCATCCCCCGCTGCCCTTTTTCTCCTGGCCACATGCAAAAGAATAAAGTTGGAGGCTGGGCATGGTG
 chrX:153096011 (-) CCATCCCCCGCTGCCCTTTTTCTCCTGGCCGGGTGCGAGGGAGCATGCCCGCGCCTGGCCTCGGCCA
 chrX:153095984

Haplotype Structure 1



Adapted from Carvalho et al. ⁵

BAB3147

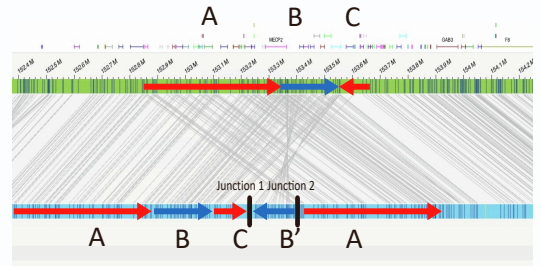
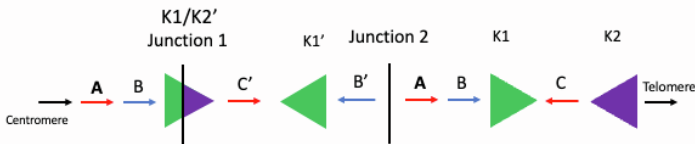


Breakpoint Junction 2:

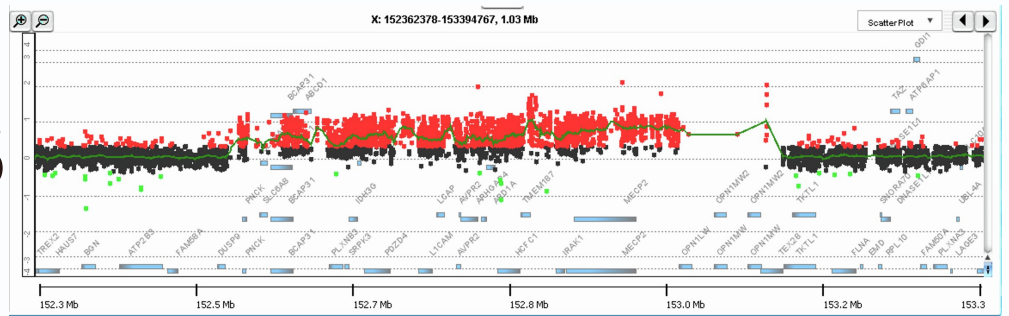
chrX:152850819

chrX:152850786 (+) GTTAGGTTCCAGAAGAAATATAGGCACGGAGG**CGTGTTC**TGGCTT**GCAATTCTGTCACCTCAGAGT**
 Junction 2 **AGGTATAGCCCAC**T**TAGTCAGTTGTGTCTAAG****CGTGTTC**TGGCTT**GCAATTCTGTCACCTCAGAGT**
 chrX:153353826 (-) **AGGTATAGCCCAC**T**TAGTCAGTTGTGTCTAAG**CACTT**GATGAACCAGTCAATCCAGTCTTCACCT**
 chrX:153353796

Haplotype Structure 6:



BAB3216



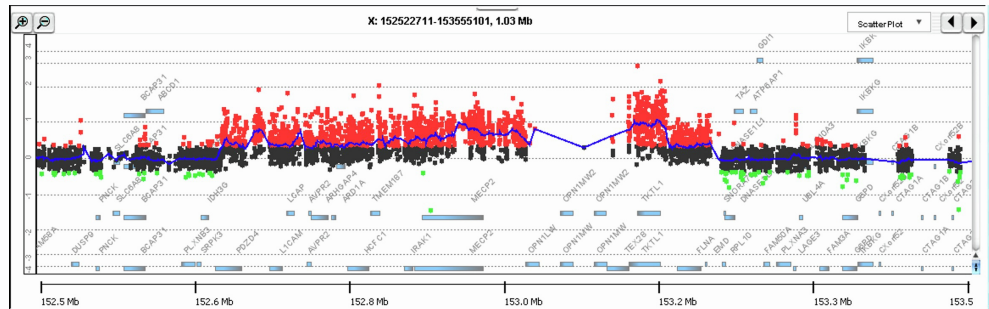
Breakpoint Junction 2:

R2_intron_TMEM187(-)	CTTTTTTTTTTTTTT AGAC GGAGTCTCACTCTGTTGCCAGGCTAGAGTGCAGTGGC
BAB3216	C- TTTTTTTTTTTTTT AGACCGATGACTTCTGGCCTCCTT CTGGATGATTGGTGGGCA
Intergenic (+)	TTACTCGTTCTTAGGAC AGAC CGATGACTTCTGGCCTCCTTACACACTGGACCAGCAAC
	 cis
Intergenic (+)	GTGTTCCACTTTTACTCGTTCTTAGGACAGACCGATGACTT CTGGCCTCCTTACACACT
2R1_intergenic (+)	ACCAGCAGCTTCTCCCATAGGCTTGAAAGGGCTCGCTCATT CTGGATGATTGGTGGGCA

Haplotype Structure: *Not Available*

Adapted from Carvalho et al. ¹⁷

BAB3255



Breakpoint Junction 2:

chrX:153073620

chrX:153073584 (+) CTCCTCGGCCCTGGGGGTAGATACAACGGGGCGCCTGCTGGAGCCCGGCCAAGGCTAGAGCCTGGAGTTTT

Junction 2 TAAATTTCAACACGAGTTTTGGGGGGACATGTACCCTGCTGGAGCCCGGCCAAGGCTAGAGCCTGGAGTTTT

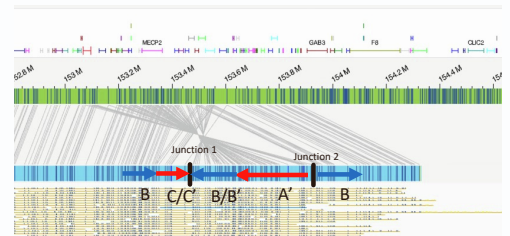
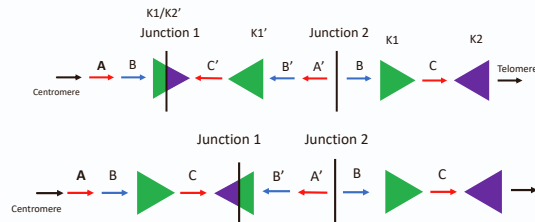
chrX:153433884 (-) TAAATTTCAACACGAGTTTTGGGGGGACATGTACCCCATAGCAGTATGCTTAACTTTTTAAGAAAGAGGAAGG

chrX:153433851

Haplotype Structure 3

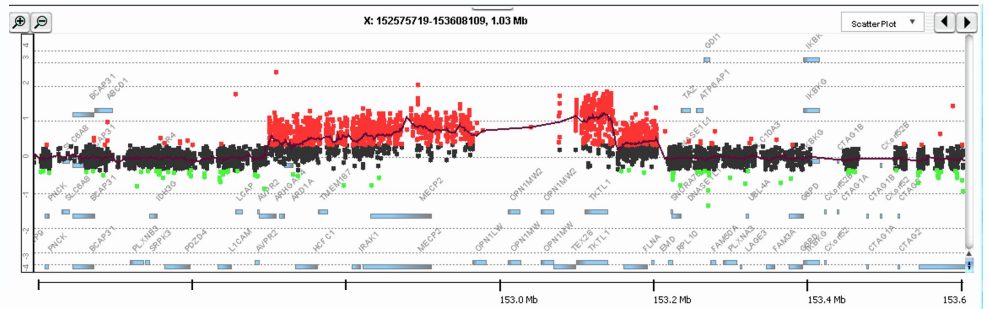
Or

Haplotype Structure 1



BAB3274

Breakpoint Junction 2:



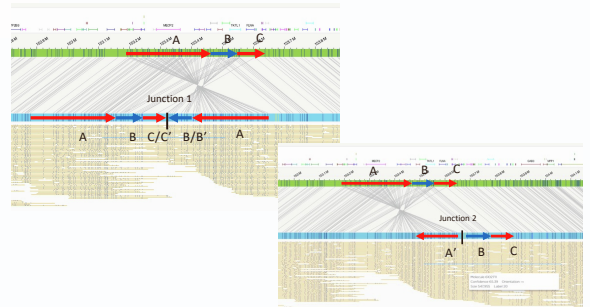
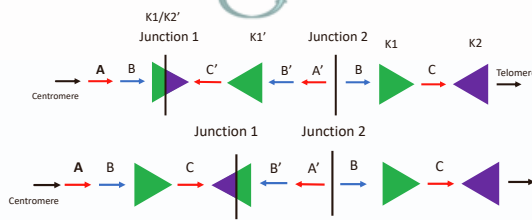
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OR2_intron_TEX28 (-) GCAGTGAGTCATGATTGCACCACTGCACTCCATTCTGGGTGACAGAGCAAGACCCT
BAB3274/BAB3275   GCAGTGAGTCA-----CACAA-----CAAAACAAGAATACAGACATTTTCA
R1_intron_ARHGAP4 (+) GCCCTGTTTCAAAAACAAACACAACAAAACAAAACAAGAATACAGACATTTTCA
  
```

Haplotype Structure 3

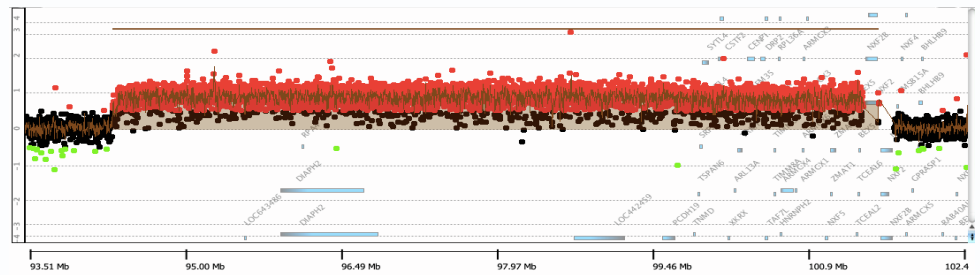
Or

Haplotype Structure 1



Adapted from Carvalho et al. ¹⁷

BAB12566



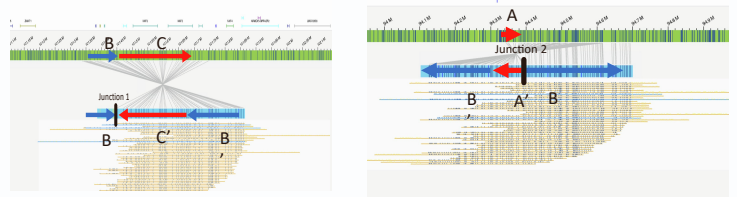
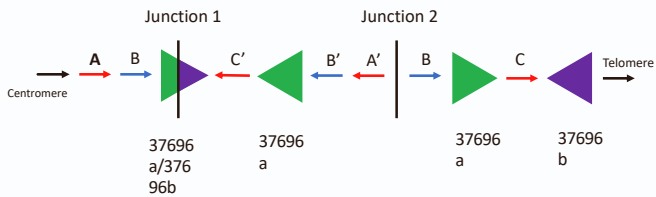
Breakpoint Junction 2:

chrX:94345302

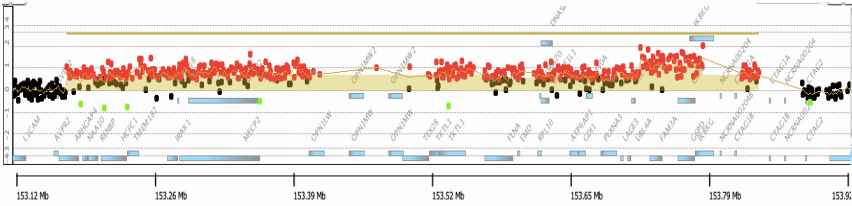
chrX:94345271 (+) AGAGAATTAAGGACAAAACTGTATGATAATTTCAATTAATGCTGAACAAACATTTAATAGAATTCAGCTTC
Junction 2 ATATTATCTATAGATAATAGATATTATCTATTTCAATTAATGCTGAACAAACATTTAATAGAATTCAGCTTC
ChrX:94394842 (-) ATAATATAGATAACAGATATTATCTATCTATATATAATATAGATAACAGATATTATCTATCTATATATAATA

ChrX:94394748

Haplotype Structure 1:



BAB14392

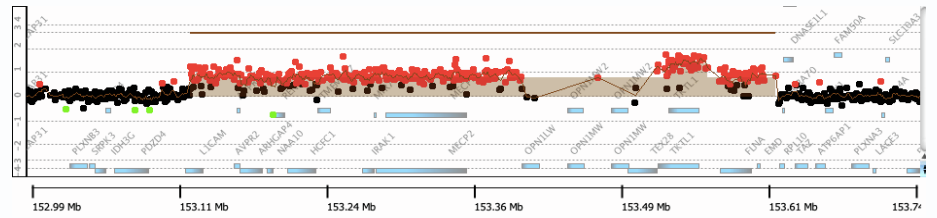


Breakpoint Junction 2:

chrX:153180013 (+) CGGTGGCATGCGCCTGTAGTCCCAGCTACTTGGGAGGCAGGCAGGGGAATCGCTTGAACCCGGGAGGCGGA
Junction 2 CACCTCACCTGGCCTCTTTATTTATCAAATACTGAGGCAGGCAGGGGAATCGCTTGAACCCGGGAGGCGGA
chrX:153722563 (-) CACCTCACCTGGCCTCTTTATTTATCAAATACTGTTTCTGGGCCAGGCACGGTGGCTTATGCCTGTAATCCCA
chrX:153180047
chrX:153722603

Haplotype Structure: *Not Available*

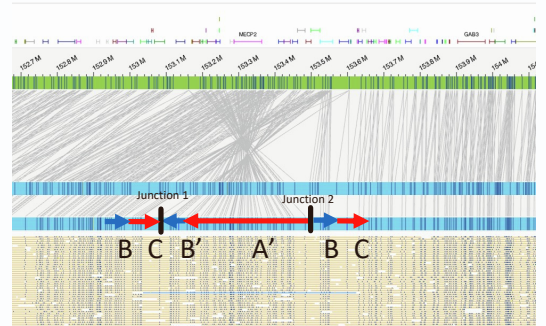
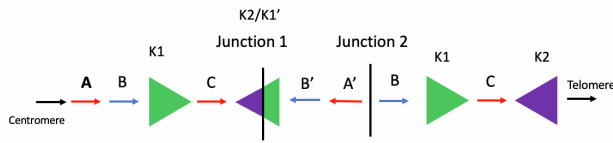
BAB14547



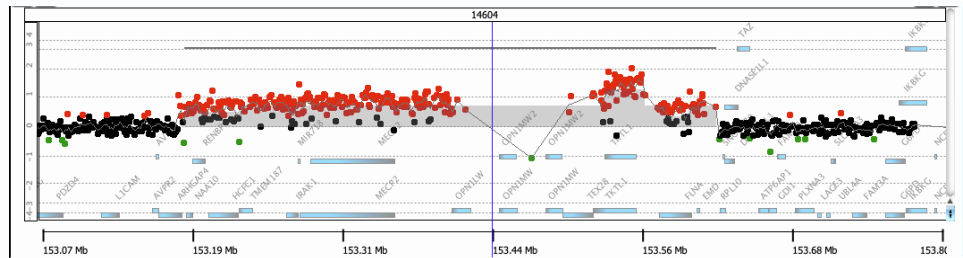
Breakpoint Junction 2:

chrX:153131049 (+) TCAACCCAAGTCTTCCAGGAGAGCGGCTGGCAGGTGGCAAAGCCCCCTCACCATCCTGTCGCTTTACCTCAGTGATCA
 Junction 2 CTCACTGCAACCTCTTGGGTTCAAGCGATTCCTCTTGCCAAAGCCCCCTCACCATCCTGTCGCTTTACCTCAGTGATCA
 chrX:153445956 (-) CTCACTGCAACCTCTTGGGTTCAAGCGATTCCTCTTGCCCTCAGCCTCCCGAGTAGCTGGGATTACAGGCGCCACCACA
 chrX:153445920

Haplotype Structure 3:



BAB14604



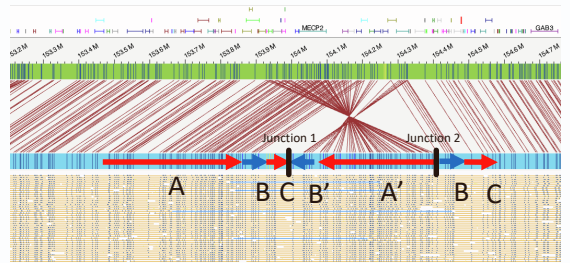
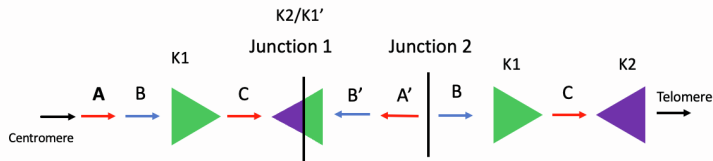
Breakpoint Junction 2:

chrX:153188686

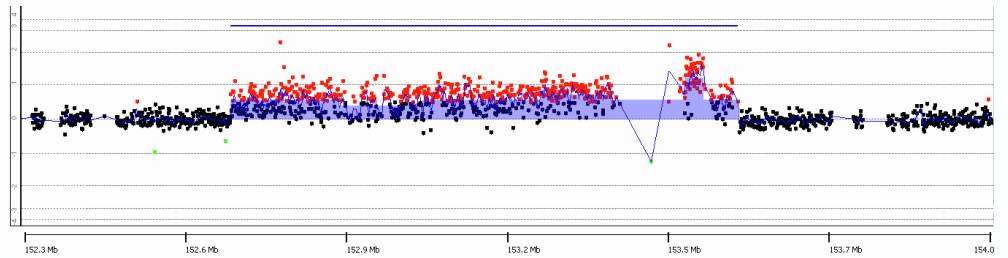
chrX:152841838 (+) CCAGGTGGCAGCTACTGTGCTAGTCCAGGCAAAA**GACGGCAGGATT**CAGACAAGCAGCAGCGAGGGAGGA
 Junction 2 **AGGGAGCGCCCTCTCAGAACCCTCCTTAGACCT****GACGGCAGGATT**CAGACCAGCAGCAGCGAGGGAGGA
 chrX:153078714 (-) **AGGGAGCGCCCTCTCAGAACCCTCCTTAGACCT**GCTTCCTCTCCTCACTTTTCCATTTCCATAGCGC

chrX:153499735

Haplotype Structure 3:



BAB14686



Breakpoint Junction 2:

chrX:152754451

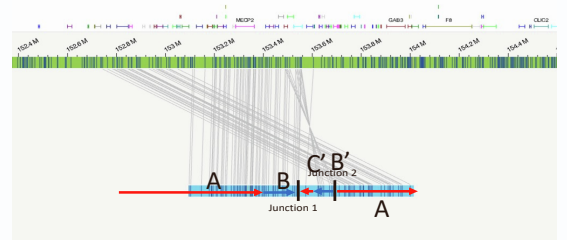
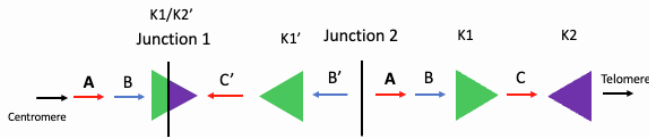
chrX:152754420 (+) CCTTCCCAACTGGGGAGGCAGGGGACTCGCACTTGGGAAATGTACCTGCGGGCTCTTGGGGTTCGTACCTGGC

Junction 2 GCAGAGCTGGAAGAAGCCAAGAGGTTCCACACTTGGGAAATGTACCTGCGGGCTCTTGGGGTTCGTACCTGGC

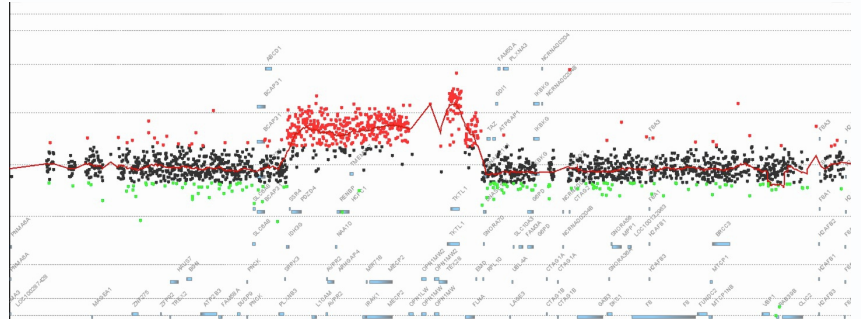
chrX:153441231 (-) GCAGAGCTGGAAGAAGCCAAGAGGTTCCACA TCAGCCTCCAGGAGTCCTATCACAGCCTAAAGGAGAGGTCTC

ChrX:153441203

Haplotype Structure 2:



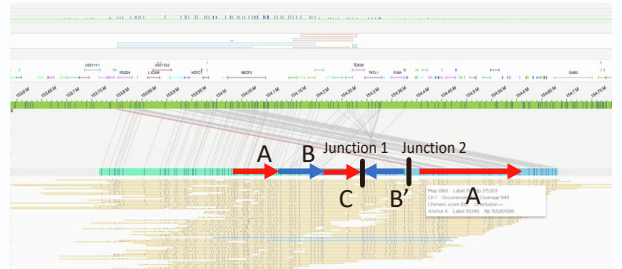
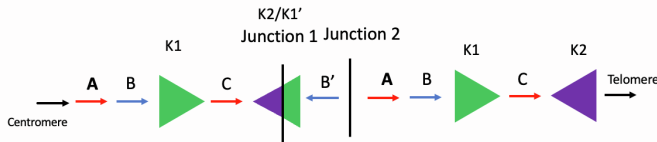
BAB15418



Breakpoint Junction 2:

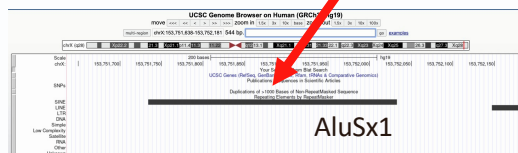
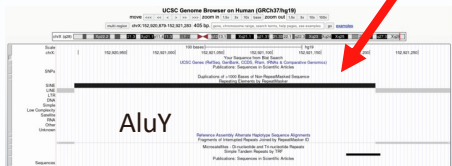
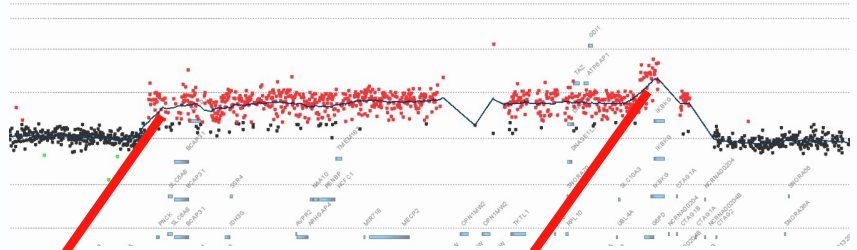
chrX:153054892 (+) GGGAGGCTGAGGCAGGAGAATCGCTTGAAACTGGAAGGCAGAGGTTGCACTGAGCCGAGATCACACCGC
 Junction 2 CTGCCTCAGCCTCTCGAGTATCTGGGACTACTGGAAGGCAGAGGTTGCACTGAGCCGAGATCACACCGC
 chrX:153413610 (-) CTGCCTCAGCCTCTCGAGTATCTGGGACTACAAGCCGTGCATTACCACAATGGCAATTTTTTTTATTTC
 chrX:153413582

Haplotype Structure 4:



BAB15428

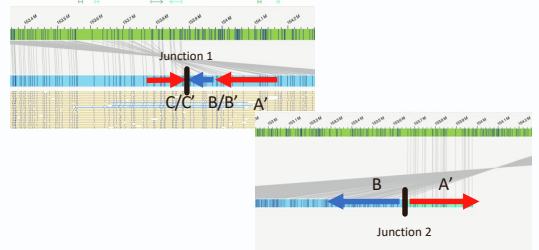
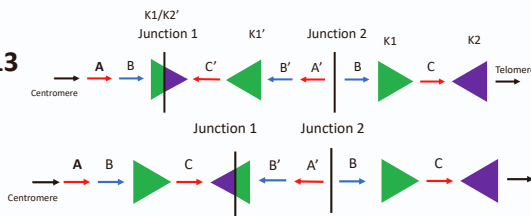
Breakpoint Junction 2:
Within *Alu* Repetitive Elements



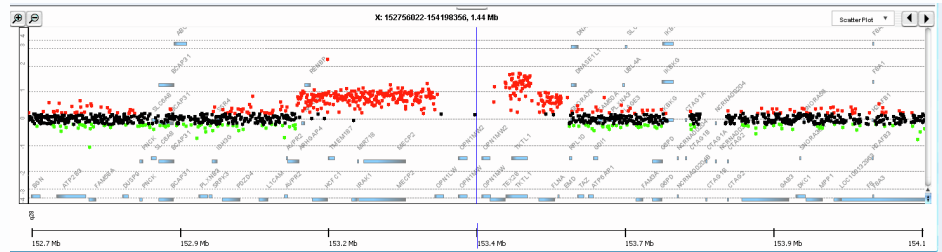
Haplotype Structure 3/13

Or

Haplotype Structure 1



BAB15702

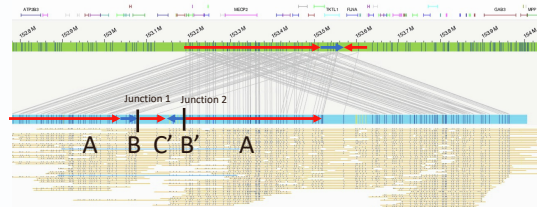
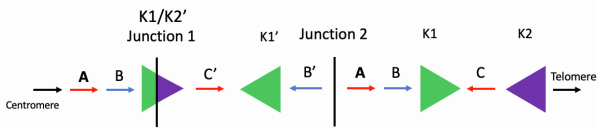


Breakpoint Junction 2:

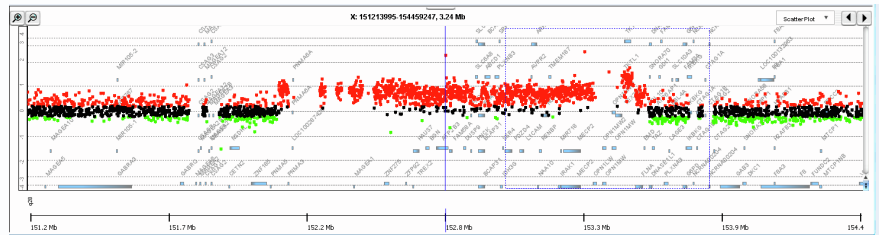
ChrX:153187761

chrX:153187715(+) GATCACAGTGCTGCTCAGGGCAAGCCTGCAGCGATCTCGCAGTCTCAGTGGCTGGGGAAGCTGCCCTCAGGCCCCAGGGGAAGG
 Junction 2 CAATCTCACTCTTGTTCAGTCAGGCTGGAGTGCAGTGGCGCAGTCTCAGTGGCTGGGGAAGCTGCCCTCAGGCCCCAGGGGAAGG
 chrX:153446662(-) CAATCTCACTCTTGTTCAGTCAGGCTGGAGTGCAGTGGCGCAGTCTCGGGTCACTGCAACCTCCGTCTCCCCGGCTCAAGCAATTCT
 ChrX:153446626

Haplotype Structure 6:



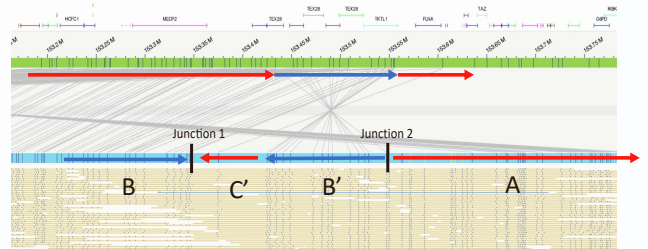
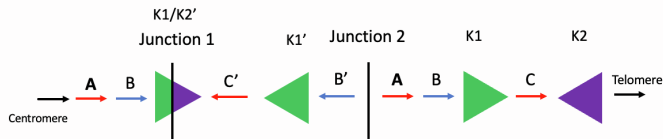
BAB15705



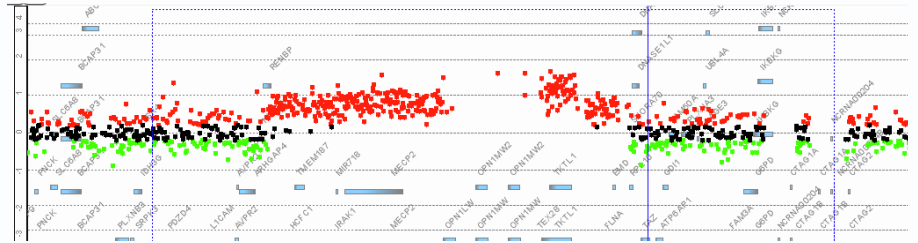
Breakpoint Junction 2:

chrX:152190603(+) ACACAATCTTCGTCAAATTTAAAGGTGTGGGAGGCTGAGG**CAGGAGAATGGCGTGAACCCGGGAGGCGGAGCTTGCA**
 Junction 2 **GTGGCACATGCCTGTAGTCCCTGCTACATGGGAGGCTGAGACAGGAGAATGGCGTGAACCCGGGAGGCGGAGCTTGCA**
 chrX:153431409(-) **GTGGCACATGCCTGTAGTCCCTGCTACATGGGAGGCTGAGACAGGAGGATCGCTTGAGCCCGAGAGTTTTATGTTGCA**
 ChrX:152190650
 ChrX:153468511

Haplotype Structure 2:



BAB15740



Breakpoint Junction 2:

ChrX:153205884

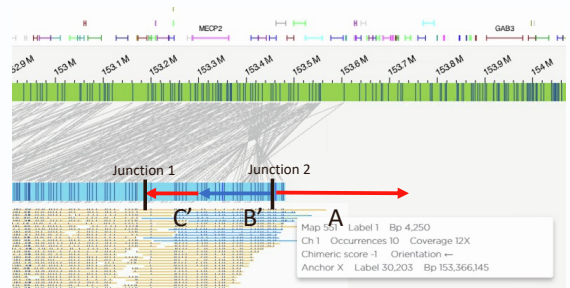
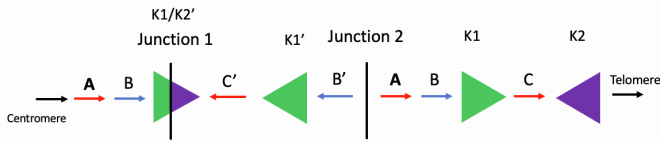
ChrX:153205843 (+) CCTGTAGATTCTTGAATAGACACCAGGGCCCCAGACGAATGGCATATATGTCTGAGCATCAGCGTTTCGCCAGCTCCCTGG

Junction 2 TTGTTTACTGACATCTGAAGAGACTCTTCAAGAATGCAC TGGCATATATGTCTGAGCATCAGCGTTTCGCCAGCTCCCTGG

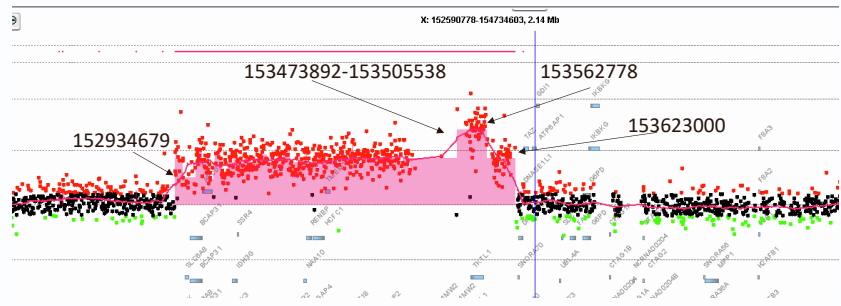
ChrX:153428346 (-) TTGTTTACTGACATCTGAAGAGACTCTTCAAGAATGCAC TGAAGAATATCTTCATGTATCTTTACCAATACATTGATGAAG

ChrX:153428308

Haplotype Structure 2:



BAB15789



Breakpoint Junction 2:

ChrX:152934497 (+)

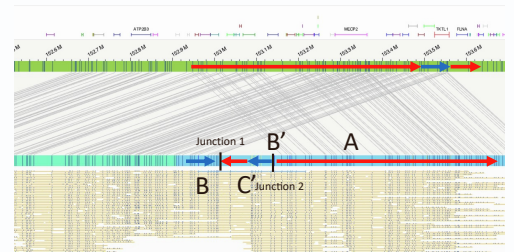
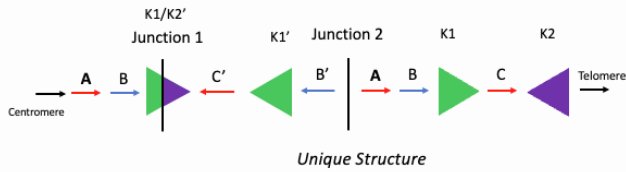
ChrX:152934457 (+) GTTGGTGGGGCTGGGGCCACTCAGACCAGAGGACAGGCC**CACGGCTCTCCACTTCAGAAGCCACCGCCAAGCTGCCACTG**

Junction 2 **TTCAGCCCTGAGTGGAGCACAGCC**TGCCCCAGGCAGAC**CACGGCTCTCCACTTCAGAAGCCACCGCCAAGCTGCCACTG**

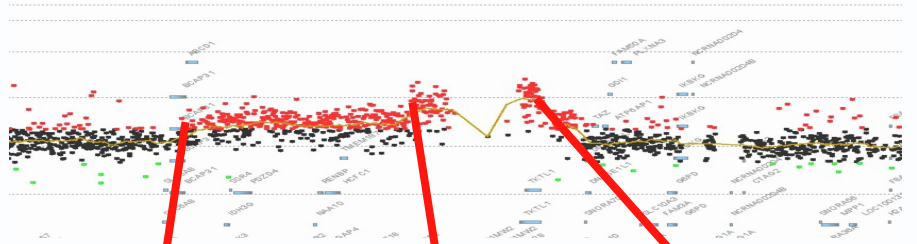
ChrX:153421664 (-) **TTCAGCCCTGAGTGGAGCACAGCCGGCCCCAGGCAGACA**TGGCTGCCTCAGGCTGCCCTGGGGGAGTTCTAGAATTG

ChrX:153421627 (-)

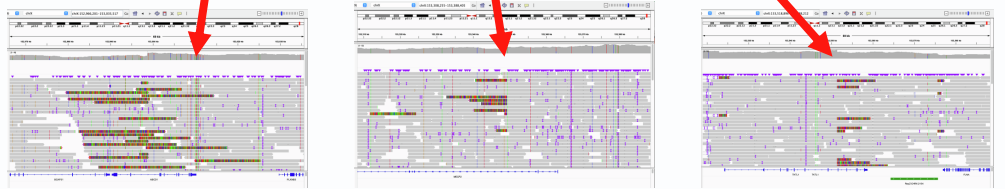
Haplotype Structure 2:



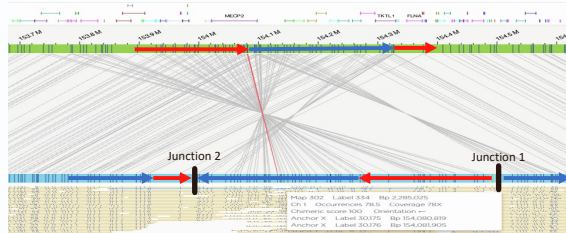
BAB15421



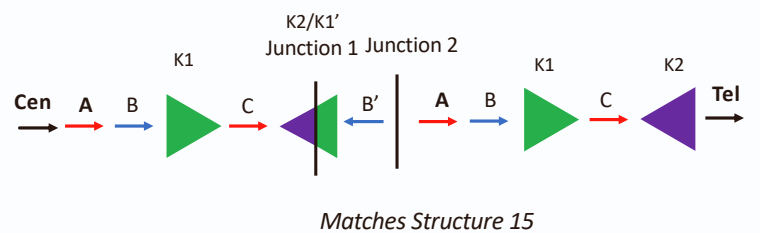
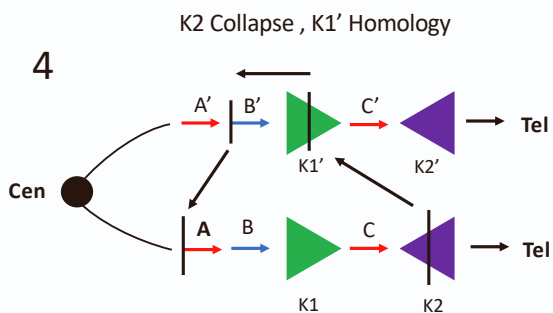
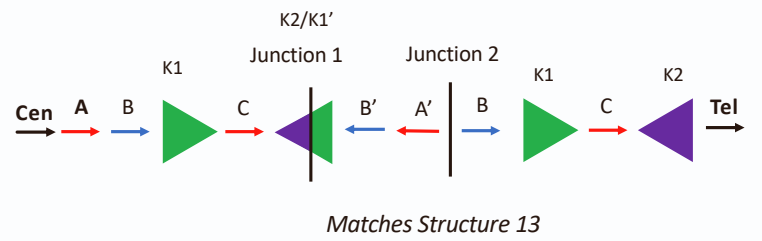
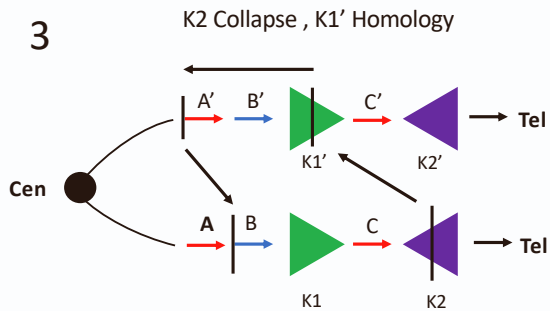
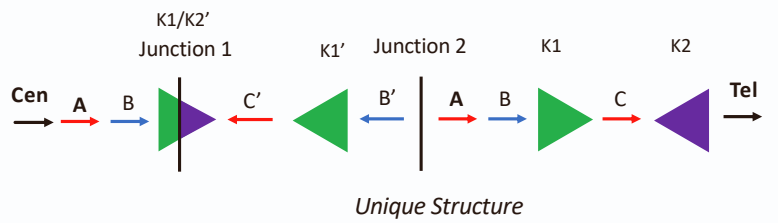
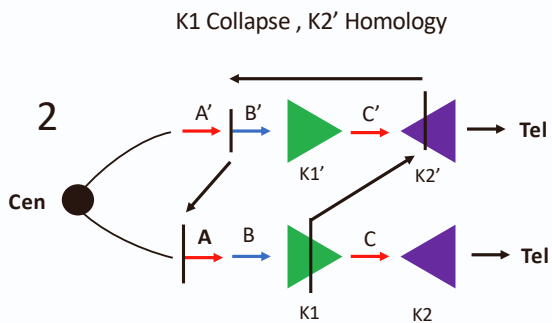
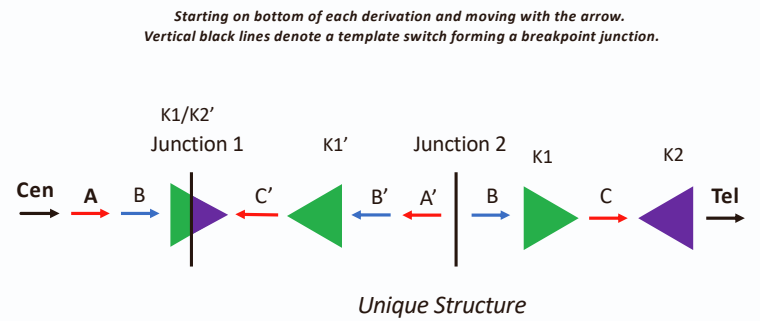
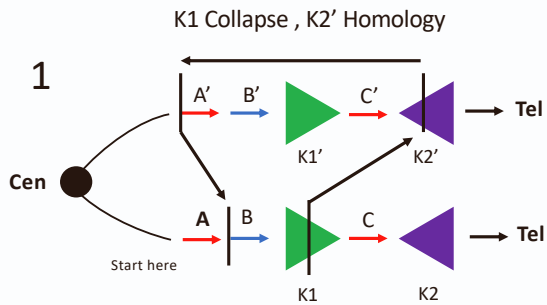
Breakpoint Junction 2:
Complex Unresolved

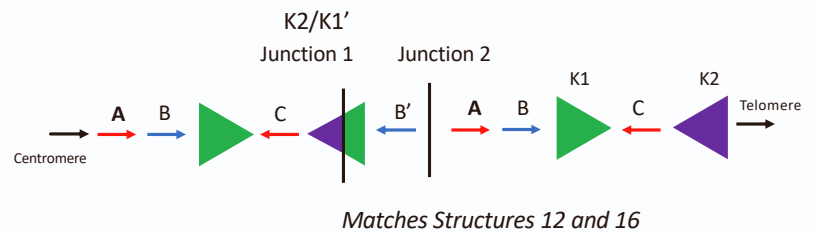
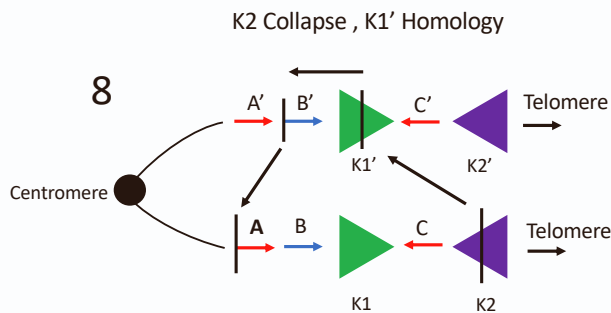
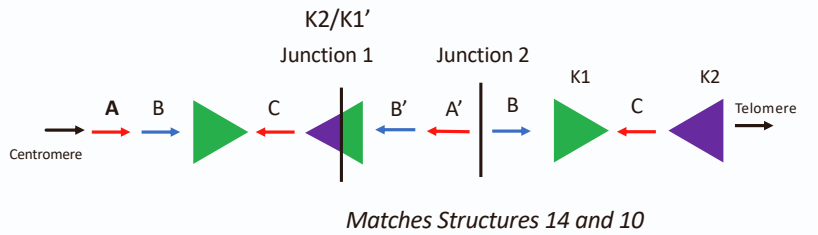
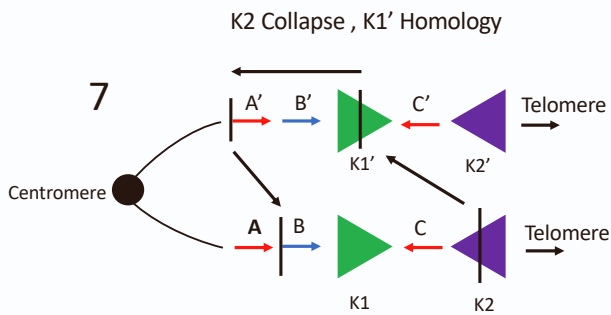
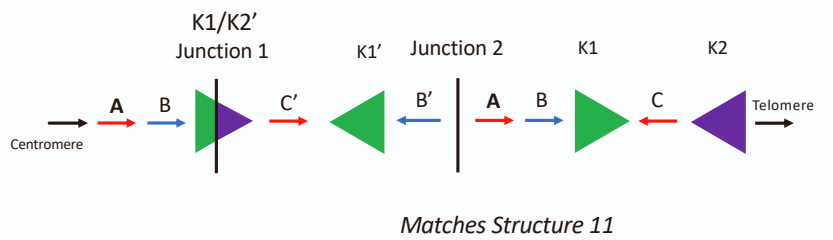
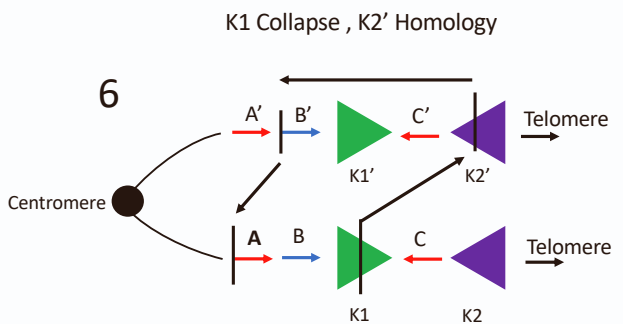
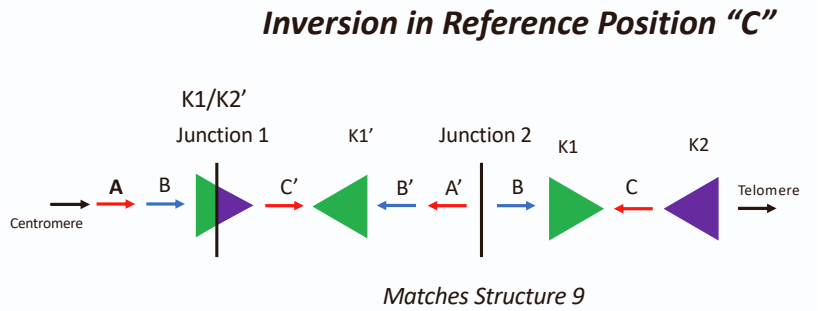
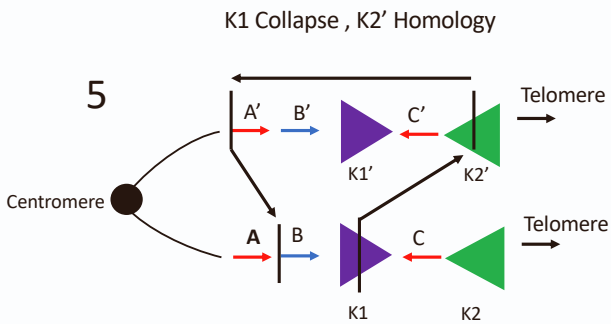


Haplotype Structure: *Complex Unresolved*



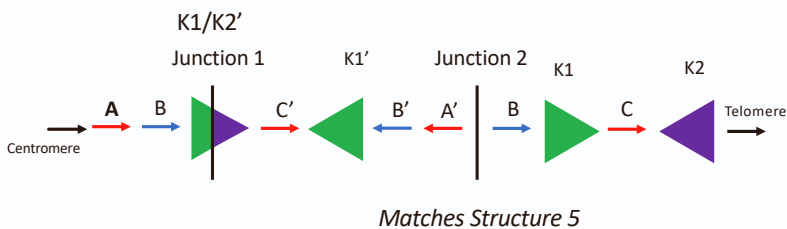
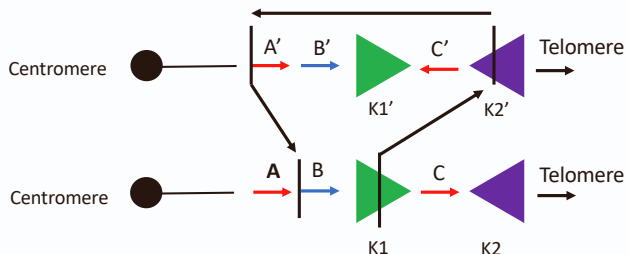
Data S2: Structural variant haplotype possibilities within a DUP-TRP/INV-DUP event during and interchromosomal and intrachromosomal template switch between inverted LCRs, related to Figure 2.



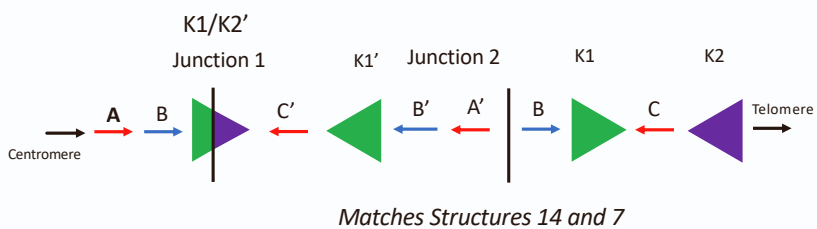
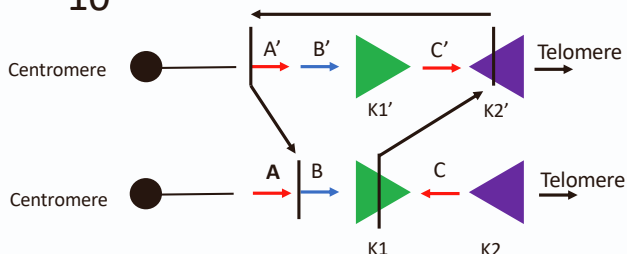


Interchromosomal Event Mixture of Inversion "C" 1/2

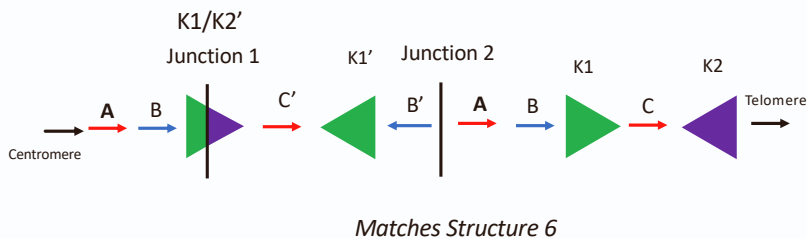
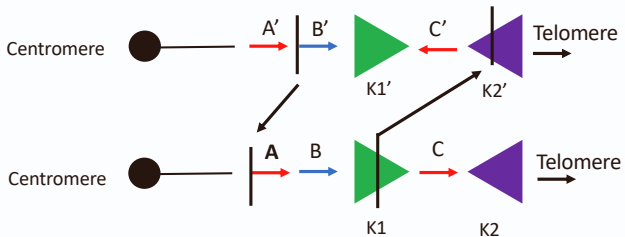
9



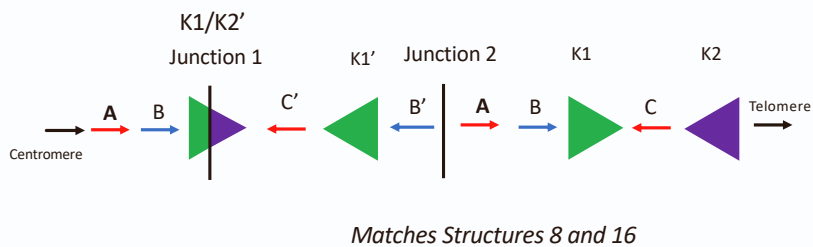
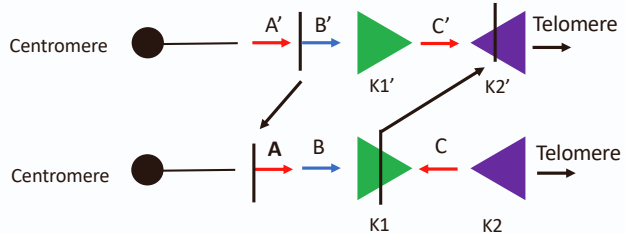
10



11

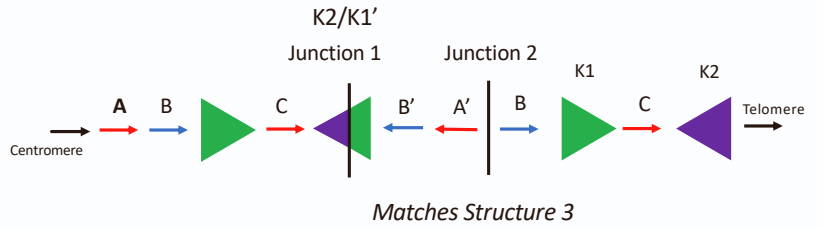
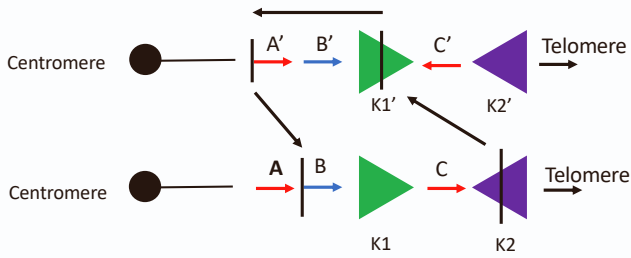


12

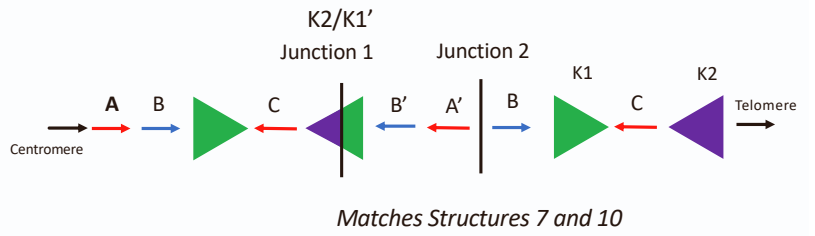
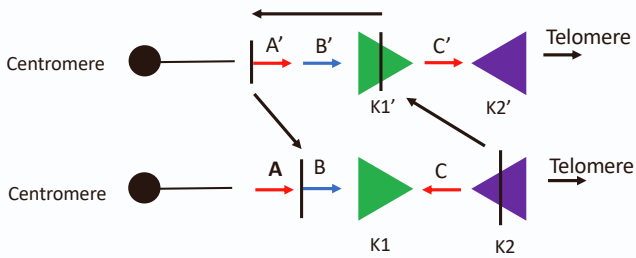


Interchromosomal Event Mixture of Inversion "C" 2/2

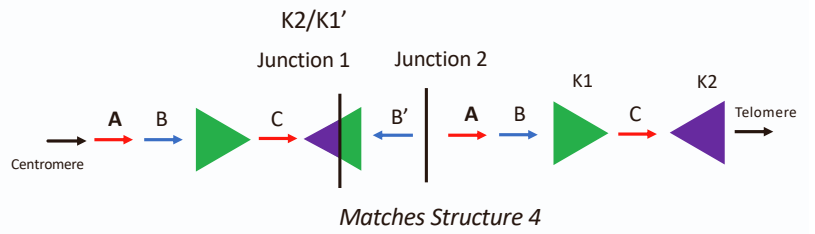
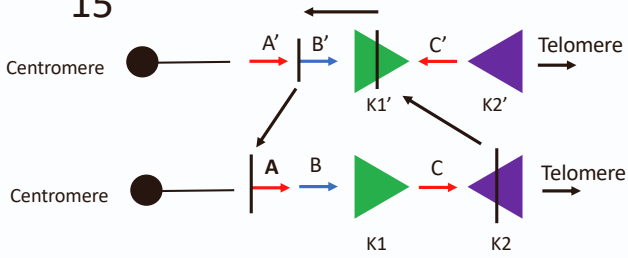
13



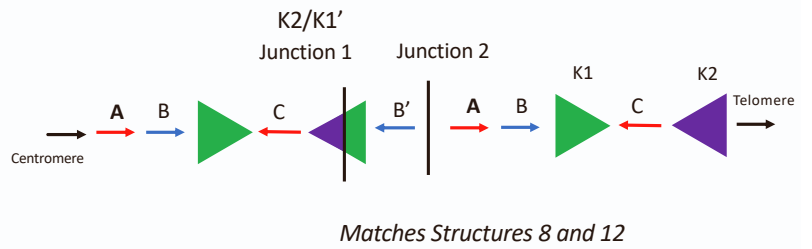
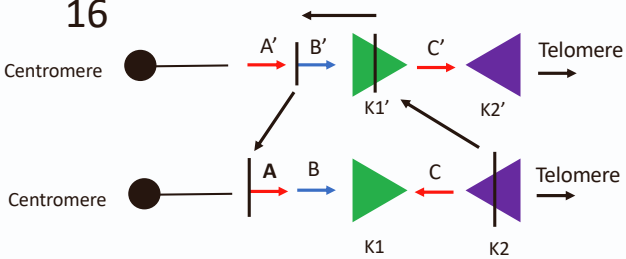
14



15



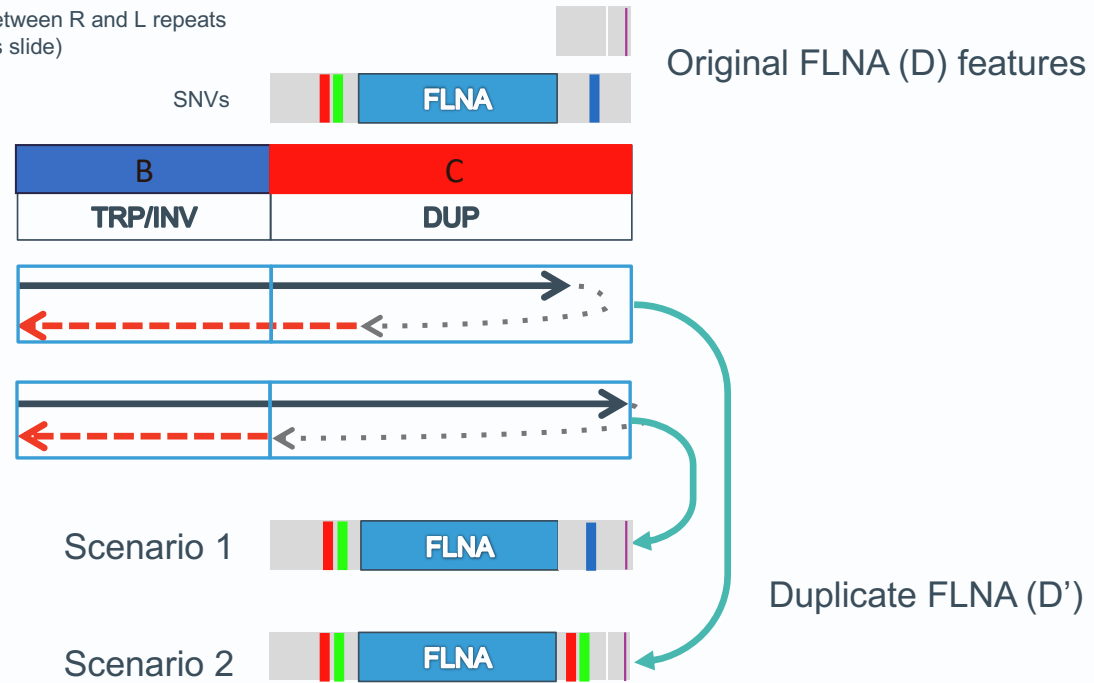
16



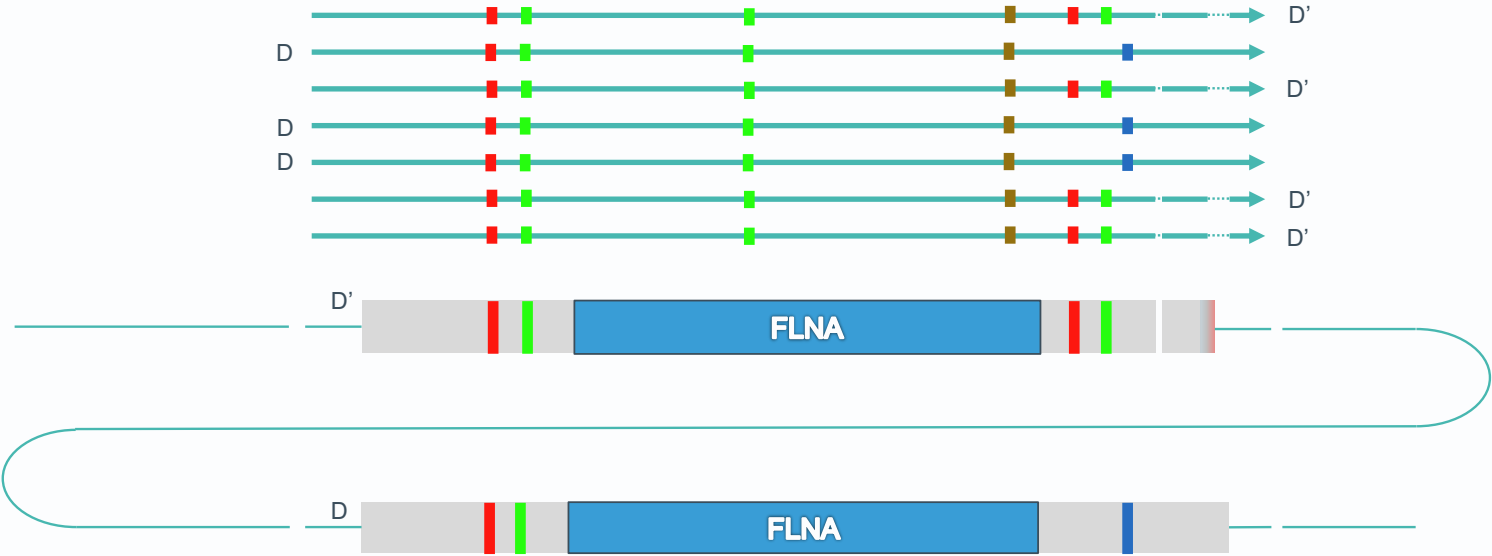
Data S3: CRISPR-Cas9 targeted ONT data for BAB3114 family, Related to Figure 5.

COMPARING THE DUPLICATED FLNA INVERTED REPEATS

Differences between R and L repeats
(from previous slide)



GENERATE PHASED VARIANTS AND HAPLOTAG



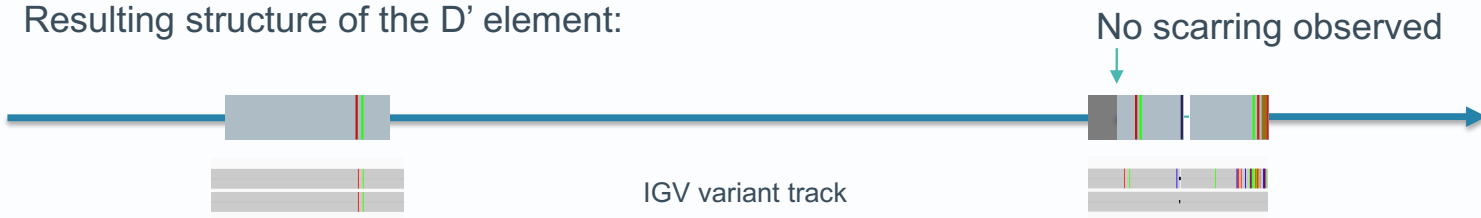
BAB3114 HAPLOTYPING SEGREGATES READS INTO D AND D'



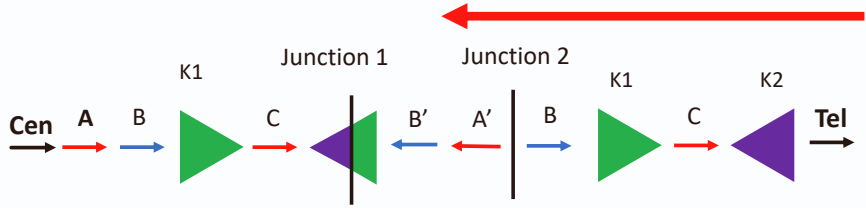
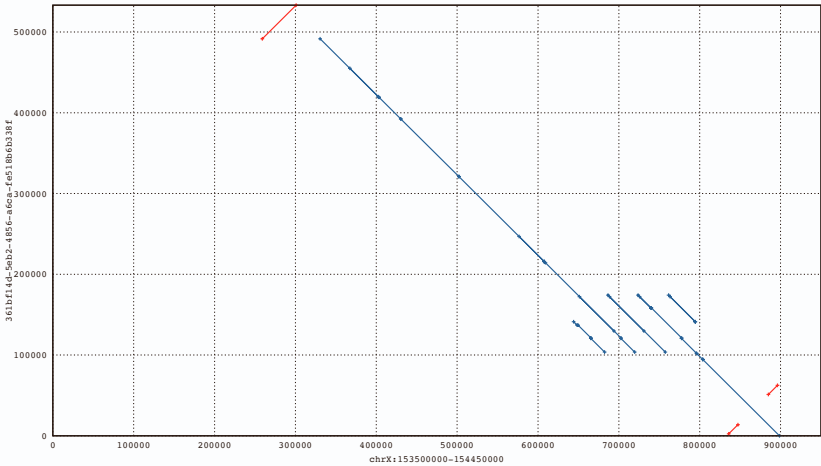
BREAKPOINT RESOLUTION CONCLUSION



Resulting structure of the D' element:



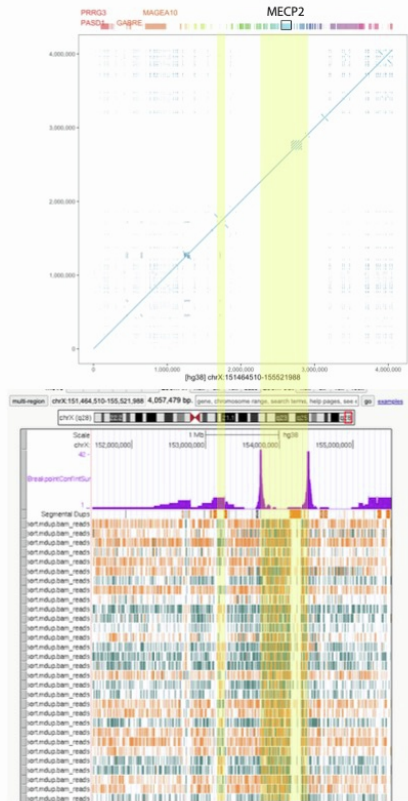
A single 530 kb read resolves opsin cluster allele in BAB3114



Single 530 kb Nanopore read spanning the region

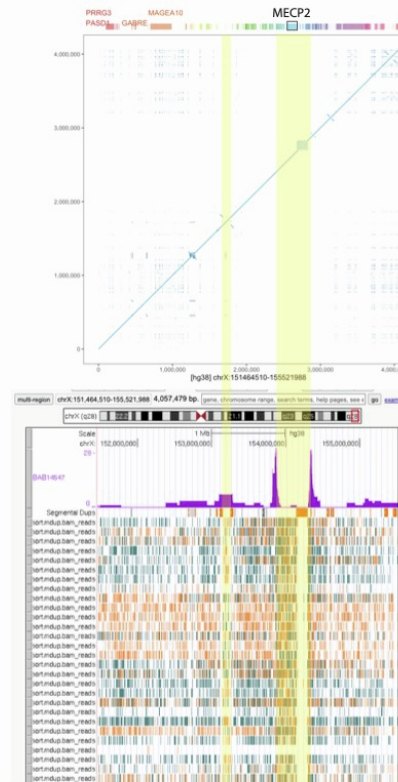
Data S4: Strand-Seq data analysis techniques for BAB14547 and BAB3114, related to Table 1.

BAB3114

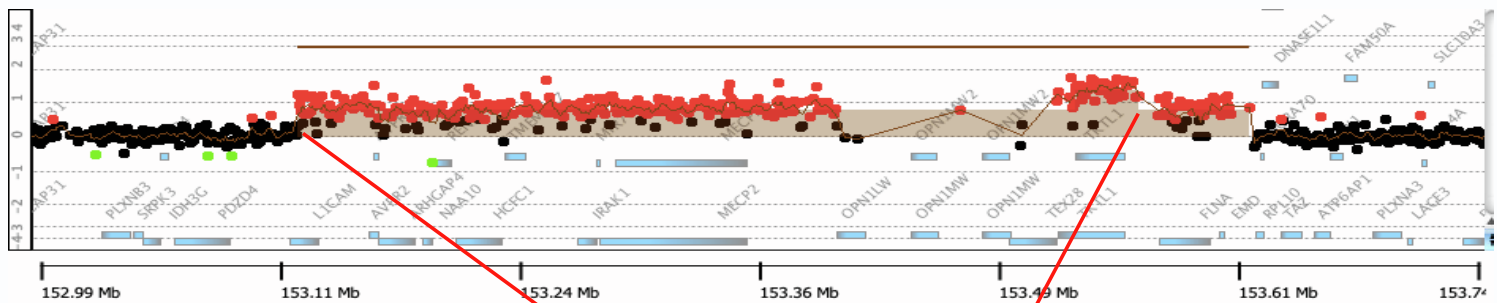


Inv InvDup

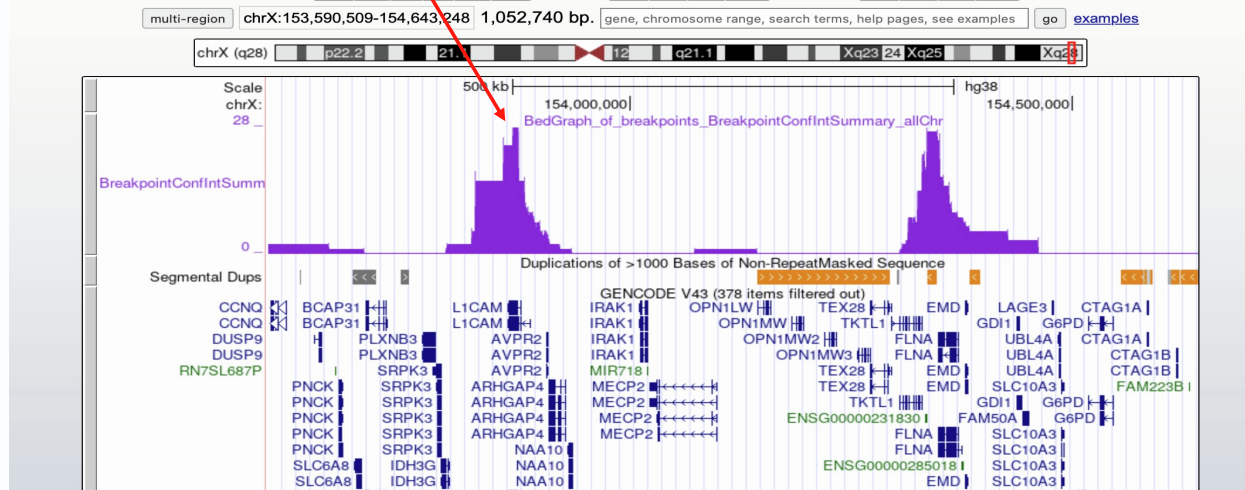
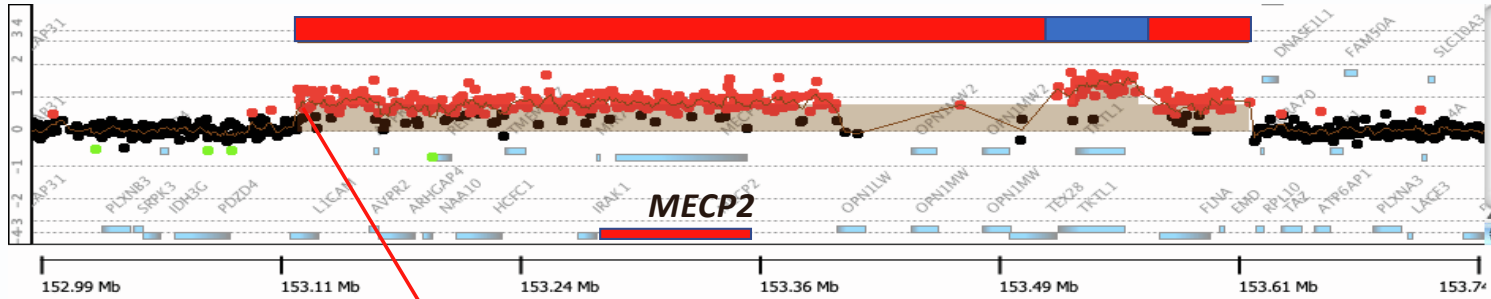
BAB14547

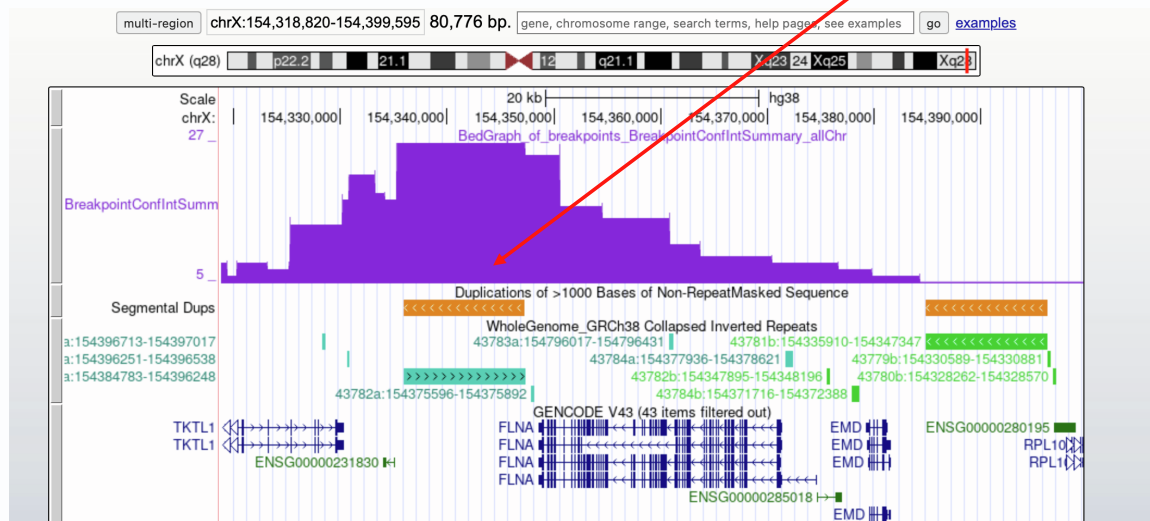
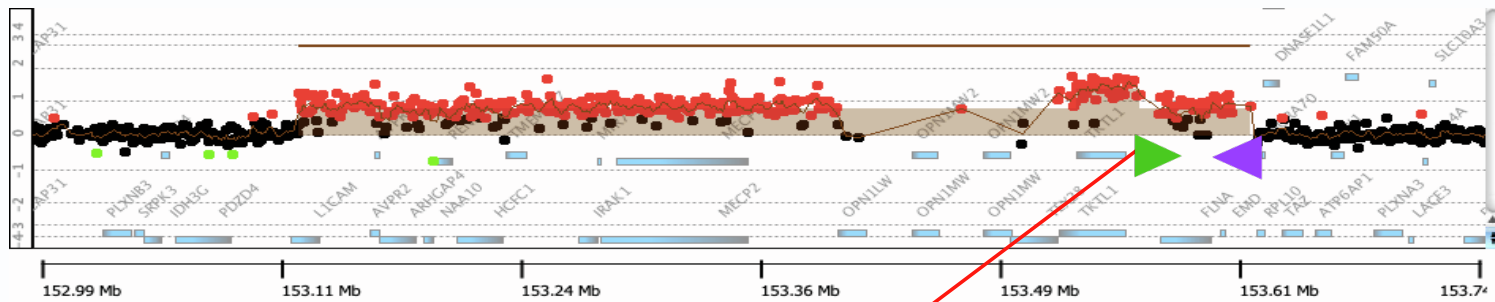


Inv InvDup

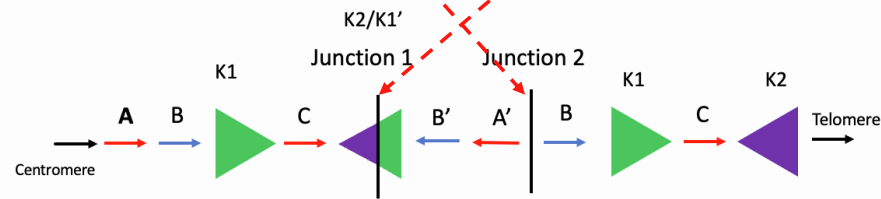
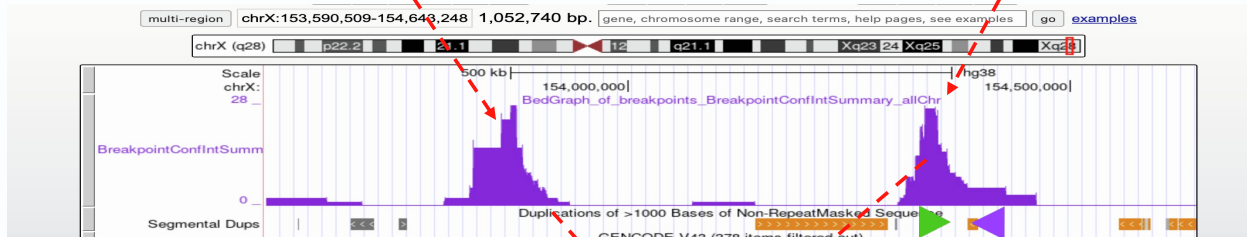
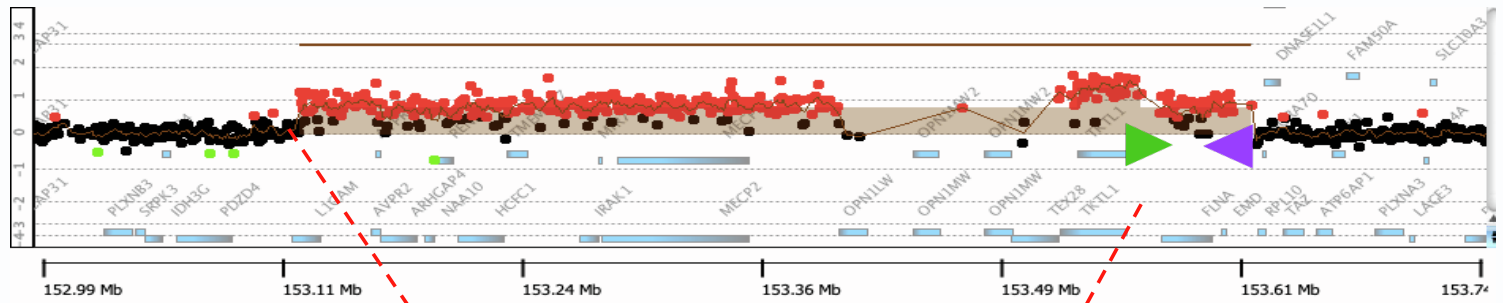


BAB14547



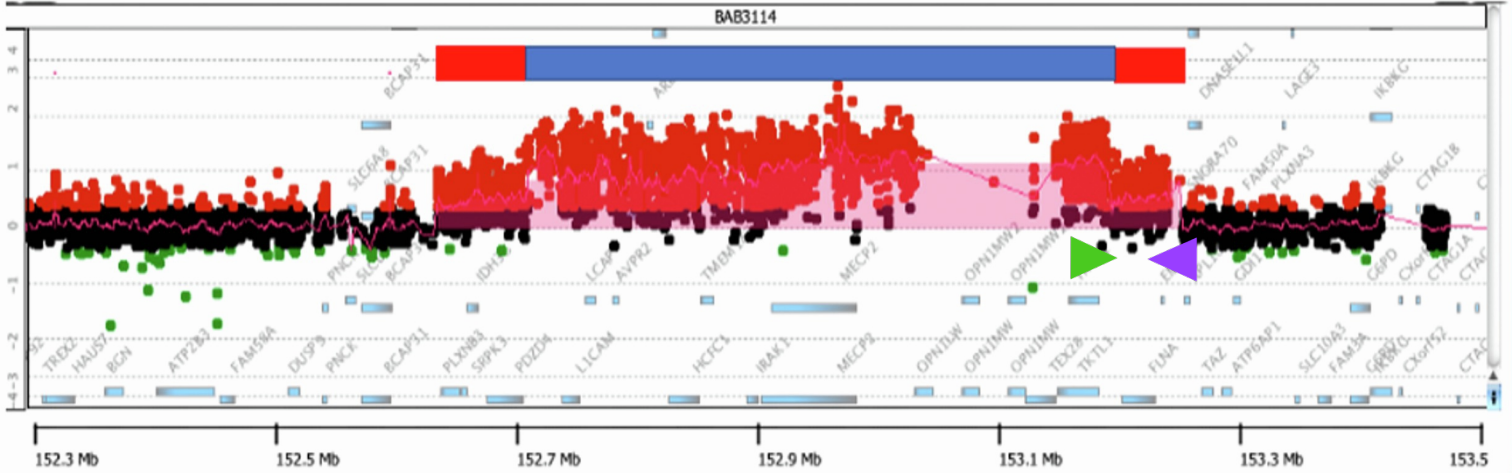


First LCR K1

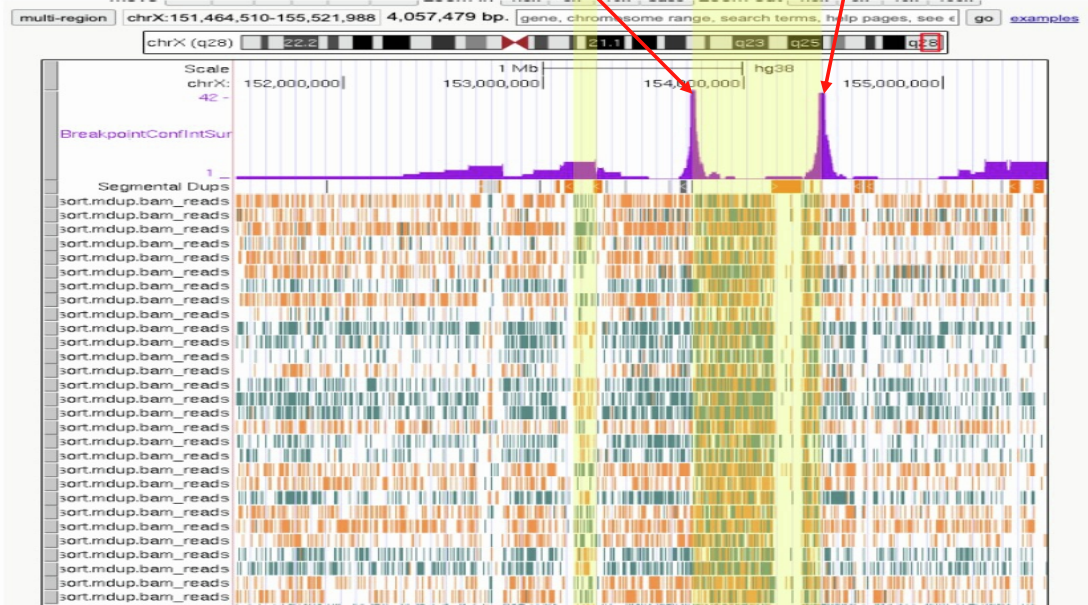
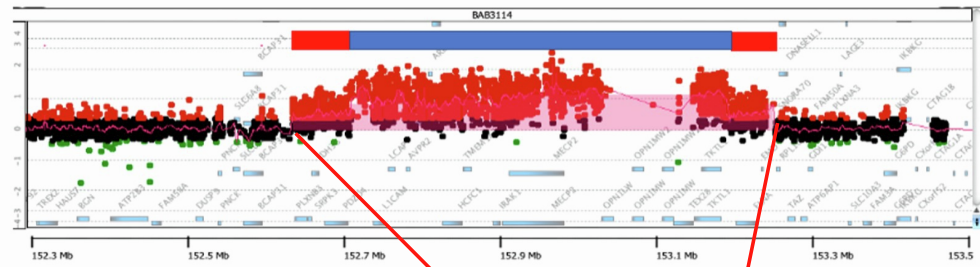


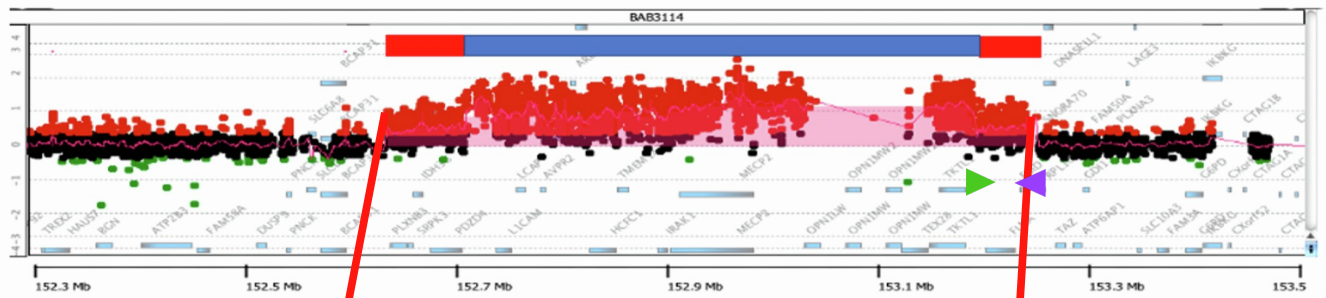
Matches
Haplotype
Structure

BAB3114



Array Comparative Genomic Hybridization

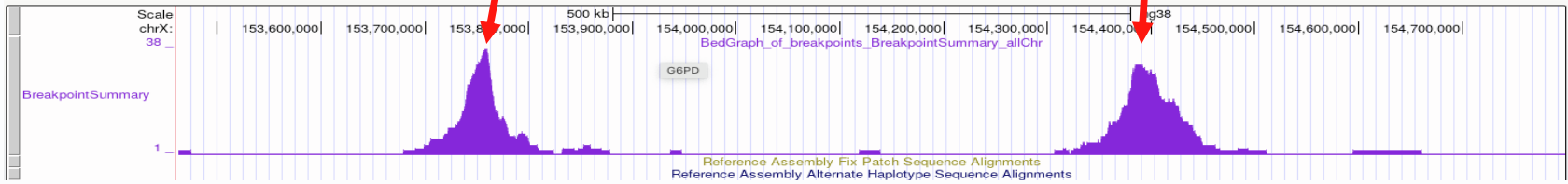
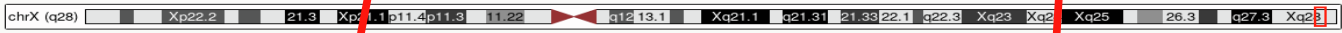




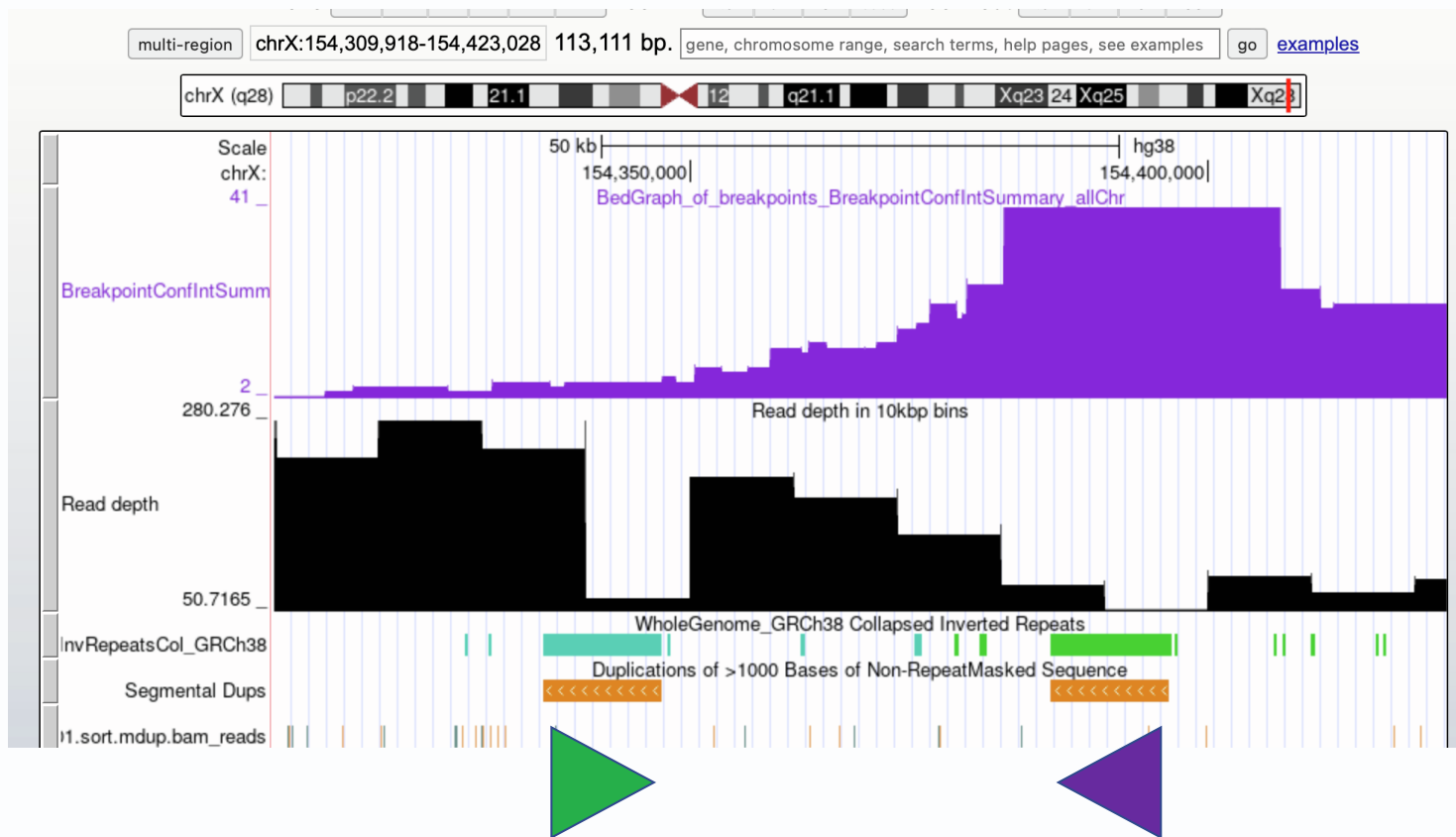
UCSC Genome Browser on Human (GRCh38/hg38)

move << < > >> zoom in 1.5x 3x 10x base zoom out 1.5x 3x 10x 100x

multi-region chrX:153,461,456-154,799,347 1,336,892 bp. gene, chromosome range, search terms, help pages, see examples go examples



BAB3114



multi-region chrX:154,329,488-154,397,941 68,454 bp. | gene, chromosome range, search terms, help pages, see examples | go [examples](#)

chrX (q28) | p22.2 | 21.1 | 12 | q21.1 | Xq23 24 Xq25 | Xq28

