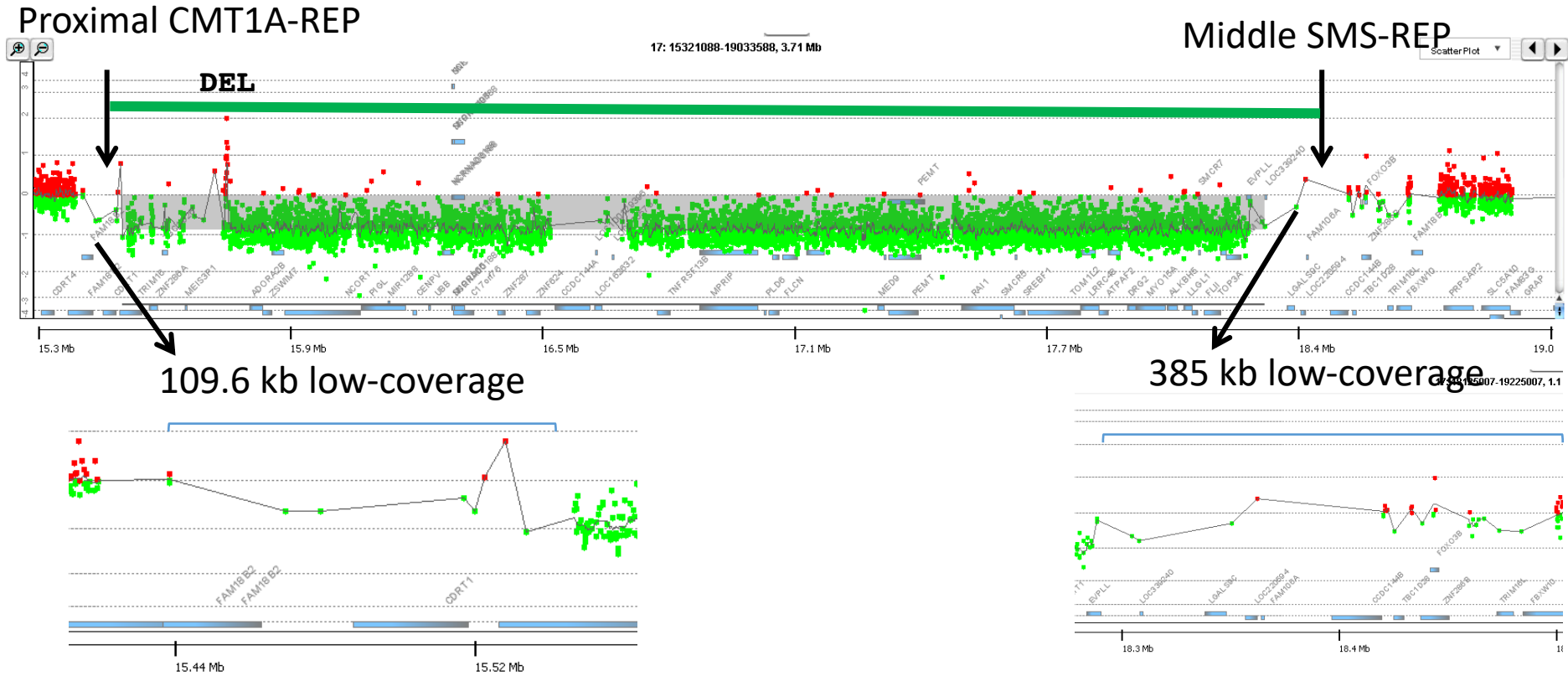
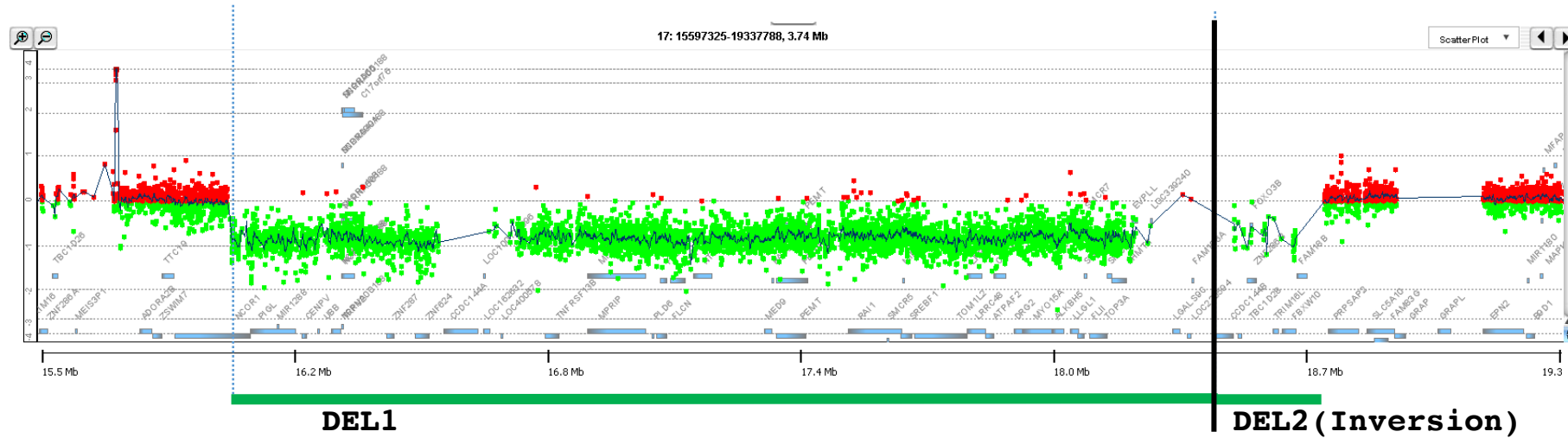


BAB572



Precise junction not mapped

BAB641



16066603

← Ref_NCOR1 LCR(LINE) (+) AAAATATCTATTAAGTCATACATCTGATGAGG**aACtAATGT**ccagaatataaaagaa

BAB641 AAAATATCTATTAAGTCATACATCTGATGAGG**-ACaAATGT**GTGATGGCATGTATATA

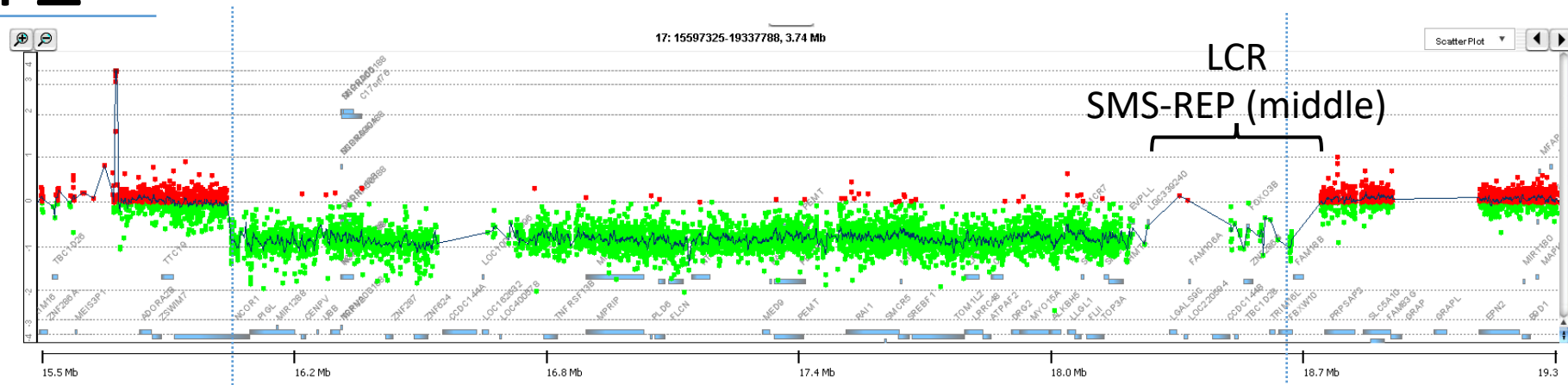
← Ref_CCDC144B (-) cactcttggtttgtacattctatggtggt**GG-ACaAATGT**GTGATGGCATGTATATA

Middle- SMS-REP

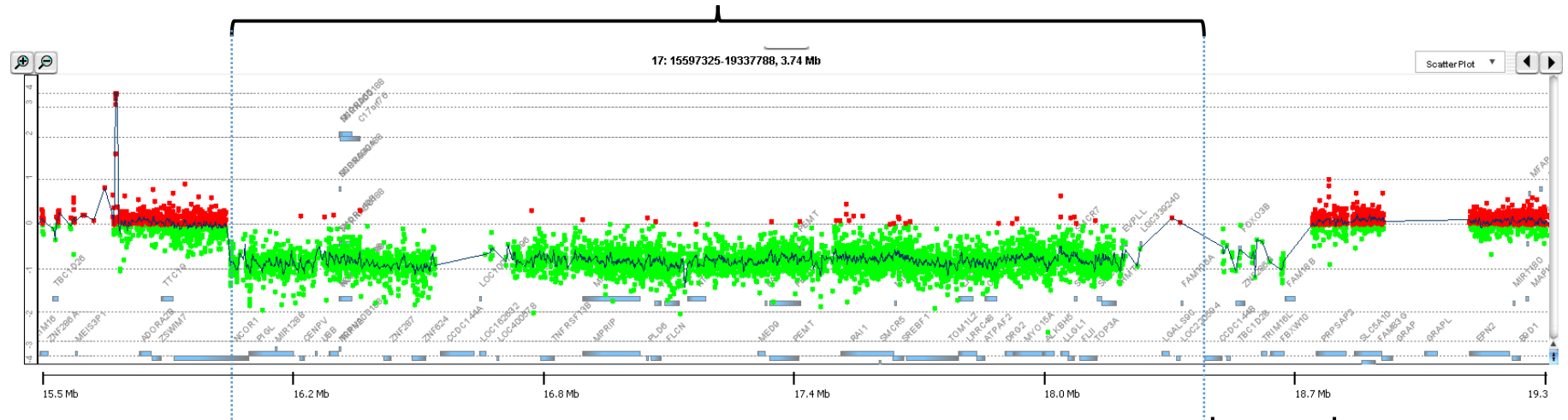
18517335

Jct contains inverted breakpoint between beginning of DEL1 and beginning of DEL2

BAB641



Jct obtained by PacBio and Sanger



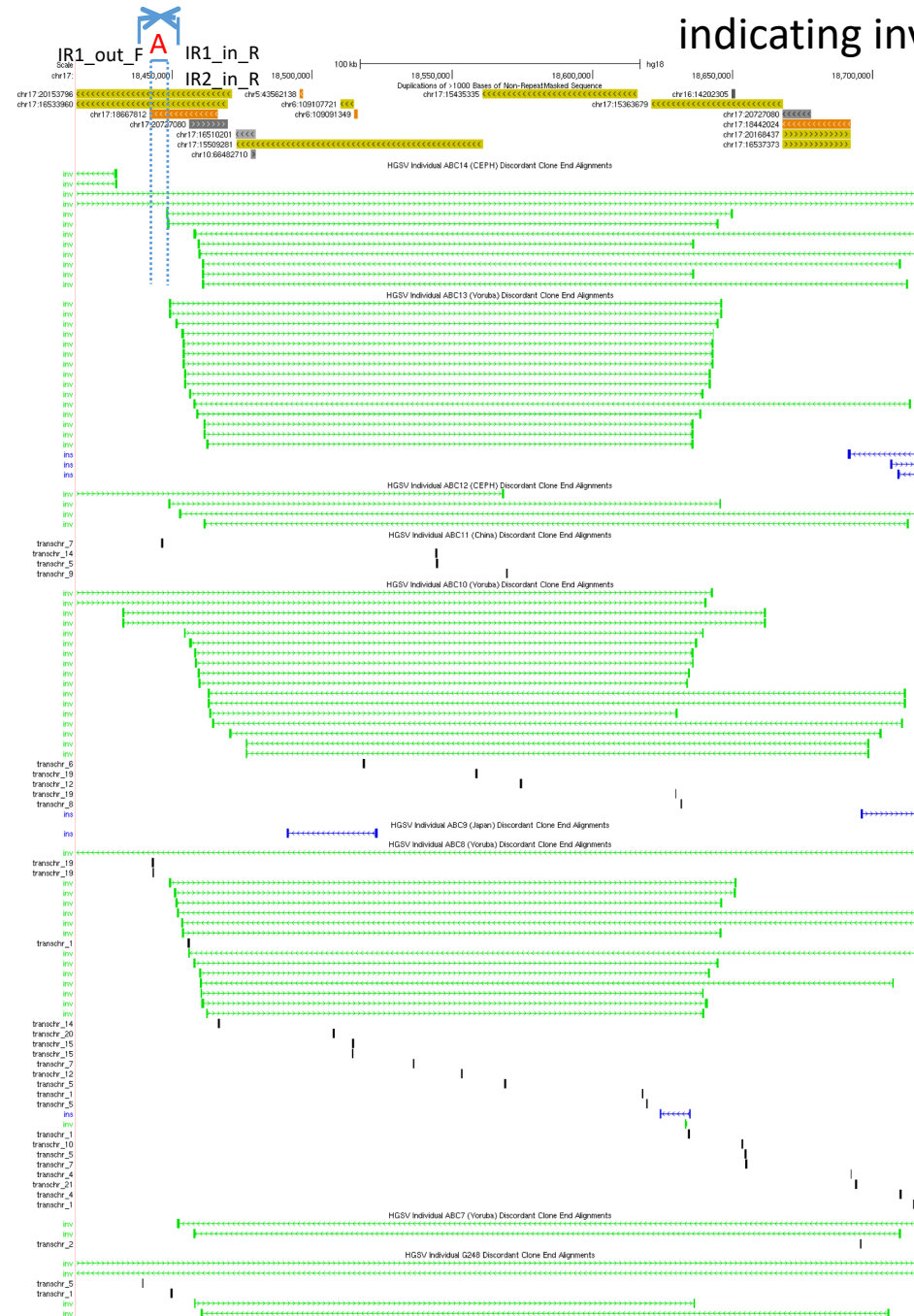
PacBio detected an INV not DEL:

Inherited inversion? ->

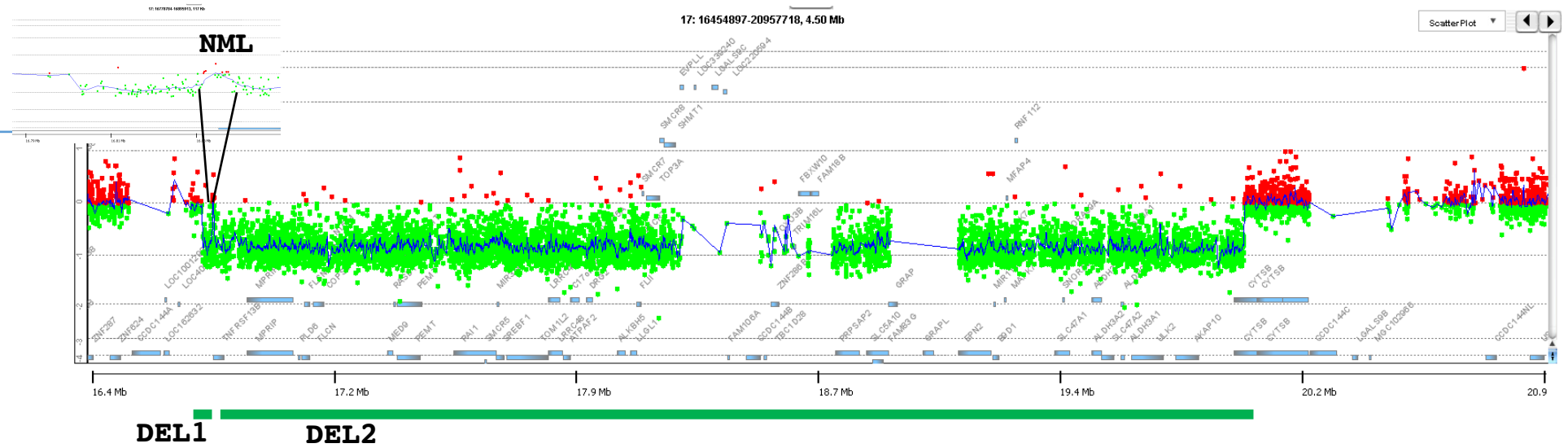
Deletion appears *de novo*; flanked by 99% identical IRs (IR1 and IR2)

BAB641

Seven individuals have discordant fosmid clones (HGSV track) indicating inversion between IR1 and IR2 is common



BAB649



16808862

LCR(LINE) (+) CGTCTTgtgtctctGTGTCTCTATGTTTAG**CTCCaATT**tataagtgagaacatacagtat

BAB649_jct1 CGTCTT GTGTCTCTATGTTTAG**CTCCaATT**AGACAGTTGCCATTGCTGCACA
 16808831 16808838

← TNFRSF13B (LINE) (-) acagggtttctgtttggtgataaaaattgt**CTCCaATT**AGACAGTTGCCATTGCTGCACA

de novo 8 bp del

16845031

16838686

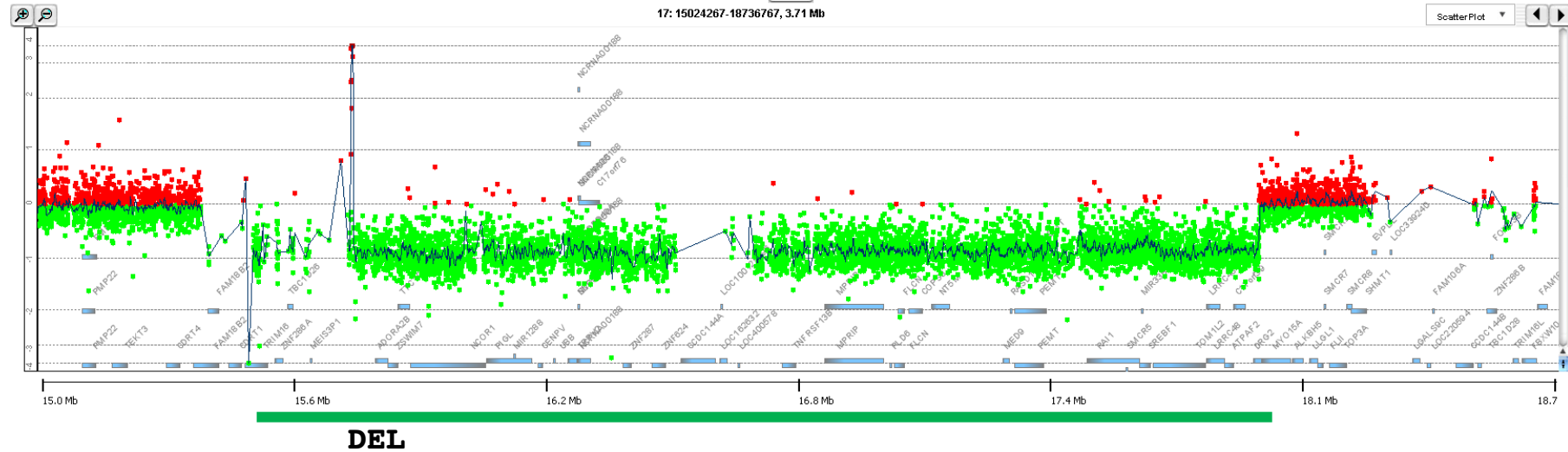
← TNFRSF13B (-) GGTGACCTCATGCAGGGGTTTGTCTTGGAGCTTGCTTGCcgccatctgtagtggggatg

BAB649_jct2 GGTGACCTCATGCAGGGGTTTGTCTTGGAGCTTGCTTGCtgctGGAACGGTGGACAAAG

→ SPECC1 (+) aaccttaggcattctggaactgaagaggtgctagggcagctcagTGAACGGTGGACAAAG

20020394

BAB765



15441705

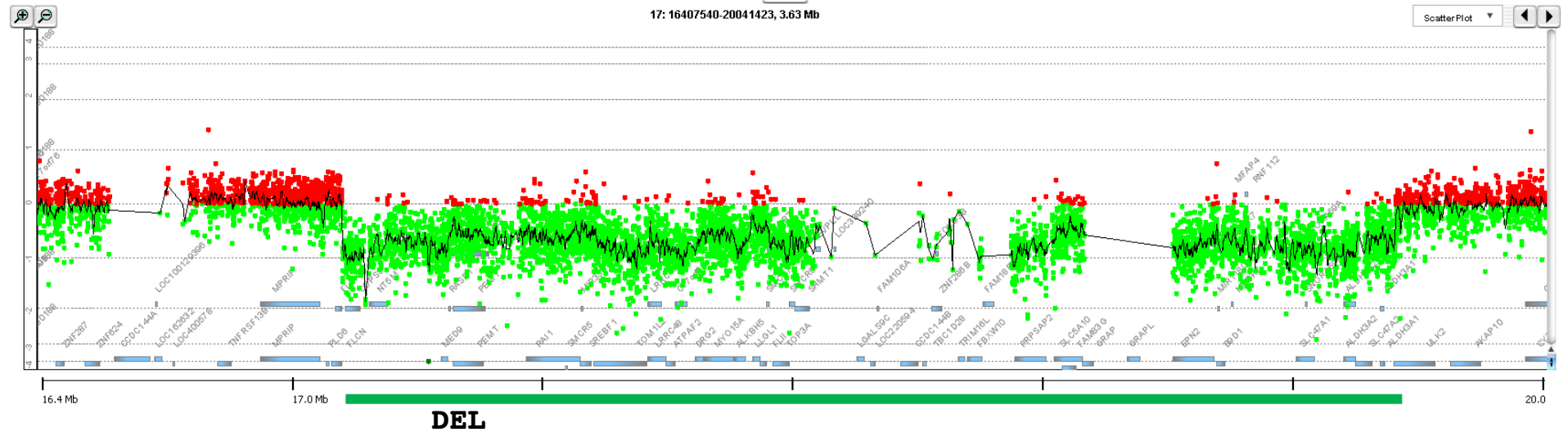
AluY (LCR) (+) AGGCCGAGGAGGGTGGATCATGAGGTCAGGAGATCGAGACcctcctggctaacacagt

BAB765 AGGCCGAGGAGGGTGGATCATGAGGTCAGGAGATCGAGACTATCCTGGCCAACATGGT

AluSg_{DRG2} (+) gggctgtggcGGGTGGATCATGAGGTCAGGAGATCGAGACTATCCTGGCCAACATGGT

18005708

BAB1354



17141946

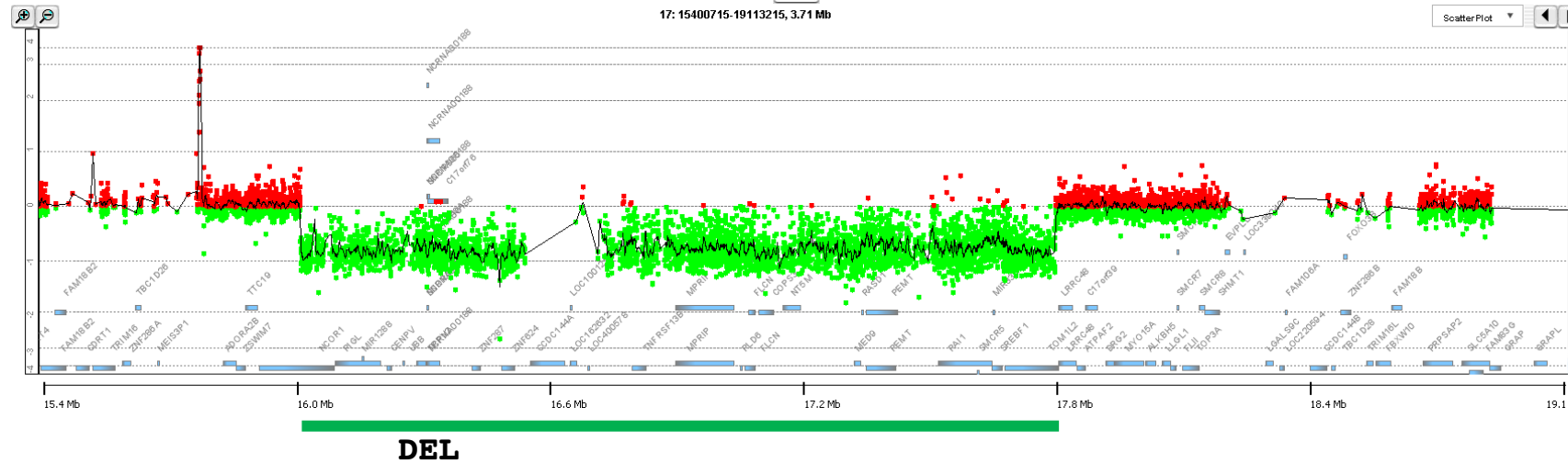
AluSc (+) **AAAAATTAGCTGGGCGTGGTGGCACACGCCTGTAGTCCAGCTACTC**tggtggctgaggcagg

BAB1354 **AAAAATTAGCTGGGCGTGGTGGCACACGCCTGTAGTCCAGCTACTC**GGGTGGCTGAGGCGGG

← AluY_ULK2 (+) **aaaaatcagccgggcgtggtggcaggCGCCTGTAGTCCAGCTACTC**GGGAGGCTGAGGCGGG

19676163

BAB2564

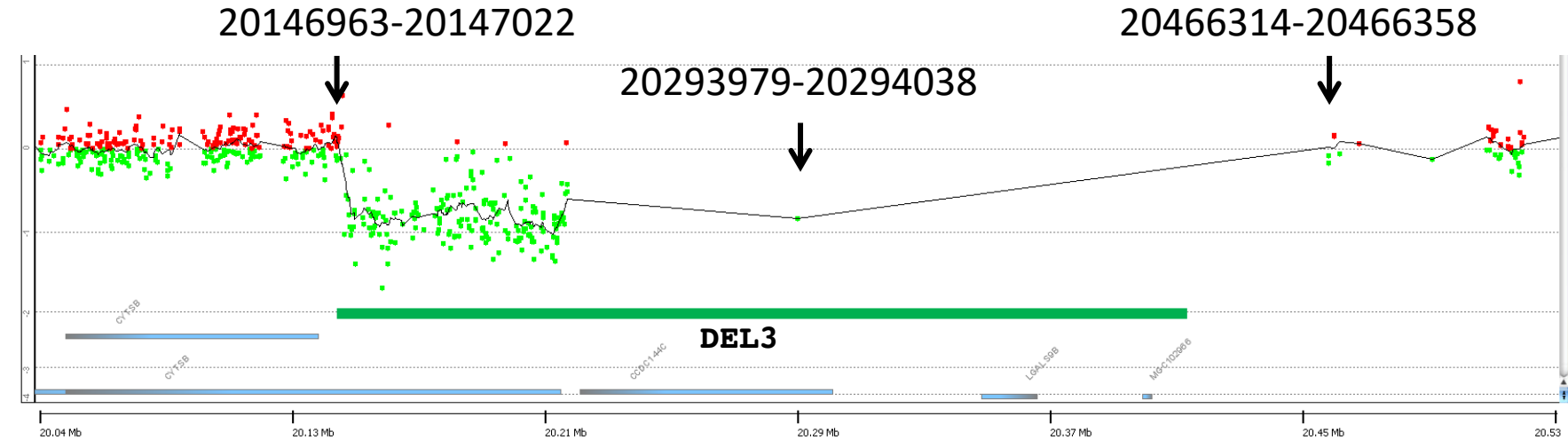
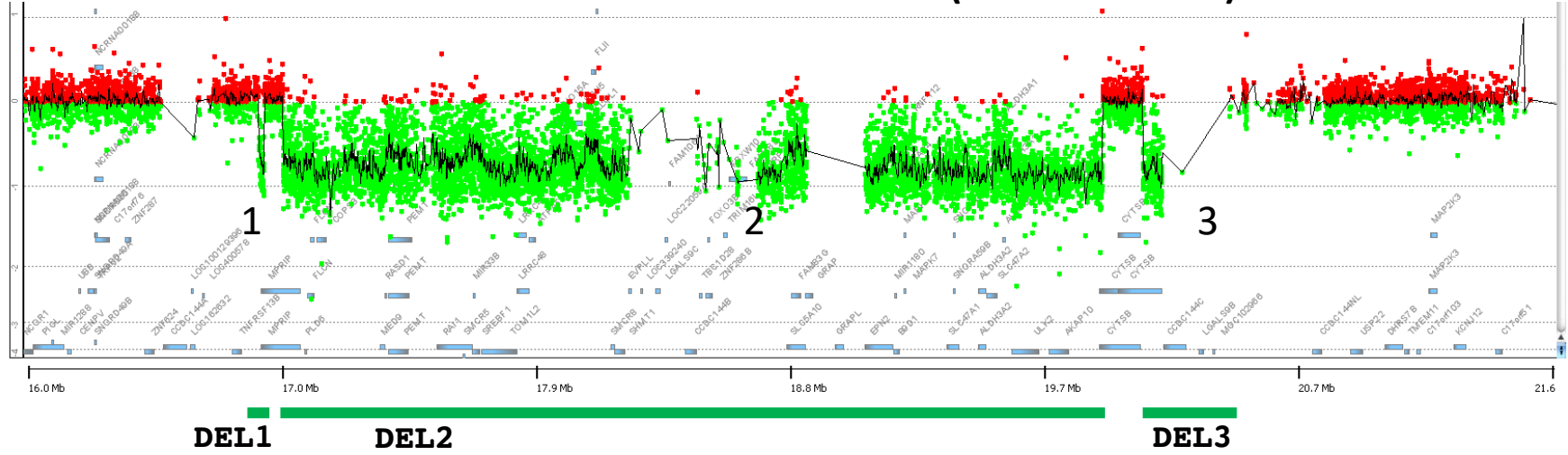


← Ref_NCOR1 (+) CATTACTTCAACAAAACATAT**AGAgAACCAc**actcactctccatggaatggtaaca
 BAB2564 CATTACTTCAACAAAACATAT**AGAgAACCAc**-CATCCTTTTCCATTTCTACACAAAC
 ← Ref_TOM1L2 (+) ttgctcaagttgtctctgcca**AGA-AACCAa**aCATCCTTTTCCATTTCTACACAAAC

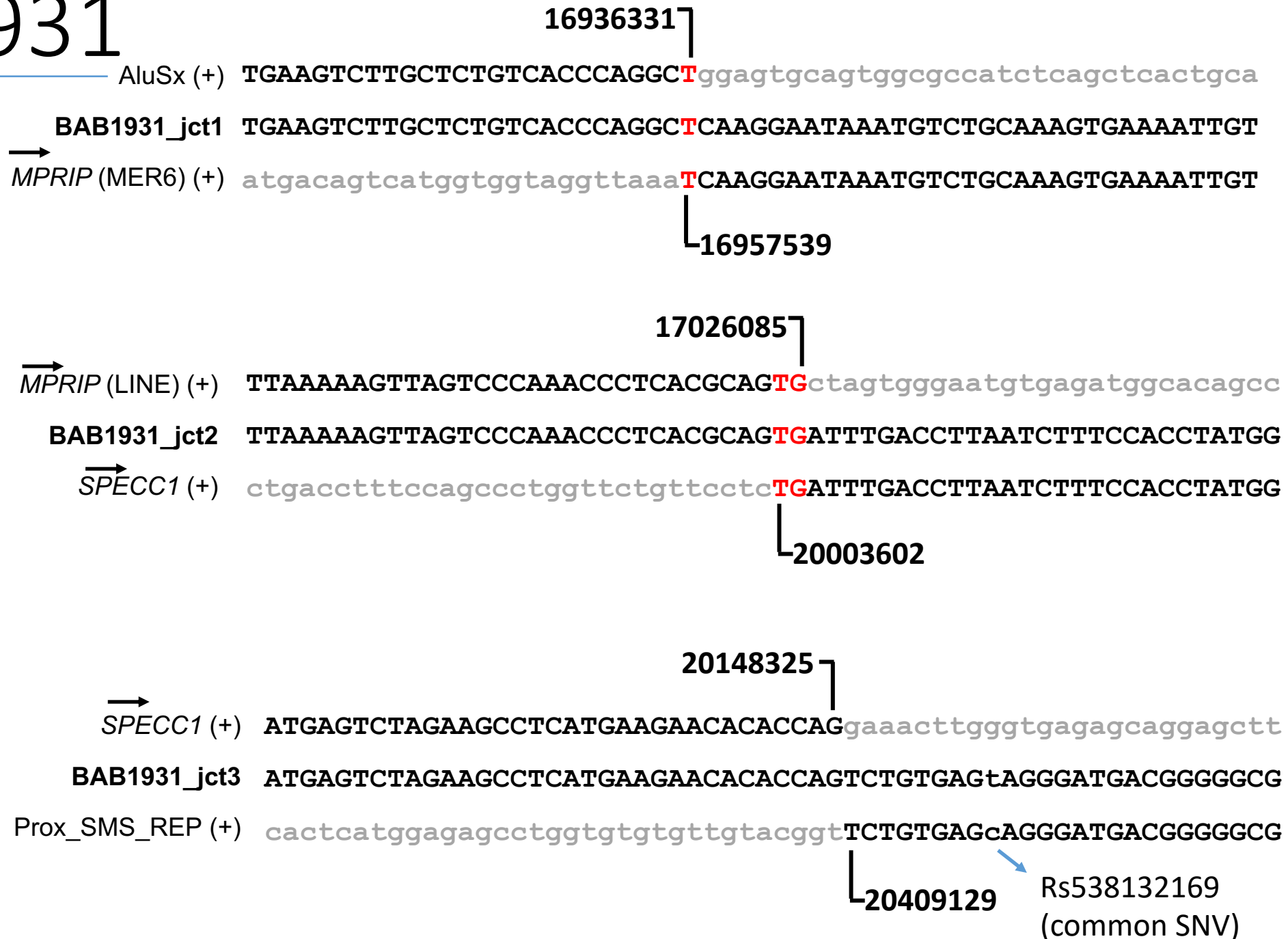
16035839 SNV called by Atlas2
 17868741

BAB1931

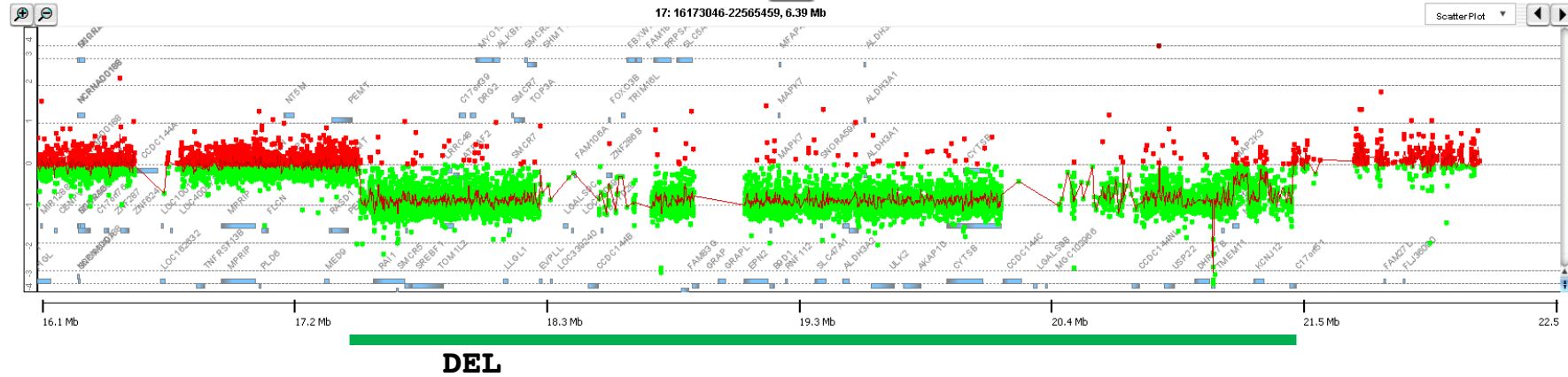
BAB1931: 3 de novo deletions (trio aCGH data)



BAB1931



BAB3031



17528139

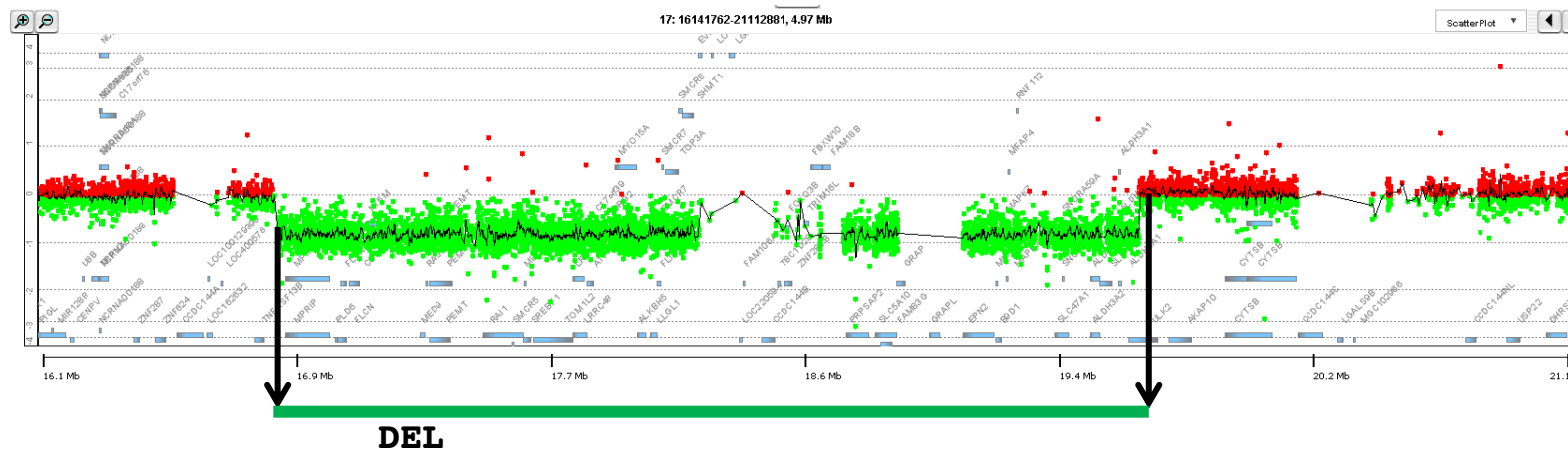
Intragenic (+) TAAGTAAAGGAGCTCCCTTGAGAA**GAG**ataggggaagtgttgagaaatatgtcagctt

BAB3031 TAAGTAAAGGAGCTCCCTTGAGAA**GAG**CAATTCTGAAATGATTTTCCATGTTAGCAA

← **C17ORF51** (LINE) (+) gcaaatttgtttcagggtatgggt**GAG**CAATTCTGAAATGATTTTCCATGTTAGCAA

21442150

BAB3133



16916256

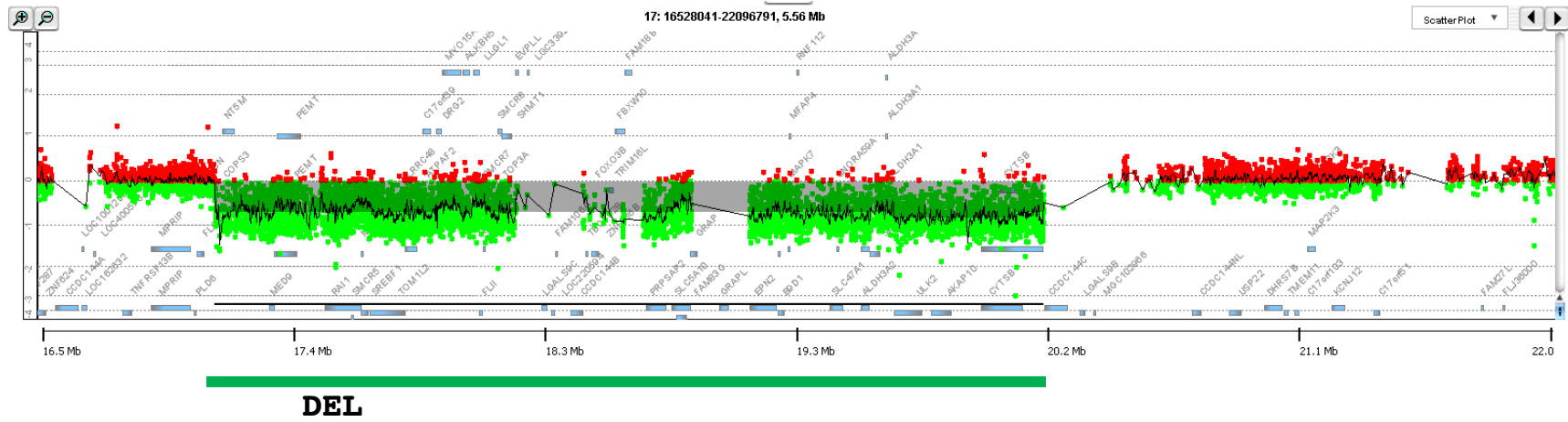
LCR_ (*AluSz*) (+) AGGTGTGAGCCACCACGCCAGCCTGTTTT**TCA**catttctatttcacatgctccagaattca

BAB3133 AGGTGTGAGCCACCACGCCAGCCTGTTTT**TCA**TGCTCATGAGGCTGTGTAAGTGATGGG

← *ULK2* (+) cagactaatgctaagccacagttgtgagc**TCA**TGCTCATGAGGCTGTGTAAGTGATGGG

19710487

BAB1615



←

17179369

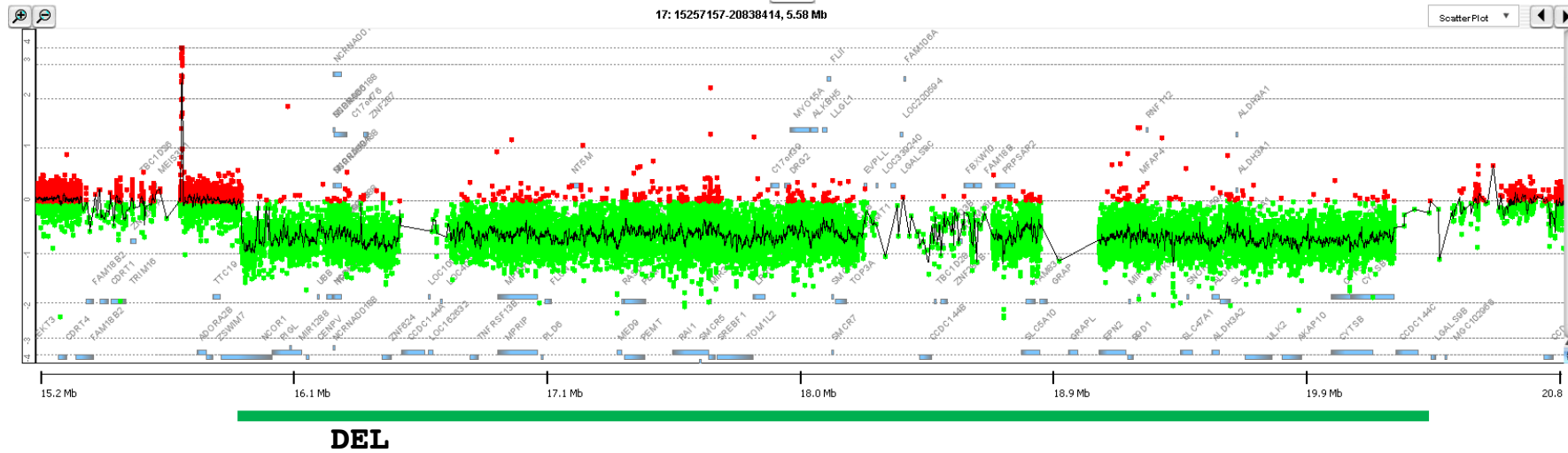
Ref_COPS3 (+) GATGCTCCTTACAAAACAGCAAGGACG**CCcaAG**gagtgttcttgtacatccagagccc

BAB1615 GATGCTCCTTACAAAACAGCAAGGACG**CCccAG**CCTTCCTGAAAACACCACCTCCC

Ref_Prox_SMS_REP (+) accctttctccatacctgctggggt**CCgcAG**CCTTCCTGAAAACACCACCTCCC

20415088

BAB6311



16008637

← Ref_NCOR1 (AluJr) (+) GCCTATGGTCCCAGATACTTGGGGGG**ATGA-GgTG**ggaaaatcgcttgagcctgggag

BAB6311 GCCTATGGTCCCAGATACTTGGGGGG**ATGA-GcTG**ACTTTAC-AAAATGTTGATGATA

Ref_Prox_SMS_REP (+) attctgtctcattcattgaaacagaa**ATGAaGcTG**ACTTTACaAAAATGTTGATGATA

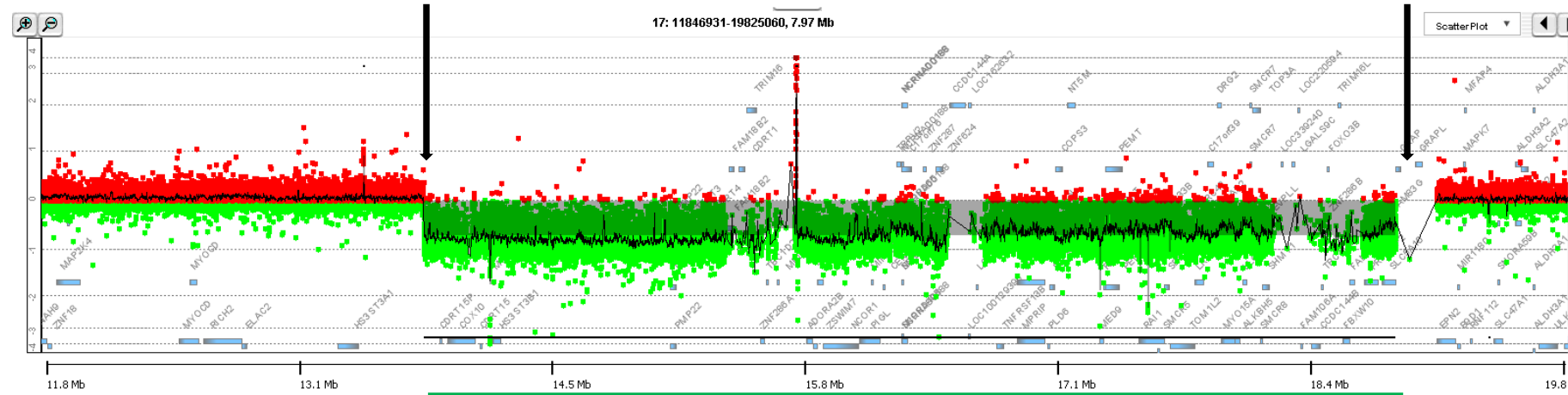
L20245447

↑ *De novo*
20245463

BAB8501

Jct outside of the captured region

Jct embedded in a LCR



Maternal origin of this DEL

DEL

13845144

Ref_Intragenic (MER58A) (+) GAGTTGGCAAGCTTTGTCTATAAAGGAC CAGT agcaaagggttcagactctgcaaaaca

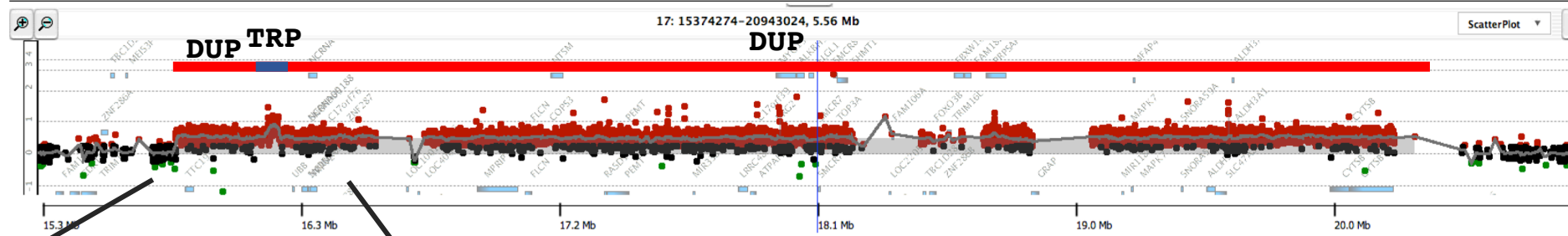
BAB8501 GAGTTGGCAAGCTTTGTCTATAAAGGAC CAGT GACTTTGCTCATCGTTGTCCCTGCGGTA

Ref_LCR_REPA/B (+) ccgtcccctctatgtcagcttcctgagg CAGT GACTTTGCTCATCGTTGTCCCTGCGGTA

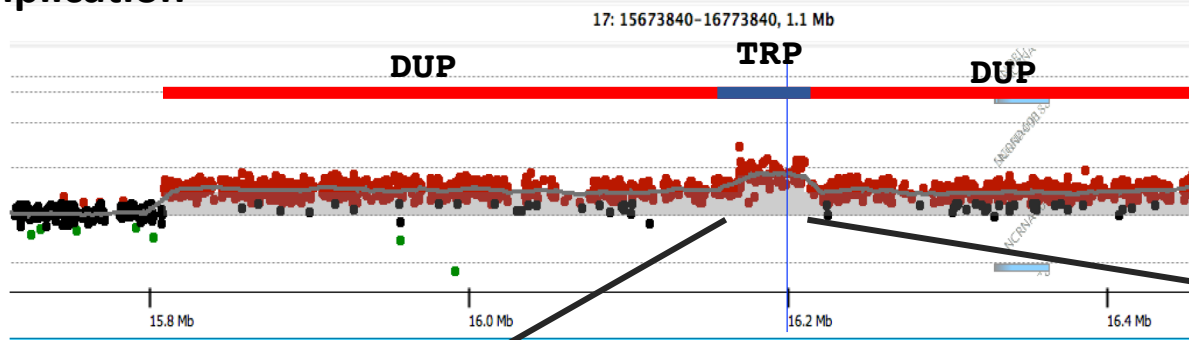
19105614*

BAB2695

Breakpoint for small triplication-40,930bp



Triplication



39 Kb Duplication

16237498 (+)

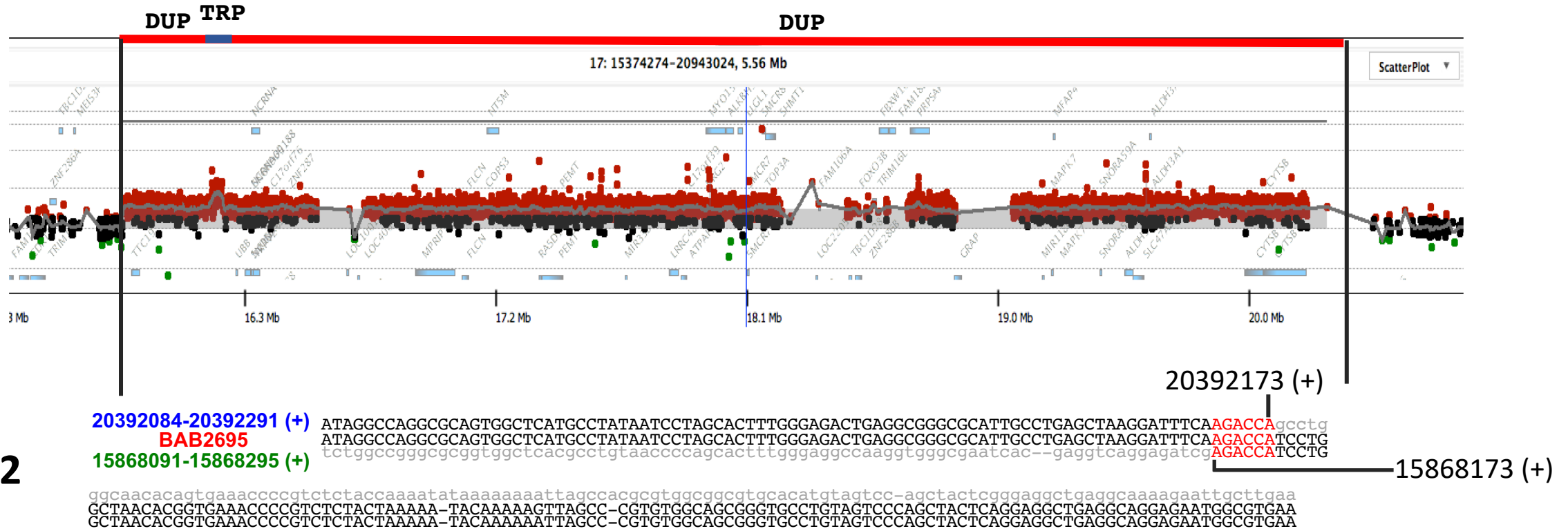
Jct1

Proximal *AluY* (+) AAATTTGATGATCACGCCGGGTGCGGTGGCTCACGCCTGTAATCCAGCACTTTGGgaggctgaggcaggcgtattacgaggtcaggagatcgaga
 BAB2695 AAATTTGATGATCACGCCGGGTGCGGTGGCTCACGCCTGTAATCCAGCACTTTGGAAAGGCCAAGGTGGGTGGATCACGAGGTCAGGAGATCCAGT
 Distal *AluY* (+) aattgcaatcttctggcctggcgcaGTGGCTCACGCCTGTAATCCAGCACTTTGGAAAGGCCAAGGTGGGTGGATCACGAGGTCAGGAGATCCAGT

16196568 (+)

BAB2695

Tandem Duplication Breakpoint in PTL5 Individual 2695

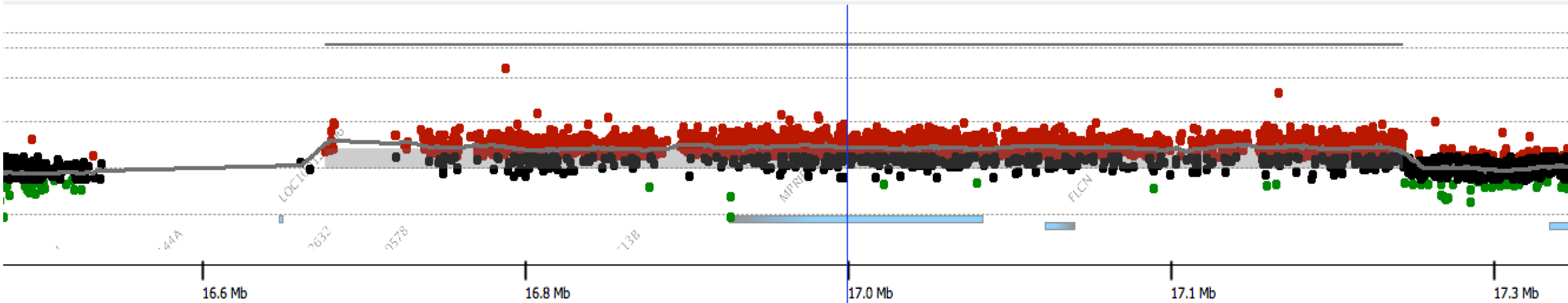


Jct2

AluSx to an *AluY* in direct orientation
 within 6 bp microhomology (in red). *AluSx* in Prox_REP

BAB3344

17: 16462095-17562095, 1.1 Mb



17326680 (+)

Exon 3 (last exon) of *SMCR9*

17326680 CAGGAGACATCATCCTAAATGTCAATGGGATGGACATTAGCAAAGTCTCTcacaactacgctctgcatctcctgcggcagccctaccagggtgttgtgactgaccatgctgcacaaccagaa

Patient CAGGAGACATCATCCTAAATGTCAATGGGATGGACATTAGCAAAGTCTCTTTATTTTTGTCTGACTGGGCTGACTCAAAGACAGGTCTTTGAGCTCTGTAATCCTTCCCTCCGCTTGGTC

16654065 aaatcccacattcctcagatgttttgttcatttttaaaatggttttTCTTTATTTTTGTCTGACTGGGCTGACTCAAAGACAGGTCTTTGAGCTCTGTAATCCTTCCCTCCGCTTGGTC

Intron of *CCDC144A*

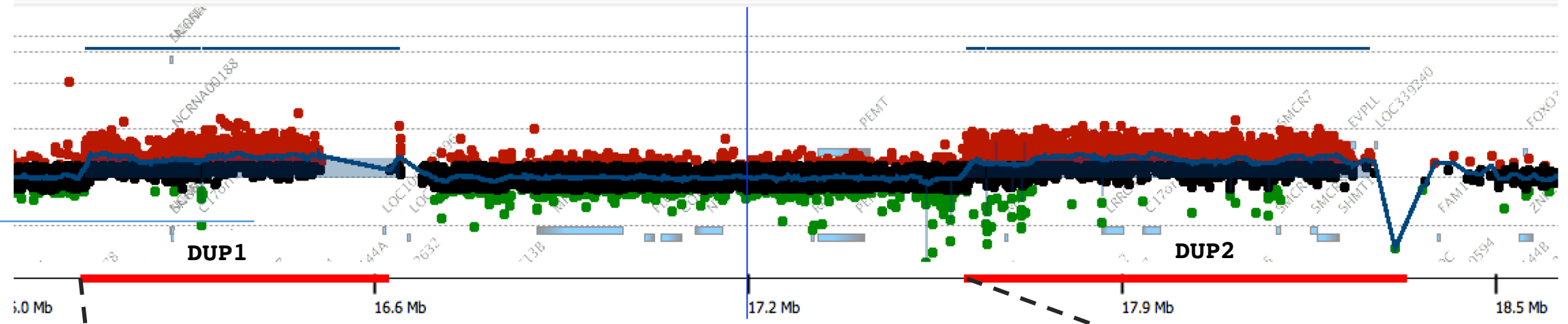
16654065 (+)

Distal side within Distal_REP

BAB3793

Beginning of DUP_1 to Beginning of DUP_2

17: 15435002-19147502, 3.71 Mb



Jct1

16203013(-)

Intron of *PIGL* ←

GAGATTTACAAACATGAAACAAC TAGAAATTAATGCATAA **TT** ccacacaacacacttttggagctttatgcttggtgctgagggttaag

BAB3793 GAGATTTACAAACATGAAACAAC TAGAAATTAATGCATAA **TT** (*****) **ATC** CAGGAGCACTTCGAGTGTGCTATTTTCTCCTCTGAG

ggcggaggtctttctgccactcttggtaacatcaaggaatgaatgagc **ATC** CAGGAGCACTTCGAGTGTGCTATTTTCTCCTCTGAG

Intron of *RAI1* →

17652760(+)

(*****) represents the insertion of 74 bp from Chr17:17662297-17662370 (from intron 2 of *RAI1*)

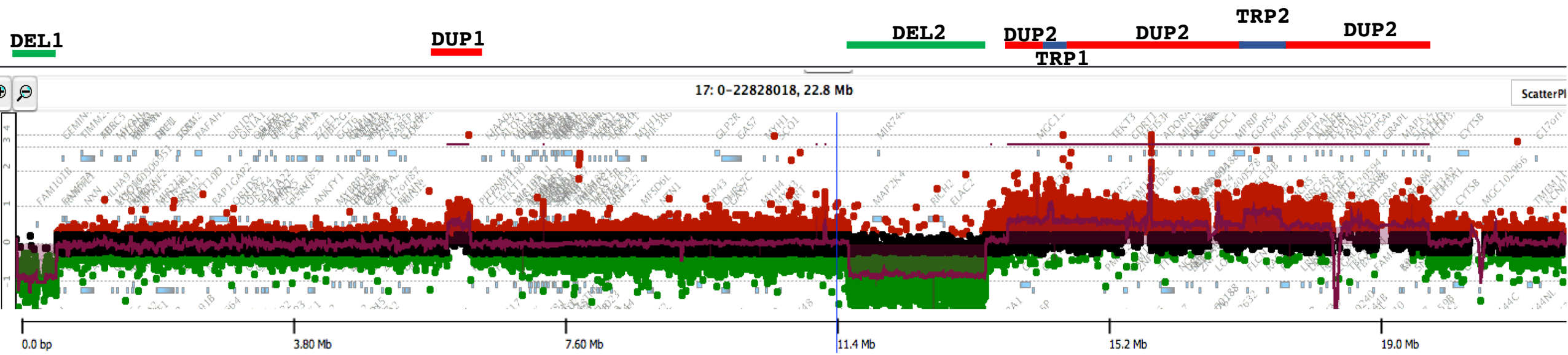
TTTACTTAGAGGTTCTGGTCGCCATGGAAGGTTTCAGCTGAGGAGGAACAAAGATTGGGTTGGGGAGTATC

Both sides are in unique regions

Jct2 (not determined at nt level) is from Distal to Middle_REP (paralogous LCRs); See Figure 3

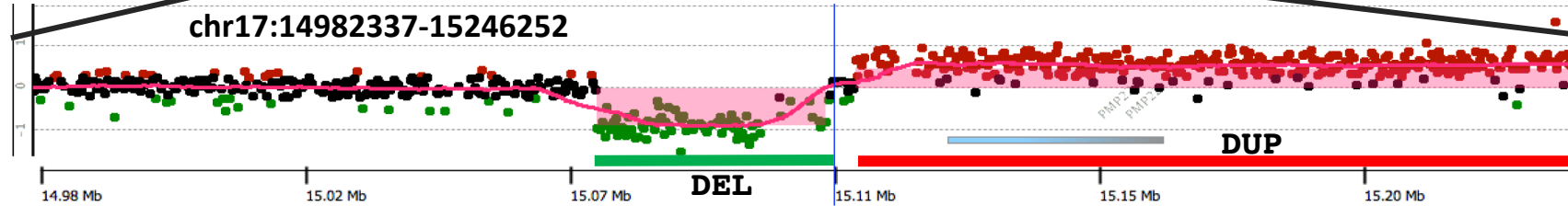
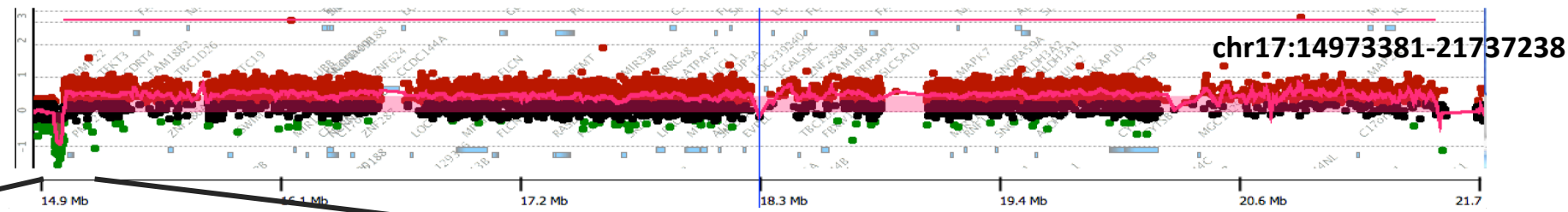
BAB2337

NR- complex (DEL-NML-DUP-NML-DEL-NML-DUP-TRP-DUP-TRP-DUP)



NR- complex (DEL-NML-
DUP-NML-DEL-NML-DUP-
TRP-DUP-TRP-DUP)

BAB1229



Jct1

39 Kb Deletion

15075315 (+)

```

TGAGCTCCACATGTGGAATCTCCCCAGACCCCATGACCTTCatacacatttcaccatttcccagagccag
TGAGCTCCACATGTGGAATCTCCCCAGACCCCATGACCTTCTGCTTCCTAACAACCACTGAGATAATTTG
attcttactgcaggggattggagtcaacataattaACCTTCTGCTTCCTAACAACCACTGAGATAATTTG
    
```

Coordinates in hg19

15113726 (+)

Jct2

6.7 Mb Dup

21968267 (+)

```

ATGGAATGGAATGGAATGGAAAGAAAGGAATAGAAggcaaaggaatcggatggaatggaatggagtcgaatgg
ATGGAATGGAATGGAATGGAAAGAAAGGAATAGAATTTTGGAATGCGCAGACTAGACTGTGAGTGATACGGTC
agtatcatttgaaattctaacaatgtagatttaAATTTTGGAATGCGCAGACTAGACTGTGAGTGATACGGTC
    
```

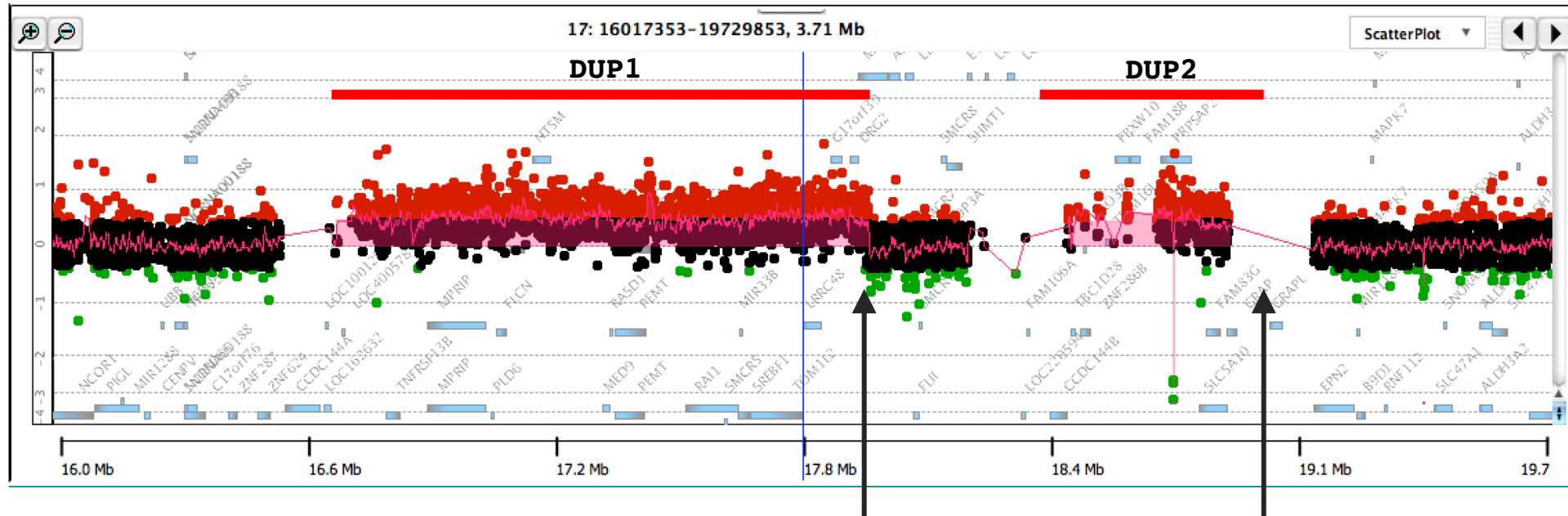
Coordinates in hg38

15214631 (+)

Deletion and duplication occurred in *cis* in the maternal haplotype as determined by GATK genotyping and B allele frequency of the regional Illumina data
 Del previously characterized in Zhang *et al.* 2009 Nat genet.

BAB2543

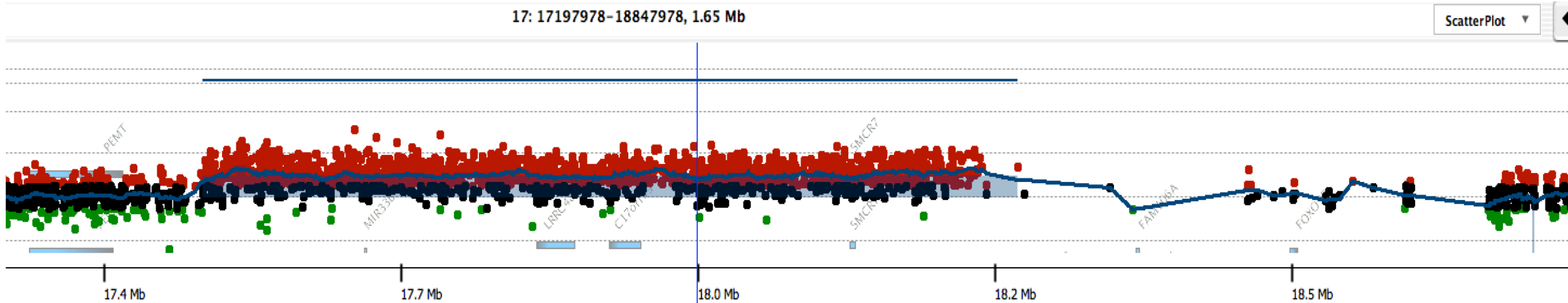
End of DUP_1 to End of DUP_2



AluSz
Jct1 18037186 (+) GCCCCACTAATTTTTGTATTTTTAGTAGAGACGGGGTTTCACCATGTTGGCCAGGCTGGTCTTGAACTCCTGAC
 19015148 (-) GCCCCACTAATTTTTGTATTTTTAGTAGAGACGGGGTTTCACCATGTTGGCCAGGCTGCTCCTGAACACCTGAG
AluSx
 19015148 (-) 29bp microhomology

Non-paralogous LCR Breakpoint is *AluSx-AluSz* mediated (Sx within REPA/B)
 Jct2 is between paralogous repeats- between Distal and Middle_REP

BAB2811



Middle_REP on proximal side of duplication

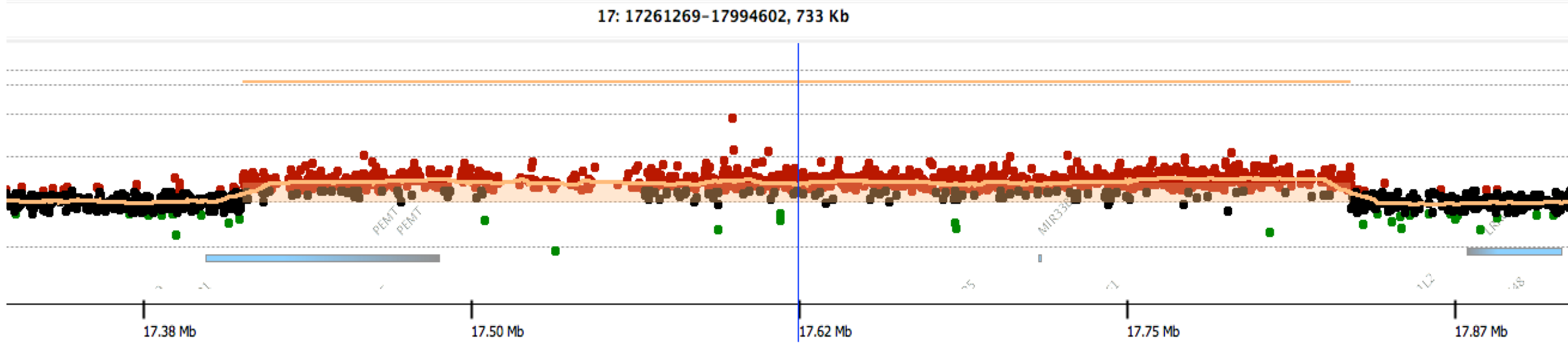
17568703 (+)

Patient

```
AGCTTGGGTGATAGAGTGAGACTCTAAAAGAAACACACACACA-CA-CACACACAATTTTAAGTTTTATTTCTTTTAAATTGATT
ACCTTGTGAAAGATTCATGTAATTGACCAGTGTTTACCAAGCA-CA-CACACACAATTTTAAGTTTTATTTCTTTTAAATTGATT
ACCTTGTGAAAGATTCATGTAATTGACCAGTGTTTACCAAGCAgCAtCAGTGTTTCAGTTTCAGTCATTGGTGATTCCGCAGTTGG
```

18320581 (+)

BAB2986



←
Intron of *TOM1L2*, downstream of *AluSp*

17833037 (+)

Patient **TTTCAGAGCAGTAGTTAACTTTTTGGATAAGATGTTAAAGGAAG**AATACATTCCCTTTGAAAATCTGATAAGACCTTTCCAGAGA
TGCCCCGGGCTCCCTGACCACCTCTGGATGCACTGCTGGTGC**A**GACCTGCTAACATAGTGACCTACTCTCCCCTGGGACCCGC

←
Intron of *PEMT*

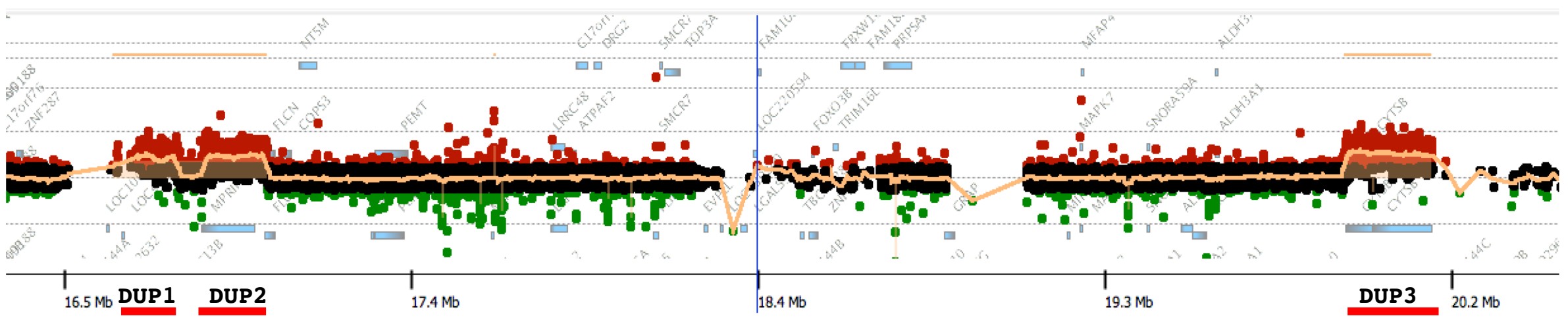
17421889 (+)

BAB8123

8123 |



17: 15638067-21206817, 5.56 Mb



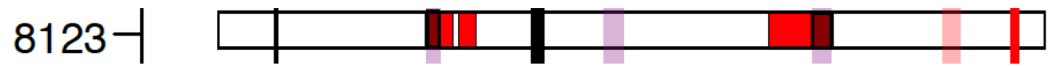
Jct1

End DUP1 to beginning of DUP2

L1ME3C, in intron of *LOC284191* ← SNP | 16881170 (+)
 GGCTCTATCTTCCAGGCCCTTTTATACTTTTATGAGTACAGTACAGAATGTTTTAGCTATGAAAAATACAGTATATAGCATTATTTTGCCTGTTATTTTTAAACATTTTATATAAAATAGCAT
 Patient GGCTCTATCTTCCAGGCCCTTTTATACT **cagg** CCGGAGGCAGAGCTTGCAGTGAGCCGAGATGGTGCCACGGCACTCCAGCCTGTGTGACAGAGC **GAGACTCCGTCTCAAAAAAAAAAAAAA**
 TS1 TACTCACGCGCTGAGGCAGGAGAATGGCGTGAACCCGGGAGGCAGAGCTTGCAGTGAGCCGAGATGGTGCCACGGCACTCCAGCCTGTGTGACAGAGC **GAGACTCCGTCTCAAAAAAAAAAAAAA**
 AluY element | 16912929 (-) | 16912841 (-)

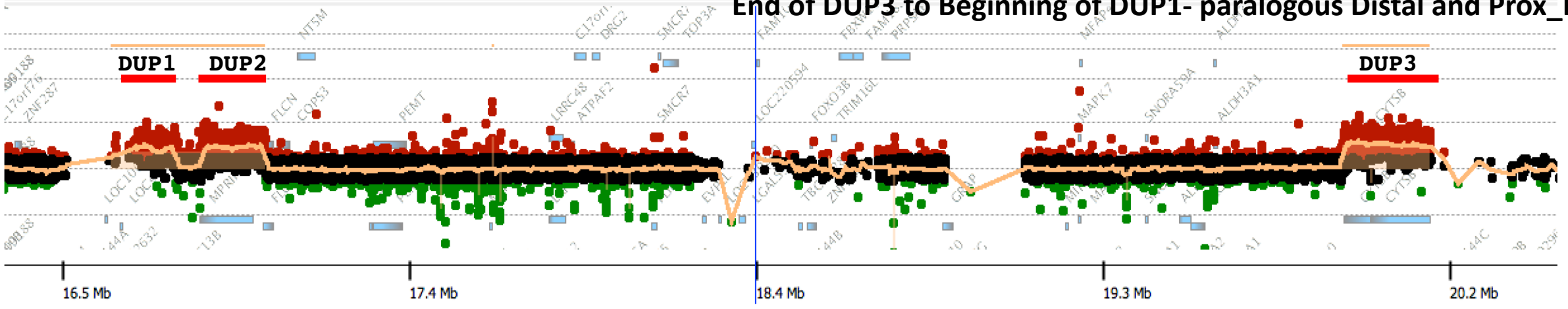
AluY element | 16912841 (-)
 25bp microhomology |
 TS1 GAGCTTGCAGTGAGCCGAGATGGTGCCACGGCACTCCAGCCTGTGTGACAGAGC **GAGACTCCGTCTCAAAAAAAAAAAAAA** CAAAAAAAAAACAGGCAAAAGTCATCTATGGAGACAGAAGTCCTG
 Patient GAGCTTGCAGTGAGCCGAGATGGTGCCACGGCACTCCAGCCTGTGTGACAGAGC **GAGACTCCGTCTCAAAAAAAAAAAAAA** AAAAAAAAAAGAGGTGAAATTAATTTTAATTATGTTTTATTTAACC
 GAGCTTGCAGTGAGCTGAGATtGcGCCACtGCACTCCAGCCTGgGcaAtAGAGt **GAGACTCCGTCTCAAAAAAAAAAAAAA** AAAAAAAAAAGAGGTGAAATTAATTTTAATTATGTTTTATTTAACC
 AluYc, in intron of *MPRIIP* | 16953616 (+)

BAB8123



17: 15638067-21206817, 5.56 Mb

End of DUP3 to Beginning of DUP1- paralogous Distal and Prox_REPs



End DUP2 to beginning of DUP3

←←
intron of *FLCN* and *PLD6*

Jct2

Patient GAGGCCTCCTCTCCACAACCCATGACAGAGATCTGGTTCCTTTGGGCCTGAGGCGTGGGGAACCTCAGCGCAGGGCATGGCCCCACAGCCCGCGGGGGCAC
 ACAAATAAACAAGAGGCTGGGCACTGTGACTCAGCCTGTAATCCAGCACTTTGGGAGGCGAGGCGGGCAGATCACCTGAGGTCAGGAGTTTGAGACCAA

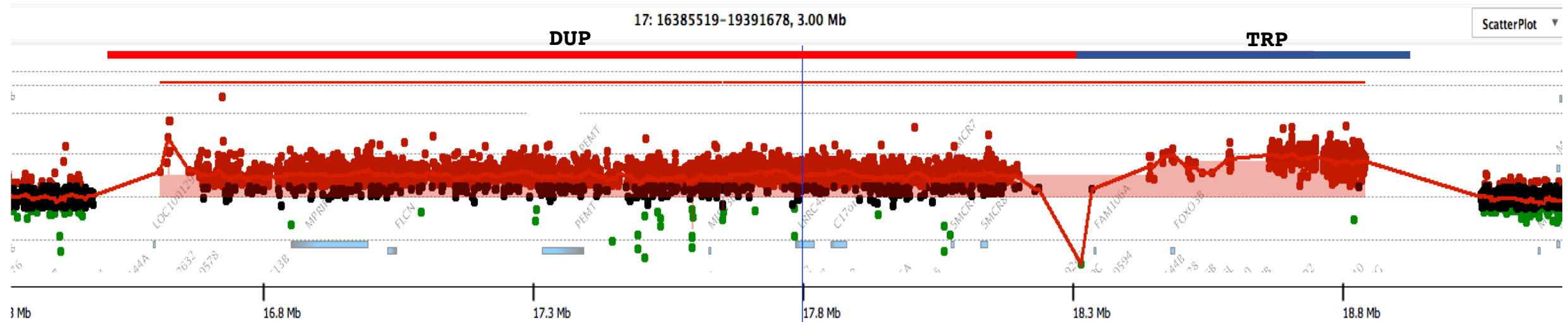
→
AluSq2, in intron of *SPECC1*

17119625(+)

19987504(+)

PTLS breakpoints Not Found

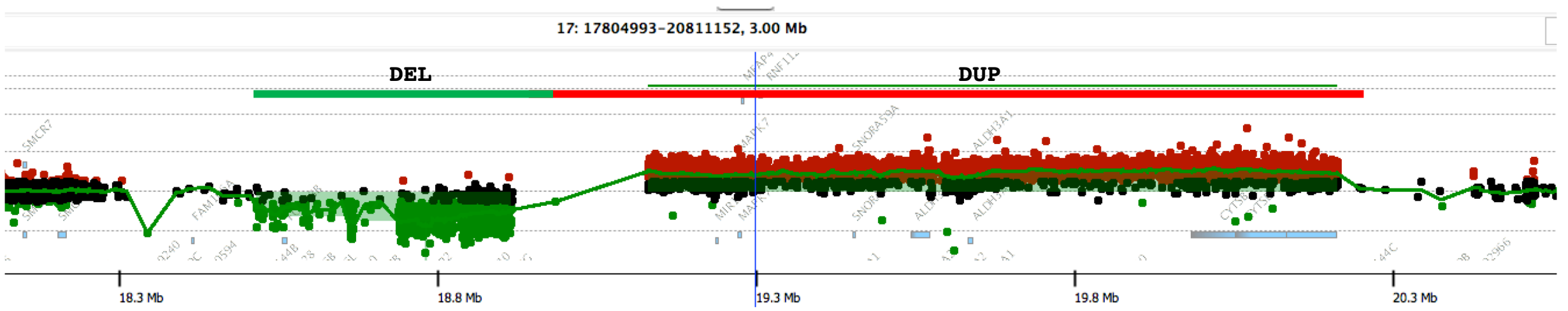
BAB2965



DUP-TRP Between Distal and Middle_REPs (paralogous LCRs) and REPA/B

PTLS breakpoints Not Found

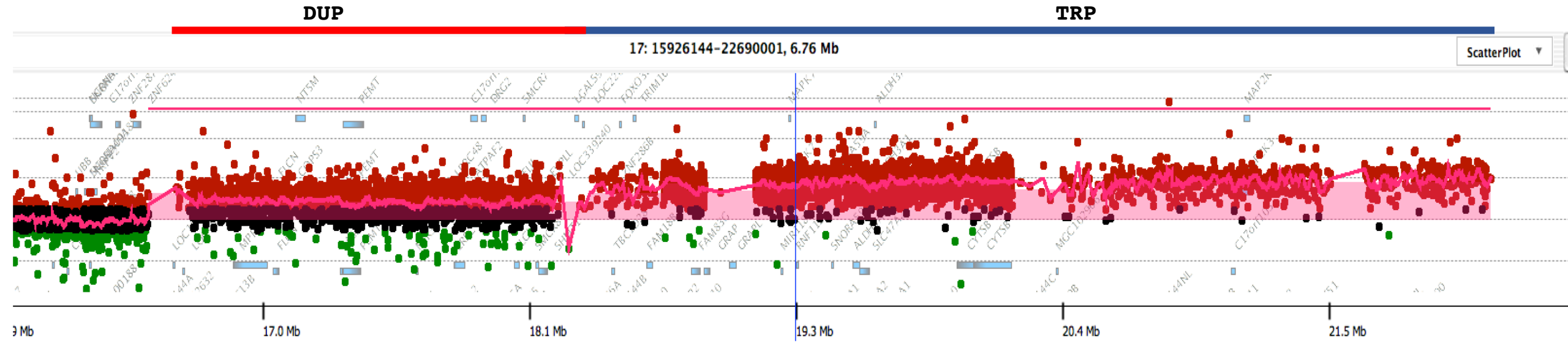
BAB6917



DEL-DUP with breakpoints between Middle_REP and REPA/B and REPA/B and Prox_REP

PTLS breakpoints Not Found

BAB8325



DUP-TRP between Distal and Middle_REP and the Centromere