## Supplementary Data

# *Alu*-mediated diverse and complex pathogenic copy-number variants within human chromosome 17 at p13.3

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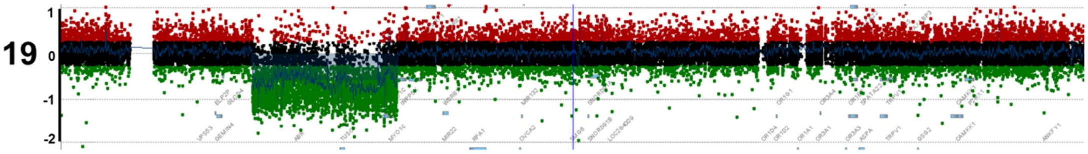
**A**



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chr17:1425666    TCGGCCAGGCGCGGTGGCTCACGCCTGTAATCCCAGCACTTTGGGAGGCCGAGGCGGGTG 1425607
                 ||||||||||||||||||||||||||||||||||||||||||||||||||||||||||||
19               TCGGCCAGGCGCGGTGGCTCACGCCTGTAATCCCAGCACTTTGGGAGGCCGAGGCGGGTG
                 |  ||| || | |||||    || |||||||||||  |||||||||||| ||| ||||||
chr17:814870     TTAGCCGGGTGTGGTGGTGGGCGGCTGTAATCCCAATACTTTGGGAGGCTGAGACGGGTG 814811

chr17:1425606    GATCATGAGGTCAGGAGATCGAGACCATCCTGGCTAACAAGGTGAAACCCCGTCTCTACT 1425547
                 ||||||||||||||||||||||||||||||||||||||| ||| |||| || |||||||
19               GATCATGAGGTCAGGAGATCGAGACCATCCTGGCTAACACGGTAAAACACCATCTCTACA
                 |||||  |||||||||||||||||||||||||||||||||||||||||||||||||||||
chr17:814810     GATCACAAGGTCAGGAGATCGAGACCATCCTGGCTAACACGGTAAAACACCATCTCTACA 814751
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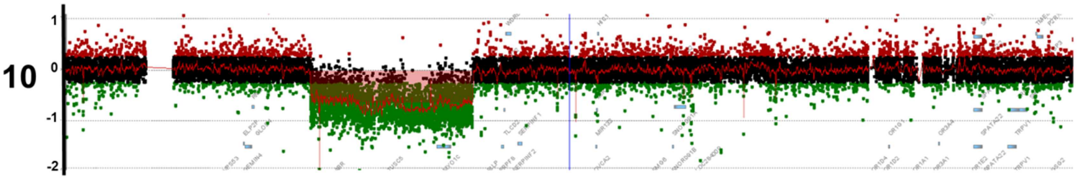
**B**



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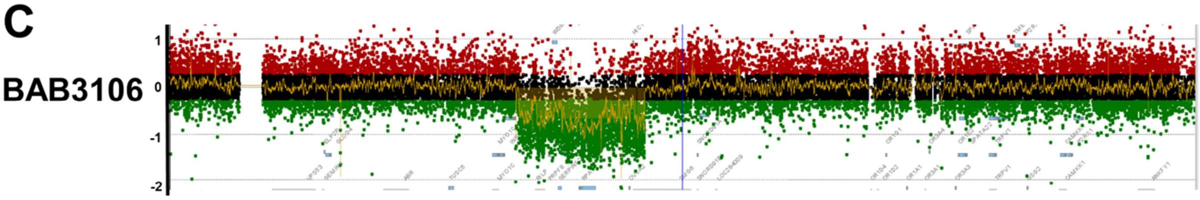
                                                             rs76608232
                                                    rs7770379**  *rs62088019
chr17:1498219    GCGTCCAGCAGATCCTGTCTCTTAAAACAAGAAAAAAAG---GTTGGGCGCGGTGGCTCA 1498163
                 |||||||||||||||||||||||||||||||||||||||   |  ||| |||||||||||
10               GCGTCCAGCAGATCCTGTCTCTTAAAACAAGAAAAAAAG---GCCGGGAGCGGTGGCTCA 57
                    |  |        |||||   |  ||    ||||| |   |      | |||||||||
chr17:903463     -AATTAAAAGACAATTGTCTAGAATTACTGAGAAAAATGTTGGCCACACGTGGTGGCTCA 903405

chr17:1498162    CGCCTGTAATCCCAGCACTTTGGGAGGCCAAGGCGGGCGGATCACGAGGTCAGGAGATCC 1498103
                 |||||||||||||||||||||||||||||||||||||||||||||||||||||||| ||
10               CGCCTGTAATCCCAGCACTTTGGGAGGCCAAGGCGGGCGGATCACGAGGTCAGGAGTTCG 117
                  |||||||||||||||||||||||||||  |||| ||| |||||||||||||||||||||
chr17:903404     TGCCTGTAATCCCAGCACTTTGGGAGGCTGAGGCAGGCAGATCACGAGGTCAGGAGTTCG 903345

chr17:1498102    AGACCATCCTGGCTAACGTGGTGAAACCCCGTCTCTACTGAAAATAC 1498056
                 ||||||             |||||||     |||||||| |||| |
10               AGACCA-------------GGTGAAATTGA-TCTCTACTAAAAAAAT 163
                 ||||||             ||||||||||| ||||||||||||||||
chr17:903344     AGACCA-------------GGTGAAATTGA-TCTCTACTAAAAAAAT 903312
```

**C**



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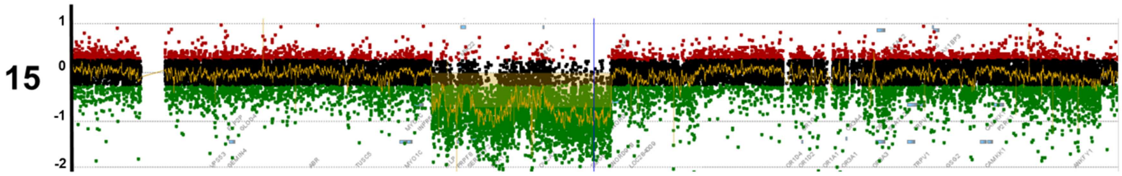
chr17:1465441    CCGGCCTCTCCTCGGGTCCACTTGGGCCTCCGAGGCCCCCGGGGCGCCTGAGGAGCCGTC 1465500
                 ||||||||||||||||||||||||||||||||
BAB3106          CCGGCCTCTCCTCGGGTCCACTTGGGCCTCCGTGTCCCTGCTGACAGGAAAGAAAGAGTG
                                           ||||||||||||||||||||||||||||||||||
chr17:2010026    AAGCTGATGACAAGGCAGACAGGAAACCTCCGTGTCCCTGCTGACAGGAAAGAAAGAGTG 2010085
```

**D**



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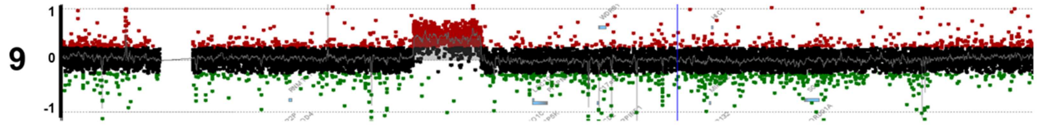
chr17:2244197   TCTGTAGAAATTA-AACACTGGGCCAGGCGTGGTGGCTCACGACTATAATCCCAGCACTT 2244139
                ||||||||||||| ||||||||||||||||||||||||||||||||||||||||||||||
15              TCTGTAGAAATTA-AACACTGGGCCAGGCGTGGTGGCTCACGACTATAATCCCAGCACTT
                ||| |  |||  | || |   ||   |||| ||||||||||| || ||||||||||||||
chr17:1498202   TCTCTTAAAACAAGAAAAAAAGGTTGGGCGCGGTGGCTCACGCCTGTAATCCCAGCACTT 1498143

chr17:2244138   TGGGAGGCCAAGGTGGGCGGATCACGAGGTCAGGAGATCGAGACCATCCTGGCTAACATG 2244079
                ||||||||||||||||||||||||||||||||||||||||||||||||||||||||| ||
15              TGGGAGGCCAAGGTGGGCGGATCACGAGGTCAGGAGATCGAGACCATCCTGGCTAACGTG
                ||||||||||||| ||||||||||||||||||||||||| ||||||||||||||||||||
chr17:1498142   TGGGAGGCCAAGGCGGGCGGATCACGAGGTCAGGAGATCCAGACCATCCTGGCTAACGTG 1498083
                                                       *rs12939032

chr17:2244078   GTGAAACCCCGTCTCTACTAAAAATACAAAAAA-TTAGCCGGGCGTGGTGGCAGGCGCCT 2244020
                ||||||||||||||||||| ||||||||||||| ||||||||| |||||||| |||||||
15              GTGAAACCCCGTCTCTACTGAAAATACAAAAAAATTAGCCGGGTGTGGTGGCGGGCGCCT
                ||||||||||||||||||||||||||||||||||||||||||||||||||||||||||||
chr17:1498082   GTGAAACCCCGTCTCTACTGAAAATACAAAAAAATTAGCCGGGTGTGGTGGCGGGCGCCT 1498023
```

**Figure S1. Array plots and breakpoint sequences of remaining interstitial deletions** (refer to Figure 1). Breakpoint junction sequence is aligned to the proximal and distal genomic references and color-matched. Microhomology at the breakpoint is indicated in red.

**A**



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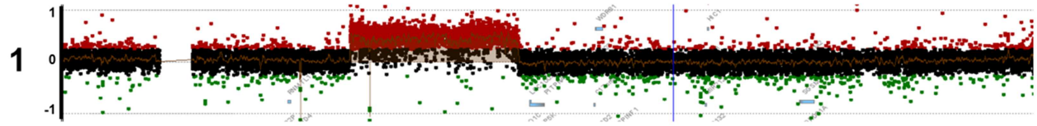
chr17:1267387   TTAACAAGACCTACAATAAGAAACATTTTATATATGTATATACACCCACAGTAAGTAAAA 1267446
                |||||||||||||||||||||||||||||||
9               TTAACAAGACCTACAATAAGAAACATTTTATTGTAGGCAGGTATTACTTTATGAGGCTCA
                                                   |||||||||||||||||||||||||
chr17:1061191   AGAATGGATTGAAAAGGATTATCTCGCCTAATAAGGGCAGGTATTACTTTATGAGGCTCA 1061250
```

**B**



```
chr17:1387172   ACTAAAAATACAAAA-TTAGCCGGGCGTGGTGGCGGGTGCCTGTAATCCCAGCTACTCGG 1387230
                ||||||||||||||| ||||||||||||||||||||||||||||||||||||||||||||
1               ACTAAAAATACAAAA-TTAGCCGGGCGCGGTGGCGGGTGCCTGTAATCCCAGCTACTCGG
                   |||||||||||| |||||| |||| |||||| || ||||||||||||||||||||||
chr17:875552    TTAAAAAATACAAAACTTAGCCAGGCGTGGTGGCTGGCGCCTGTAATCCCAGCTACTCGG 875611

chr17:1387231   GAGGCTGAGGCAGGAGAATCGCTTGAACCCGGGGGGAGGAGGTTACAGTGAGCTGAGATC 1387290
                |||||||||||||||||||||||||||||| || || ||||||| |||||||| ||||||
1               GAGGCTGAGGCAGGAGAATCGCTTGAACCCAGGAGGCGGAGGTTGCAGTGAGCCGAGATC
                ||||||||||||||||||||||||||||||||||||||||||||||||||||||||||||
chr17:875612    GAGGCTGAGGCAGGAGAATCGCTTGAACCCAGGAGGCGGAGGTTGCAGTGAGCCGAGATC 875671
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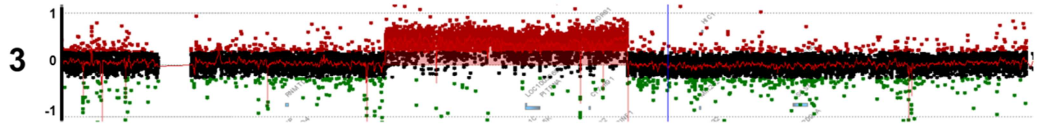
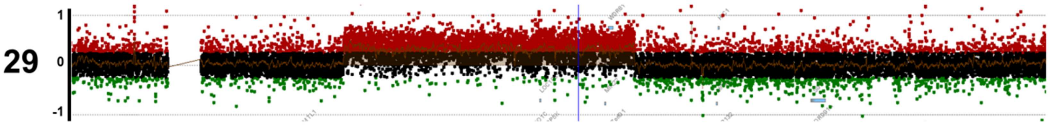
**C**



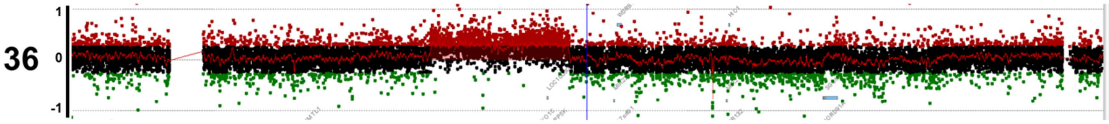
```
chr17:1733657   ATAAACGCTTGCCTTTCTCTGCATCTTCCACCTGCGGACACTTTCTGTCTC 1733707
                |||||||||||||||||||||||||||||||||||
3               ATAAACGCTTGCCTTTCTCTGCATCTTCCACCTGCAGCCCCAGGGGCCTCA
                                               ||||||||||||||||||||
chr17:991317    CTGACCCGGCTCCCCCCGGGCCCTCCCAAATCTGCAGCCCCAGGGGCCTCA 991367
```

**D**



```
chr17:1706253   CTGACCAACATGGTGAAACGCCATCTCTACTTAAAATTAGCCGGGCGTGGTGGTGCACGC 1706312
                ||||||||||||||||||||||||||||||||||||
29              CTGACCAACATGGTGAAACGCCATCTCTACTTAAAACCTCAGGTGATCCGCCCACCTCGG
                                                  ||||||||||||||||||||||||||
chr17:827671    TTCACCATGTTGGCCAGGCTGCTCTTGAACTCCCAACCTCAGGTGATCCGCCCACCTCGG 827730
```

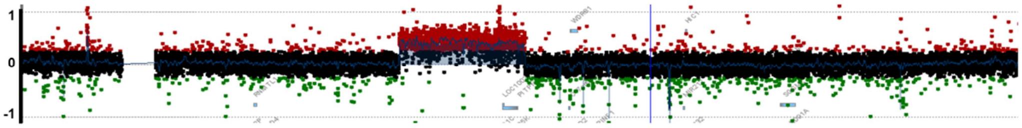
**E**



```
chr17:1073447   ACCAACTGGGCTGCCTCTTCCTAGGGGCATGAGCCTAAGGAGGTCTCCTCATGGGCACTT 1073388
                ||||||||||||||||||||||||||
36              ACCAACTGGGCTGCCTCTTCCTAGGGATTACAGGTGCGCACCATCACGCCCAGCTAATTT
                                       |||||||||||||||||||||||||||||||||||||
chr17:1486280   CTGCCTCAGCCTCCCGAGTAGCTGGGATTACAGGTGCGCACCATCACGCCCAGCTAATTT 1486221
```

**F**

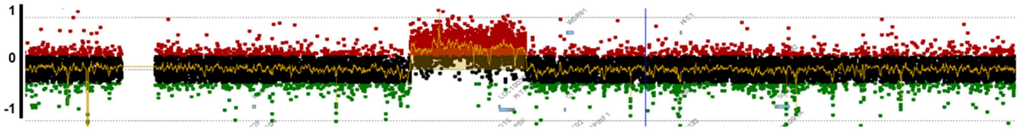
**7**



```
chr17:1485898   CCACGGTTATGCAAGACGCCCCCAAAACCAAGACATGCCTTCCGGGGAGGAGTCCCTTTG 1485957
                ||||||||||||||||||||||||||||||||||||||||||||||||||
7               CCACGGTTATGCAAGACGCCCCCAAAACCAAGACATGCCTTCCGGGGAGGCTGAGACAGG
                                                            ||||||||||||||||
chr17:1116032   AATTAGCCAAGTGTGGTGGCGGACGCCTGTAATCTCAGCTACTTGGGAGGCTGAGACAGG 1116091
```

**G**

**22**



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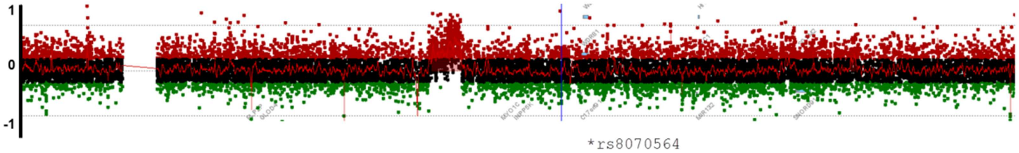
chr17:1498114   CCTCGTGATCCGCCCGCCTTGGCCTCCCAAAGTGCTGGGATTACAGGCGTGAGCCACCGC 1498173
                ||||||||||||||||||||||||||||||||||||||||||||||||||||||||||||
22              CCTCGTGATCCGCCCGCCTTGGCCTCCCAAAGTGCTGGGATTACAGGCGTGAGCCACCGC
                |||||||||| |||| | ||||||||||||||||||||||||||||||||||||||||||
chr17:1156180   CCTCGTGATCTGCCCACTTTGGCCTCCCAAAGTGCTGGGATTACAGGCGTGAGCCACCGC 1156239

chr17:1498174   GCCCAACCTTT----TTTTCTTGTTTTAAGAGAC----AGGATCTGCTGGACGCAG---T 1498222
                ||||  ||  |    || | |  |  ||| |  |    ||||| ||    | | |    |
22              GCCCGGCCCCTAAAGTTATTTCTTAATAACAAGCCGGAAGGATTTGTGAAAAGGAAAACT
                ||||||||||||||||||||||||||||||||||||||||||||||||||||||||||||
chr17:1156240   GCCCGGCCCCTAAAGTTATTTCTTAATAACAAGCCGGAAGGATTTGTGAAAAGGAAAACT 1156299
```

**H**

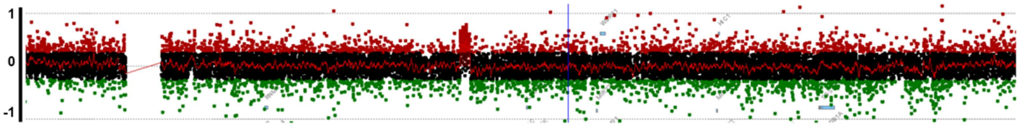
**BAB3054**



```
                                                     *rs8070564
chr17:1181273   GGGAGGCCAAGGTGGGCAGATCACGAGGTCAGGAGATTGAGACCATCCTGACCAATGTGG 1181214
                ||||||||||||||||||||||||||||||||||||| |||||| ||||| ||||   ||
3054            GGGAGGCCAAGGTGGGCAGATCACGAGGTCAGGAGATCGAGACCGTCCTGGCCAACACGG
                |||||||||||| |||| |||||| ||||||||||||||||||||||||||| |||| ||
chr17:1273684   GGGAGGCCAAGGCGGGCGGATCACAAGGTCAGGAGATCGAGACCGTCCTGGCTAACATGG 1273625
                                                            rs62087941*  *rs78971562
```

**I**

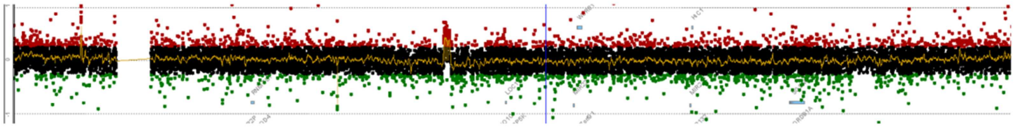
**31**



```
chr17:1263037   GGTGAAGAGAAAGTTTTCAAATAGTGGTGATTACACAACACTGTCAATGTATTCGGTGCC 1263096
                |||||||||||||||||||||||||||||||||||||||||||||
31              GGTGAAGAGAAAGTTTTCAAATAGTGGTGATTACACAACACTGTCGCTTGAACCCGGGAG
                                                           |||||||||||||||||
chr17:1234571   CATGCCTGTAATCCCAGCTACTCGGGAGGCTGAGGCAGGAGAATCGCTTGAACCCGGGAG 1234630
```

**J**

**26**



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chr17:1260559   TGGTGGCGGGCGCCTGTAGTCCCAGCTACTCGGGAGGCTGAGGCAGGAGAATGGCGTGAA 1260618
                ||||||||||||||||||||||||||||||||||||||||||||||||||||  | | ||
26              TGGTGGCGGGCGCCTGTAGTCCCAGCTACTCGGGAGGCTGAGGCAGGAGAATCACTTCAA
                |||||| ||||||||||| |||||||||||||||||||||||||||||||||||||||||
chr17:1242683   TGGTGGTGGGCGCCTGTAATCCCAGCTACTCGGGAGGCTGAGGCAGGAGAATCACTTCAA 1242742
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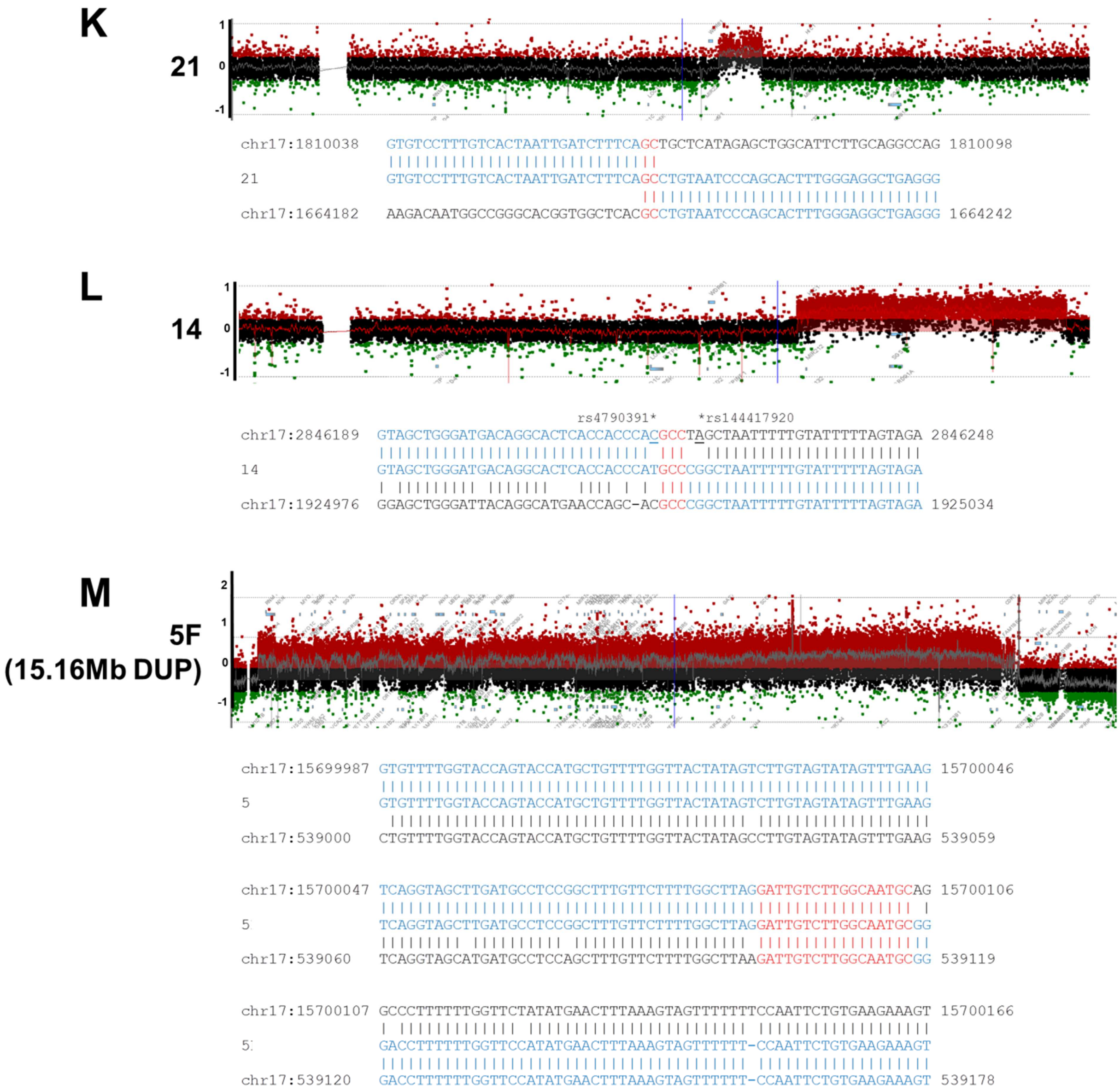

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chr17:1810038   GTGTCCTTTGTCACTAATTGATCTTTCAGCTGCTCATAGAGCTGGCATTCTTGCAGGCCAG 1810098
                ||||||||||||||||||||||||||||||
21              GTGTCCTTTGTCACTAATTGATCTTTCAGCCTGTAATCCCAGCACTTTGGGAGGCTGAGGG
                                            |||||||||||||||||||||||||||||||||
chr17:1664182   AAGACAATGGCCGGGCACGGTGGCTCACGCCTGTAATCCCAGCACTTTGGGAGGCTGAGGG 1664242
```

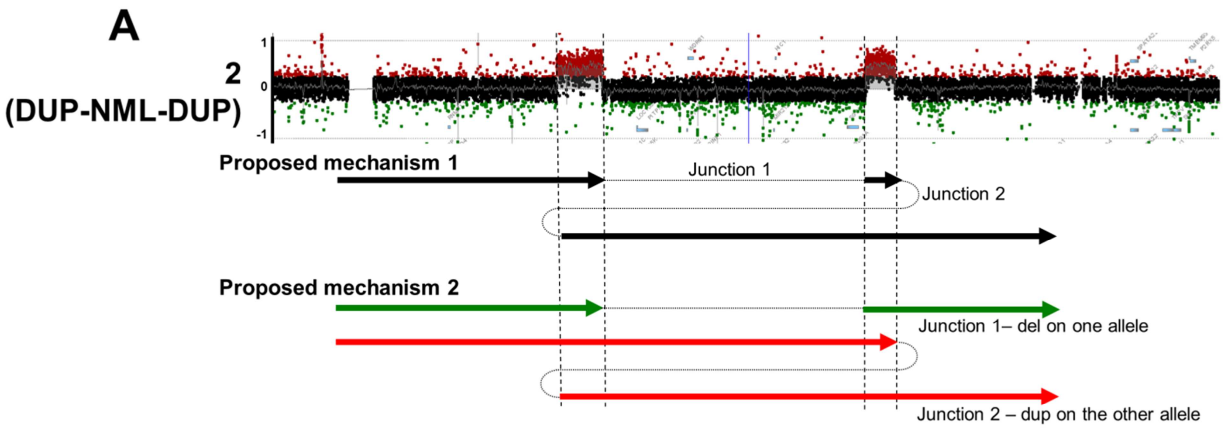
```
                                     rs4790391*    *rs144417920
chr17:2846189   GTAGCTGGGATGACAGGCACTCACCACCCACGCCTAGCTAATTTTTGTATTTTTAGTAGA 2846248
                |||||||||||||||||||||||||||||| |||  ||||||||||||||||||||||||
14              GTAGCTGGGATGACAGGCACTCACCACCCATGCCCGGCTAATTTTTGTATTTTTAGTAGA
                | ||||||||| |||||||   |||| | | |||||||||||||||||||||||||||||
chr17:1924976   GGAGCTGGGATTACAGGCATGAACCAGC-ACGCCCGGCTAATTTTTGTATTTTTAGTAGA 1925034
```

```
chr17:15699987  GTGTTTTGGTACCAGTACCATGCTGTTTTGGTTACTATAGTCTTGTAGTATAGTTTGAAG 15700046
                ||||||||||||||||||||||||||||||||||||||||||||||||||||||||||||
5               GTGTTTTGGTACCAGTACCATGCTGTTTTGGTTACTATAGTCTTGTAGTATAGTTTGAAG
                 ||||||||||||||||||||||||||||||||||||||| |||||||||||||||||||
chr17:539000    CTGTTTTGGTACCAGTACCATGCTGTTTTGGTTACTATAGCCTTGTAGTATAGTTTGAAG 539059

chr17:15700047  TCAGGTAGCTTGATGCCTCCGGCTTTGTTCTTTTGGCTTAGGATTGTCTTGGCAATGCAG 15700106
                |||||||||||||||||||||||||||||||||||||||||||||||||||||||||| |
5               TCAGGTAGCTTGATGCCTCCGGCTTTGTTCTTTTGGCTTAGGATTGTCTTGGCAATGCGG
                ||||||||| |||||||||| ||||||||||||||||||| |||||||||||||||||||
chr17:539060    TCAGGTAGCATGATGCCTCCAGCTTTGTTCTTTTGGCTTAAGATTGTCTTGGCAATGCGG 539119

chr17:15700107  GCCCTTTTTTGGTTCTATATGAACTTTAAAGTAGTTTTTTTCCAATTCTGTGAAGAAAGT 15700166
                | ||||||||||||| |||||||||||||||||||||||| |||||||||||||||||||
5               GACCTTTTTTGGTTCCATATGAACTTTAAAGTAGTTTTTT-CCAATTCTGTGAAGAAAGT
                |||||||||||||||||||||||||||||||||||||||| |||||||||||||||||||
chr17:539120    GACCTTTTTTGGTTCCATATGAACTTTAAAGTAGTTTTTT-CCAATTCTGTGAAGAAAGT 539178
```

**Figure S2. Array plots and breakpoint sequences of remaining tandem duplications** (refer to Figure 2). Breakpoint junction sequence is aligned to the proximal and distal genomic references and color-matched. Microhomology at the breakpoint is indicated in red. The 4 bp small insertion at the breakpoint of 9M (S2A) is indicated by a black box, and may be copied from the proximal reference sequence as indicated by a red box.



**Junction 1**

```
                                  *rs77376275
chr17:1287744   GGGAGGCCAAGGCGGGCAGATCACAACGTCAGGAGATCAAGACCATCCTGGCTAACACGG 1287803
                ||||||||||||||||| ||||||||||||||||||||||||||||||||||||||||||
2_1             GGGAGGCCAAGGCGGGCGGATCACAACGTCAGGAGATCAAGACCATCCTGGCTAACACGG
                |||||||| ||| |||| |||||| | ||||||||||| |||||||||||||||||||||
chr17:2311717   GGGAGGCCGAGGTGGGCGGATCACGAGGTCAGGAGATCGAGACCATCCTGGCTAACACGG 2311776

chr17:1287804   TGAAACCCTGTCTCTACTAAAAATACAAAAAAAATTAGCCAGGCATGGTGGCAGGCACCT 1287863
                |||||||| ||||||||||||||||||||||   |||||| ||| ||||||||||| |||
2_1             TGAAACCCCGTCTCTACTAAAAATACAAAAA---TTAGCCGGGCGTGGTGGCAGGCGCCT
                |||||||||||| ||||||||||||||||||   ||||||||||||||||||| || |||
chr17:2311777   TGAAACCCCGTCGCTACTAAAAATACAAAAA---TTAGCCGGGCGTGGTGGCATGCACCT 2311833
                           *rs12452380                     rs12450730*  *rs12450250
```

**Junction 2**

```
chr17:1108955   AA--------CCGGTCCGCGCCGCCGAGTGCGGACATTAGGGTCGTGAGACCCTCCGGGT 1109006
                ||        |||||||||||| ||||||| ||||||||||||||||| |||||||||||
2_2             AATTTTTTATCCGGTCCGCGCCACCGAGTGTGGACATTAGGGTCGTGAAACCCTCCGGGT
                |||||||||||||||||||||||||||||||||||||||||||||||||||||||||| |
chr17:2426932   AATTTTTTATCCGGTCCGCGCCACCGAGTGTGGACATTAGGGTCGTGAAACCCTCCGGTT 2426991
```

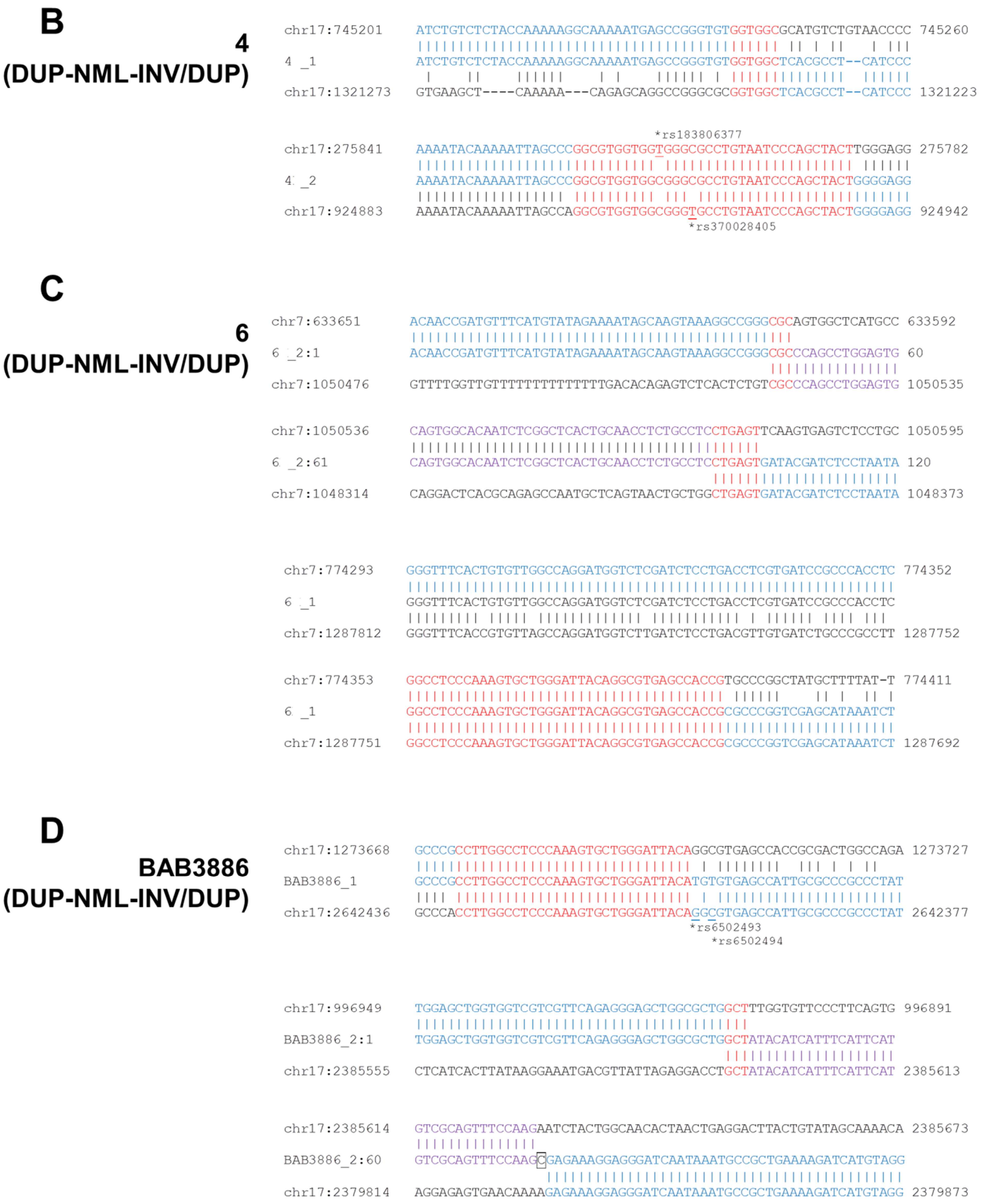


**Figure S3. Array plots and breakpoint sequences of complex rearrangements with DUP-NML-DUP pattern** (refer to Figure 3). Breakpoint junction sequence is aligned to the proximal and distal genomic references and color-matched. Microhomology at the breakpoint is indicated in red. Sequence in purple indicates inserted sequences at the breakpoint junction.

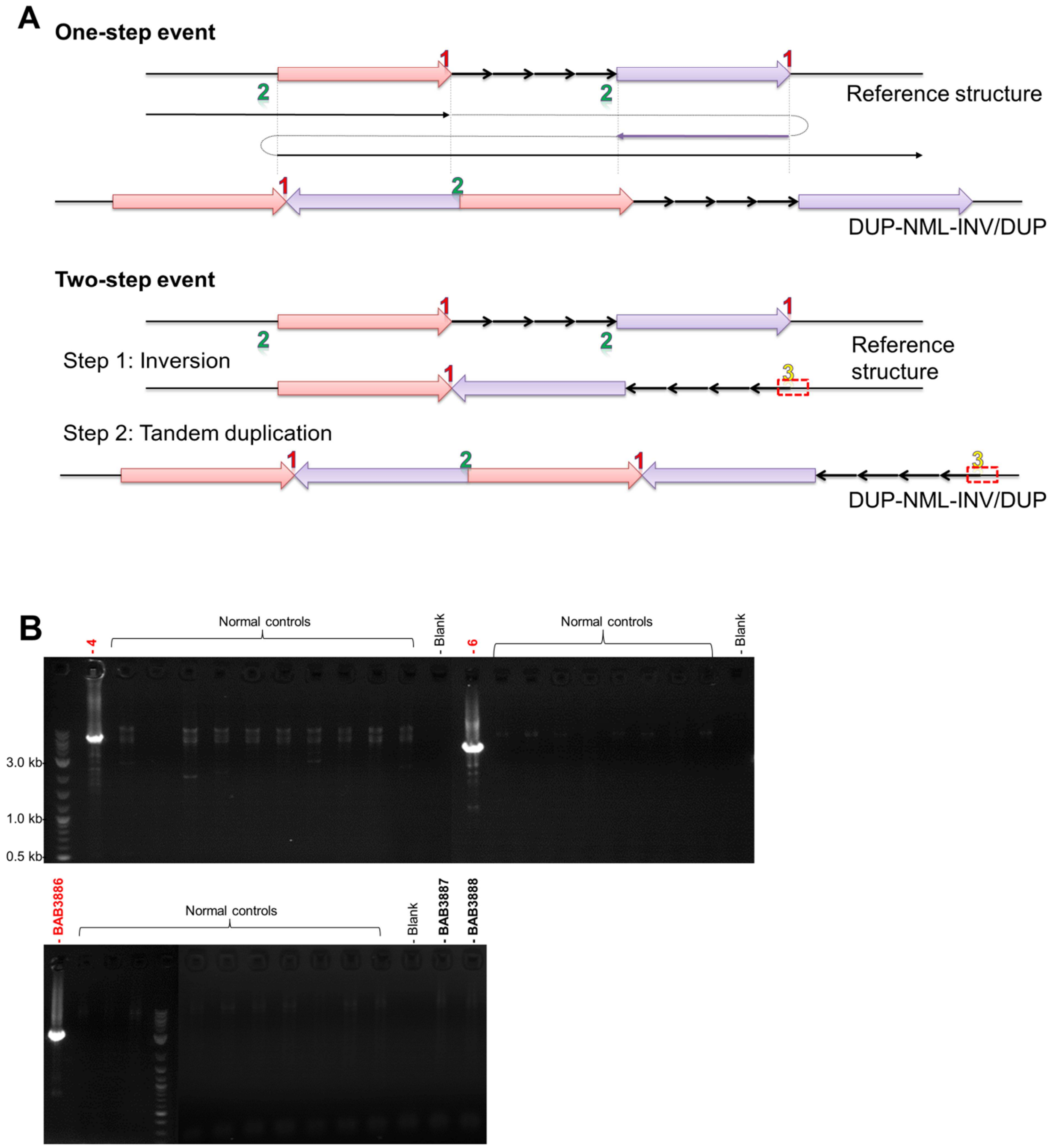
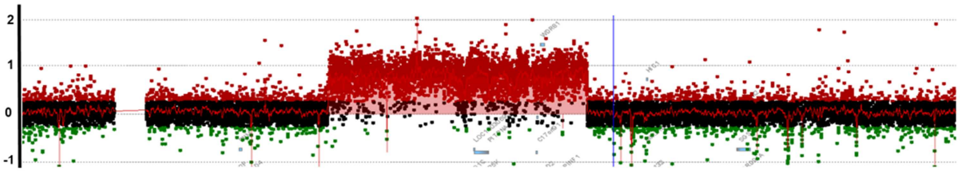


**Figure S4**. (*A*). Two possible rearrangements to generate the DUP-NML-INV/DUP pattern in samples 4F, 6M and BAB3886. (*B*). Long-range PCR detected "breakpoint junction 1" as shown in Figure S4A only existed in proband 4F, 6M and BAB3886, but not in any of the available parental samples (BAB3887 and BAB3888 as mother and father of BAB38886, respectively) or normal individuals ("Normal controls").

**A**

**20 (TRP)**



```
chr17:1774264   TGTTCTATTTCTTTCCAGCCACTTAGCCCCAATACTATTTCCTGAGCAACTCATCTTTT 1774322
                |||||||||||||||||||||
20              TGTTCTATTTCTTTCCAGCCAGCGAGACTCTACCTCAAAAAGAAAGAAAAAGAATCTGA
                                    |||||||||||||||||||||||||||||||||||||||
chr17:963614    CACTCCAGCCTGGGTGACAGAGCGAGACTCTACCTCAAAAAGAAAGAAAAAGAATCTGA 963672
```

**B**

**K2 (DUP-TRP-DUP)**

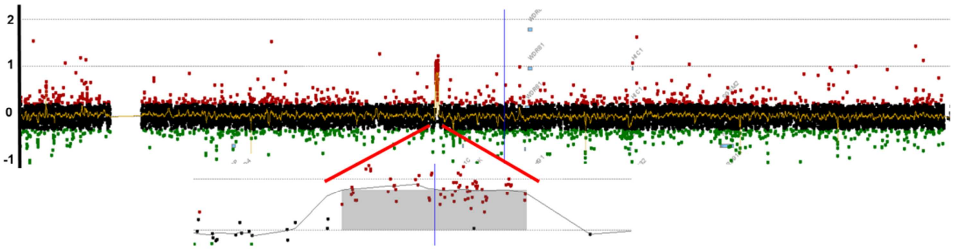
```
chr17:1118797   CTCGGGAGGCCGAGGCTGGCAGATCACTCGCGGCTAGGA-GCTGGAGACCAGCCCGGCCA 1118739
                ||||||||   ||| || |||| | |  || |     |   ||| |  |||||| |||||
K2_1            CTCGGGAGATGGAG-CTTGCAG-TGAGCCGAGATCGCGCTACTGCACTCCAGCCTGGCCA
                |||||||||||||| ||||||| |||||||||||||||||||||||||||||||||||||
chr17:1439677   CTTGGGAGATGGAG-CTTGCAG-TGAGCCGAGATCGCGCTACTGCACTCCAGCCTGGCCA 1439620


chr17:1121165   AATACAA-AAATTAGCTGAGCATGGTGGCACGTGCCTGTAATCCCAGCTACTCAGGAGGC 1121107
                ||||||| ||||||||||||||||||||||||||||||||||||||||||||||||||||
K2_2            AATACAA-AAATTAGCTGAGCATGGTGGCACGTGCCTGTAATCCCAGCTACTCAGGAGGC
                ||||||| | ||||||   || |||||||| ||||||||||||||||||||| |||||||
chr17:1440535   AATACAACAGATTAGCCAGGCGTGGTGGCAGGTGCCTGTAATCCCAGCTACTTAGGAGAC 1440476

chr17:1121106   TGAGGCAGGAGAATCGCTTGAACCTGGGAGGTGGAGGTTGCAGTGAACTGAGATTGCACC 1121047
                ||||||||||||||||||||||||  ||||| |||||||||||||| | |||||    ||
K2_2            TGAGGCAGGAGAATCGCTTGAACCCAGGAGGCGGAGGTTGCAGTGAGCCGAGATCATGCC
                ||||||||||||||||||||||||||||||||||||||||||||||||||||||||||||
chr17:1440475   TGAGGCAGGAGAATCGCTTGAACCCAGGAGGCGGAGGTTGCAGTGAGCCGAGATCATGCC 1440416
```

**C**

**18 (TRP)**



```
chr17:1345010   TCCTGACCTC--GTGA----TCCATCCACCTCGGCCTCCCAAAGTGCTGGGATTACAGGC 1345063
                ||||||||||  ||||    ||||||||||||||||||||||||||||||||||||||||
18              TCCTGACCTC--GTGA----TCCATCCACCTCGGCCTCCCAAAGTGCTGGGATTACAGGC
                ||||||||||  ||||    | || |||||||| ||||||||||||||||||||||||||
chr17:1333562   TCCTGACCTCAGGTGAAGAGTTCACCCACCTCGACCTCCCAAAGTGCTGGGATTACAGGC 1333621

chr17:1345064   GTGAGCCACTGTGCCCGGCCCTTCACCCAGCTGATTTTGAAATTTTTTTTATAGAGACAG 1345123
                |||| ||||||| || ||           ||| ||| |||        ||||      ||
18              GTGAACCACTGTACCTGG-----------GCTCATTCTGA--------TTAT-----TAG
                ||||||||||||||||||           |||||||||||        ||||     |||
chr17:1333622   GTGAACCACTGTACCTGG-----------GCTCATTCTGA--------TTAT-----TAG 1333657
```

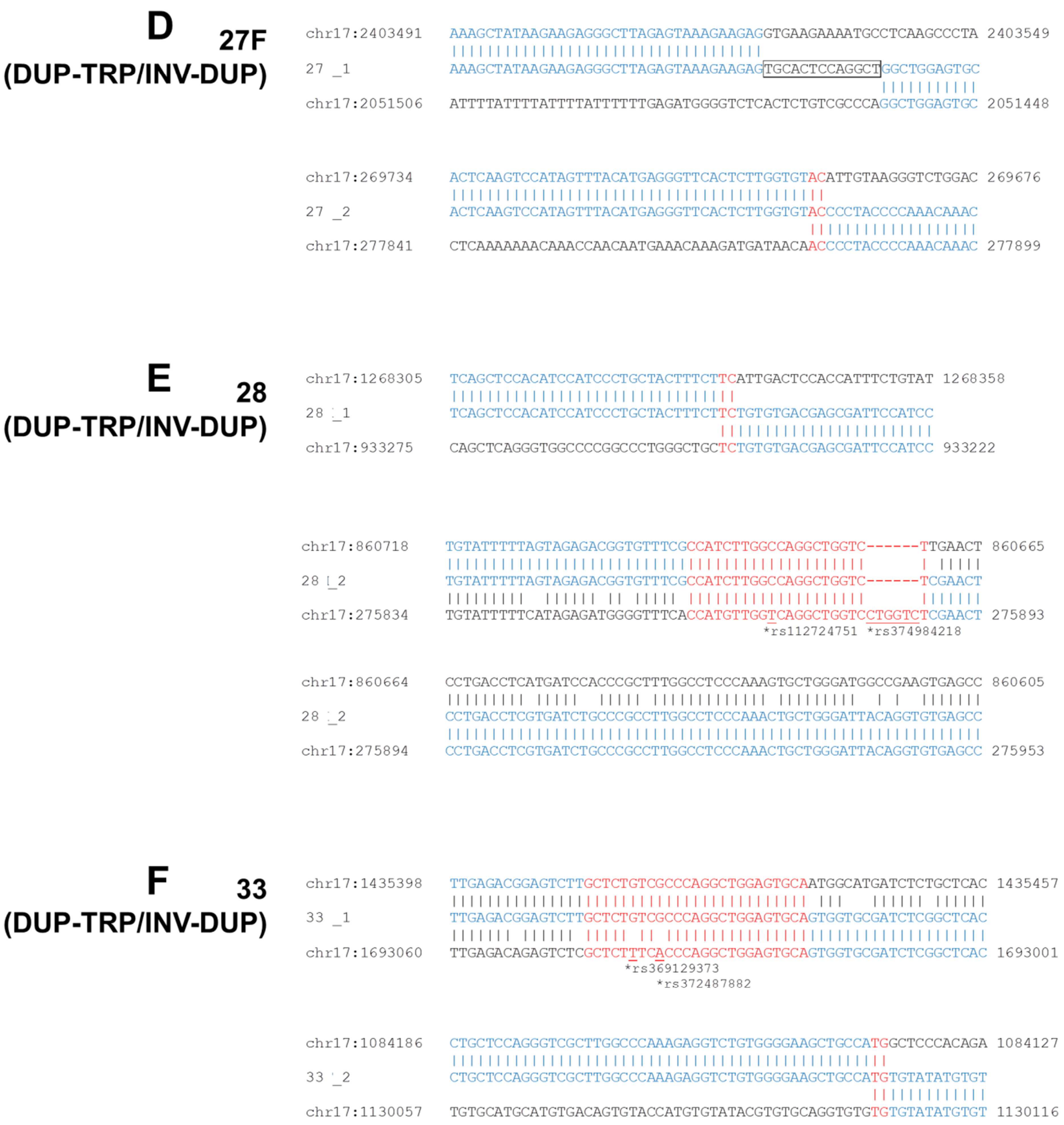


**Figure S5. Array plots and breakpoint sequences of complex rearrangements with triplication** (refer to Figure 4). Breakpoint junction sequence is aligned to the proximal and distal genomic references and color-matched. Microhomology at the breakpoint is indicated in red. Small inserted sequence is indicated by a black box in individual 27F (Figure S5D).

```
Junction 1
chr17:4334048      AGTTCAAGACCAGCCTGGCGAAGACAGTGAAACCCTGTCTCTACTAAAACTACAAAAAAATTA 4333986
                   ||||||||||
23 _1              AGTTCAAGACACATTTGCCTGTGTTTTGGATCAGTGTGTCGCGATGATCACAATGCCTTTTTT
                                                                            ||||||
chr17:99539        GCAGCGATGGAGCTTGGTTTAACCTAGTGCTTTTCAAGAGTATGTGATTAGAAACCTTTTTTT 99477

Junction 2
chr17:4359046      CCCAGGGTCCATTCTCCCCGAATGCCAGATGACATGCATCTGAGTGTCCTAAAGAC 4359101
                   |||||||||||||||
23 _2              CCCAGGGTCCATTCTGACCAGCAGCCTCCCAAAGCGCTAGGATTACAGGCGTGAGC
                                ||||||||||||||||||||||||||||||||||||||||||
chr17:5986795      GTGACCTTTATAACTGACCAGCAGCCTCCCAAAGCGCTAGGATTACAGGCGTGAGC 5986850

Junction 3
chr17:6000851      TCTCACGTCGCGACACAAAGTGCTTTCTCATTCTTTTTACCATTGCACGGAGGTCCTAT 6000910
                   |||||||||||||||||||
23 _3              TCTCACGTCGCGACACAAACAGCAACCTGTTTGTGTCCTGGAACCACACTTCCCAACAAG
                                                     ||||||||||||||||||||||||||
chr17:1966846      AAAGAAAGTTCTGAGGCTGGGGTGGGGCCCAGCAGTCCTGGAACCACACTTCCCAACAAG 1966905

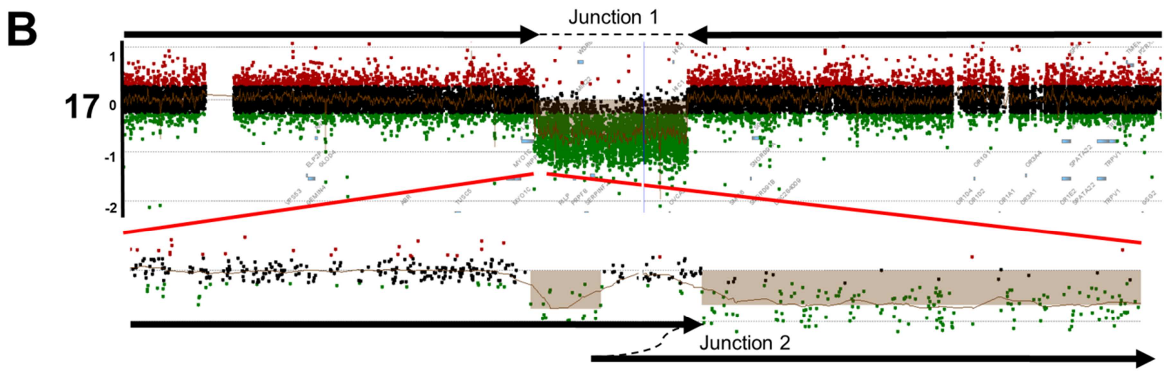
Junction 4
chr17:6451754      TTACCACAAGGGATGACTCTAGAGTTGCTCGGCTGGGACTAGCCTCACGGGGGGCAGGAA 6451813
                   ||||||||||||||||||||||||||||||
23 _4              TTACCACAAGGGATGACTCTAGAGTTGCTCCGGGCCCACCGAGAGGCTCAGGCTCCCCAA
                                                                       ||||||||
chr17:7697573      AAATGTTCCTGATGGCTGCCATGGGCCCCCCTGGGGGTGGACGGACTGTCATCTCCCCAA 7697632

                   CGGGCCCACCGAGAGGCTCAGG : chr17:8958078-8958057

Junction 5
                               *rs142763445
chr17:9856287      TTCGACCCAGCAATCCCATTACTGGGTATATACCCAAAGGAATATAAATCTTTCCACATA 9856346
                   ||||||||||||| |||||||||||||||||||||||||||||||||||
23 _5              TTCGACCCAGCAAGCCCATTACTGGGTATATACCCAAAGGAATATAAATATAAAAGAAAA
                                                                     ||||||||||
chr17:9845519      CAAGACTGCGCAACTACACTCCAGCCTGGATGACAGAGCGAGACTCTGTCTAAAAGAAAA 9845578

Junction 6
chr17:10091013     TGGCTGTTTCCAAAAAAAATCAAATCCACCCCTCAAAAGACAAAGATTTGCTGTTACT 10091070
                   ||||||||||||||
23 _6:1            TGGCTGTTTCCAAACAGACTGAGGCTATGTTTTAAAGAGGAATGACATGGCACTGGAA 58
                                          |||||||||||||||||||||||||||||||||||
chr17:1512390      TCGTTTTTCATAGGATCAGAGTGGCTATGTTTTAAAGAGGAATGACATGGCACTGGAA 1512447

23 _6:59           GGGACATGGAGGAAAAGACACTCTGTACACGTACTCGTGTACGTGCTGGTGGGC 99
                   |||||||||||||||||||||||             ||||||||||||||||||
chr17:1512448      GGGACATGGAGGAAAAGACACTC-------------GTGTACGTGCTGGTGGGC 1512488
```

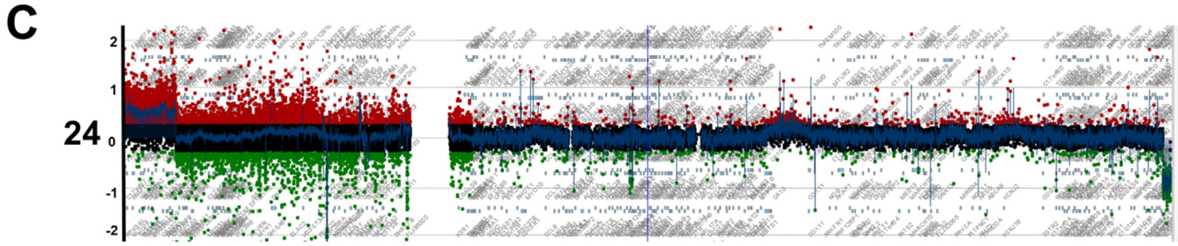


Junction 1

```
chr17:1465452  TCGGGTCCACTTGGGCCTCCGAGGCCCCCGGGGCGCCTGAGGAGCCGTCCGCCCGGGTTG 1465511
               |||||||||||||||||||||
17 '_1         TCGGGTCCACTTGGGCCTCCGTGTCCCTGCTGACAGGAAAGAAAGAGTGAAAGAAATTCT
                              |||||||||||||||||||||||||||||||||||||||||||||
chr17:2010037  AAGGCAGACAGGAAACCTCCGTGTCCCTGCTGACAGGAAAGAAAGAGTGAAAGAAATTCT 2010096
```

Junction 2

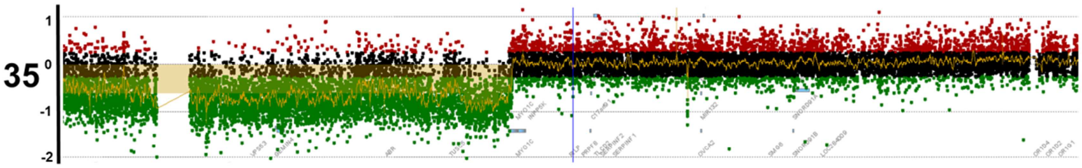
```
chr17:1478998  CAGATCAGGTAGAGCGGGAGGCAGAAGCCCAGCAGGCTGAGAAGGAGCAGCCCCACGTTCACCT 1479061
               ||||||||||||||||||||||
17 _2          CAGATCAGGTAGAGCGGGAGGCTACTACCACCACGCCCGGCTACTTTTTTGTATTTTTAGTAGA
                                           ||||||||||||||||||||||||||||||||||||
chr17:1471932  CCGAGTAGCTGGGAGTACAGGCACCCGTCACCACGCCCGGCTACTTTTTTGTATTTTTAGTAGA 1471995
```



```
chr17:80508491  GCCTCAGCCTCCCGAGTAGCTGGGACTACAGGCGCCCGCCACC--ACGCCCGGCTAATTT 80508548
                |||||||||||||||||||||||||||||||||||||||||||  | | | ||  | ||
24              GCCTCAGCCTCCCGAGTAGCTGGGACTACAGGCGCCCGCCACCTGAGGTCAGG--AGTTC
                  | ||||     | |  || | | |   ||||      ||||||||||||||  |||||
chr17:4096993   ATCCCAGCACTTTGGGAGGCCGAGGC---AGGCAG--ATCACCTGAGGTCAGG--AGTTC 4096941
```

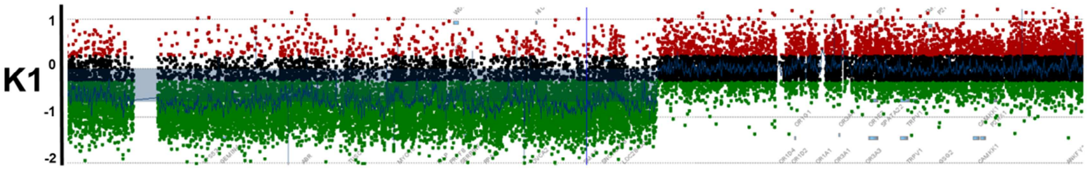
**Figure S6. Array plots and breakpoint sequences of additional cases with complex rearrangements.** Breakpoint junction sequence is aligned to the proximal and distal genomic references and color-matched. Microhomology at the breakpoint is indicated in red. Inserted sequences at the breakpoint junctions are underlined.

**A**



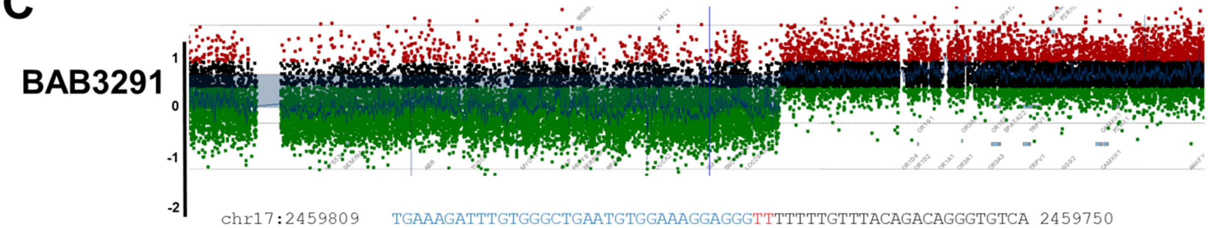
```
chr17:1964448     TCTCGCCTGGGGACGCAGCCACTTTCTCCAGTGGAGACAGGGCAGGGGTTCTGAGTTTCCGTCA 1964385
                  ||||||||||||||||||||||||||||||||||||||||||
35                TCTCGCCTGGGGACGCAGCCACTTTCTCCAGTGGAGACAGGGTTAGGGTTAGGGTTAGGGTTAG
                                                        ||||||||||||||||||||||||||
TEL               TTAGGGTTAGGGTTAGGGTTAGGGTTAGGGTTAGGGTTAGGGTTAGGGTTAGGGTTAGGGTTAG
```

**B**

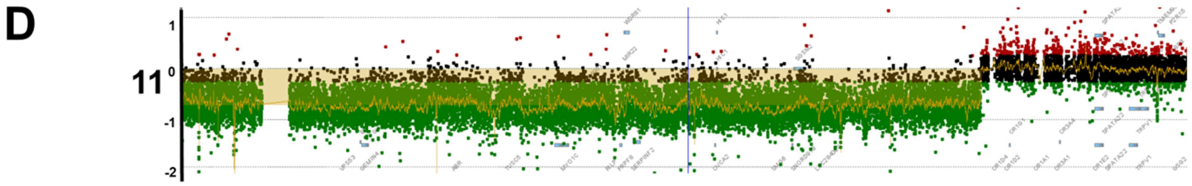


```
chr17:2455955     TTTTTTTTTTTTGAGATGGAGTTTCGCTCTTGTTGCACAGGCTGGAGTGCAATGGCATAATCT 2455893
                  |||||||||||||||||||||||||||||||||||||||||||
K1                TTTTTTTTTTTTGAGATGGAGTTTCGCTCTTGTTGCACAGGCTTAGGGTTAGGGTTAGGGTTA
                                                            |||||||||||||||||||||
TEL               TTAGGGTTAGGGTTAGGGTTAGGGTTAGGGTTAGGGTTAGGGTTAGGGTTAGGGTTAGGGTTA
```

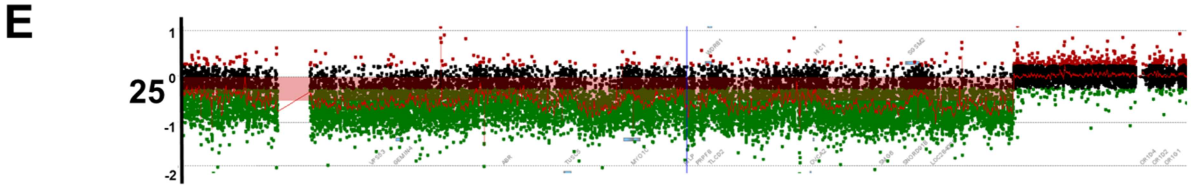
**C**

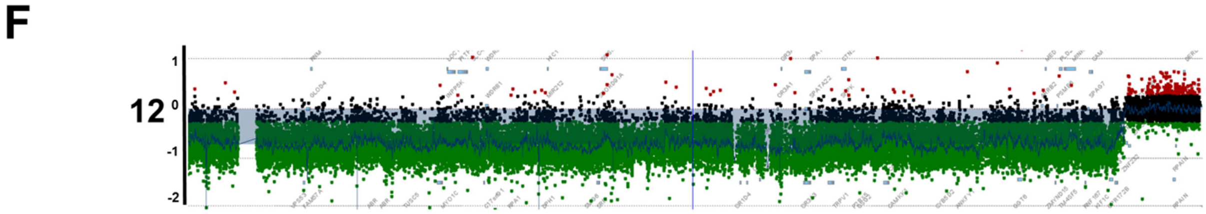


```
chr17:2459809     TGAAAGATTTGTGGGCTGAATGTGGAAAGGAGGGTTTTTTTGTTTACAGACAGGGTGTCA 2459750
                  ||||||||||||||||||||||||||||||||||||
BAB3291           TGAAAGATTTGTGGGCTGAATGTGGAAAGGAGGGTTAGGGTTAGGGTTAGGGTTAGGGTT
                                                    ||||||||||||||||||||||||||
TEL               AGGGTTAGGGTTAGGGTTAGGGTTAGGGTTAGGGTTAGGGTTAGGGTTAGGGTTAGGGTT
```



```
chr17:2924445   GTGATGGATATTTCTCTTACCTTGATTTGATCATTATGCATTTTATCAAAATATCACACGTACCC 2924381
                ||||||||||||||||||||||||||||||||||||||
11              GTGATGGATATTTCTCTTACCTTGATTTGATCATTATGGTTAGGGTTAGGGTTAGGGTTAGGGTT
                                                       ||||||||||||||||||||||||||
TEL             GGGTTAGGGTTAGGGTTAGGGTTAGGGTTAGGGTTAGGGTTAGGGTTAGGGTTAGGGTTAGGGTT
```





```
chr17:5061250   AACAATTGTTCAGGGTAAAAAGCCAGCCTGGGCCGGGTGCAGTGGCTCACGCCT  5061197
                ||||||||||||||||||||||||||||||||||||||||||||||
12 :1           AACAATTGTTCAGGGTAAAAAGCCAGCCTGGGCCGGGTGCAGTGGCCGAGGGAT  54
                                                              ||||
chr17:5059818   CTCAAATTCCCAACCTCAGGTGATCCGCCTGCCTCAGCCTCCCAAAGTGCGGAT  5059869

chr17:5059874   TACAGGCGTGAGCCACCTCACCCAGCCTGTAGATCATTTTCTTGCCATCATGGT 5059927
                ||||||||||||||||||||||||||||||||||||||||||||||||||||||
12 :55          TACAGGCGTGAGCCACCTCACCCAGCCTGTAGATCATTTTCTTGCCATCATGGT  108

chr17:5059928   ACAAATGAGAGTTCTGTTTTATGGTAACTAAAAAAAGTTCTTTACTTATTTGTT 5059981
                |||||||||||||||||||||||||||||||||||||||
12 :109         ACAAATGAGAGTTCTGTTTTATGGTAACTAAAAAAAGTTAGGGTTAGGGTTAGG  162
                                                    ||||||||||||||||||
TEL             GTTAGGGTTAGGGTTAGGGTTAGGGTTAGGGTTAGGGTTAGGGTTAGGGTTAGG
```

**Figure S7. Array plots and breakpoint sequences of additional cases with terminal deletion** (refer to Figure 6). Breakpoint junction sequence is aligned to the proximal and distal genomic references and color-matched. Microhomology at the breakpoint is indicated in red.