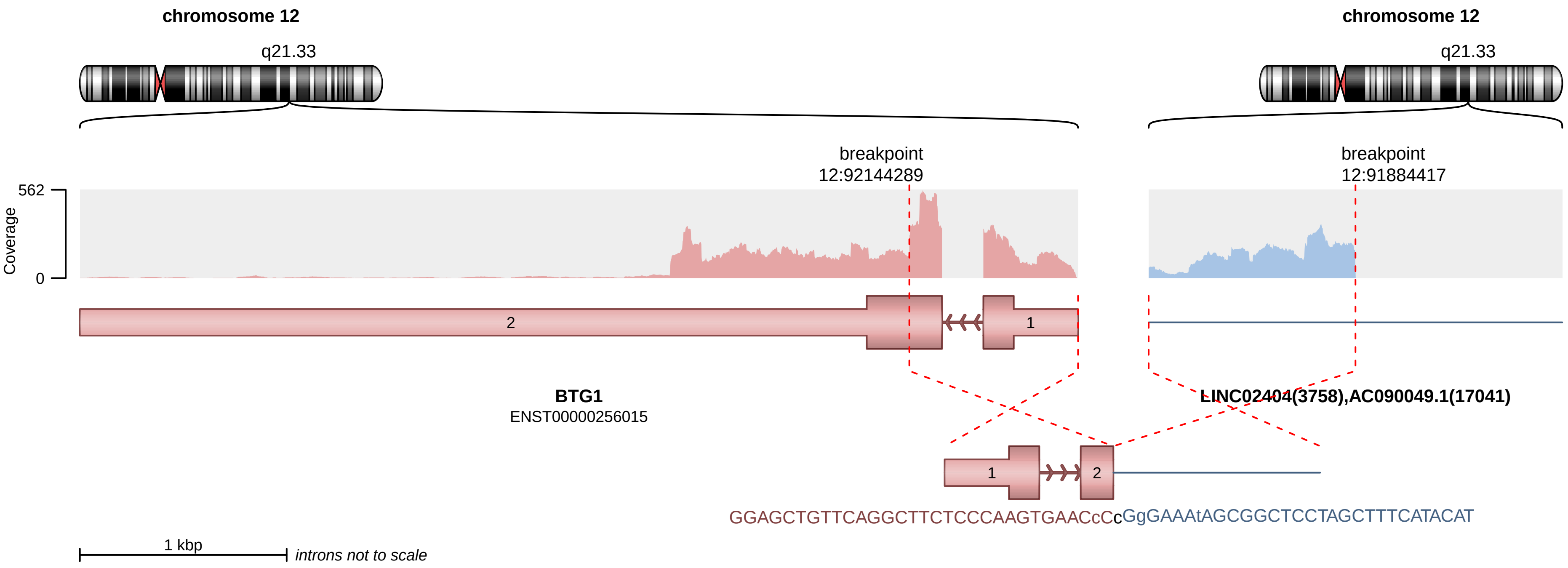
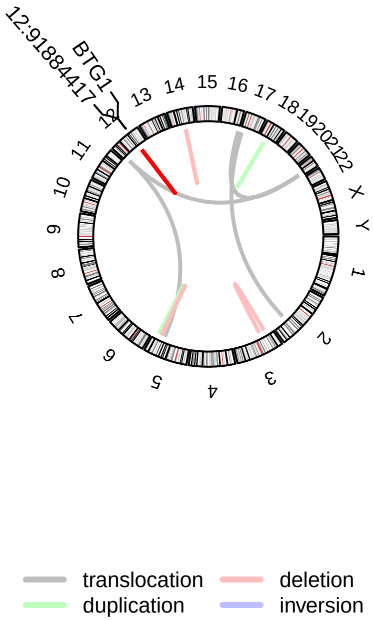
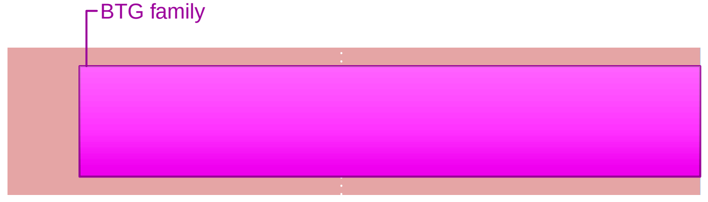

chromosome 12
q21.33
chromosome 12
q21.33
breakpoint
12:92144289
breakpoint
12:91884417
562
Coverage
0
2
1
BTG1
ENST00000256015
LINC02404(3758),AC090049.1(17041)
1
2
GGAGCTGTTCAGGCTTCTCCCAAGTGAACcCcGgGAAAtAGCGGCTCCTAGCTTTCATACAT
1 kbp
introns not to scale


12:91884417
BTG1
12
13
14
15
16
17
18
19
20
21
22
X
Y
1
2
3
4
5
6
7
8
9
10
11
translocation
deletion
duplication
inversion

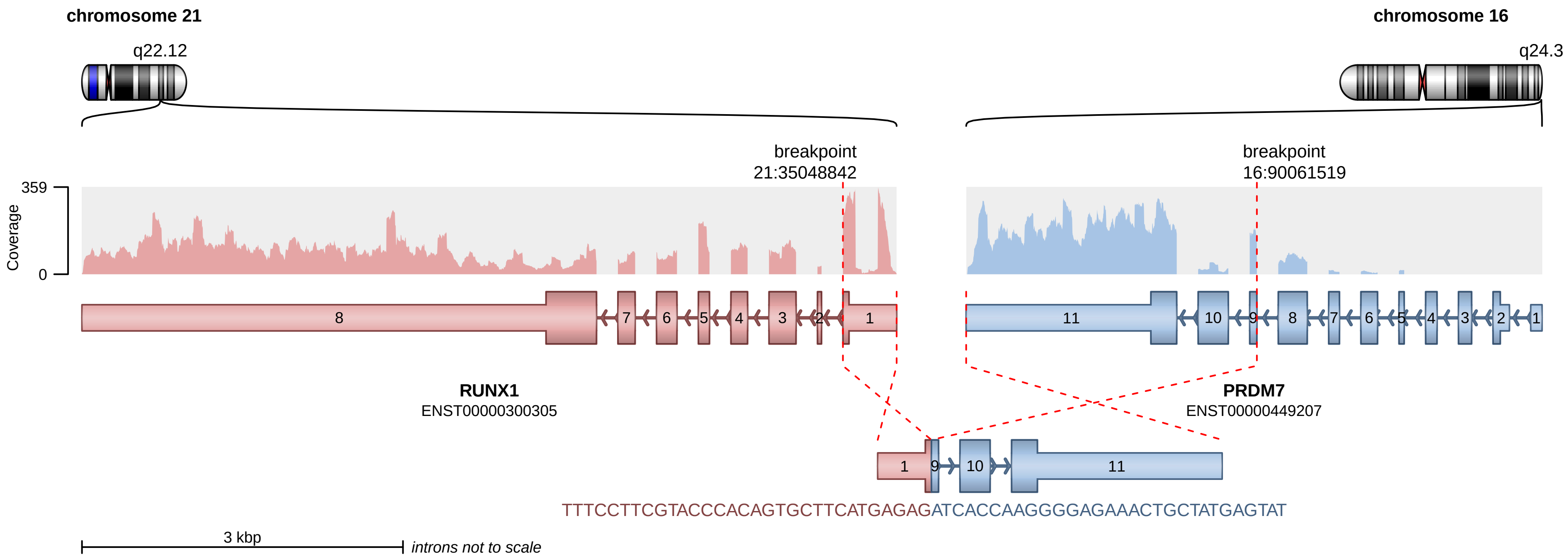
RETAINED PROTEIN DOMAINS
out−of−frame fusion

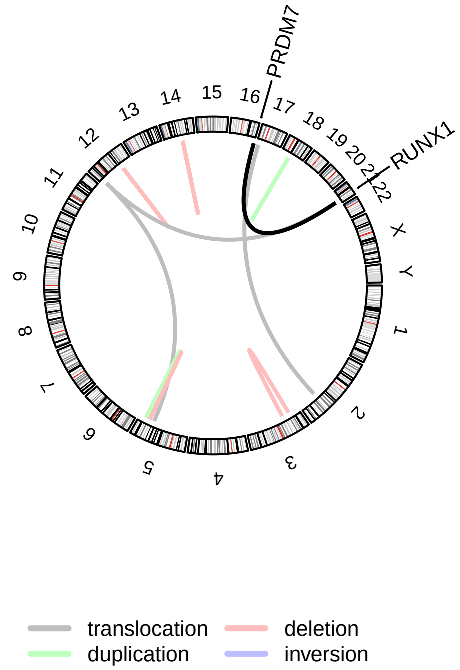


BTG1

SUPPORTING READ COUNT

Split reads = 88
Discordant mates = 32

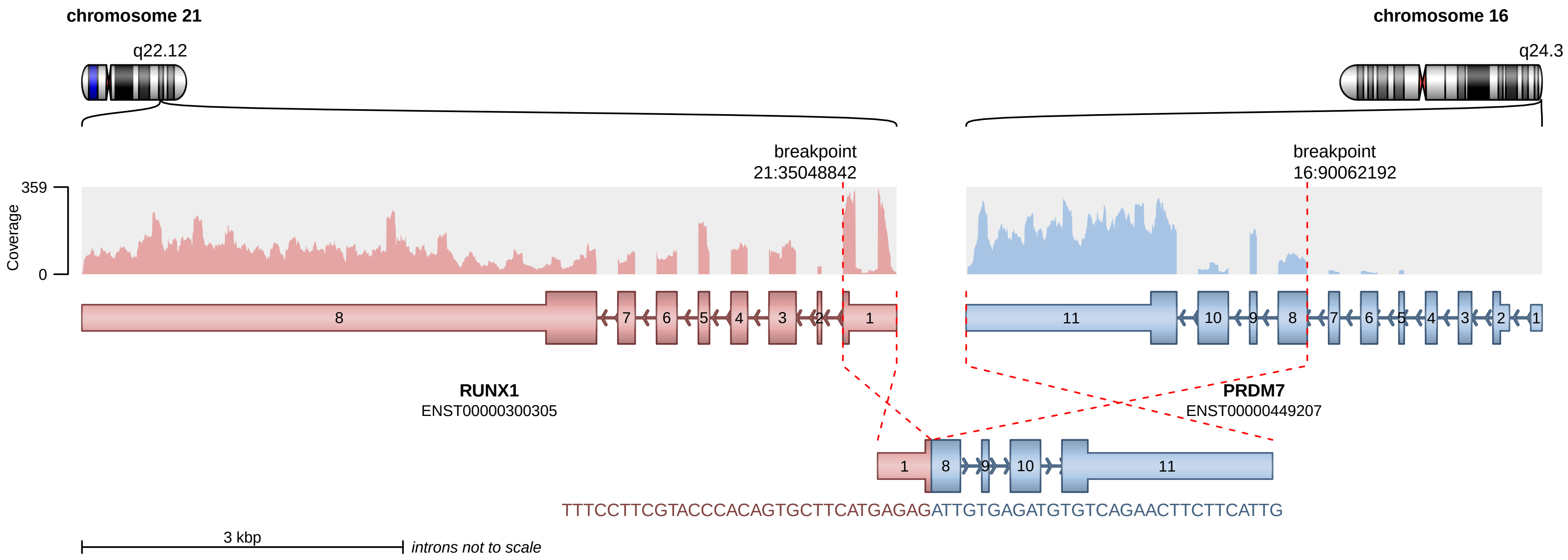

chromosome 21
q22.12
chromosome 16
q24.3
breakpoint
21:35048842
breakpoint
16:90061519
Coverage
359
0
8
7
6
5
4
3
2
1
11
10
9
8
7
6
5
4
3
2
1
RUNX1
ENST00000300305
PRDM7
ENST00000449207
1
9
10
11
TTTCCTTCGTACCCACAGTGCTTCATGAGAGATCACCAAGGGGAGAAACTGCTATGAGTAT
3 kbp
introns not to scale

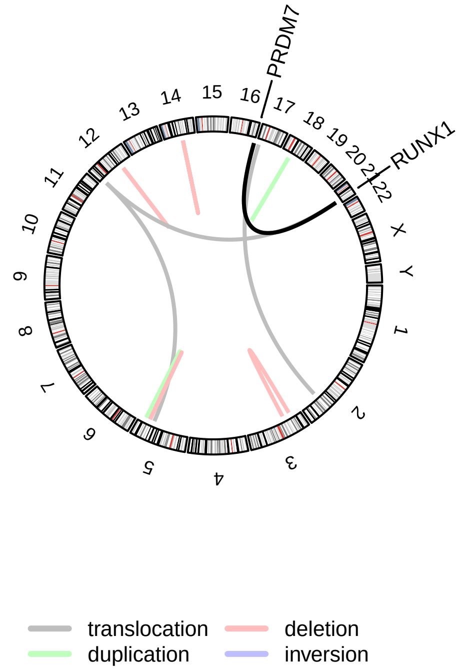

PRDM7
RUNX1
1
2
3
4
5
6
7
8
9
10
11
12
13
14
15
16
17
18
19
20
21
22
X
Y
translocation
deletion
duplication
inversion

No protein domains retained in fusion.

SUPPORTING READ COUNT

Split reads = 54
Discordant mates = 16

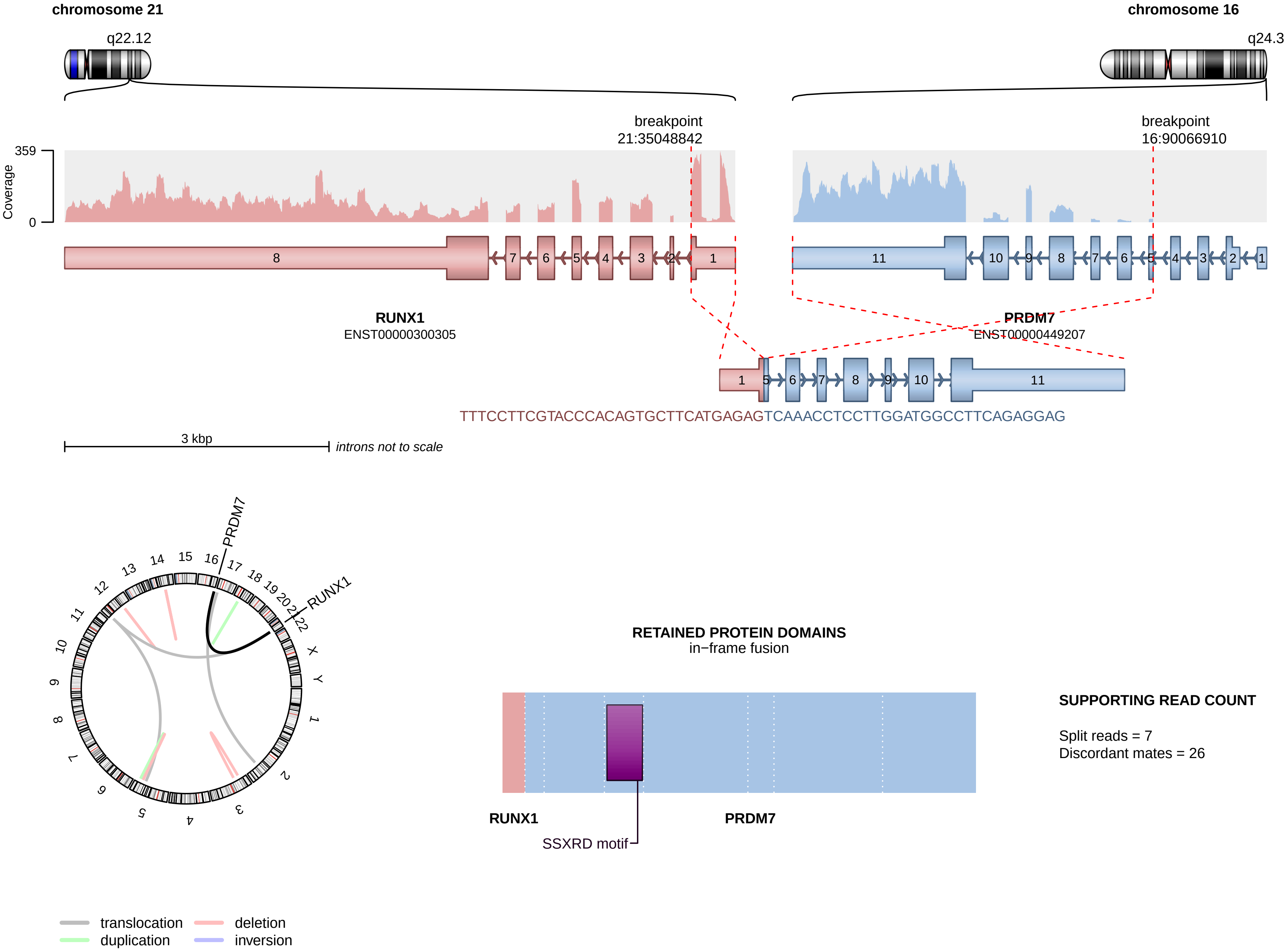

chromosome 21
q22.12
chromosome 16
q24.3
breakpoint
21:35048842
breakpoint
16:90062192
Coverage
359
0
8
7
6
5
4
3
2
1
11
10
9
8
7
6
5
4
3
2
1
RUNX1
ENST00000300305
PRDM7
ENST00000449207
1
8
9
10
11
TTTCCTTCGTACCCACAGTGCTTCATGAGAGATTGTGAGATGTGTCAGAACTTCTTCATTG
3 kbp
introns not to scale

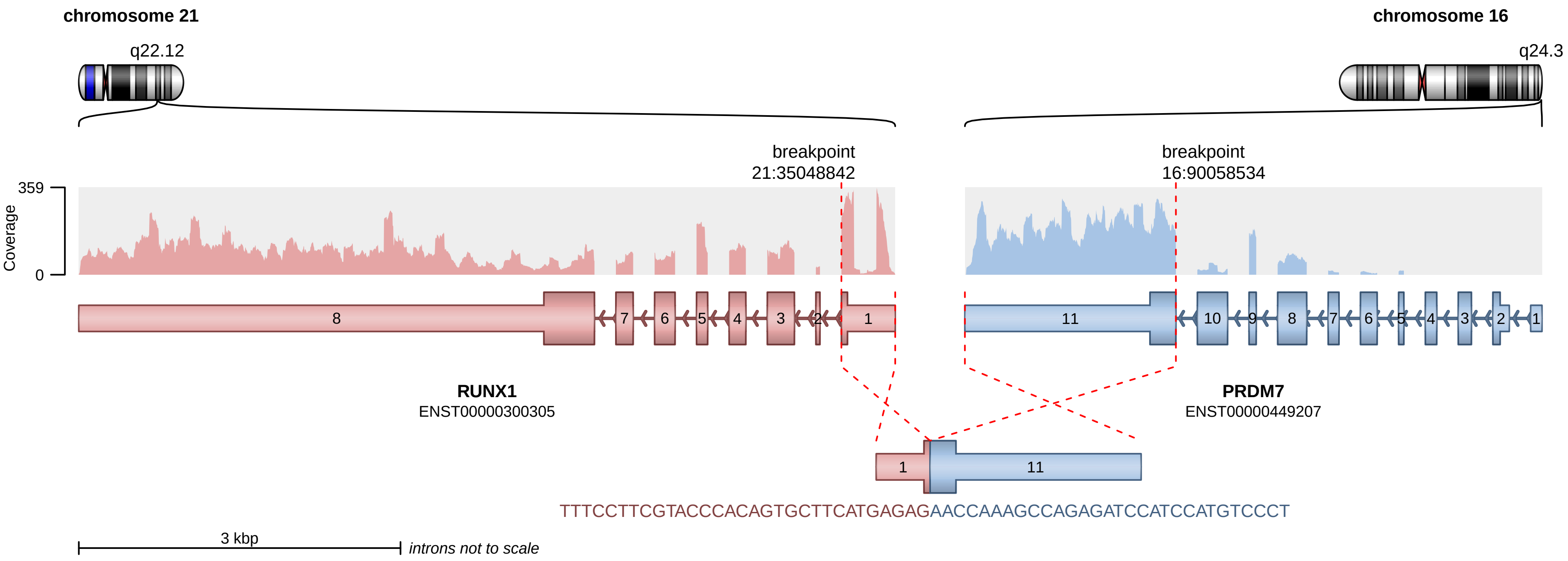

PRDM7
RUNX1
1
2
3
4
5
6
7
8
9
10
11
12
13
14
15
16
17
18
19
20
21
22
X
Y
translocation
deletion
duplication
inversion

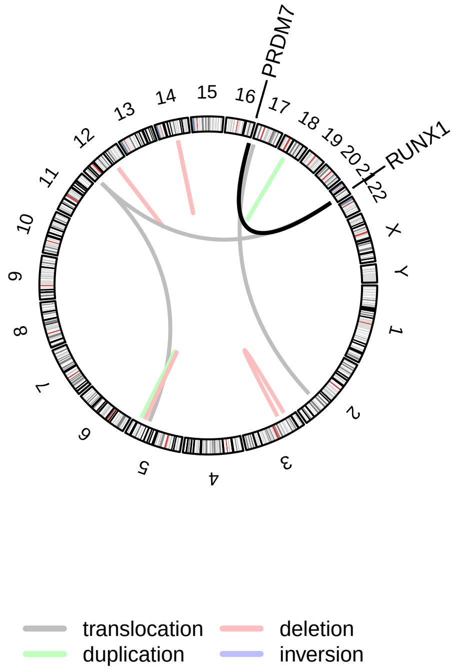
No protein domains retained in fusion.

SUPPORTING READ COUNT

Split reads = 12
Discordant mates = 24


chromosome 21
q22.12
chromosome 16
q24.3
breakpoint
21:35048842
breakpoint
16:90066910
Coverage
359
0
8
7
6
5
4
3
2
1
RUNX1
ENST00000300305
11
10
9
8
7
6
5
4
3
2
1
PRDM7
ENST00000449207
1
5
6
7
8
9
10
11
TTTCCTTCGTACCCACAGTGCTTCATGAGAGTCAAACCTCCTTGGATGGCCTTCAGAGGAG
3 kbp
introns not to scale
PRDM7
RUNX1
1
2
3
4
5
6
7
8
9
10
11
12
13
14
15
16
17
18
19
20
21
22
X
Y
translocation
duplication
deletion
inversion
RETAINED PROTEIN DOMAINS
in−frame fusion
RUNX1
PRDM7
SSXRD motif
SUPPORTING READ COUNT
Split reads = 7
Discordant mates = 26

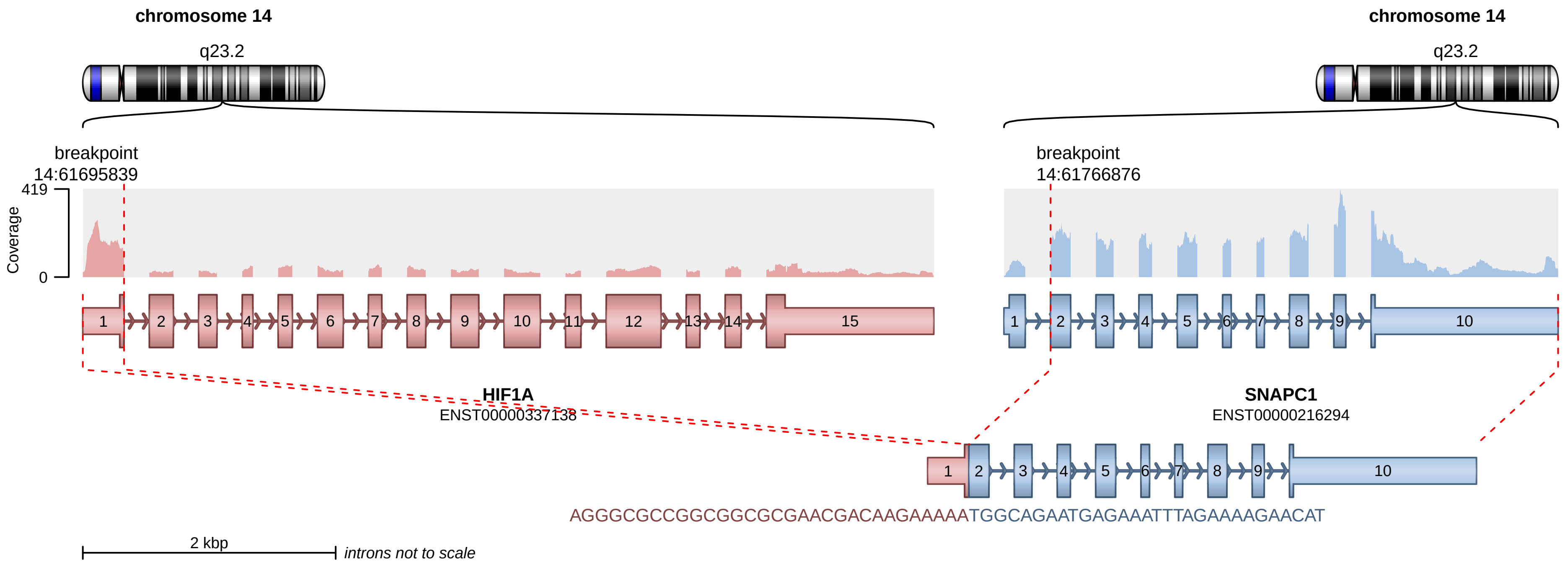

chromosome 21
q22.12
chromosome 16
q24.3
breakpoint
21:35048842
breakpoint
16:90058534
Coverage
359
0
8
7
6
5
4
3
2
1
11
10
9
8
7
6
5
4
3
2
1
RUNX1
ENST00000300305
PRDM7
ENST00000449207
1
11
TTTCCTTCGTACCCACAGTGCTTCATGAGAGAACCAAAGCCAGAGATCCATCCATGTCCCT
3 kbp
introns not to scale

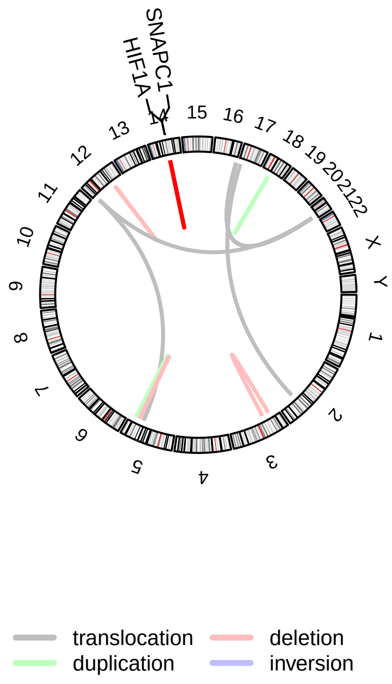

PRDM7
RUNX1
16
17
18
19
20
21
22
X
Y
1
2
3
4
5
6
7
8
9
10
11
12
13
14
15
translocation
duplication
deletion
inversion

No protein domains retained in fusion.

SUPPORTING READ COUNT

Split reads = 1
Discordant mates = 3

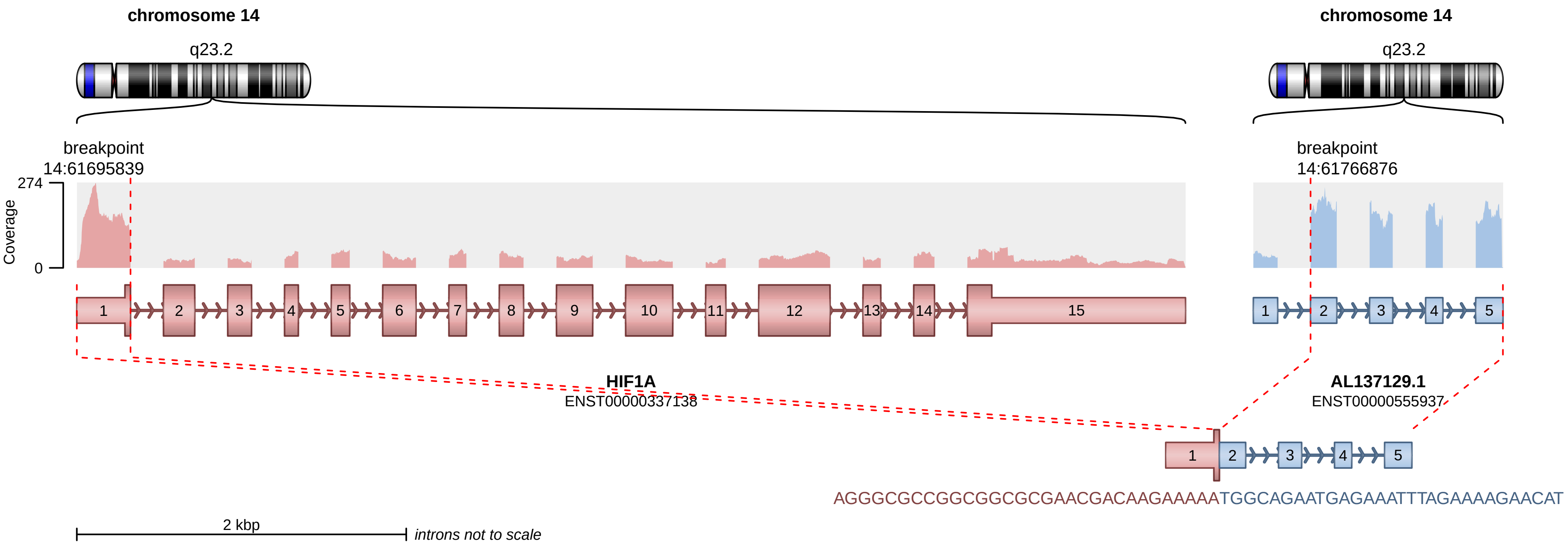

chromosome 14
q23.2
chromosome 14
q23.2
breakpoint
14:61695839
breakpoint
14:61766876
Coverage
419
0
HIF1A
ENST00000337138
SNAPC1
ENST00000216294
AGGGCGCCGGCGGCGCGAACGACAAGAAAAATGGCAGAATGAGAAATTTAGAAAAGAACAT
2 kbp
introns not to scale

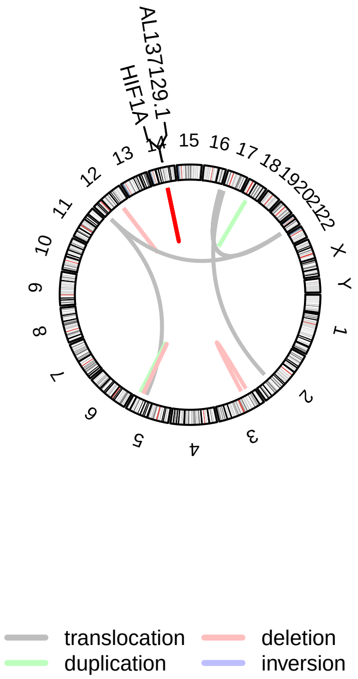


No protein domains retained in fusion.

SUPPORTING READ COUNT

Split reads = 45
Discordant mates = 20

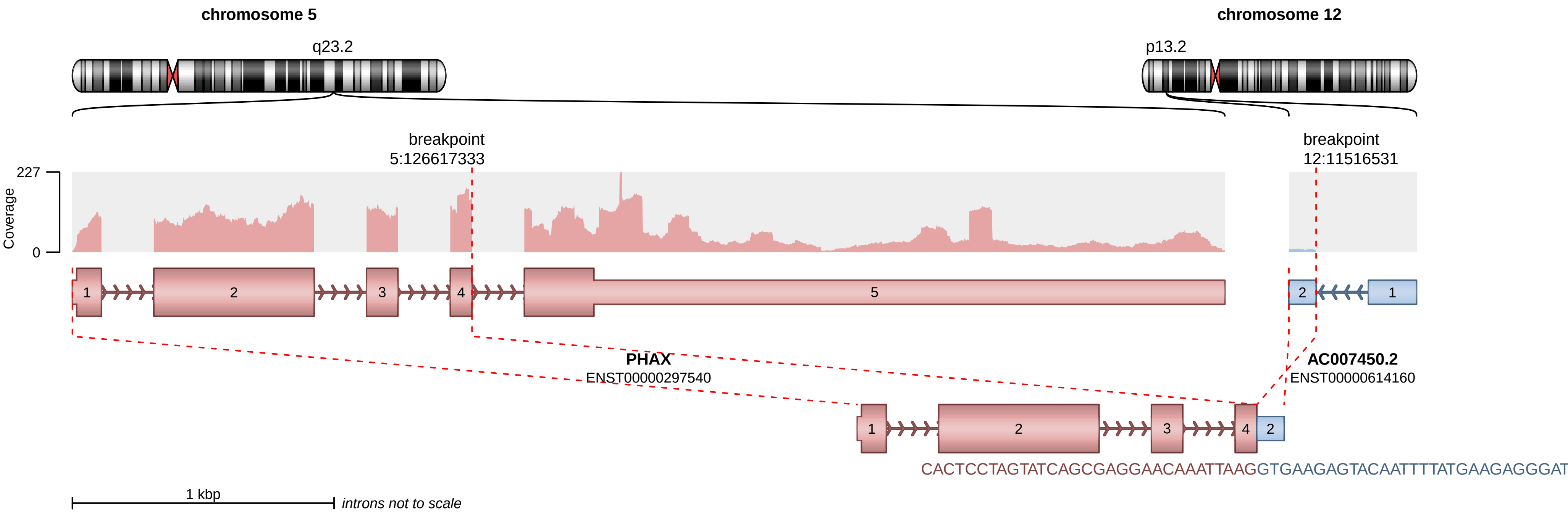

chromosome 14
q23.2
chromosome 14
q23.2
breakpoint
14:61695839
breakpoint
14:61766876
Coverage
274
0
1
2
3
4
5
6
7
8
9
10
11
12
13
14
15
1
2
3
4
5
HIF1A
ENST00000337138
AL137129.1
ENST00000555937
1
2
3
4
5
AGGGCGCCGGCGGCGCGAACGACAAGAAAAATGGCAGAATGAGAAATTTAGAAAAGAACAT
2 kbp
introns not to scale

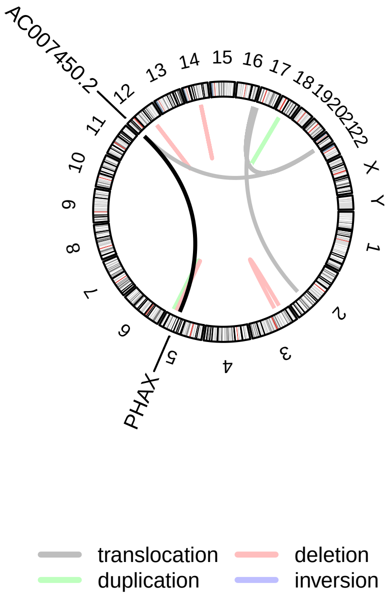

AL137129.1
HIF1A
1
2
3
4
5
6
7
8
9
10
11
12
13
14
15
16
17
18
19
20
21
22
X
Y
translocation
duplication
deletion
inversion

No protein domains retained in fusion.

SUPPORTING READ COUNT

Split reads = 45
Discordant mates = 19

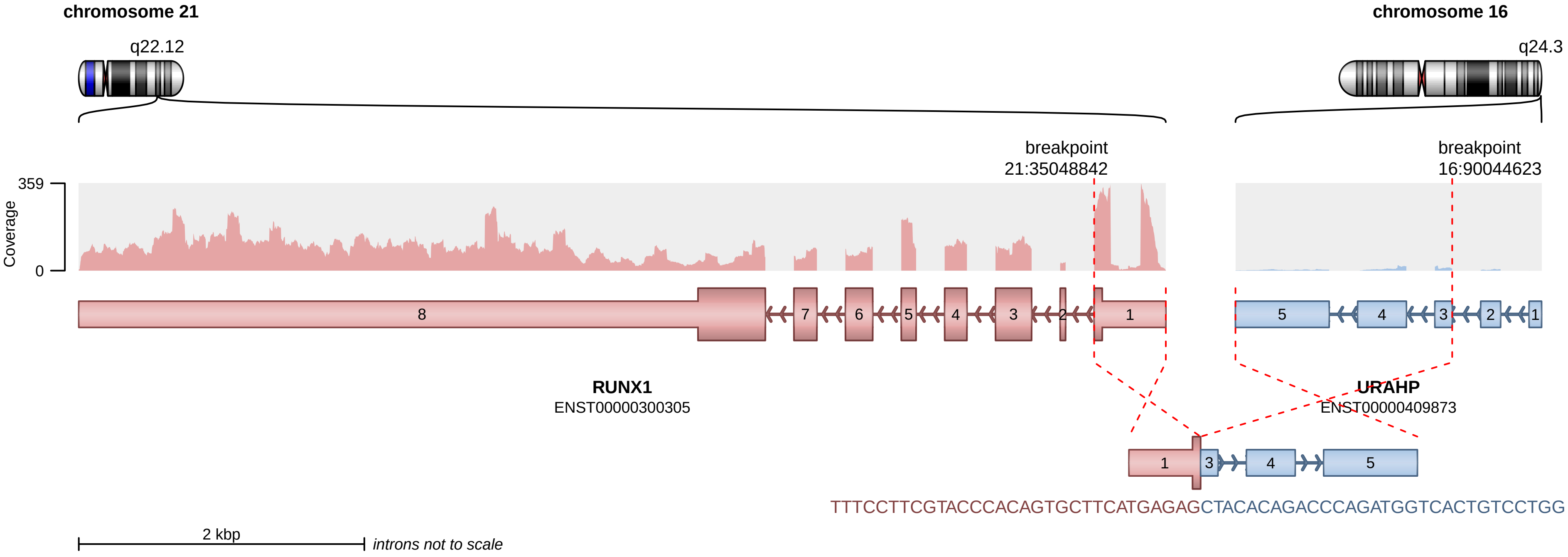

chromosome 5
q23.2
chromosome 12
p13.2
breakpoint
5:126617333
breakpoint
12:11516531
Coverage
227
0
1
2
3
4
5
2
1
PHAX
ENST00000297540
AC007450.2
ENST00000614160
1
2
3
4
2
CACTCCTAGTATCAGCGAGGAACAAATTAAGGTGAAGAGTACAATTTTATGAAGAGGGAT
1 kbp
introns not to scale

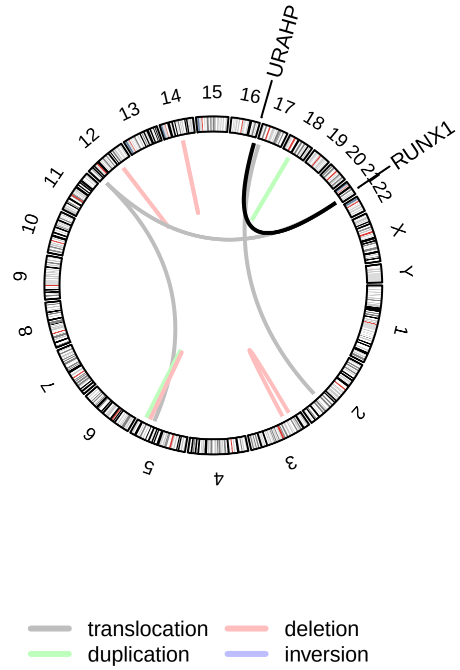

AC007450.2
PHAX
12
13
14
15
16
17
18
19
20
21
22
X
Y
1
2
3
4
5
6
7
8
9
10
11
translocation
deletion
duplication
inversion

No protein domains retained in fusion.

SUPPORTING READ COUNT

Split reads = 5
Discordant mates = 1

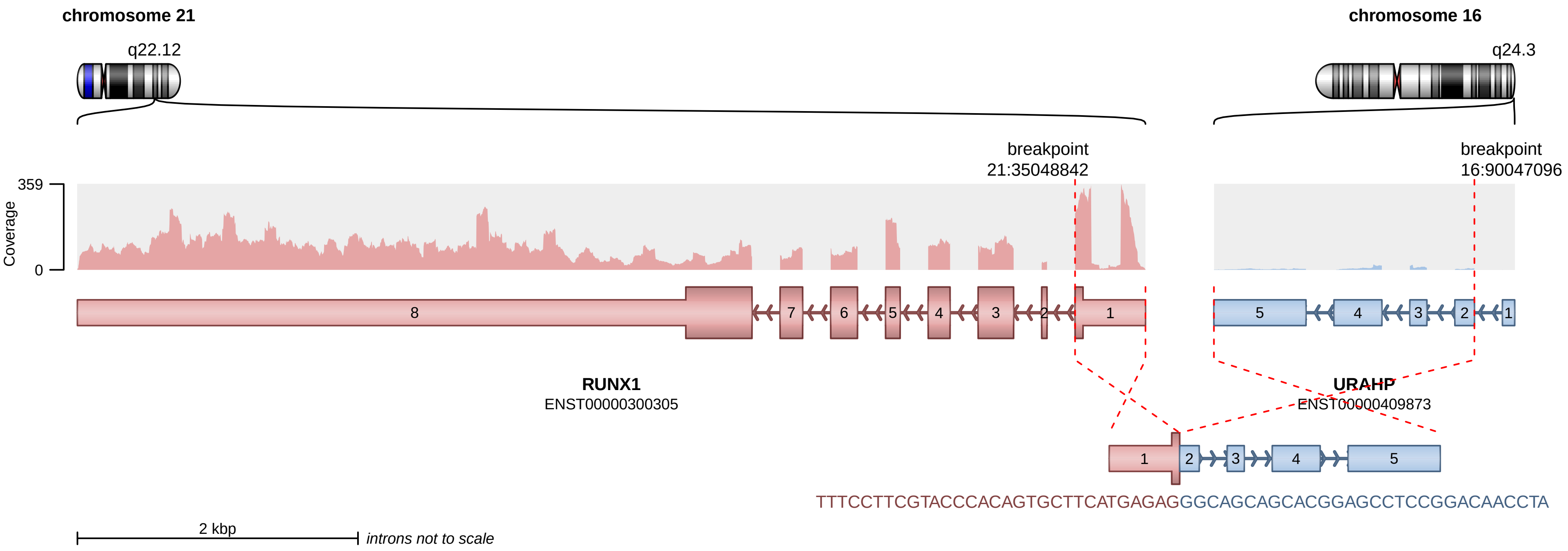

chromosome 21
q22.12
chromosome 16
q24.3
Coverage
359
0
breakpoint
21:35048842
breakpoint
16:90044623
8
7
6
5
4
3
2
1
RUNX1
ENST00000300305
5
4
3
2
1
URAHP
ENST00000409873
1
3
4
5
TTTCCTTCGTACCCACAGTGCTTCATGAGAGCTACACAGACCCAGATGGTCACTGTCCTGG
2 kbp
introns not to scale

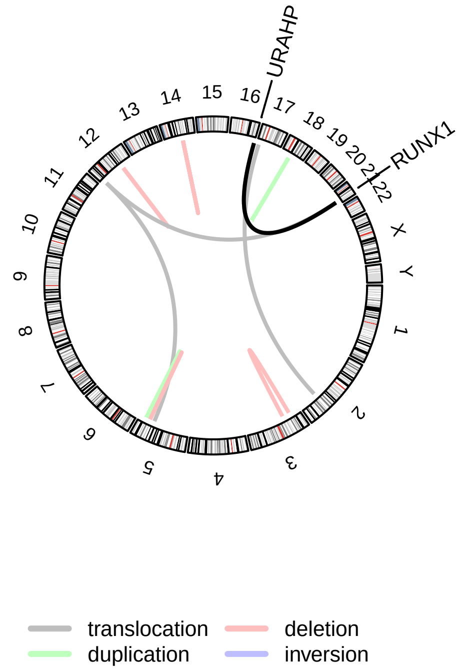


No protein domains retained in fusion.

SUPPORTING READ COUNT

Split reads = 4
Discordant mates = 0

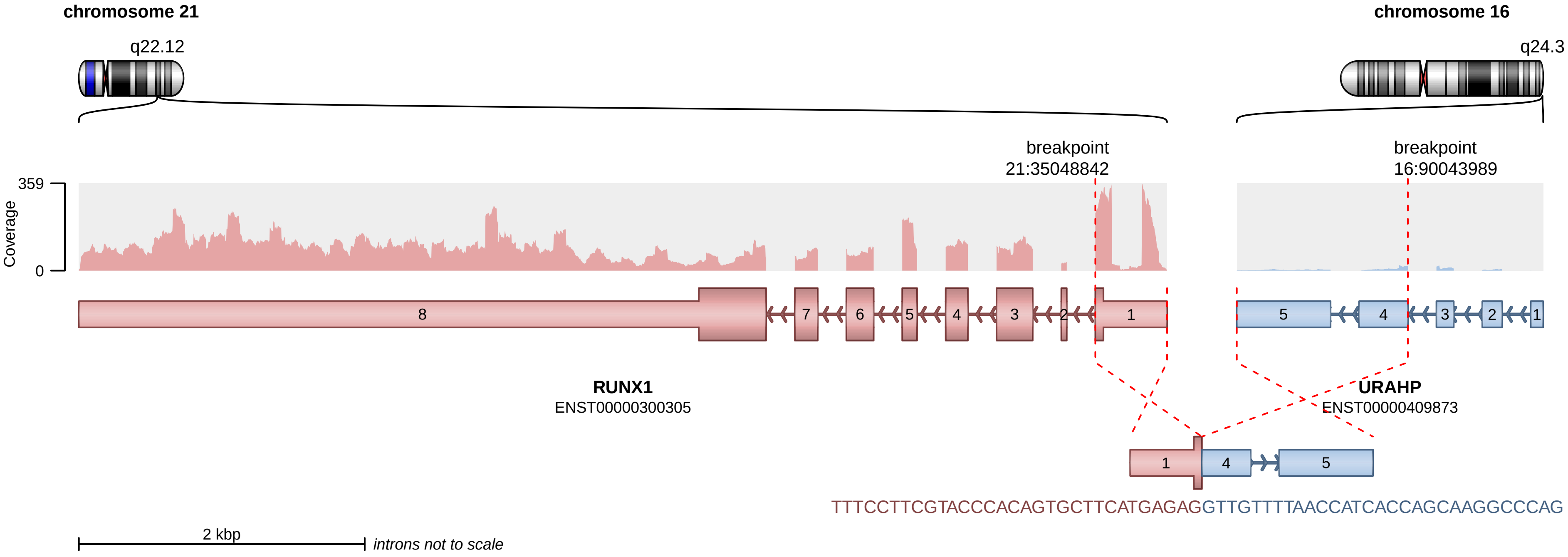

chromosome 21
q22.12
chromosome 16
q24.3
breakpoint
21:35048842
breakpoint
16:90047096
Coverage
359
0
8
7
6
5
4
3
2
1
5
4
3
2
1
RUNX1
ENST00000300305
URAHP
ENST00000409873
1
2
3
4
5
TTTCCTTCGTACCCACAGTGCTTCATGAGAGGGCAGCAGCACGGAGCCTCCGGACAACCTA
2 kbp
introns not to scale

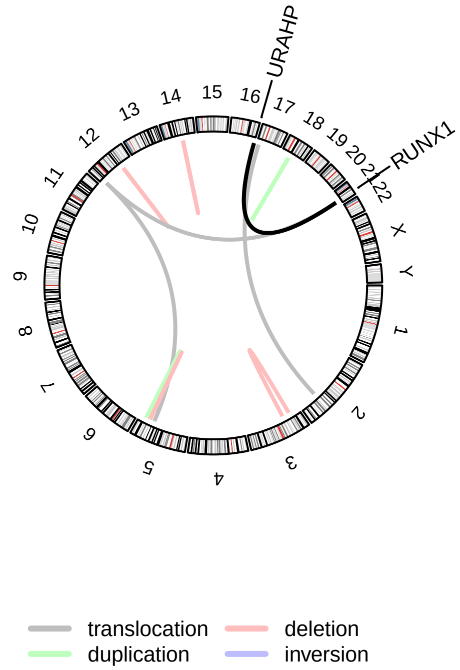

URAHP
RUNX1
16
17
18
19
20
21
22
X
Y
1
2
3
4
5
6
7
8
9
10
11
12
13
14
15
translocation
duplication
deletion
inversion

No protein domains retained in fusion.

SUPPORTING READ COUNT

Split reads = 2
Discordant mates = 0

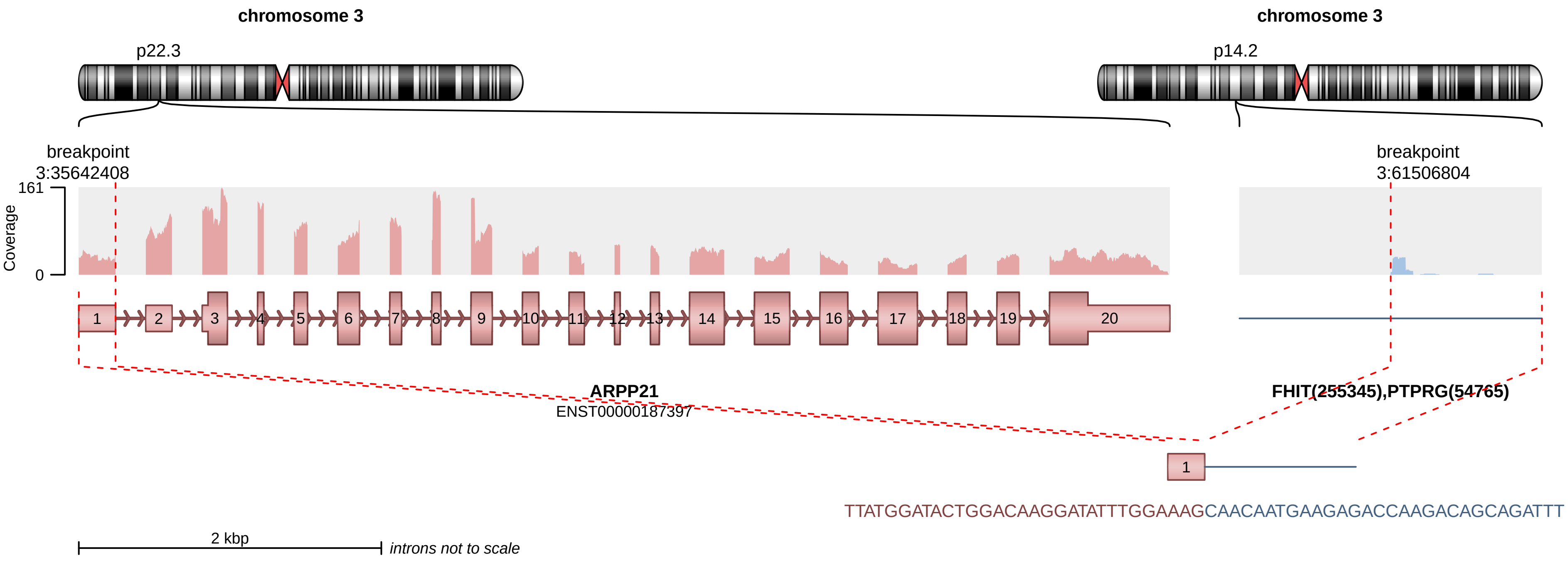

chromosome 21
q22.12
chromosome 16
q24.3
breakpoint
21:35048842
breakpoint
16:90043989
Coverage
359
0
8
7
6
5
4
3
2
1
5
4
3
2
1
RUNX1
ENST00000300305
URAHP
ENST00000409873
1
4
5
TTTCCTTCGTACCCACAGTGCTTCATGAGAGGTTGTTTTAACCATCACCAGCAAGGCCCAG
2 kbp
introns not to scale

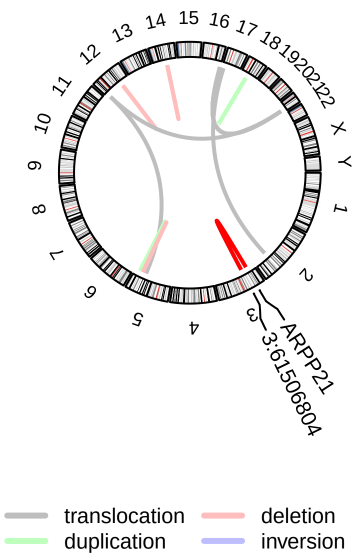


No protein domains retained in fusion.

SUPPORTING READ COUNT

Split reads = 2
Discordant mates = 0

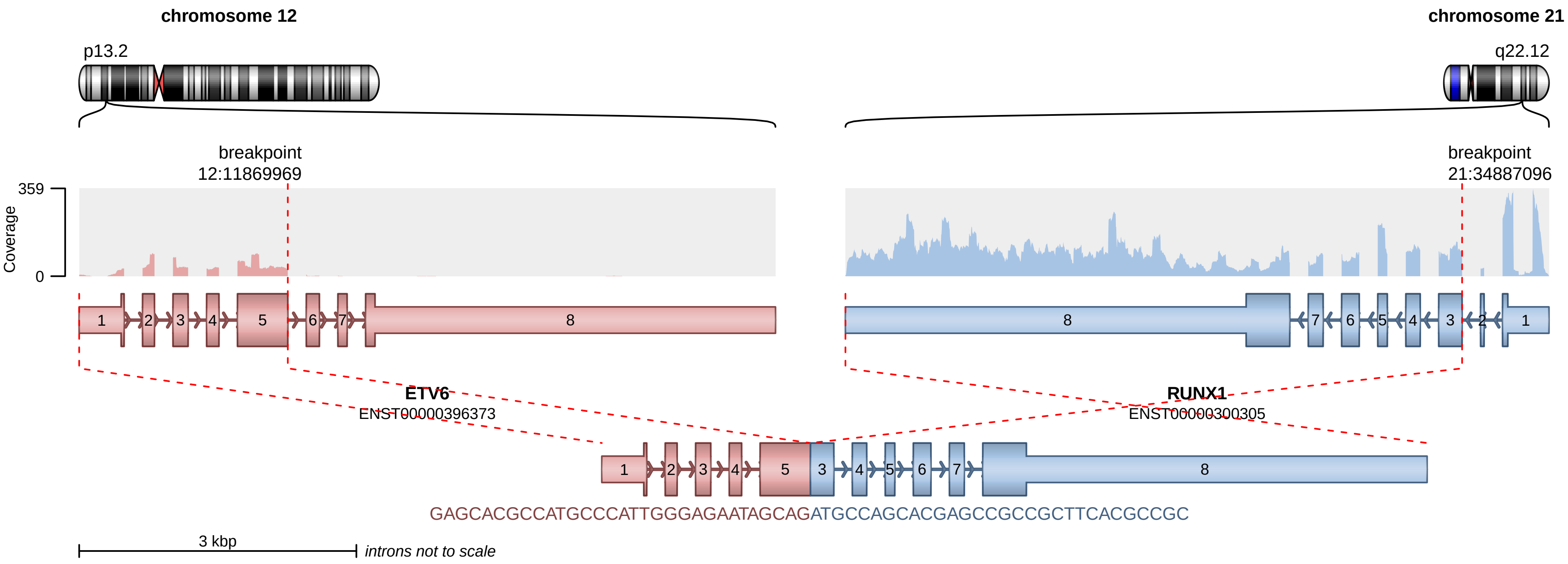

chromosome 3
p22.3
chromosome 3
p14.2
breakpoint
3:35642408
breakpoint
3:61506804
Coverage
161
0
1
2
3
4
5
6
7
8
9
10
11
12
13
14
15
16
17
18
19
20
ARPP21
ENST00000187397
FHIT(255345),PTPRG(54765)
1
TTATGGATACTGGACAAGGATATTTGGAAAGCAACAATGAAGAGACCAAGACAGCAGATTT
2 kbp
introns not to scale

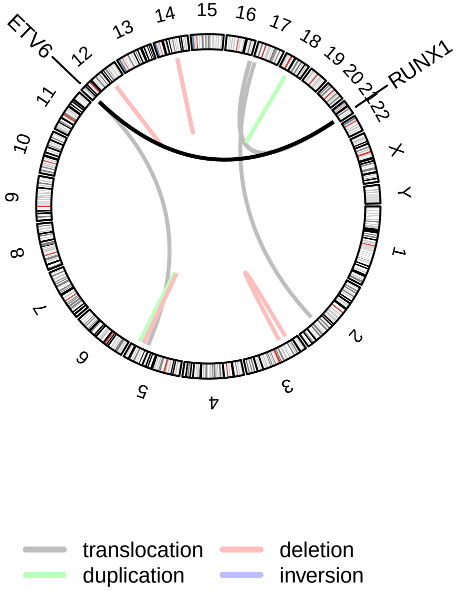

13
14
15
16
17
18
19
20
21
22
X
Y
1
2
3
4
5
6
7
8
9
10
11
12
ARPP21
3:61506804
translocation
duplication
deletion
inversion

No coding regions retained in fusion transcript.

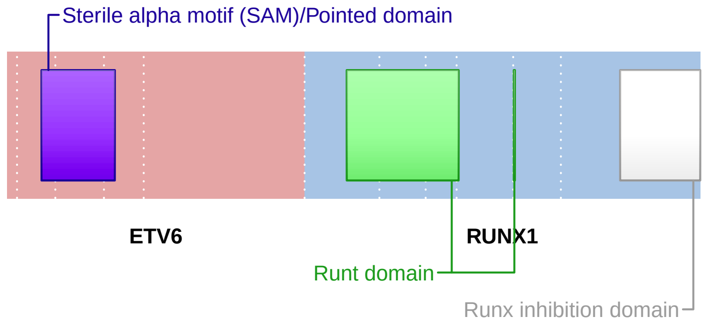
SUPPORTING READ COUNT

Split reads = 2
Discordant mates = 1


chromosome 12
p13.2
chromosome 21
q22.12
breakpoint
12:11869969
breakpoint
21:34887096
Coverage
359
0
1
2
3
4
5
6
7
8
ETV6
ENST00000396373
RUNX1
ENST00000300305
GAGCACGCCATGCCCATTGGGAGAATAGCAGATGCCAGCACGAGCCGCCGCTTCACGCCGC
3 kbp
introns not to scale

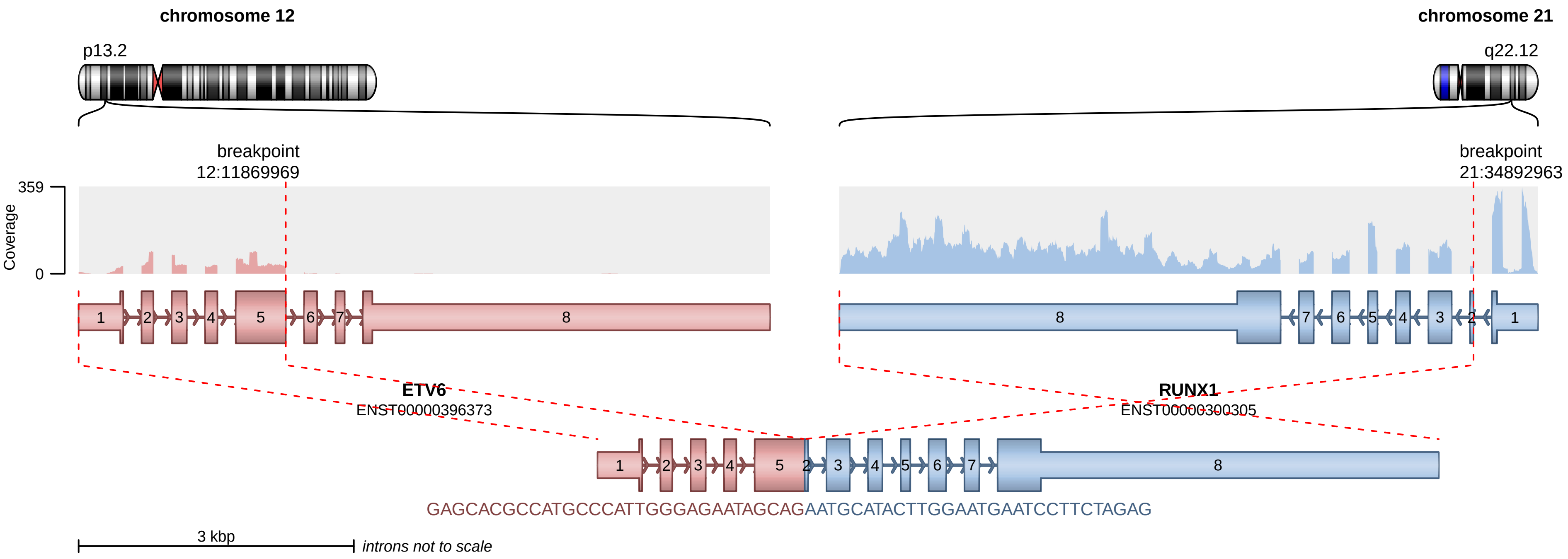

ETV6
RUNX1
1
2
3
4
5
6
7
8
9
10
11
12
13
14
15
16
17
18
19
20
21
22
X
Y
translocation
deletion
duplication
inversion

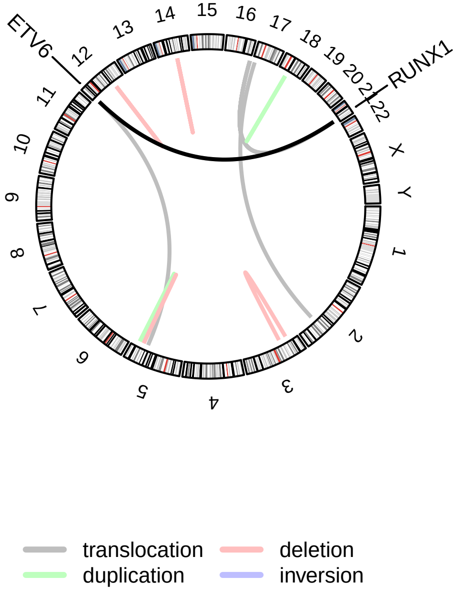
RETAINED PROTEIN DOMAINS
in−frame fusion


Sterile alpha motif (SAM)/Pointed domain
ETV6
RUNX1
Runt domain
Runx inhibition domain

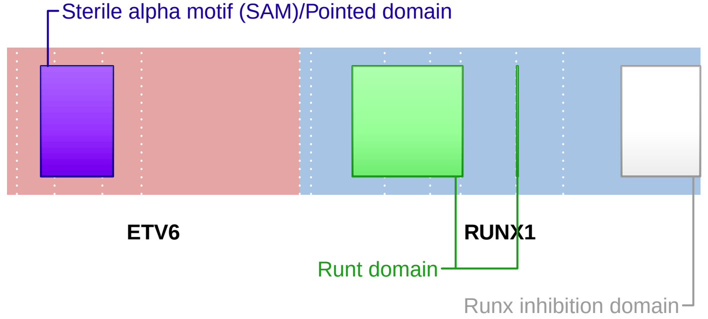
SUPPORTING READ COUNT

Split reads = 3
Discordant mates = 0


chromosome 12
p13.2
breakpoint
12:11869969
359
Coverage
0
1 2 3 4 5 6 7 8
ETV6
ENST00000396373
chromosome 21
q22.12
breakpoint
21:34892963
8 7 6 5 4 3 2 1
RUNX1
ENST00000300305
1 2 3 4 5 2 3 4 5 6 7 8
GAGCACGCCATGCCCATTGGGAGAATAGCAGAATGCATACTTGGAATGAATCCTTCTAGAG
3 kbp
introns not to scale

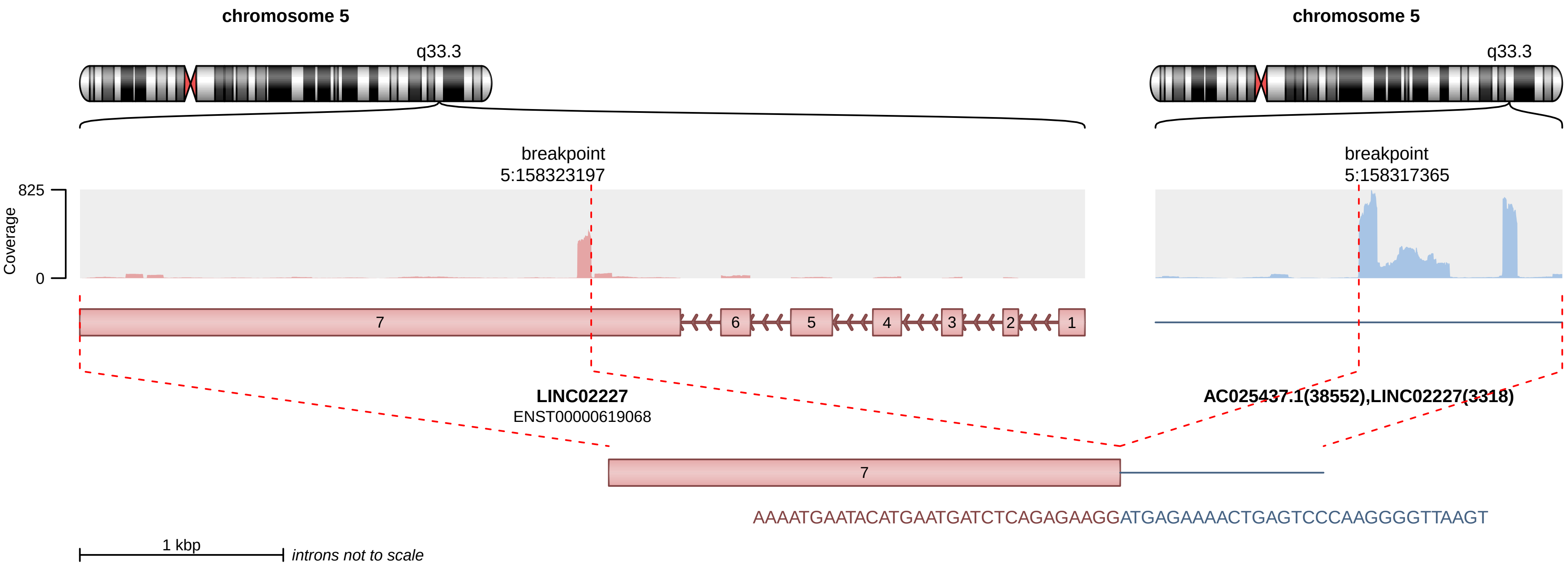


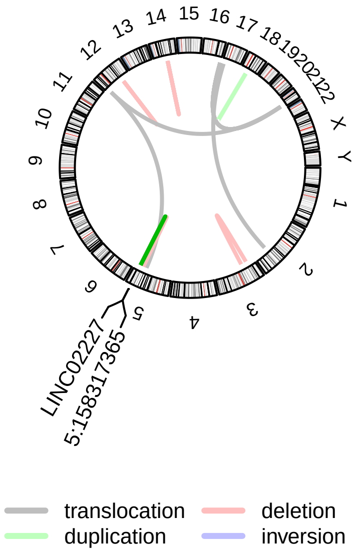
RETAINED PROTEIN DOMAINS
in−frame fusion


Sterile alpha motif (SAM)/Pointed domain
ETV6
RUNX1
Runt domain
Runx inhibition domain

SUPPORTING READ COUNT

Split reads = 1
Discordant mates = 0

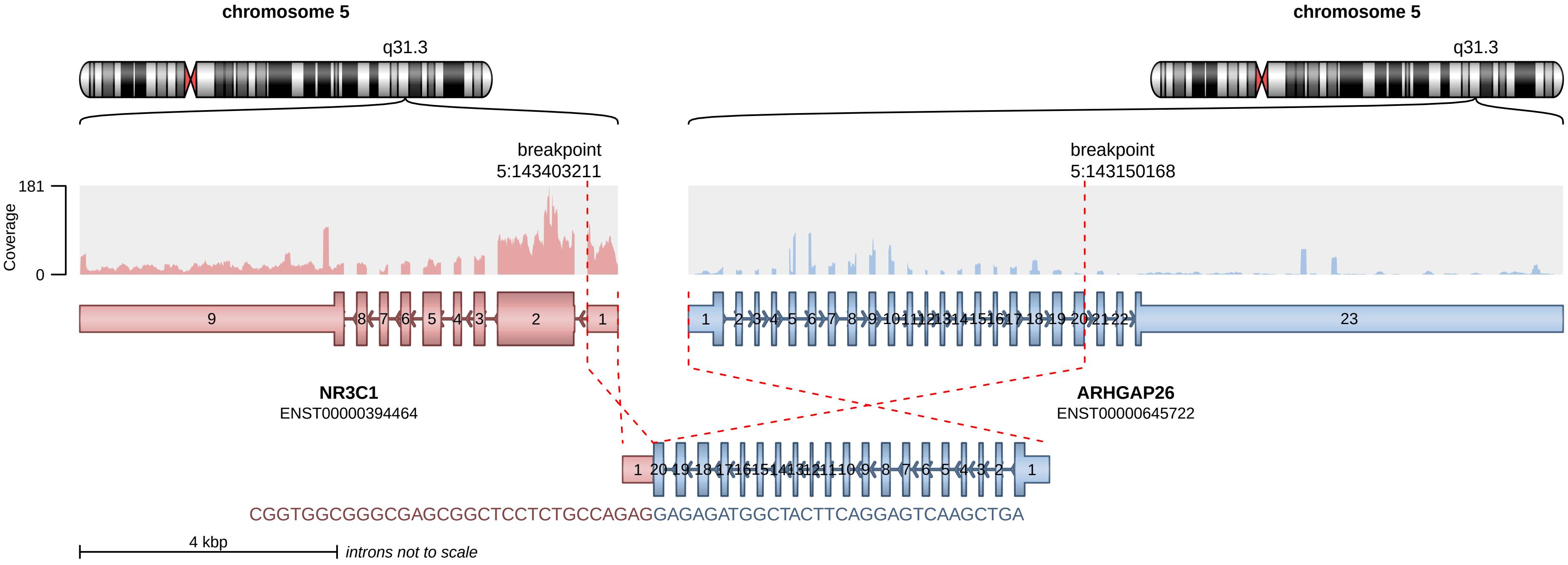

chromosome 5
q33.3
chromosome 5
q33.3
breakpoint
5:158323197
breakpoint
5:158317365
Coverage
825
0
7
6
5
4
3
2
1
LINC02227
ENST00000619068
AC025437.1(38552),LINC02227(3318)
7
AAAATGAATACATGAATGATCTCAGAGAAGGATGAGAAAACTGAGTCCCAAGGGGTTAAGT
1 kbp
introns not to scale

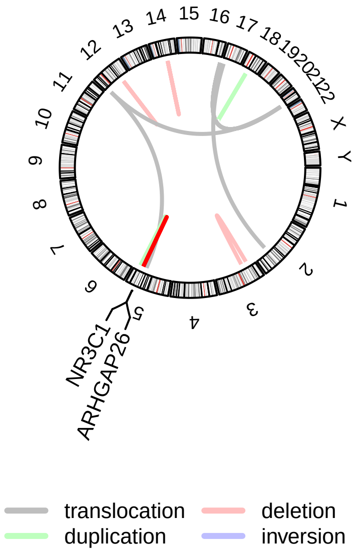


Genes are not protein−coding.

SUPPORTING READ COUNT

Split reads = 50
Discordant mates = 2

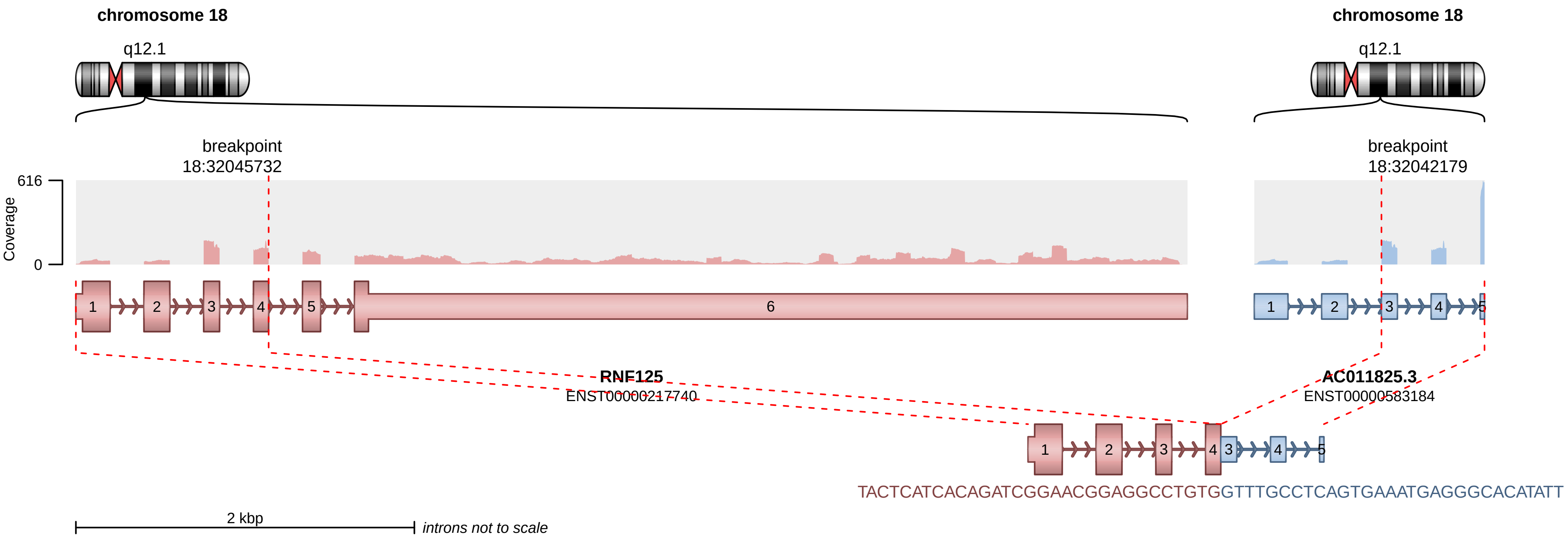

chromosome 5
q31.3
chromosome 5
q31.3
breakpoint
5:143403211
breakpoint
5:143150168
181
Coverage
0
9
8
7
6
5
4
3
2
1
1
2
3
4
5
6
7
8
9
10
11
12
13
14
15
16
17
18
19
20
21
22
23
NR3C1
ENST00000394464
ARHGAP26
ENST00000645722
CGGTGGCGGGCGAGCGGCTCCTCTGCCAGAGGAGAGATGGCTACTTCAGGAGTCAAGCTGA
4 kbp
introns not to scale

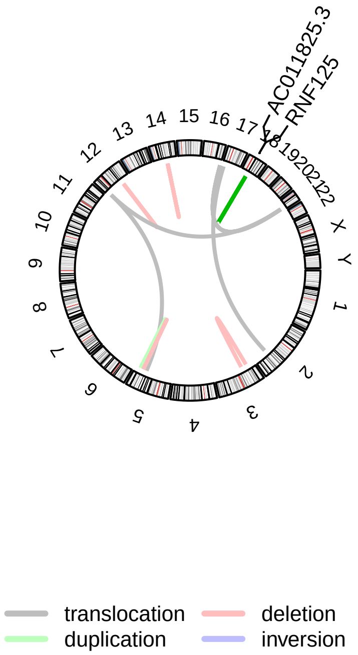

13
14
15
16
17
18
19
20
21
22
X
Y
1
2
3
4
5
6
7
8
9
10
11
12
NR3C1
ARHGAP26
translocation
duplication
deletion
inversion

No coding regions due to antisense transcription.

SUPPORTING READ COUNT

Split reads = 8
Discordant mates = 2

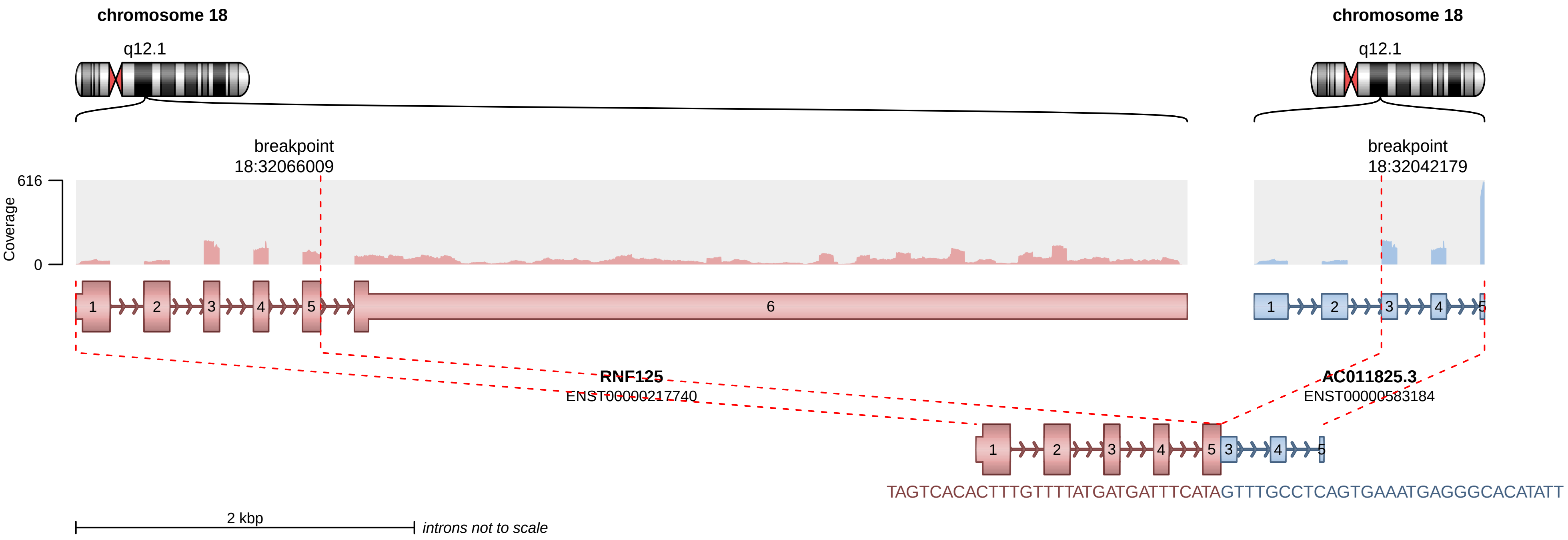

chromosome 18
q12.1
chromosome 18
q12.1
breakpoint
18:32045732
breakpoint
18:32042179
616
Coverage
0
1
2
3
4
5
6
1
2
3
4
5
RNF125
ENST00000217740
AC011825.3
ENST00000583184
1
2
3
4
3
4
5
TACTCATCACAGATCGGAACGGAGGCCTGTGGTTTGCCTCAGTGAAATGAGGGCACATATT
2 kbp
introns not to scale

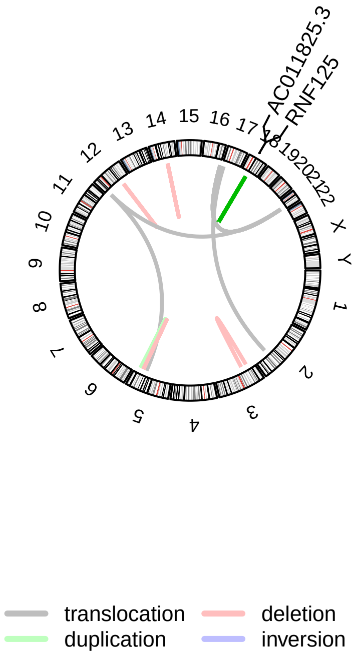


No protein domains retained in fusion.

SUPPORTING READ COUNT

Split reads = 10
Discordant mates = 0

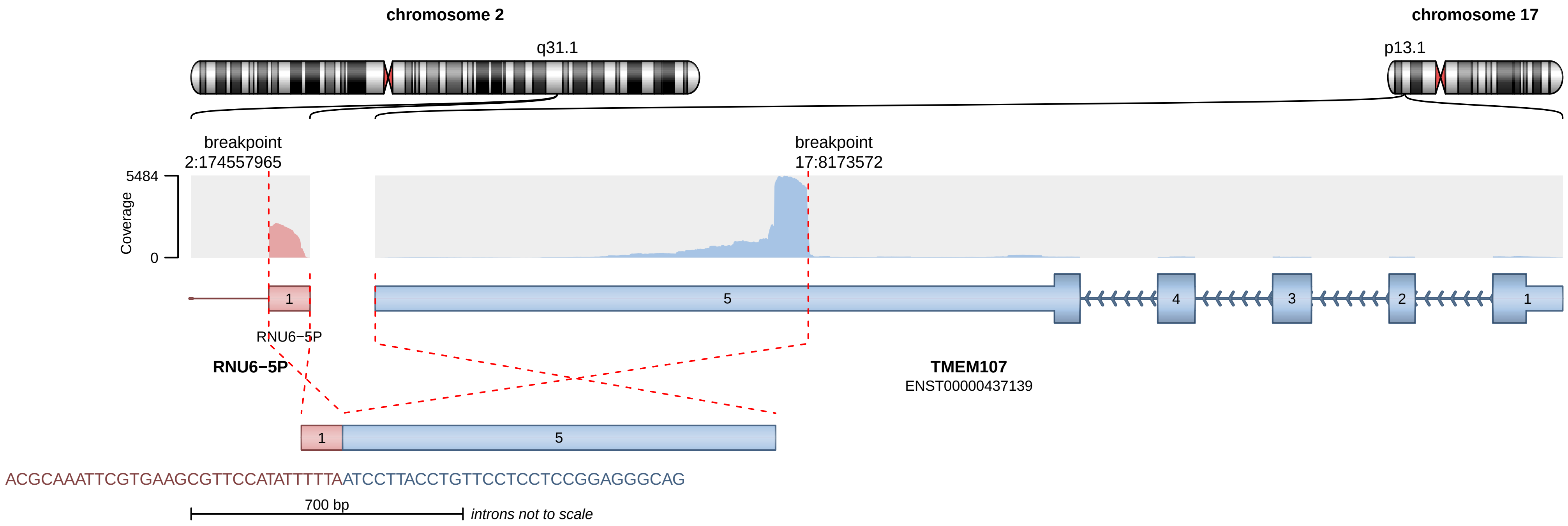

chromosome 18
q12.1
chromosome 18
q12.1
breakpoint
18:32066009
breakpoint
18:32042179
Coverage
616
0
1
2
3
4
5
6
1
2
3
4
5
RNF125
ENST00000217740
AC011825.3
ENST00000583184
1
2
3
4
5
3
4
5
TAGTCACACTTTGTTTTATGATGATTTCATAGTTTGCCTCAGTGAAATGAGGGCACATATT
2 kbp
introns not to scale

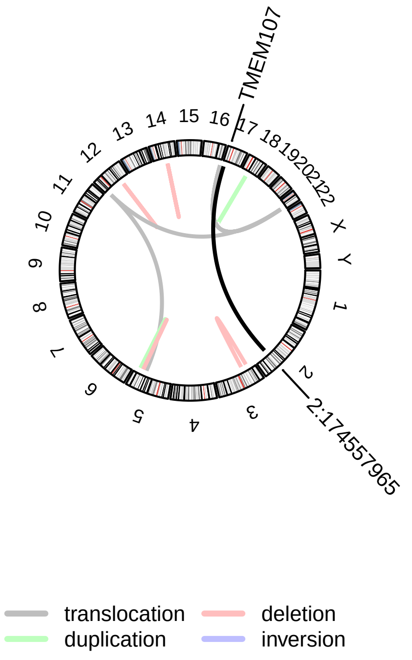


No protein domains retained in fusion.

SUPPORTING READ COUNT

Split reads = 7
Discordant mates = 1

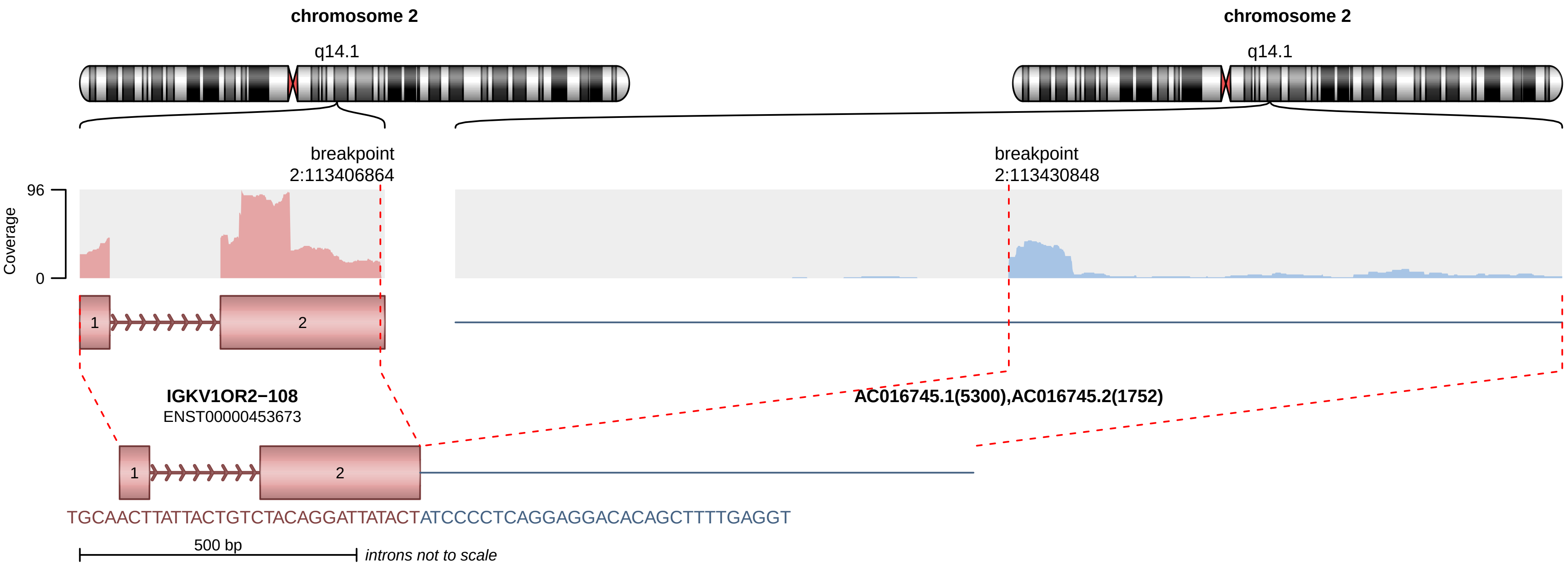

chromosome 2
q31.1
chromosome 17
p13.1
breakpoint
2:174557965
breakpoint
17:8173572
5484
Coverage
0
1
5
4
3
2
1
RNU6−5P
RNU6−5P
TMEM107
ENST00000437139
1
5
ACGCAAATTCGTGAAGCGTTCCATATTTTTAATCCTTACCTGTTCCTCCTCCGGAGGGCAG
700 bp
introns not to scale

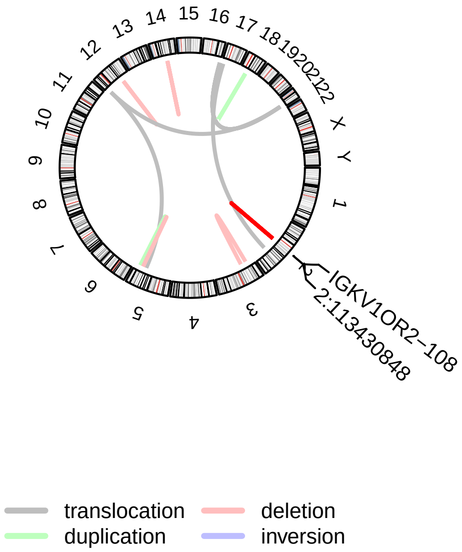


No coding regions retained in fusion transcript.

SUPPORTING READ COUNT

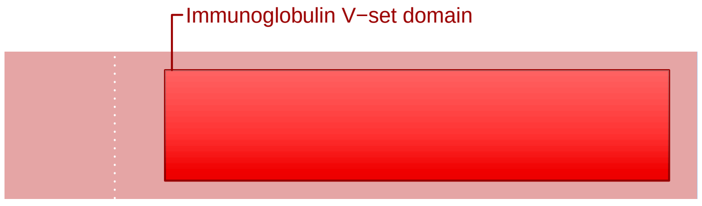
Split reads = 3
Discordant mates = 0


chromosome 2
q14.1
chromosome 2
q14.1
breakpoint
2:113406864
breakpoint
2:113430848
Coverage
96
0
1
2
IGKV1OR2−108
ENST00000453673
AC016745.1(5300),AC016745.2(1752)
1
2
TGCAACTTATTACTGTCTACAGGATTATACTATCCCCTCAGGAGGACACAGCTTTTGAGGT
500 bp
introns not to scale


1
3
4
5
6
7
8
9
10
11
12
13
14
15
16
17
18
19
20
21
22
X
Y
IGKV1OR2−108
2:113430848
translocation
duplication
deletion
inversion

RETAINED PROTEIN DOMAINS

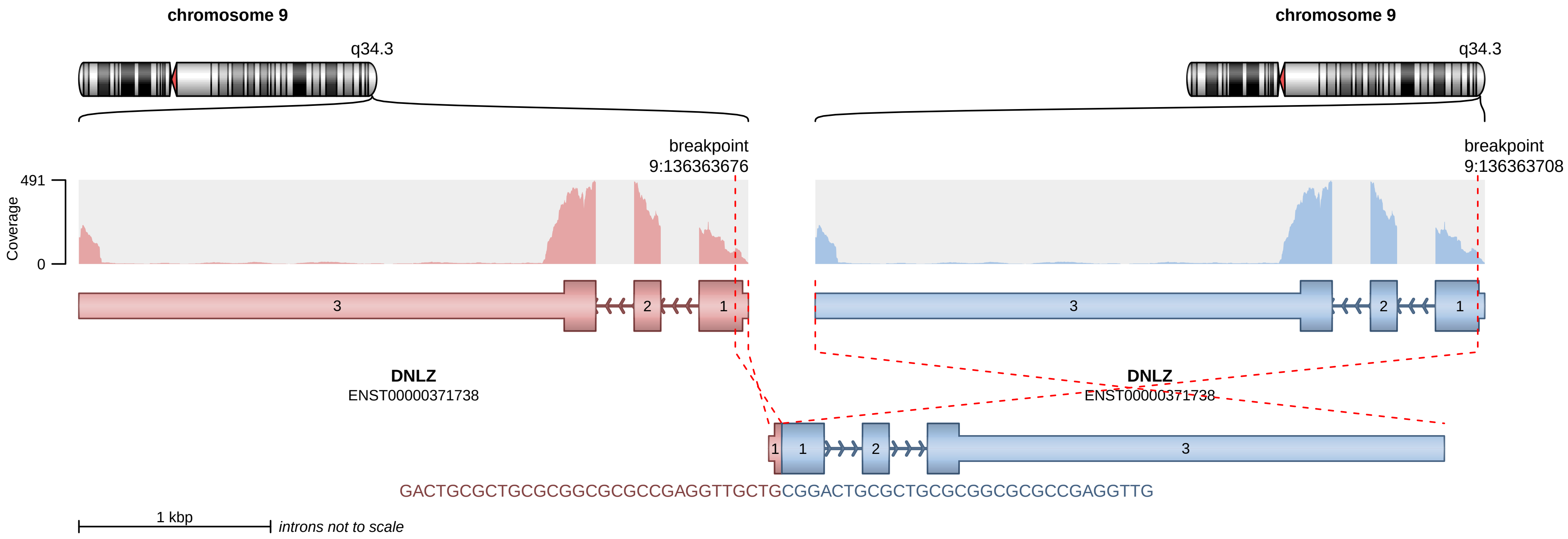
out−of−frame fusion

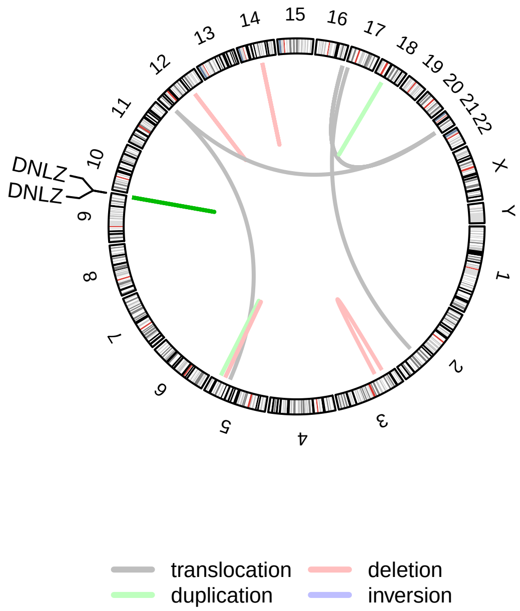

Immunoglobulin V−set domain

IGKV1OR2−108

SUPPORTING READ COUNT

Split reads = 8
Discordant mates = 10

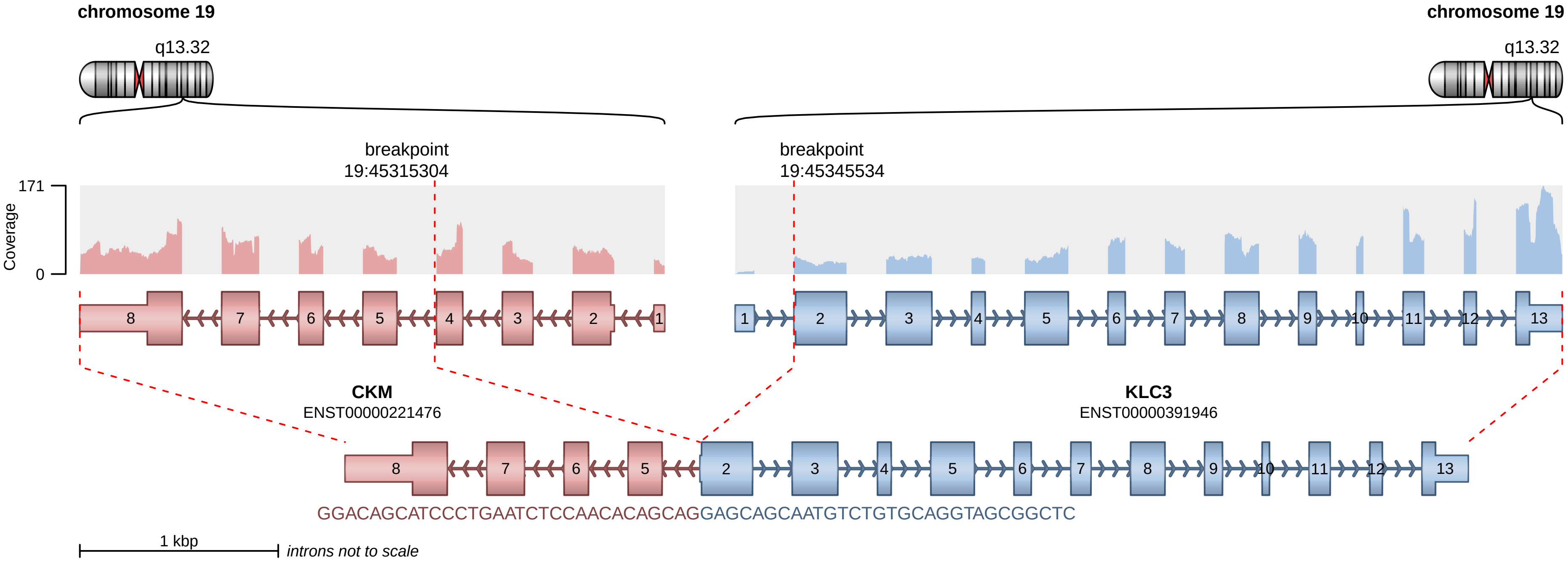

chromosome 9
q34.3
chromosome 9
q34.3
breakpoint
9:136363676
breakpoint
9:136363708
Coverage
491
0
3
2
1
3
2
1
DNLZ
ENST00000371738
DNLZ
ENST00000371738
1
1
2
3
GACTGCGCTGCGCGGCGCGCCGAGGTTGCTGCGGACTGCGCTGCGCGGCGCGCCGAGGTTG
1 kbp
introns not to scale

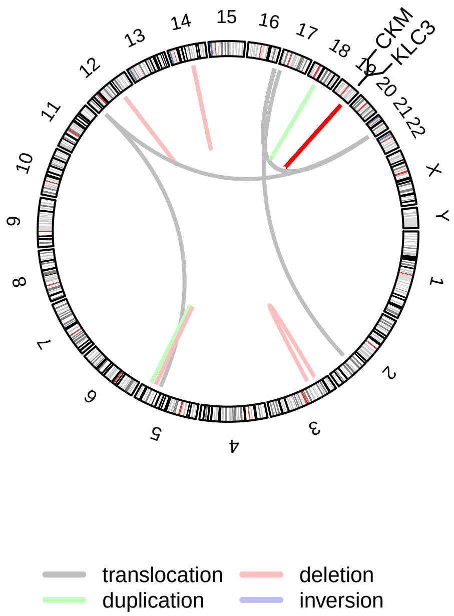

DNLZ
DNLZ
1
2
3
4
5
6
7
8
9
10
11
12
13
14
15
16
17
18
19
20
21
22
X
Y
translocation
duplication
deletion
inversion

No protein domains retained in fusion.

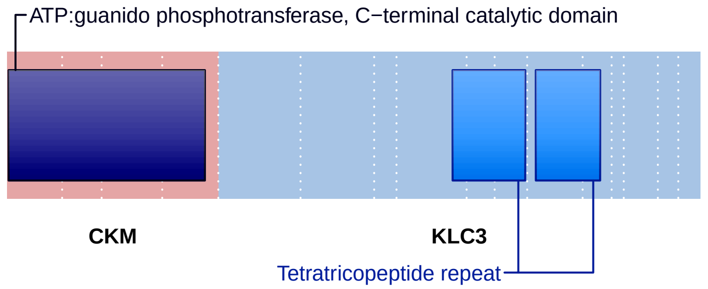
SUPPORTING READ COUNT

Split reads = 13
Discordant mates = 0


chromosome 19
q13.32
chromosome 19
q13.32
breakpoint
19:45315304
breakpoint
19:45345534
Coverage
171
0
8
7
6
5
4
3
2
1
1
2
3
4
5
6
7
8
9
10
11
12
13
CKM
ENST00000221476
KLC3
ENST00000391946
8
7
6
5
2
3
4
5
6
7
8
9
10
11
12
13
GGACAGCATCCCTGAATCTCCAACACAGCAGGAGCAGCAATGTCTGTGCAGGTAGCGGCTC
1 kbp
introns not to scale

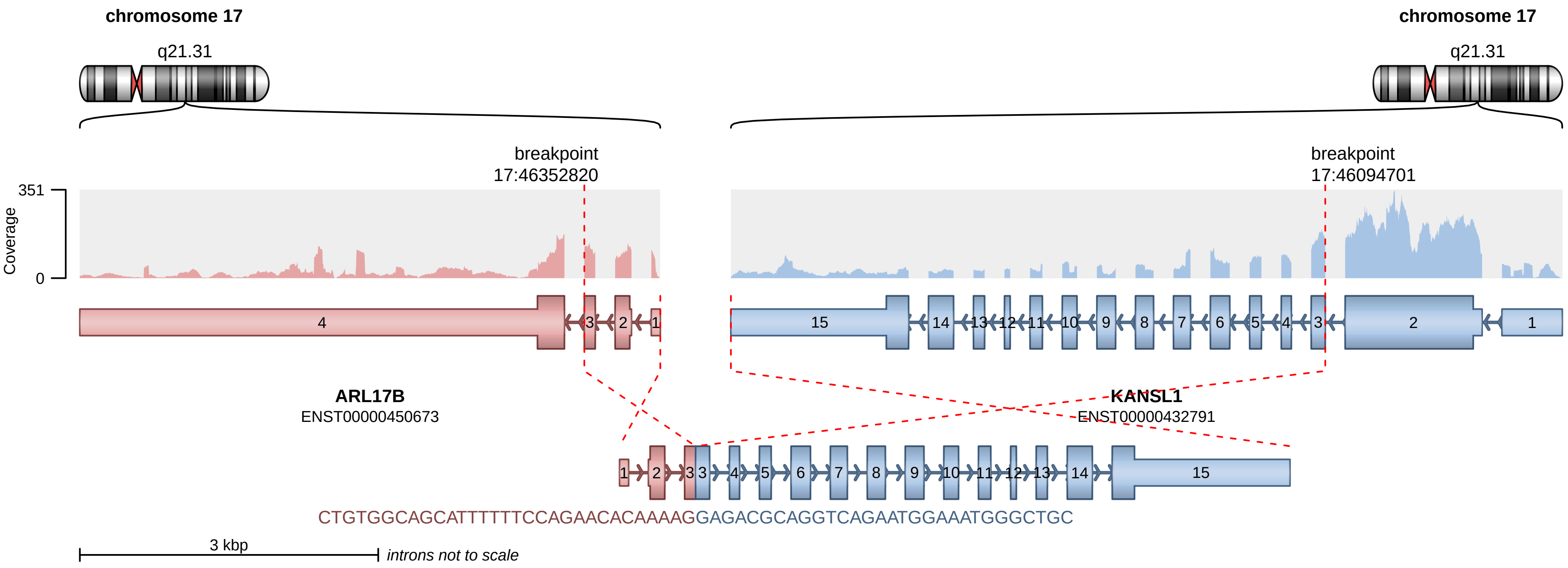


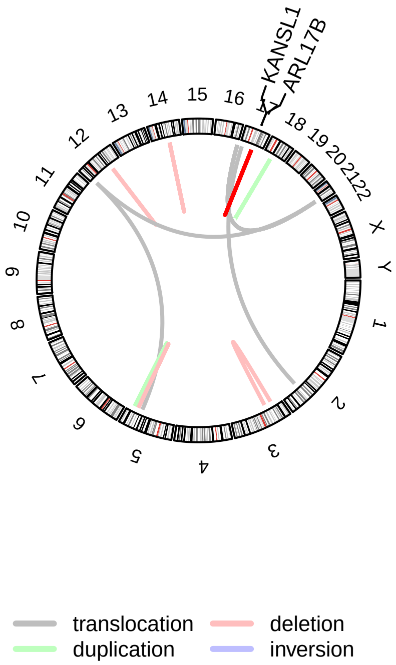
RETAINED PROTEIN DOMAINS
reading frame unclear

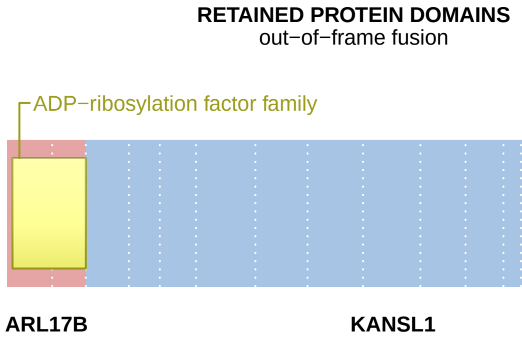

ATP:guanido phosphotransferase, C−terminal catalytic domain
CKM
KLC3
Tetratricopeptide repeat

SUPPORTING READ COUNT

Split reads = 8
Discordant mates = 1

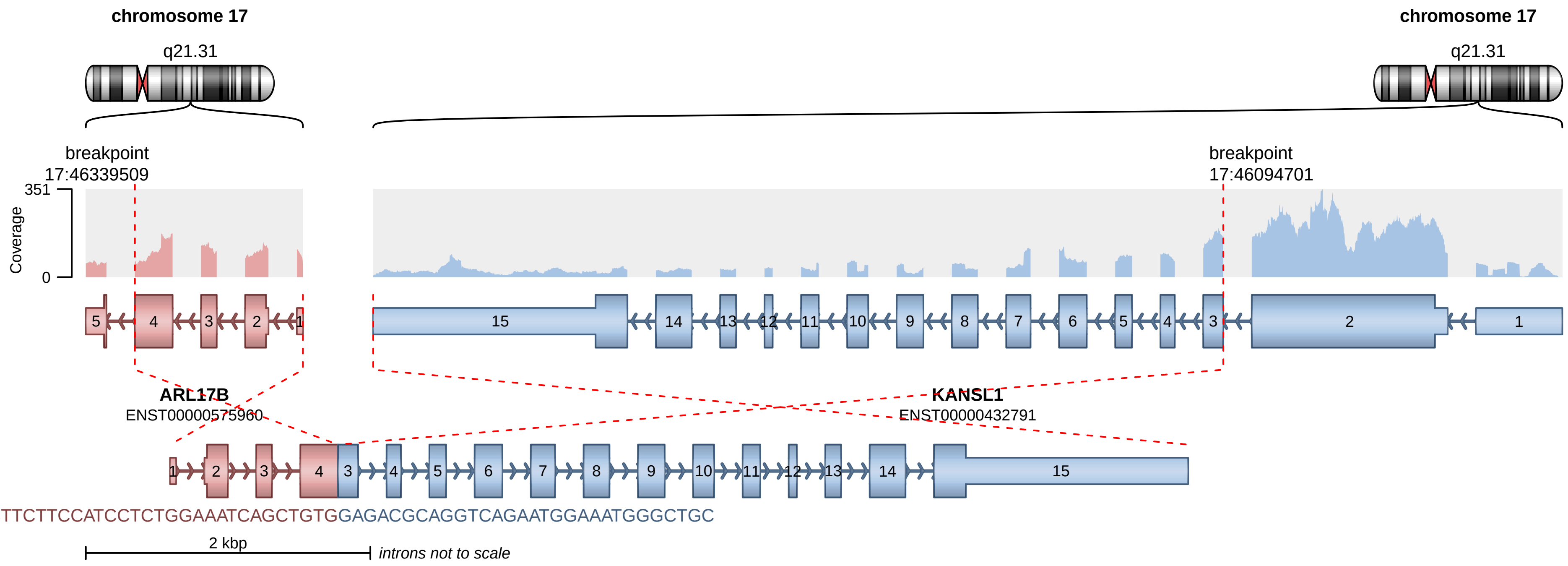

chromosome 17
q21.31
chromosome 17
q21.31
breakpoint
17:46352820
breakpoint
17:46094701
351
Coverage
0
4
3
2
1
15
14
13
12
11
10
9
8
7
6
5
4
3
2
1
ARL17B
ENST00000450673
KANSL1
ENST00000432791
1
2
3
3
4
5
6
7
8
9
10
11
12
13
14
15
CTGTGGCAGCATTTTTTCCAGAACACAAAAGGAGACGCAGGTCAGAATGGAAATGGGCTGC
3 kbp
introns not to scale

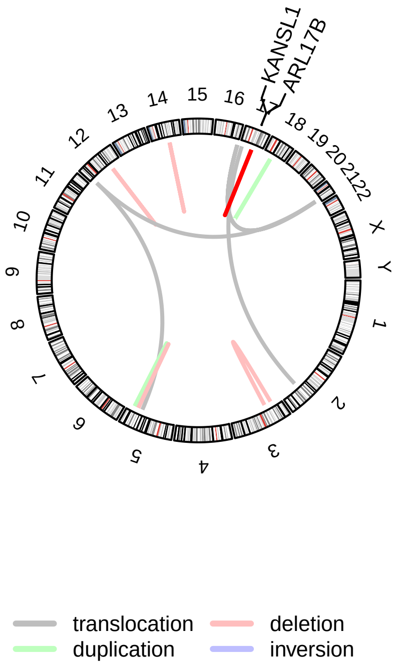



RETAINED PROTEIN DOMAINS
out−of−frame fusion
ADP−ribosylation factor family
ARL17B
KANSL1

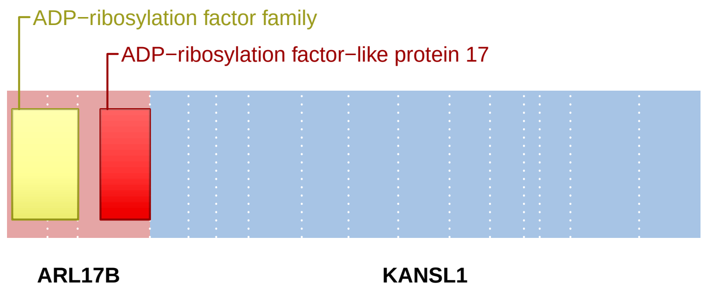
SUPPORTING READ COUNT

Split reads = 7
Discordant mates = 0


chromosome 17
q21.31
chromosome 17
q21.31
breakpoint
17:46339509
breakpoint
17:46094701
Coverage
351
0
5
4
3
2
1
15
14
13
12
11
10
9
8
7
6
5
4
3
2
1
ARL17B
ENST00000575960
KANSL1
ENST00000432791
1
2
3
4
3
4
5
6
7
8
9
10
11
12
13
14
15
GACTTCTTCCATCCTCTGGAAATCAGCTGTGGAGACGCAGGTCAGAATGGAAATGGGCTGC
2 kbp
introns not to scale

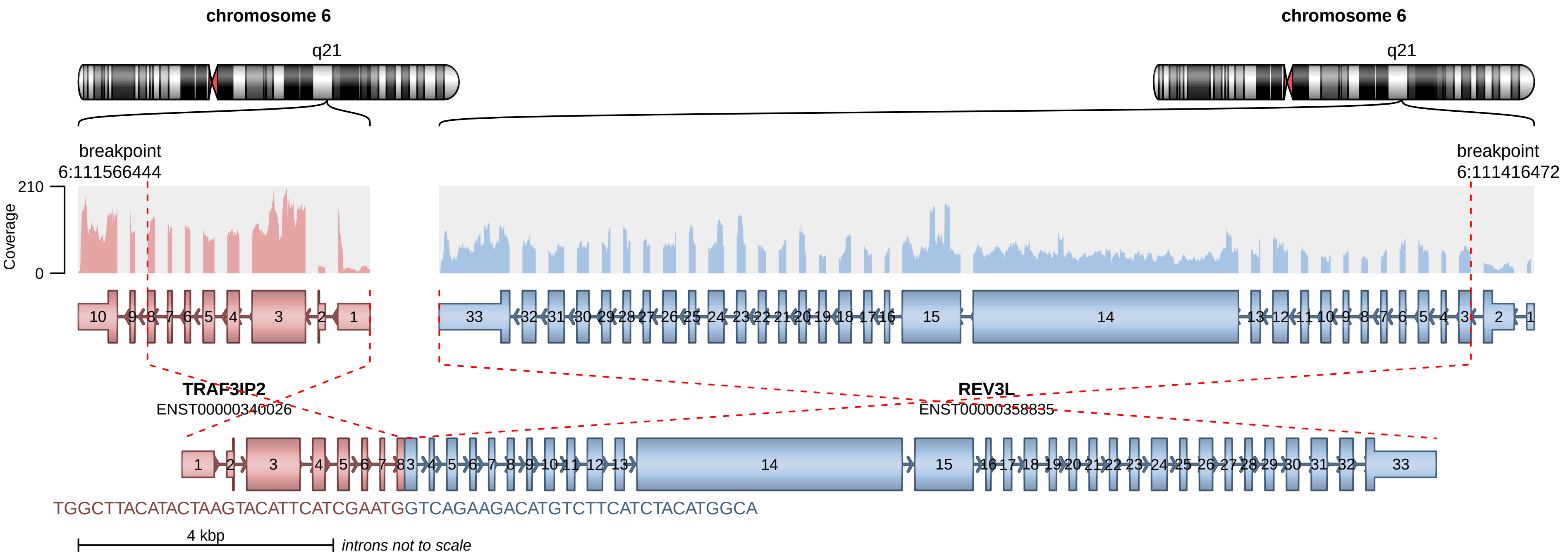


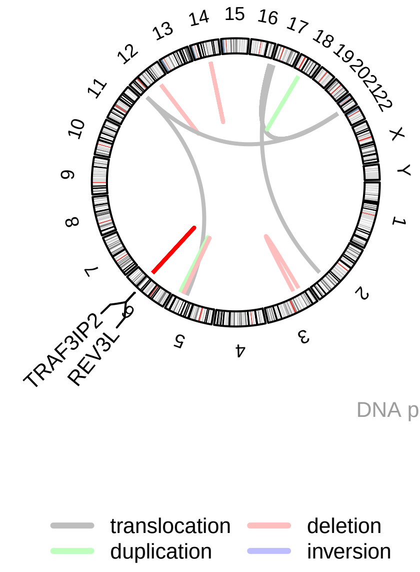
RETAINED PROTEIN DOMAINS
reading frame unclear

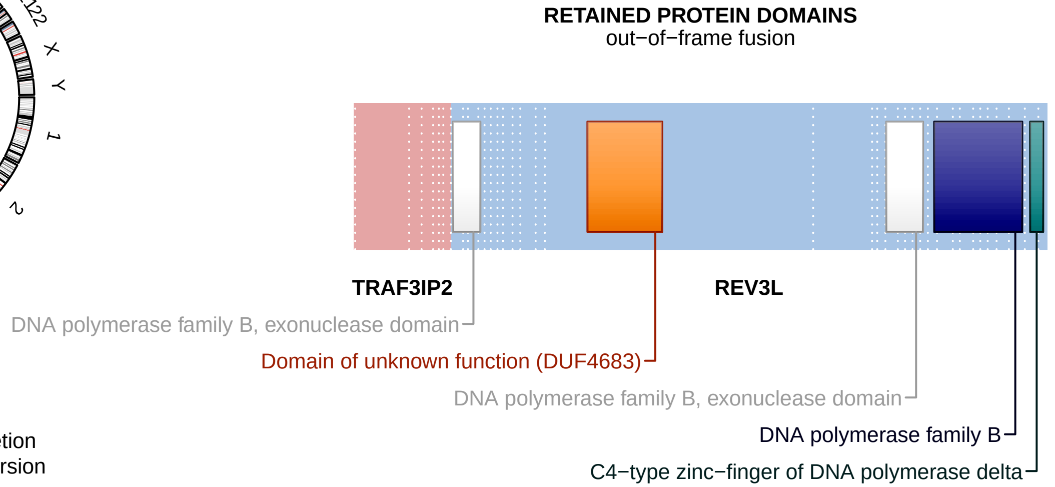

ADP−ribosylation factor family
ADP−ribosylation factor−like protein 17
ARL17B
KANSL1

SUPPORTING READ COUNT

Split reads = 1
Discordant mates = 0

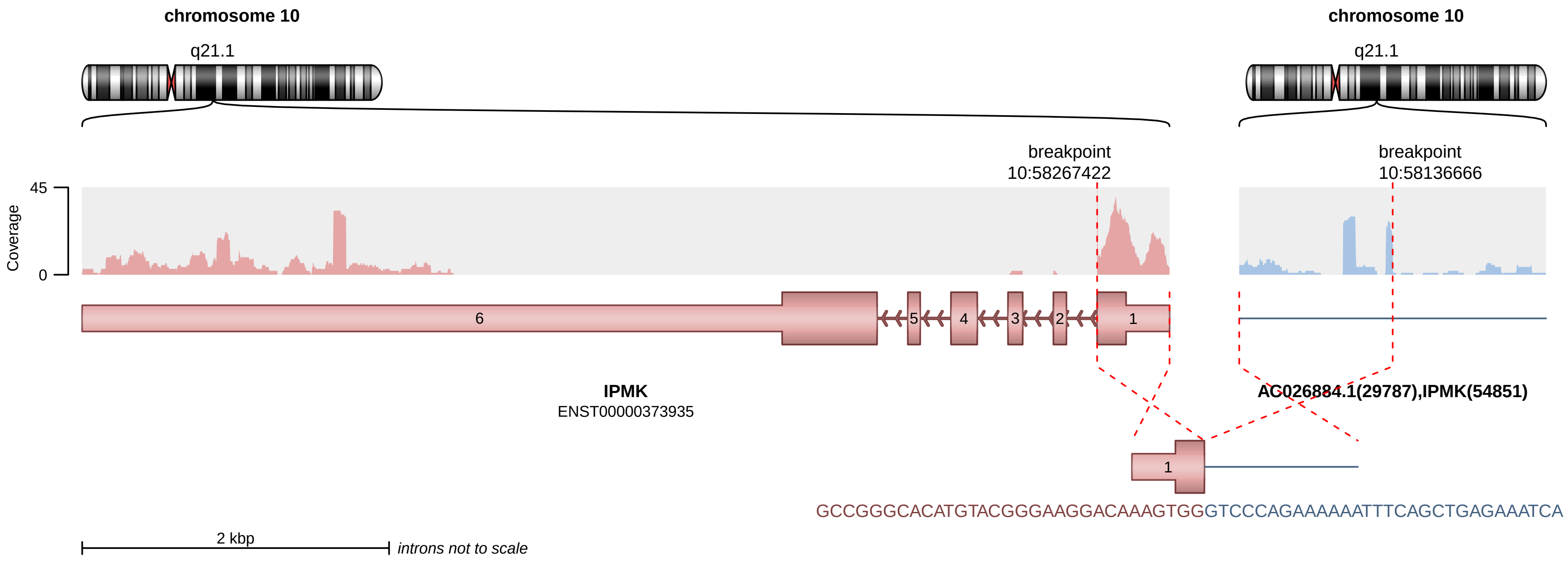

chromosome 6
q21
chromosome 6
q21
breakpoint
6:111566444
breakpoint
6:111416472
210
Coverage
0
10 9 8 7 6 5 4 3 2 1
33 32 31 30 29 28 27 26 25 24 23 22 21 20 19 18 17 16 15 14 13 12 11 10 9 8 7 6 5 4 3 2 1
TRAF3IP2
ENST00000340026
REV3L
ENST00000358835
1 2 3 4 5 6 7 8 3 4 5 6 7 8 9 10 11 12 13 14 15 16 17 18 19 20 21 22 23 24 25 26 27 28 29 30 31 32 33
TGGCTTACATACTAAGTACATTCATCGAATGGTCAGAAGACATGTCTTCATCTACATGGCA
4 kbp
introns not to scale

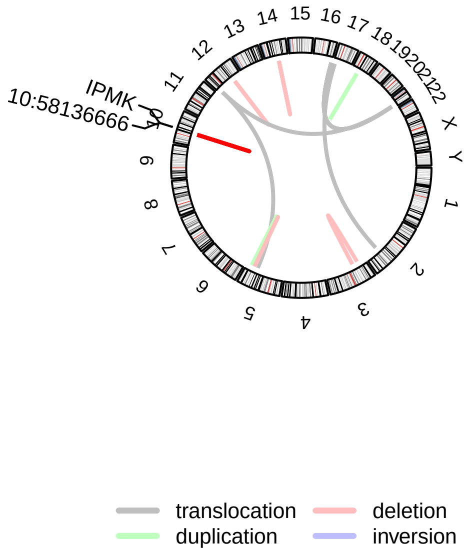

1 2 3 4 5 6 7 8 9 10 11 12 13 14 15 16 17 18 19 20 21 22 X Y
TRAF3IP2
REV3L
translocation
duplication
deletion
inversion


RETAINED PROTEIN DOMAINS
out−of−frame fusion
TRAF3IP2
REV3L
DNA polymerase family B, exonuclease domain
Domain of unknown function (DUF4683)
DNA polymerase family B, exonuclease domain
DNA polymerase family B
C4−type zinc−finger of DNA polymerase delta

SUPPORTING READ COUNT

Split reads = 5
Discordant mates = 1

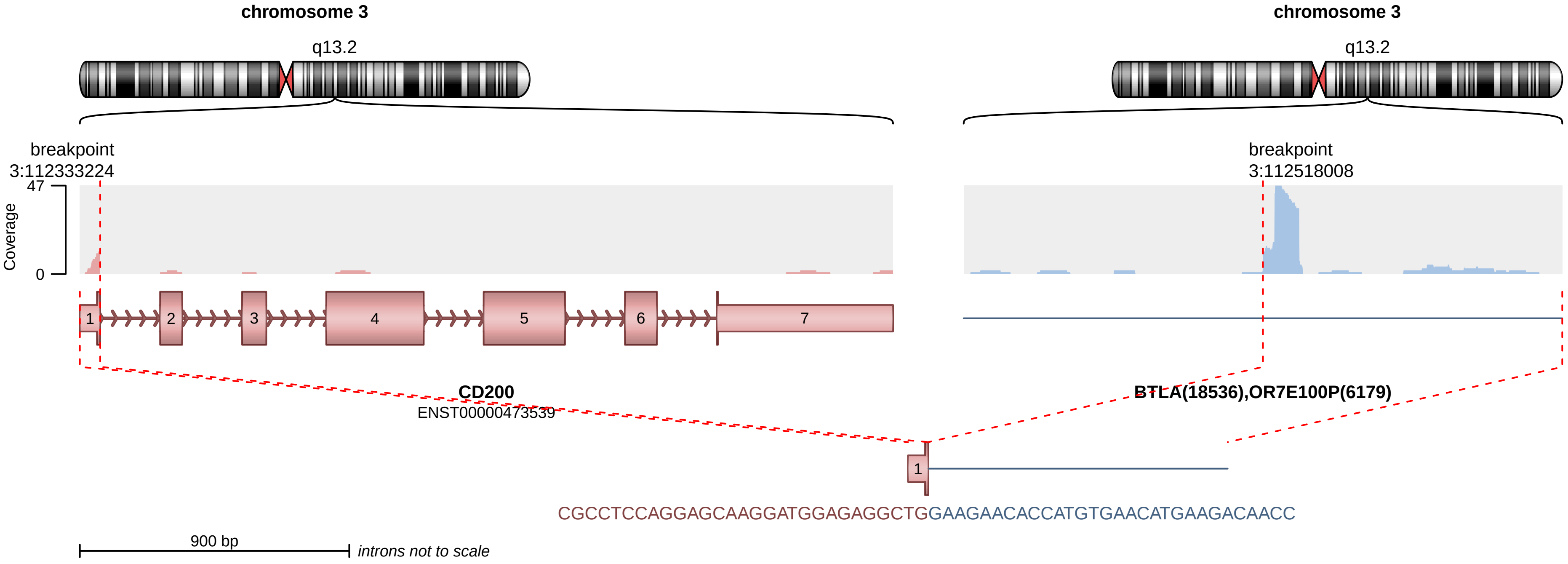

chromosome 10
q21.1
chromosome 10
q21.1
breakpoint
10:58267422
breakpoint
10:58136666
Coverage
45
0
6
5
4
3
2
1
IPMK
ENST00000373935
AC026884.1(29787),IPMK(54851)
1
GCCGGGCACATGTACGGGAAGGACAAAGTGGGTCCCAGAAAAAATTTCAGCTGAGAAATCA
2 kbp
introns not to scale

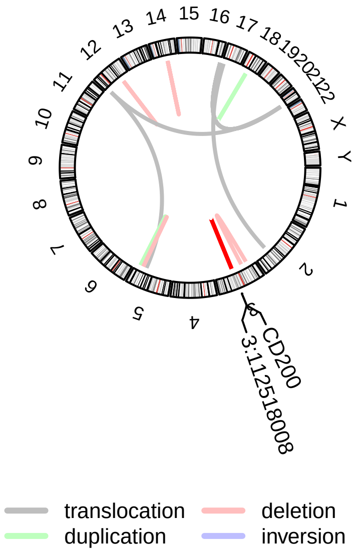

IPMK
10:58136666
1
2
3
4
5
6
7
8
9
10
11
12
13
14
15
16
17
18
19
20
21
22
X
Y
translocation
duplication
deletion
inversion

No protein domains retained in fusion.

SUPPORTING READ COUNT

Split reads = 1
Discordant mates = 3

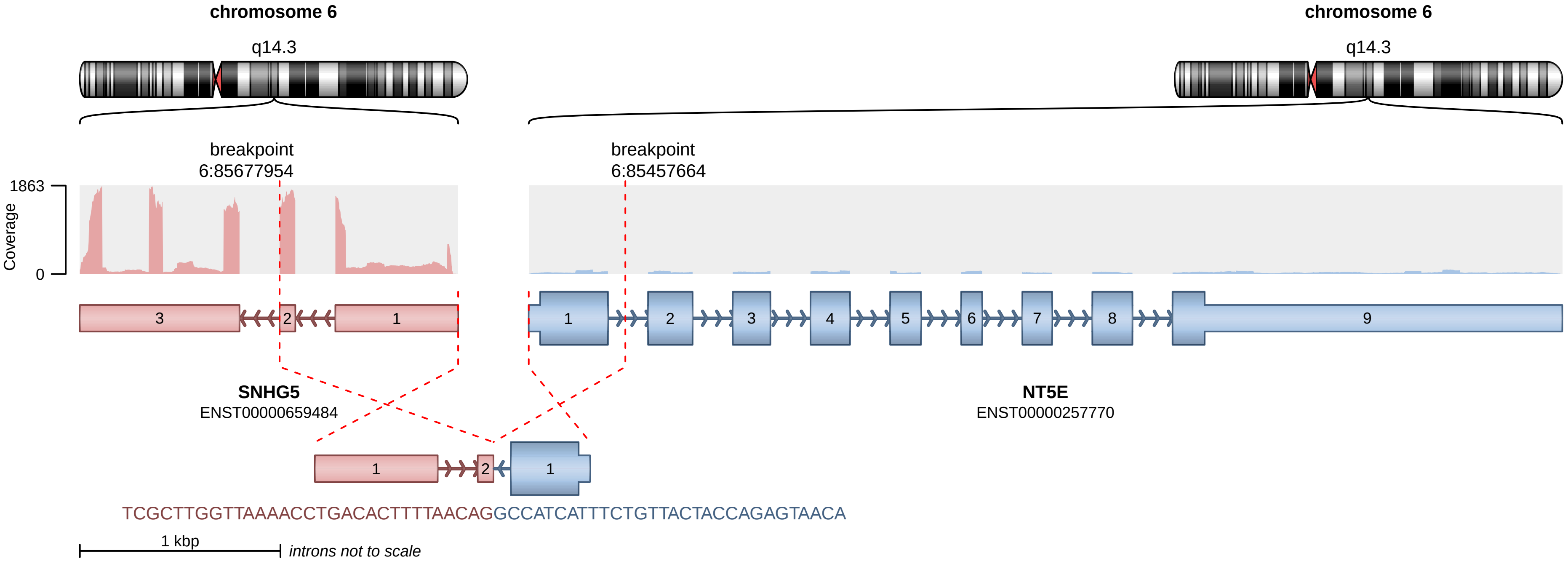

chromosome 3
q13.2
chromosome 3
q13.2
breakpoint
3:112333224
47
Coverage
0
breakpoint
3:112518008
1
2
3
4
5
6
7
CD200
ENST00000473539
BTLA(18536),OR7E100P(6179)
1
CGCCTCCAGGAGCAAGGATGGAGAGGCTGGAAGAACACCATGTGAACATGAAGACAACC
900 bp
introns not to scale

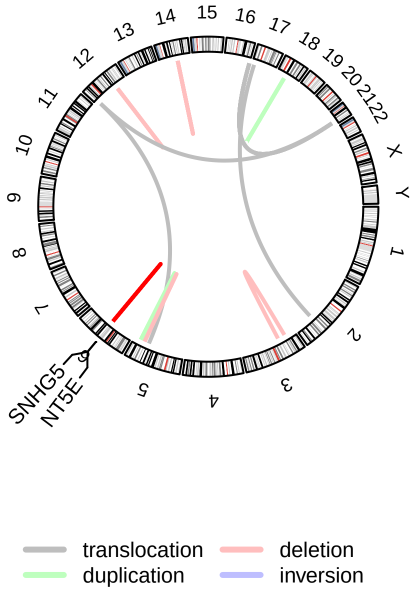


No protein domains retained in fusion.

SUPPORTING READ COUNT

Split reads = 3
Discordant mates = 0

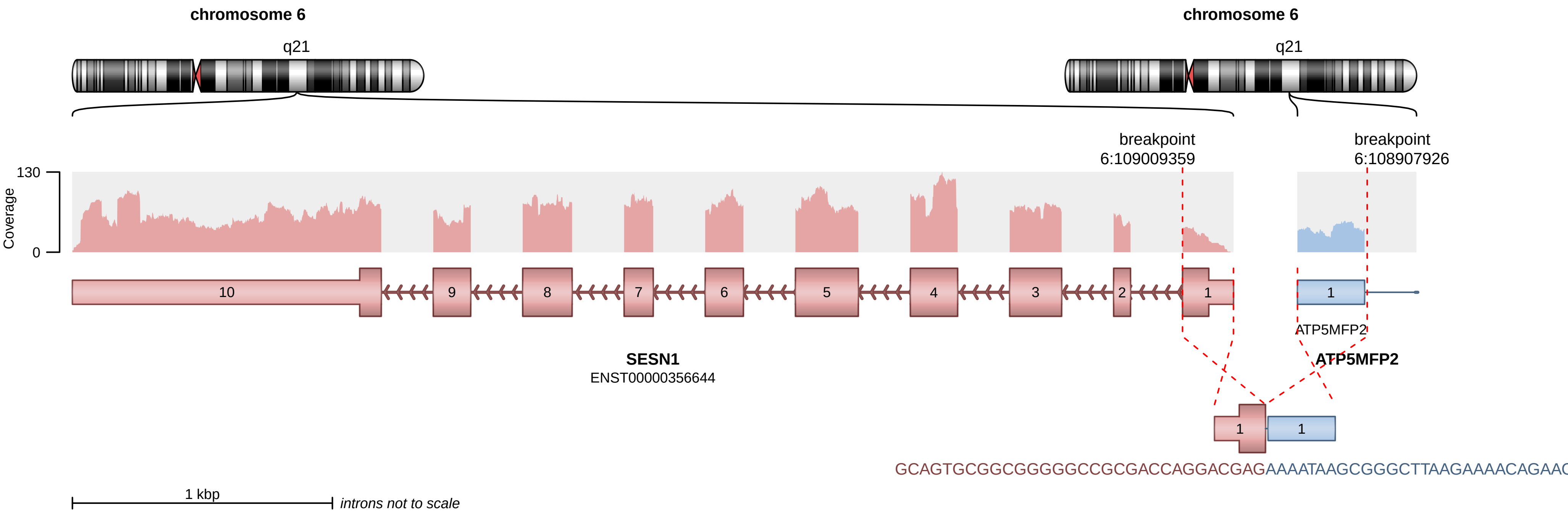

chromosome 6
q14.3
chromosome 6
q14.3
breakpoint
6:85677954
breakpoint
6:85457664
1863
Coverage
0
3
2
1
1
2
3
4
5
6
7
8
9
SNHG5
ENST00000659484
NT5E
ENST00000257770
1
2
1
TCGCTTGGTTAAAACCTGACACTTTTAACAGGCCATCATTTCTGTTACTACCAGAGTAACA
1 kbp
introns not to scale

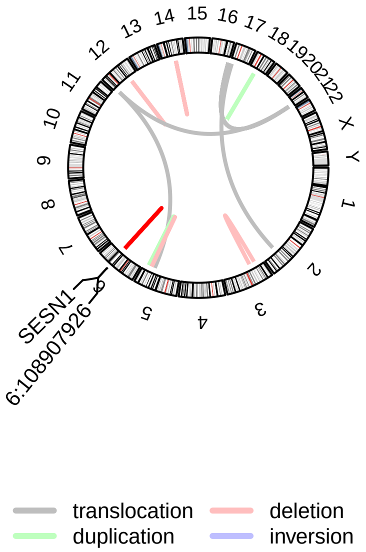

15
16
17
18
19
20
21
22
X
Y
1
2
3
4
5
SNHG5
NT5E
6
7
8
9
10
11
12
13
14
translocation
duplication
deletion
inversion

No coding regions due to antisense transcription.

SUPPORTING READ COUNT

Split reads = 2
Discordant mates = 1

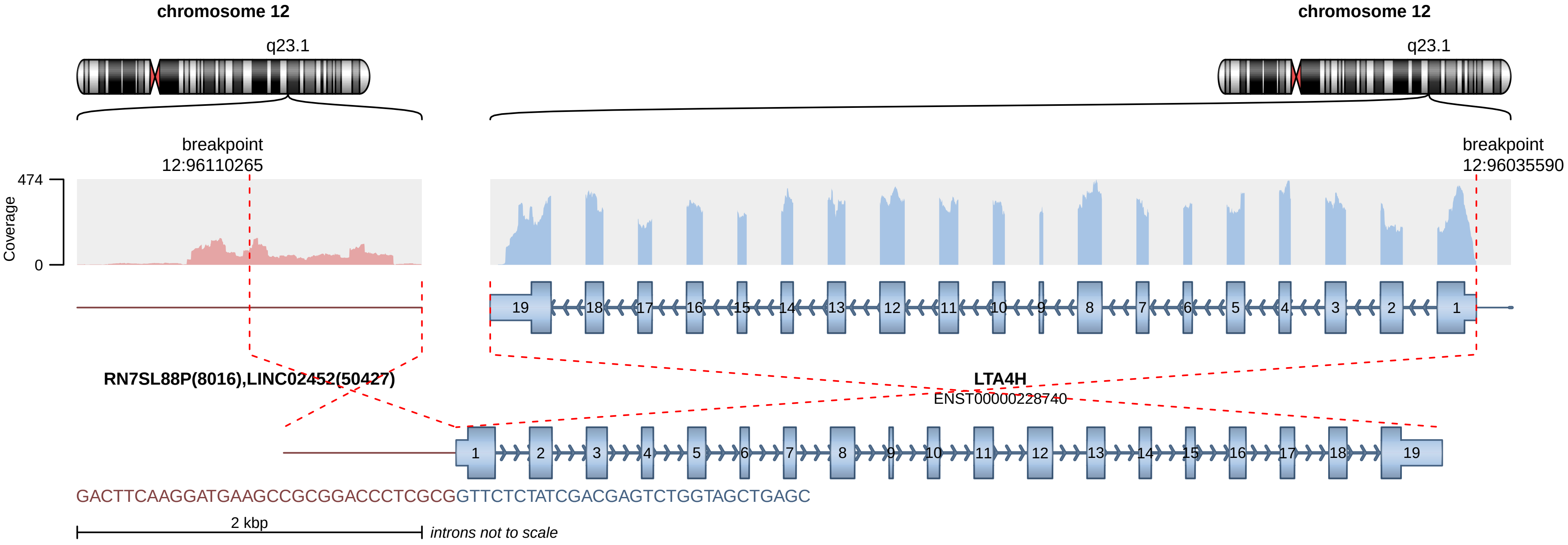

chromosome 6
q21
chromosome 6
q21
breakpoint
6:109009359
breakpoint
6:108907926
Coverage
130
0
10
9
8
7
6
5
4
3
2
1
1
ATP5MFP2
SESN1
ENST00000356644
ATP5MFP2
1
1
GCAGTGCGGCGGGGGCCGCGACCAGGACGAGAAAATAAGCGGGCTTAAGAAAACAGAA
1 kbp
introns not to scale

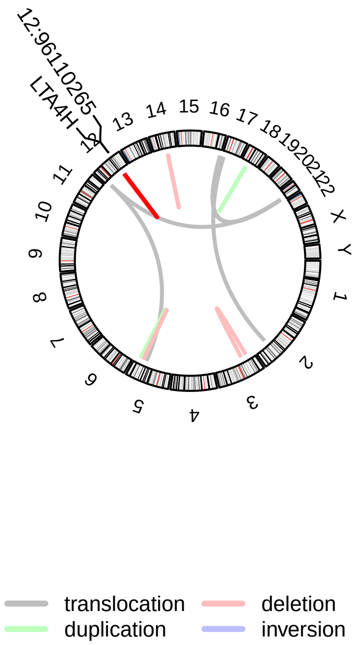


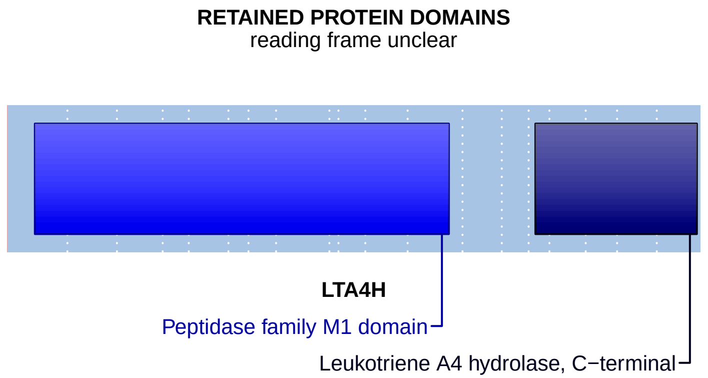
No protein domains retained in fusion.

SUPPORTING READ COUNT

Split reads = 2
Discordant mates = 1

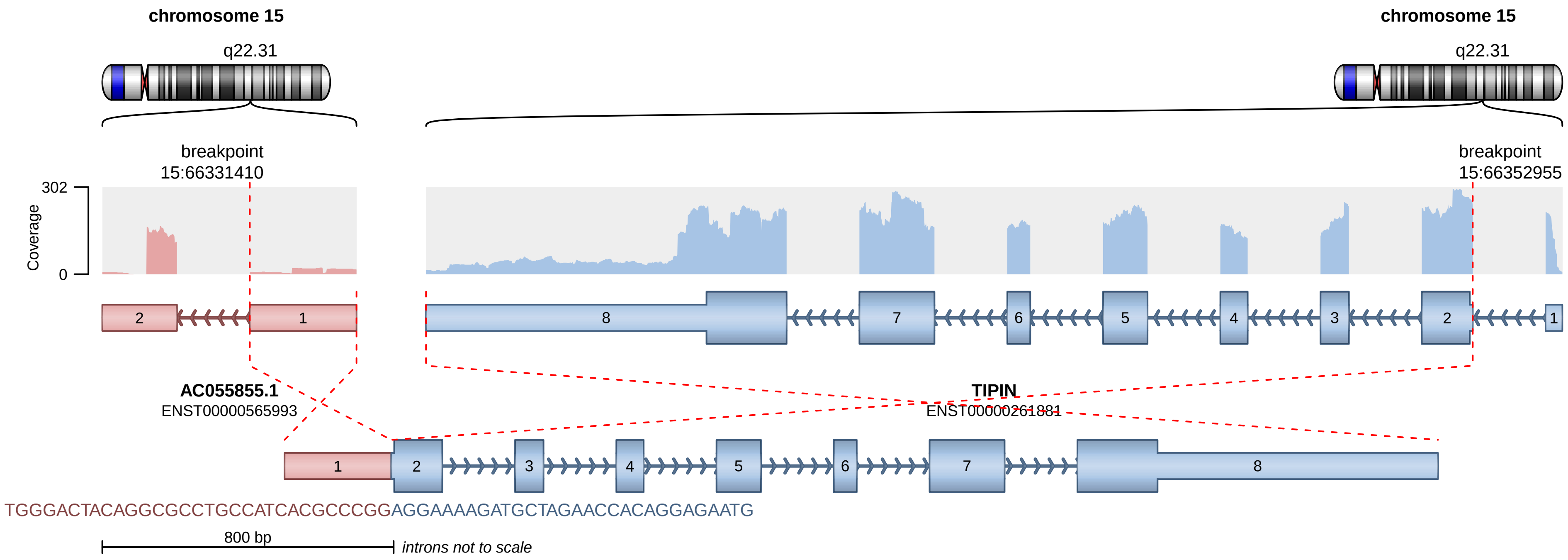

chromosome 12
q23.1
chromosome 12
q23.1
breakpoint
12:96110265
breakpoint
12:96035590
474
Coverage
0
19 18 17 16 15 14 13 12 11 10 9 8 7 6 5 4 3 2 1
RN7SL88P(8016),LINC02452(50427)
LTA4H
ENST00000228740
1 2 3 4 5 6 7 8 9 10 11 12 13 14 15 16 17 18 19
GACTTCAAGGATGAAGCCGCGGACCCTCGCGGTTCTCTATCGACGAGTCTGGTAGCTGAGC
2 kbp
introns not to scale

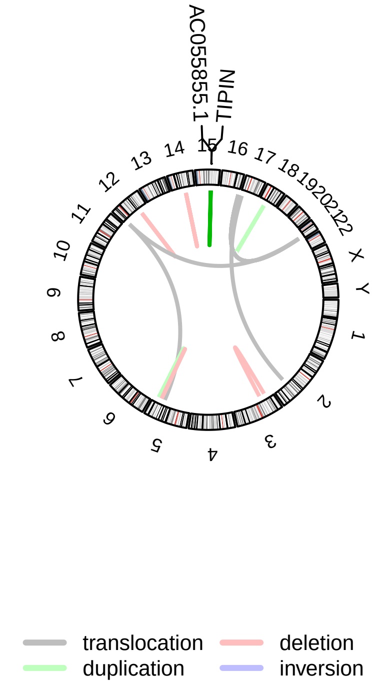

12:96110265
LTA4H
13 14 15 16 17 18 19 20 21 22
X
Y
1
2
3
4
5
6
7
8
9
10
11
12
translocation
deletion
duplication
inversion


RETAINED PROTEIN DOMAINS
reading frame unclear
LTA4H
Peptidase family M1 domain
Leukotriene A4 hydrolase, C−terminal

SUPPORTING READ COUNT

Split reads = 3
Discordant mates = 0

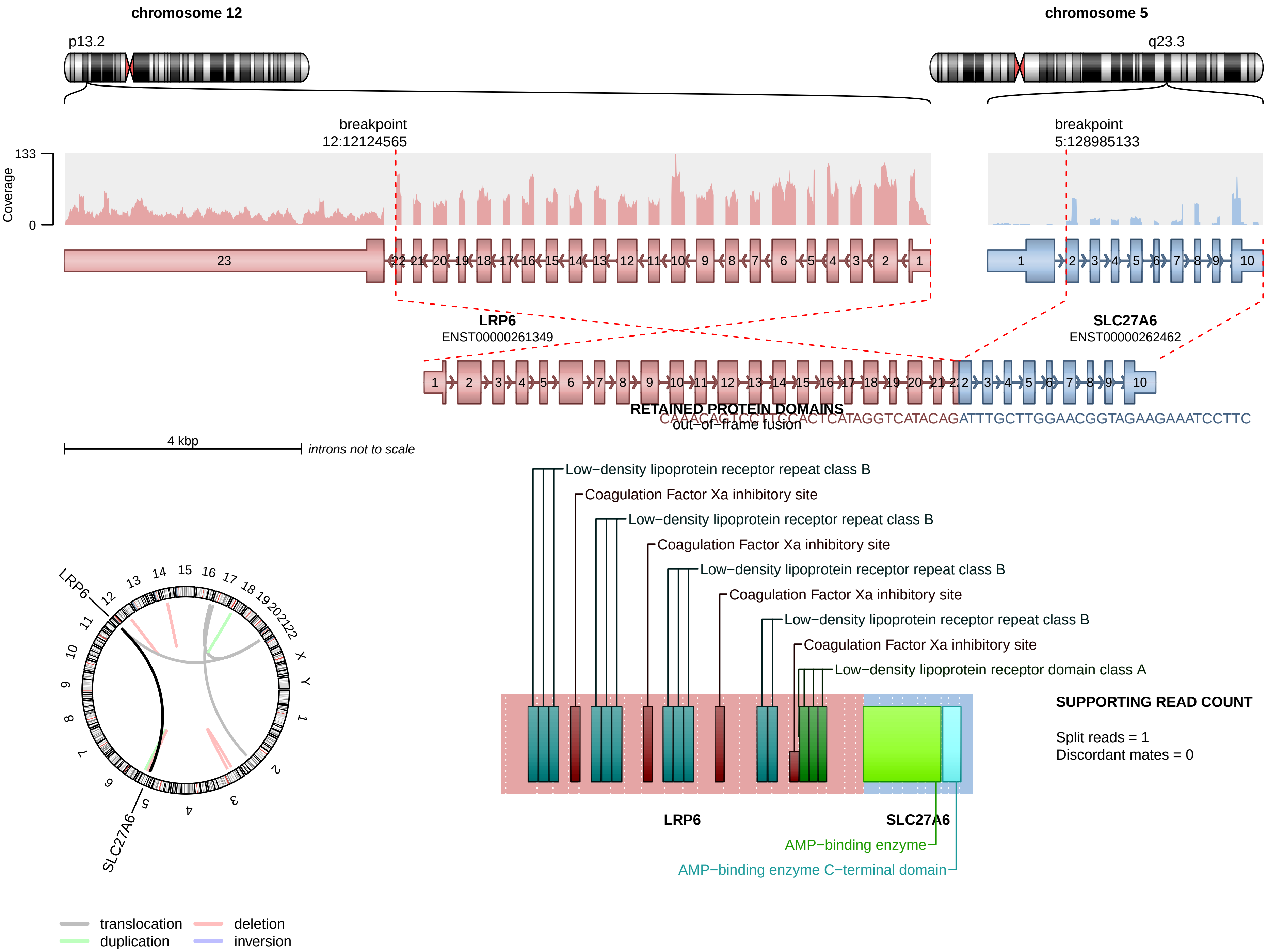

chromosome 15
q22.31
chromosome 15
q22.31
breakpoint
15:66331410
breakpoint
15:66352955
Coverage
302
0
2
1
8
7
6
5
4
3
2
1
AC055855.1
ENST00000565993
TIPIN
ENST00000261881
1
2
3
4
5
6
7
8
TGGGACTACAGGCGCCTGCCATCACGCCCGGAGGAAAAGATGCTAGAACCACAGGAGAATG
800 bp
introns not to scale


AC055855.1
TIPIN
15
16
17
18
19
20
21
22
X
Y
1
2
3
4
5
6
7
8
9
10
11
12
13
14
translocation
deletion
duplication
inversion

No protein domains retained in fusion.

SUPPORTING READ COUNT

Split reads = 1
Discordant mates = 1


chromosome 12
p13.2
chromosome 5
q23.3
breakpoint
12:12124565
breakpoint
5:128985133
Coverage
133
0
23
22 21 20 19 18 17 16 15 14 13 12 11 10 9 8 7 6 5 4 3 2 1
1 2 3 4 5 6 7 8 9 10
LRP6
ENST00000261349
SLC27A6
ENST00000262462
1 2 3 4 5 6 7 8 9 10 11 12 13 14 15 16 17 18 19 20 21 22 2 3 4 5 6 7 8 9 10
RETAINED PROTEIN DOMAINS
CAAACAGTCCTTCCACTCATAGGTCATACAGATTTGCTTGGAACGGTAGAAGAAATCCTTC
out−of−frame fusion
4 kbp
introns not to scale
Low−density lipoprotein receptor repeat class B
Coagulation Factor Xa inhibitory site
Low−density lipoprotein receptor repeat class B
Coagulation Factor Xa inhibitory site
Low−density lipoprotein receptor repeat class B
Coagulation Factor Xa inhibitory site
Low−density lipoprotein receptor repeat class B
Coagulation Factor Xa inhibitory site
Low−density lipoprotein receptor domain class A
LRP6
SLC27A6
AMP−binding enzyme
AMP−binding enzyme C−terminal domain
LRP6
SLC27A6
12 13 14 15 16 17 18 19 20 21 22 X Y 1 2 3 4 5 6 7 8 9 10 11
translocation
deletion
duplication
inversion

SUPPORTING READ COUNT

Split reads = 1
Discordant mates = 0