Patient_6

chromosome 4

q12

chromosome 4

p16.3

breakpoint1
chr4:55475963

breakpoint2
chr4:4391475

Coverage
120
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

CLOCK
ENST00000513440.6

NSG1
ENST00000397958.5

1 2 3 4 5 6 7 4 5 6
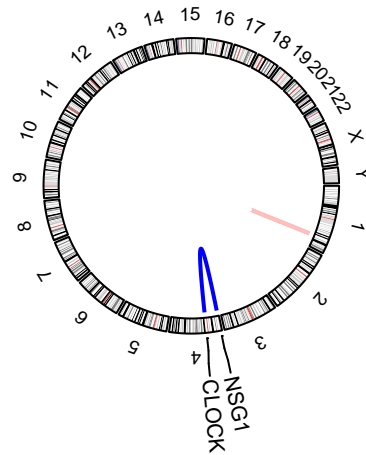
TAATGAAGAGTTTACACAATTAATGTTAGAGGTGGTCGTGAAAACTAAGACCGAGTATGAA

4 kbp
introns not to scale

RETAINED PROTEIN DOMAINS
in-frame fusion

Helix-loop-helix DNA-binding domain

PAS fold

CLOCK

NSG1

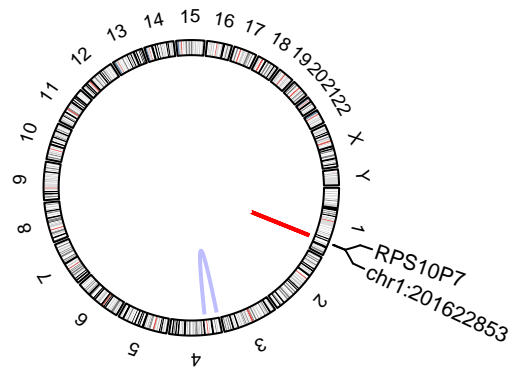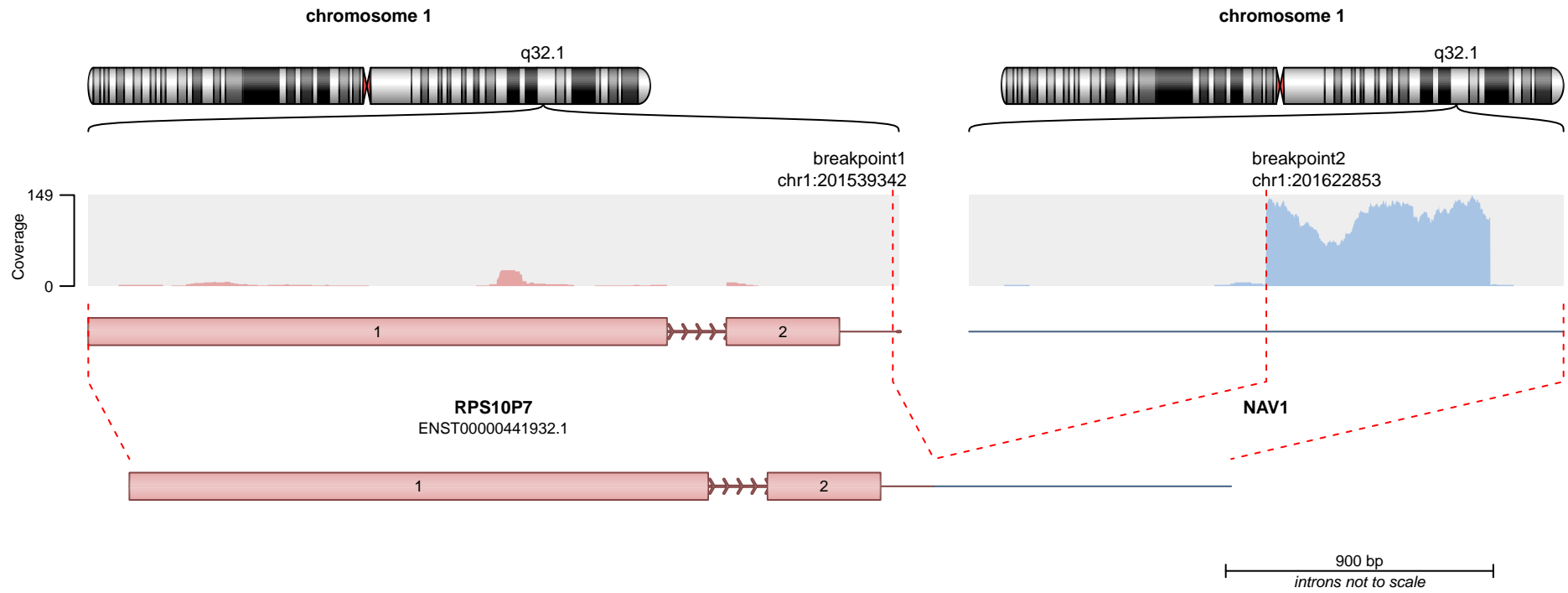D1 dopamine receptor-interacting protein (calcyon)
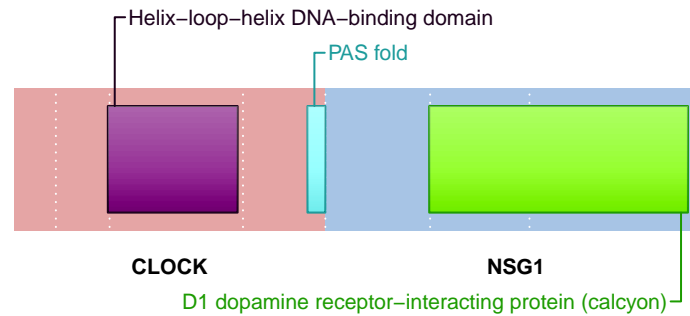
SUPPORTING READ COUNT

Split reads at breakpoint1 = 2
Split reads at breakpoint2 = 1
Discordant mates = 0

translocation   deletion
duplication   inversion

Patient_6

chromosome 1

q32.1

breakpoint1
chr1:201539342

149

Coverage

0

1

2

RPS10P7
ENST00000441932.1

1

2

chromosome 1

q32.1

breakpoint2
chr1:201622853

NAV1

900 bp
introns not to scale

RPS10P7
chr1:201622853

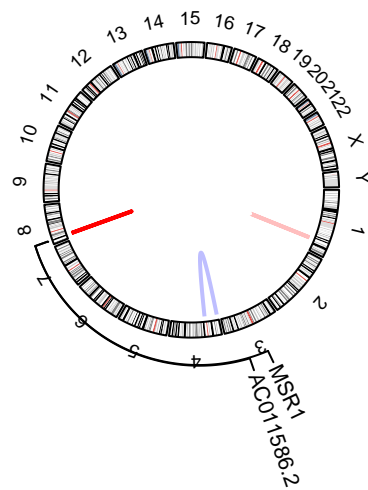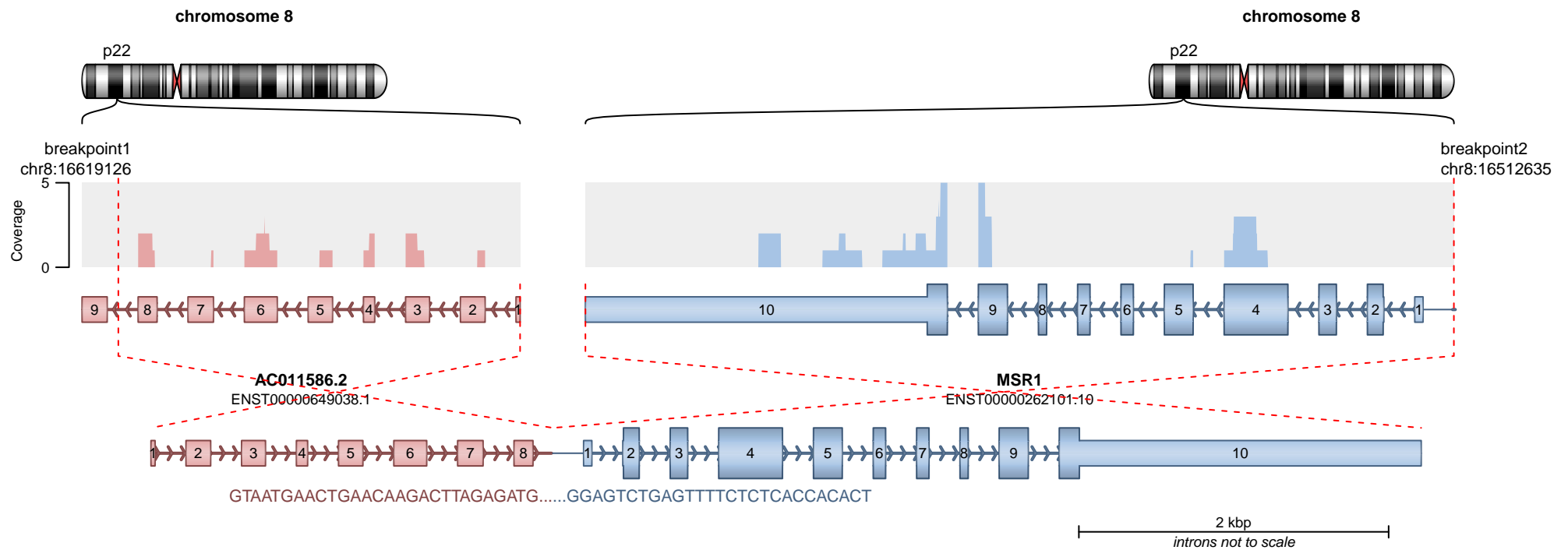Genes are not protein–coding.

SUPPORTING READ COUNT

Split reads at breakpoint1 = 17
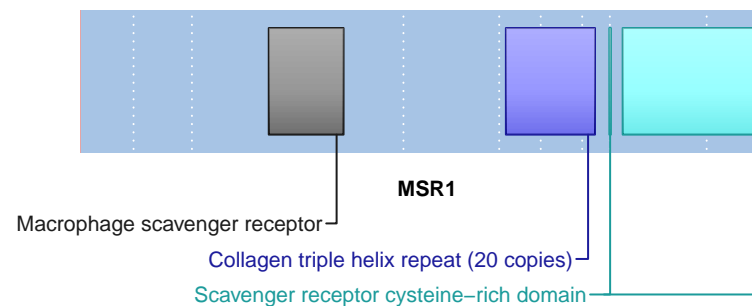Split reads at breakpoint2 = 2
Discordant mates = 5

translocation   deletion
duplication   inversion

Patient_6

chromosome 8

p22

breakpoint1
chr8:16619126

breakpoint2
chr8:16512635

Coverage

AC011586.2
ENST00000649038.1

MSR1
ENST00000262101.10

GTAATGAACTGAACAAGACTTAGAGATG......GGAGTCTGAGTTTTCTCTCACCACACT

2 kbp
introns not to scale

RETAINED PROTEIN DOMAINS
reading frame unclear

MSR1

Macrophage scavenger receptor

Collagen triple helix repeat (20 copies)

Scavenger receptor cysteine−rich domain

SUPPORTING READ COUNT

Split reads at breakpoint1 = 0
Split reads at breakpoint2 = 0
Discordant mates = 2

MSR1
AC011586.2

translocation   deletion
duplication   inversion