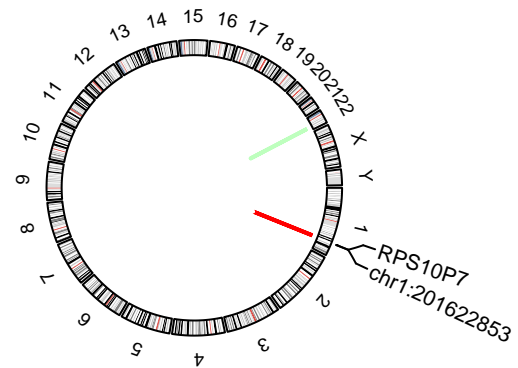
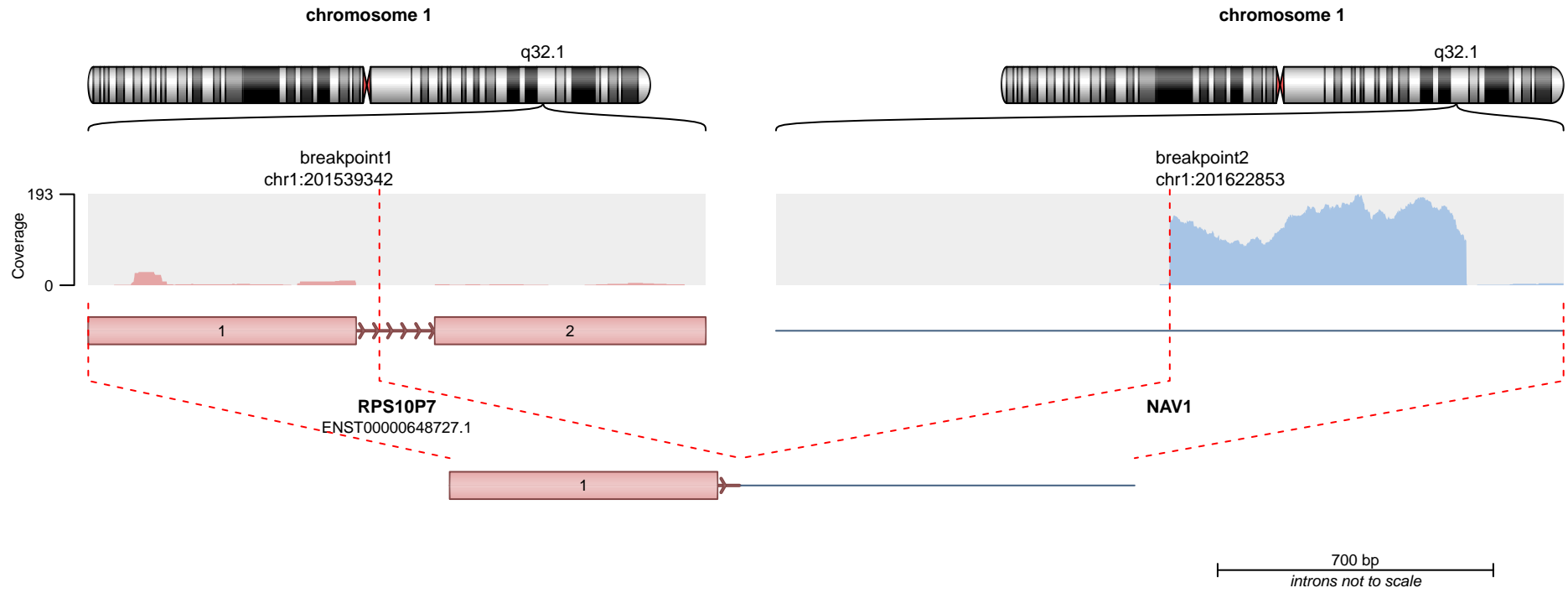


# Patient\_9



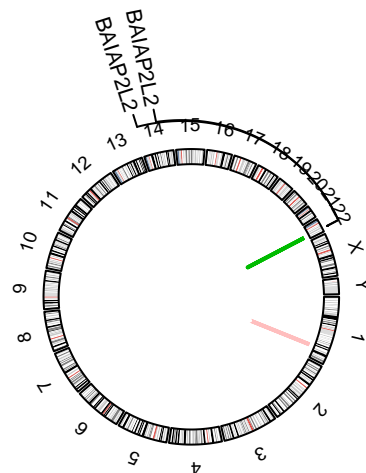
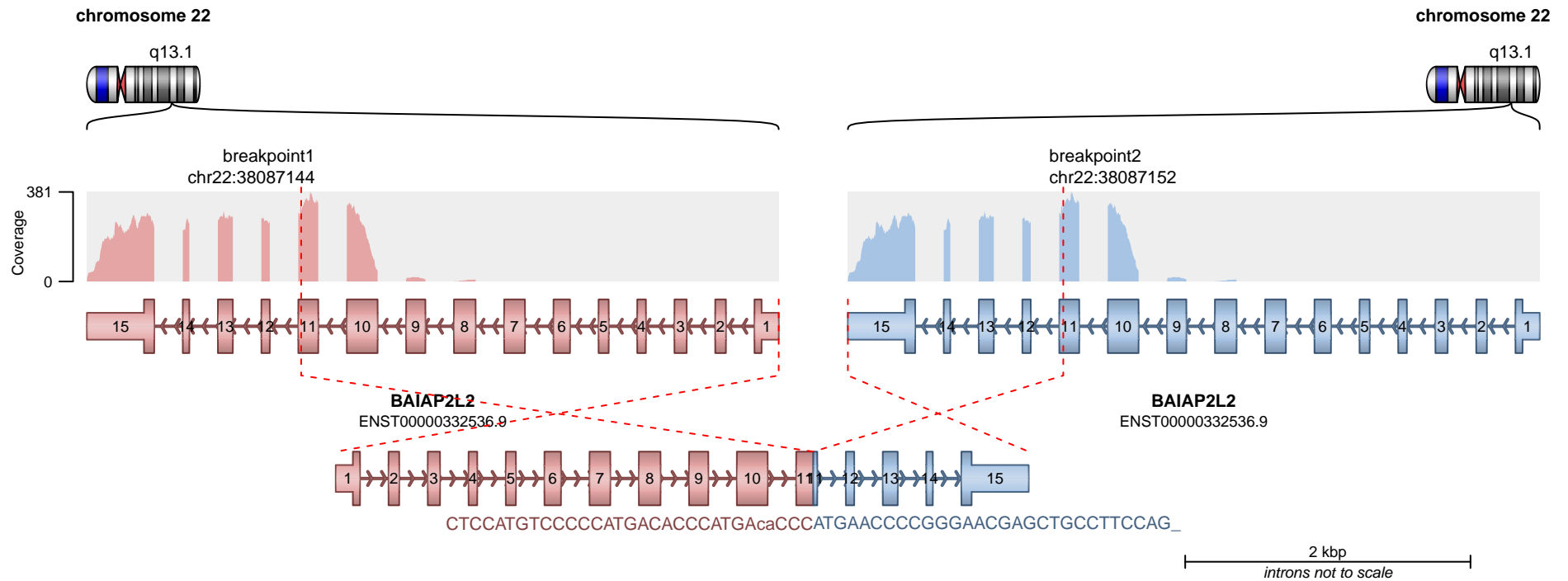
Genes are not protein-coding.

## SUPPORTING READ COUNT

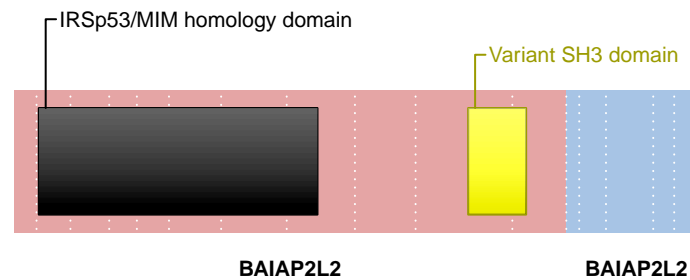
Split reads at breakpoint1 = 20  
Split reads at breakpoint2 = 16  
Discordant mates = 8

— translocation — deletion  
— duplication — inversion

# Patient\_9



## RETAINED PROTEIN DOMAINS in-frame fusion

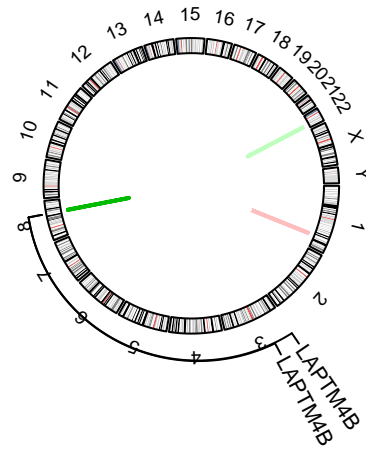
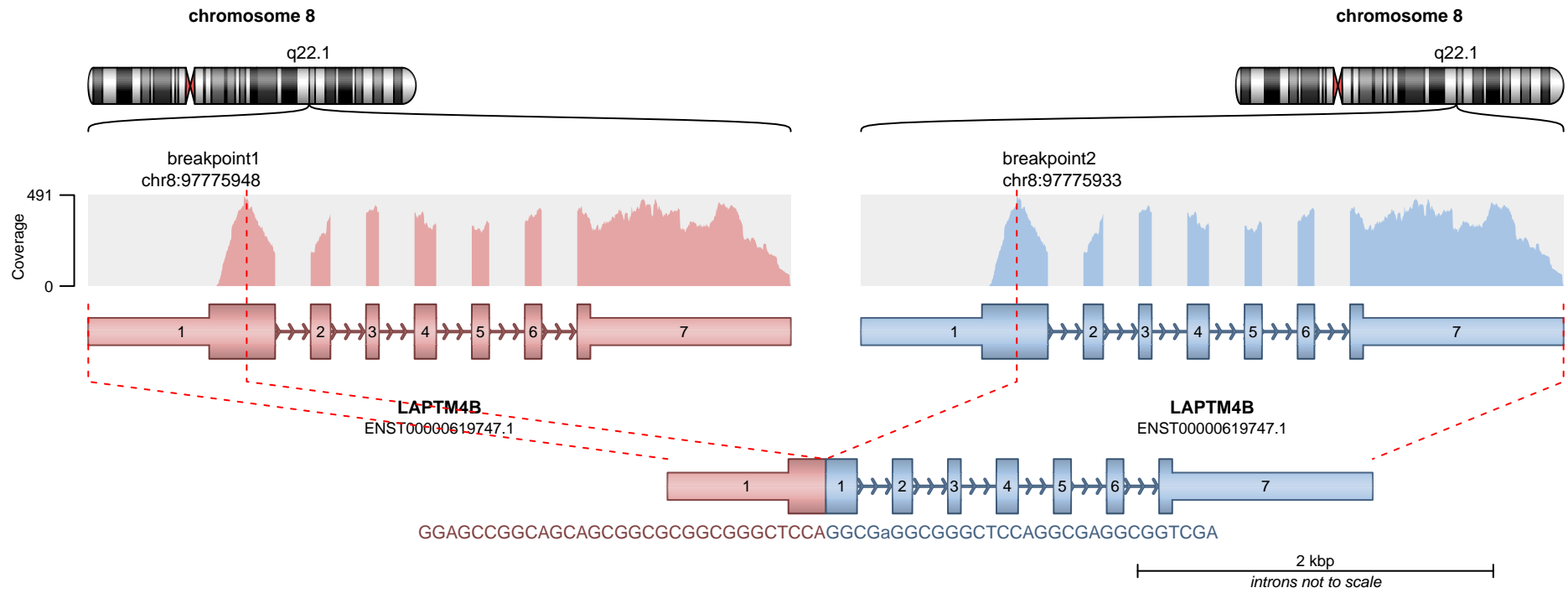


## SUPPORTING READ COUNT

Split reads at breakpoint1 = 13  
Split reads at breakpoint2 = 9  
Discordant mates = 0

translocation deletion  
duplication inversion

# Patient\_9



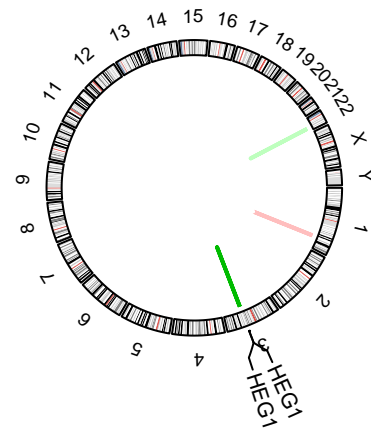
translocation deletion  
duplication inversion

No protein domains retained in fusion.

## SUPPORTING READ COUNT

Split reads at breakpoint1 = 26  
Split reads at breakpoint2 = 5  
Discordant mates = 0

The figure displays two alternative splicing events on chromosome 3, specifically at the q21.2 region. The top tracks show the genomic context with a red 'X' indicating the splice site. Below these are two tracks: 'breakpoint1' (chr3:125013564) and 'breakpoint2' (chr3:125013581). Each breakpoint track includes a signal plot (red for breakpoint1, blue for breakpoint2) and a gene model. The gene models show exons as numbered boxes (1-17) and introns as lines with arrows. The HEG1 gene is identified with the ENST00000311127.9 transcript. A detailed view of the DNA sequence is provided at the bottom, showing the junction between exon 6 and exon 7. The sequence is: CCTCTTCCTCCTCCTCCTCcTCTTCcTCcTCCTCCTCTCTCTTCTTCTTCAGGGCCTCCTTT. A scale bar indicates 5 kbp, and a note states 'introns not to scale'.



— translocation    — deletion  
— duplication    — inversion

HEG1

HEG1

EGF-like domain

Calcium-binding EGF domain

Split reads at breakpoint1 = 5  
Split reads at breakpoint2 = 9  
Discordant mates = 0