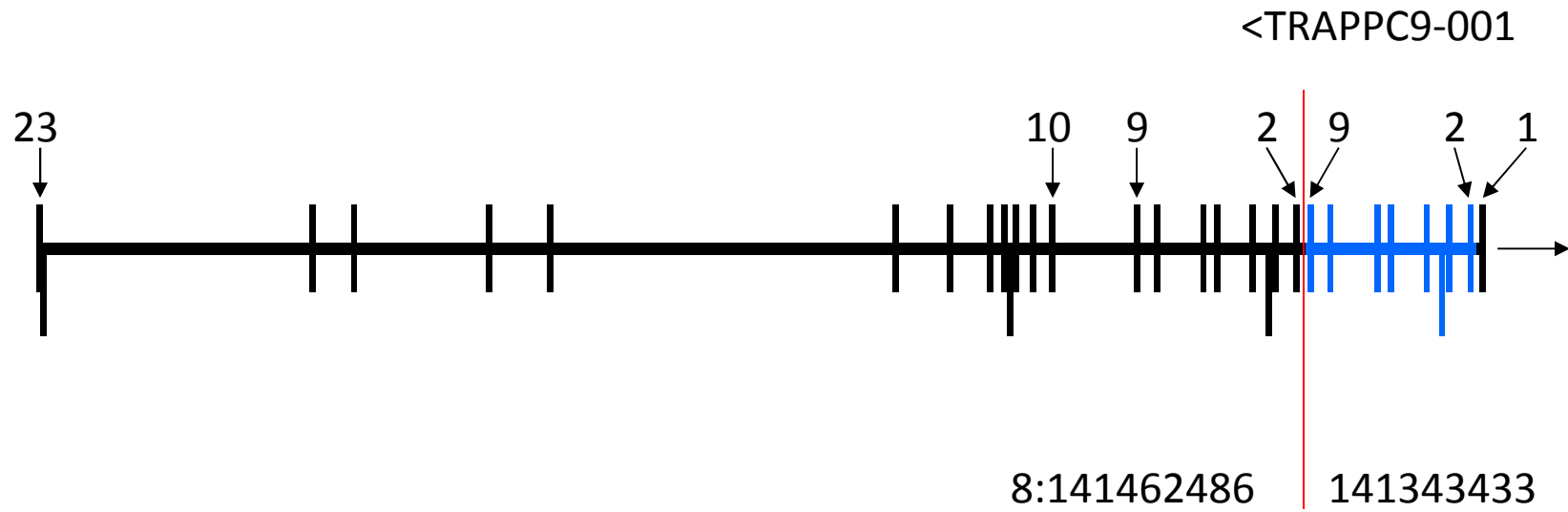
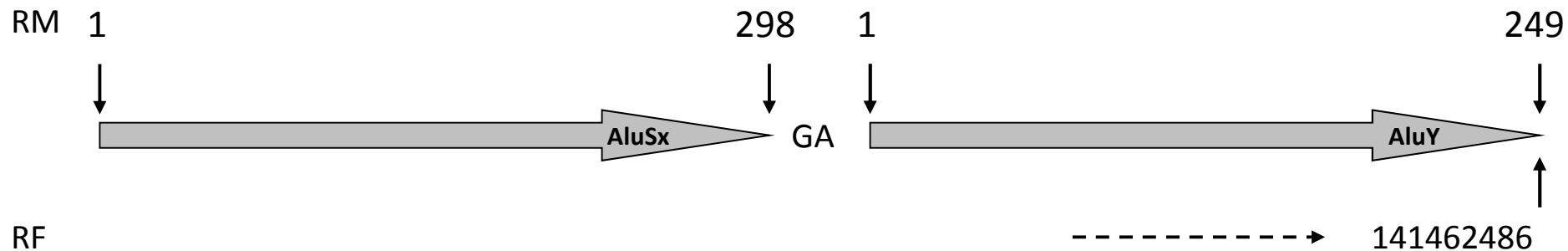


PATIENT 3: HOMOZYGOUS DUPLICATION OF *TRAPPC9* GENE

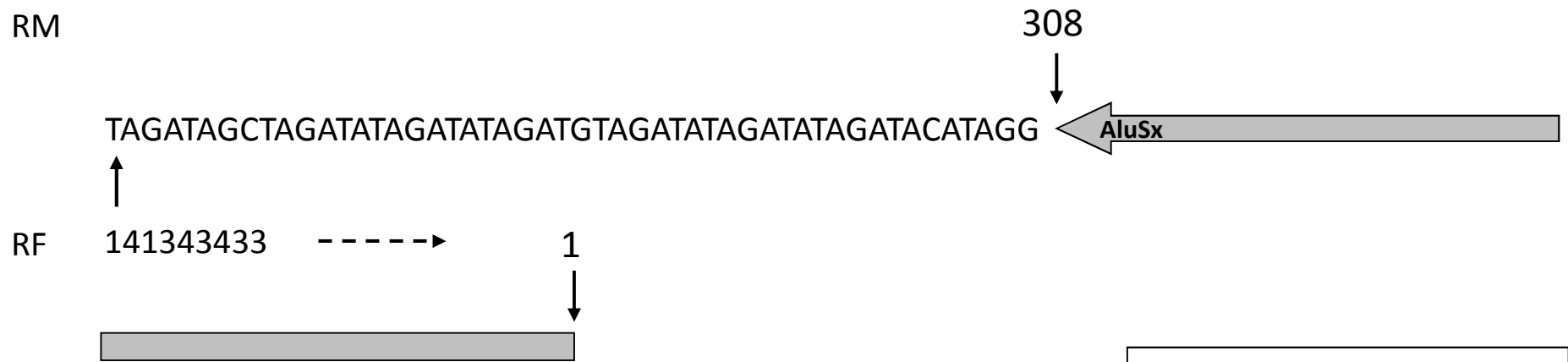


All nucleotide positions in hg 19

PATIENT 3: REPEATED SEQUENCES AT THE DUPLICATION JUNCTION

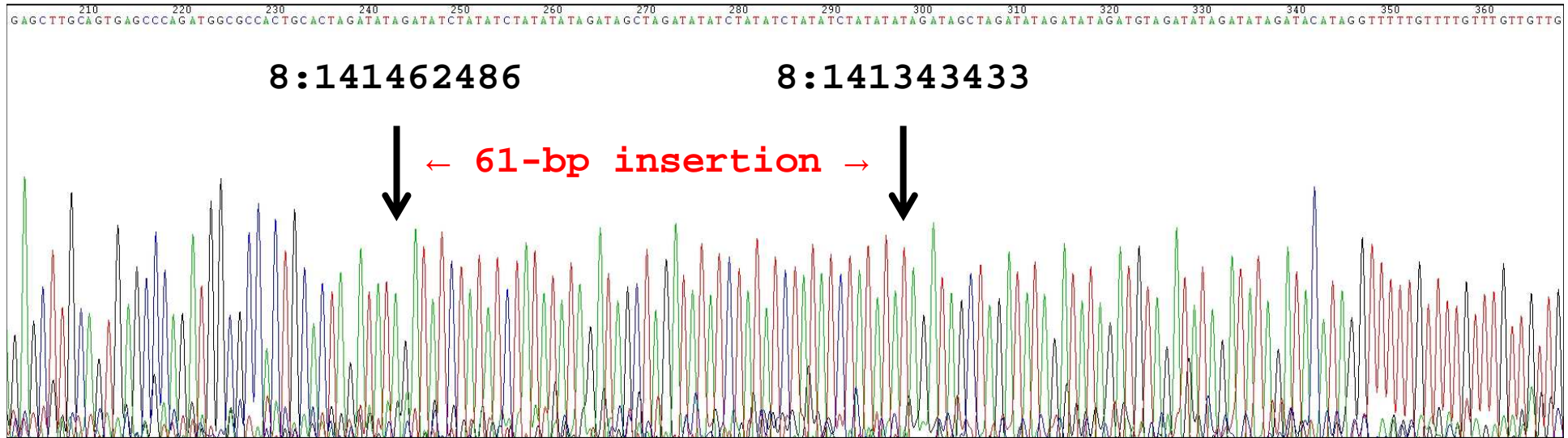


AGATATAGATATCTATATCTATATATAGATAGCTAGATATATCTATATCTATATA



RM: RepeatMasker
RF: Reference Sequence

PATIENT 3: GENOMIC SEQUENCE AT THE DUPLICATION JUNCTION



AAATTAGCCCGGCGTGGTGGCAGGCACCTGTAGTCCAGCTACTTGGGAGGCTGAGGCAG

8:141462486 ↓

GAGAATGGCATGAACCAGGGAGGCAGAGCTTGCAGTGAGCCCAGATGGCGCCACTGCACT

AGATATAGATATCTATATCTATATATAGCTAGATATATCTATATCTATATCTATATA

← 61-bp insertion →

TAGATAGCTAGATATAGATATAGATGTAGATATAGATATAGATACATAGGTTTTTGT

↑ 8:141343433

GTTTGTGTTGTTGTTGTTGAGACGGAGTCTCACTCTTGTGCGCCAGGCTGGAGTGCAATGGT

PATIENT 3: *TRAPPC9* DUPLICATION cDNA SEQUENCE



← Exon 2 AGTGGTGCCTGTAGCGGATGTAGAGGACTCGCTGGGAGTCCCGCACGCTGA
TCTGACTCACAGAGCAAATCCTCTTATAGATCCTGAAGAAGTTCTCCTCGGAGACGATGC
CCACAGGCTGGACCACCACGAGCAGCGTCTGGTGGTCCTCAGCACACTGCATGTAGTCAG
GGACGCTCATTTTGAAGTCC_CCTGATCCGACAAGAAGTCCAGCATGGTCTGTAGAAGGA
AGGACAGGTGTCTGACAGAGAGGGCAGGGTTCCCCATCCTTCGGGAGGCGTAGACCAATT
CATGGAGCAAACGCATCTGG ← Exon 9 (310 bp)

cDNA PRIMERS	SEQUENCES	LOCATION (hg19)	Exon
TRAPPC9_ex2_F1	AGTGGTGCCTGTAGCGGATG	8:141461292-141461310	2
TRAPPC9_ex9_R1	CCAGATGCGTTTGCTCCATG	8:141370248-141370267	9