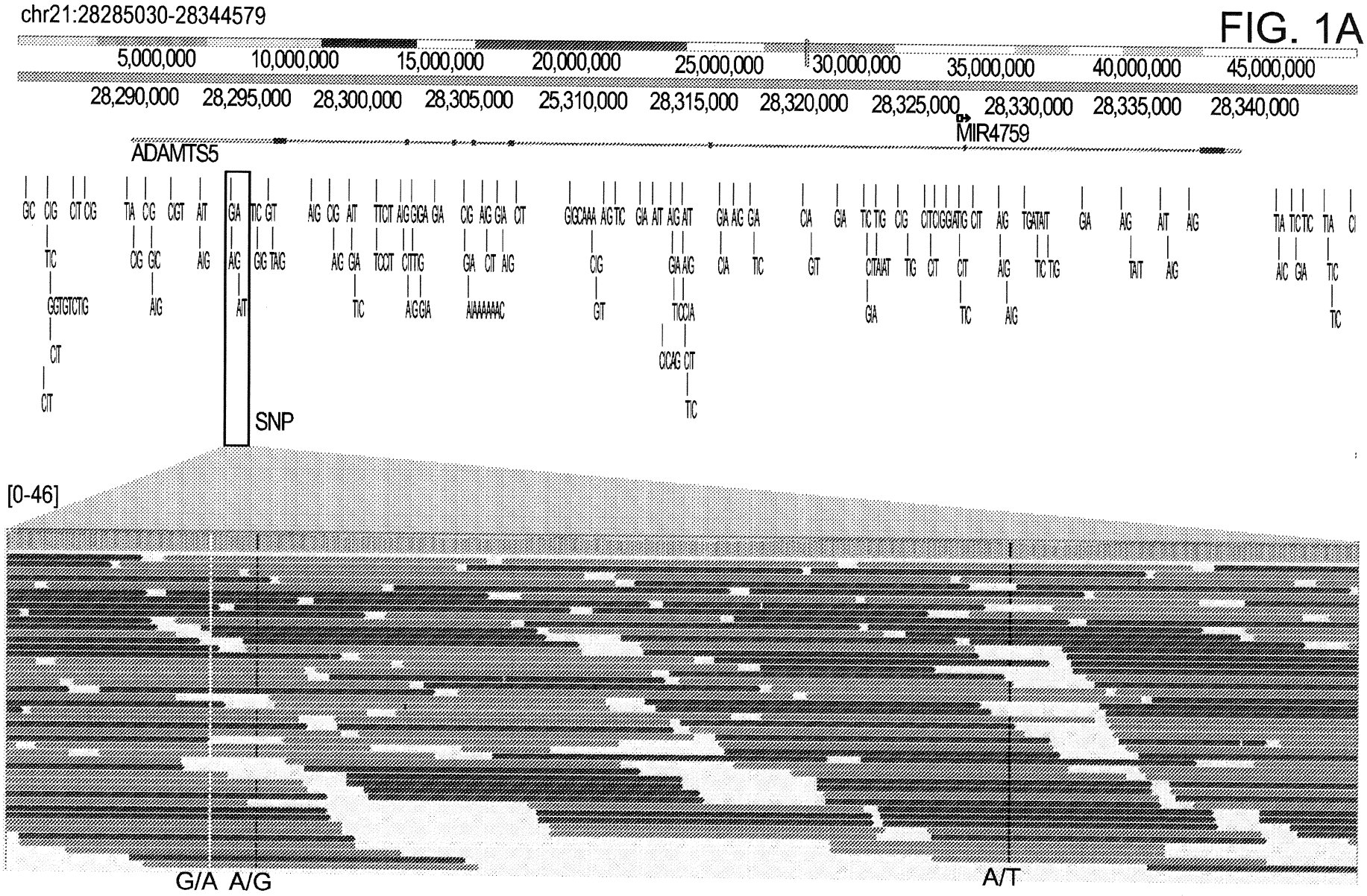
FIG. 1A


chr21:28285030-28344579
5,000,000
10,000,000
15,000,000
20,000,000
25,000,000
30,000,000
35,000,000
40,000,000
45,000,000
28,290,000
28,295,000
28,300,000
28,305,000
25,310,000
28,315,000
28,320,000
28,325,000
28,330,000
28,335,000
28,340,000
MIR4759
ADAMTS5
SNP
[0-46]
G/A A/G
A/T

chr21:28285030-28344579

FIG. 1B

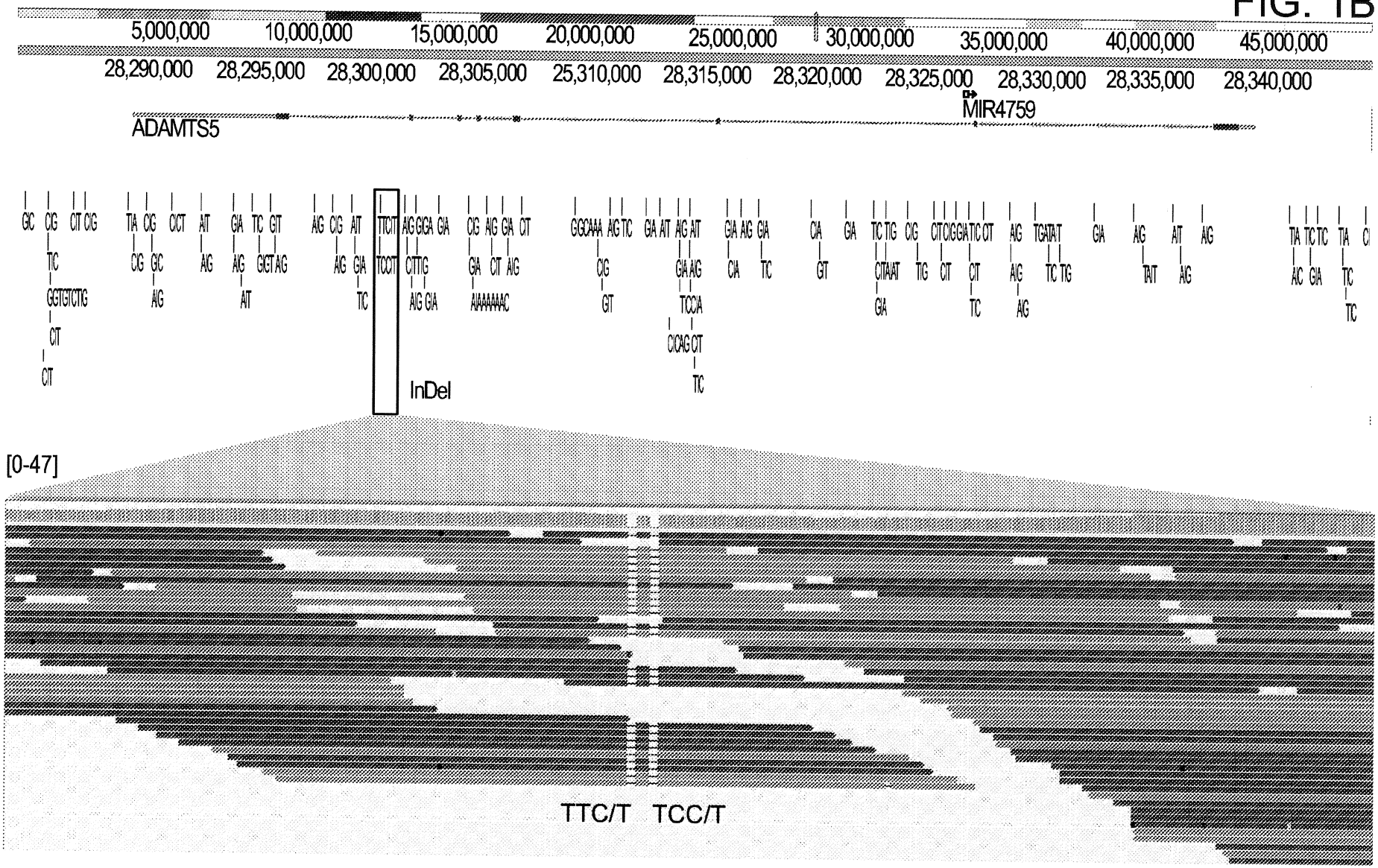


FIG. 2

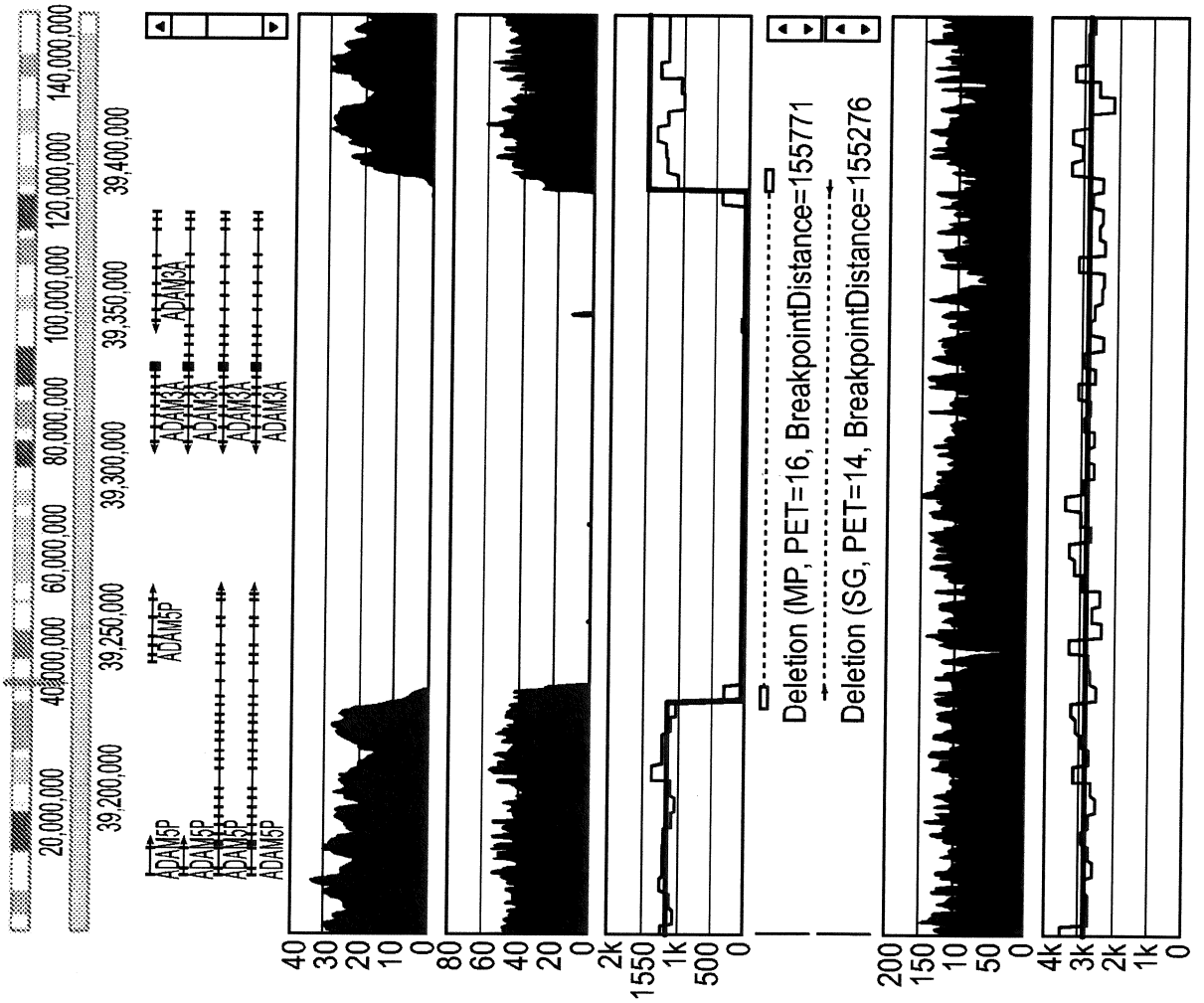

chr8:39154557-39442371,
Homozygous Deletion
20,000,000
40,000,000
60,000,000
80,000,000
100,000,000
120,000,000
140,000,000
39,200,000
39,250,000
39,300,000
39,350,000
39,400,000
ADAM5P
ADAM3A
P46107
(Patient sample)
Mate-pair
Physical coverage
Shotgun
Base pair coverage
CNV
Mate-pair cluster
Shotgun cluster
Deletion (MP, PET=16, BreakpointDistance=155771
Deletion (SG, PET=14, BreakpointDistance=155276
JAX0001
(Control sample)
Shotgun
Base pair coverage
CNV

FIG. 3

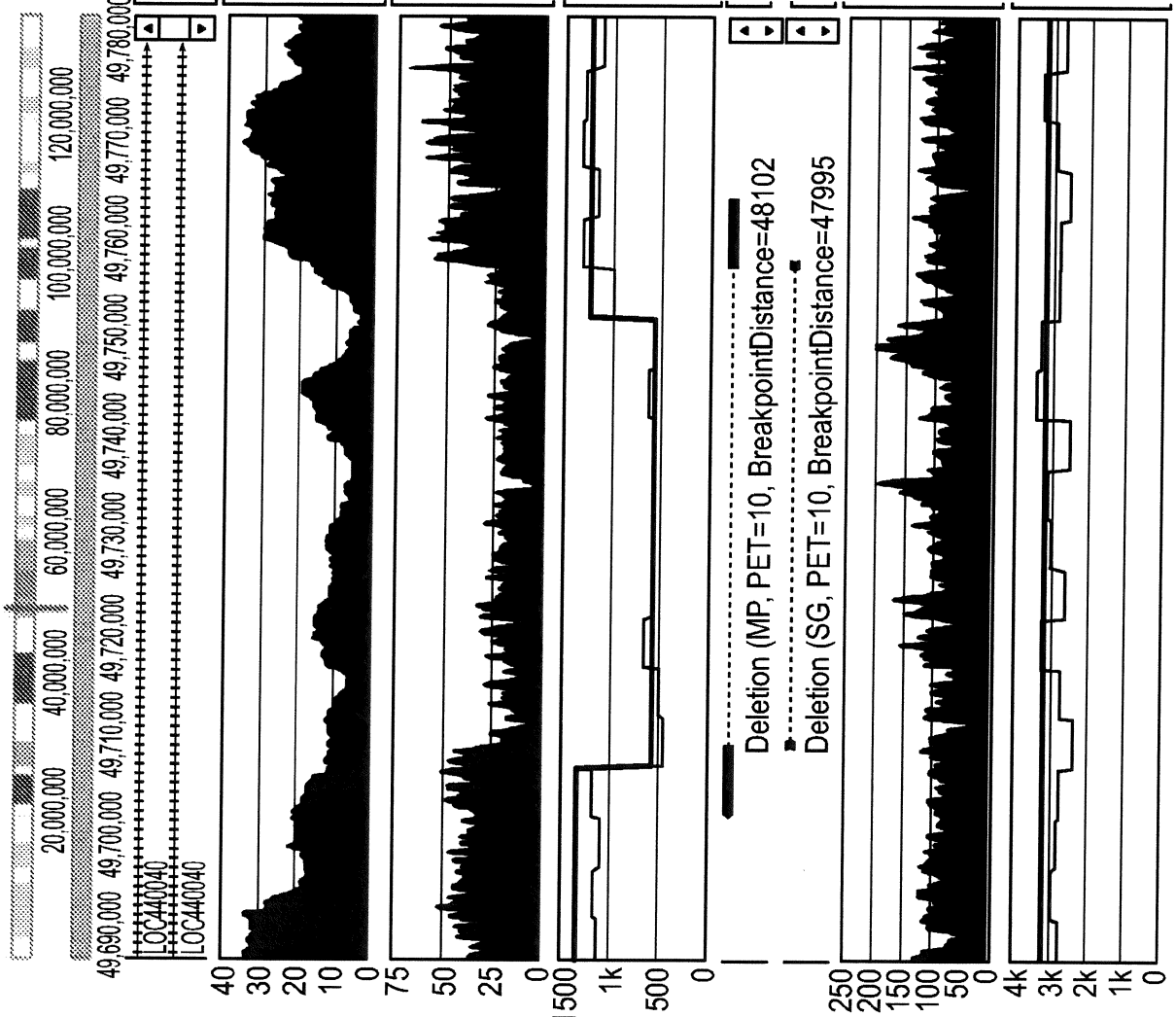
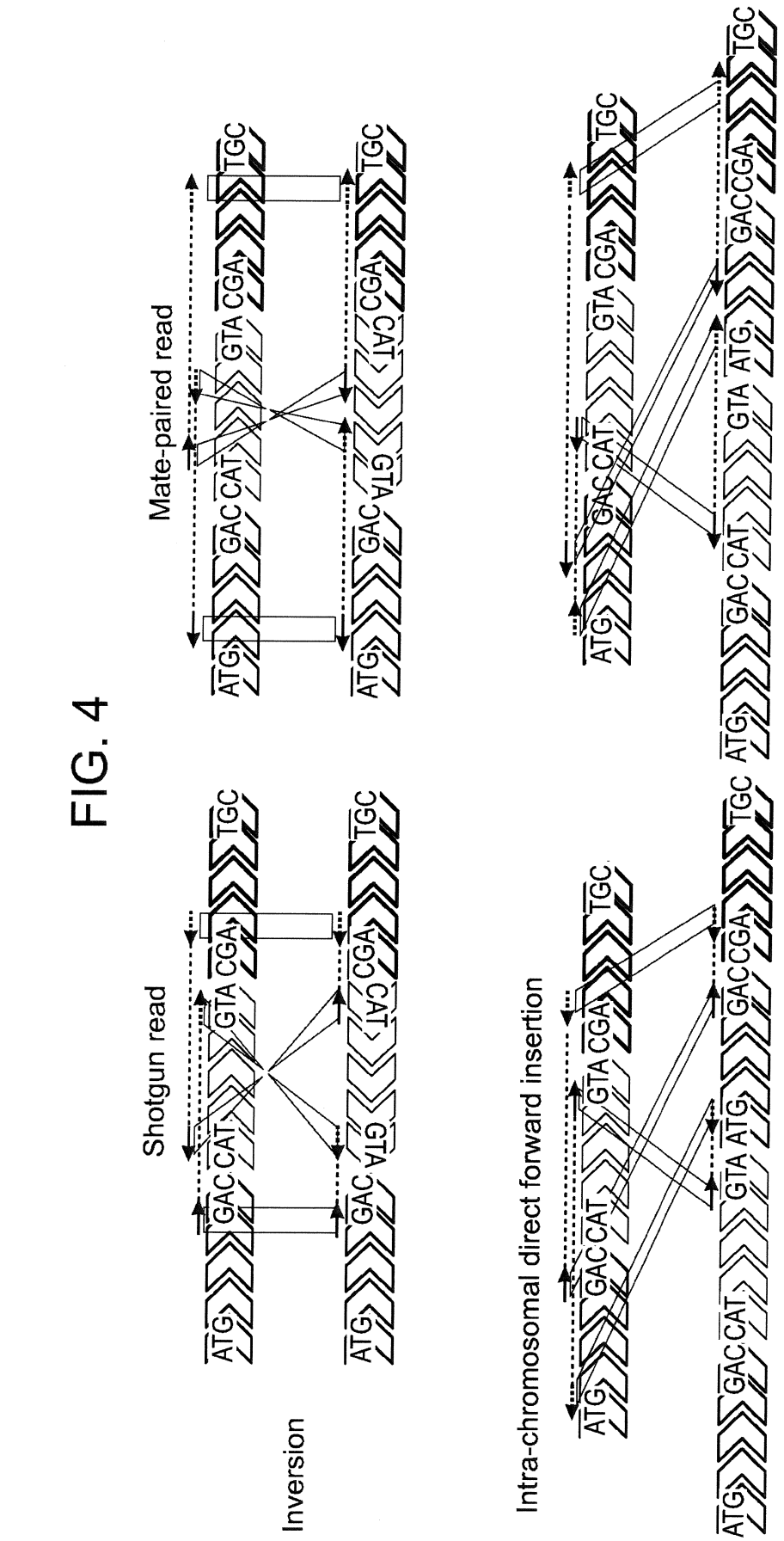

chr11: 49688366-49782733,
Heterozygous Deletion
P46107
(Patient sample)
JAX0001
(Control sample)
Mate-pair
Physical coverage
Shotgun
Base pair coverage
CNV
Mate-pair cluster
Shotgun cluster
Deletion (MP, PET=10, BreakpointDistance=48102
Deletion (SG, PET=10, BreakpointDistance=47995
20,000,000
40,000,000
60,000,000
80,000,000
100,000,000
120,000,000
49,690,000
49,700,000
49,710,000
49,720,000
49,730,000
49,740,000
49,750,000
49,760,000
49,770,000
49,780,000
LOC440040
LOC440040
40
30
20
10
0
75
50
25
0
1500
1k
500
0
250
200
150
100
50
0
4k
3k
2k
1k
0

FIG. 4


Mate-paired read
Shotgun read
Inversion
Intra-chromosomal direct forward insertion
ATG
GAC
CAT
GTA
CGA
TGC

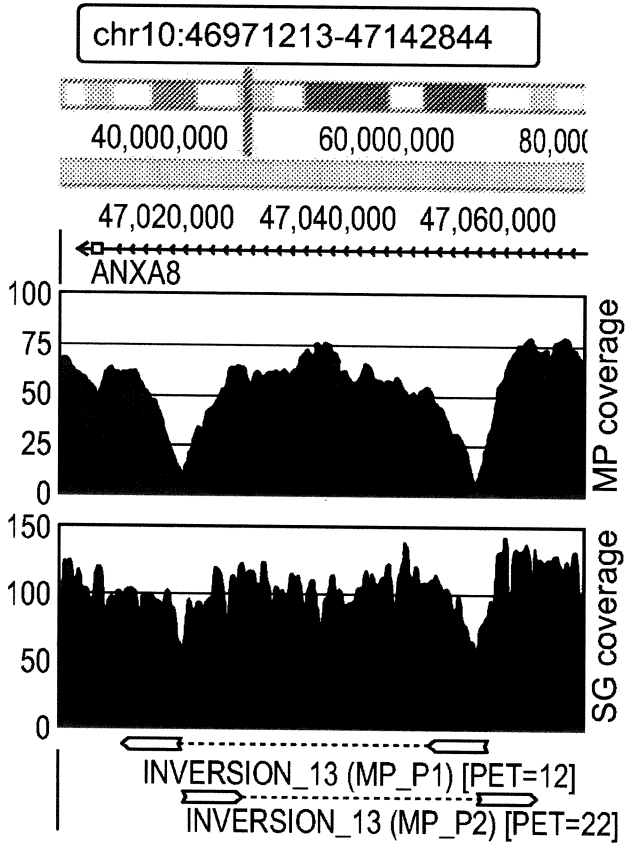
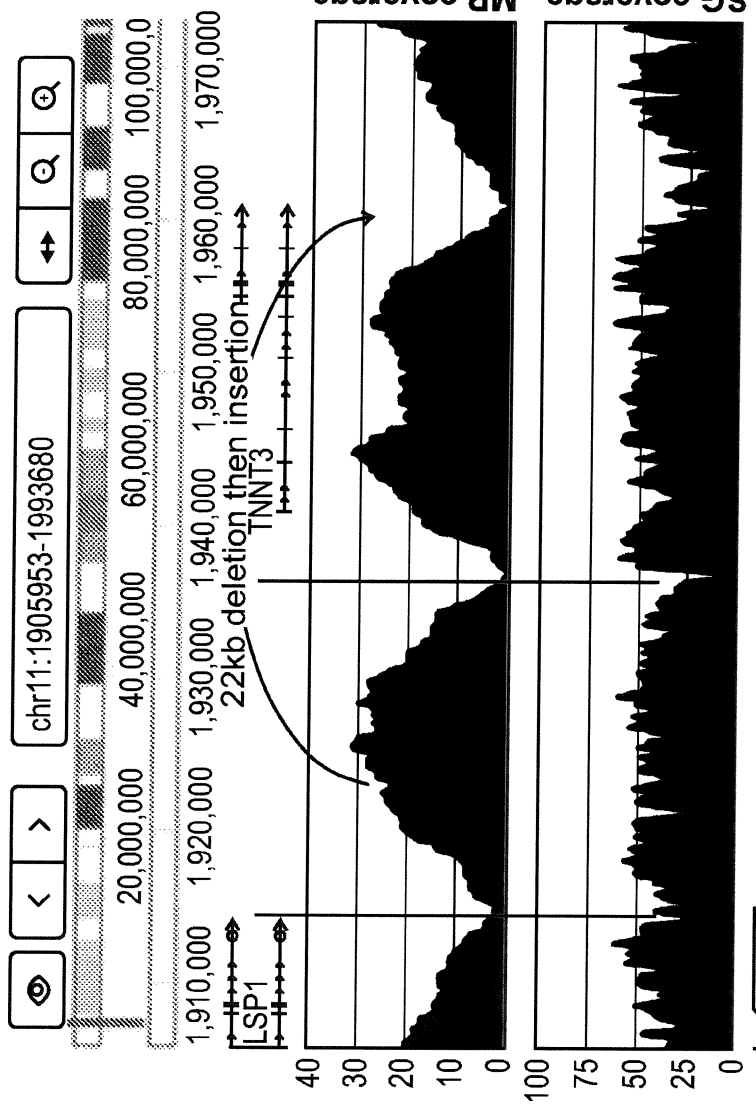


FIG. 5

SVStartRegion	SVTranslocated Region	SVEndRegion	SVStartBreak point	SVEndBreak point	Breakpoint Distance	Translocated Span
chr10:47016481..47022581	chr10:47023194..47059388	chr10:47059601..47066456	chr10:47022581	chr10:47059601	37020	36194

ClusterID	ChrL	StrandL	StartL	EndL	SpanL	ChrR	StrandR	StartR	EndR	SpanR	Size
84573_1	10	+	47016481	47022581	6101	10	-	47054482	47059388	4907	12
87751_1	10	-	47023194	47030036	6843	10	+	47059601	47066456	6856	22

FIG. 6



SVType	SVStartRegion	SVTranslocatedRegion	SVEnd Region	SVStartBreakpoint	SVEndBreakpoint	Breakpoint Distance	Translocated Span
INSERTION-INTRA-INV_6	chr11:1957607..1960251	chr11:19169 43..1935852	chr11:19640 95..1965562	chr11:196025 1	chr11:196409 5	3844	18909

FIG. 7

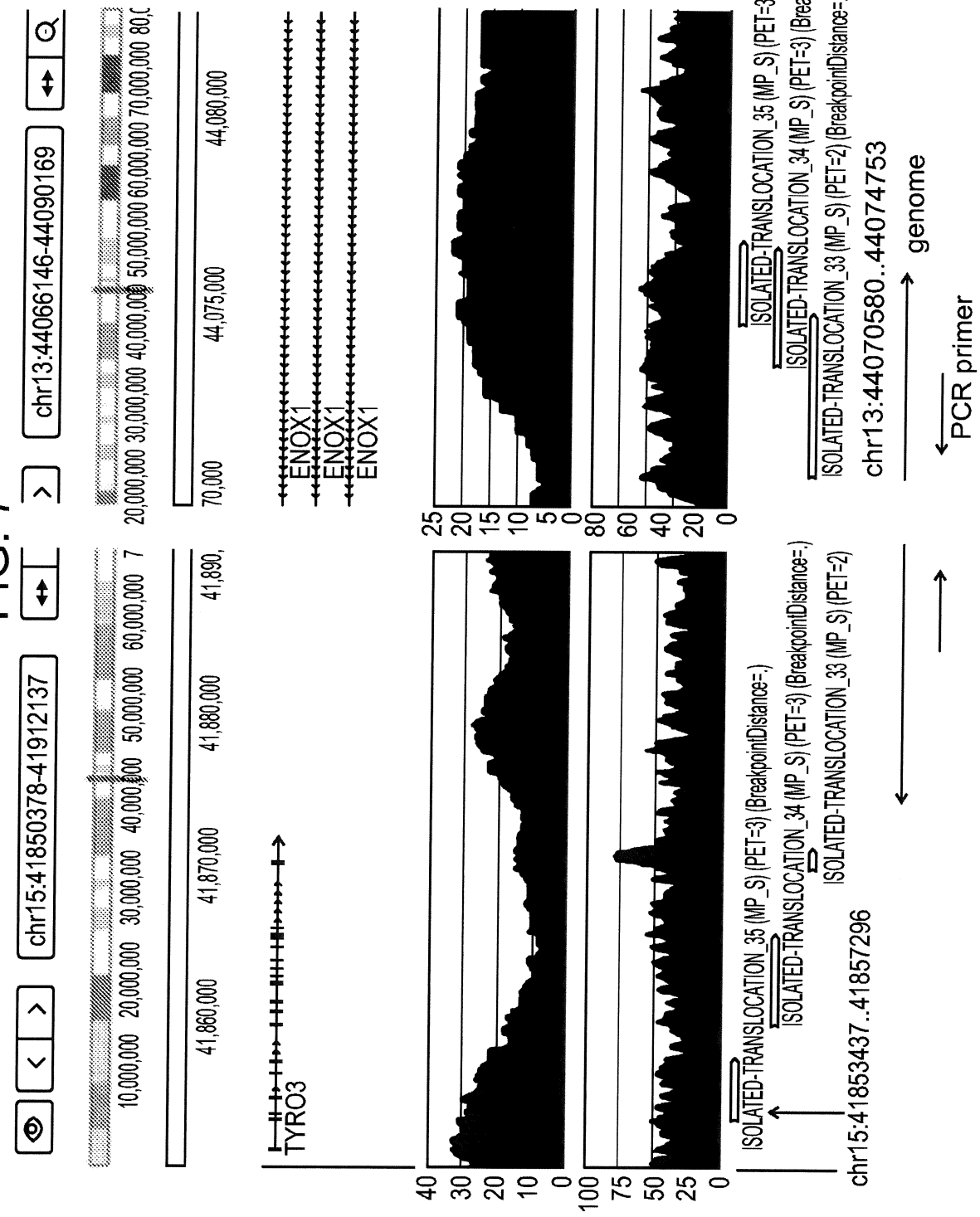

chr15:41850378-41912137
chr13:44066146-44090169
10,000,000 20,000,000 30,000,000 40,000,000 50,000,000 60,000,000 7
20,000,000 30,000,000 40,000,000 50,000,000 60,000,000 70,000,000 80,(
41,860,000 41,870,000 41,880,000 41,890,
70,000 44,075,000 44,080,000
TYRO3
ENOX1
ENOX1
ENOX1
MP coverage
SG coverage
ISOLATED-TRANSLOCATION_35 (MP_S) (PET=3) (BreakpointDistance=.)
ISOLATED-TRANSLOCATION_34 (MP_S) (PET=3) (BreakpointDistance=.)
ISOLATED-TRANSLOCATION_33 (MP_S) (PET=2)
chr15:41853437..41857296
chr13:44070580..44074753
genome
PCR primer

FIG. 8

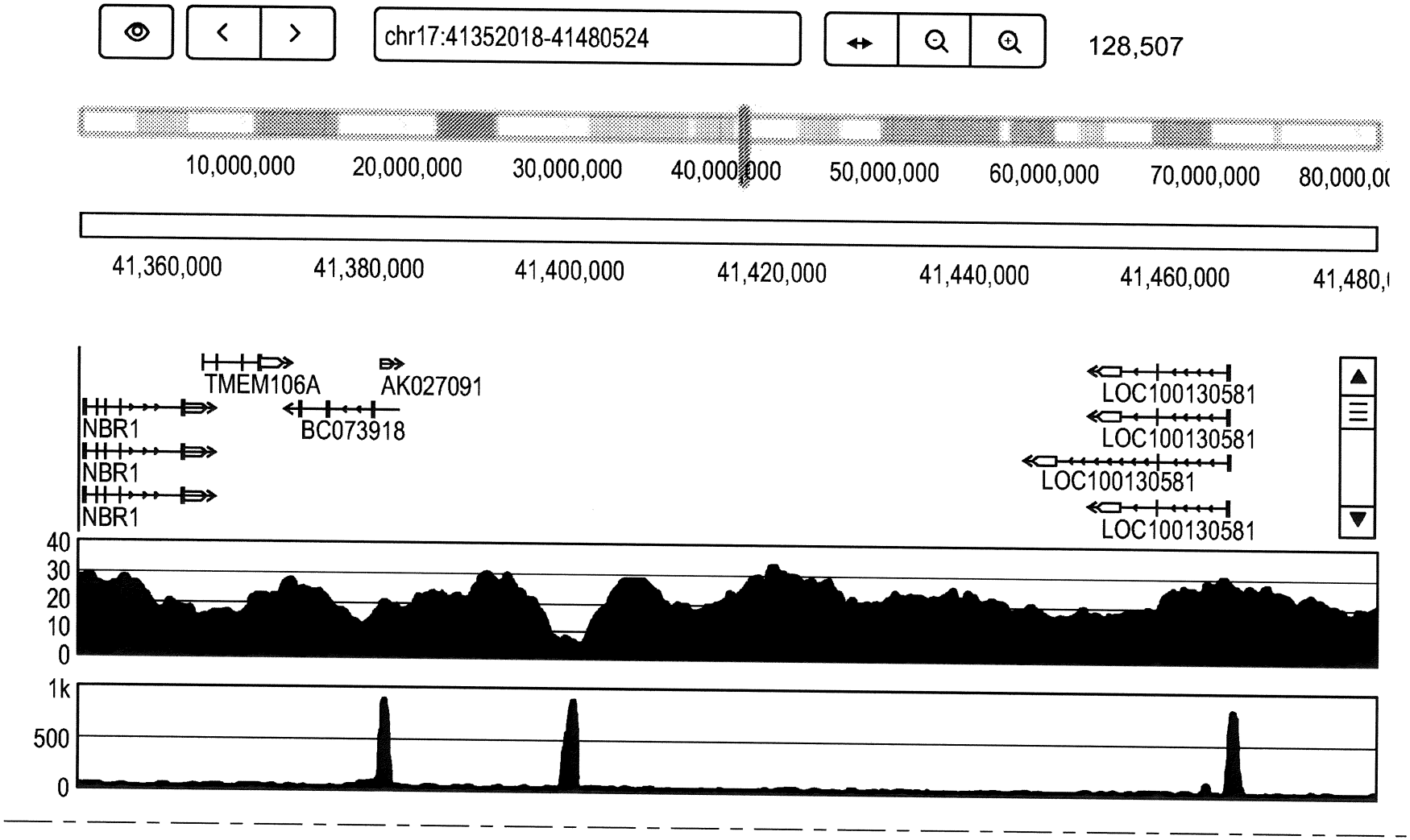

chr17:41352018-41480524
128,507
10,000,000
20,000,000
30,000,000
40,000,000
50,000,000
60,000,000
70,000,000
41,360,000
41,380,000
41,400,000
41,420,000
41,440,000
41,460,000
TMEM106A
AK027091
NBR1
BC073918
NBR1
NBR1
LOC100130581
LOC100130581
LOC100130581
LOC100130581
40
30
20
10
0
1k
500
0

FIG. 8 Cont.

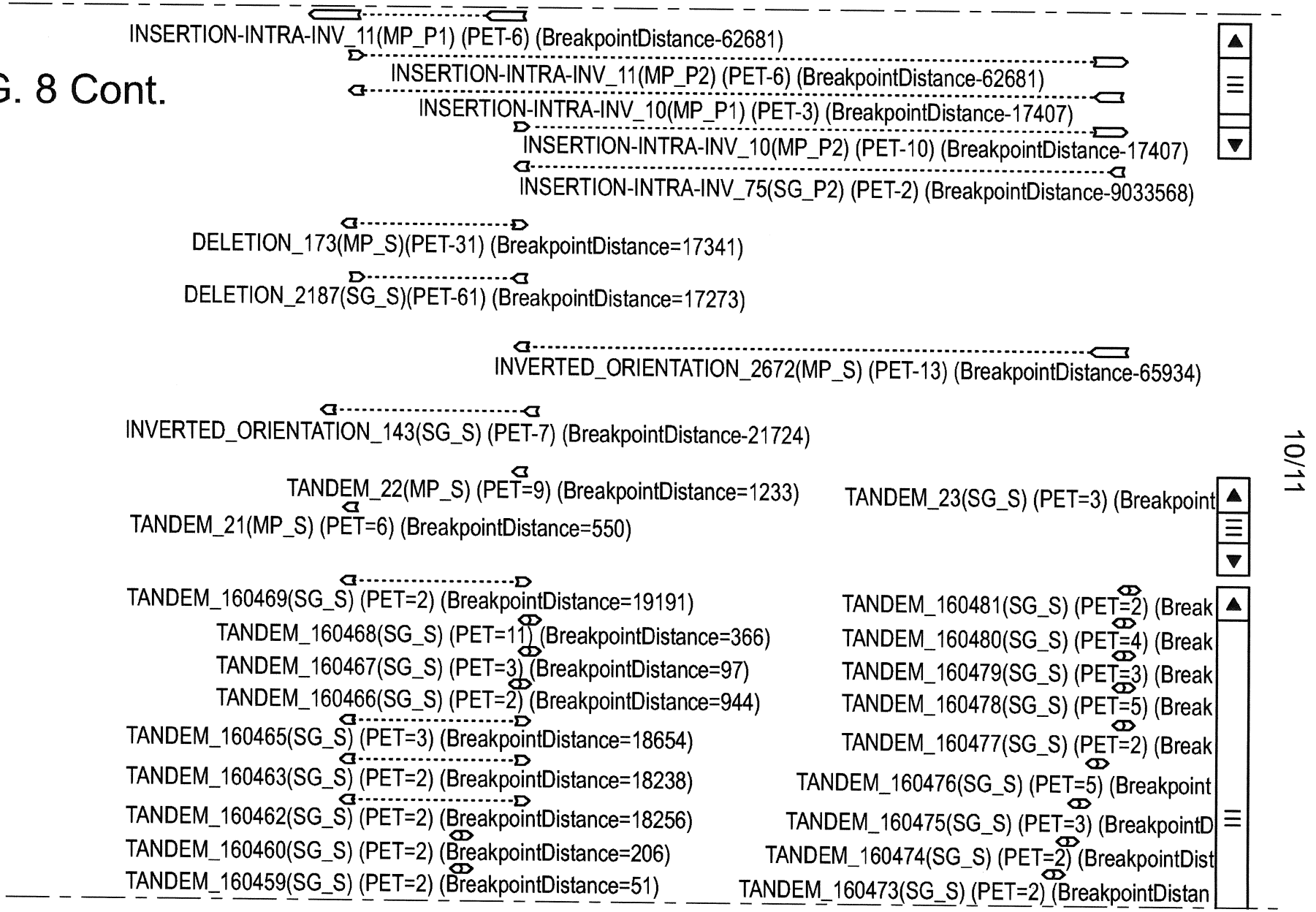
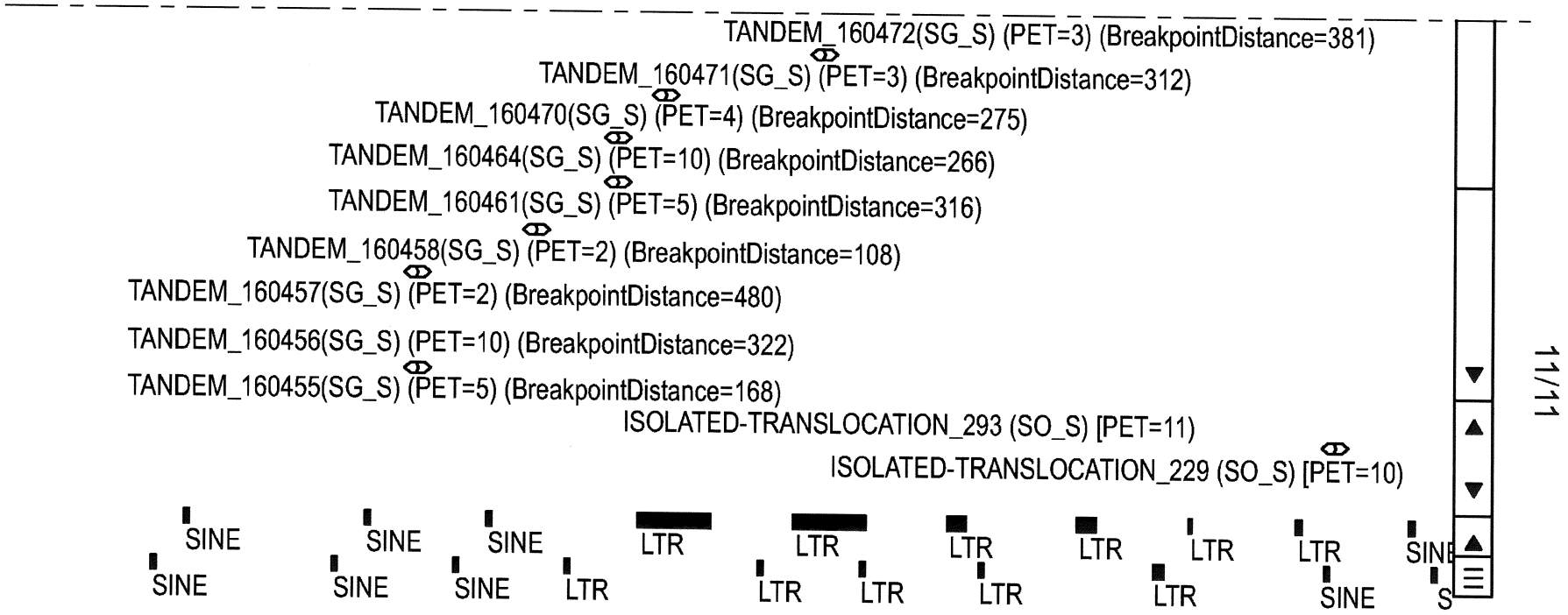

INSERTION-INTRA-INV_11(MP_P1) (PET-6) (BreakpointDistance-62681)
INSERTION-INTRA-INV_11(MP_P2) (PET-6) (BreakpointDistance-62681)
INSERTION-INTRA-INV_10(MP_P1) (PET-3) (BreakpointDistance-17407)
INSERTION-INTRA-INV_10(MP_P2) (PET-10) (BreakpointDistance-17407)
INSERTION-INTRA-INV_75(SG_P2) (PET-2) (BreakpointDistance-9033568)
DELETION_173(MP_S)(PET-31) (BreakpointDistance=17341)
DELETION_2187(SG_S)(PET-61) (BreakpointDistance=17273)
INVERTED_ORIENTATION_2672(MP_S) (PET-13) (BreakpointDistance-65934)
INVERTED_ORIENTATION_143(SG_S) (PET-7) (BreakpointDistance-21724)
TANDEM_22(MP_S) (PET=9) (BreakpointDistance=1233)
TANDEM_21(MP_S) (PET=6) (BreakpointDistance=550)
TANDEM_23(SG_S) (PET=3) (Breakpoint
TANDEM_160469(SG_S) (PET=2) (BreakpointDistance=19191)
TANDEM_160468(SG_S) (PET=11) (BreakpointDistance=366)
TANDEM_160467(SG_S) (PET=3) (BreakpointDistance=97)
TANDEM_160466(SG_S) (PET=2) (BreakpointDistance=944)
TANDEM_160465(SG_S) (PET=3) (BreakpointDistance=18654)
TANDEM_160463(SG_S) (PET=2) (BreakpointDistance=18238)
TANDEM_160462(SG_S) (PET=2) (BreakpointDistance=18256)
TANDEM_160460(SG_S) (PET=2) (BreakpointDistance=206)
TANDEM_160459(SG_S) (PET=2) (BreakpointDistance=51)
TANDEM_160481(SG_S) (PET=2) (Break
TANDEM_160480(SG_S) (PET=4) (Break
TANDEM_160479(SG_S) (PET=3) (Break
TANDEM_160478(SG_S) (PET=5) (Break
TANDEM_160477(SG_S) (PET=2) (Break
TANDEM_160476(SG_S) (PET=5) (Breakpoint
TANDEM_160475(SG_S) (PET=3) (BreakpointD
TANDEM_160474(SG_S) (PET=2) (BreakpointDist
TANDEM_160473(SG_S) (PET=2) (BreakpointDistan

FIG. 8 Cont.


TANDEM_160472(SG_S) (PET=3) (BreakpointDistance=381)
TANDEM_160471(SG_S) (PET=3) (BreakpointDistance=312)
TANDEM_160470(SG_S) (PET=4) (BreakpointDistance=275)
TANDEM_160464(SG_S) (PET=10) (BreakpointDistance=266)
TANDEM_160461(SG_S) (PET=5) (BreakpointDistance=316)
TANDEM_160458(SG_S) (PET=2) (BreakpointDistance=108)
TANDEM_160457(SG_S) (PET=2) (BreakpointDistance=480)
TANDEM_160456(SG_S) (PET=10) (BreakpointDistance=322)
TANDEM_160455(SG_S) (PET=5) (BreakpointDistance=168)
ISOLATED-TRANSLOCATION_293 (SO_S) [PET=11)
ISOLATED-TRANSLOCATION_229 (SO_S) [PET=10)
SINE
SINE
SINE
LTR
LTR
LTR
LTR
LTR
LTR
SINE
SINE
SINE
SINE
LTR
LTR
LTR
LTR
LTR
SINE
S