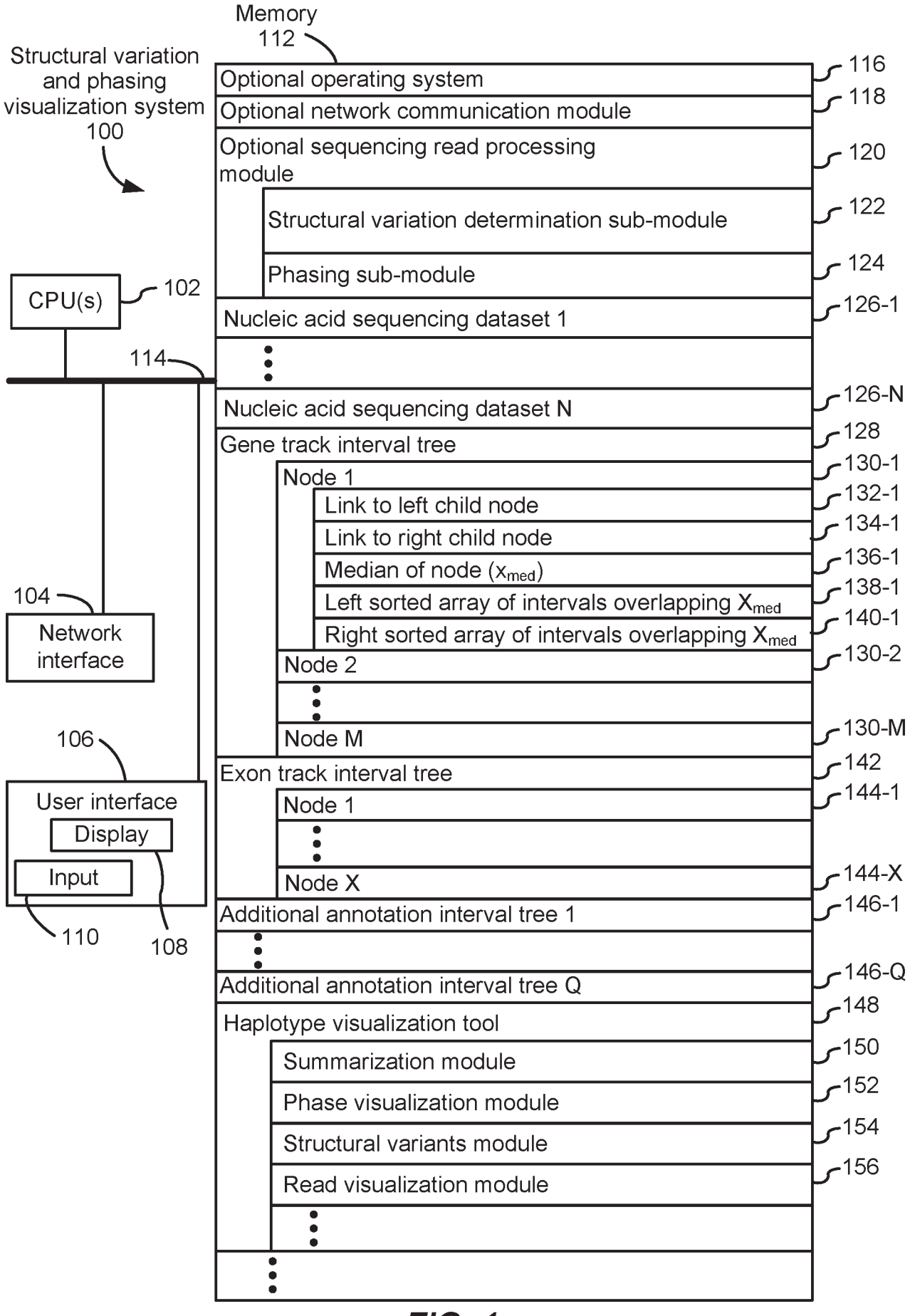

Structural variation and phasing visualization system 100
CPU(s)
102
114
104
Network interface
106
User interface
Display
Input
110
108
Memory 112
Optional operating system
116
Optional network communication module
118
Optional sequencing read processing module
120
Structural variation determination sub-module
122
Phasing sub-module
124
Nucleic acid sequencing dataset 1
126-1
Nucleic acid sequencing dataset N
126-N
Gene track interval tree
128
Node 1
130-1
Link to left child node
132-1
Link to right child node
134-1
Median of node (x_{med})
136-1
Left sorted array of intervals overlapping X_{med}
138-1
Right sorted array of intervals overlapping X_{med}
140-1
Node 2
130-2
Node M
130-M
Exon track interval tree
142
Node 1
144-1
Node X
144-X
Additional annotation interval tree 1
146-1
Additional annotation interval tree Q
146-Q
Haplotype visualization tool
148
Summarization module
150
Phase visualization module
152
Structural variants module
154
Read visualization module
156

FIG. 1

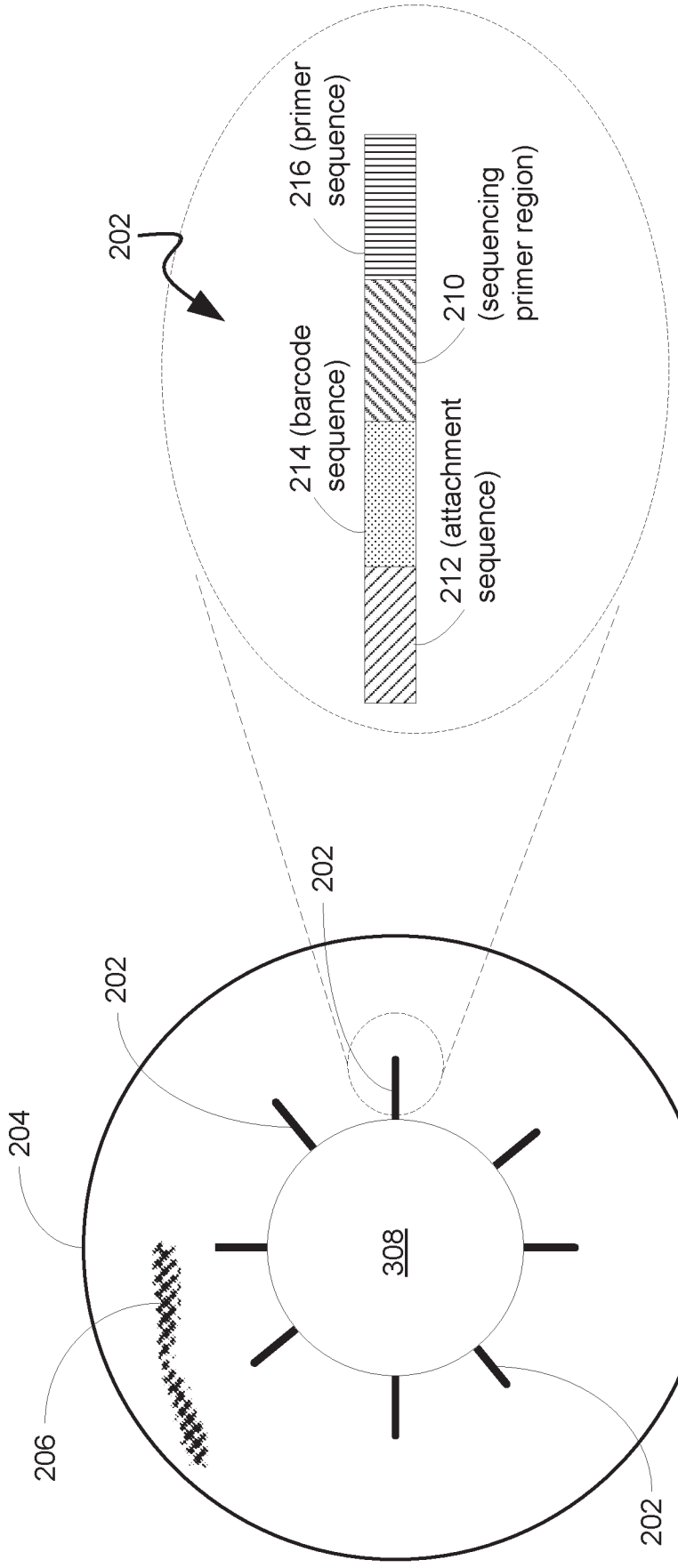

202
216 (primer sequence)
214 (barcode sequence)
210 (sequencing primer region)
212 (attachment sequence)
(B)
202
202
204
308
206
202
(A)

FIG. 2

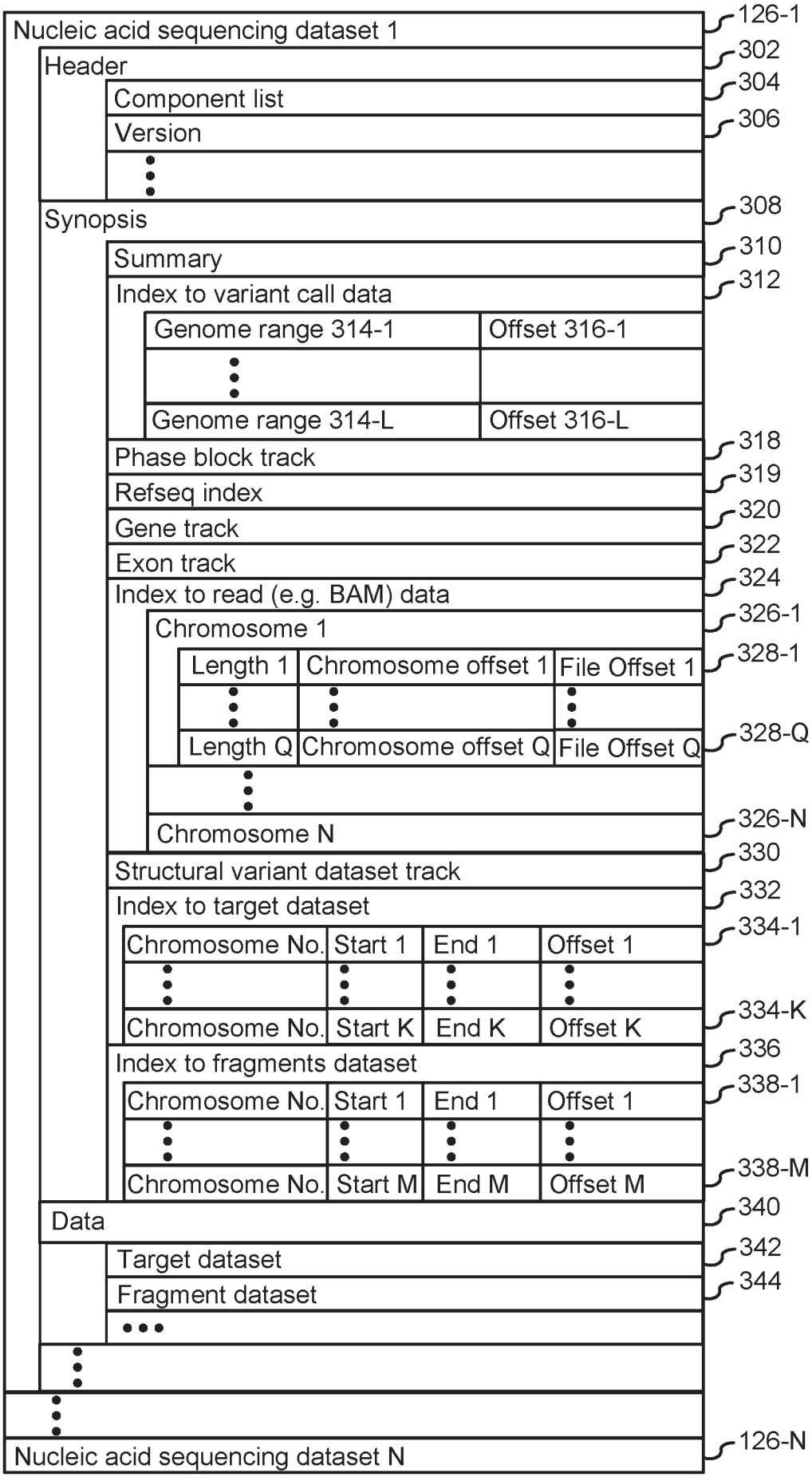

Nucleic acid sequencing dataset 1
126-1
Header
302
Component list
304
Version
306
Synopsis
308
Summary
310
Index to variant call data
312
Genome range 314-1
Offset 316-1
Genome range 314-L
Offset 316-L
Phase block track
318
Refseq index
319
Gene track
320
Exon track
322
Index to read (e.g. BAM) data
324
Chromosome 1
326-1
Length 1
Chromosome offset 1
File Offset 1
328-1
Length Q
Chromosome offset Q
File Offset Q
328-Q
Chromosome N
326-N
Structural variant dataset track
330
Index to target dataset
332
Chromosome No.
Start 1
End 1
Offset 1
334-1
Chromosome No.
Start K
End K
Offset K
334-K
Index to fragments dataset
336
Chromosome No.
Start 1
End 1
Offset 1
338-1
Chromosome No.
Start M
End M
Offset M
338-M
Data
340
Target dataset
342
Fragment dataset
344
Nucleic acid sequencing dataset N
126-N

FIG. 3

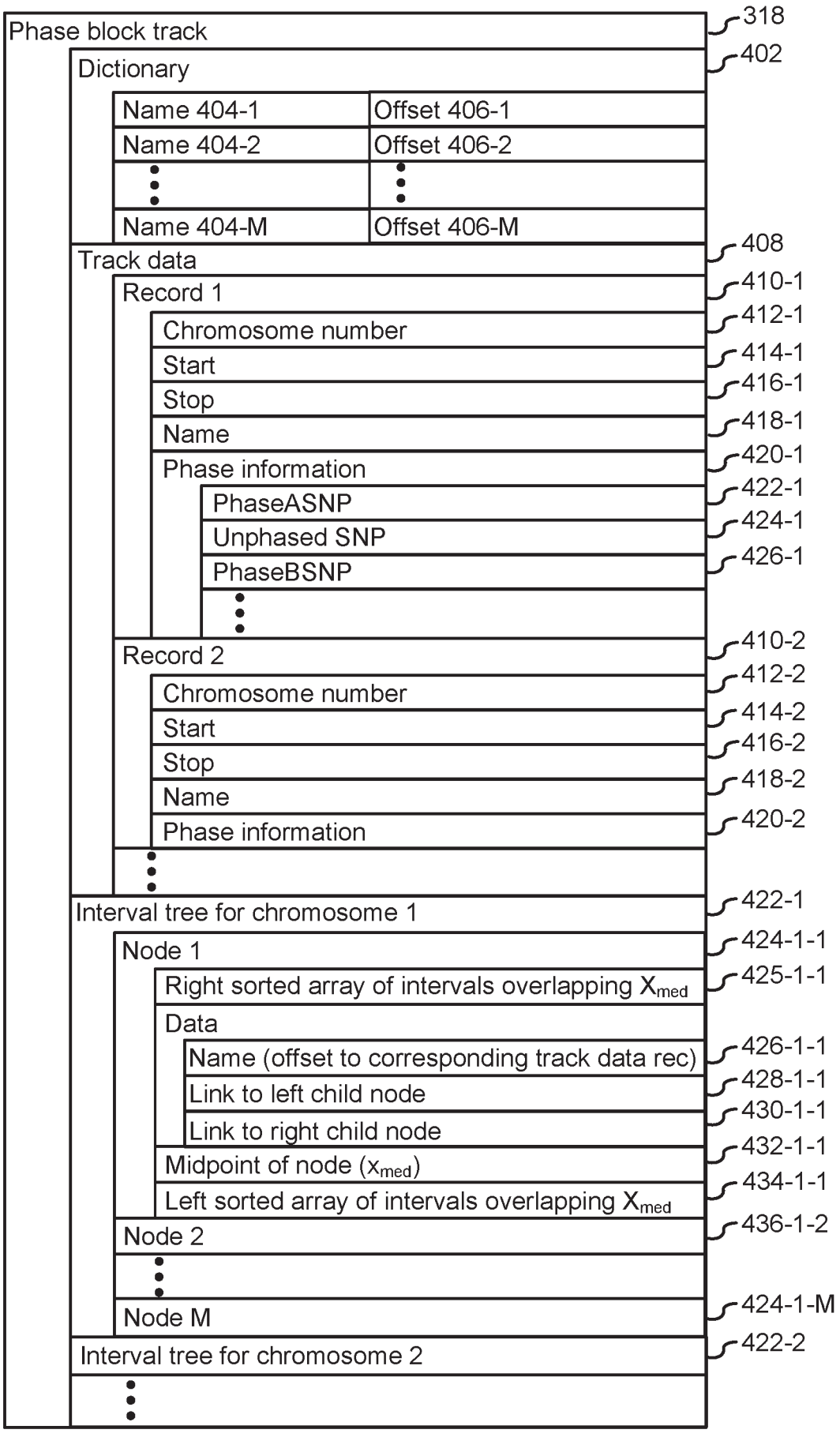

Phase block track
318
Dictionary
402
Name 404-1
Offset 406-1
Name 404-2
Offset 406-2
Name 404-M
Offset 406-M
Track data
408
Record 1
410-1
Chromosome number
412-1
Start
414-1
Stop
416-1
Name
418-1
Phase information
420-1
PhaseASNP
422-1
Unphased SNP
424-1
PhaseBSNP
426-1
Record 2
410-2
Chromosome number
412-2
Start
414-2
Stop
416-2
Name
418-2
Phase information
420-2
Interval tree for chromosome 1
422-1
Node 1
424-1-1
Right sorted array of intervals overlapping X_{med}
425-1-1
Data
Name (offset to corresponding track data rec)
426-1-1
Link to left child node
428-1-1
Link to right child node
430-1-1
Midpoint of node (x_{med})
432-1-1
Left sorted array of intervals overlapping X_{med}
434-1-1
Node 2
436-1-2
Node M
424-1-M
Interval tree for chromosome 2
422-2

FIG. 4

318

```
{
        "Name" : "phase_data",                          (402)
        "TrackData" : [                                 (408)
            {                                           (410-1)
                "Info" : {
                        "PhaseASNP" : 0,
                        "UnPhasedSNP" : 0,
                        "PhaseBSNP" : 5
                },
                "Name" : "3",
                "Stop" : 35,
                "Chromosome" : "chr1",
                "Start" : 30
            },
            {                                           (410-2)
                "Start" : 37,
                "Info" : {
                        "UnPhasedSNP" : 0,
                        "PhaseBSNP" : 3,
                        "PhaseASNP" : 0
                 },
                "Stop" : 40,
                "Chromosome" :  "chr1",
                "Name" : "6"
            }
],
"Itree" : {
        "chrl" : {                                      422-1
                "RightTree" : null,
                "MidPoint" : 38,
                "LeftTree" : {
                        "RightTree" : null,
                        "MidPoint" : 32 ,
                        "LeftTree" : null,
                        "Data Here" : [
                            {
                                "Left" : 30,
                                "Right" : 35,
                                "Name" : "0"
                            },
                        ]
        },
        "DataHere" : [
                {
                        "Right" : 40,
                        "Left":  37,
                        "Name" : "1"
                }
        ]
},
"Dictionary" {
        "3" :  0,
        "6" : 1
}
```

FIG. 5

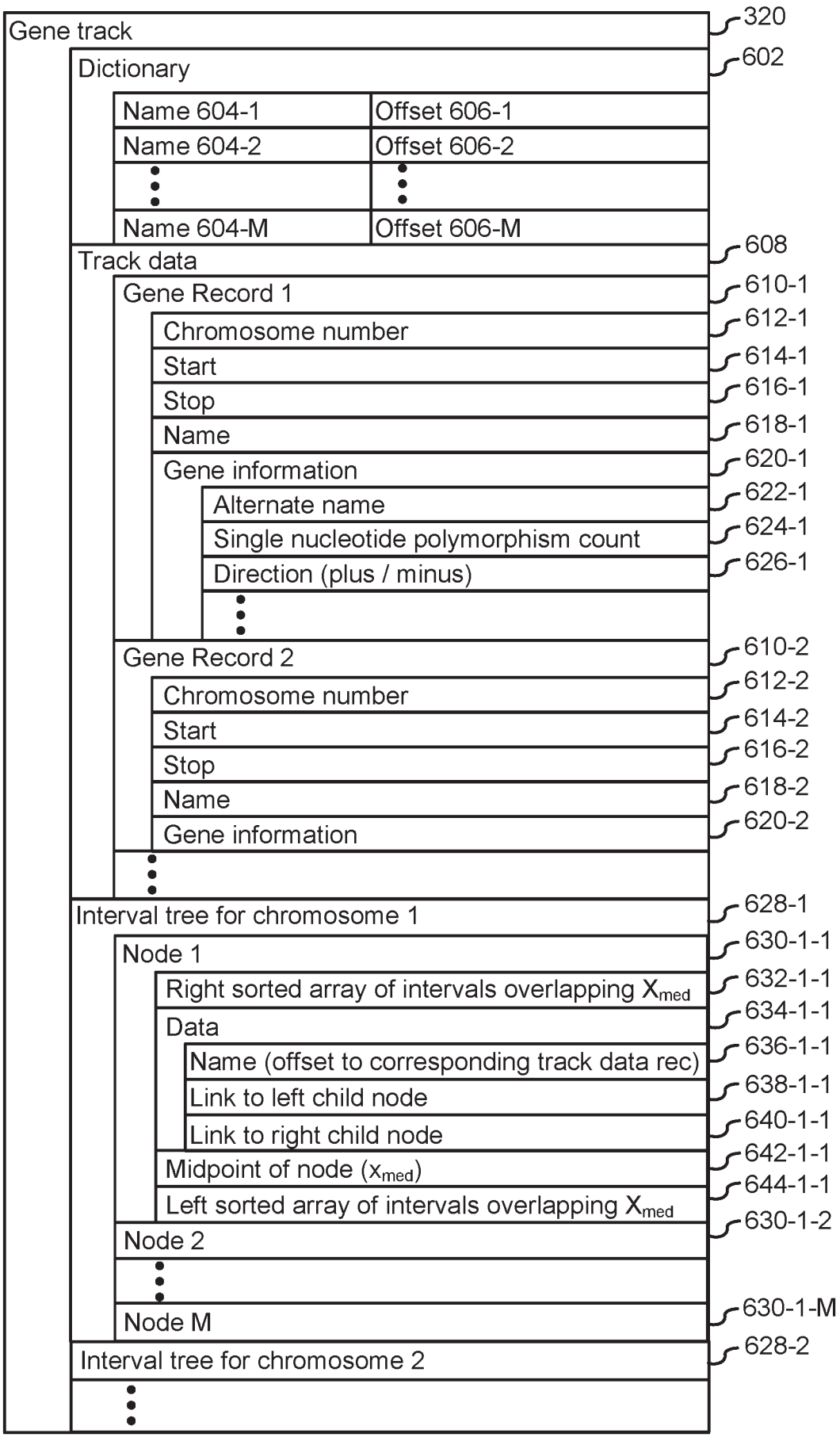

Gene track
320
Dictionary
602
Name 604-1
Offset 606-1
Name 604-2
Offset 606-2
Name 604-M
Offset 606-M
Track data
608
Gene Record 1
610-1
Chromosome number
612-1
Start
614-1
Stop
616-1
Name
618-1
Gene information
620-1
Alternate name
622-1
Single nucleotide polymorphism count
624-1
Direction (plus / minus)
626-1
Gene Record 2
610-2
Chromosome number
612-2
Start
614-2
Stop
616-2
Name
618-2
Gene information
620-2
Interval tree for chromosome 1
628-1
Node 1
630-1-1
Right sorted array of intervals overlapping X_{med}
632-1-1
Data
634-1-1
Name (offset to corresponding track data rec)
636-1-1
Link to left child node
638-1-1
Link to right child node
640-1-1
Midpoint of node (x_{med})
642-1-1
Left sorted array of intervals overlapping X_{med}
644-1-1
Node 2
630-1-2
Node M
630-1-M
Interval tree for chromosome 2
628-2

FIG. 6

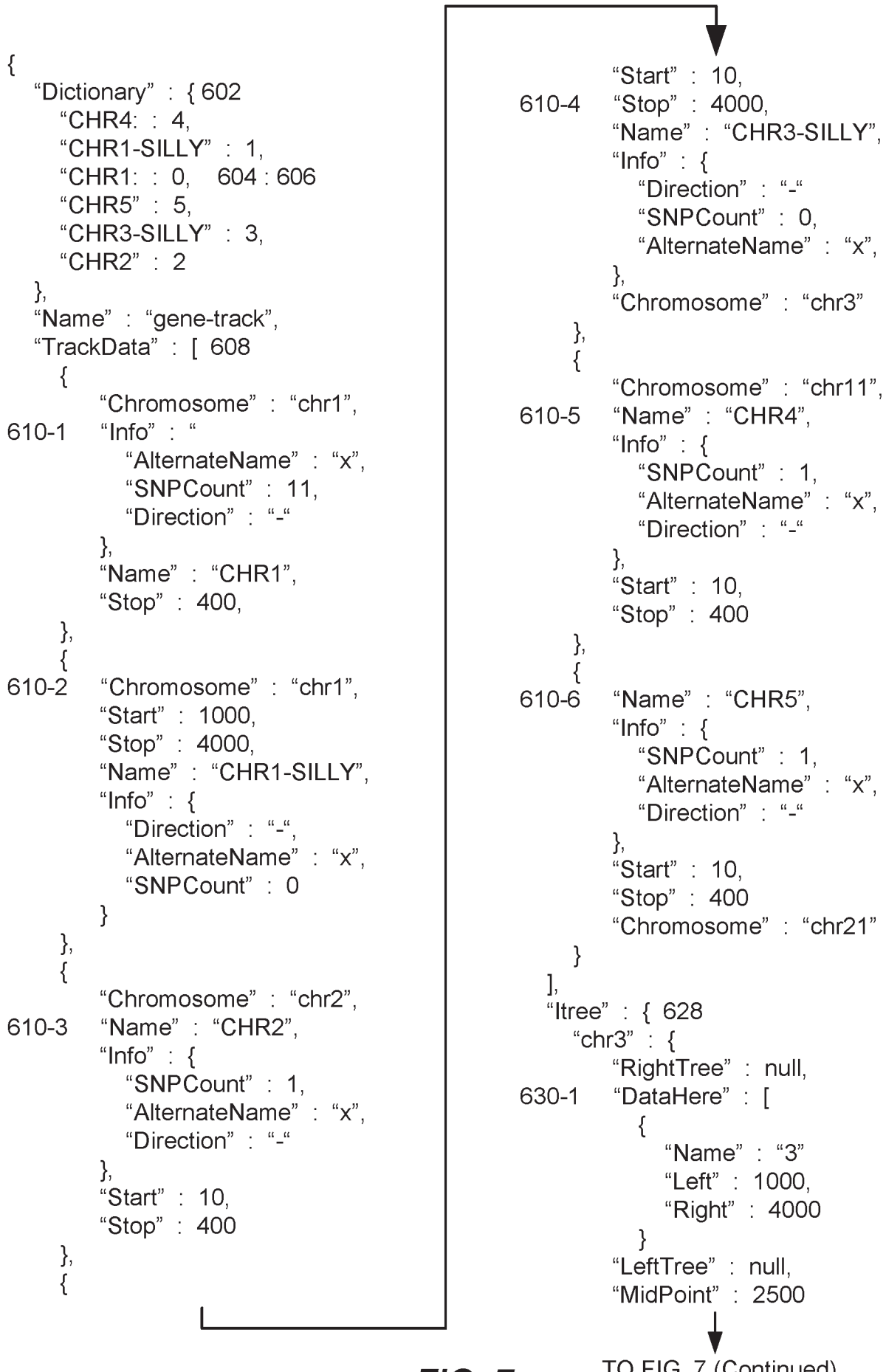
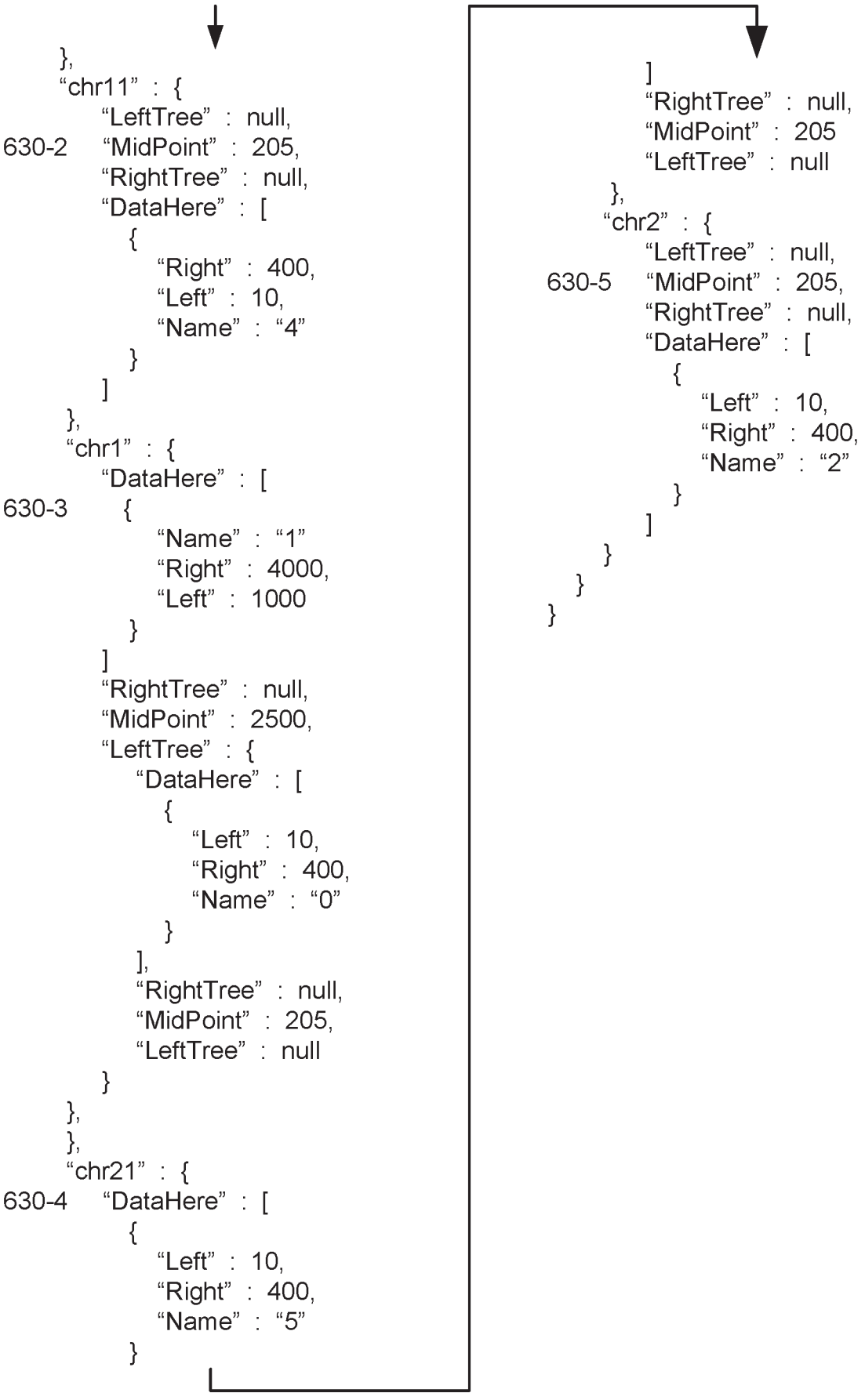

{
"Dictionary" : { 602
"CHR4: : 4,
"CHR1-SILLY" : 1,
"CHR1: : 0, 604 : 606
"CHR5" : 5,
"CHR3-SILLY" : 3,
"CHR2" : 2
},
"Name" : "gene-track",
"TrackData" : [608
{
"Chromosome" : "chr1",
610-1 "Info" : "
"AlternateName" : "x",
"SNPCount" : 11,
"Direction" : "-"
},
"Name" : "CHR1",
"Stop" : 400,
},
{
610-2 "Chromosome" : "chr1",
"Start" : 1000,
"Stop" : 4000,
"Name" : "CHR1-SILLY",
"Info" : {
"Direction" : "-",
"AlternateName" : "x",
"SNPCount" : 0
}
},
{
"Chromosome" : "chr2",
610-3 "Name" : "CHR2",
"Info" : {
"SNPCount" : 1,
"AlternateName" : "x",
"Direction" : "-"
},
"Start" : 10,
"Stop" : 400
},
{
"Start" : 10,
610-4 "Stop" : 4000,
"Name" : "CHR3-SILLY",
"Info" : {
"Direction" : "-"
"SNPCount" : 0,
"AlternateName" : "x",
},
"Chromosome" : "chr3"
},
{
"Chromosome" : "chr11",
610-5 "Name" : "CHR4",
"Info" : {
"SNPCount" : 1,
"AlternateName" : "x",
"Direction" : "-"
},
"Start" : 10,
"Stop" : 400
},
{
610-6 "Name" : "CHR5",
"Info" : {
"SNPCount" : 1,
"AlternateName" : "x",
"Direction" : "-"
},
"Start" : 10,
"Stop" : 400
"Chromosome" : "chr21"
}
],
"Itree" : { 628
"chr3" : {
"RightTree" : null,
630-1 "DataHere" : [
{
"Name" : "3"
"Left" : 1000,
"Right" : 4000
}
"LeftTree" : null,
"MidPoint" : 2500
TO FIG. 7 (Continued)

FIG. 7

FROM FIG. 7



```
},
"chr11" : {
    "LeftTree" : null,
630-2   "MidPoint" : 205,
    "RightTree" : null,
    "DataHere" : [
      {
        "Right" : 400,
        "Left" : 10,
        "Name" : "4"
      }
    ]
},
"chr1" : {
    "DataHere" : [
630-3   {
        "Name" : "1"
        "Right" : 4000,
        "Left" : 1000
      }
    ]
    "RightTree" : null,
    "MidPoint" : 2500,
    "LeftTree" : {
      "DataHere" : [
        {
          "Left" : 10,
          "Right" : 400,
          "Name" : "0"
        }
      ],
      "RightTree" : null,
      "MidPoint" : 205,
      "LeftTree" : null
    }
},
},
"chr21" : {
630-4   "DataHere" : [
      {
        "Left" : 10,
        "Right" : 400,
        "Name" : "5"
      }
    ]
    "RightTree" : null,
    "MidPoint" : 205
    "LeftTree" : null
},
"chr2" : {
    "LeftTree" : null,
630-5   "MidPoint" : 205,
    "RightTree" : null,
    "DataHere" : [
      {
        "Left" : 10,
        "Right" : 400,
        "Name" : "2"
      }
    ]
  }
}
```

FIG. 7
(Continued)

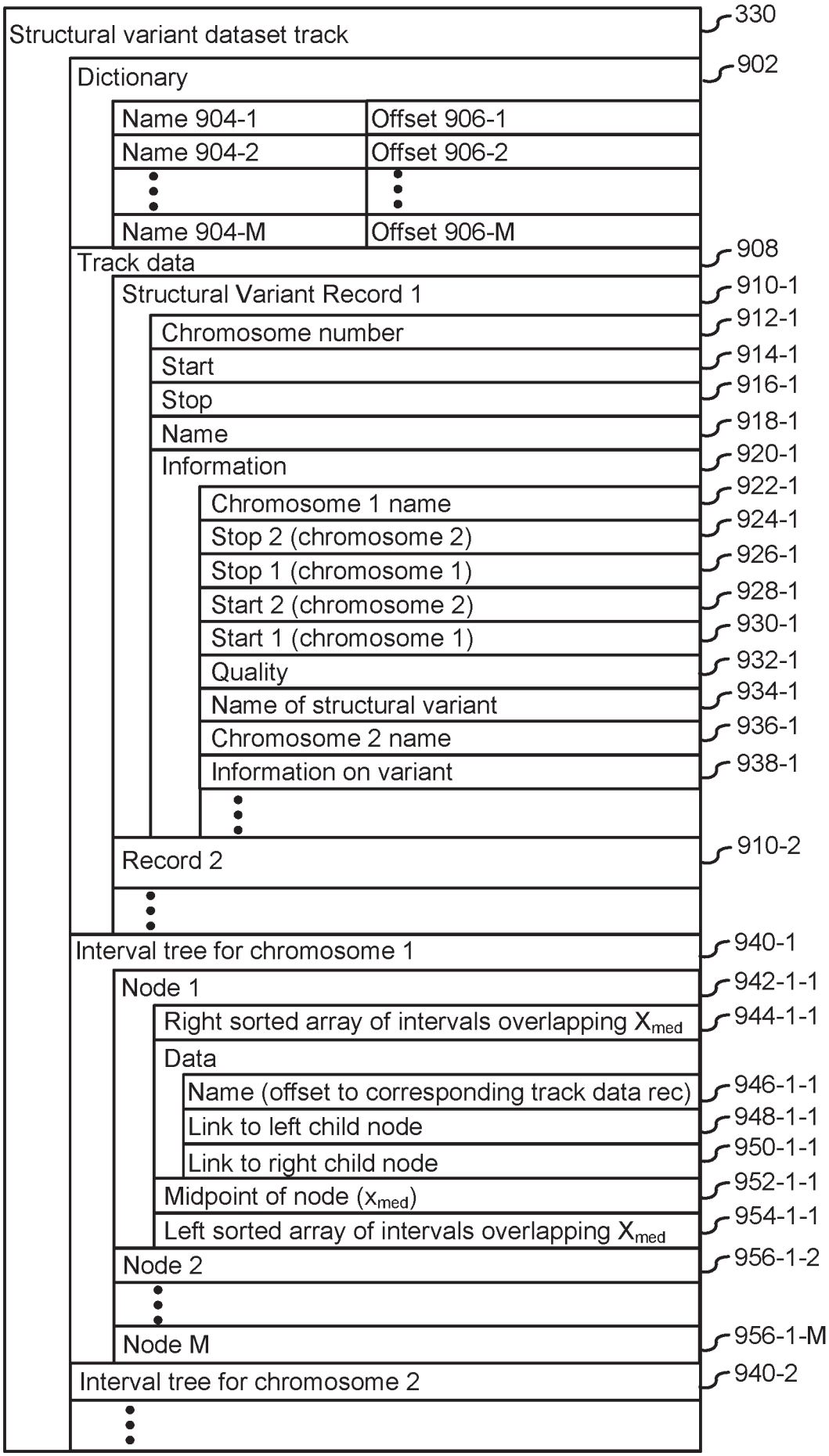

Structural variant dataset track
330
Dictionary
902
Name 904-1
Offset 906-1
Name 904-2
Offset 906-2
Name 904-M
Offset 906-M
Track data
908
Structural Variant Record 1
910-1
Chromosome number
912-1
Start
914-1
Stop
916-1
Name
918-1
Information
920-1
Chromosome 1 name
922-1
Stop 2 (chromosome 2)
924-1
Stop 1 (chromosome 1)
926-1
Start 2 (chromosome 2)
928-1
Start 1 (chromosome 1)
930-1
Quality
932-1
Name of structural variant
934-1
Chromosome 2 name
936-1
Information on variant
938-1
Record 2
910-2
Interval tree for chromosome 1
940-1
Node 1
942-1-1
Right sorted array of intervals overlapping X_{med}
944-1-1
Data
Name (offset to corresponding track data rec)
946-1-1
Link to left child node
948-1-1
Link to right child node
950-1-1
Midpoint of node (x_{med})
952-1-1
Left sorted array of intervals overlapping X_{med}
954-1-1
Node 2
956-1-2
Node M
956-1-M
Interval tree for chromosome 2
940-2

FIG. 8

```
810-1 {
  812  “Chromosome” : “chr16”,
  816  “Stop” : 71320834,
       “Name” : “BRK_chr16_71316475_chr5_115237536”,
  820  “Info” : {
          “Chromosomel” : “chr16”
          “Stop2” : 115250206,
          “Stop1” : 71320834,
          “Start2” : 115237536,
          “Start1” : 71316475,
          “Quality” : 73,
          “Name” : “BRK_chr16_71316475_chr5_115237536”,
          “Chromosome2” : “chr5”,
          “Info” : {
             “SEG_DUP” : “.”,
             “NMATES1 : “2”,
             “NSPLIT” : “0”,
             “BLACK_FRAC” : “nan”,
             “BCOV” : “17”,
             “BLACK_DIST2” : “10366.0”,
             “TYPE” : “UNK”,
             “BLACK_DIST1” : “58024.0”,
             “NBCS1” : “270”,
             “NMATES2” : “1”,
             “MATCHES” : “.”,
             “NPAIRS” : “0”,
             “NBCS2” : “217”,
             “RP_LR” : “0.0”,
             “ORIGINALQ” : “53”
          },
          “Filters” : [ ]
       },
  814  “Start” : 71316475
    },
```


FIG. 9

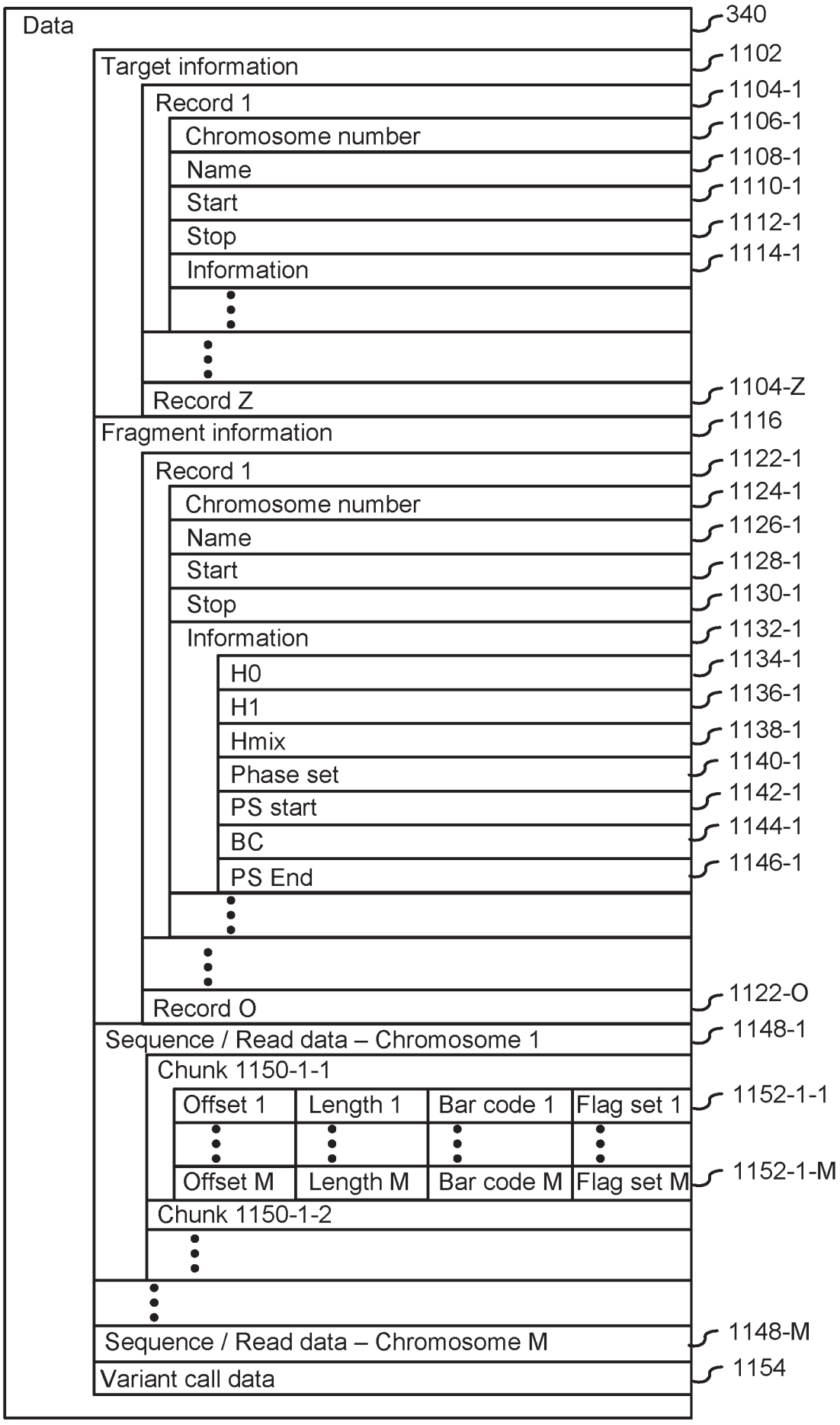

Data
340
Target information
1102
Record 1
1104-1
Chromosome number
1106-1
Name
1108-1
Start
1110-1
Stop
1112-1
Information
1114-1
Record Z
1104-Z
Fragment information
1116
Record 1
1122-1
Chromosome number
1124-1
Name
1126-1
Start
1128-1
Stop
1130-1
Information
1132-1
H0
1134-1
H1
1136-1
Hmix
1138-1
Phase set
1140-1
PS start
1142-1
BC
1144-1
PS End
1146-1
Record O
1122-O
Sequence / Read data – Chromosome 1
1148-1
Chunk 1150-1-1
Offset 1
Length 1
Bar code 1
Flag set 1
1152-1-1
Offset M
Length M
Bar code M
Flag set M
1152-1-M
Chunk 1150-1-2
Sequence / Read data – Chromosome M
1148-M
Variant call data
1154

FIG. 10

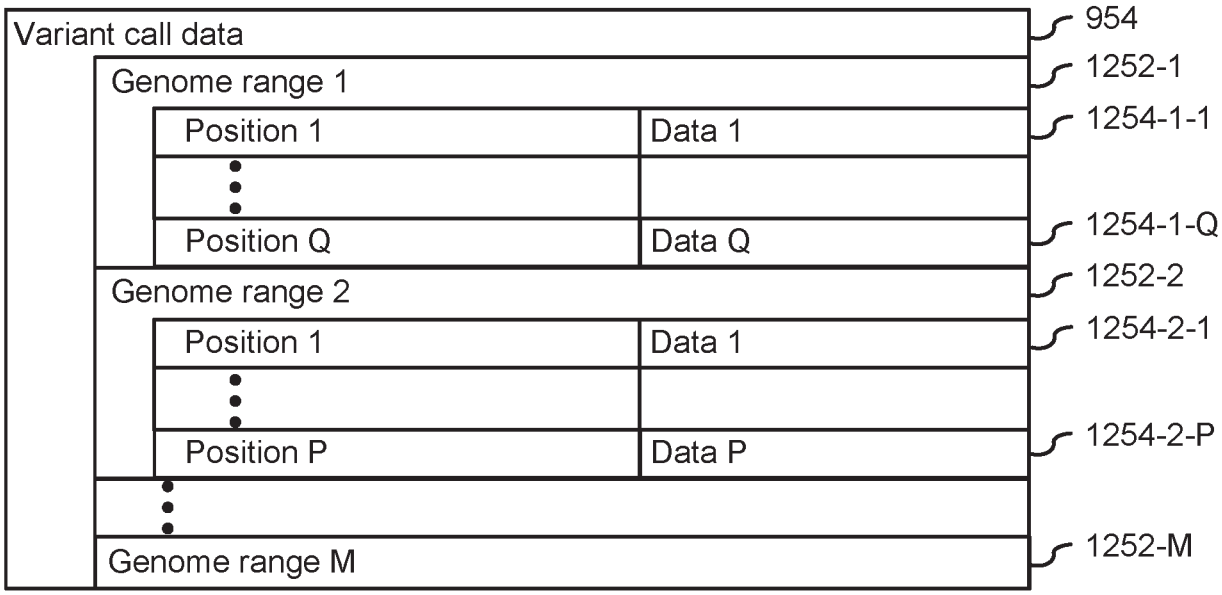

Variant call data
954
Genome range 1
1252-1
Position 1
Data 1
1254-1-1
Position Q
Data Q
1254-1-Q
Genome range 2
1252-2
Position 1
Data 1
1254-2-1
Position P
Data P
1254-2-P
Genome range M
1252-M

FIG. 11

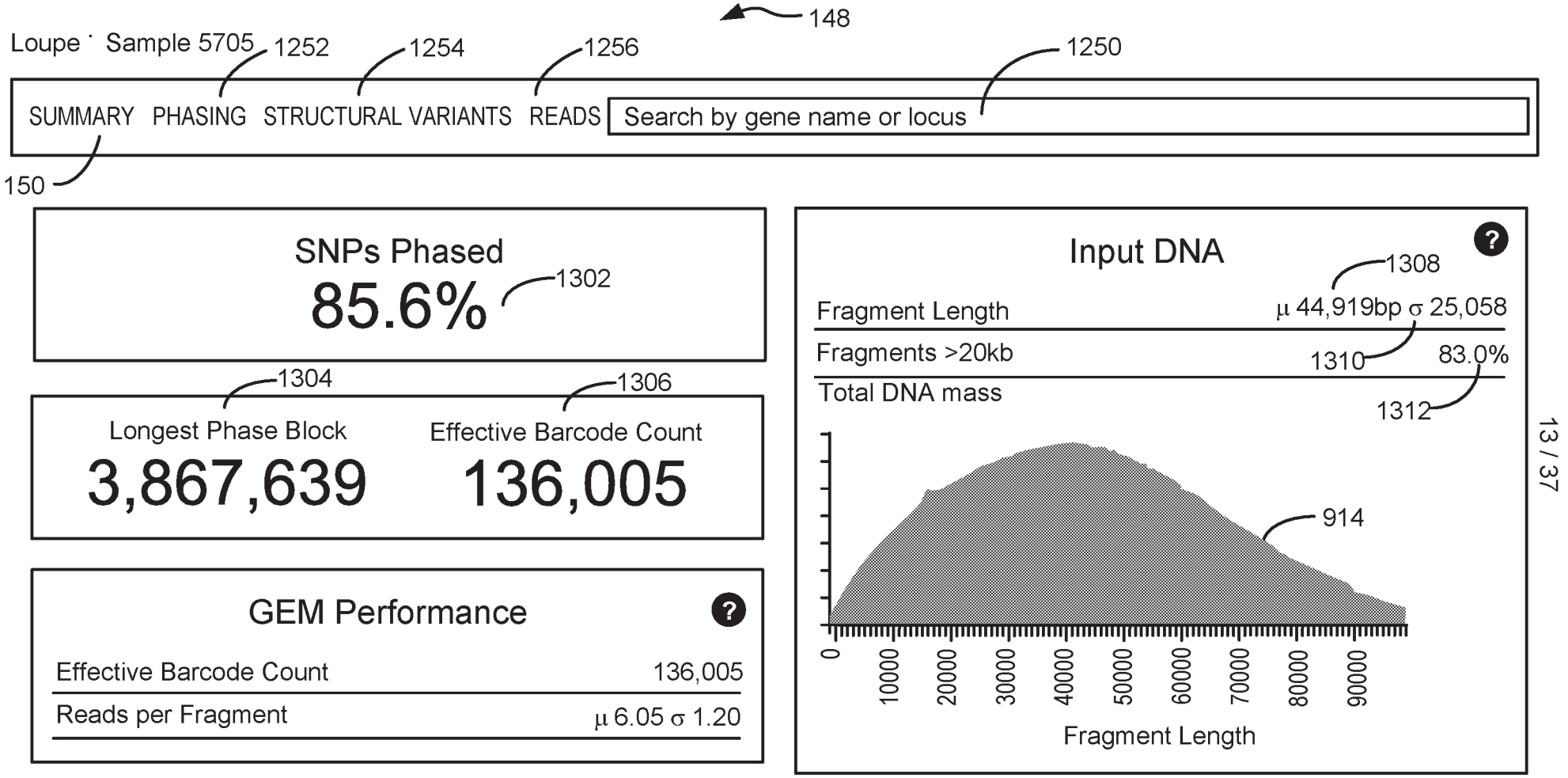

148
Loupe · Sample 5705
1252
1254
1256
1250
SUMMARY
PHASING
STRUCTURAL VARIANTS
READS
Search by gene name or locus
150
SNPs Phased
85.6%
1302
1304
1306
Longest Phase Block
3,867,639
Effective Barcode Count
136,005
GEM Performance
Effective Barcode Count
136,005
Reads per Fragment
μ 6.05 σ 1.20
Input DNA
1308
Fragment Length
μ 44,919bp σ 25,058
Fragments >20kb
1310
83.0%
Total DNA mass
1312
914
0
10000
20000
30000
40000
50000
60000
70000
80000
90000
Fragment Length

FIG. 12

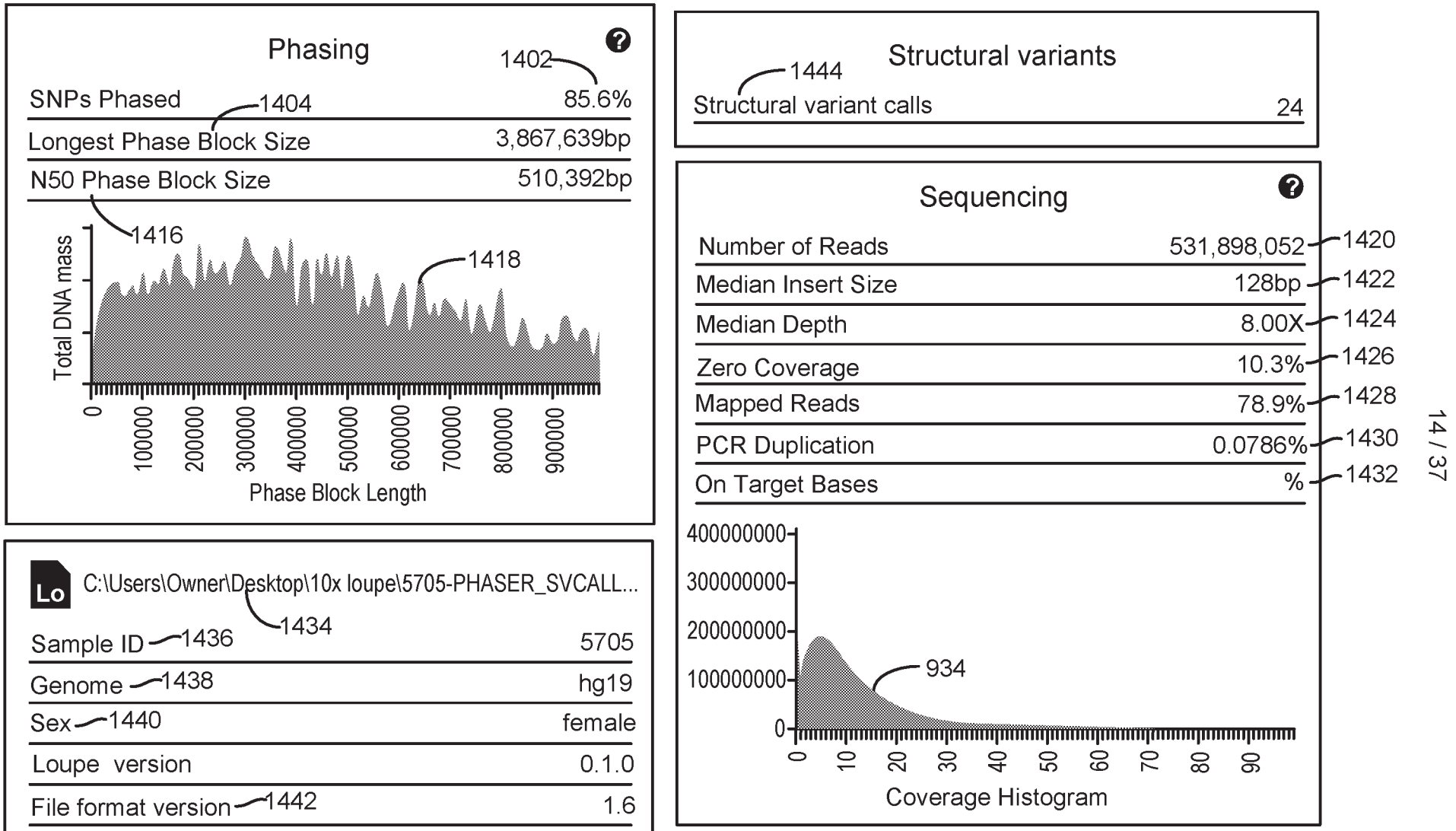


FIG. 12
(Continued)

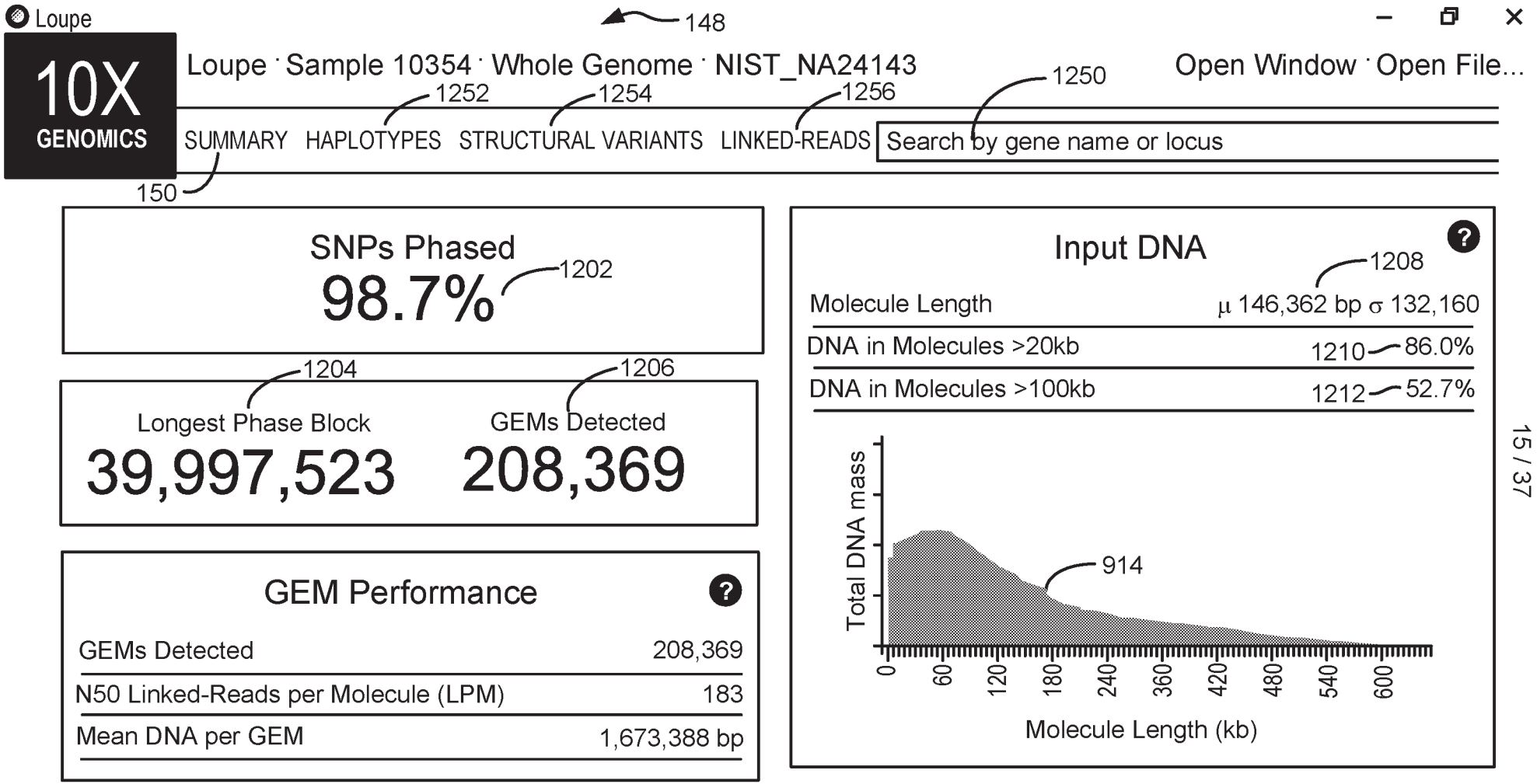

Loupe
148
10X GENOMICS
Loupe · Sample 10354 · Whole Genome · NIST_NA24143
Open Window · Open File...
1250
1252
1254
1256
SUMMARY
HAPLOTYPES
STRUCTURAL VARIANTS
LINKED-READS
Search by gene name or locus
150
SNPs Phased
98.7%
1202
1204
1206
Longest Phase Block
39,997,523
GEMs Detected
208,369
GEM Performance
GEMs Detected 208,369
N50 Linked-Reads per Molecule (LPM) 183
Mean DNA per GEM 1,673,388 bp
Input DNA
1208
Molecule Length μ 146,362 bp σ 132,160
DNA in Molecules >20kb 1210 86.0%
DNA in Molecules >100kb 1212 52.7%
Total DNA mass
914
0
60
120
180
240
360
420
480
540
600
Molecule Length (kb)

FIG. 13

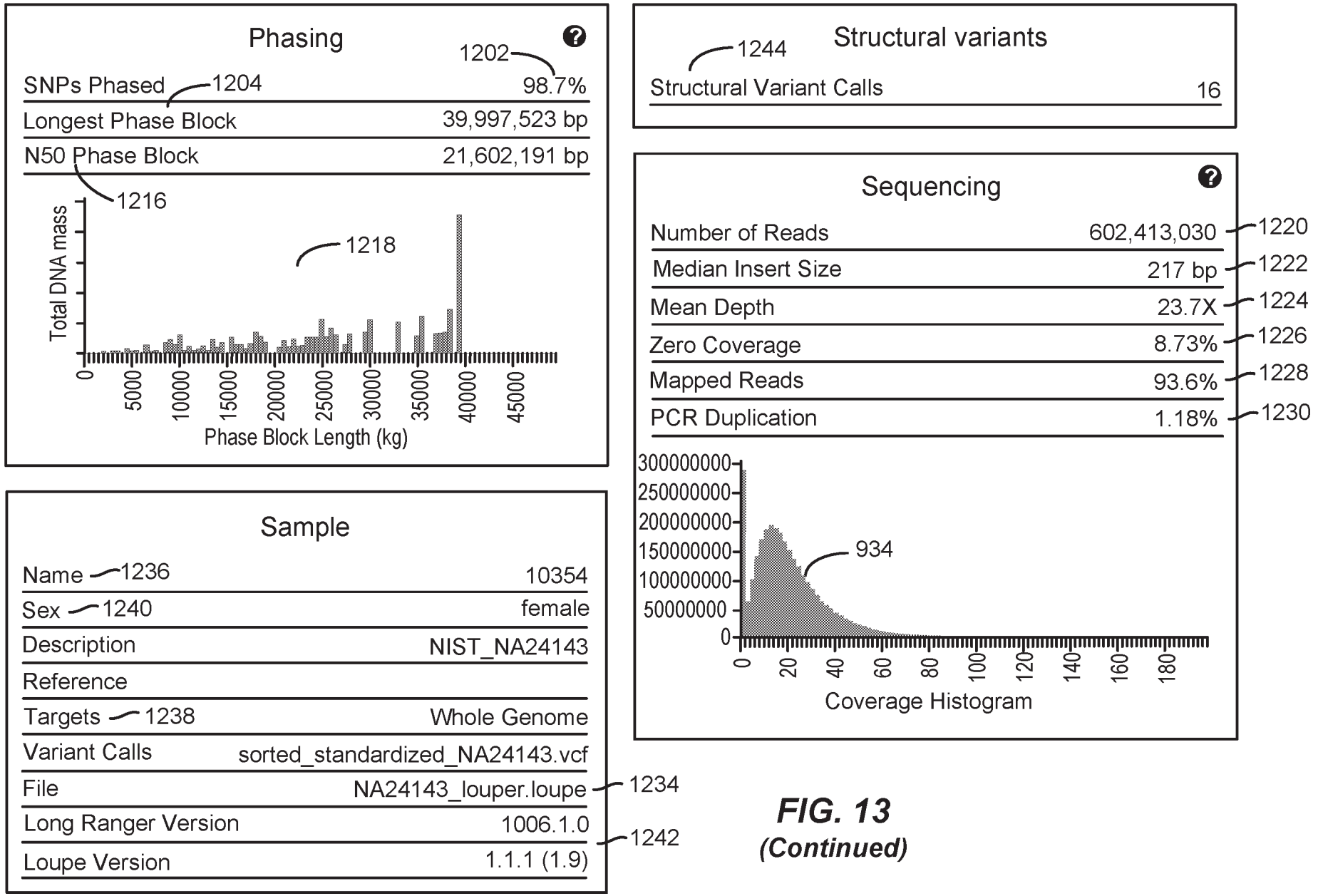


FIG. 13
(Continued)

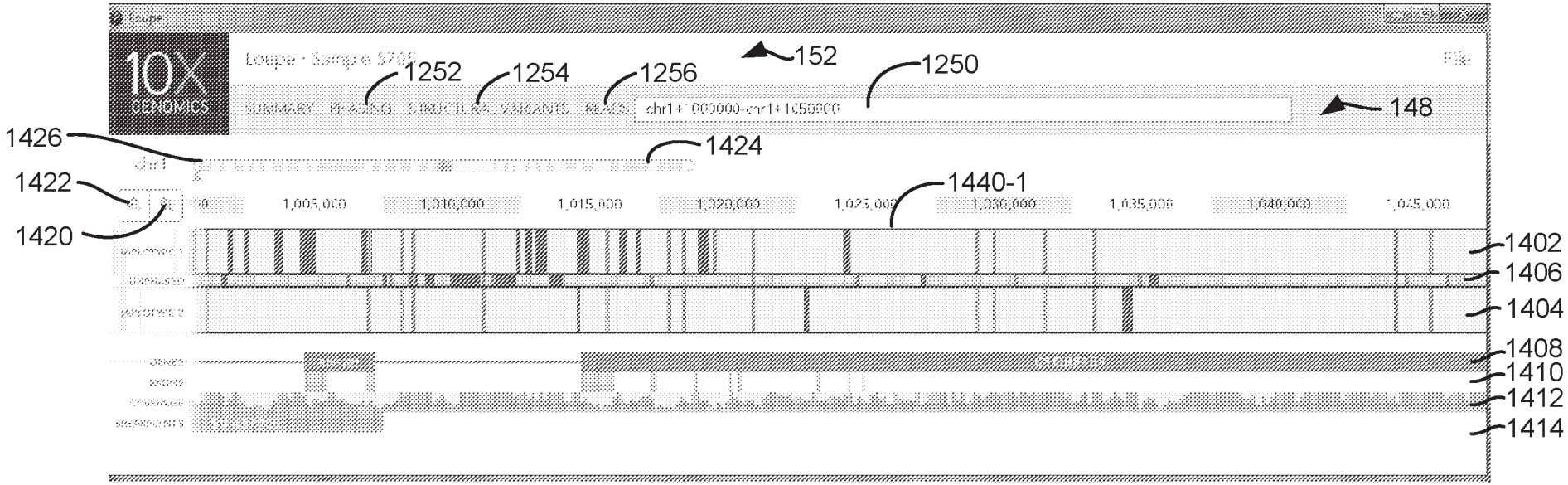


FIG. 14A

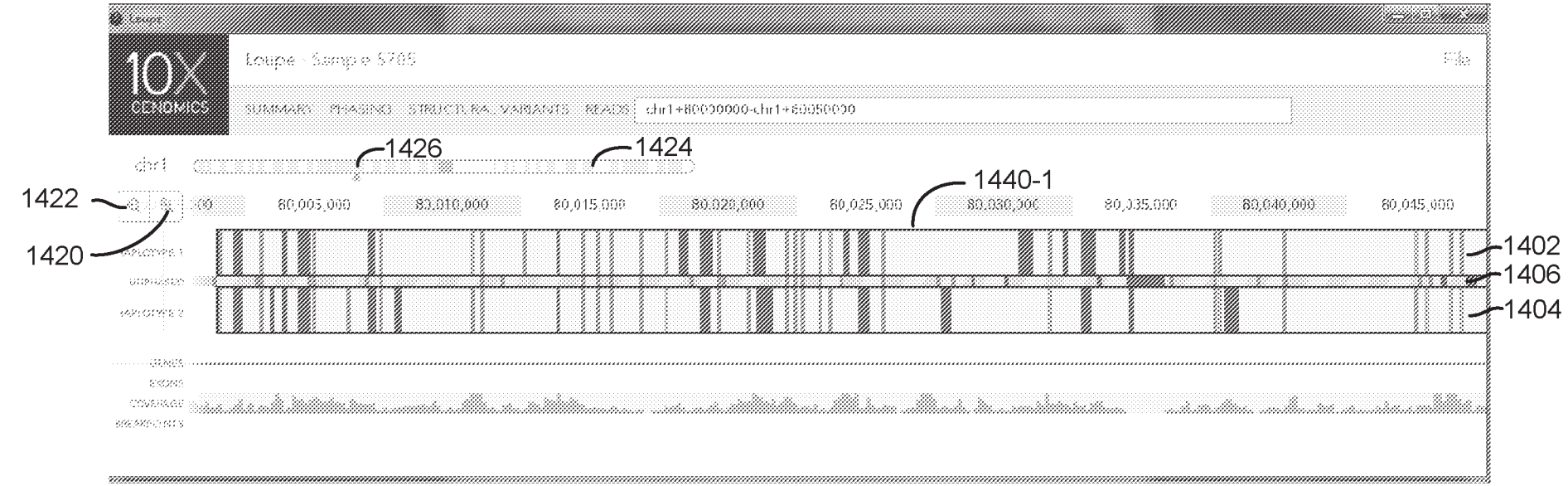


FIG. 14B

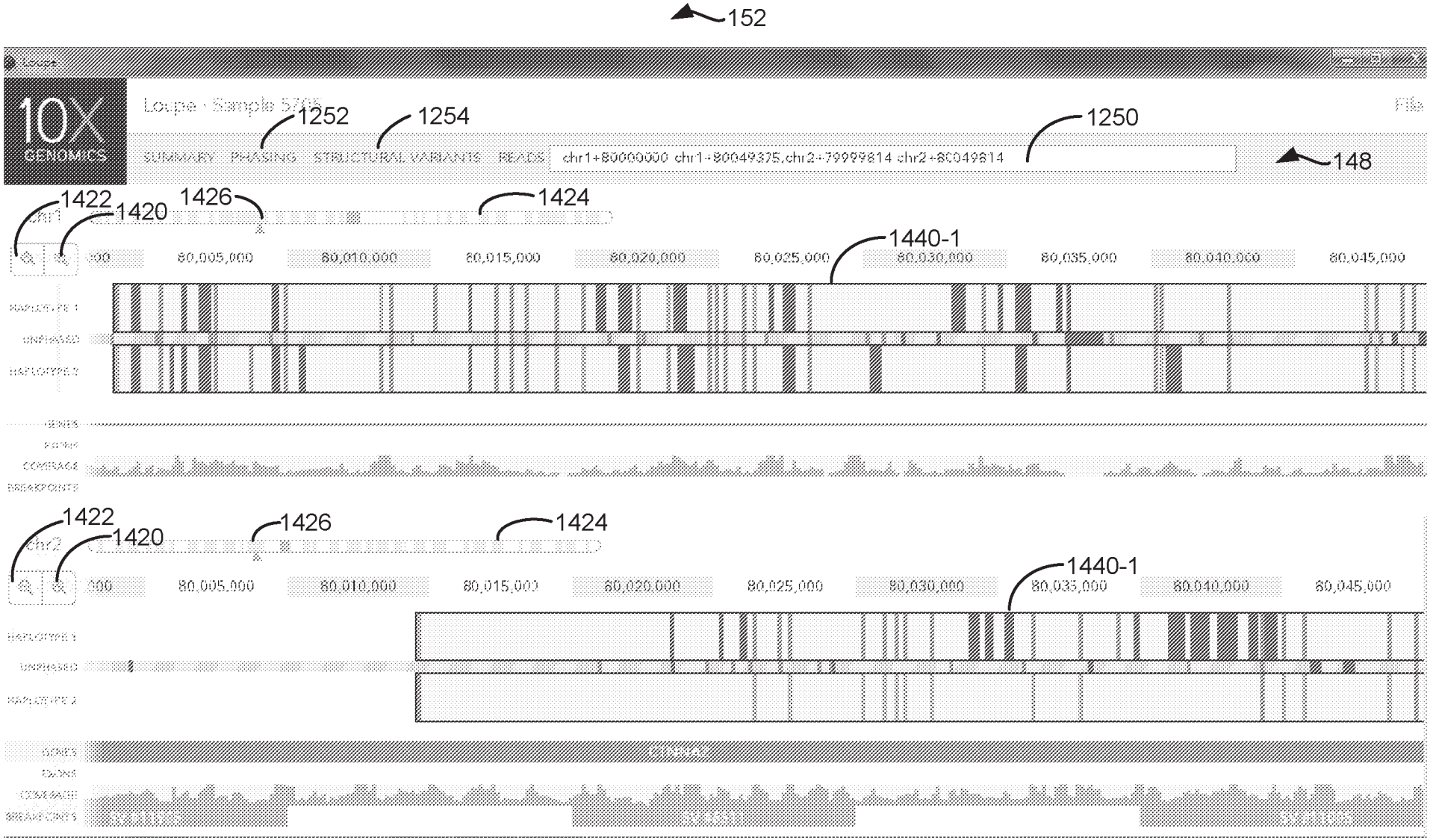

152
10X GENOMICS
Loupe · Sample 57f6
File
SUMMARY
PHASING
STRUCTURAL VARIANTS
READS
chr1+80000000 chr1+80049375,chr2+79999814 chr2+80049814
148
1252
1254
1250
1422
1420
1426
1424
1440-1
chr1
chr2
80,005,000
80,010,000
80,015,000
80,020,000
80,025,000
80,030,000
80,035,000
80,040,000
80,045,000
HAPLOTYPE 1
UNPHASED
HAPLOTYPE 2
GENES
READS
COVERAGE
BREAKPOINTS

FIG. 15

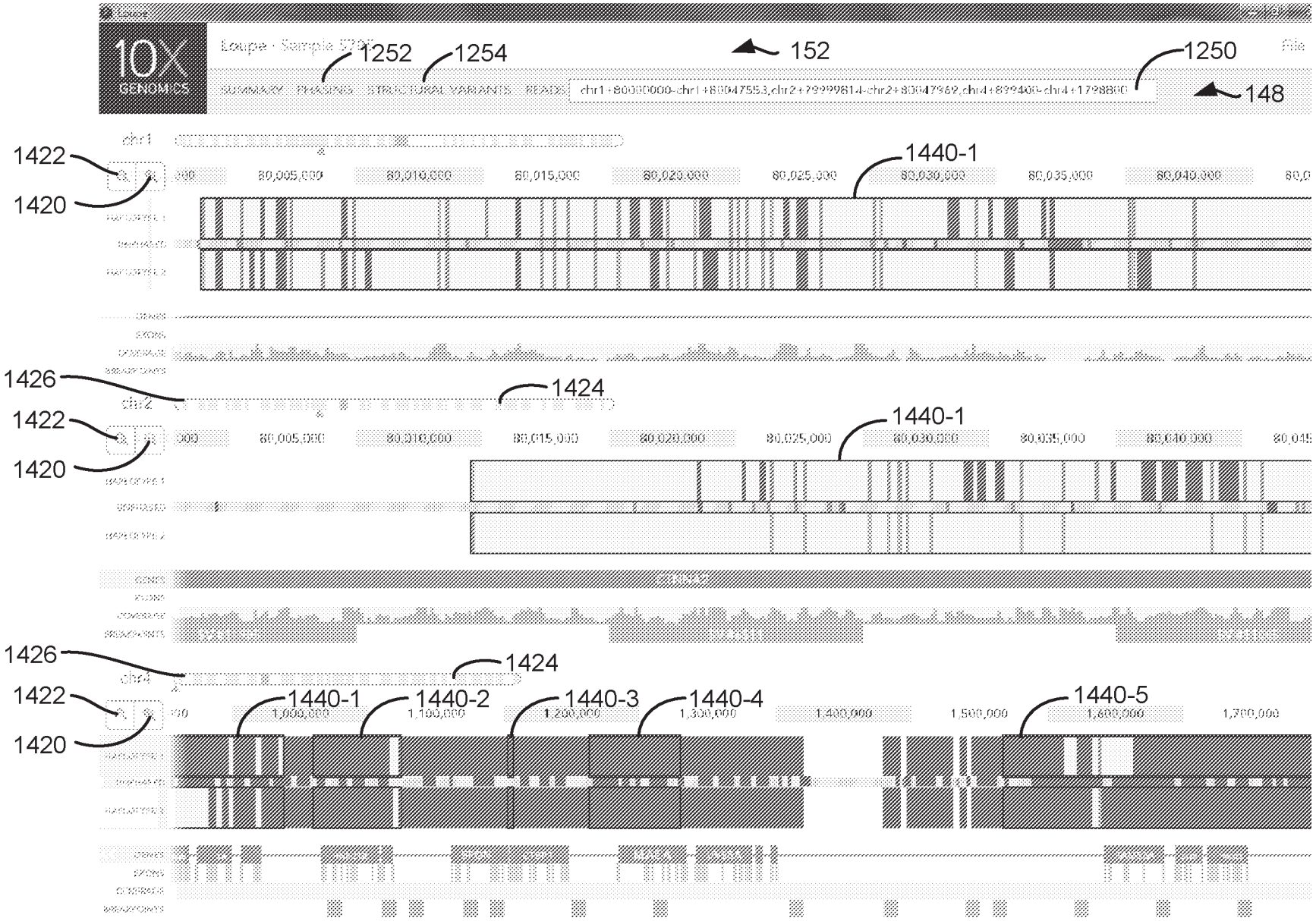

10X GENOMICS
Loupe - Sample 579
SUMMARY
PHASING
STRUCTURAL VARIANTS
READS
chr1:80000000-chr1:80047553,chr2:79999814-chr2:80047962,chr4:839400-chr4:1798800
152
1250
148
1252
1254
1422
1420
1426
1424
1440-1
1440-2
1440-3
1440-4
1440-5
chr1
chr2
chr4

FIG. 16

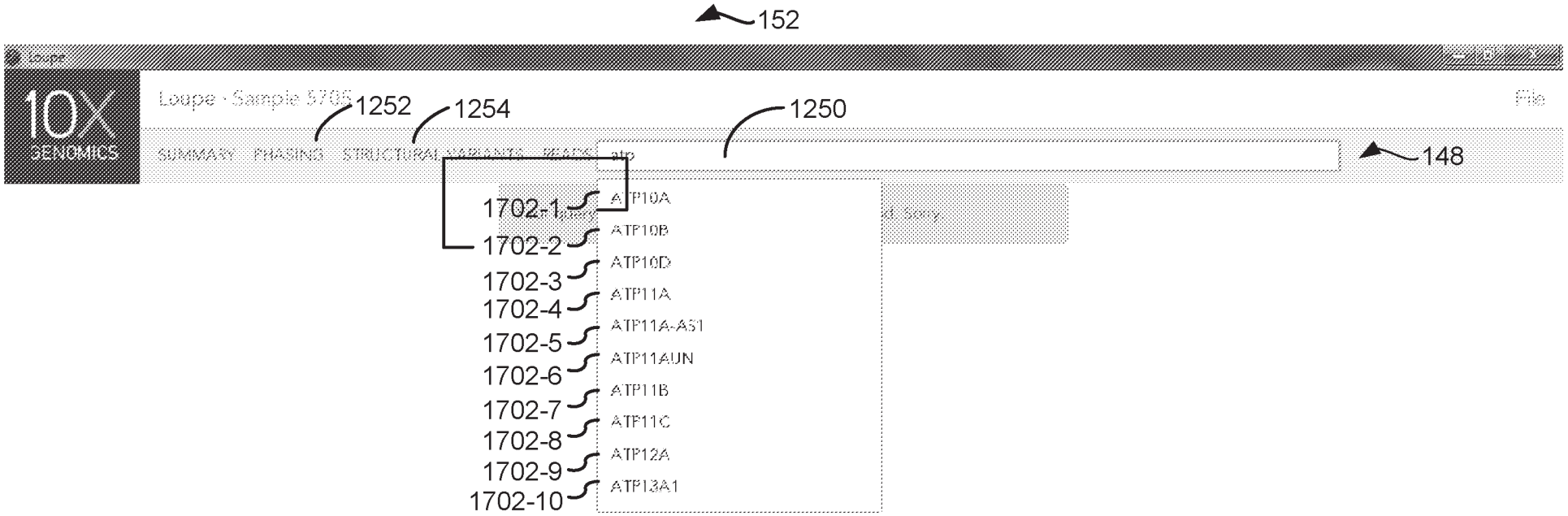

152
10X GENOMICS
Loupe · Sample 5705
File
SUMMARY
PHASING
STRUCTURAL
1252
1254
1250
148
atp
1702-1
1702-2
1702-3
1702-4
1702-5
1702-6
1702-7
1702-8
1702-9
1702-10
ATP10A
ATP10B
ATP10D
ATP11A
ATP11A-AS1
ATP11AUN
ATP11B
ATP11C
ATP12A
ATP13A1

FIG. 17

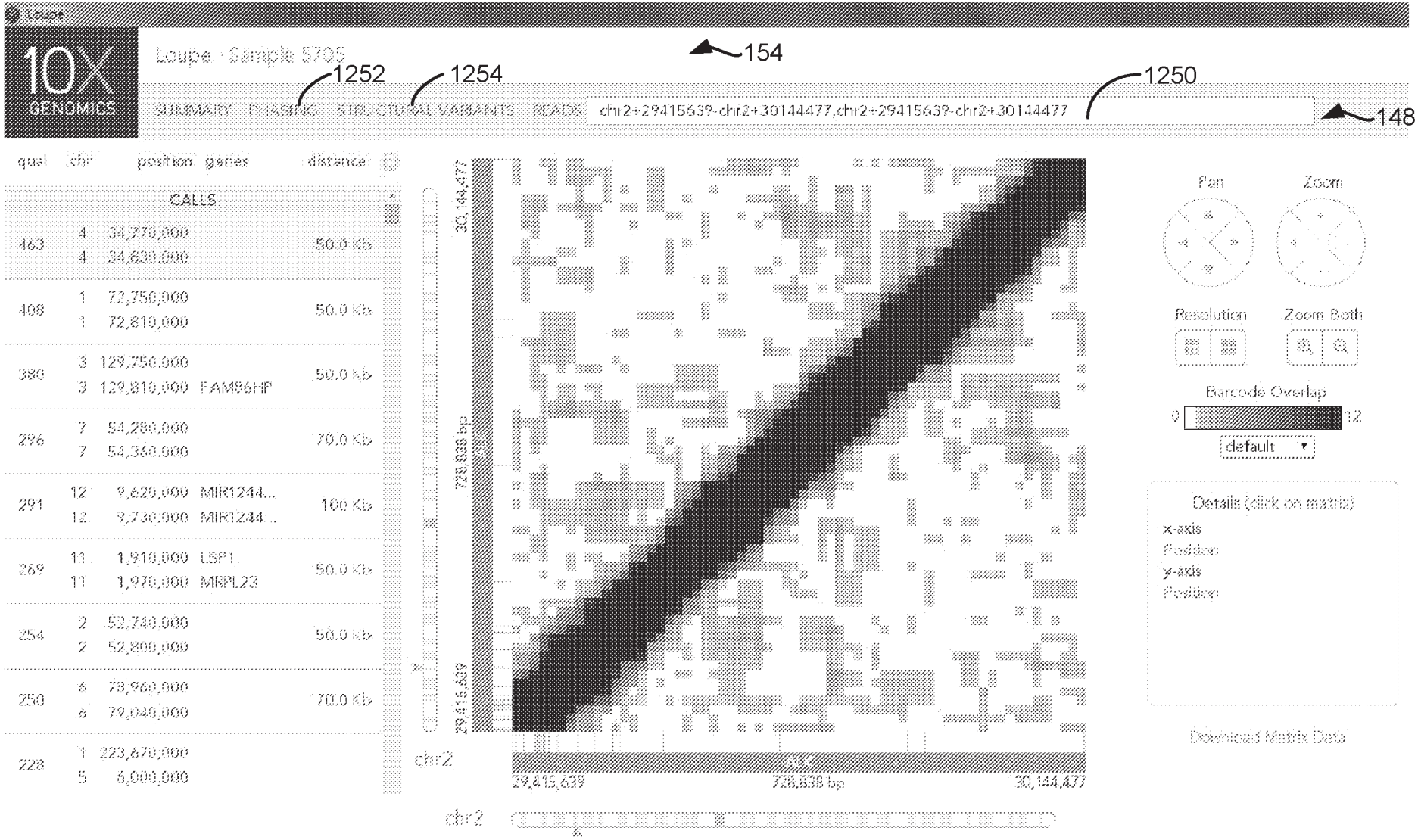

Loupe
10X GENOMICS
Loupe - Sample 5705
154
1252
1254
1250
148
SUMMARY
PHASING
STRUCTURAL VARIANTS
READS
chr2+29415639-chr2+30144477,chr2+29415639-chr2+30144477
qual
chr
position
genes
distance
CALLS
463 4 34,770,000 4 34,630,000 50.0 Kb
408 1 72,750,000 1 72,810,000 50.0 Kb
380 3 129,750,000 3 129,810,000 FAM86HP 50.0 Kb
296 7 54,280,000 7 54,360,000 70.0 Kb
291 12 9,620,000 MIR1244... 12 9,730,000 MIR1244 .. 100 Kb
269 11 1,910,000 LSP1 11 1,970,000 MRPL23 50.0 Kb
254 2 52,740,000 2 52,800,000 50.0 Kb
250 6 78,960,000 6 79,040,000 70.0 Kb
228 1 223,670,000 5 6,000,000
30,144,477
728,838 bp
29,415,639
chr2
chr2
29,415,639
728,838 bp
30,144,477
Pan
Zoom
Resolution
Zoom Both
Barcode Overlap
0
12
default
Details (click on matrix)
x-axis
Position
y-axis
Position
Download Matrix Data

FIG. 18

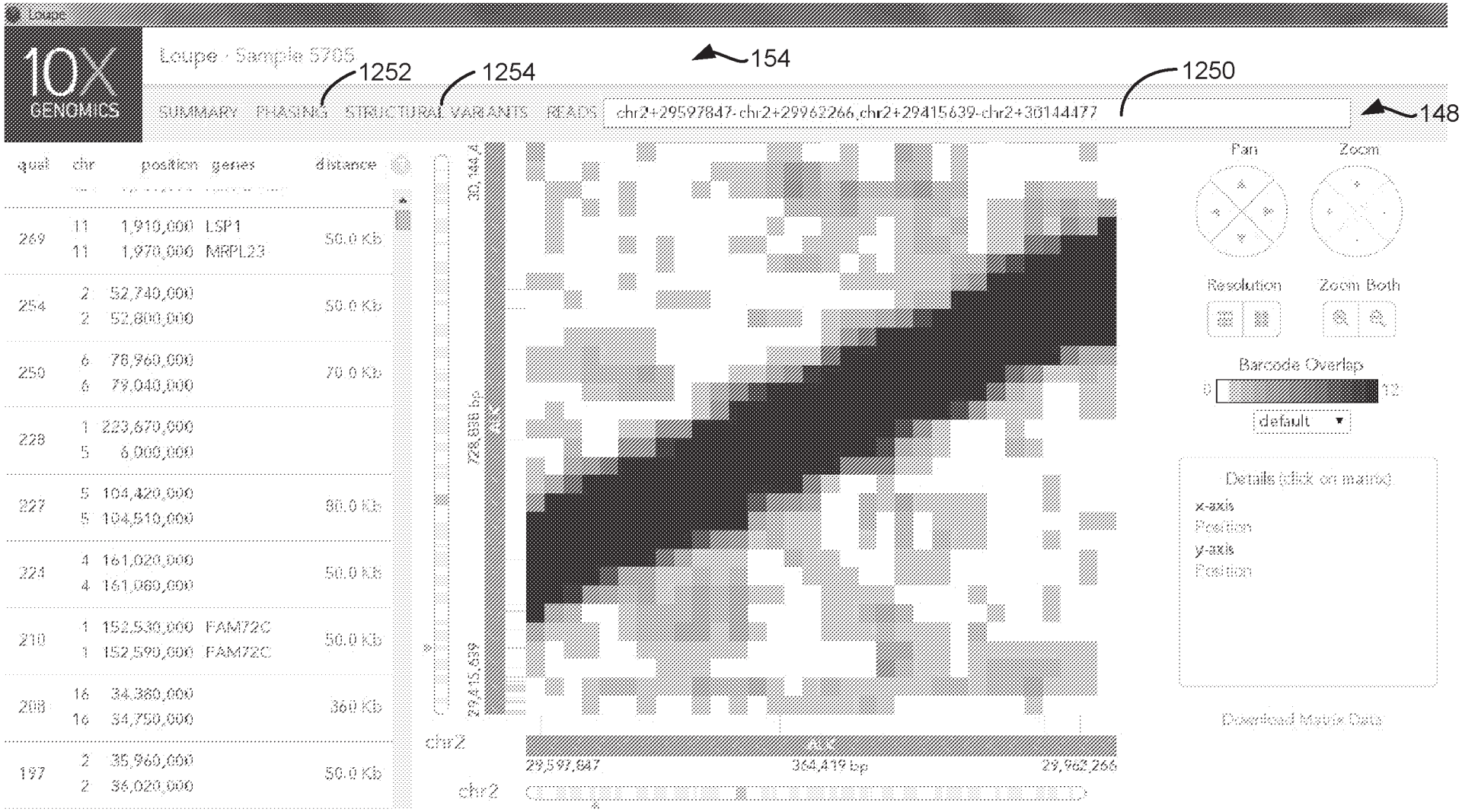

Loupe
10X GENOMICS
Loupe - Sample 5705
154
1252
1254
1250
148
SUMMARY
PHASING
STRUCTURAL VARIANTS
READS
chr2+29597847-chr2+29962266,chr2+29415639-chr2+30144477
qual
chr
position
genes
distance
269
11 1,910,000 LSP1
11 1,970,000 MRPL23
50.0 Kb
254
2 52,740,000
2 52,800,000
50.0 Kb
250
6 78,960,000
6 79,040,000
70.0 Kb
228
1 223,670,000
5 6,000,000
227
5 104,420,000
5 104,510,000
90.0 Kb
224
4 161,020,000
4 161,060,000
50.0 Kb
210
1 152,530,000 FAM72C
1 152,590,000 FAM72C
50.0 Kb
208
16 34,360,000
16 34,750,000
360 Kb
197
2 35,960,000
2 36,020,000
50.0 Kb
30,144,477 bp
728,838 bp
29,415,639
chr2
29,597,847
364,419 bp
29,962,266
chr2
Pan
Zoom
Resolution
Zoom Both
Barcode Overlap
0
12
default
Details (click on matrix)
x-axis
Position
y-axis
Position
Download Matrix Data

FIG. 19

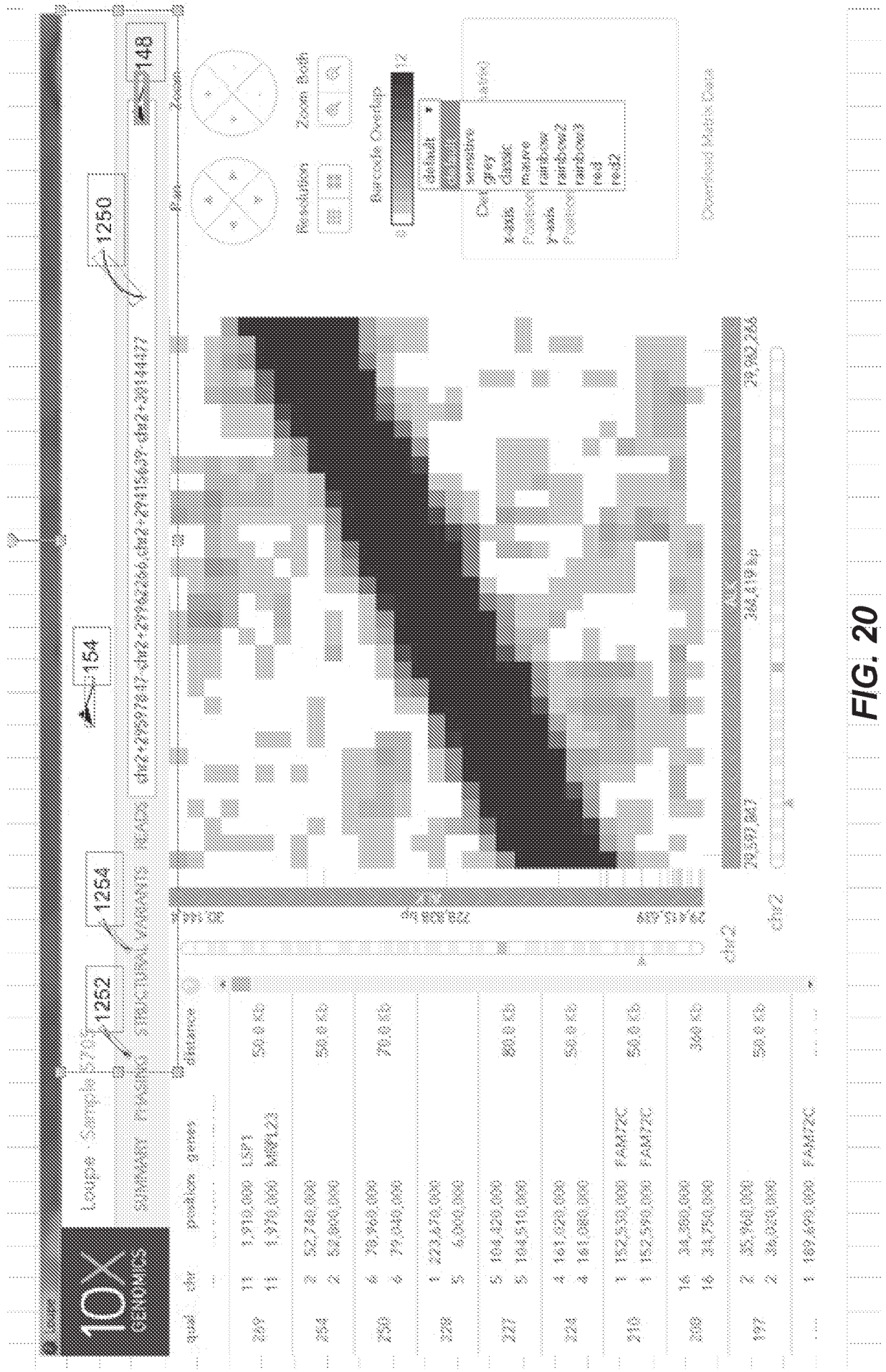

10X GENOMICS
Loupe · Sample 5705
SUMMARY
PHASING
STRUCTURAL VARIANTS
BEAM
chr2:27997847-chr2:27998247-chr2:27998266-chr2:27998277-chr2:29144477
148
Pan
Zoom
Resolution
Zoom Both
Barcode Overlap
default
sensitive
Color
grey
classic
maroon
rainbow
rainbow2
rainbow3
red
red2
Position
Position
Overlaid Molecules
154
1254
1252
1250
chr2
29,997,847
29,998,266
384,419 bp
distance
qual
chr
position
genes
50.0 Kb
80.0 Kb
70.0 Kb
366 Kb
50.0 Kb
50.0 Kb
LSP1
MBP1L3
FAM72C

FIG. 20

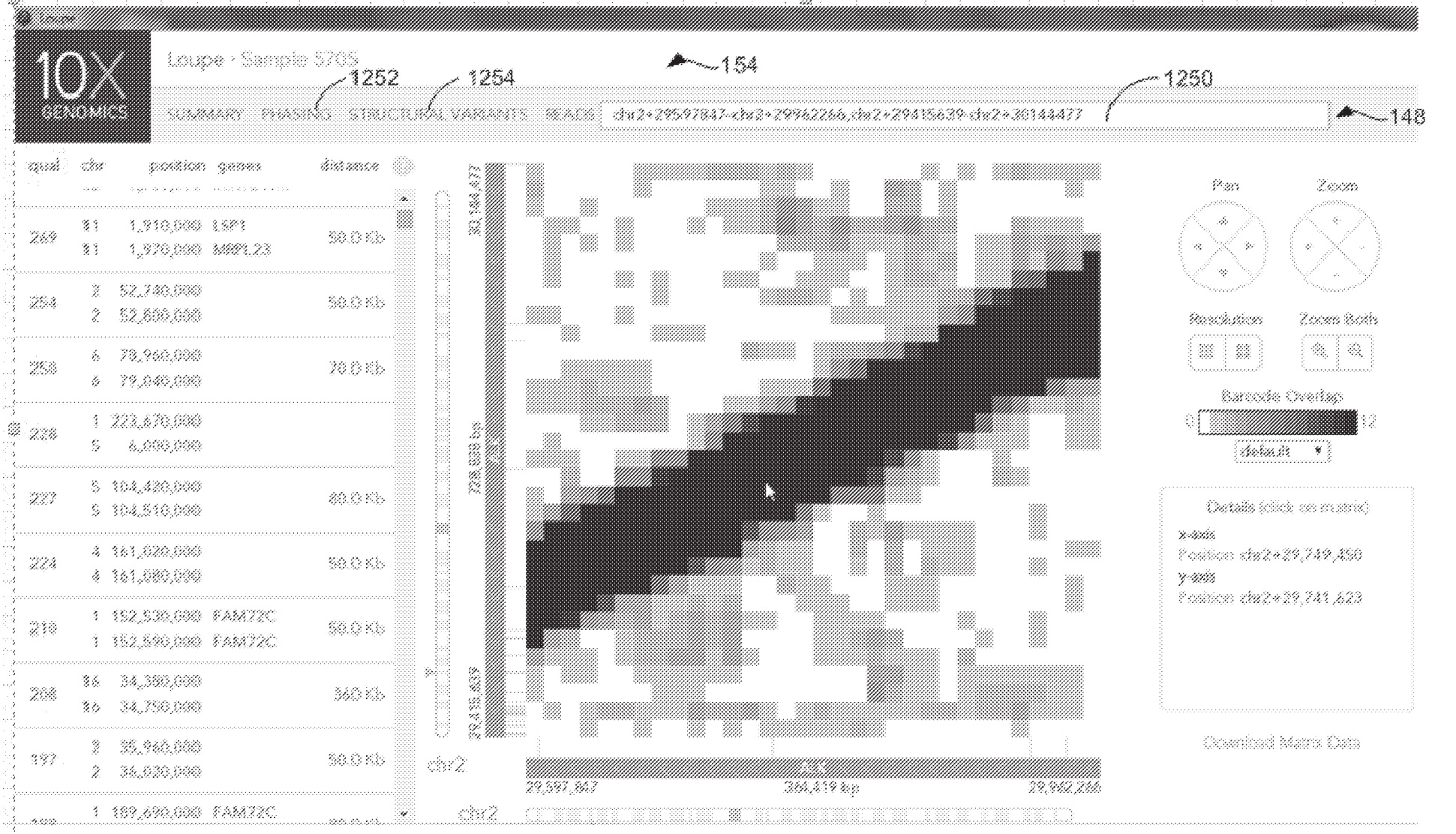

10X GENOMICS
Loupe - Sample 5705
1252
1254
154
1250
148
SUMMARY
PHASING
STRUCTURAL VARIANTS
READS
chr2:29597847-chr2:29962266,chr2:29415639-chr2:30144477
qual
chr
position
genes
distance
269
11 1,910,000 LSP1
11 1,970,000 MRPL23
50.0 Kb
254
2 52,740,000
2 52,600,000
50.0 Kb
258
6 78,960,000
6 79,040,000
70.0 Kb
228
1 223,670,000
5 6,000,000
227
5 104,420,000
5 104,510,000
60.0 Kb
224
4 161,020,000
4 161,080,000
50.0 Kb
218
1 152,530,000 FAM72C
1 152,590,000 FAM72C
50.0 Kb
208
16 34,380,000
16 34,750,000
360 Kb
197
2 35,960,000
2 36,020,000
50.0 Kb
1 189,690,000 FAM72C
chr2
chr2
29,597,847
364,419 bp
29,962,266
Pan
Zoom
Resolution
Zoom Both
Barcode Overlap
default
Details (click on matrix)
x-axis
Position chr2:29,749,450
y-axis
Position chr2:29,741,623
Download Matrix Data

FIG. 21

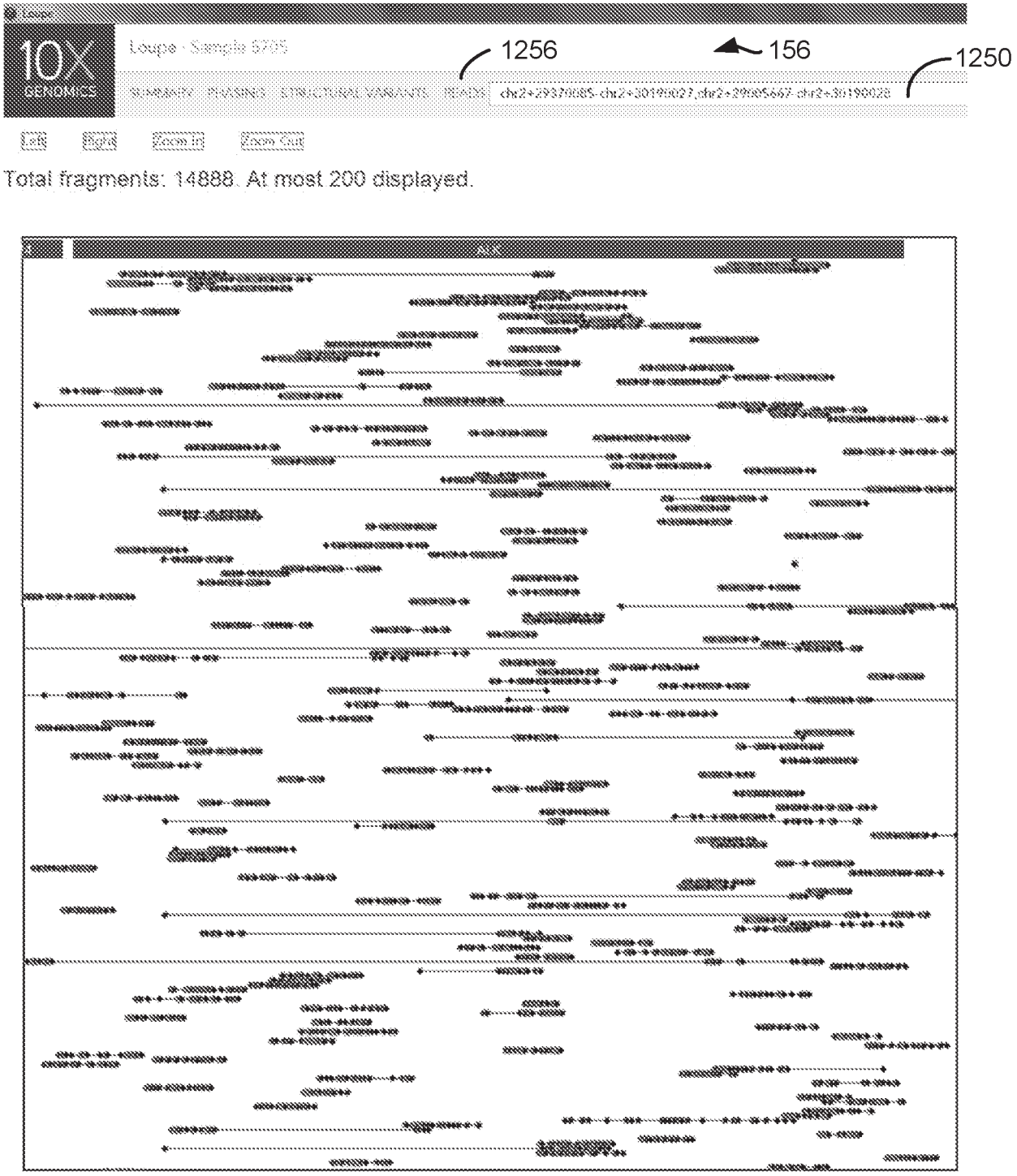

Loupe
10X GENOMICS
Loupe - Sample 5705
1256
156
1250
SUMMARY PHASING STRUCTURAL VARIANTS READS
chr2+29379085-chr2+30199027,chr2+29905667-chr2+30199028
Left
Right
Zoom In
Zoom Out
Total fragments: 14888. At most 200 displayed.

FIG. 22

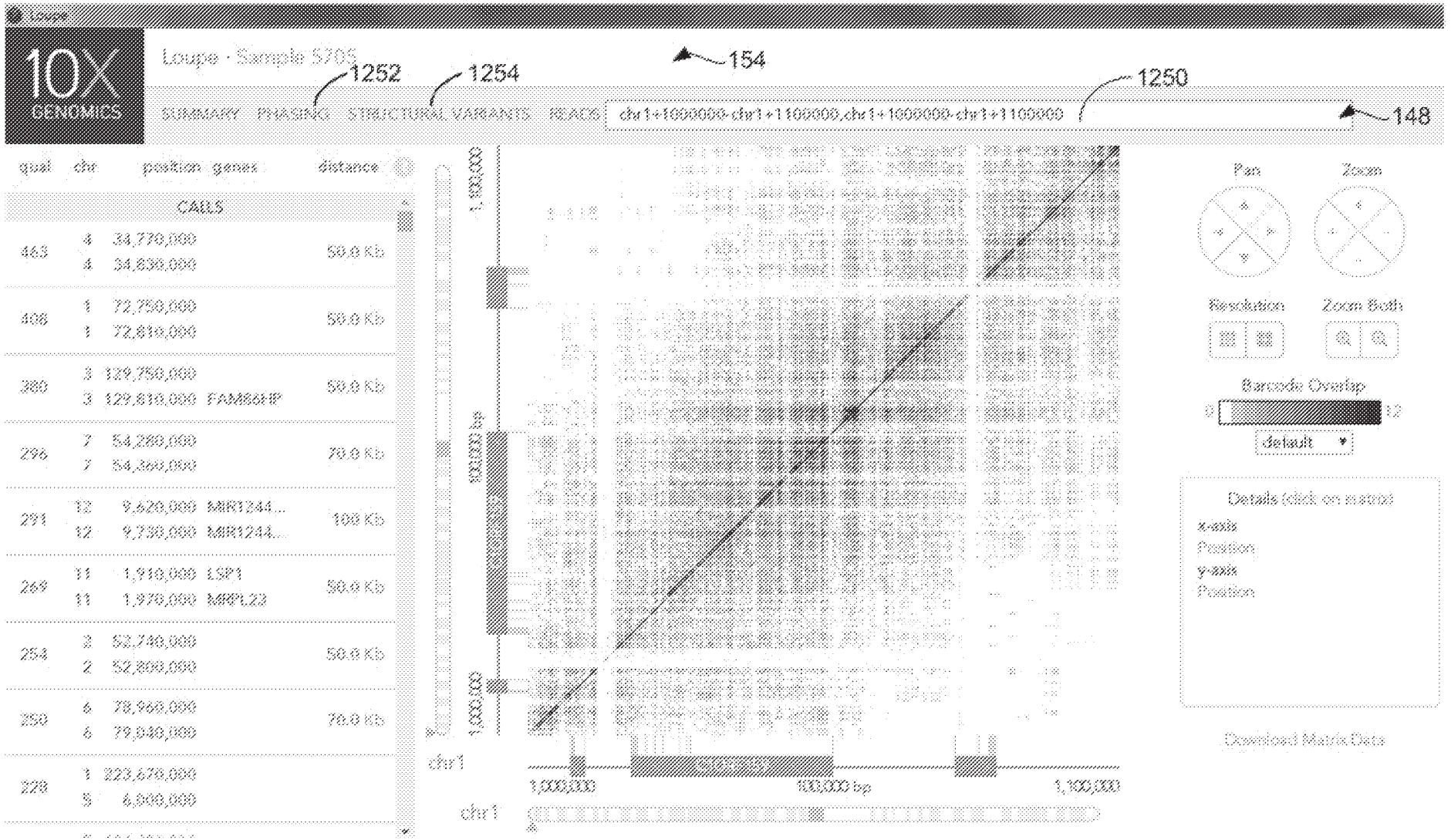

10X GENOMICS
Loupe · Sample 5705
1252
1254
154
1250
148
SUMMARY
PHASING
STRUCTURAL VARIANTS
READS
chr1:1000000-chr1:1100000,chr1:1000000-chr1:1100000
qual
chr
position
genes
distance
CALLS
463
4 34,770,000
4 34,830,000
50.0 Kb
408
1 72,750,000
1 72,810,000
50.0 Kb
380
3 129,750,000
3 129,810,000
FAM86HP
50.0 Kb
296
7 54,280,000
7 54,300,000
70.0 Kb
291
12 9,620,000
12 9,730,000
MIR1244...
MIR1244...
100 Kb
269
11 1,910,000
11 1,970,000
LSP1
MRPL23
50.0 Kb
254
2 52,740,000
2 52,800,000
50.0 Kb
250
6 78,960,000
6 79,030,000
70.0 Kb
228
1 223,670,000
5 6,900,000
1,100,000 bp
1,000,000
chr1
1,000,000
100,000 bp
1,100,000
chr1
Pan
Zoom
Resolution
Zoom Both
Barcode Overlap
default
Details (click on a plot)
x-axis
Position
y-axis
Position
Download Matrix Data

FIG. 23

10X GENOMICS
Loupe - Sample S705
1252
1254
154
1250
148
SUMMARY
PHASING
STRUCTURAL VARIANTS
READS
chr4:34675000-chr4:34875000,chr4:34735000-chr4:34935000
qual
chr
position
genes
distance
CALLS
Pan
Zoom
Resolution
Zoom Both
Barcode Overlap
default
Details (click on matrix)
x-axis
Position
y-axis
Position
Download Matrix Data
chr4
chr4
34,675,000
300,000 bp
34,875,000

FIG. 24

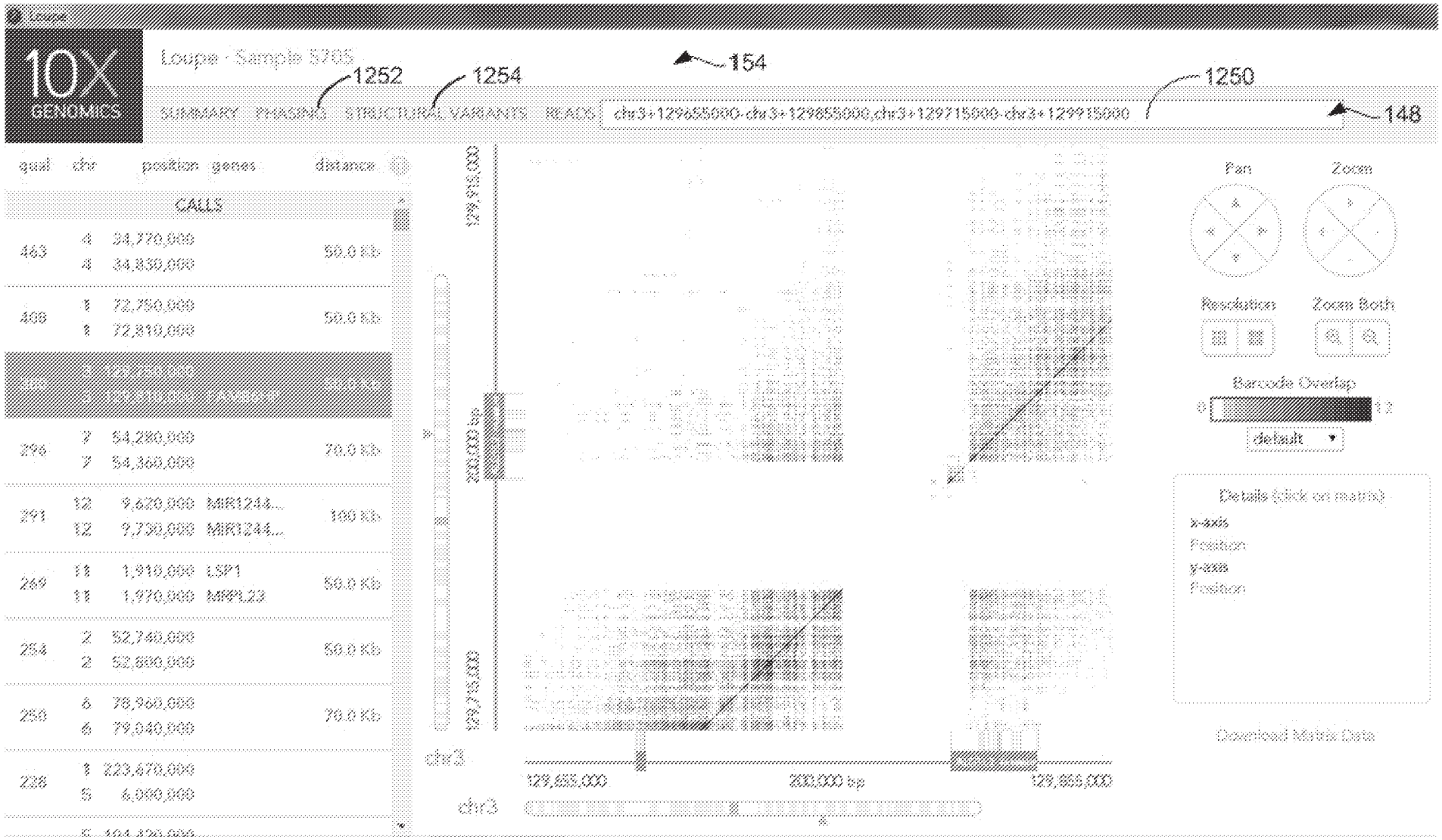

10X GENOMICS
Loupe · Sample 5705
154
1252
1254
1250
148
SUMMARY
PHASING
STRUCTURAL VARIANTS
READS
chr3:129655000-chr3:129855000,chr3:129715000-chr3:129915000
qual
chr
position
genes
distance
CALLS
463
4 34,770,000
4 34,830,000
50.0 Kb
408
1 72,750,000
1 72,810,000
50.0 Kb
296
7 54,280,000
7 54,360,000
70.0 Kb
291
12 9,620,000 MIR1244...
12 9,730,000 MIR1244...
100 Kb
269
11 1,910,000 LSP1
11 1,970,000 MRPL23
50.0 Kb
254
2 52,740,000
2 52,800,000
50.0 Kb
250
6 78,960,000
6 79,040,000
70.0 Kb
228
1 223,670,000
5 6,080,000
129,915,000
200,000 bp
129,715,000
chr3
129,655,000
200,000 bp
129,855,000
chr3
Pan
Zoom
Resolution
Zoom Both
Barcode Overlap
default
Details (click on matrix)
x-axis
Position
y-axis
Position
Download Matrix Data

FIG. 25

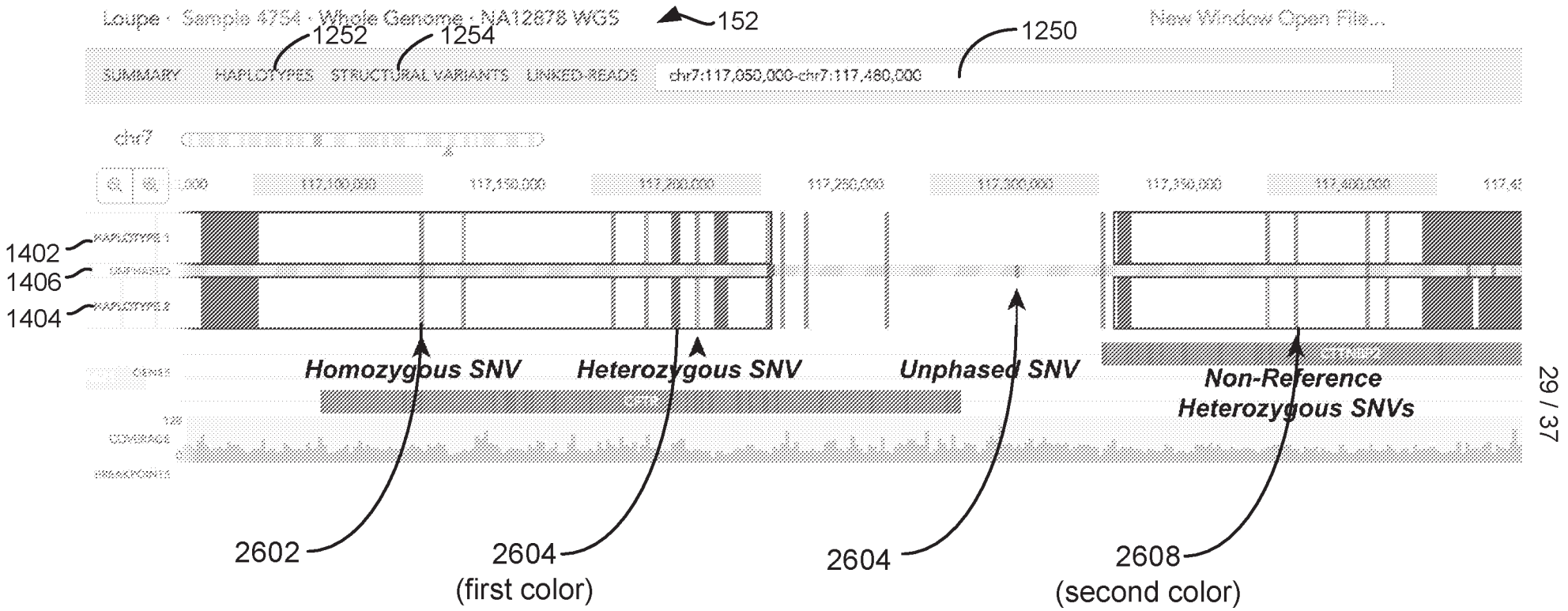

Loupe · Sample 4754 · Whole Genome · NA12878 WGS
152
New Window Open File...
1252
1254
1250
SUMMARY
HAPLOTYPES
STRUCTURAL VARIANTS
LINKED-READS
chr7:117,050,000-chr7:117,480,000
chr7
117,100,000
117,150,000
117,200,000
117,250,000
117,300,000
117,350,000
117,400,000
1402
1406
1404
Homozygous SNV
Heterozygous SNV
Unphased SNV
Non-Reference
Heterozygous SNVs
2602
2604
(first color)
2604
2608
(second color)

FIG. 26

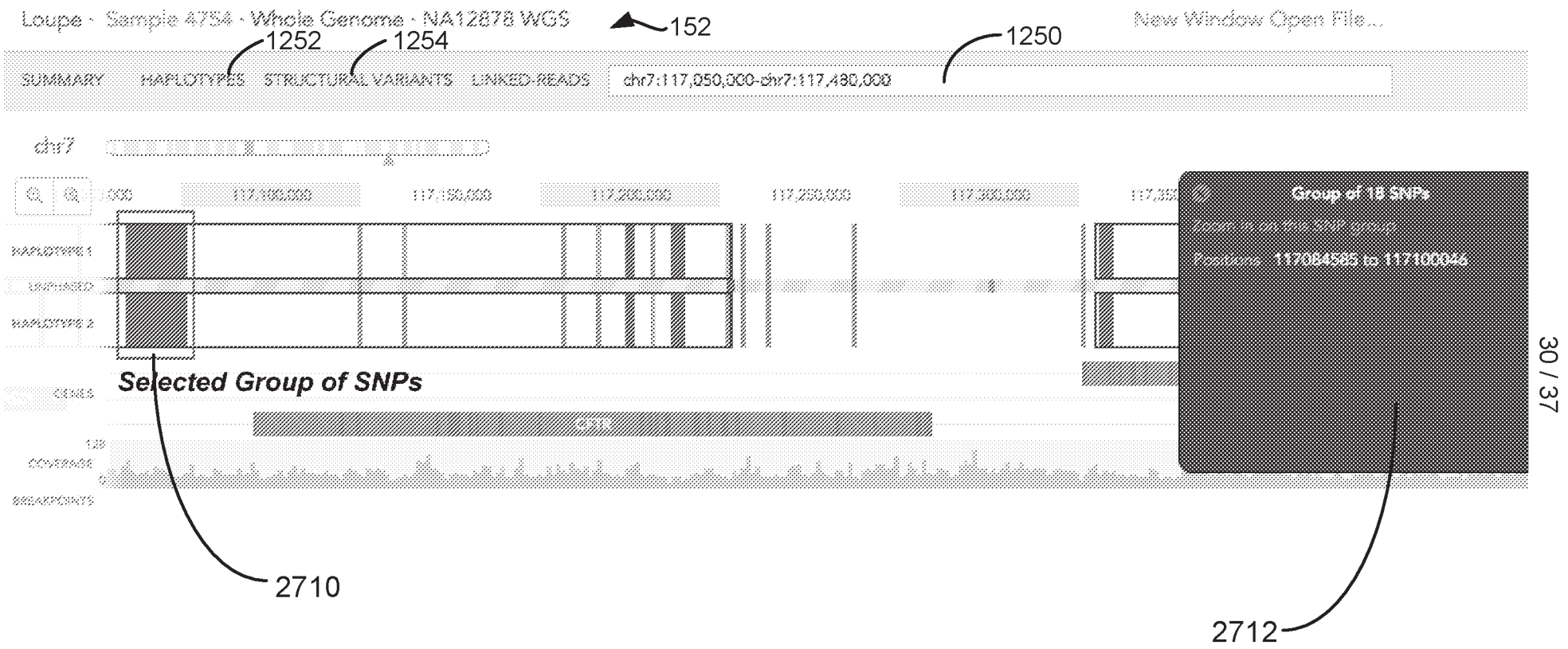

Loupe - Sample 4754 - Whole Genome - NA12878 WGS
152
New Window Open File...
1252
1254
1250
SUMMARY
HAPLOTYPES
STRUCTURAL VARIANTS
LINKED-READS
chr7:117,050,000-chr7:117,480,000
chr7
Group of 18 SNPs
Positions: 117084585 to 117100046
Selected Group of SNPs
2710
2712
30 / 37

FIG. 27

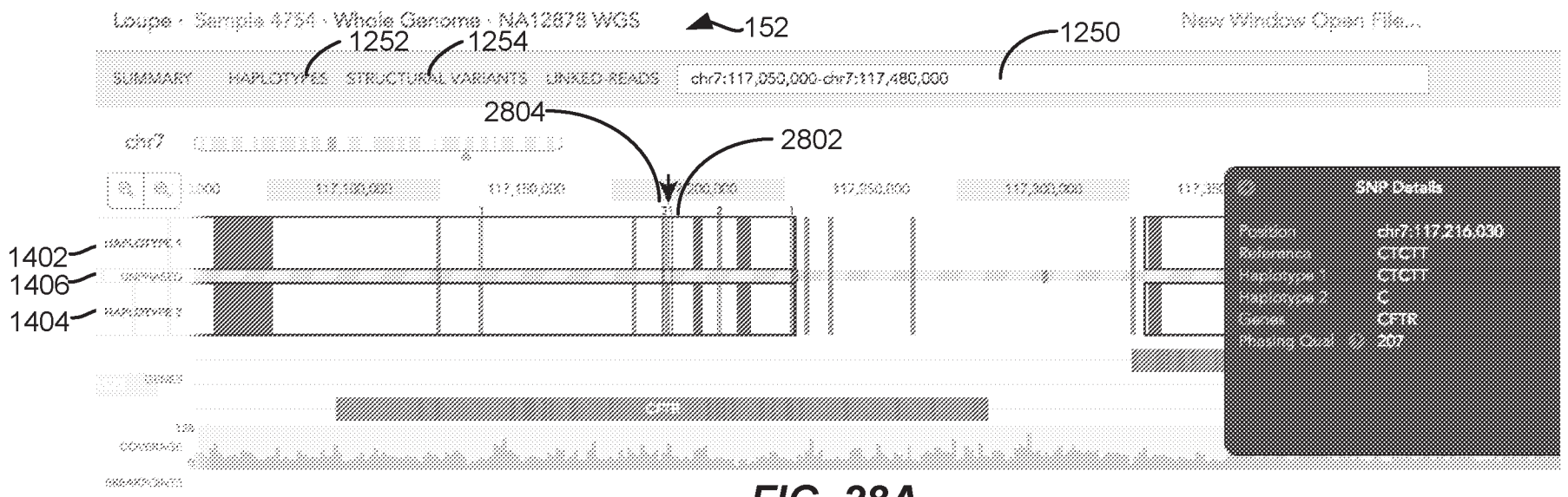


FIG. 28A

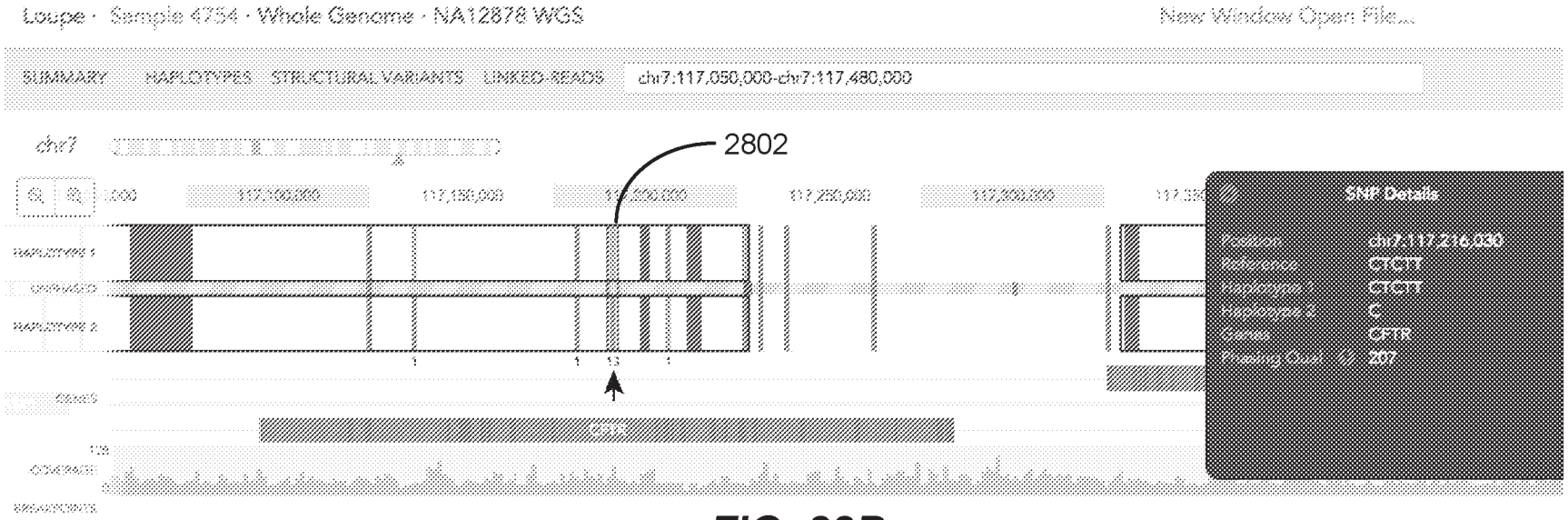


FIG. 28B

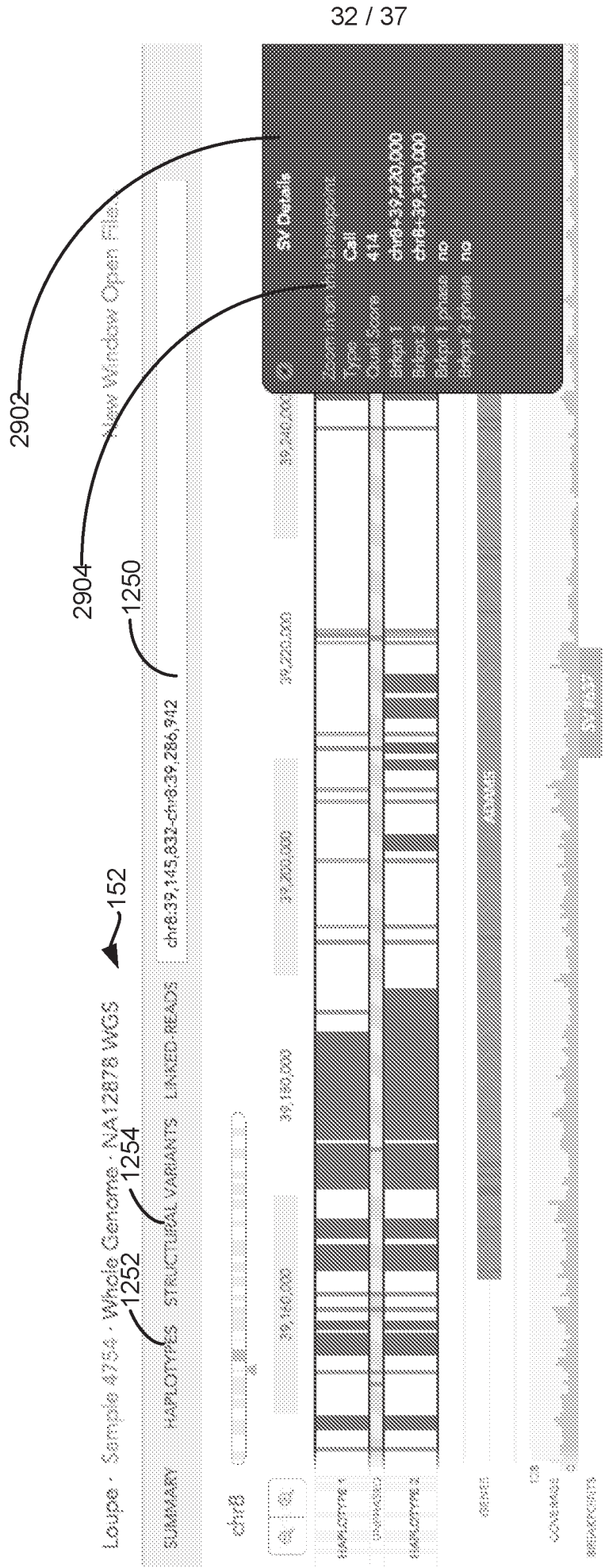

2902
2904
1250
152
1252
1254
Loupe · Sample 4754 · Whole Genome · NA12878 WGS
New Window Open File
SUMMARY
HAPLOTYPES
STRUCTURAL VARIANTS
LINKED-READS
chr8:39,145,832-chr8:39,286,942
SV Details
Call
414
chr8:39,220,000
chr8:39,260,000
no
no
chr8
39,140,000
39,180,000
39,200,000
39,220,000
39,240,000
genes
coverage

FIG. 29

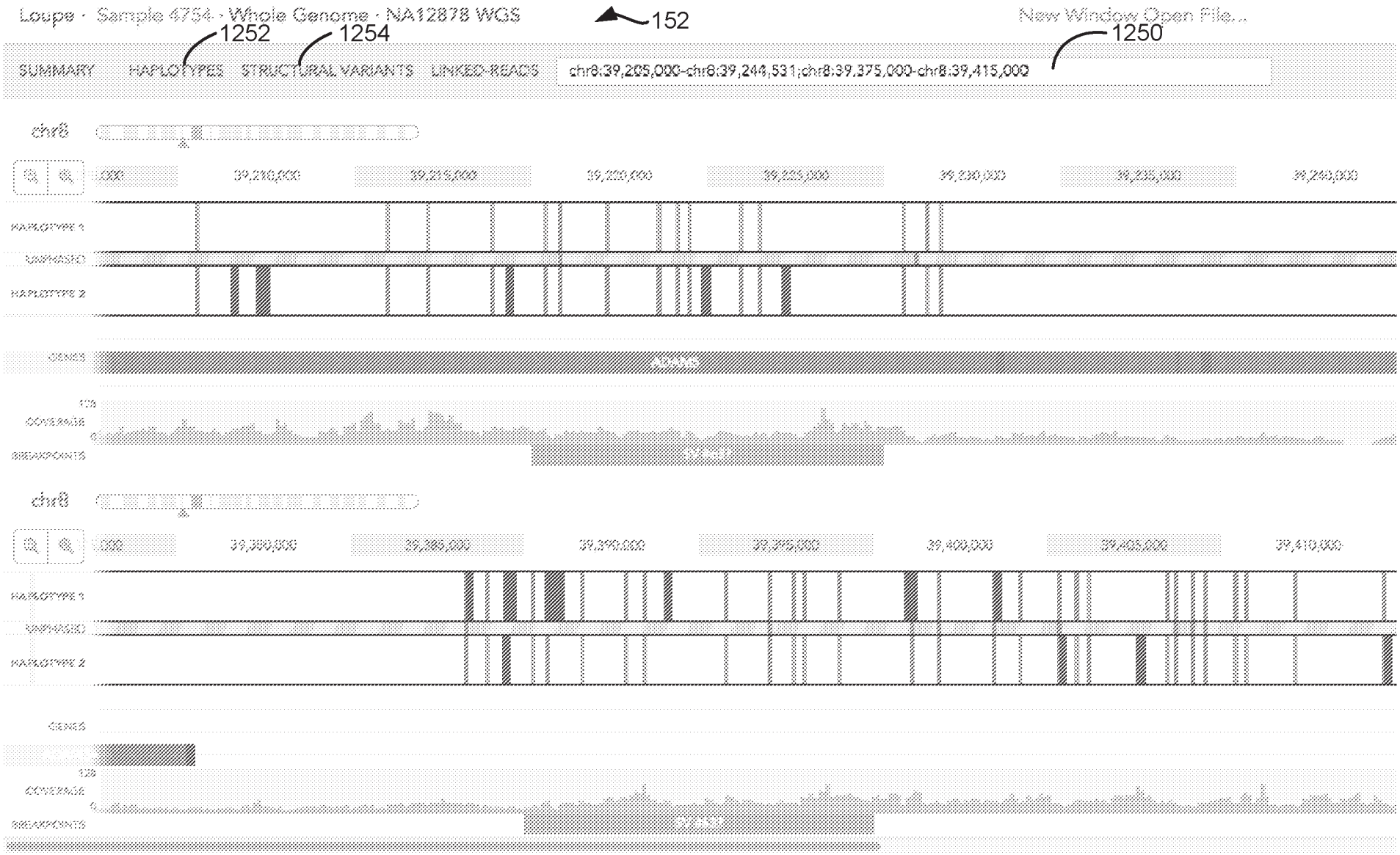

Loupe · Sample 4754 · Whole Genome · NA12878 WGS
152
New Window Open File...
1252
1254
1250
SUMMARY
HAPLOTYPES
STRUCTURAL VARIANTS
LINKED-READS
chr8:39,205,000-chr8:39,244,531;chr8:39,375,000-chr8:39,415,000
chr8
39,210,000
39,215,000
39,220,000
39,225,000
39,230,000
39,235,000
39,240,000
HAPLOTYPE 1
UNPHASED
HAPLOTYPE 2
GENES
COVERAGE
BREAKPOINTS
chr8
39,380,000
39,385,000
39,390,000
39,395,000
39,400,000
39,405,000
39,410,000
HAPLOTYPE 1
UNPHASED
HAPLOTYPE 2
GENES
COVERAGE
BREAKPOINTS

FIG. 30

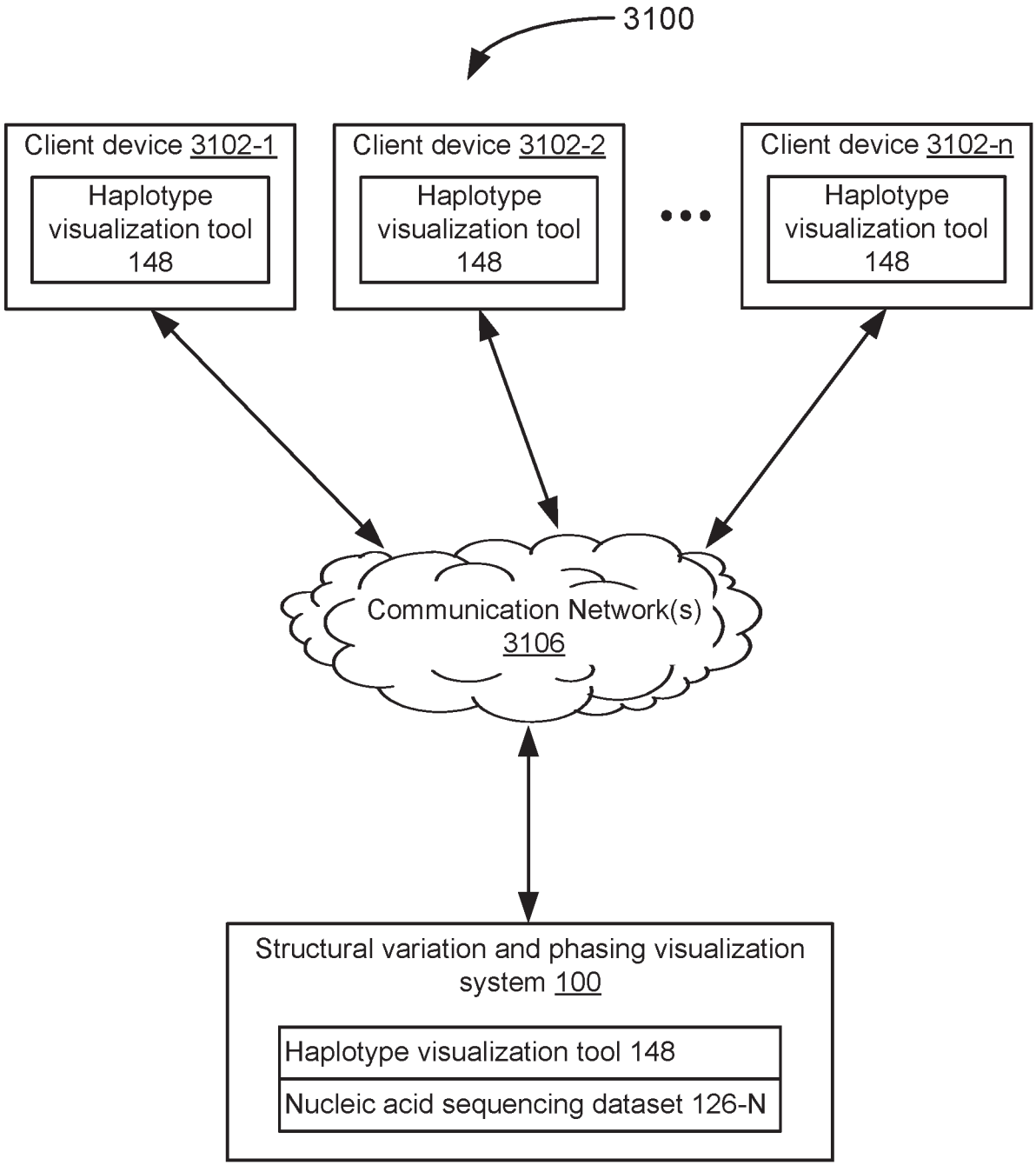

3100
Client device 3102-1
Haplotype visualization tool 148
Client device 3102-2
Haplotype visualization tool 148
Client device 3102-n
Haplotype visualization tool 148
Communication Network(s) 3106
Structural variation and phasing visualization system 100
Haplotype visualization tool 148
Nucleic acid sequencing dataset 126-N

FIG. 31

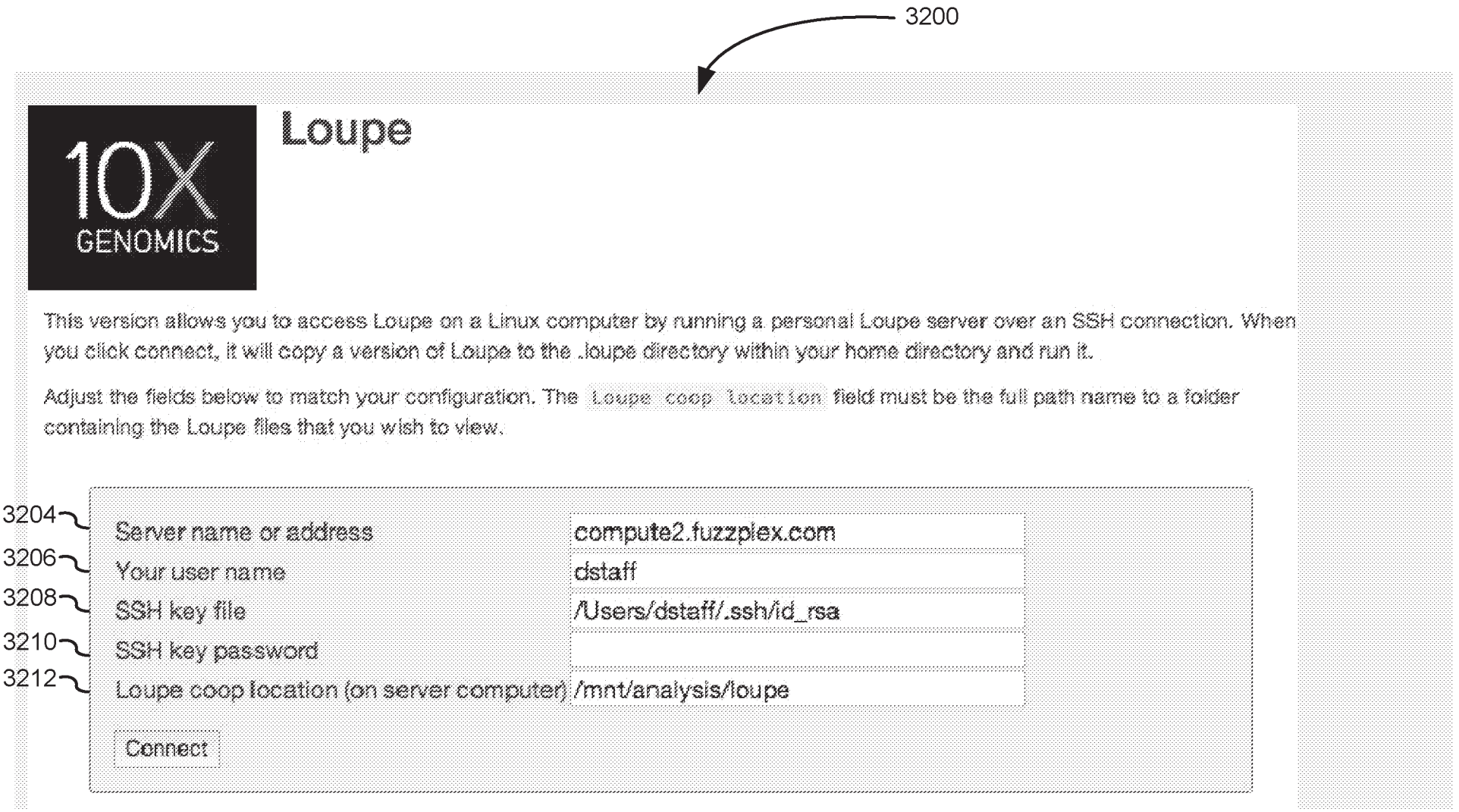

3200
10X GENOMICS
Loupe
This version allows you to access Loupe on a Linux computer by running a personal Loupe server over an SSH connection. When you click connect, it will copy a version of Loupe to the .loupe directory within your home directory and run it.
Adjust the fields below to match your configuration. The Loupe coop location field must be the full path name to a folder containing the Loupe files that you wish to view.
3204
Server name or address
compute2.fuzzplex.com
3206
Your user name
dstaff
3208
SSH key file
/Users/dstaff/.ssh/id_rsa
3210
SSH key password
3212
Loupe coop location (on server computer)
/mnt/analysis/loupe
Connect

FIG. 32

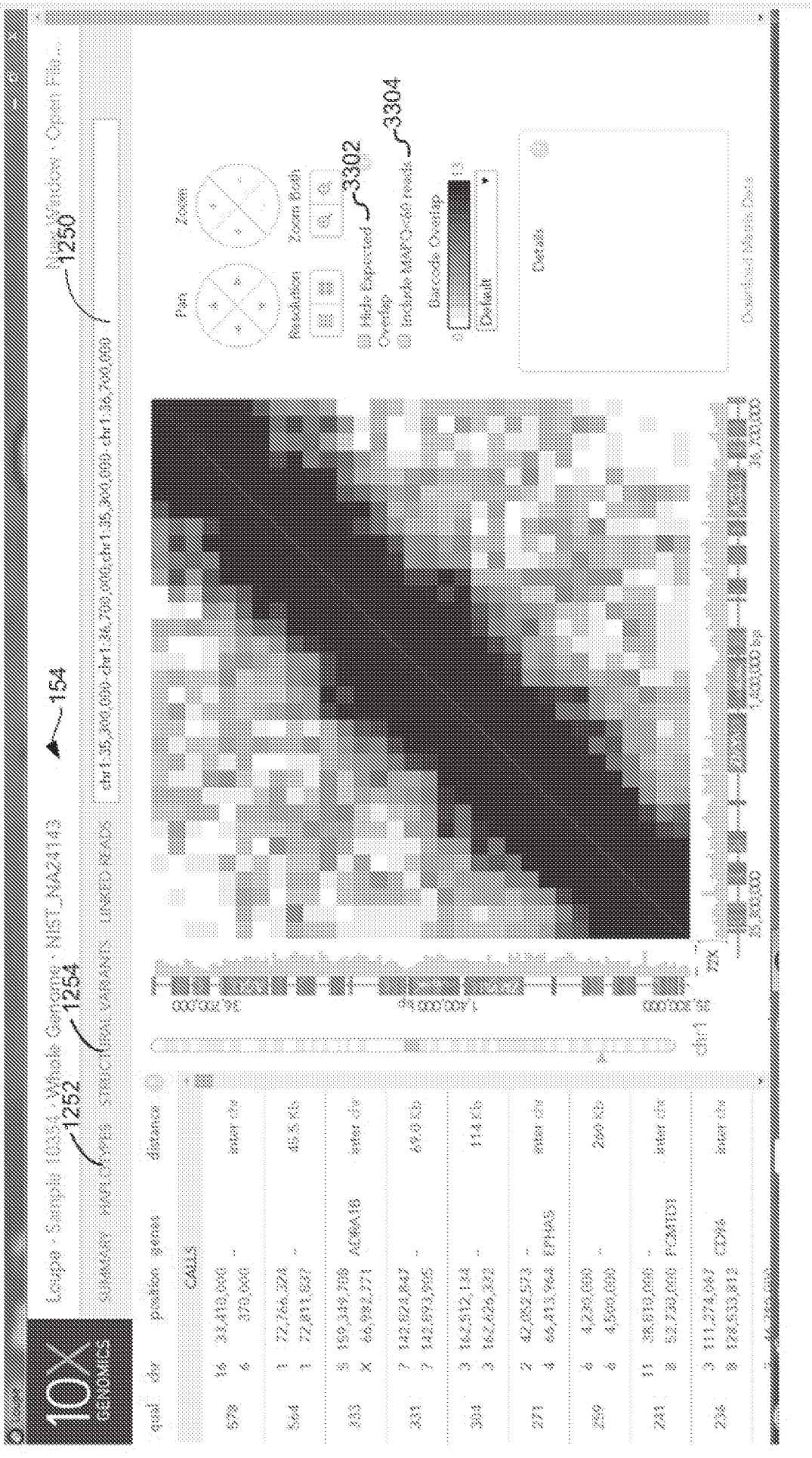

10X GENOMICS
Loupe - Sample ID354 - Whole Genome - NIST_NA24143
1252
1254
154
1250
STRUCTURAL VARIANTS
LINKED READS
chr1:35,300,000-chr1:36,700,000,chr1:35,300,000-chr1:36,700,000
Pan
Zoom
Reset
Zoom Both
Hide Expected Overlap
3302
Include MAPQ<60 reads
3304
Barcode Overlap
Default
Details
qual
chr
position
genes
distance
CALLS
578
16
33,419,000
-
inter chr
6
370,000
544
1
72,766,324
-
45.5 Kb
1
72,811,827
233
9
159,349,708
ADAM18
inter chr
X
46,992,771
231
7
142,824,847
-
69.8 Kb
7
142,894,645
303
3
162,312,184
-
114 Kb
3
162,426,333
271
2
42,202,573
-
inter chr
4
66,413,964
EPHA5
259
6
4,226,000
-
200 Kb
6
4,500,000
241
11
38,819,000
-
inter chr
8
52,739,000
FGFR1OP2
236
3
111,274,047
-
inter chr
8
128,533,813
CDH6

FIG. 33

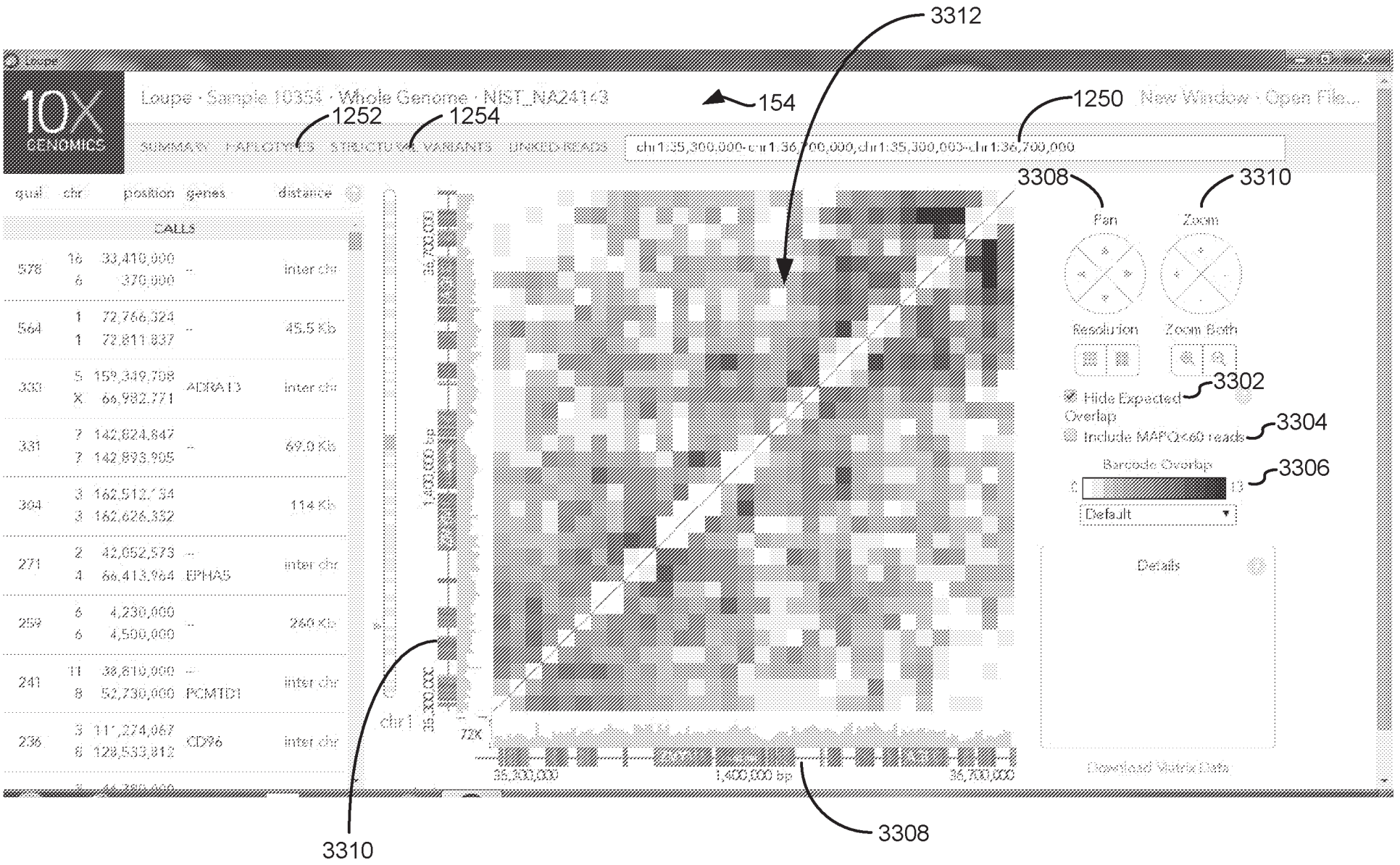

3312
10X GENOMICS
Loupe · Sample 10354 · Whole Genome · NIST_NA24143
154
1250
New Window · Open File...
1252
1254
SUMMARY
HAPLOTYPES
STRUCTURAL VARIANTS
LINKED-READS
chr1:35,300,000-chr1:36,700,000,chr1:35,300,000-chr1:36,700,000
3308
3310
Pan
Zoom
Resolution
Zoom Both
3302
Hide Expected Overlap
3304
Include MAPQ<60 reads
Barcode Overlap
3306
Default
Details
Download Matrix Data
qual
chr
position
genes
distance
CALLS
578
16 33,410,000
6 370,000
inter chr
564
1 72,766,324
1 72,811,837
45.5 Kb
333
5 159,339,708
X 66,982,771
ADRA1D
inter chr
331
7 142,824,847
7 142,893,905
69.0 Kb
304
3 162,512,734
3 162,626,332
114 Kb
271
2 42,052,573
4 66,413,964
EPHA5
inter chr
259
6 4,230,000
6 4,500,000
240 Kb
241
11 38,810,000
8 52,730,000
PCMTD1
inter chr
236
3 117,274,067
8 128,533,812
CD96
inter chr
chr 1
35,300,000
1,400,000 bp
36,700,000
72K
3310
3308

FIG. 34