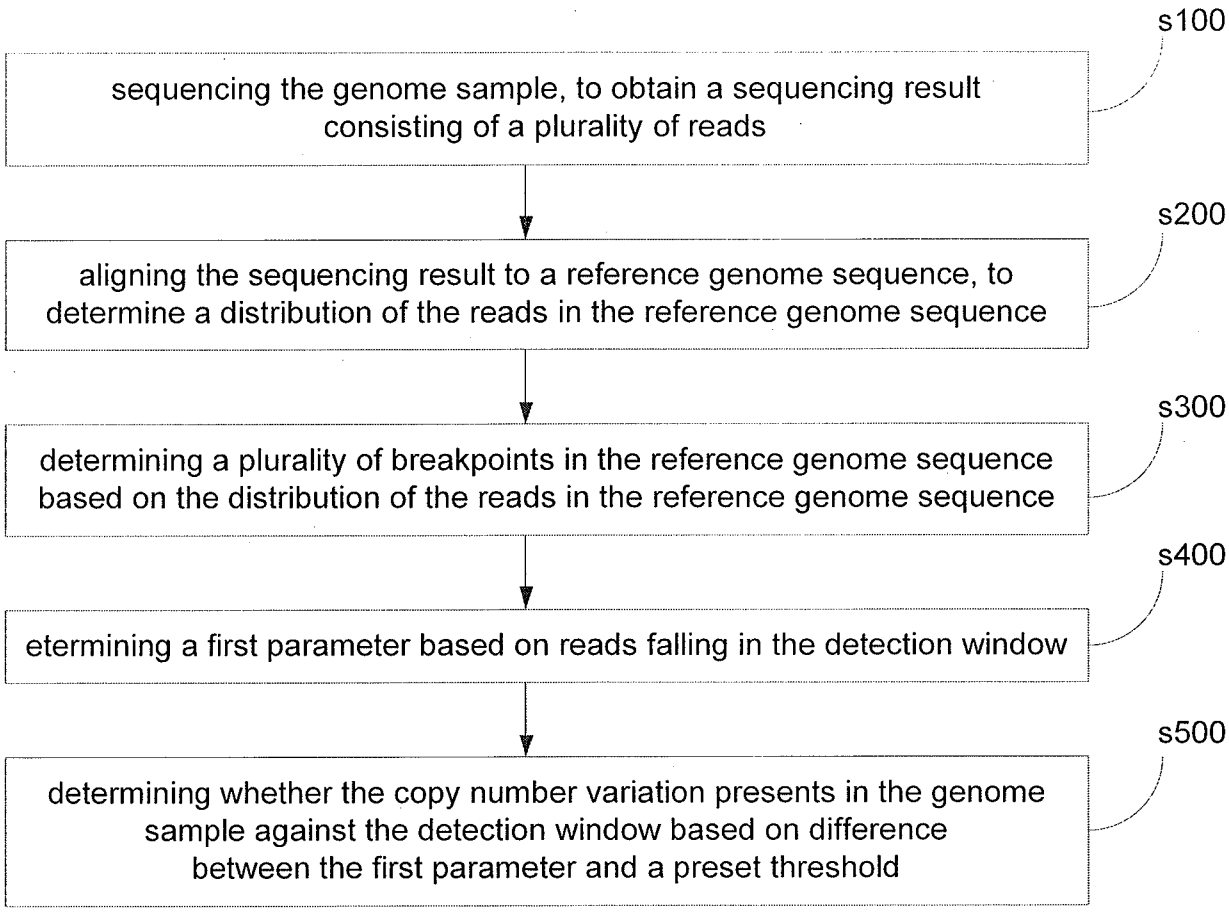
s100
sequencing the genome sample, to obtain a sequencing result consisting of a plurality of reads
s200
aligning the sequencing result to a reference genome sequence, to determine a distribution of the reads in the reference genome sequence
s300
determining a plurality of breakpoints in the reference genome sequence based on the distribution of the reads in the reference genome sequence
s400
etermining a first parameter based on reads falling in the detection window
s500
determining whether the copy number variation presents in the genome sample against the detection window based on difference between the first parameter and a preset threshold

FIG.1

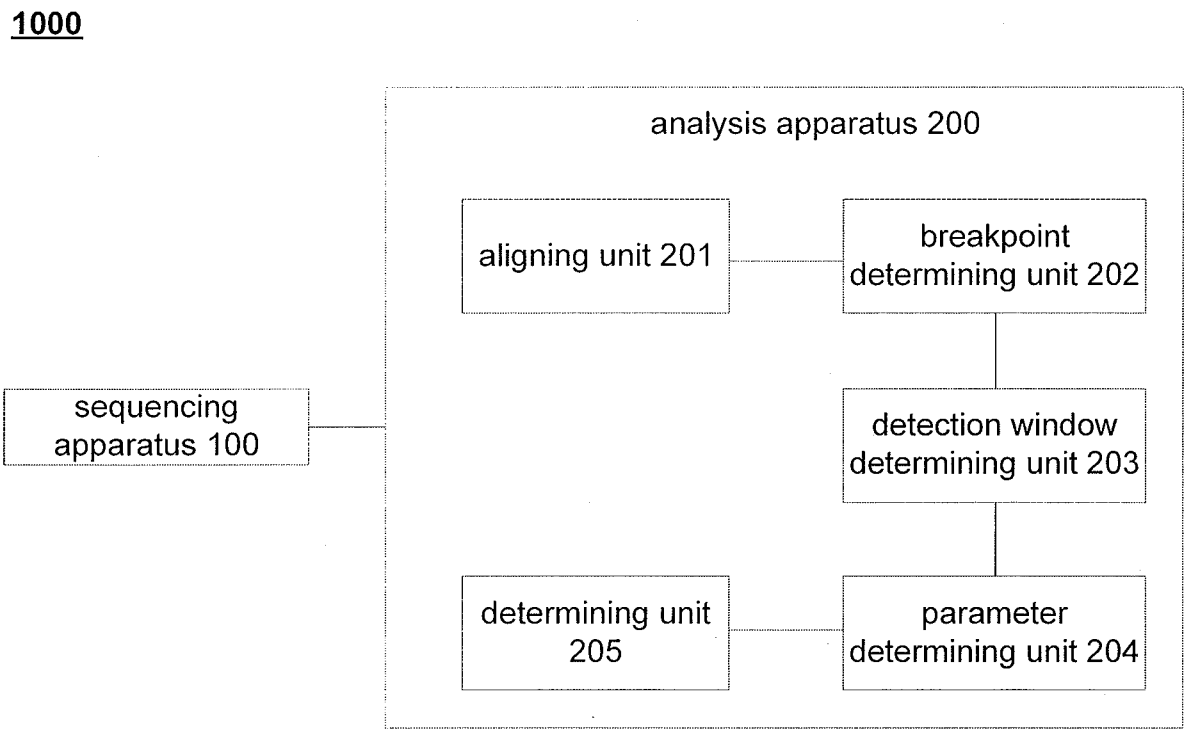
1000
analysis apparatus 200
aligning unit 201
breakpoint determining unit 202
sequencing apparatus 100
detection window determining unit 203
determining unit 205
parameter determining unit 204

FIG.2

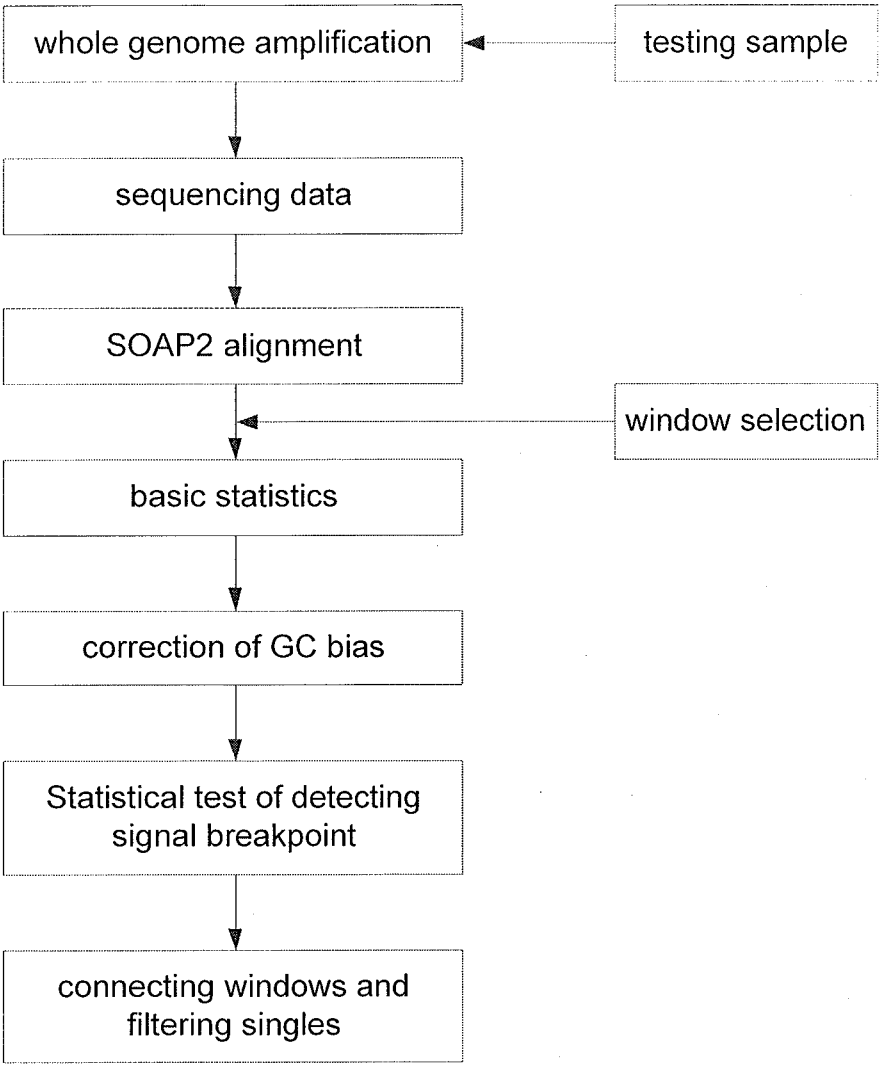
whole genome amplification
testing sample
sequencing data
SOAP2 alignment
window selection
basic statistics
correction of GC bias
Statistical test of detecting signal breakpoint
connecting windows and filtering singles

Fig.3

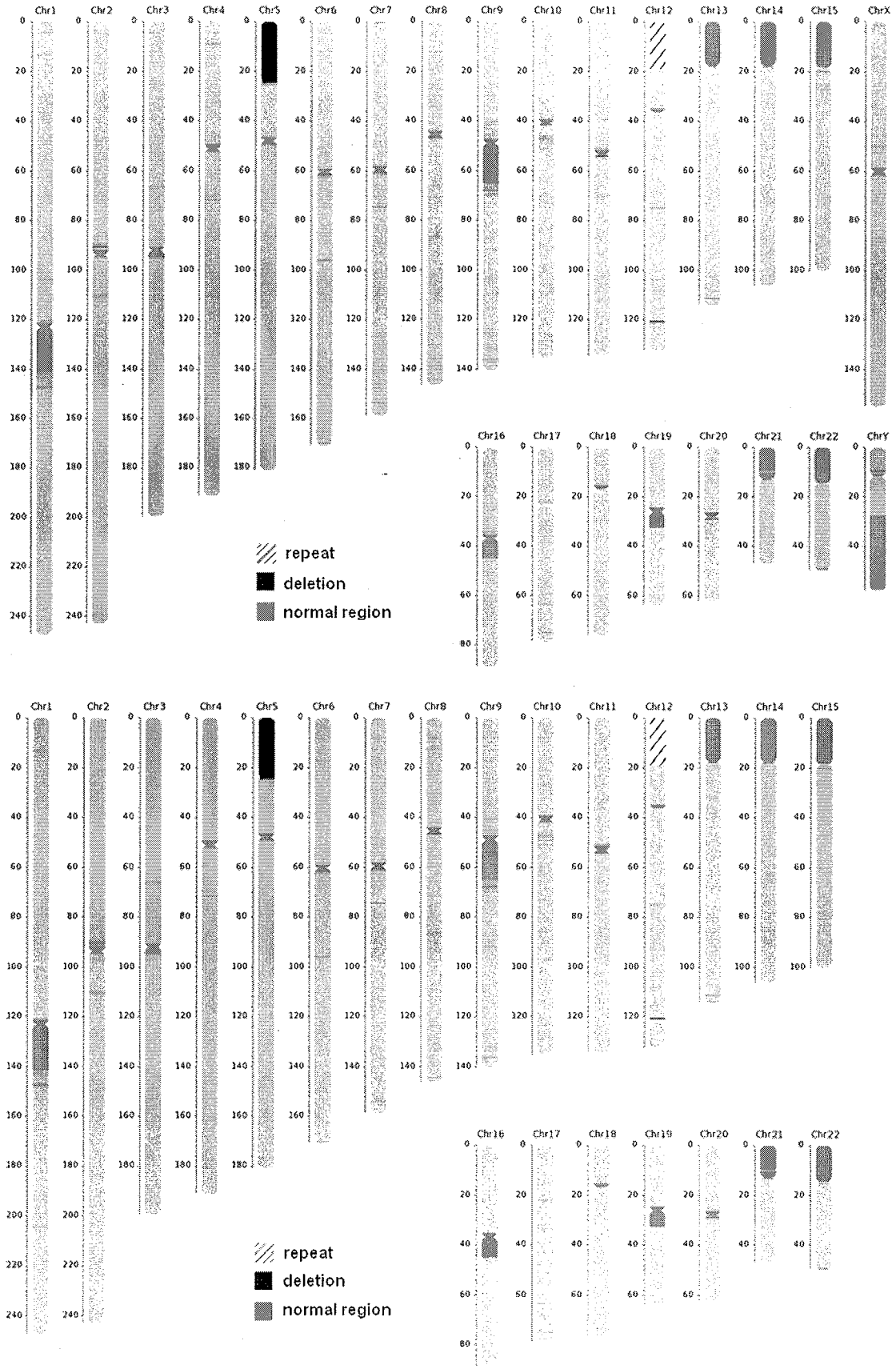
Chr1
Chr2
Chr3
Chr4
Chr5
Chr6
Chr7
Chr8
Chr9
Chr10
Chr11
Chr12
Chr13
Chr14
Chr15
ChrX
Chr16
Chr17
Chr18
Chr19
Chr20
Chr21
Chr22
ChrY
repeat
deletion
normal region
Chr1
Chr2
Chr3
Chr4
Chr5
Chr6
Chr7
Chr8
Chr9
Chr10
Chr11
Chr12
Chr13
Chr14
Chr15
Chr16
Chr17
Chr18
Chr19
Chr20
Chr21
Chr22
repeat
deletion
normal region

Fig.4

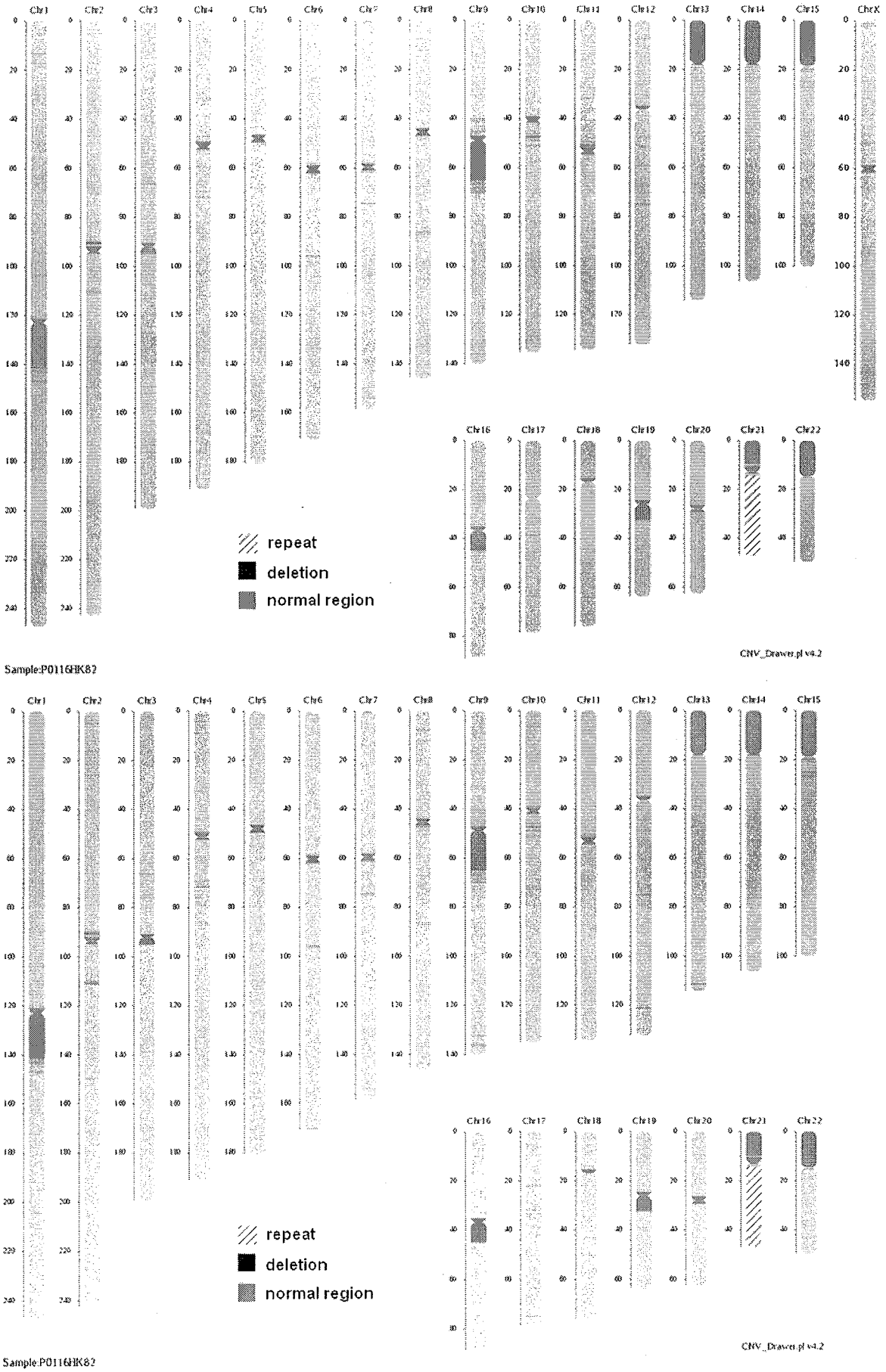
repeat
deletion
normal region
CNV_Drawer.pl v4.2
Sample:P0116HK82

Fig.5