

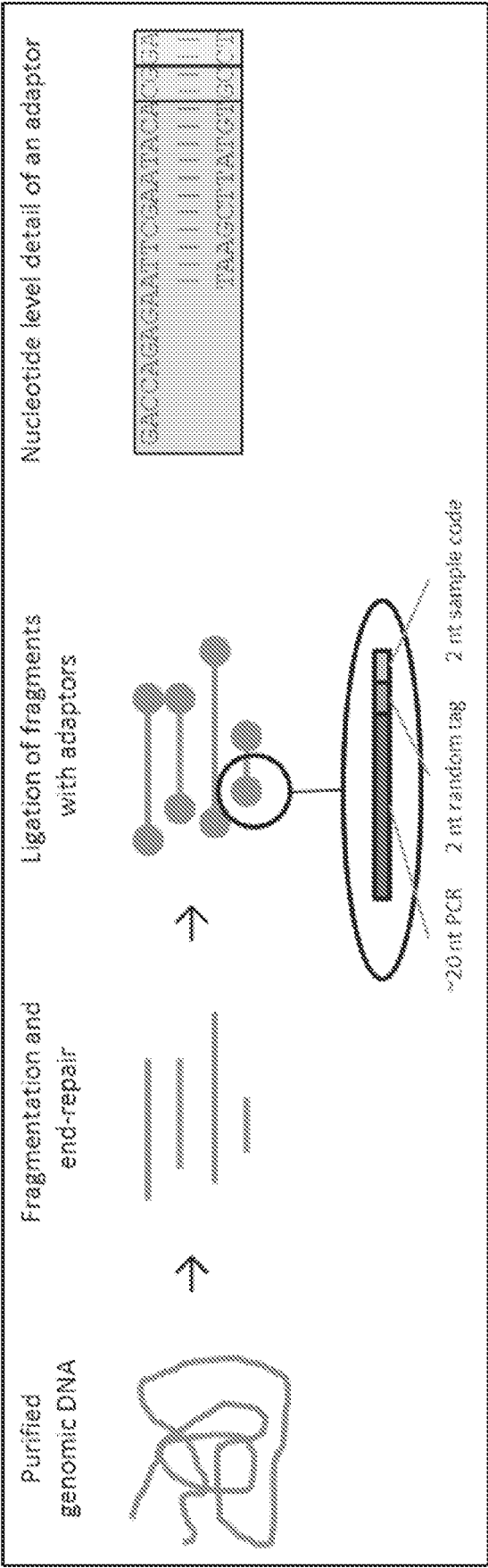


FIG. 1

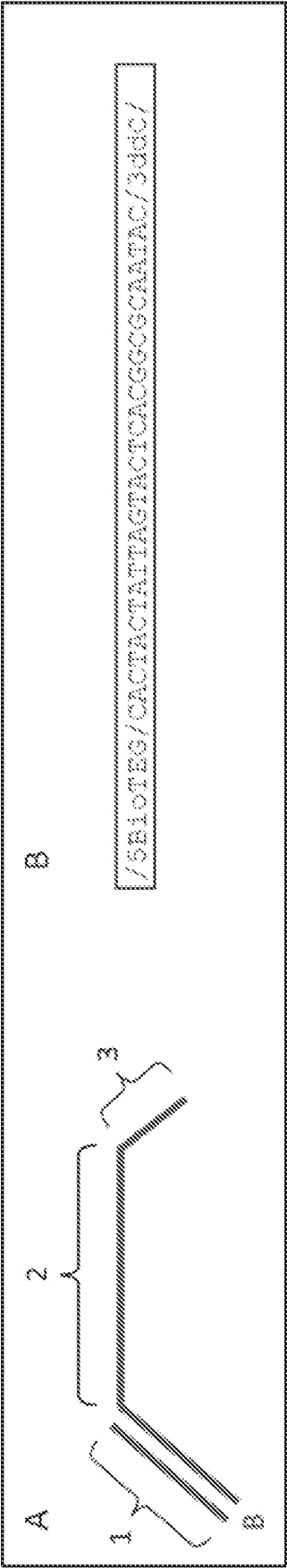


FIG. 2

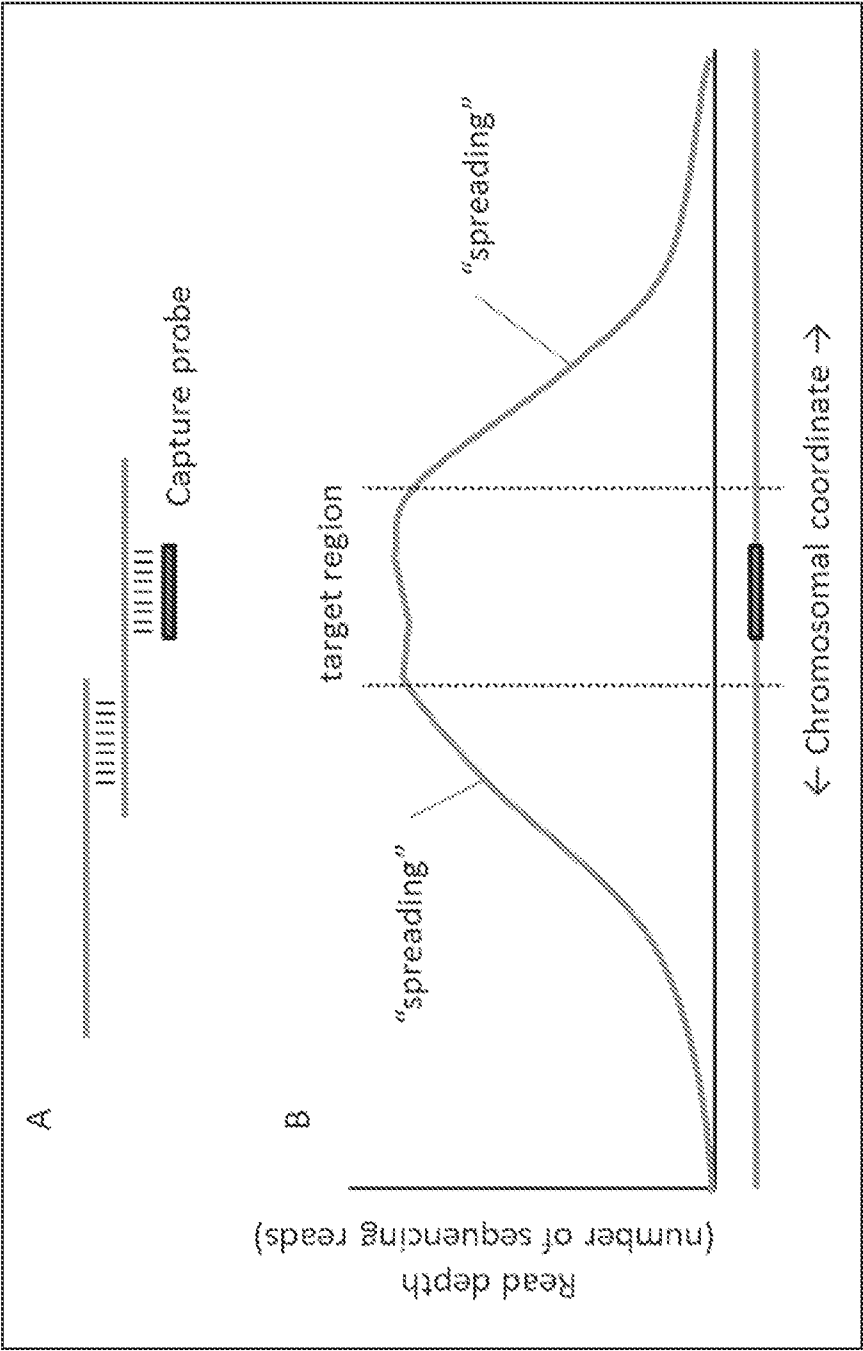


FIG. 3

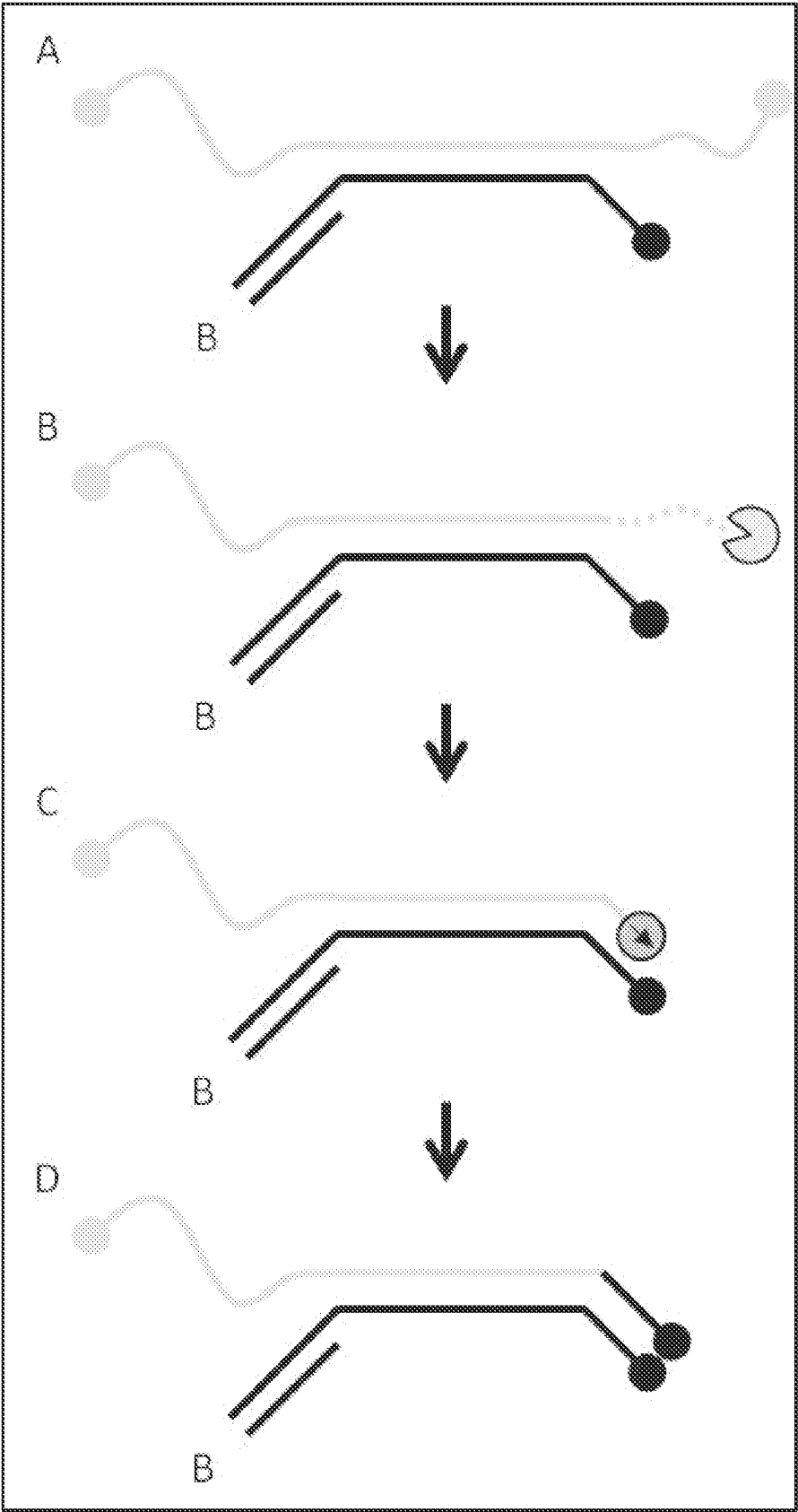


FIG. 4

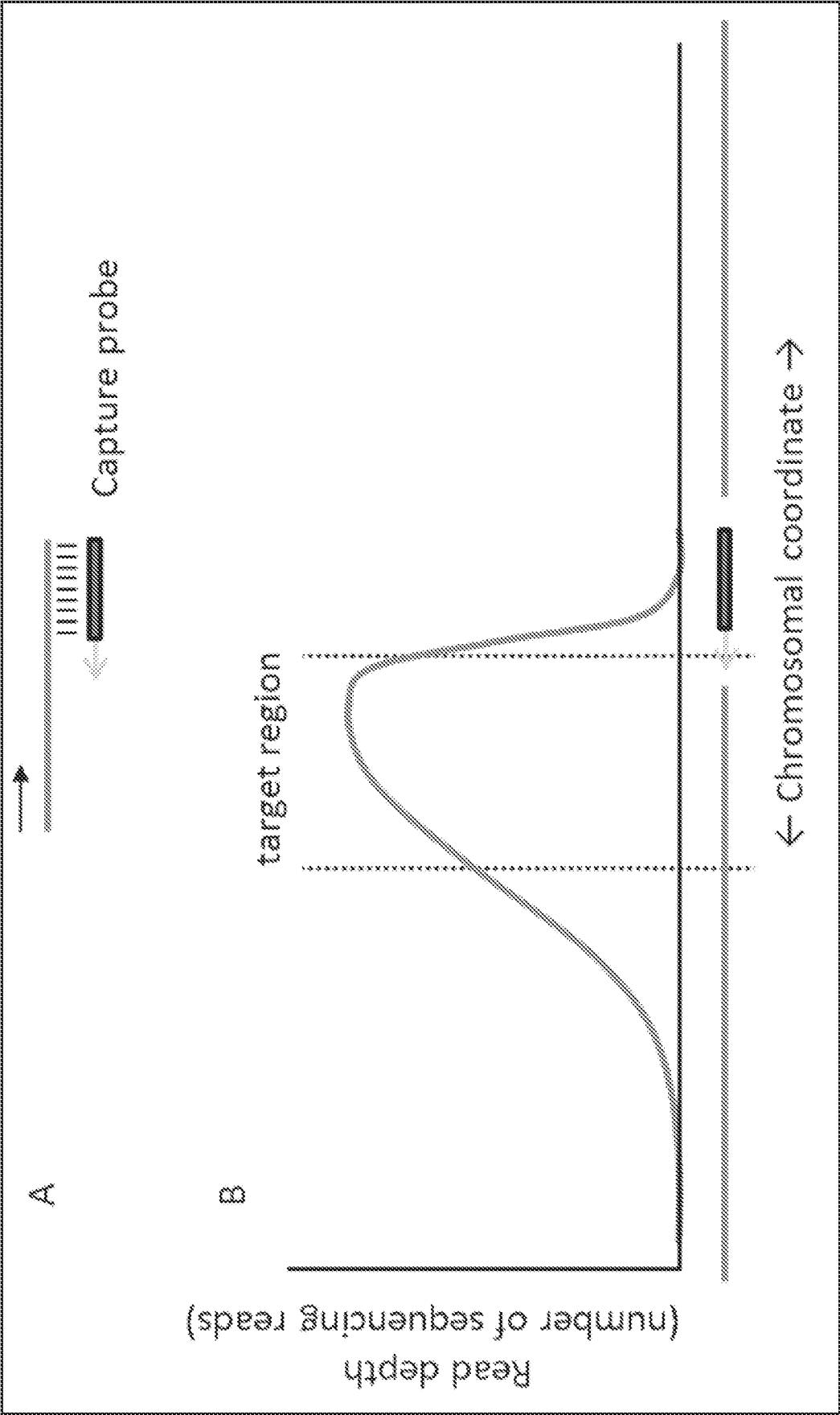


FIG. 5

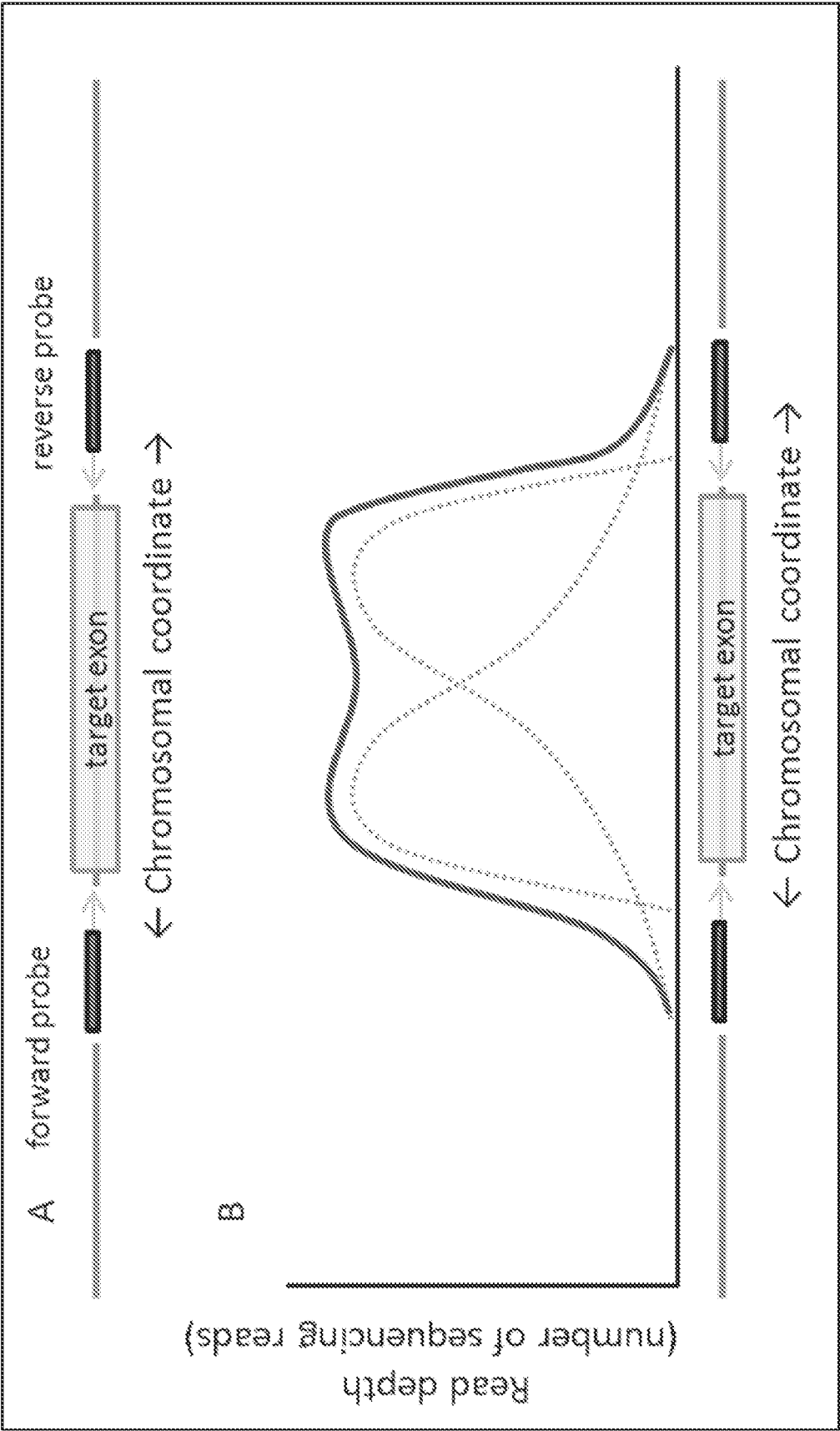


FIG. 6

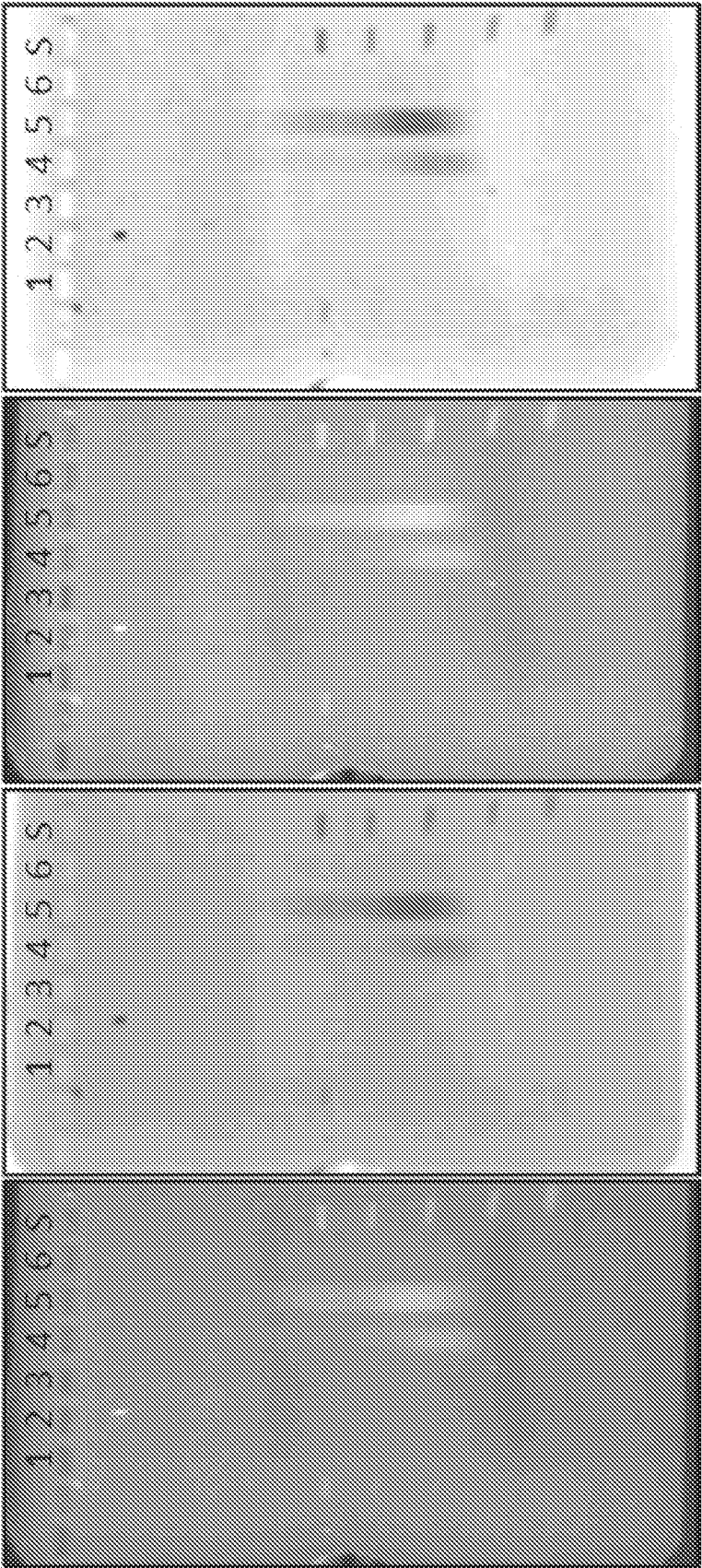


FIG. 7

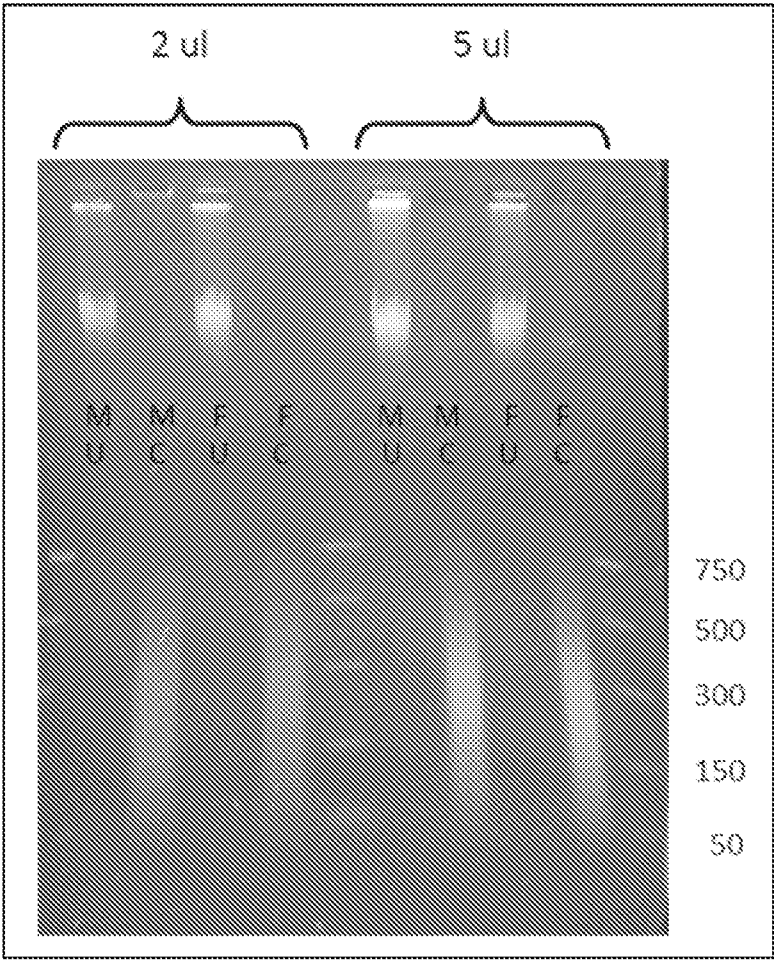


FIG. 8

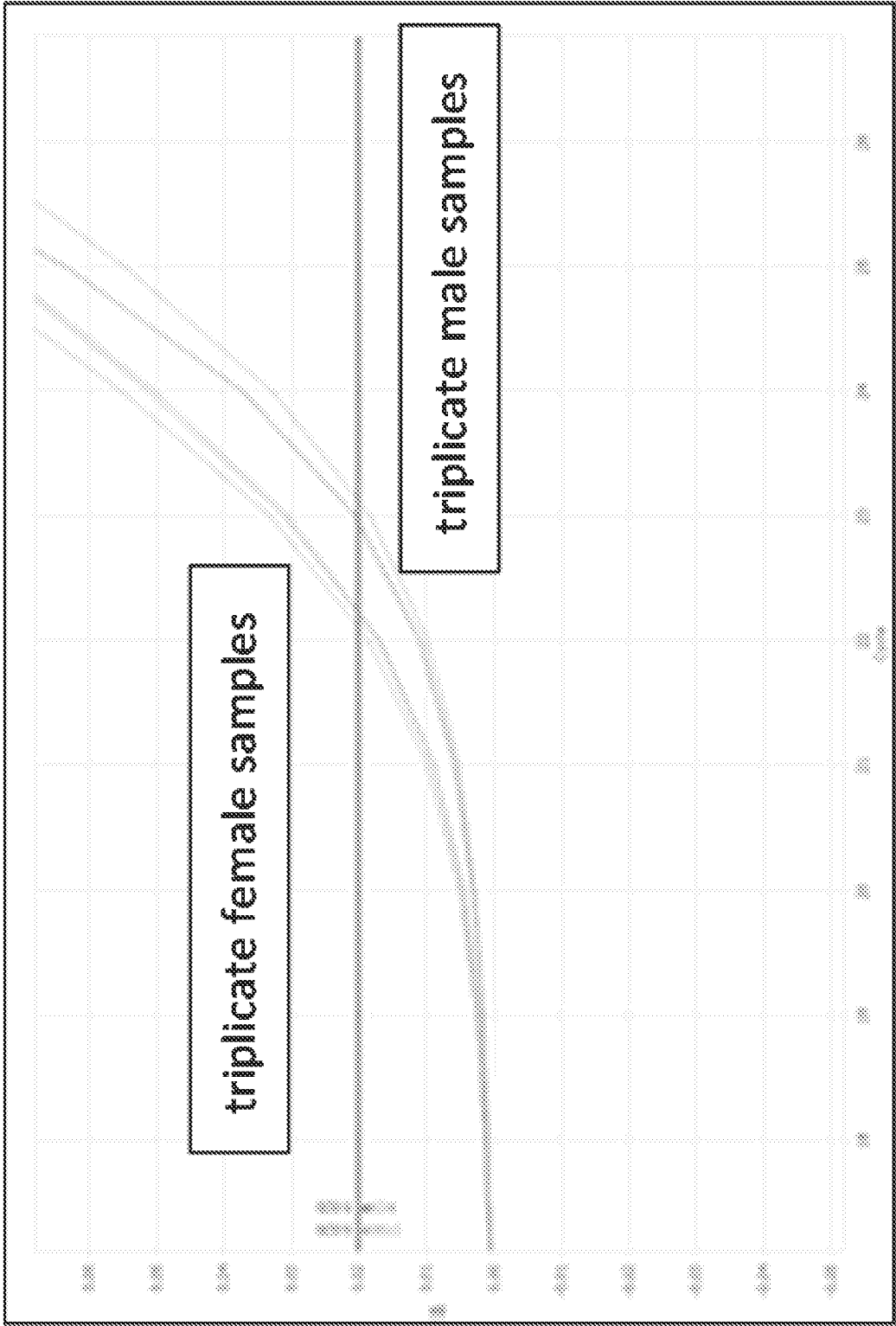


FIG. 9

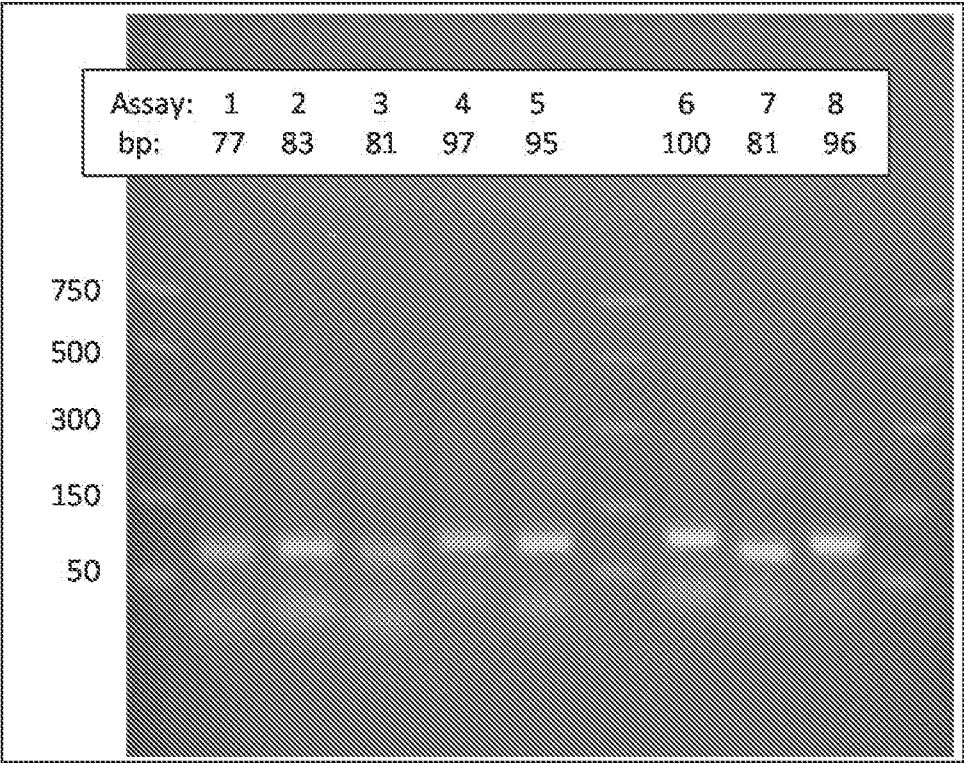


FIG. 10

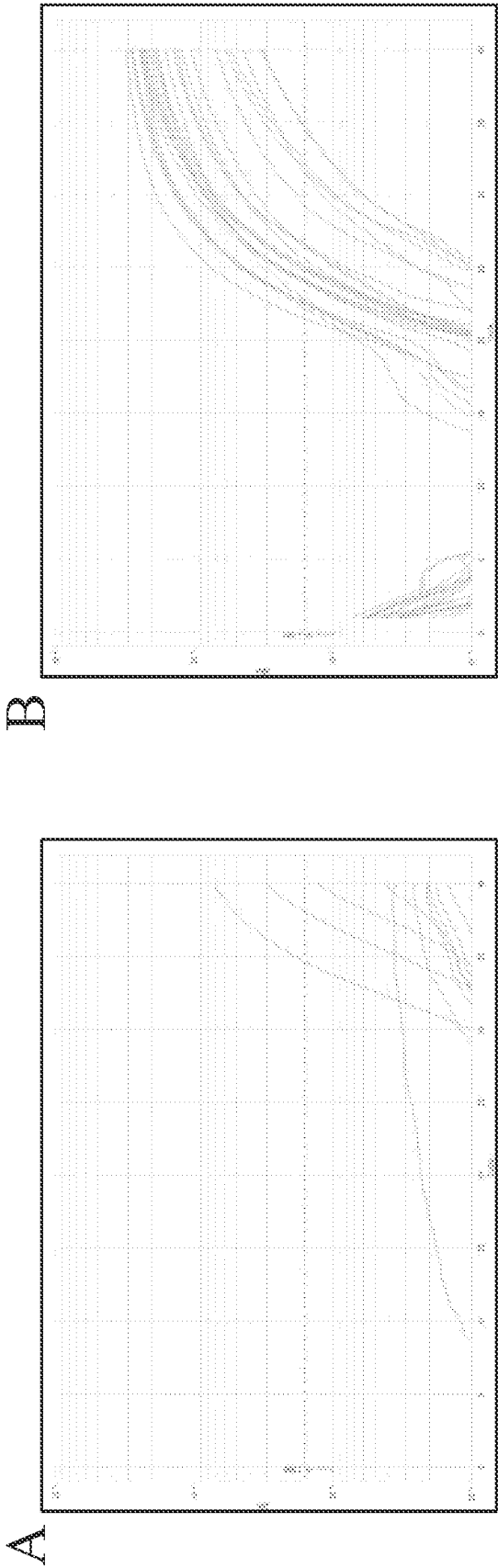


FIG. 11

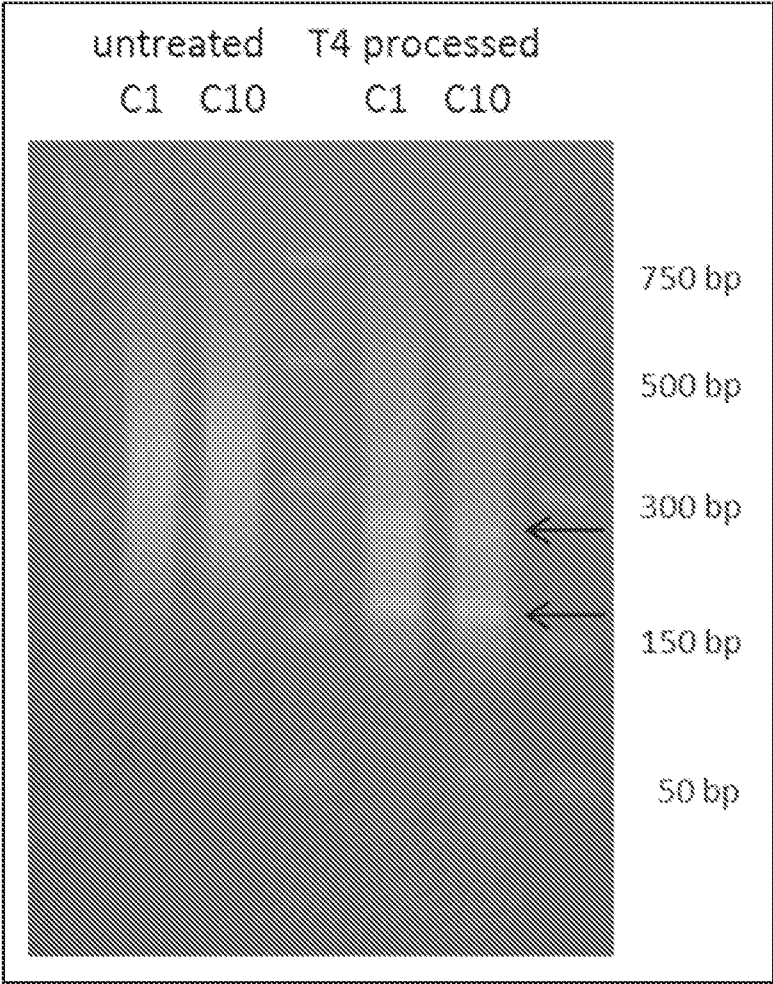


FIG. 12

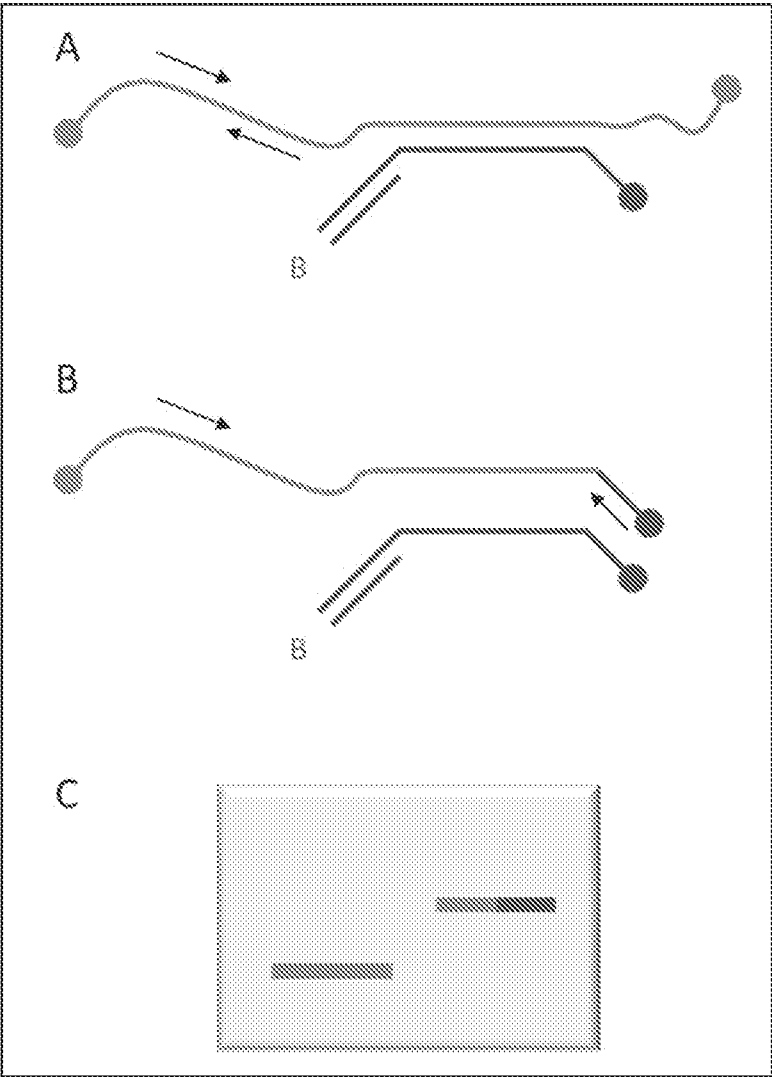
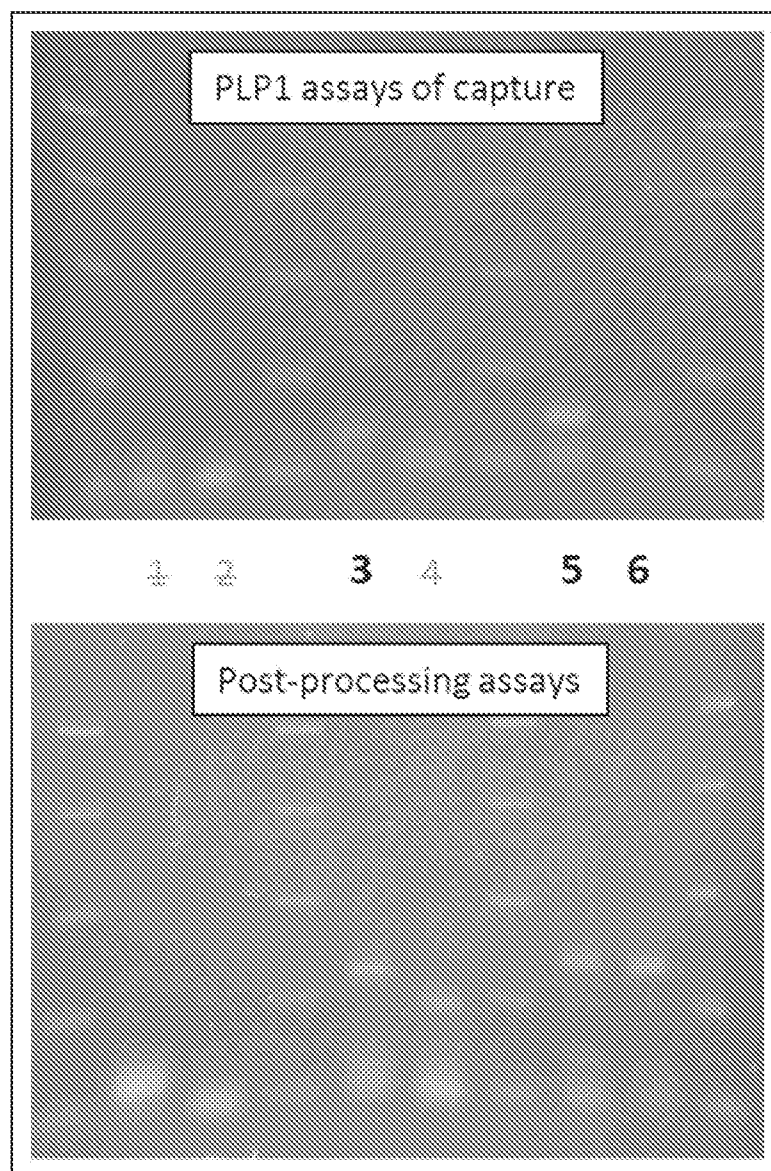


FIG. 13

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*FIG. 14*

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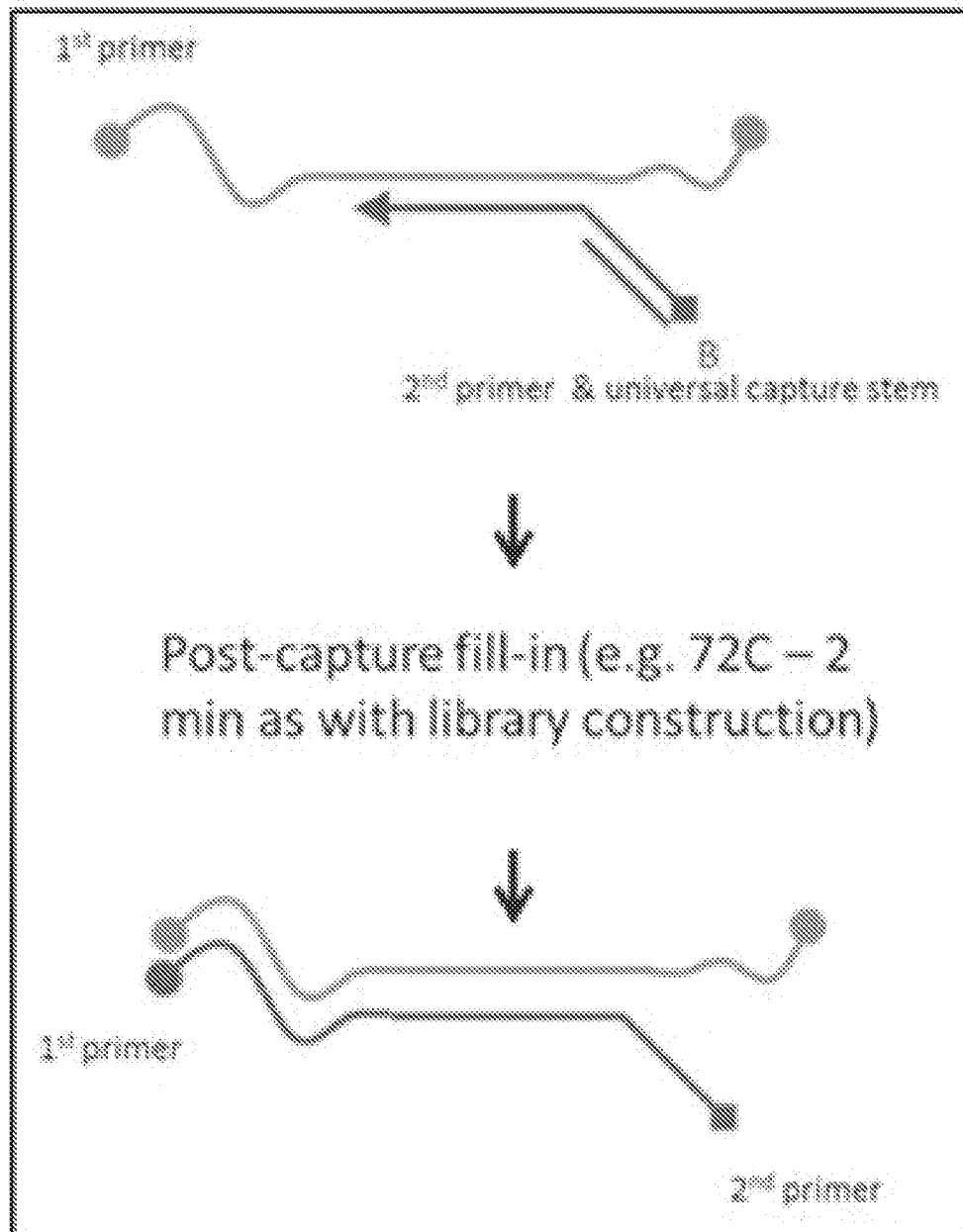
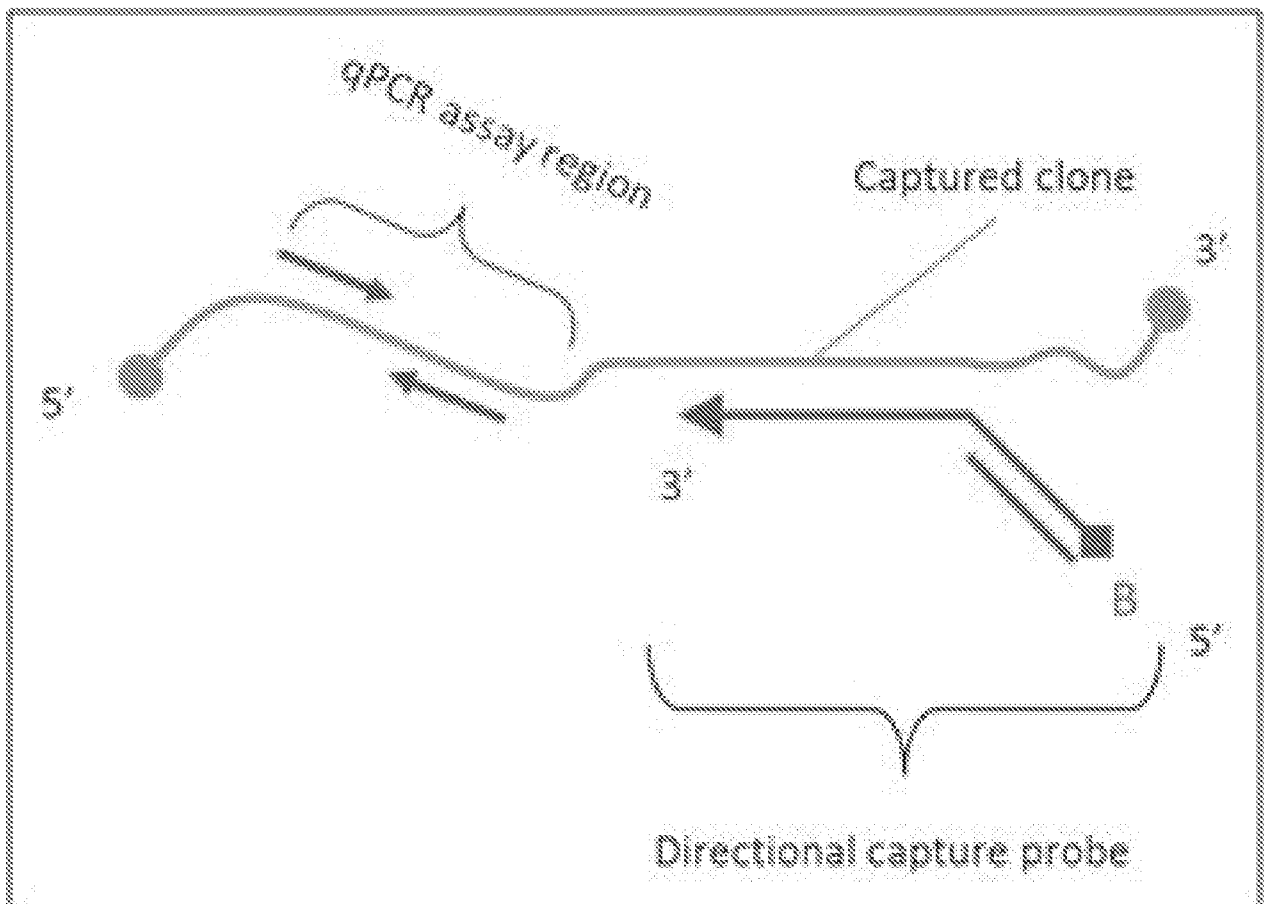


FIG. 15

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*FIG. 16*

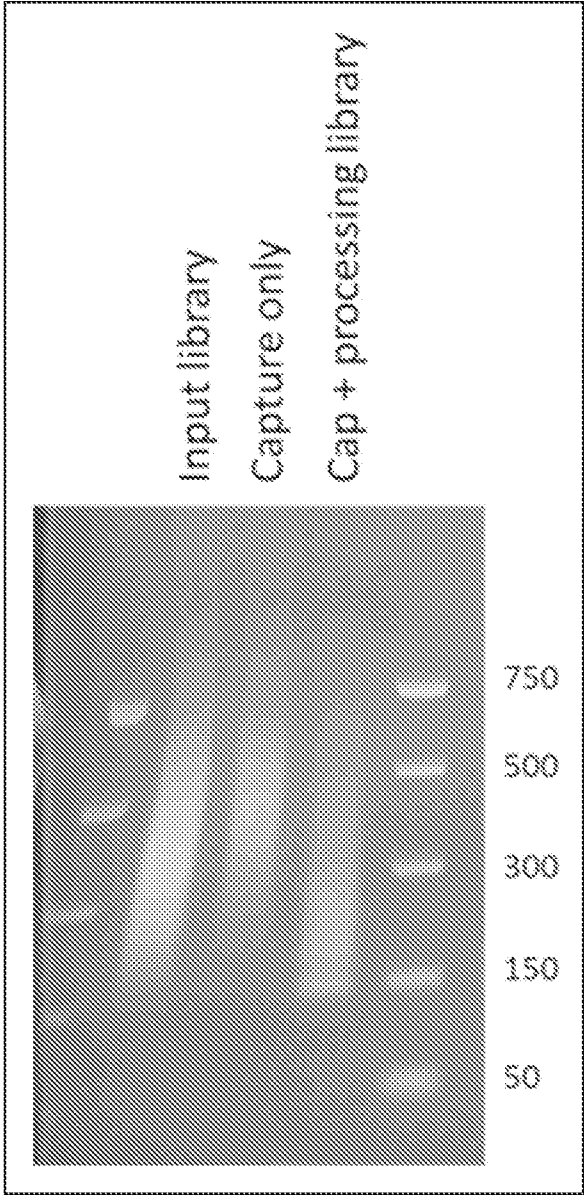
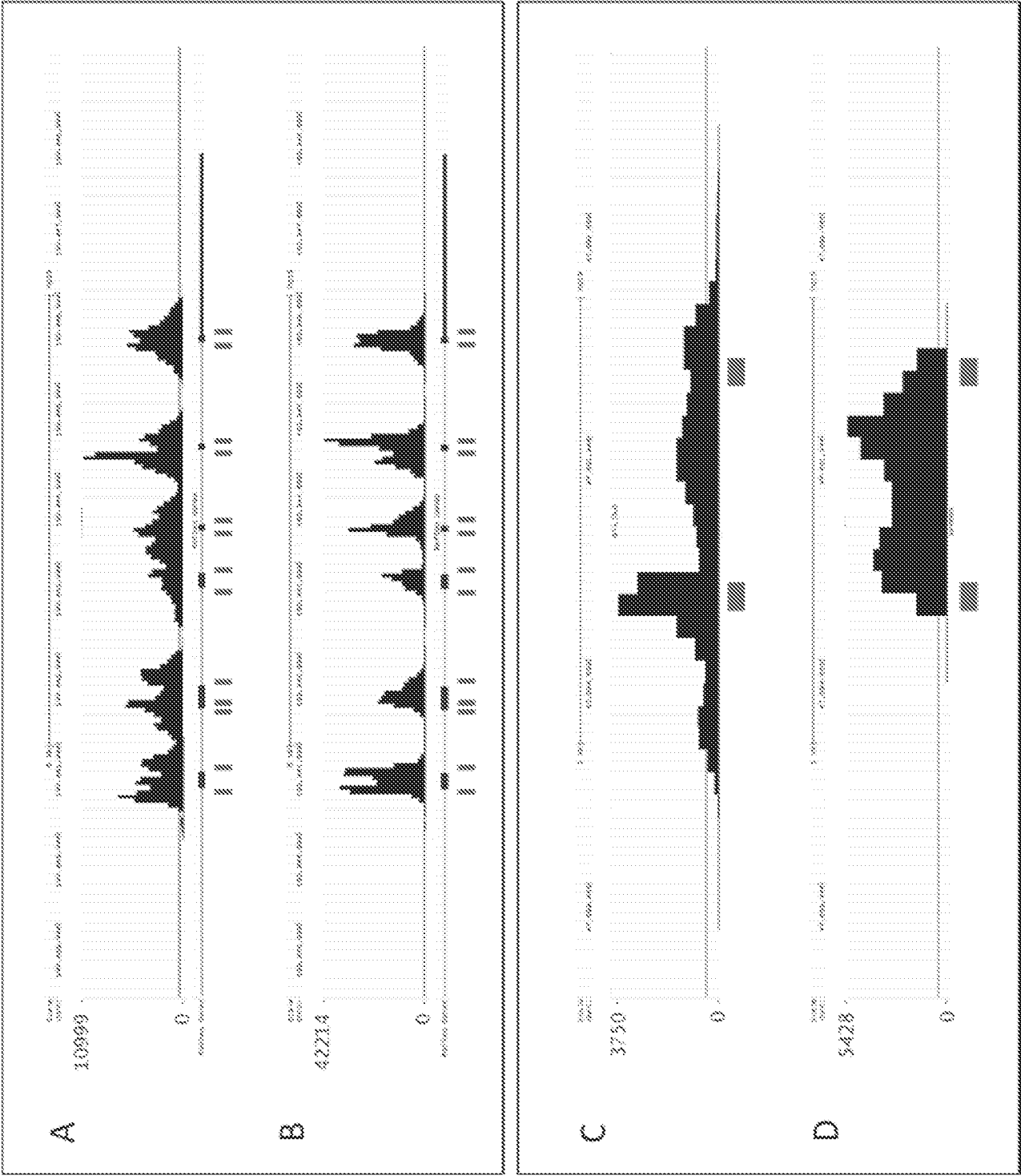


FIG. 17



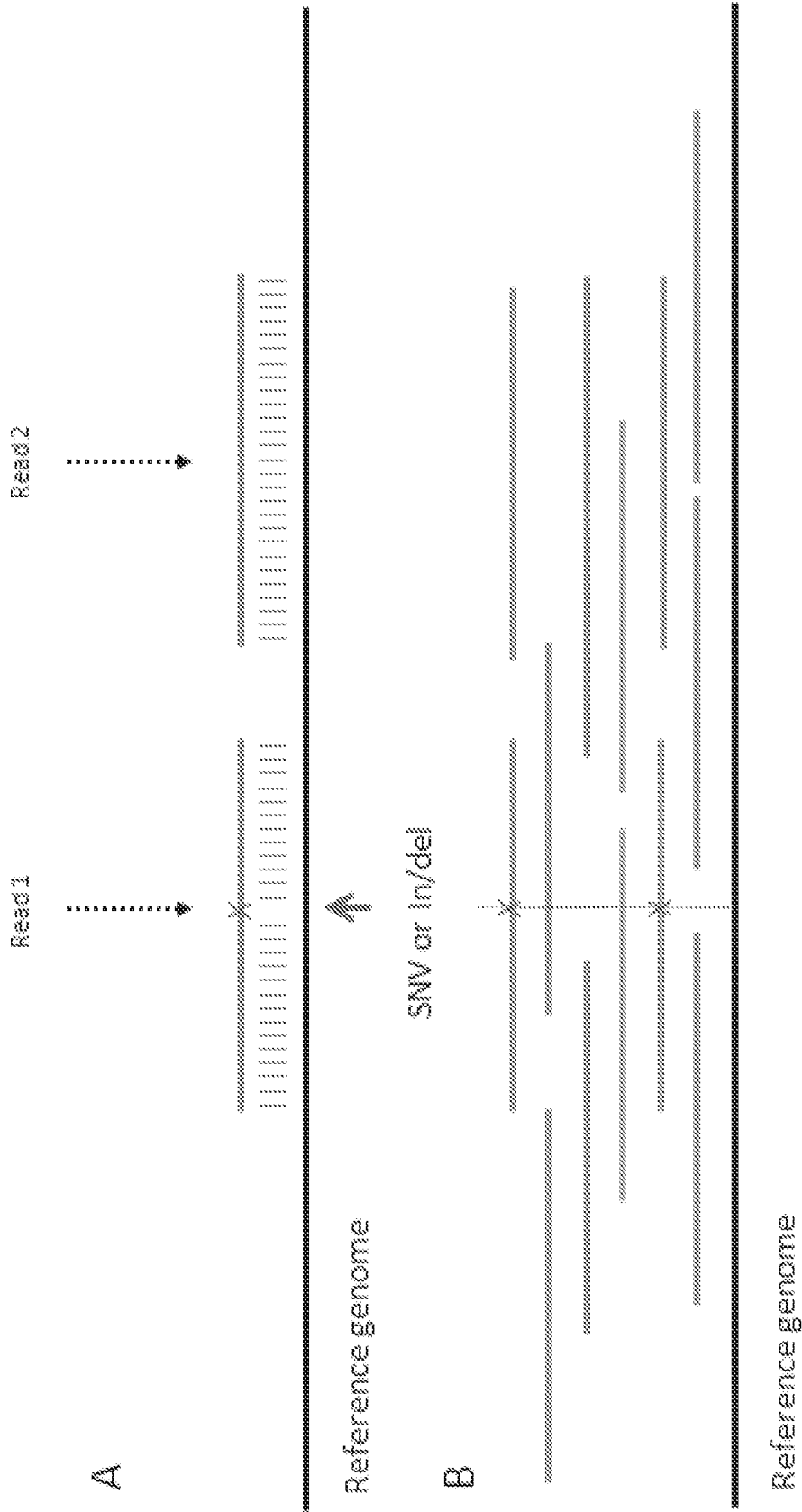


FIG. 19

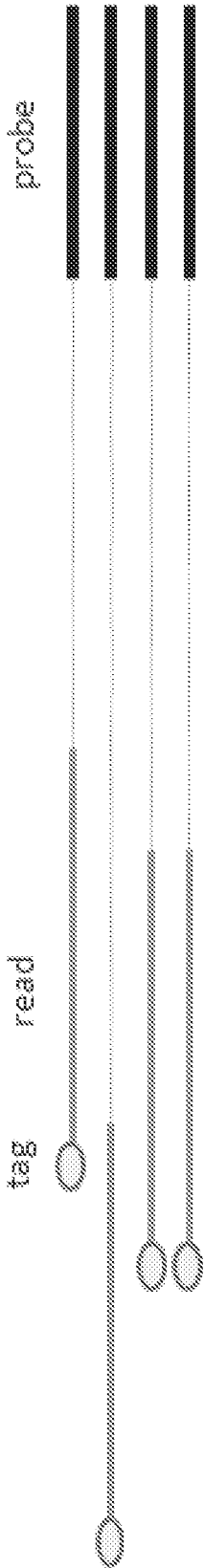


FIG. 20

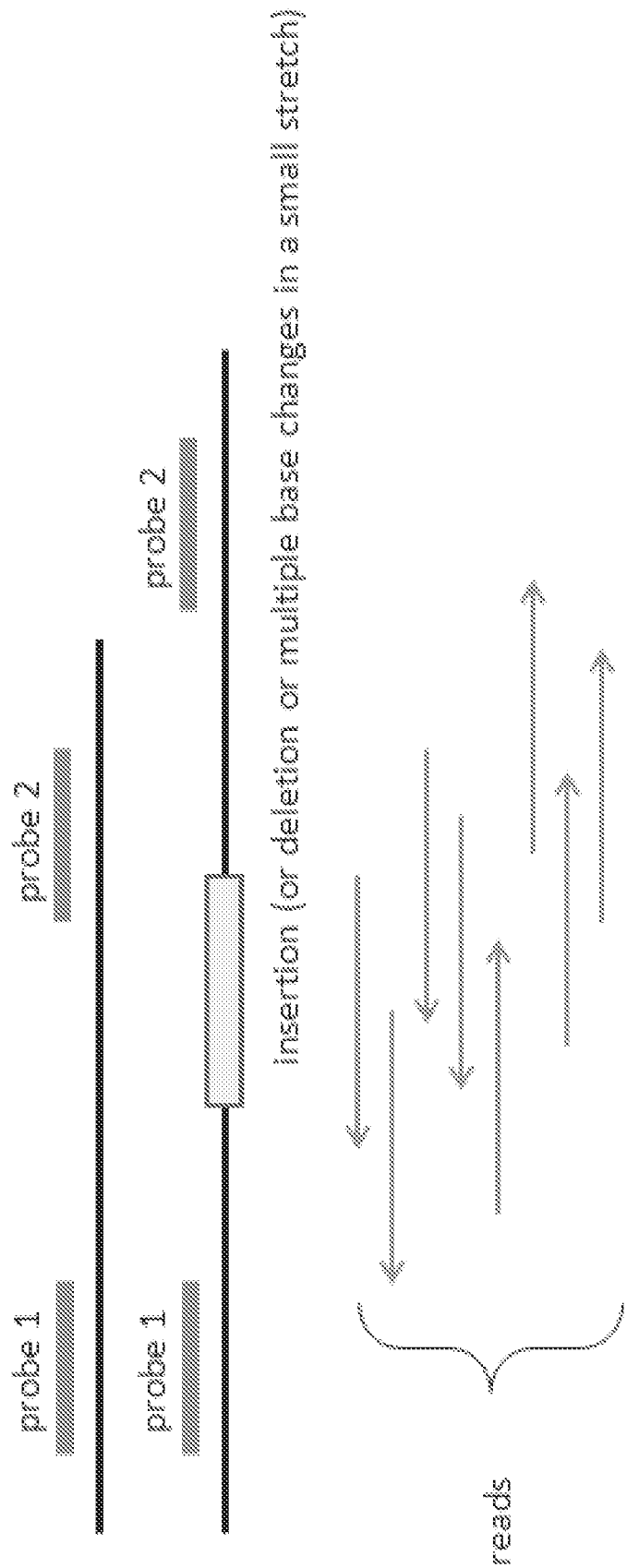


FIG. 21

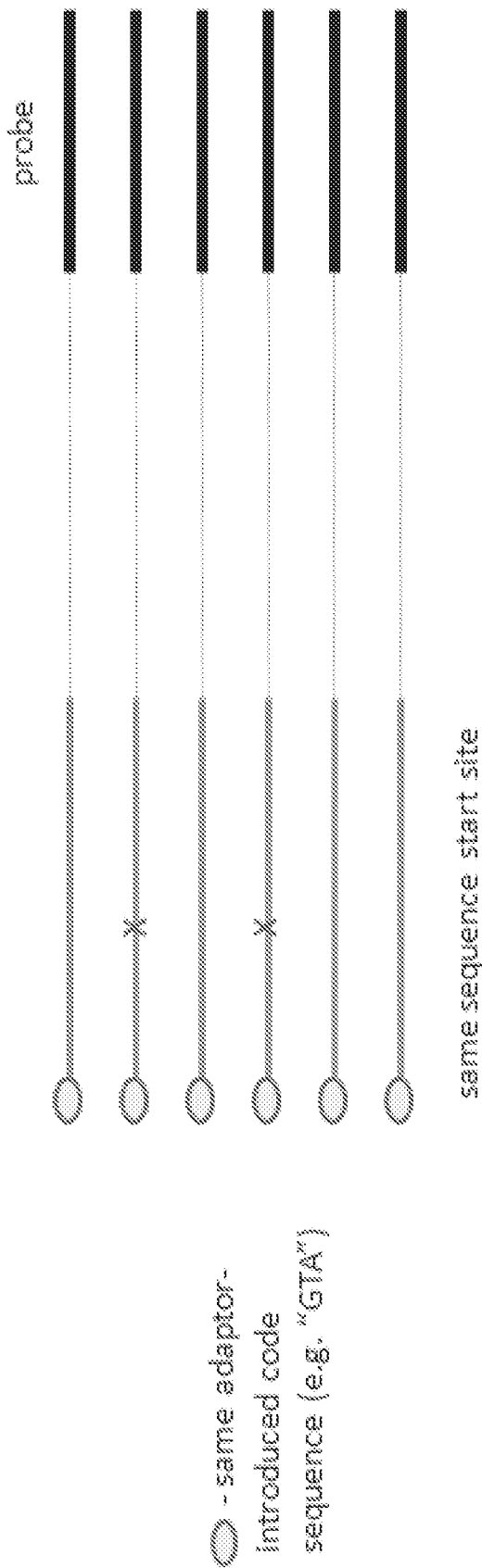


FIG. 22

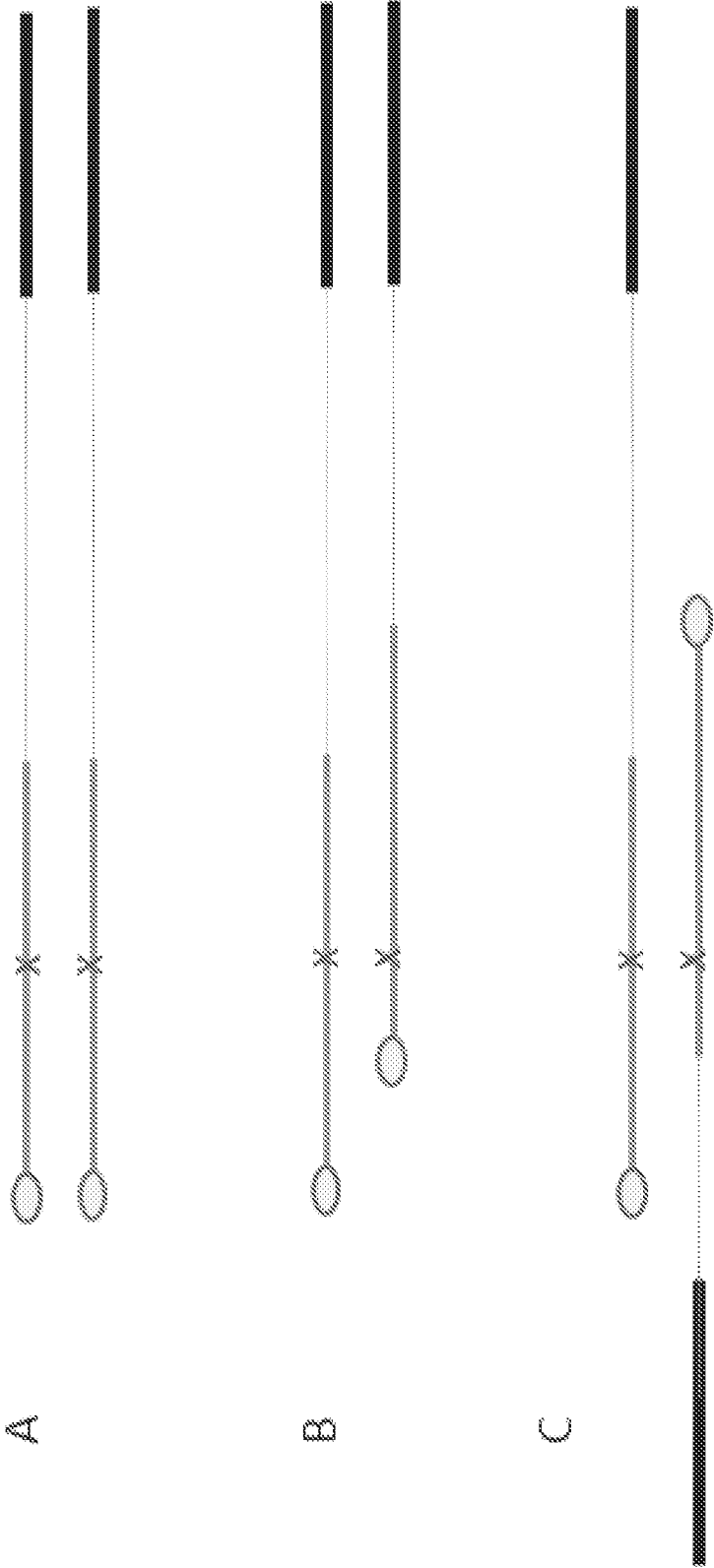


FIG. 23

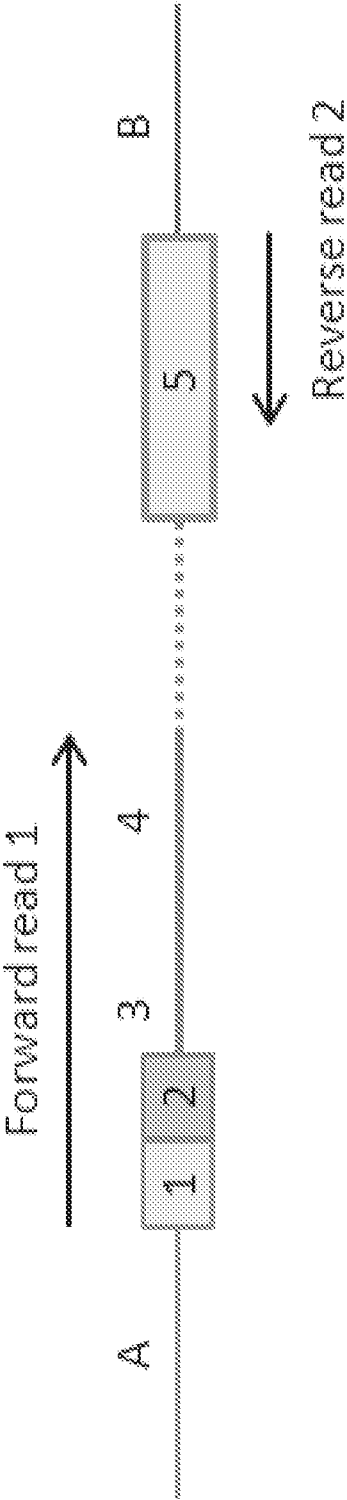


FIG. 24

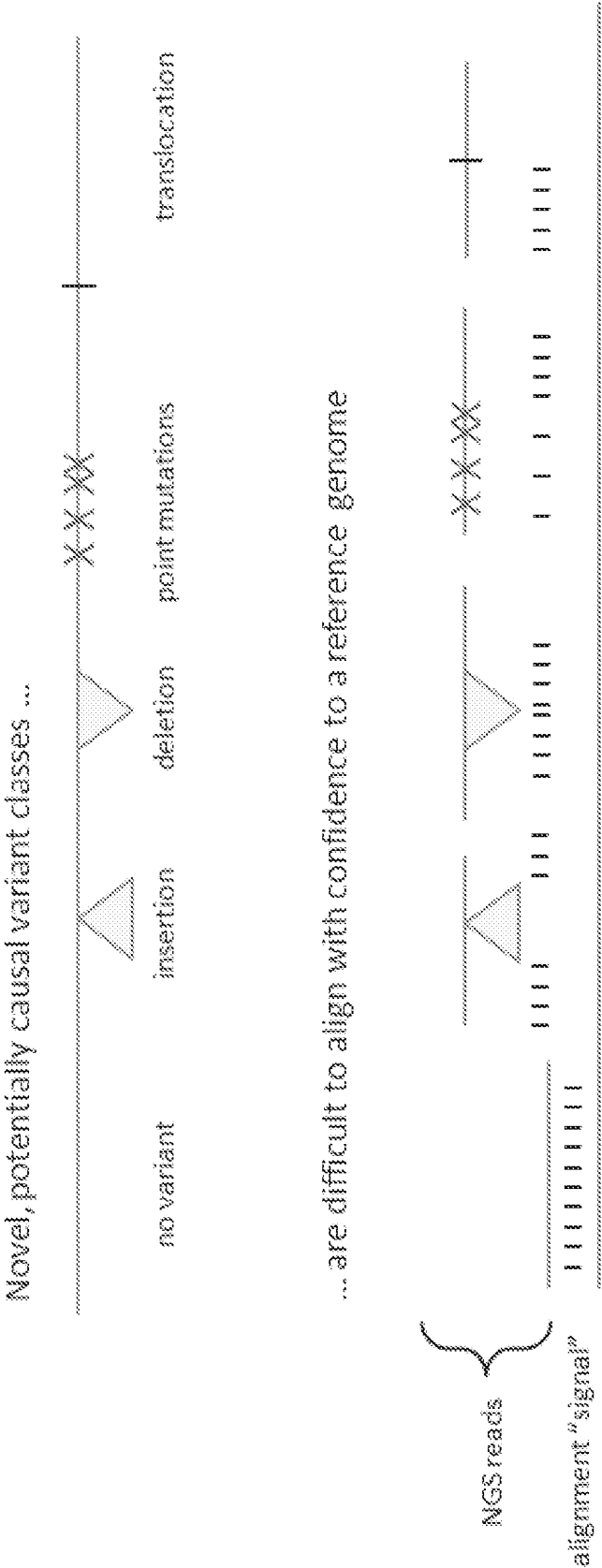


FIG. 25

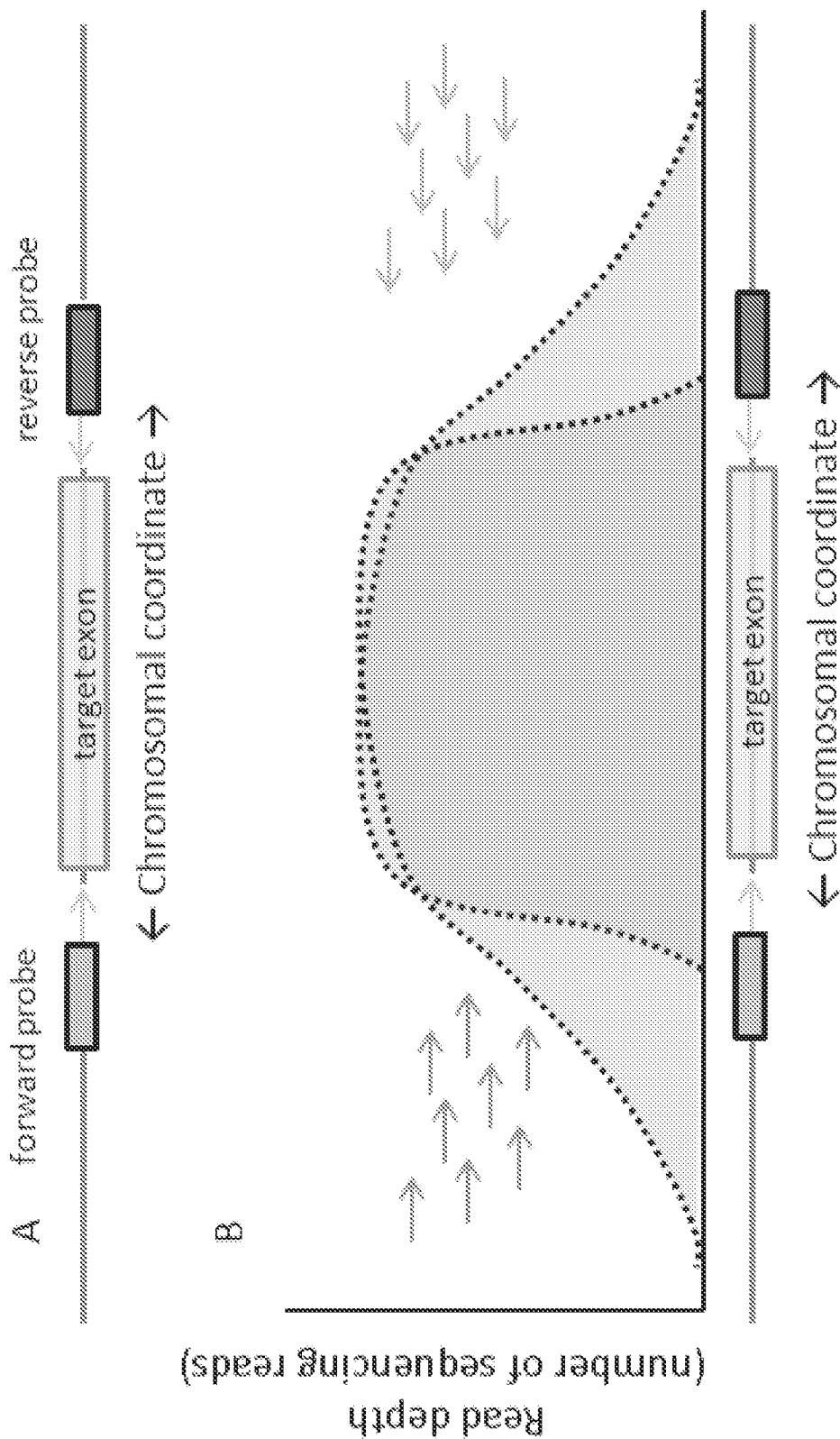


FIG. 26

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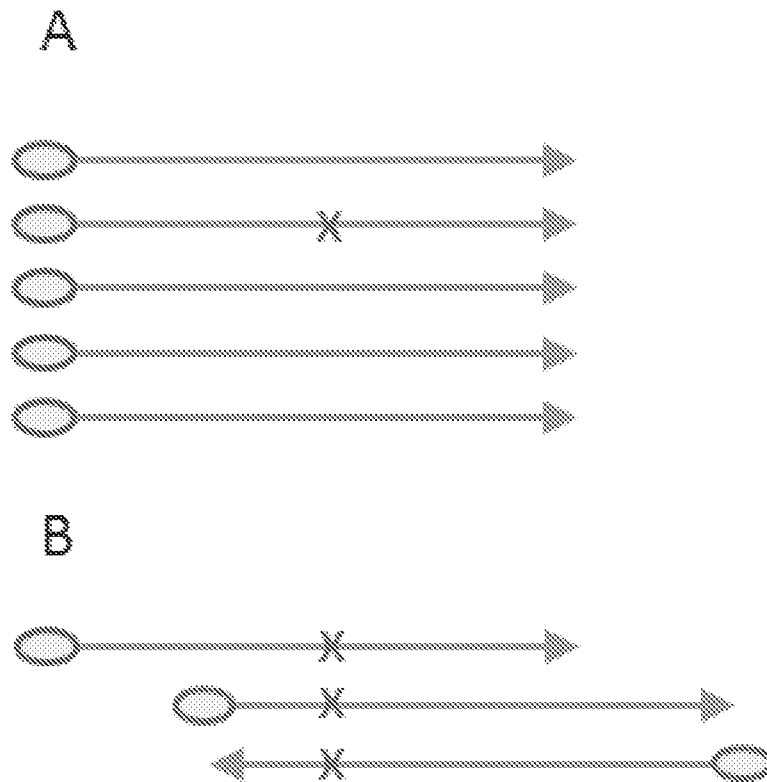
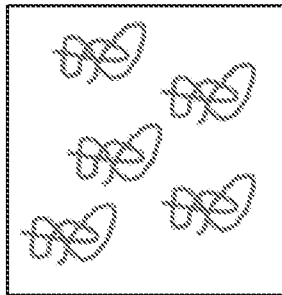
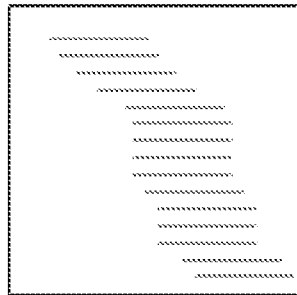


FIG. 27

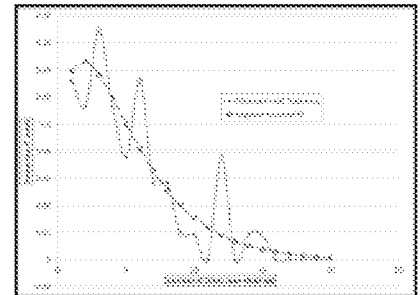
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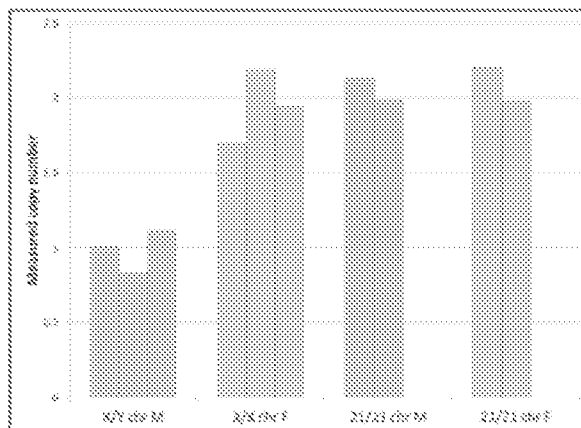
Low input genome libraries (e. g. ~50-100 genome equivalents) created with molecular annotation and subsequently captured with targeting probes.



Variant filtering for true positive read set on a probe-by-probe basis. This allows the number and frequency of observed unique reads to be determined.



Observed read frequencies are fitted to a negative binomial distribution, which allows the "inferred class" of unobserved reads to be estimated. The number of observed reads + inferred reads = copy number.



Representative copy determination at three probes for the X-chromosomal gene PLP1 in a male and a female sample. For comparison, two probes for the autosomal chromosome 21 gene TNFRSF14 gene are also shown.

FIG. 28

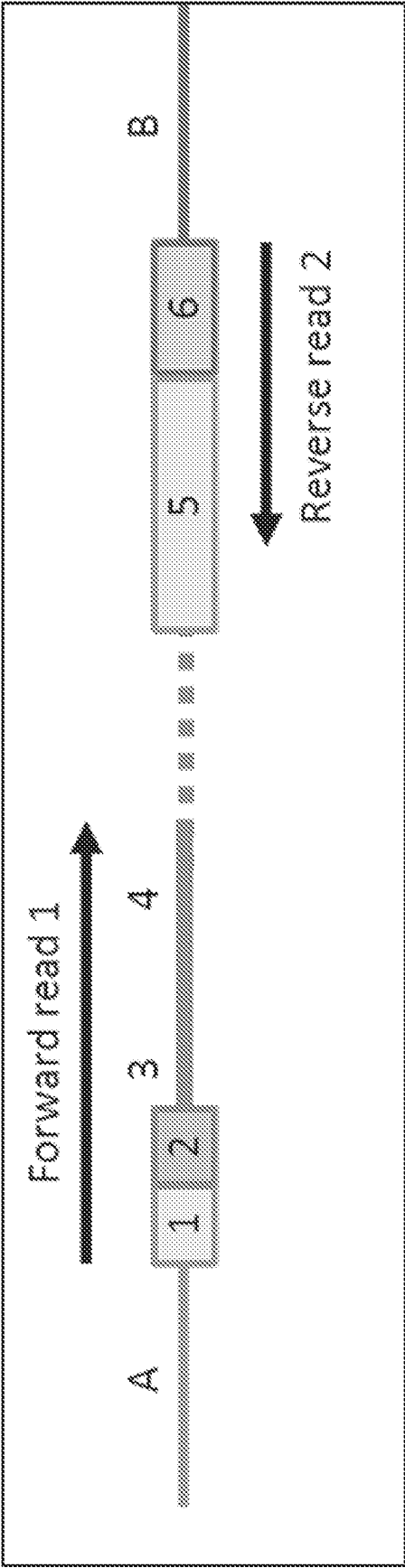


FIG. 29

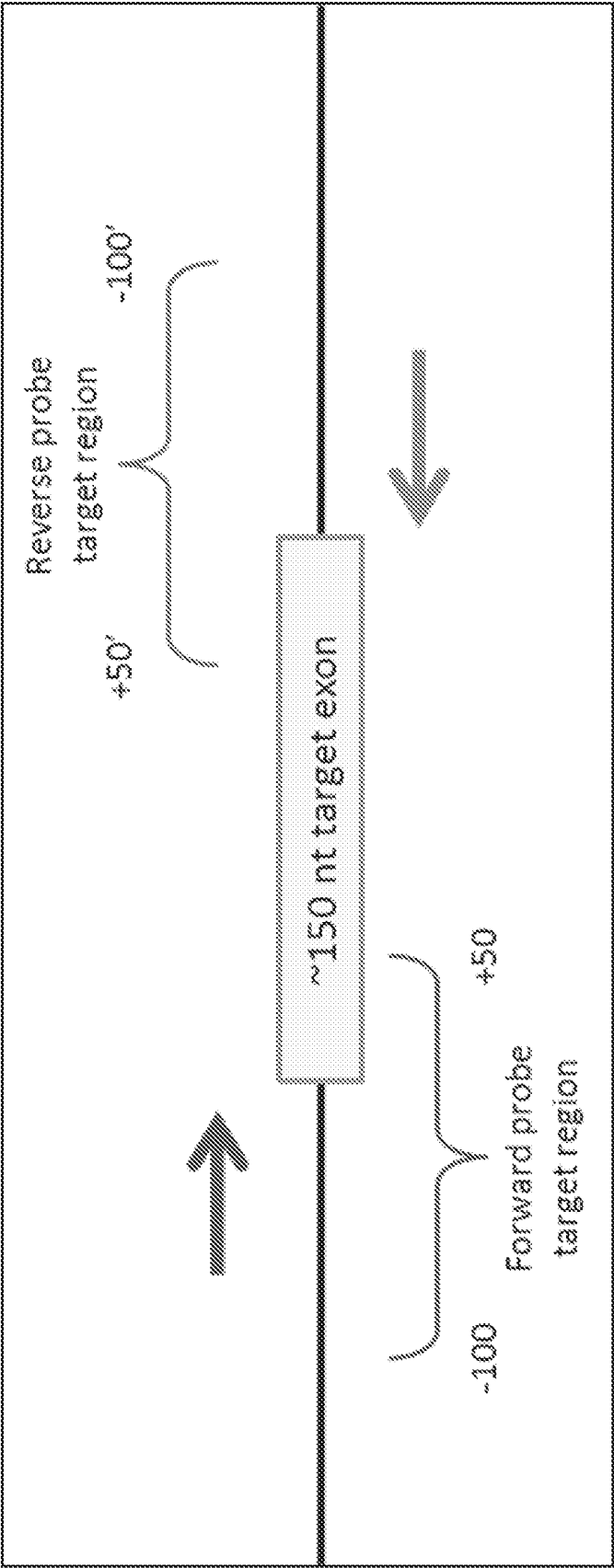


FIG. 30

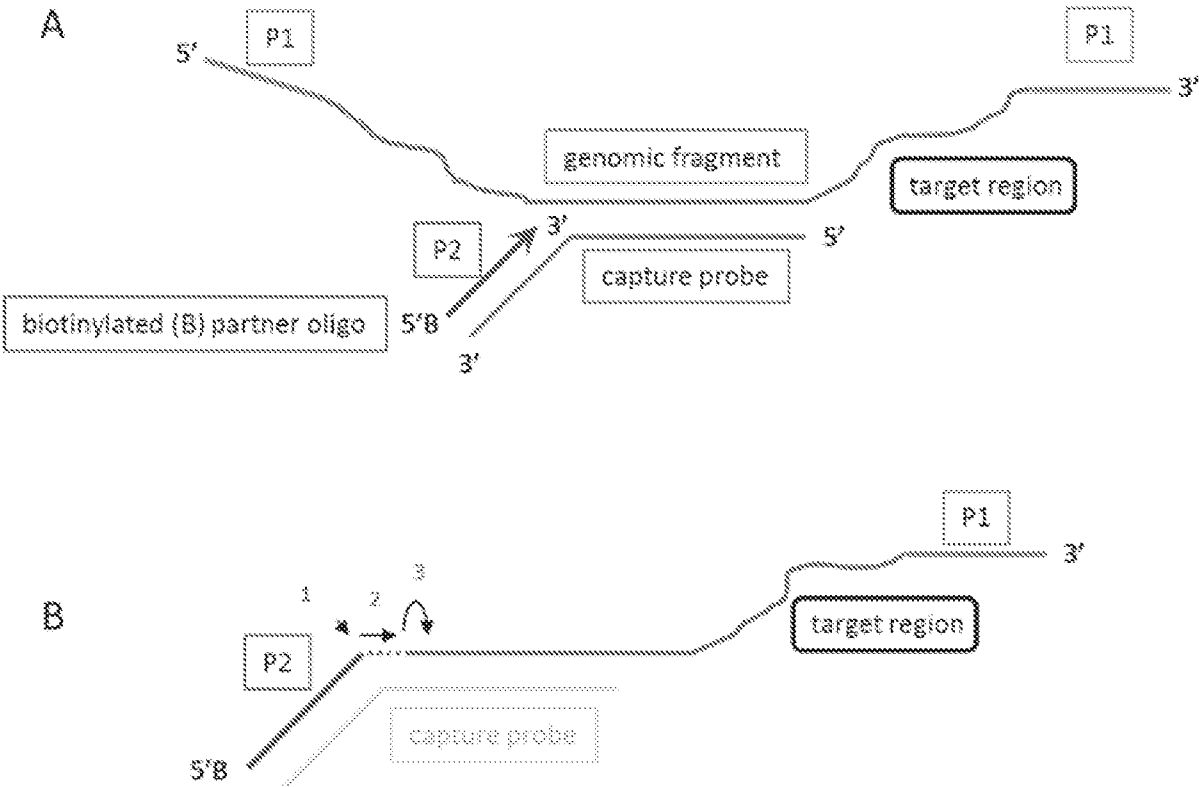


FIG. 31

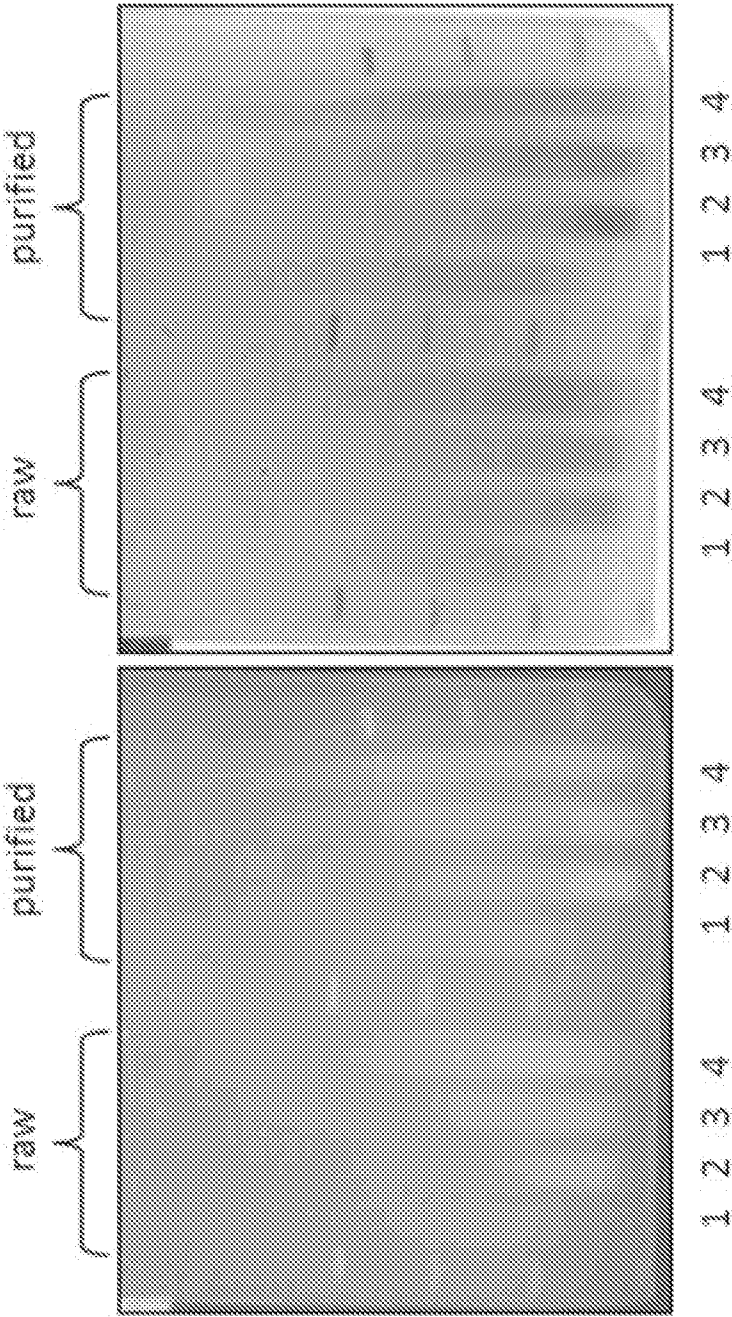


FIG. 32

Step 1 -- fragment gDNA to ~400 bp
or
perform 1st strand cDNA synthesis with (N)₁₅ in the presence of ddNTPs
please see slide 4 for special comments about cDNA profiling

Step 2 -- melt gDNA
anneal gDNA or cDNA with labeled capture probes, purify

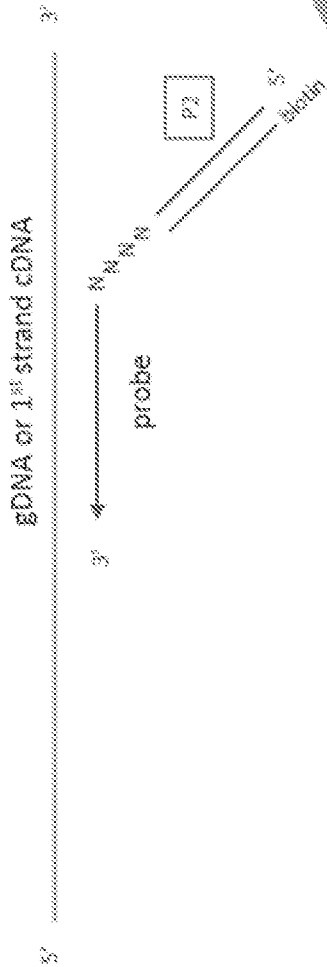


FIG. 33

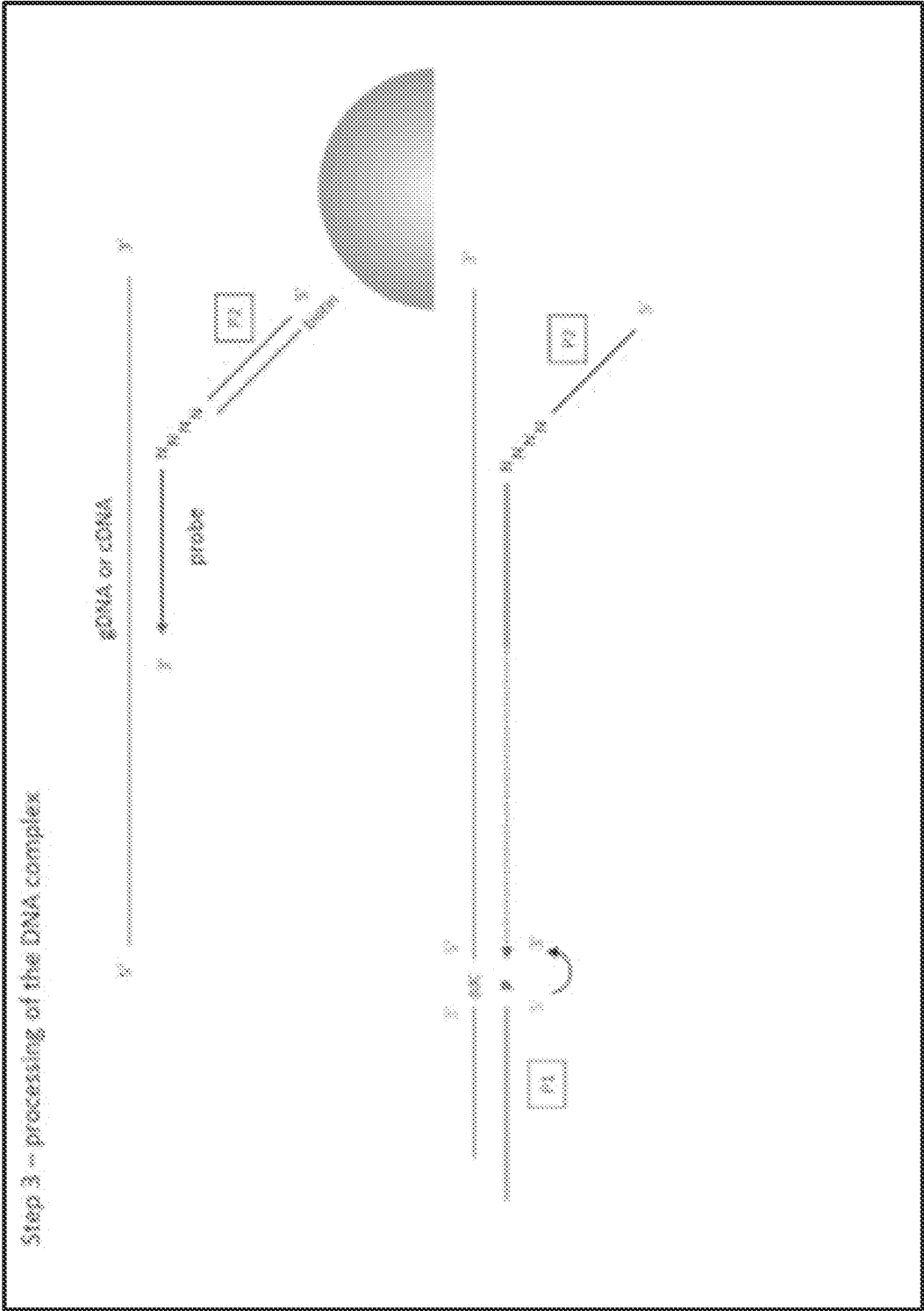


FIG. 34

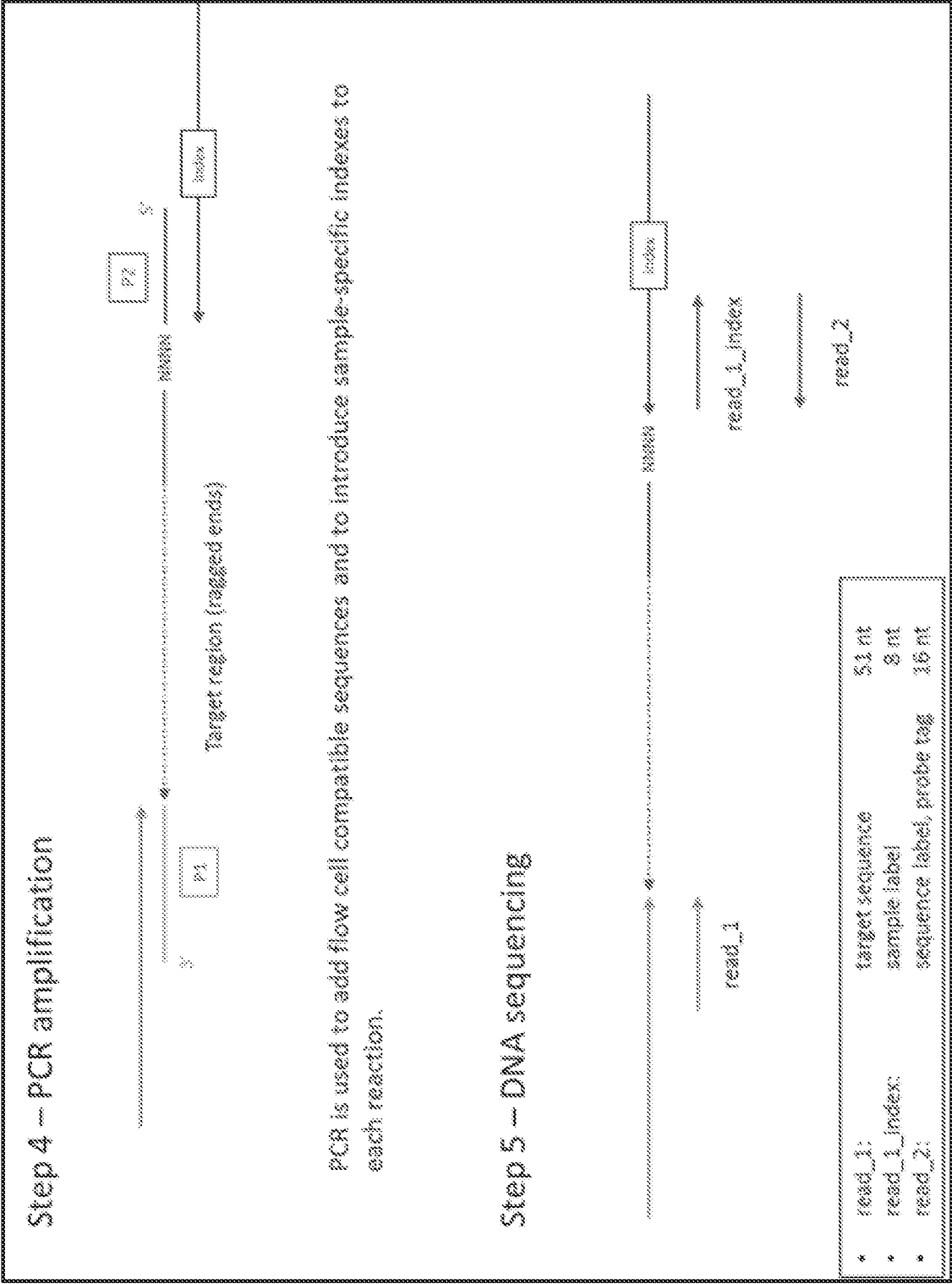


FIG. 35

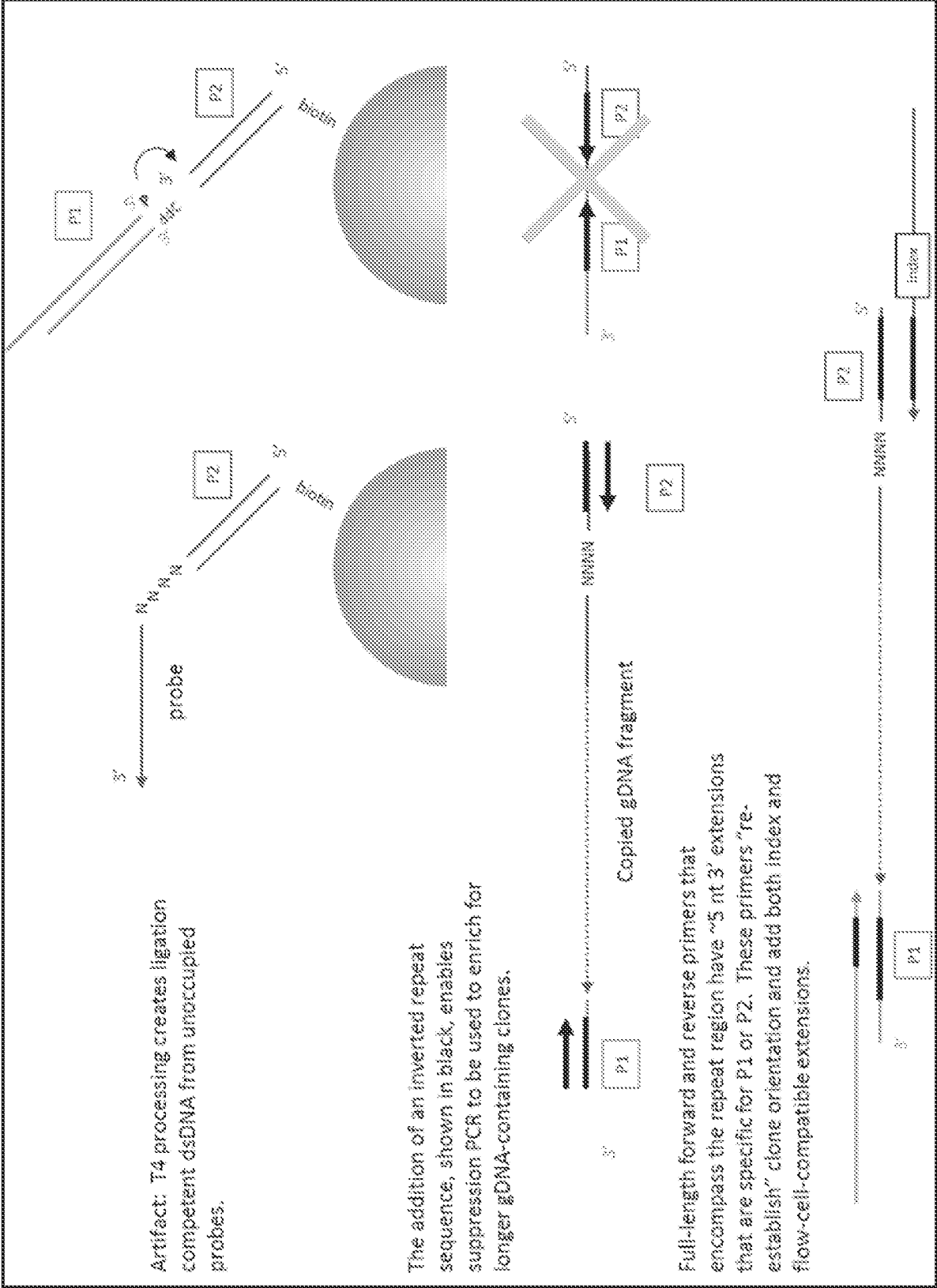


FIG. 36

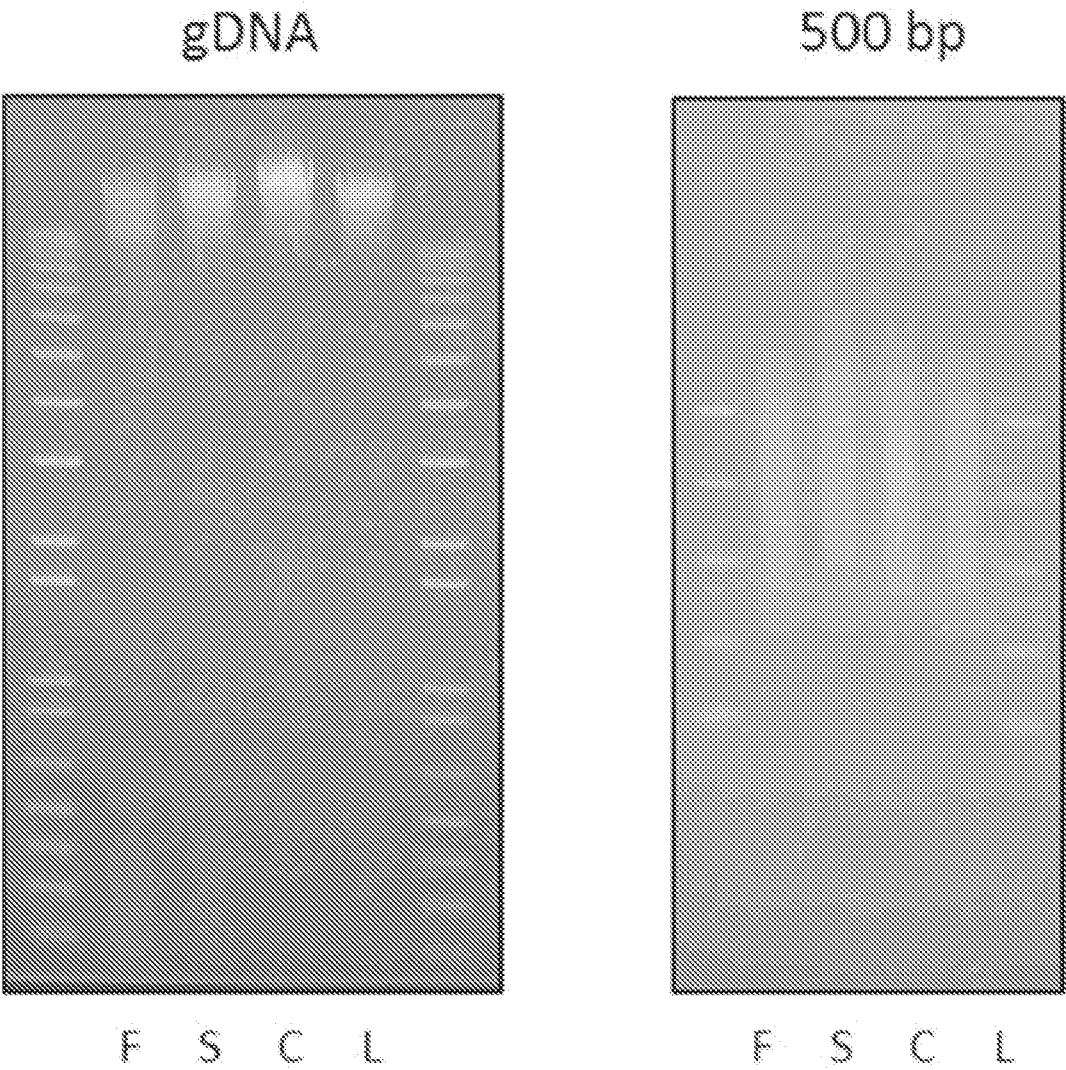


FIG. 37

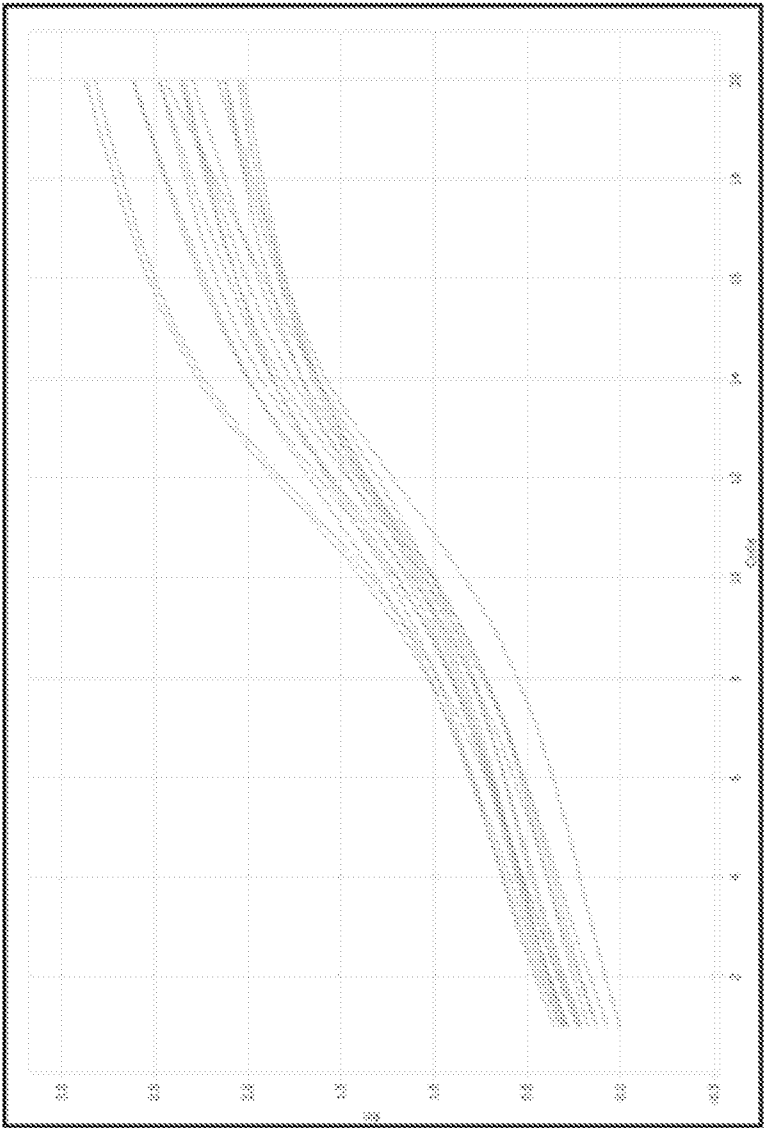
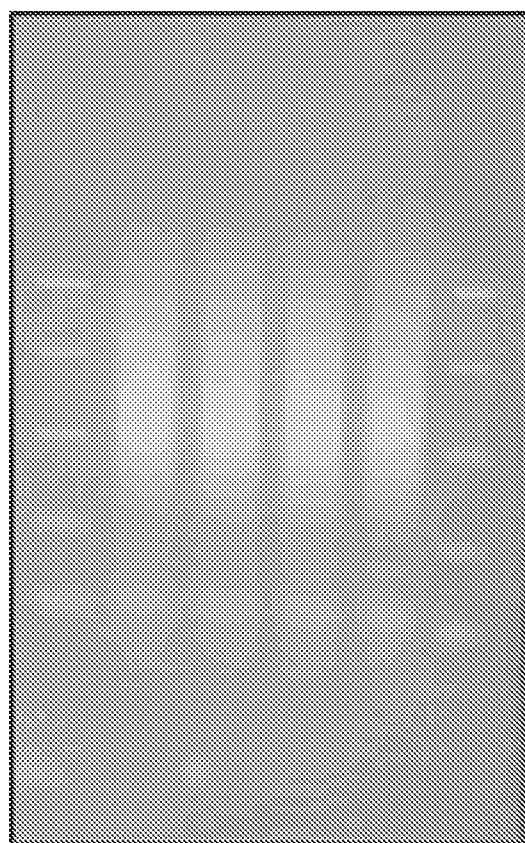


FIG. 38

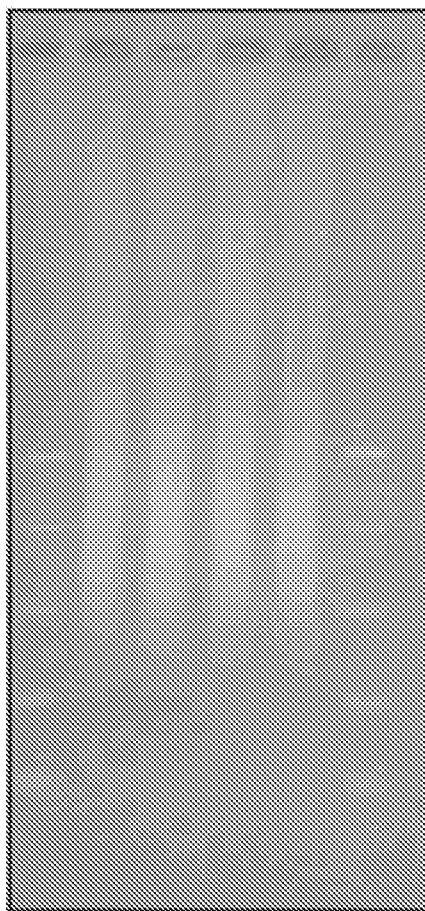
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F S C L

FIG. 39

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F S C L

FIG. 40

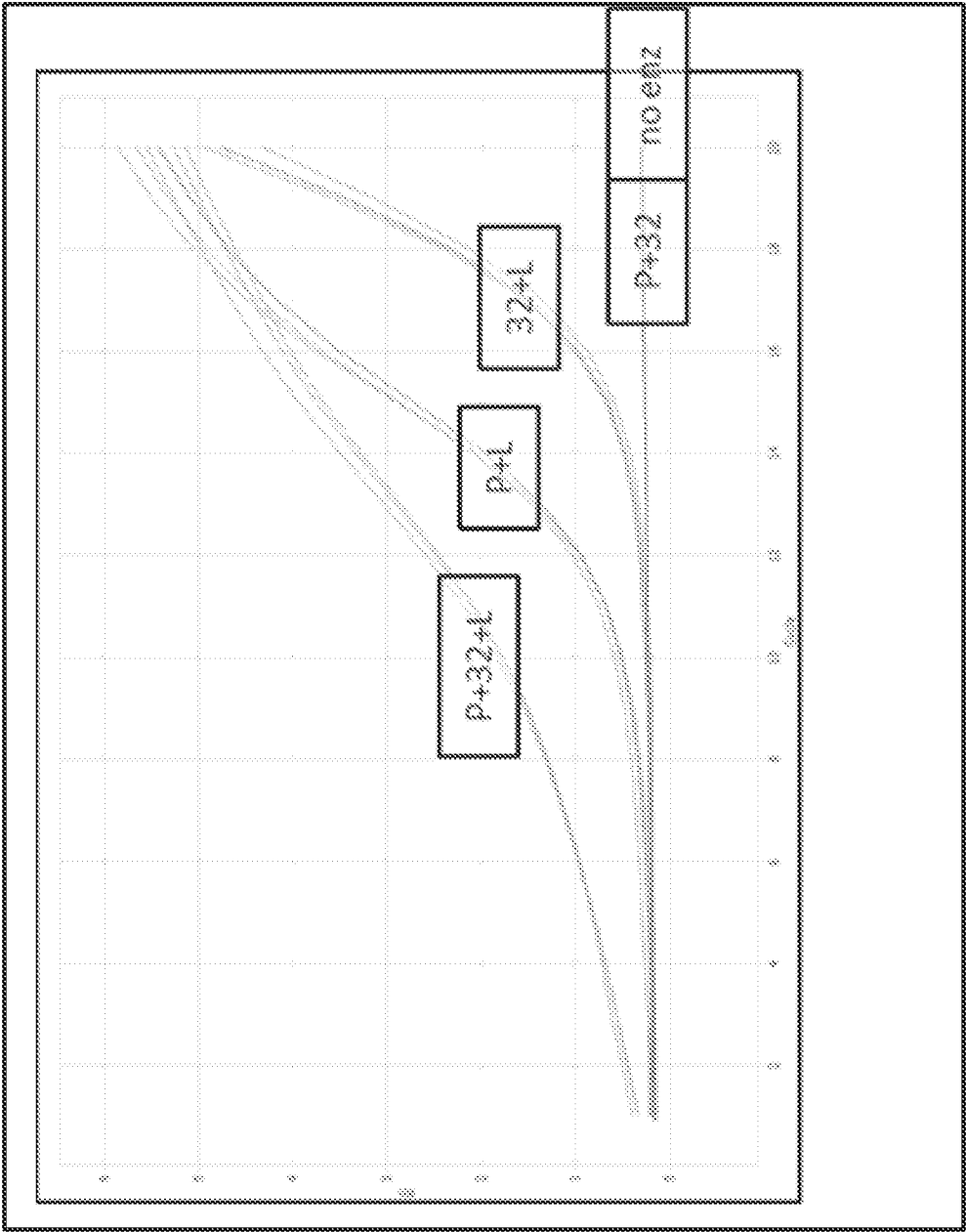


FIG. 41

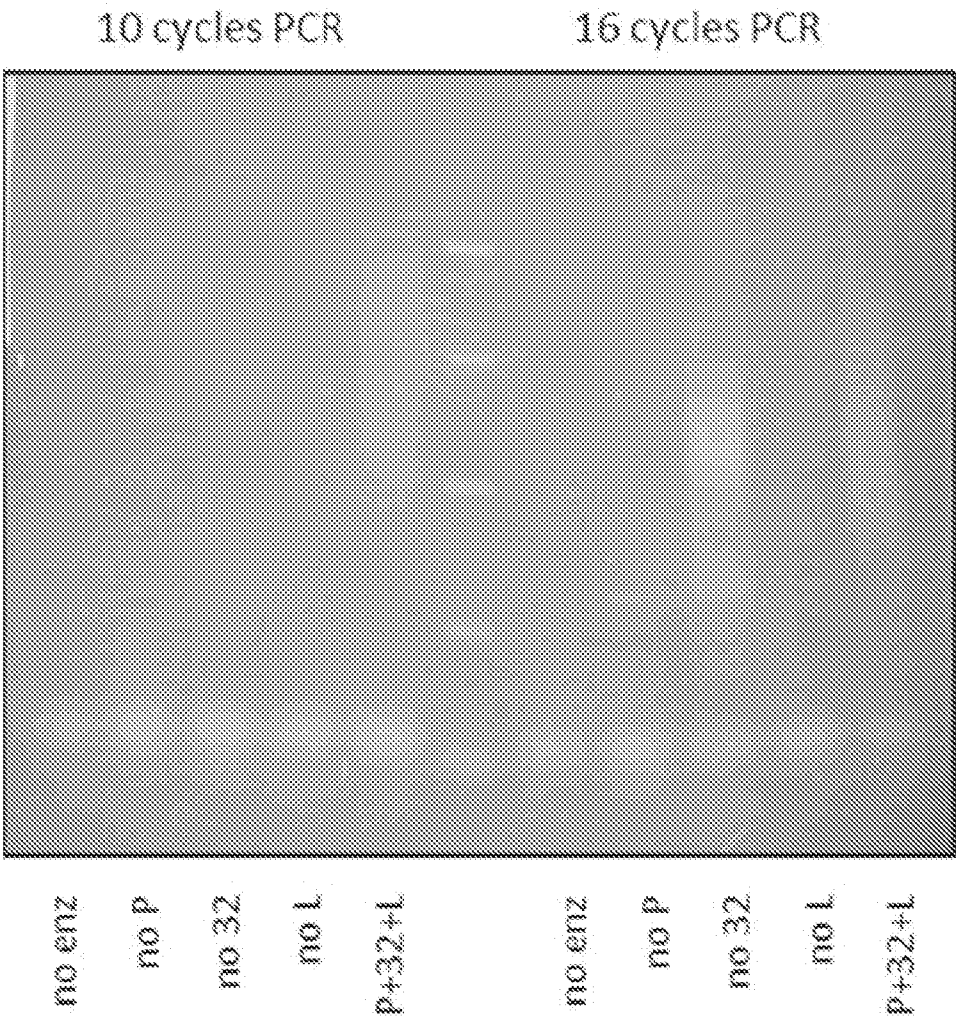
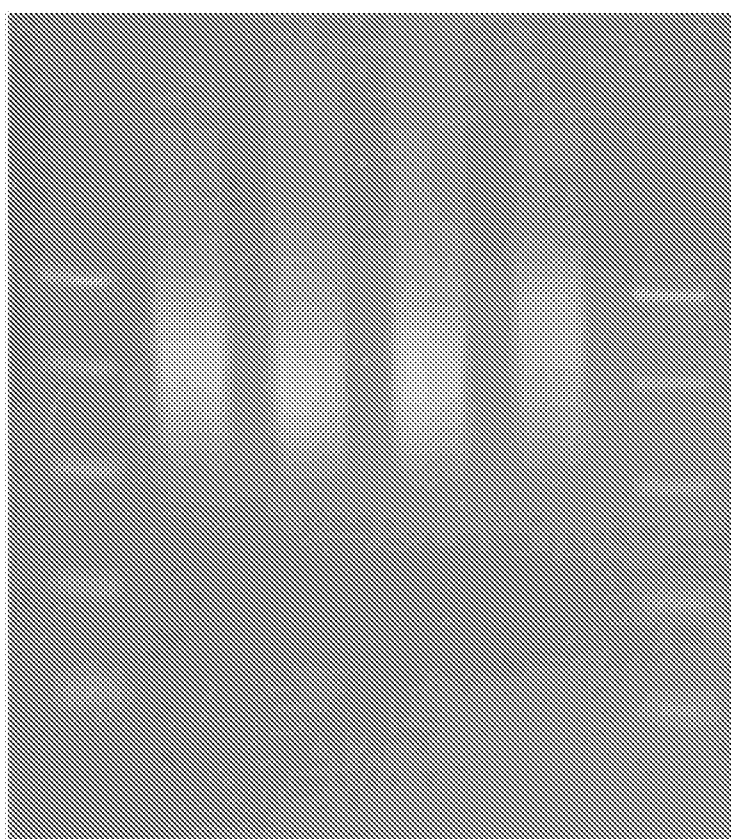


FIG. 42

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11 13 15 16

FIG. 43

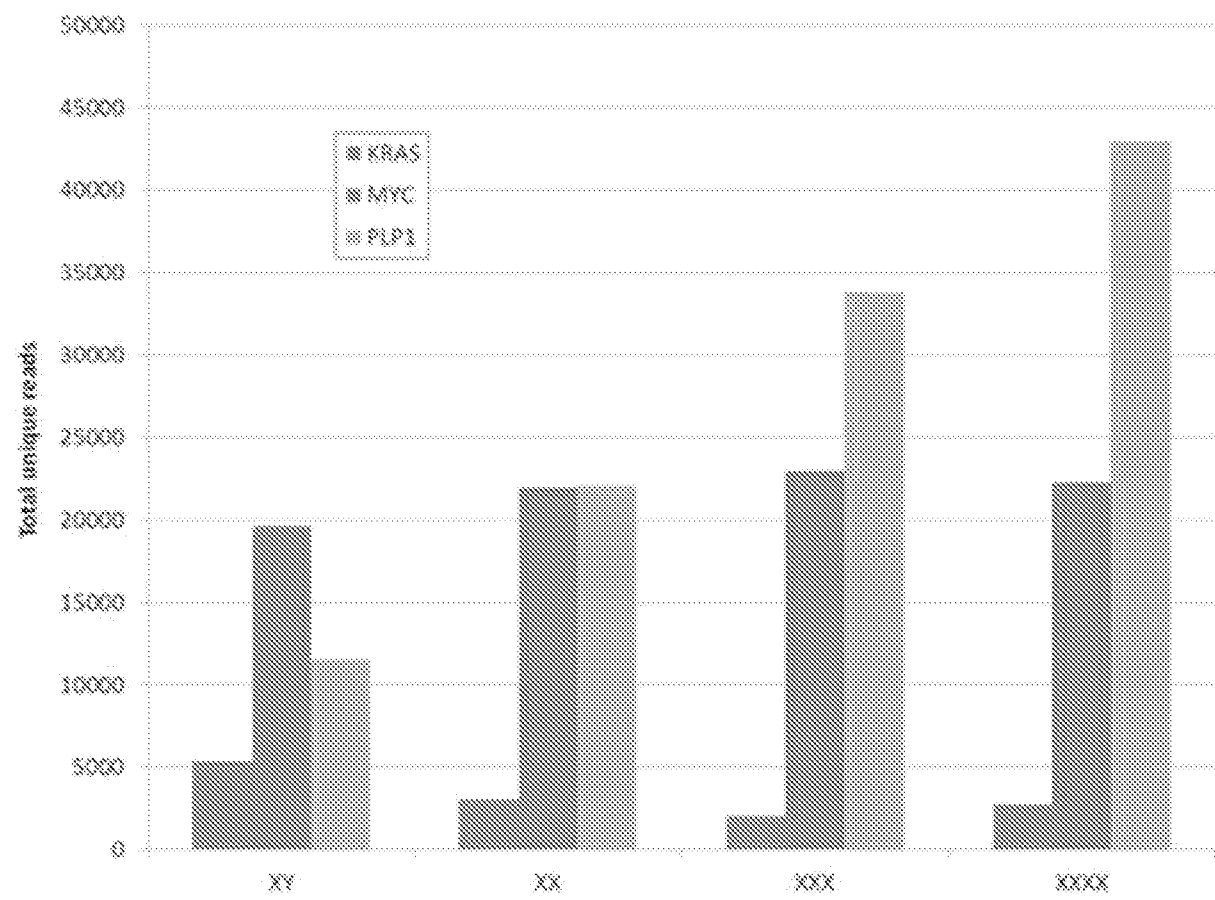


FIG. 44

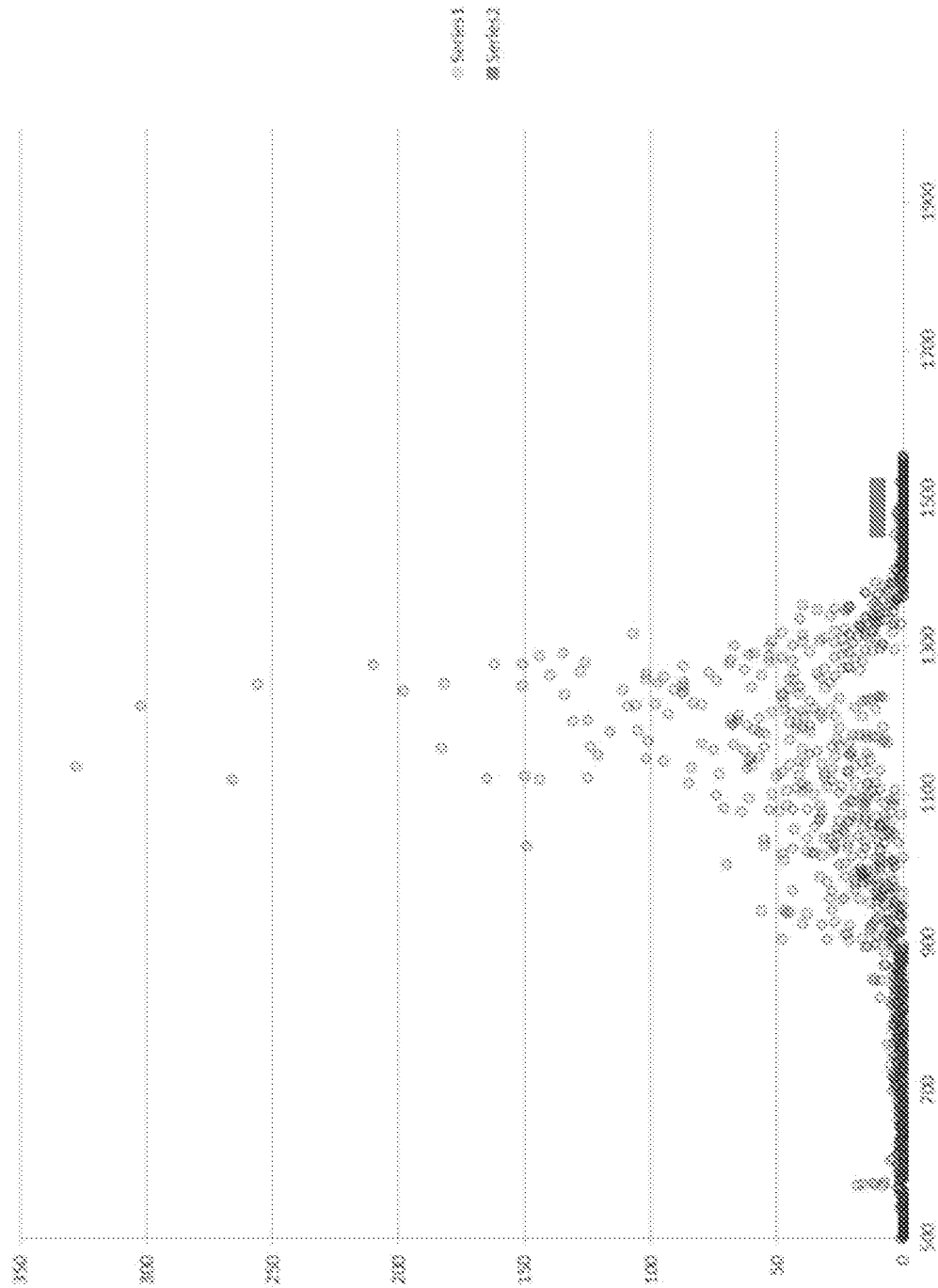


FIG. 45

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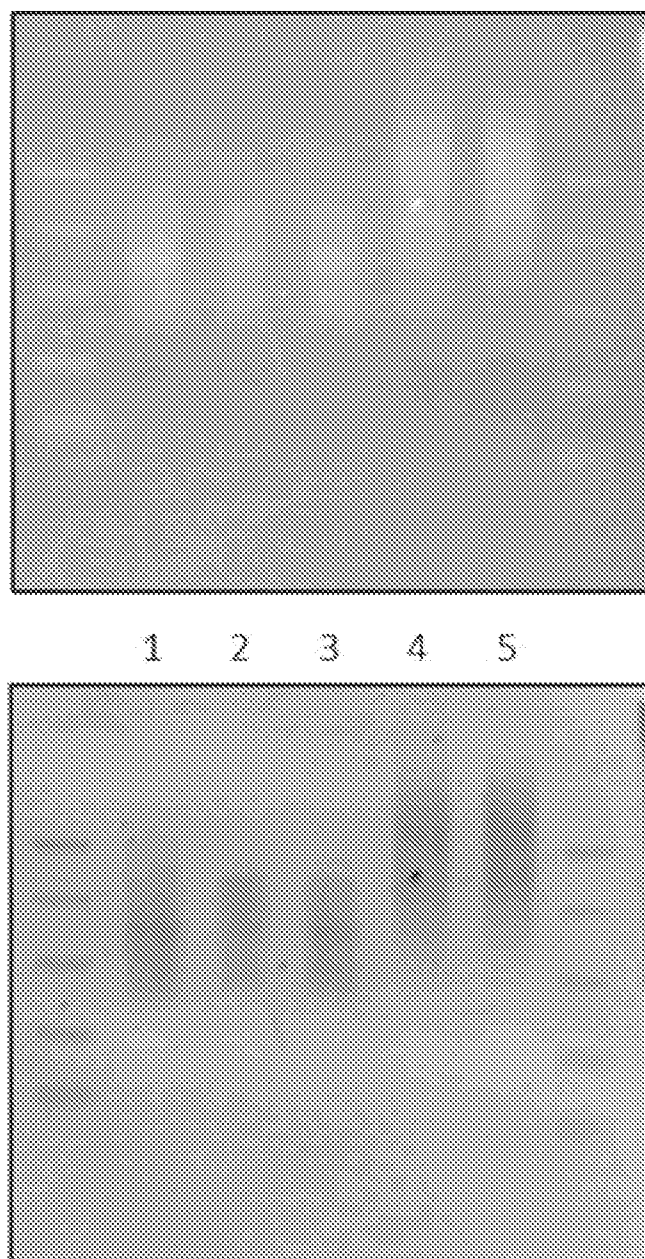


Figure 1. DNA gel of RNA-seq samples.

1. Post capture RNA-seq C+P
2. Pippin prep lane 4
3. Pippin prep lane 5
4. ACA2-20 dT library
5. ACA2_FLFP + exome CAC3_FLRP dT library

FIG. 46

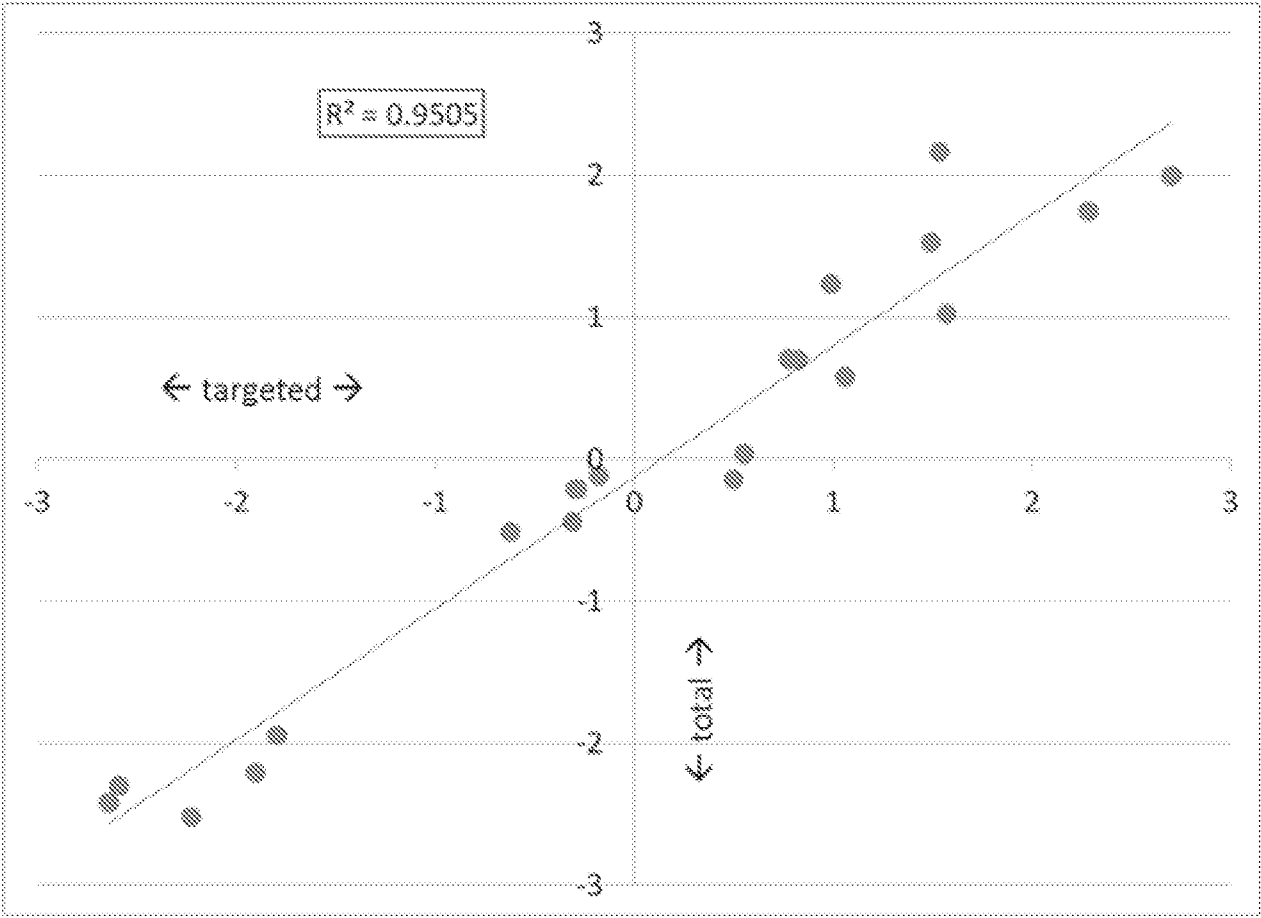


FIG. 47

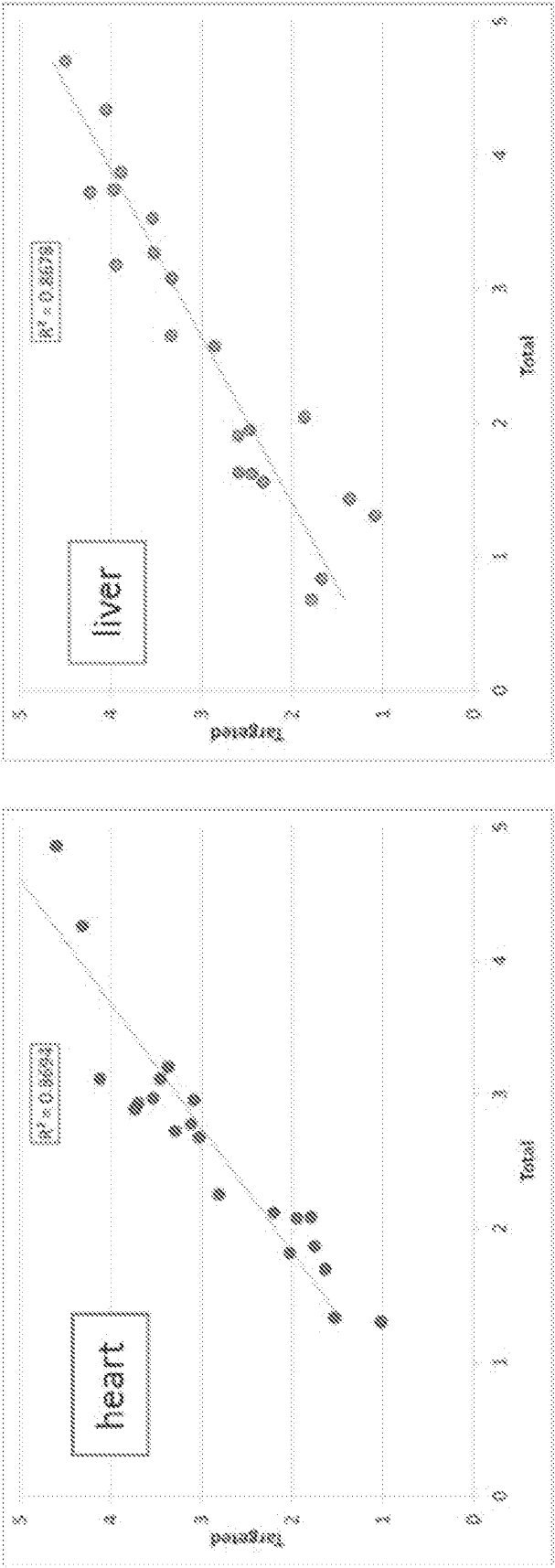


FIG. 48