

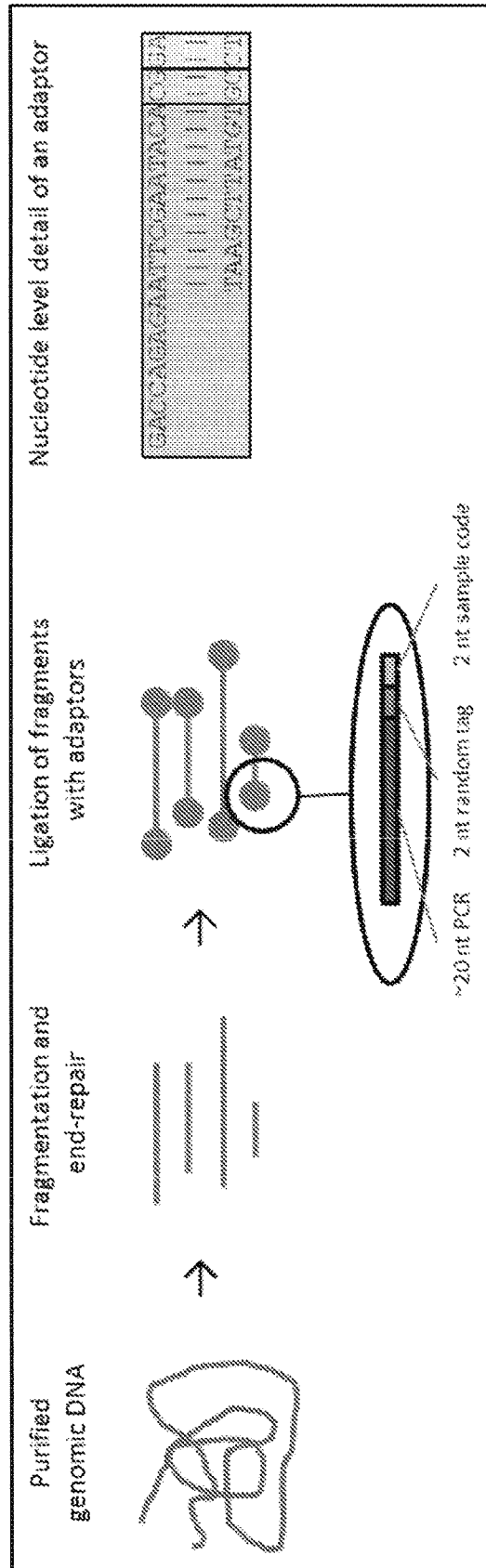


FIG. 1

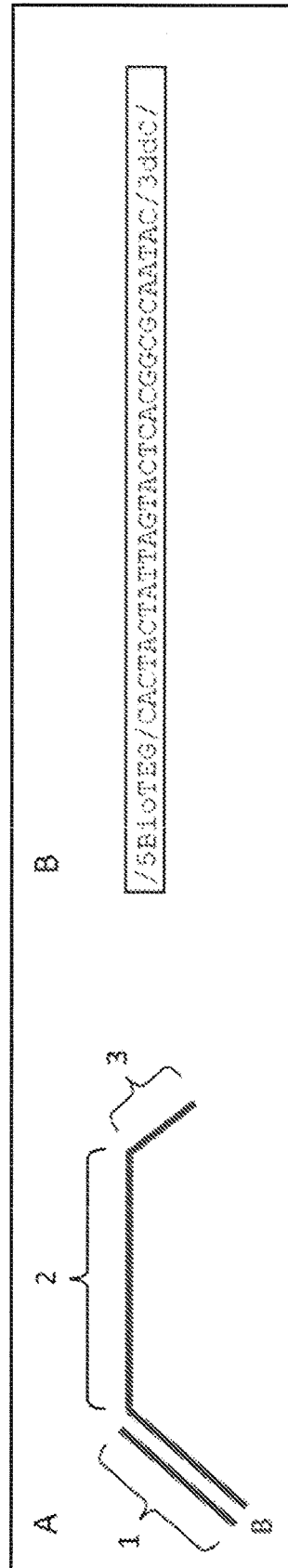
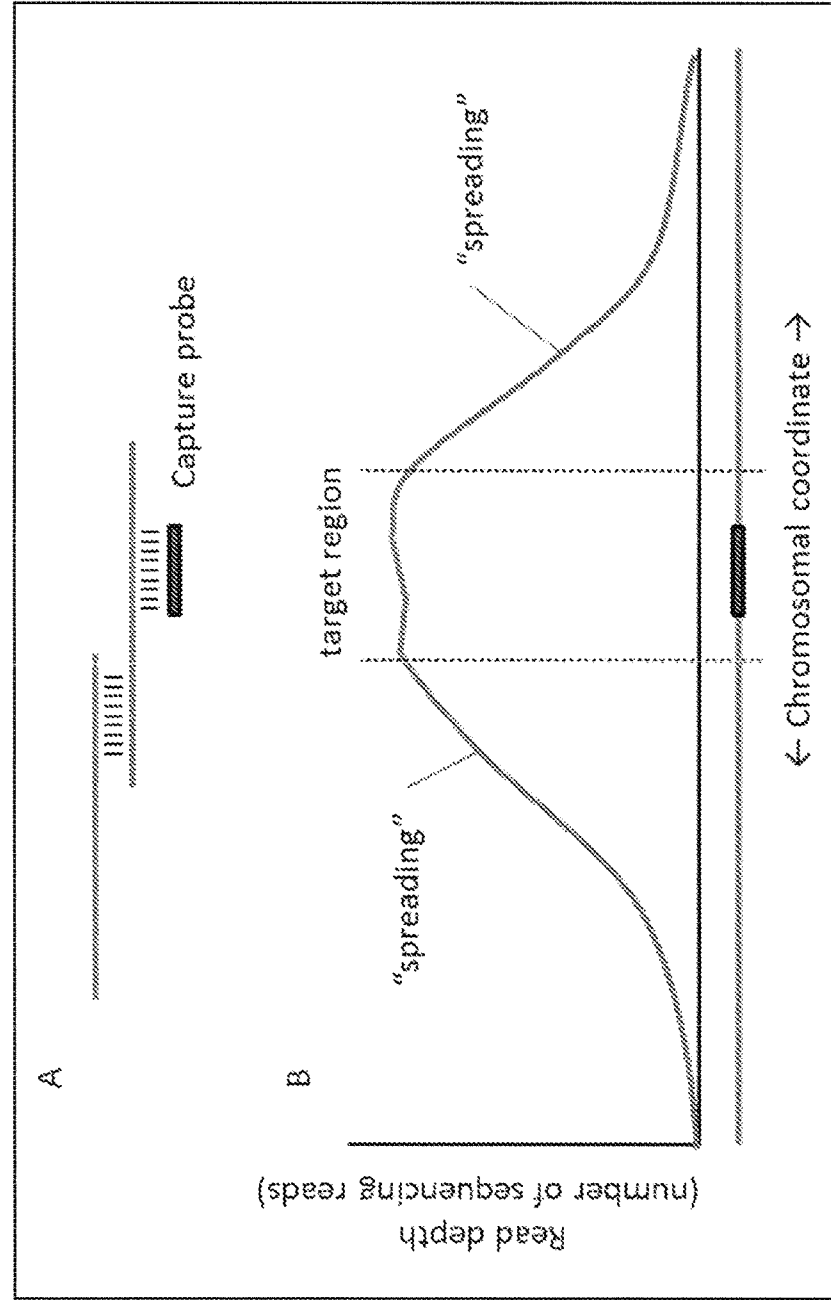


FIG. 2

*FIG. 3*

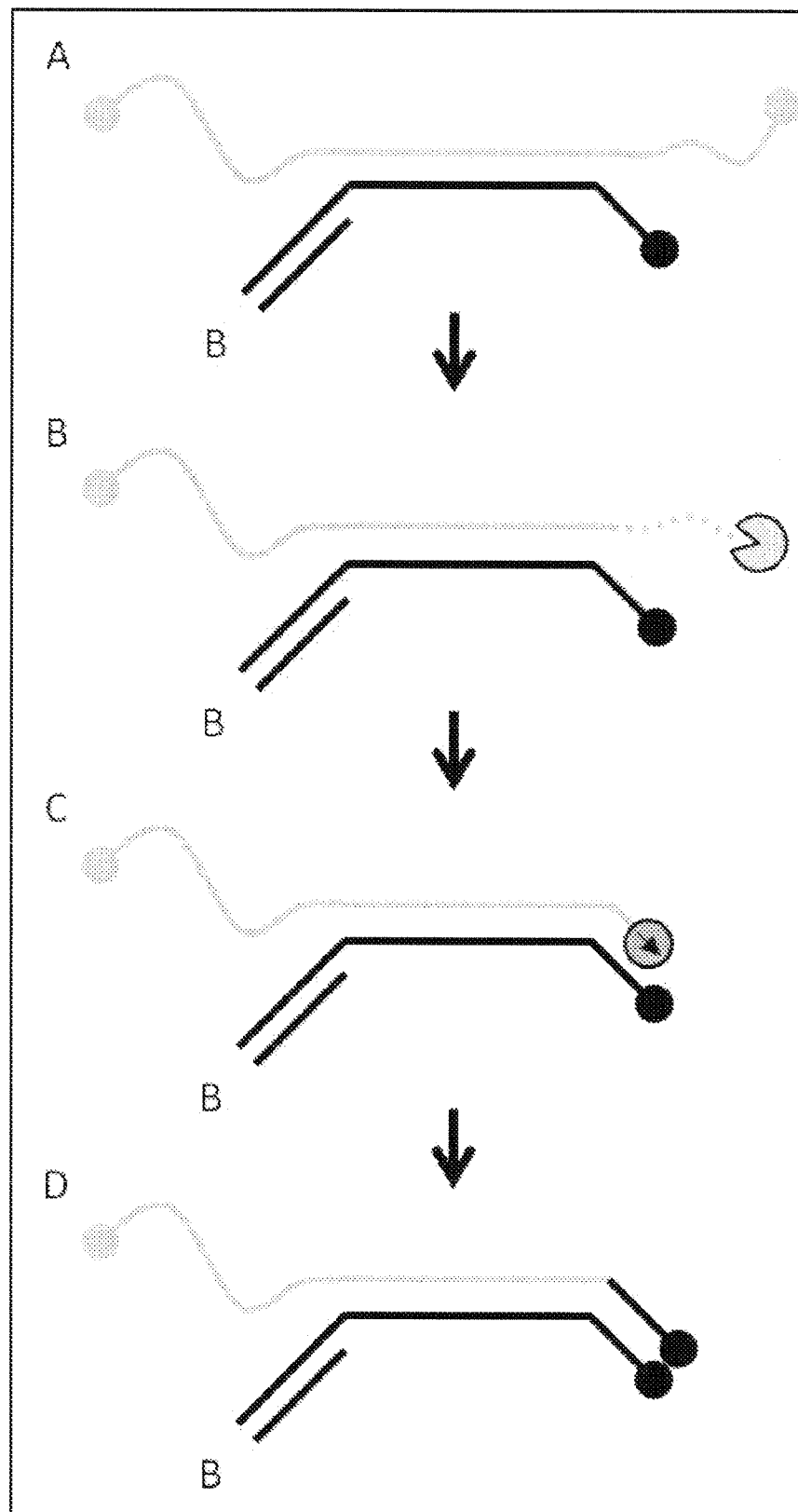
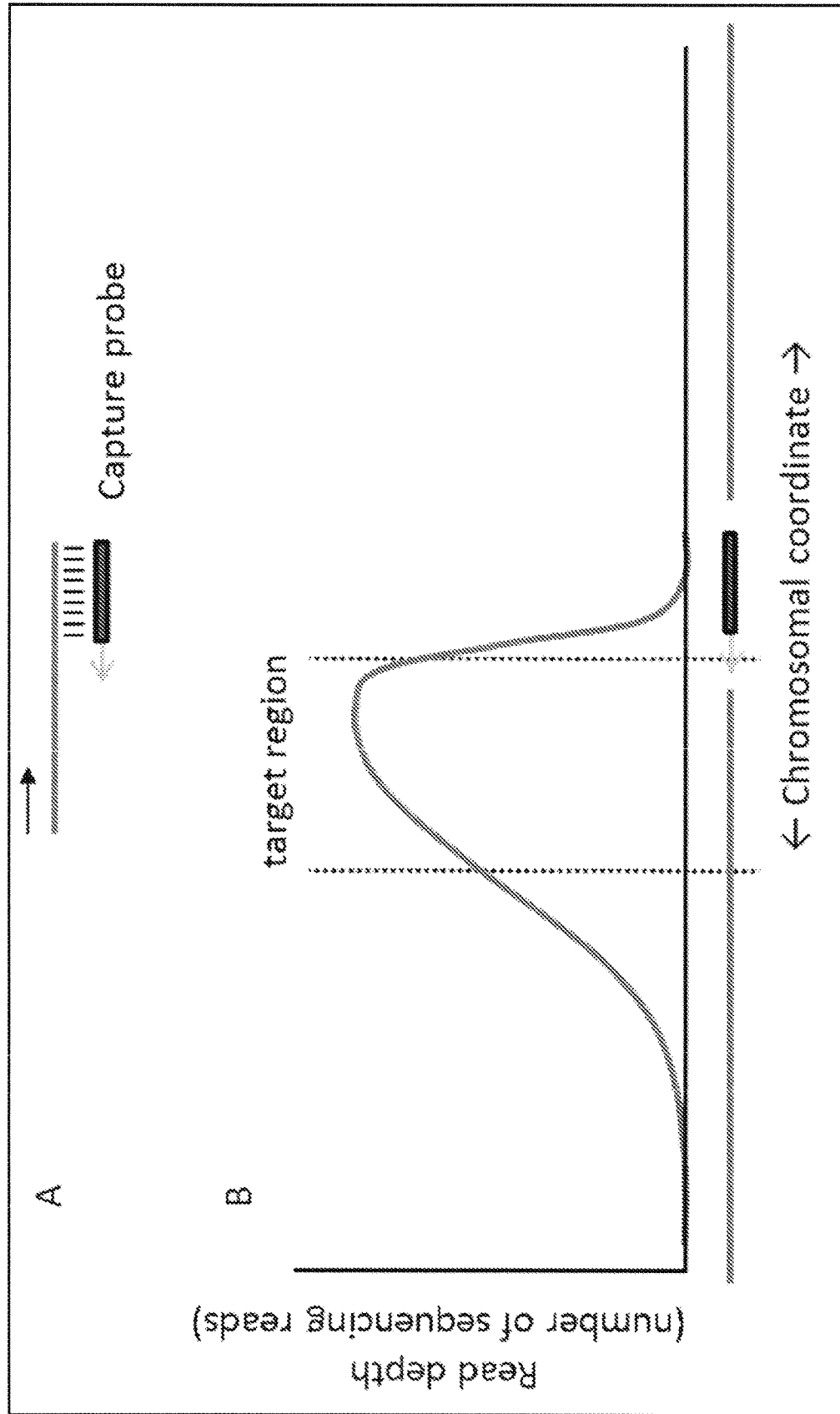


FIG. 4

*FIG. 5*

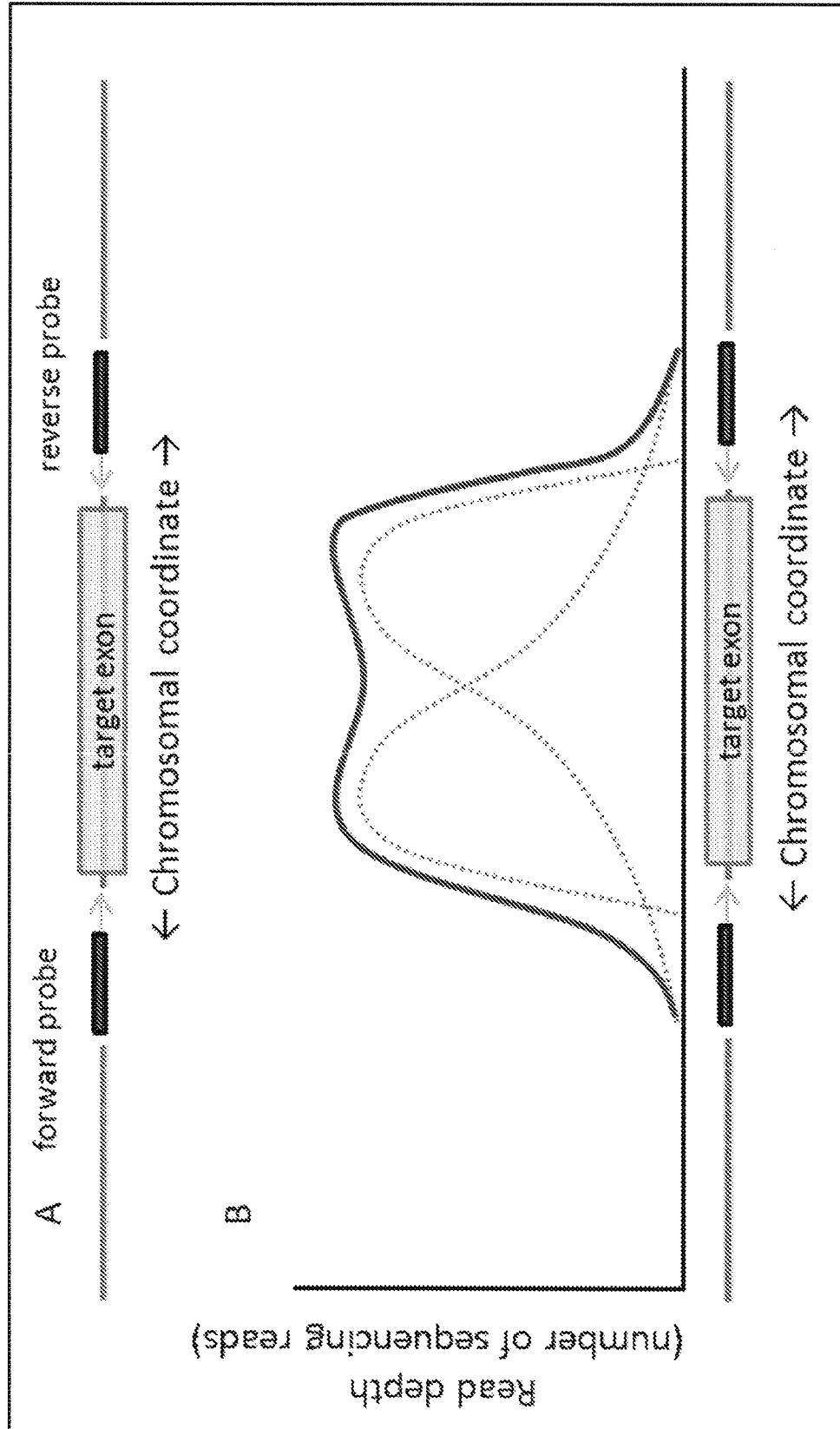


FIG. 6

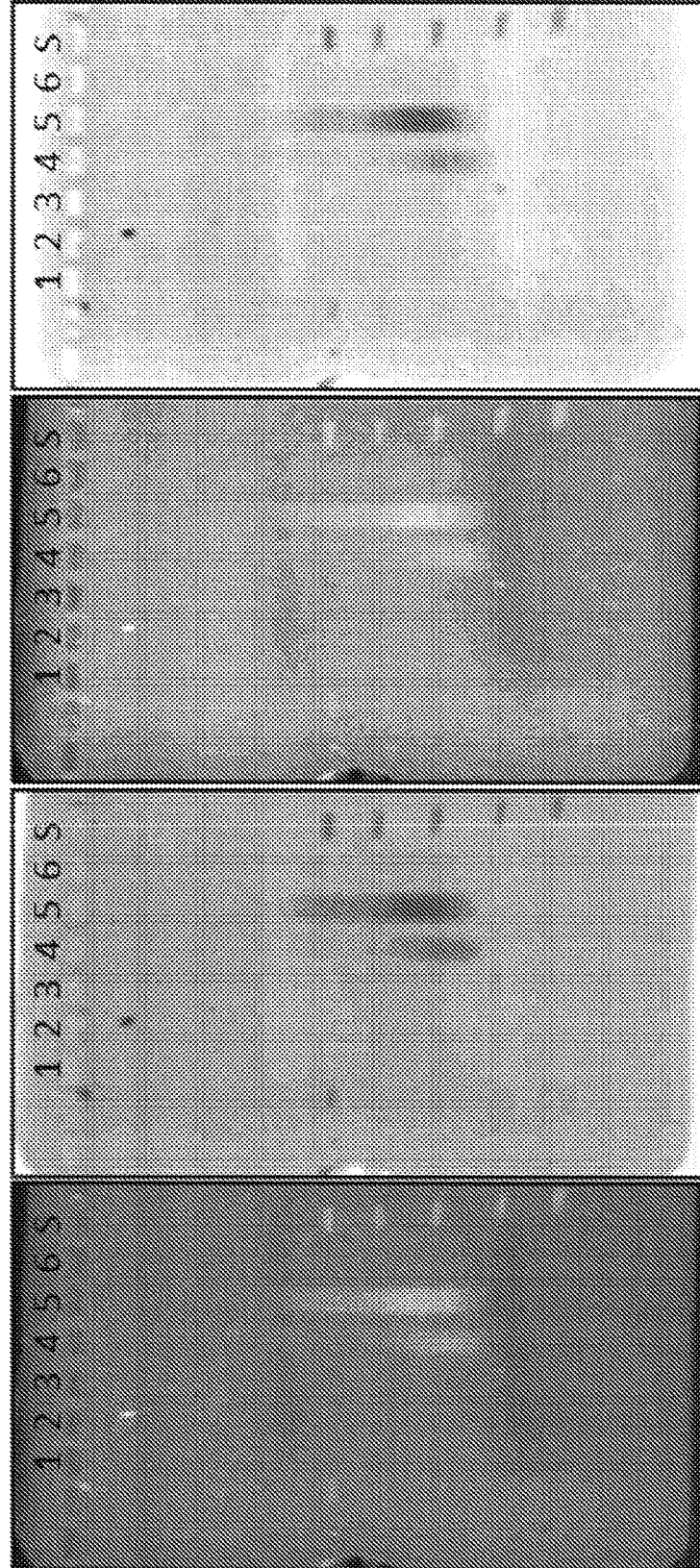
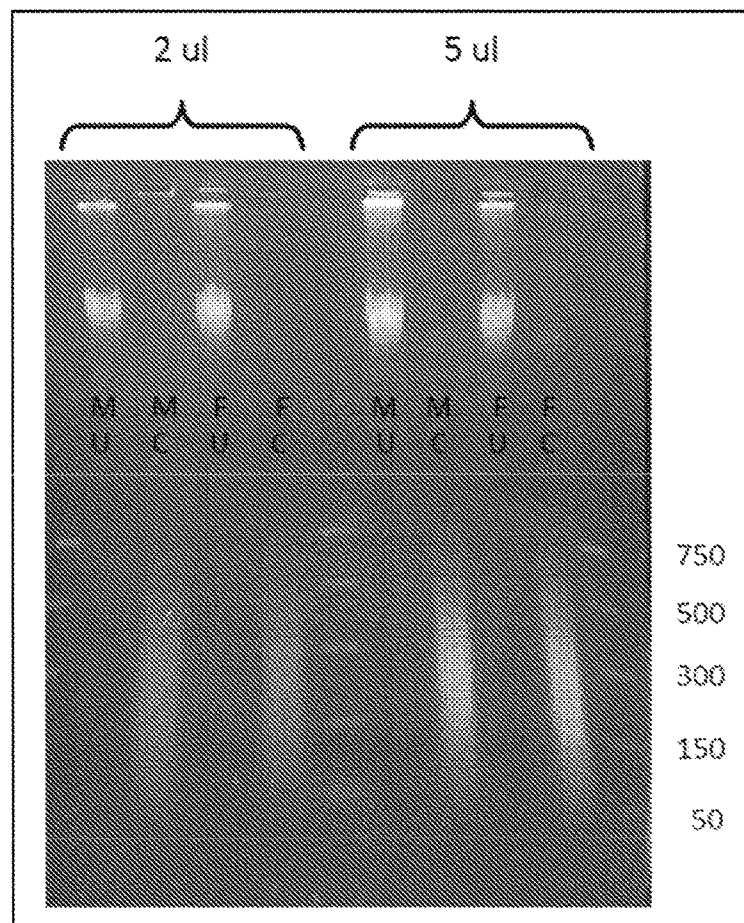


FIG. 7

*FIG. 8*

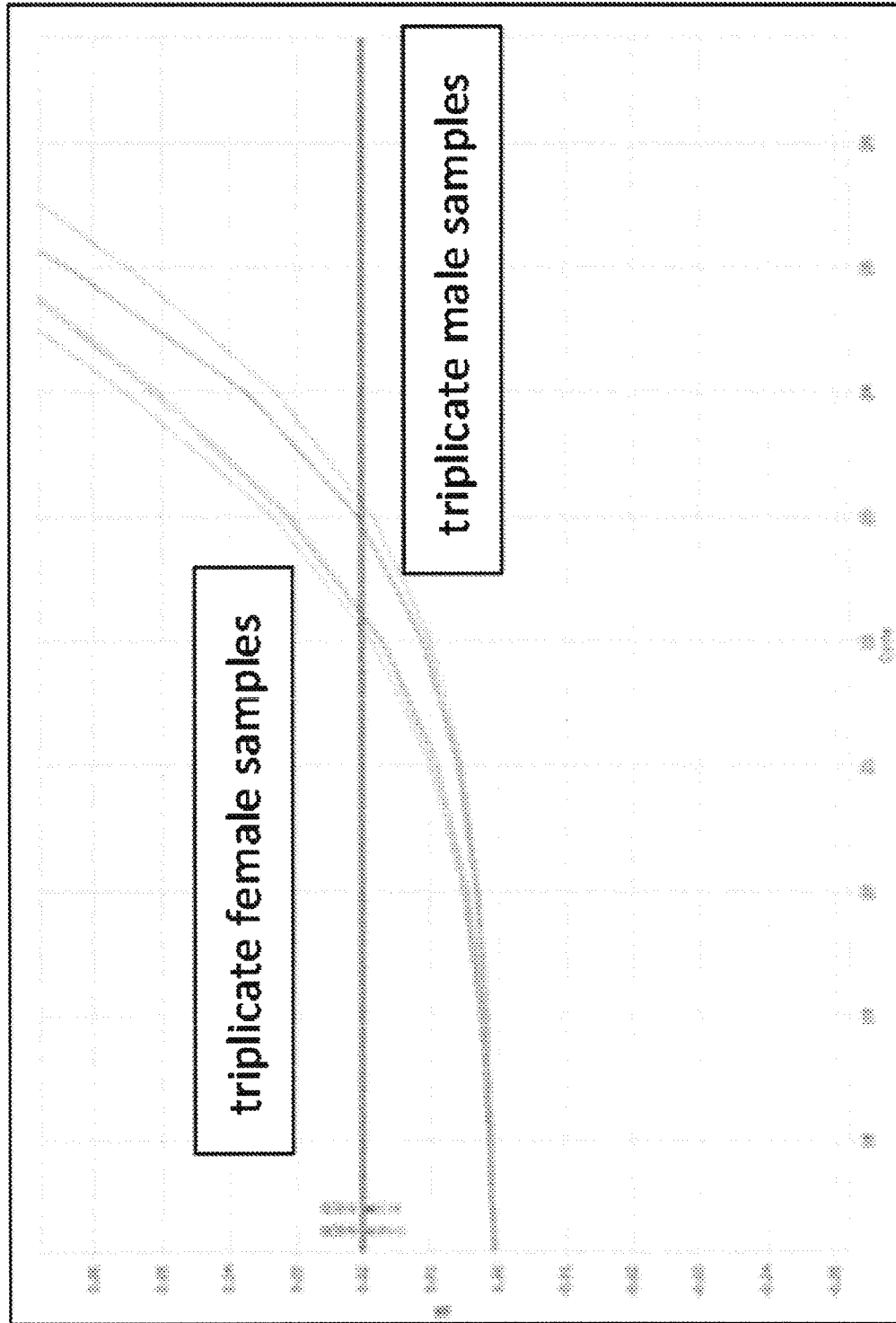
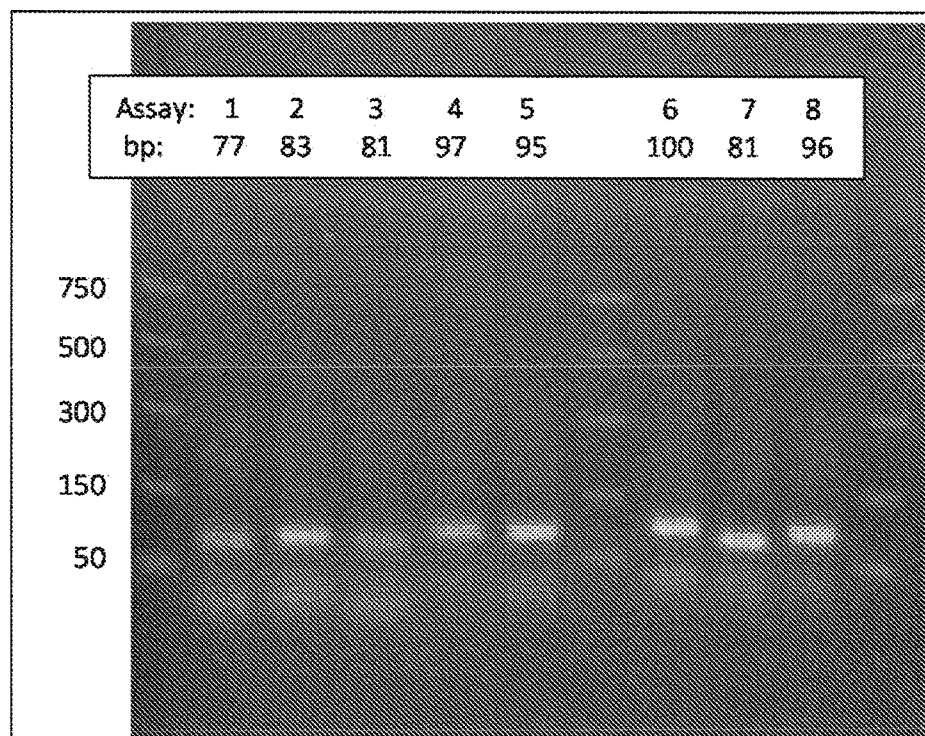
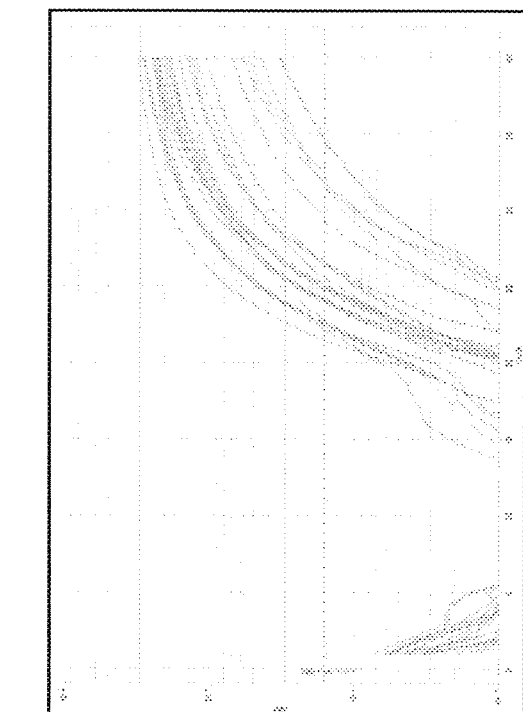
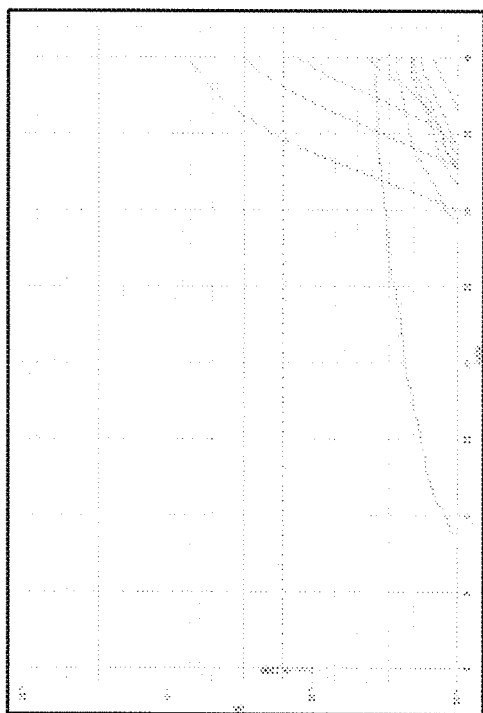


FIG. 9

*FIG. 10*

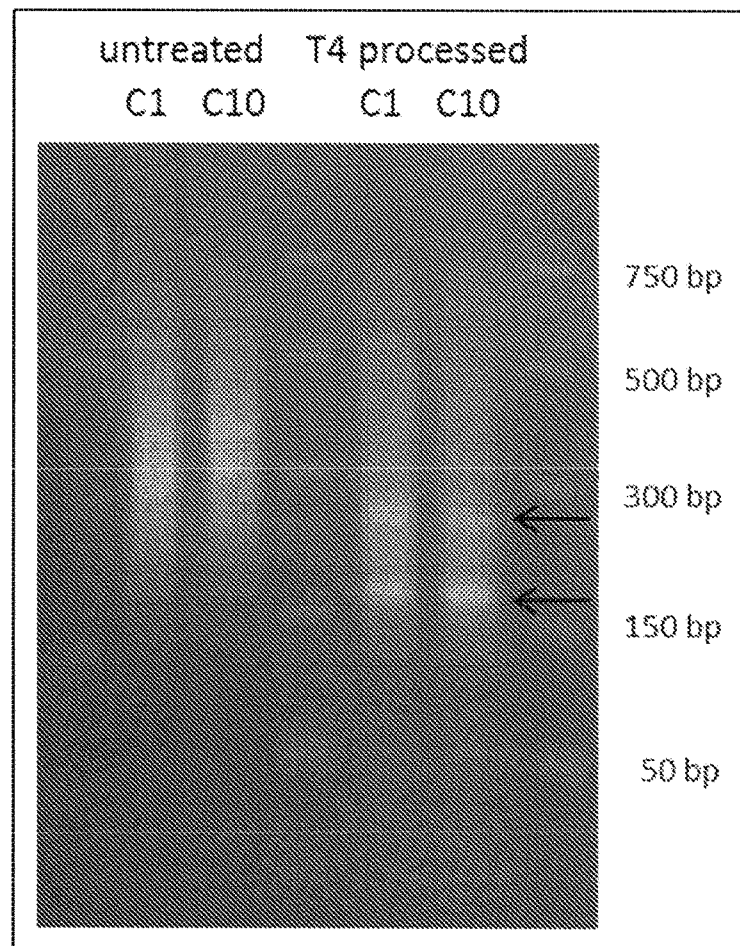


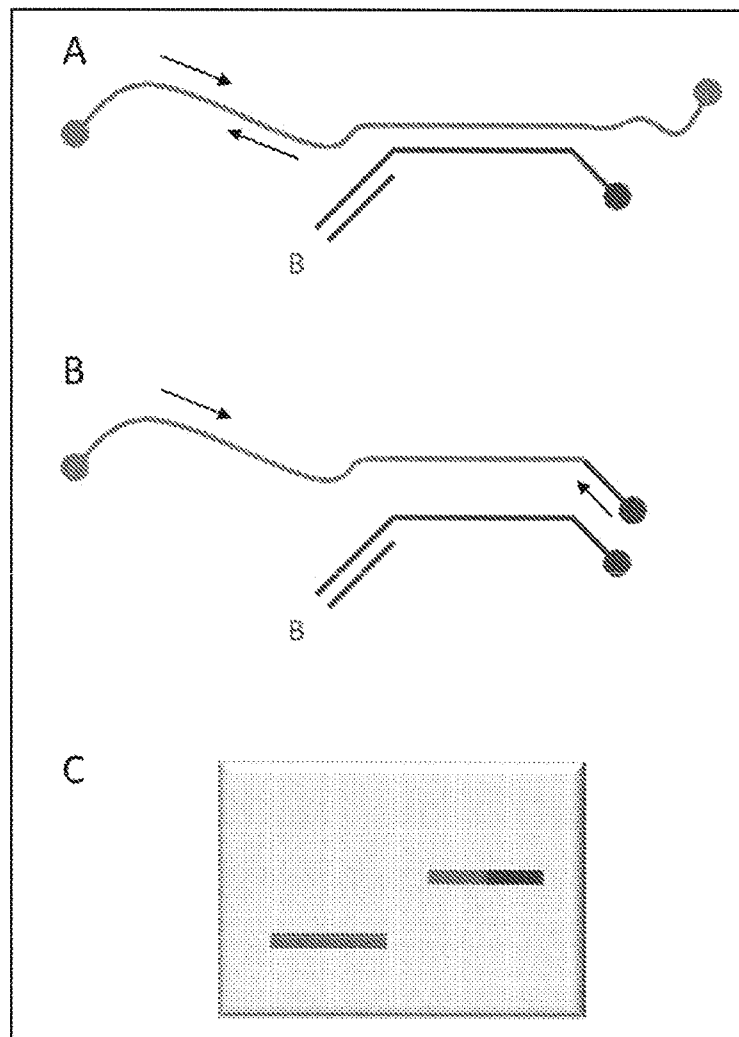
B



A

FIG. 11

*FIG. 12*

*FIG. 13*

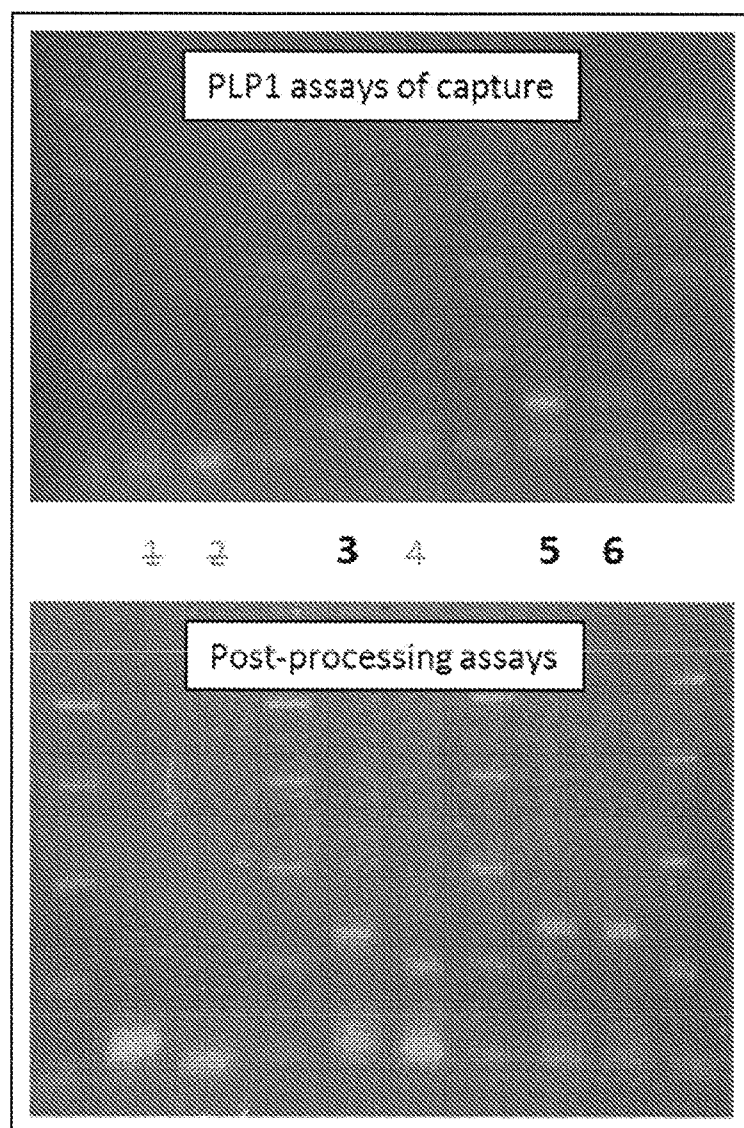
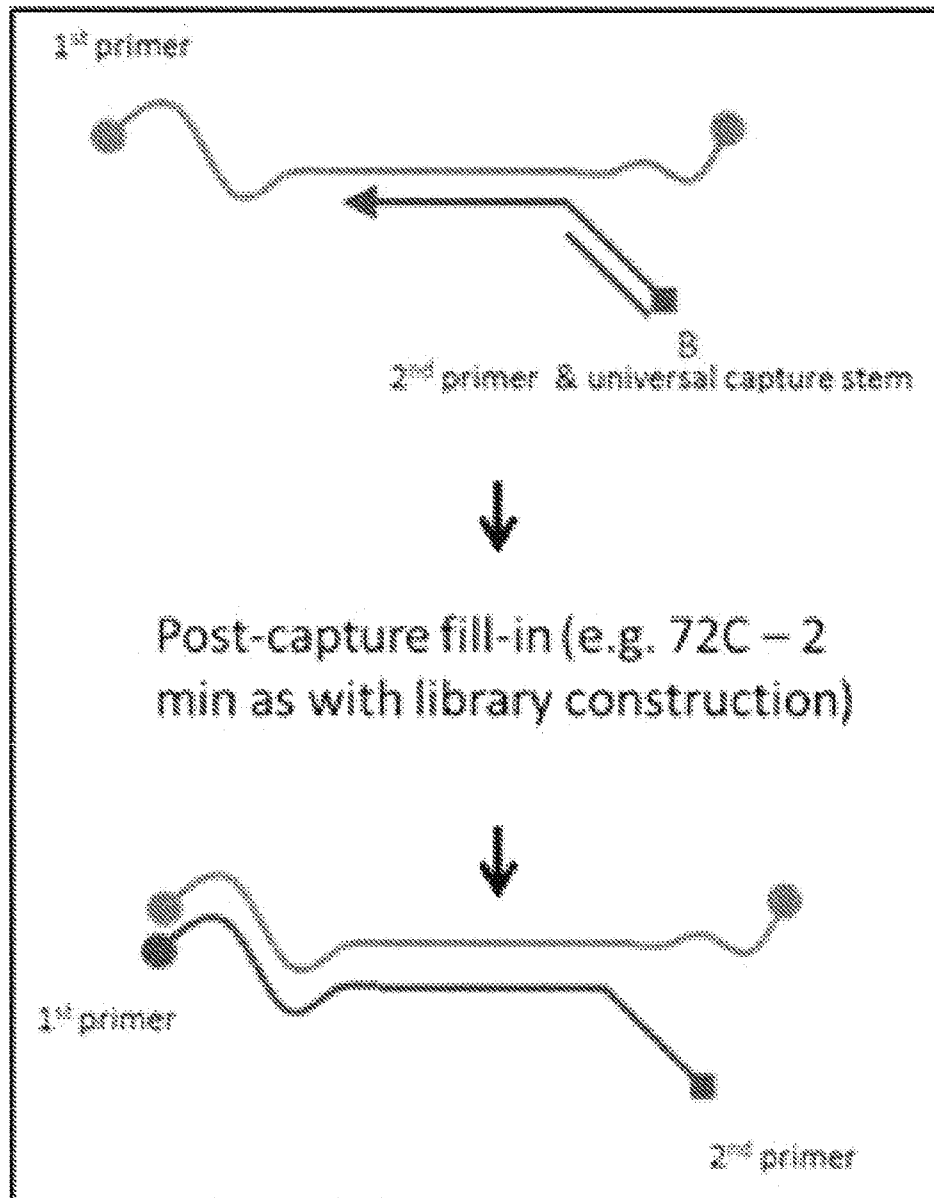
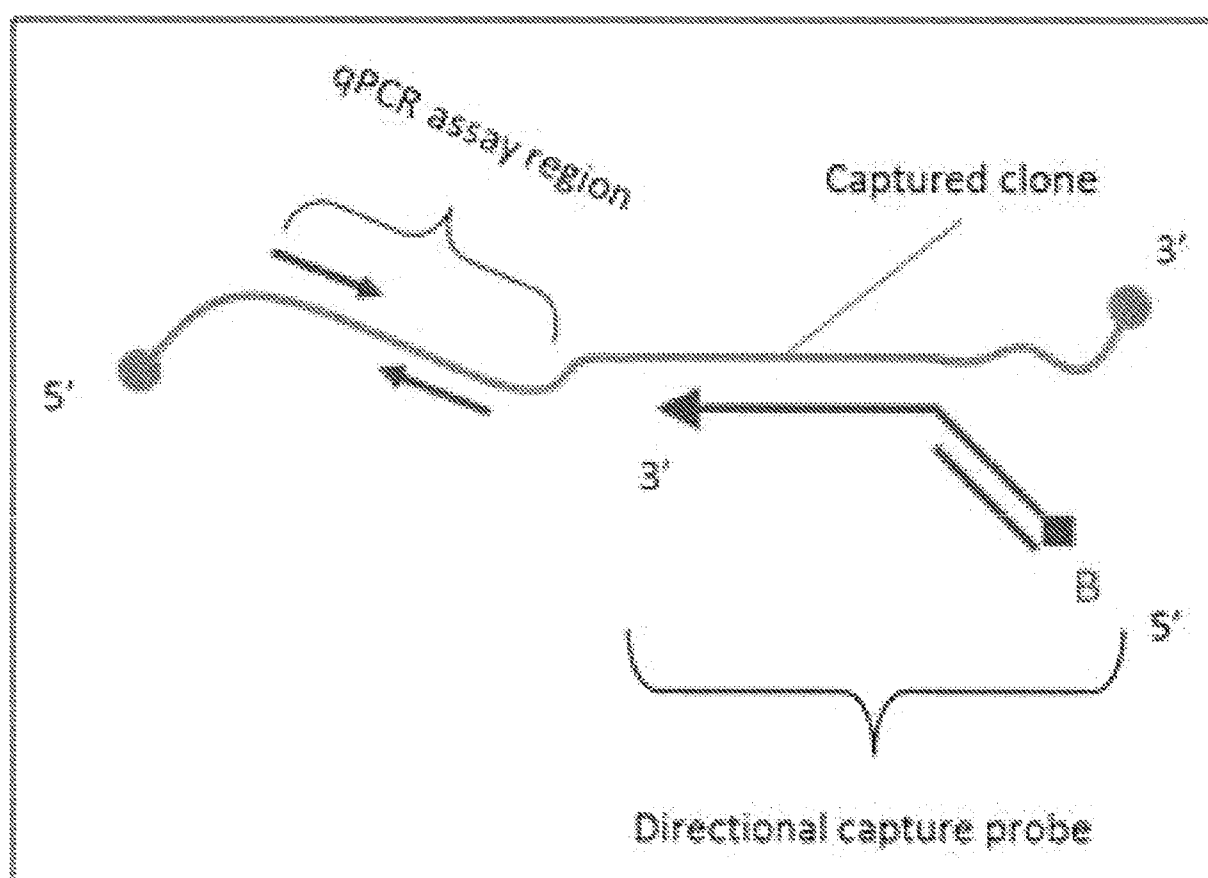
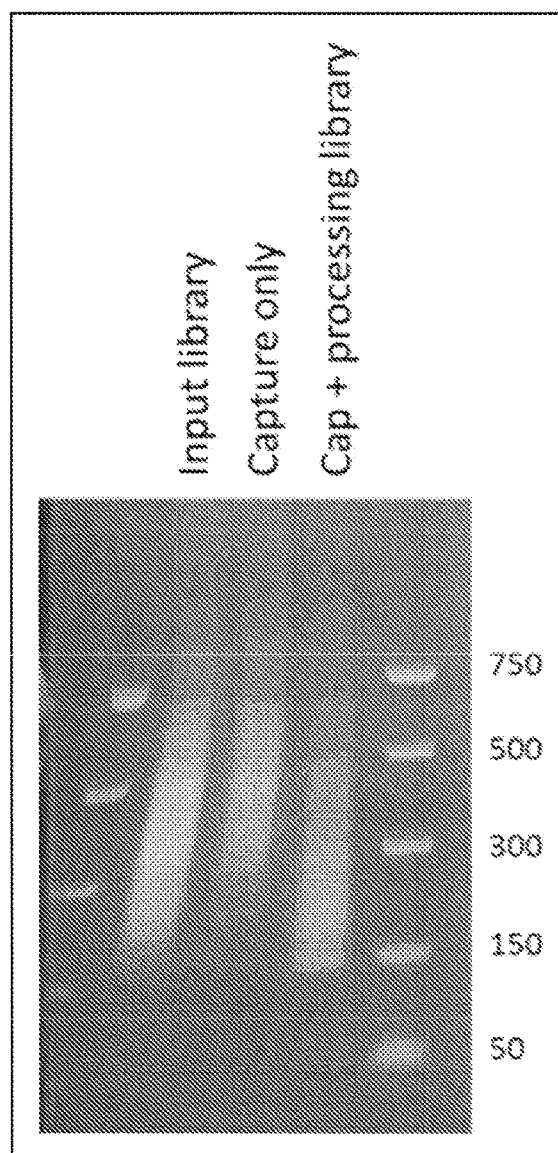


*FIG. 14*

*FIG. 15*

*FIG. 16*

*FIG. 17*

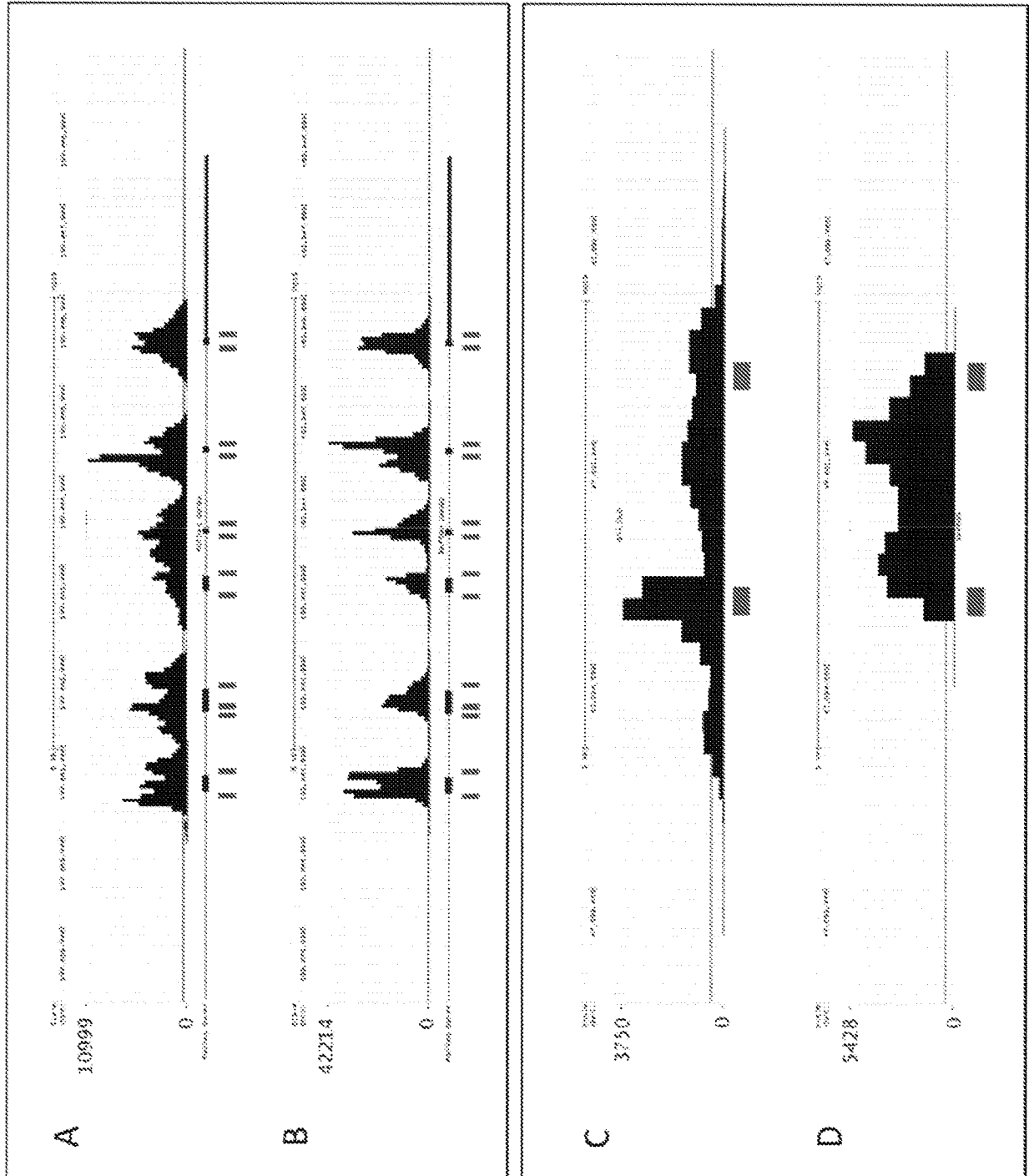


FIG. 18

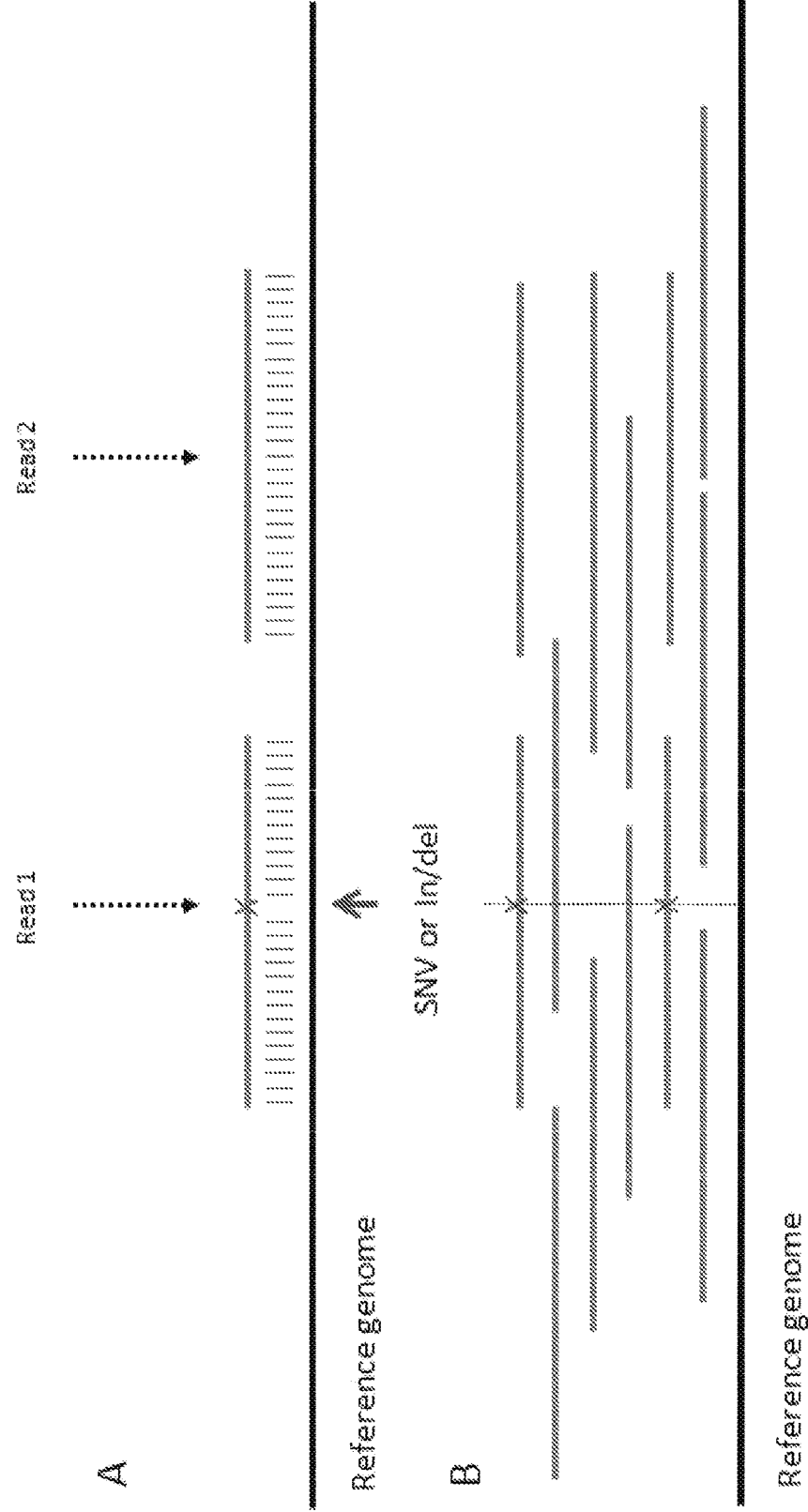
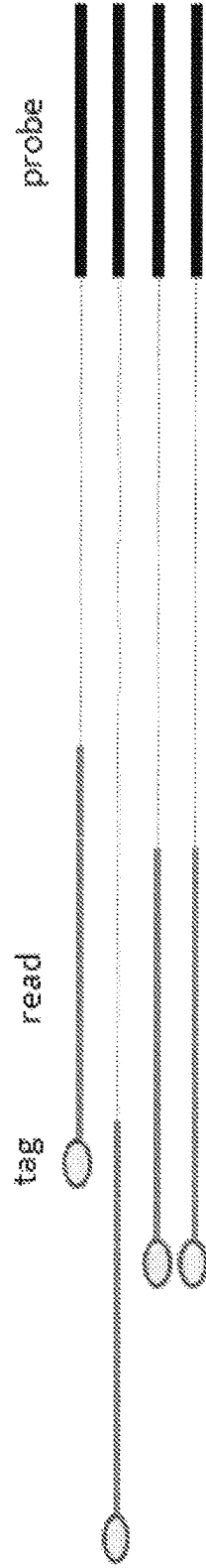
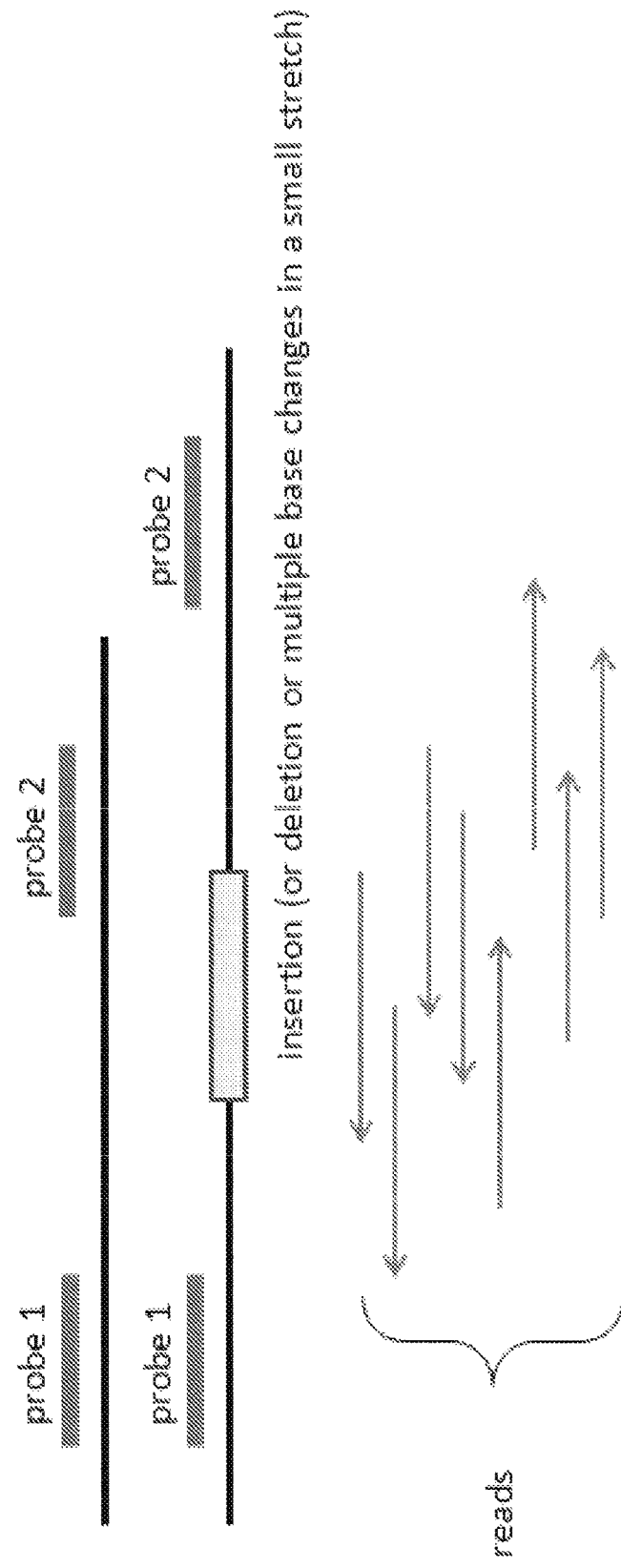
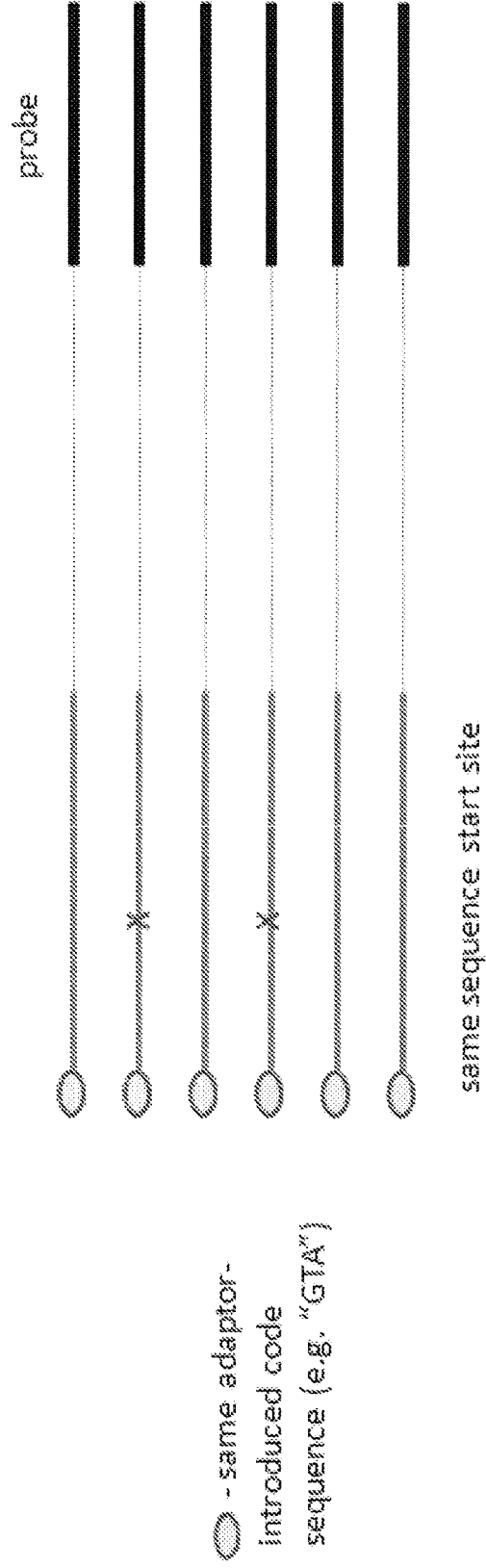


FIG. 19

*FIG. 20*

*FIG. 21*

*FIG. 22*

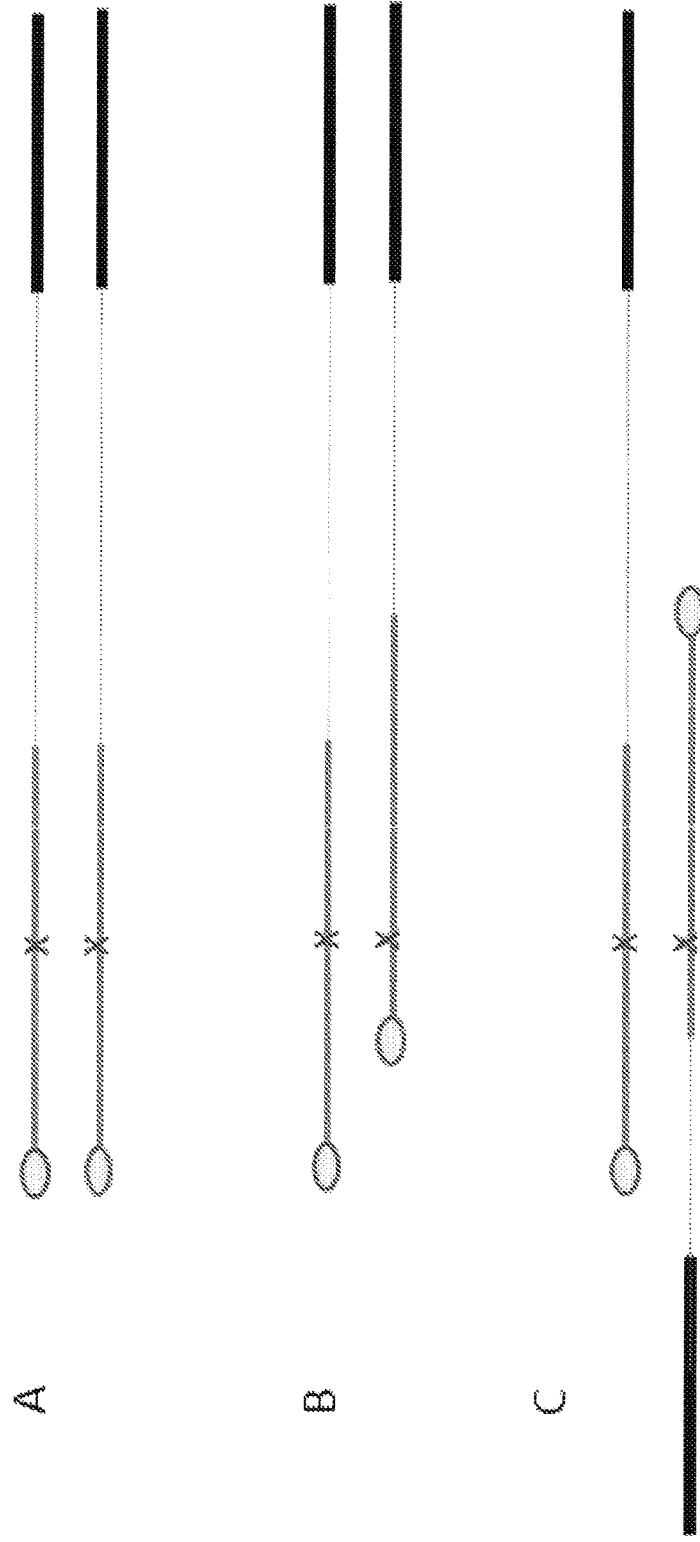


FIG. 23

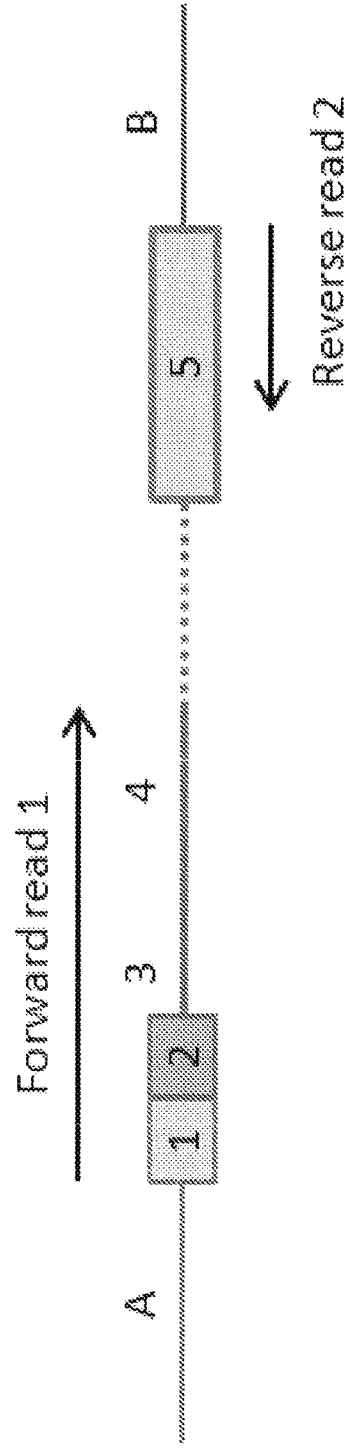


FIG. 24

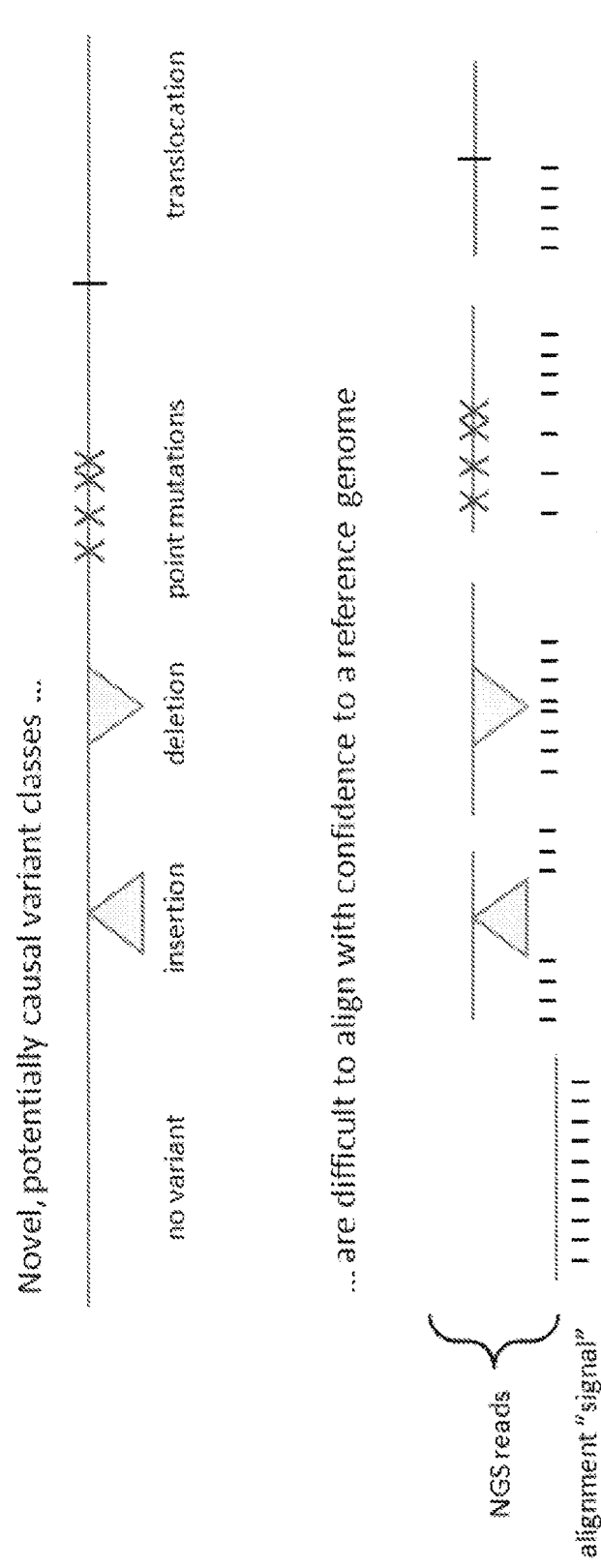


FIG. 25

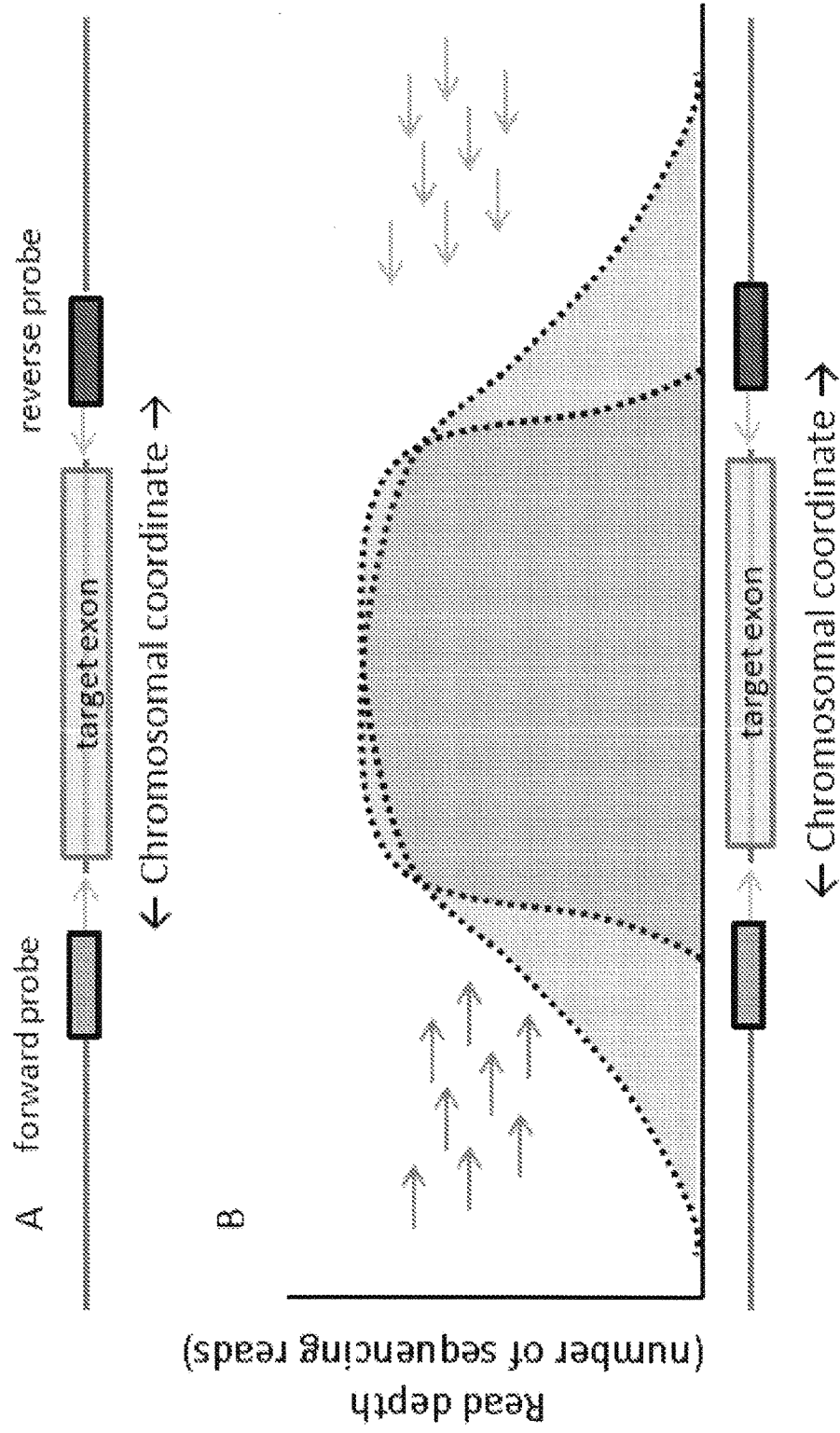
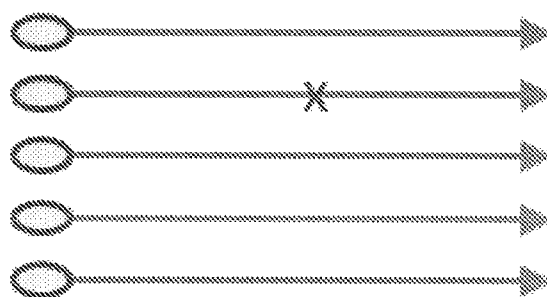


FIG. 26

A



B

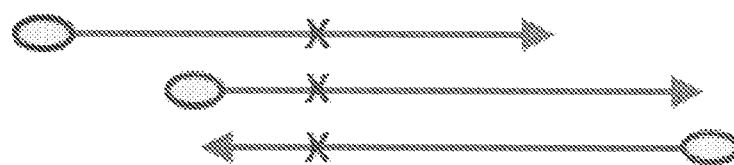
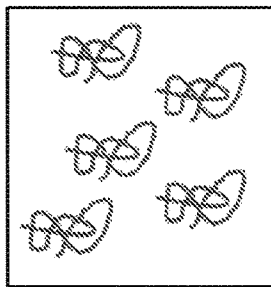
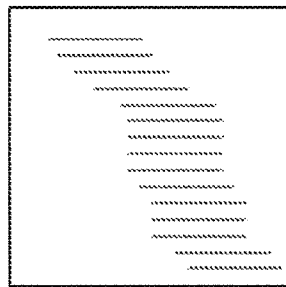


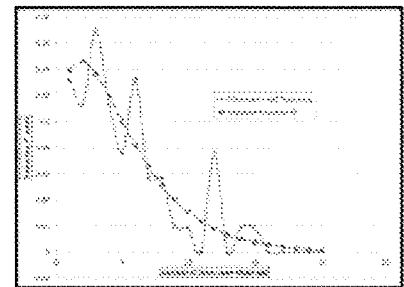
FIG. 27



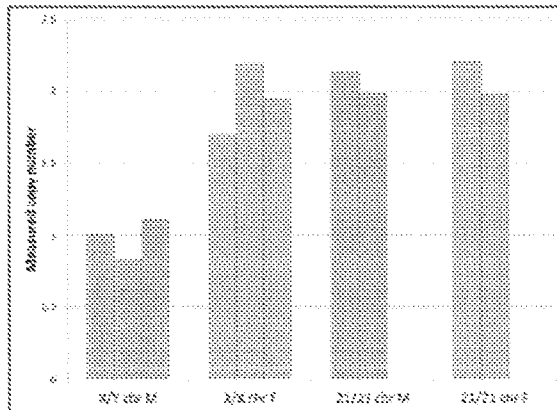
Low input genome libraries (e. g. ~50-100 genome equivalents) created with molecular annotation and subsequently captured with targeting probes.



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.

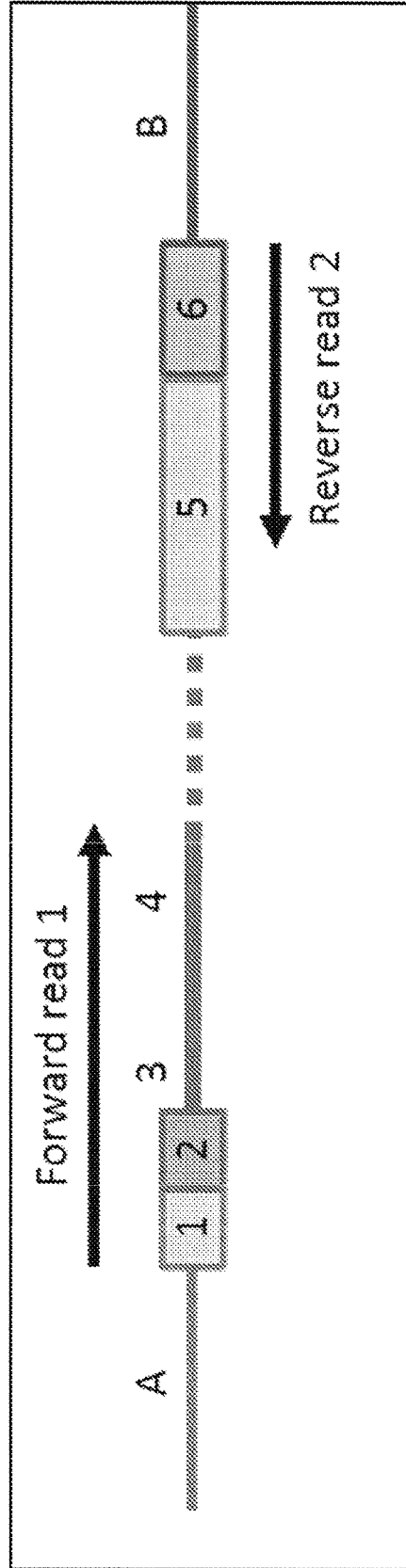


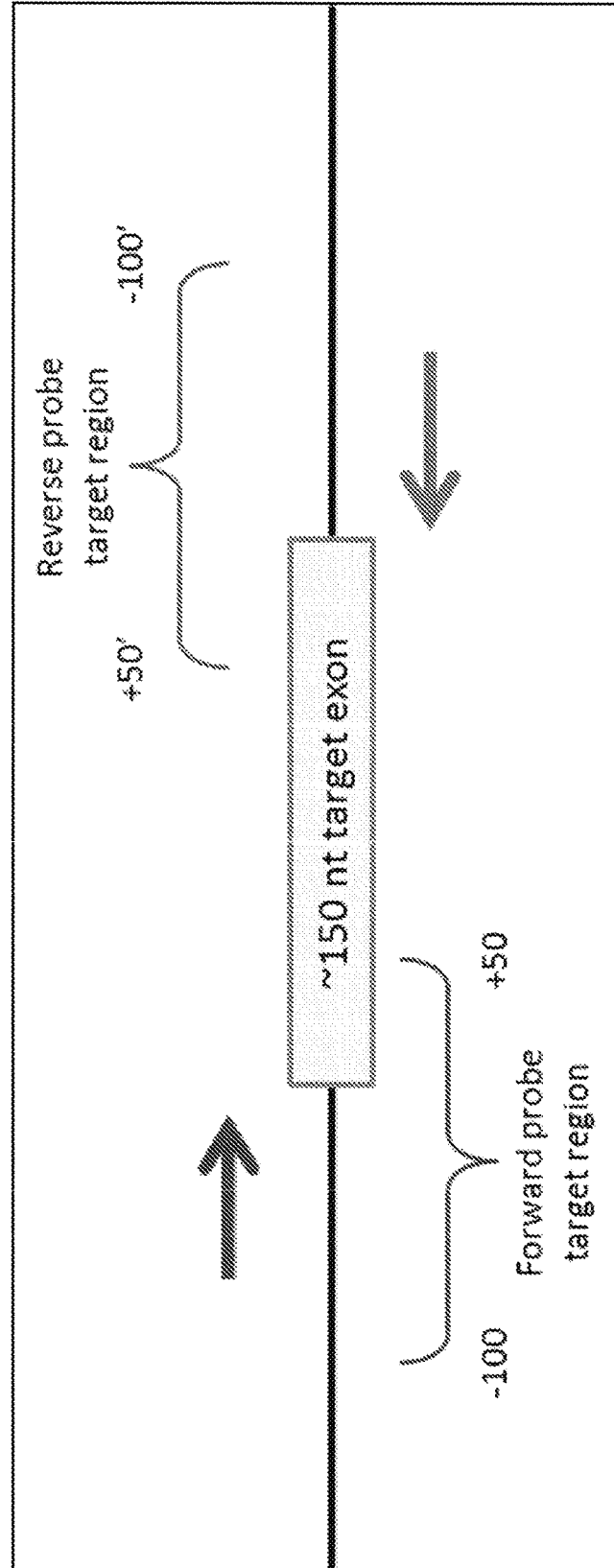
Observed read frequencies are fitted to a negative binomial distribution, which allows the "inferred class" of unobserved reads to be estimated. The number of observed reads + inferred reads = copy number.

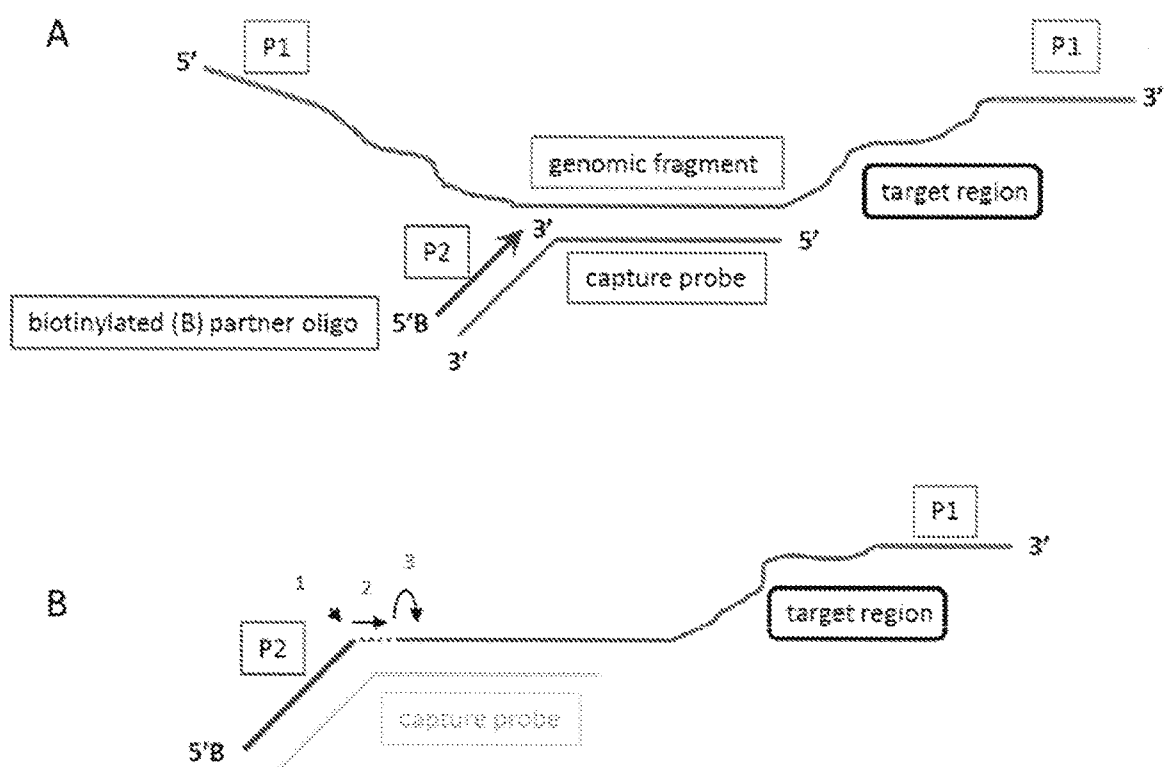


Representative copy determination at three probes for the X-chromosomal gene PLP1 in a male and a female sample. For comparison, two probes for the autosomal chromosome 21 gene TNFRSF14 gene are also shown.

*FIG. 28*

*FIG. 29*

*FIG. 30*

*FIG. 31*

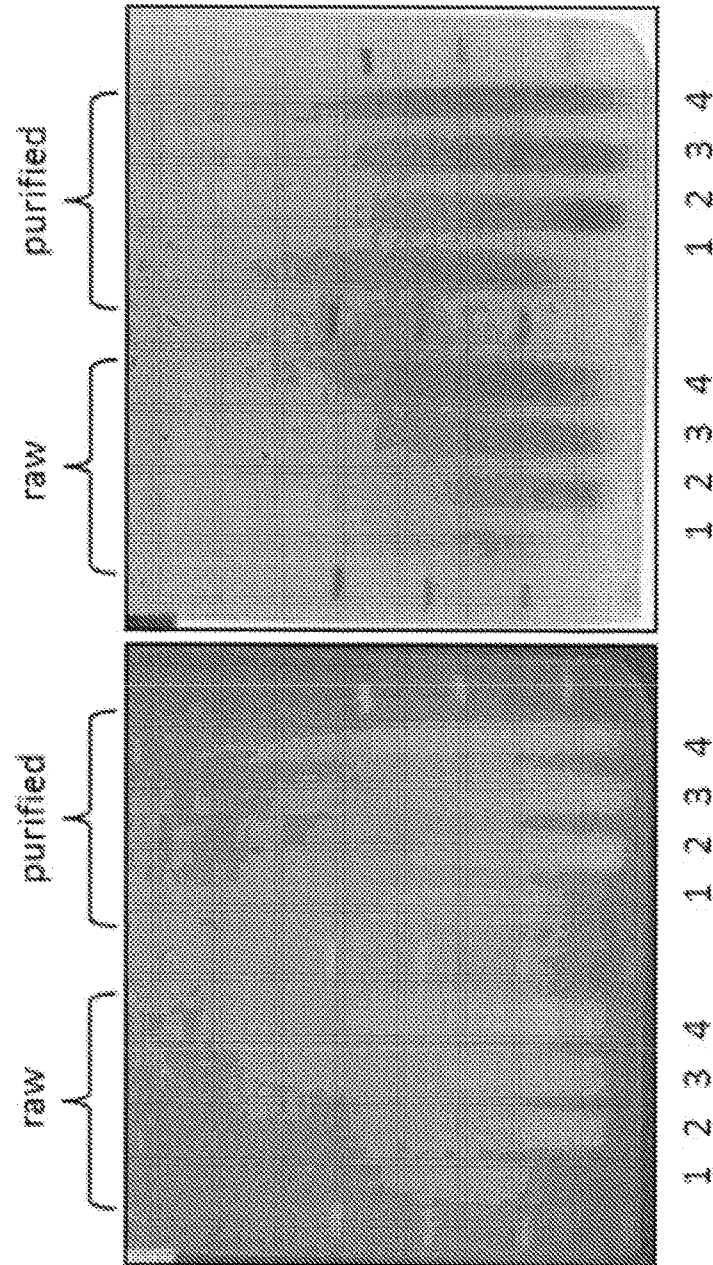


FIG. 32

Step 1 -- fragment gDNA to ~400 bp  
or  
perform 1<sup>st</sup> strand cDNA synthesis with (N)<sub>15</sub> 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

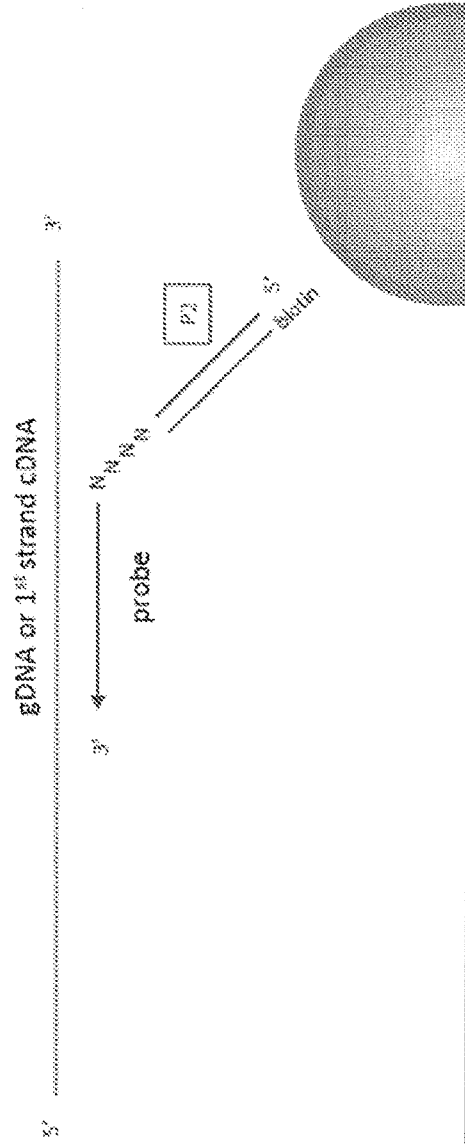


FIG. 33

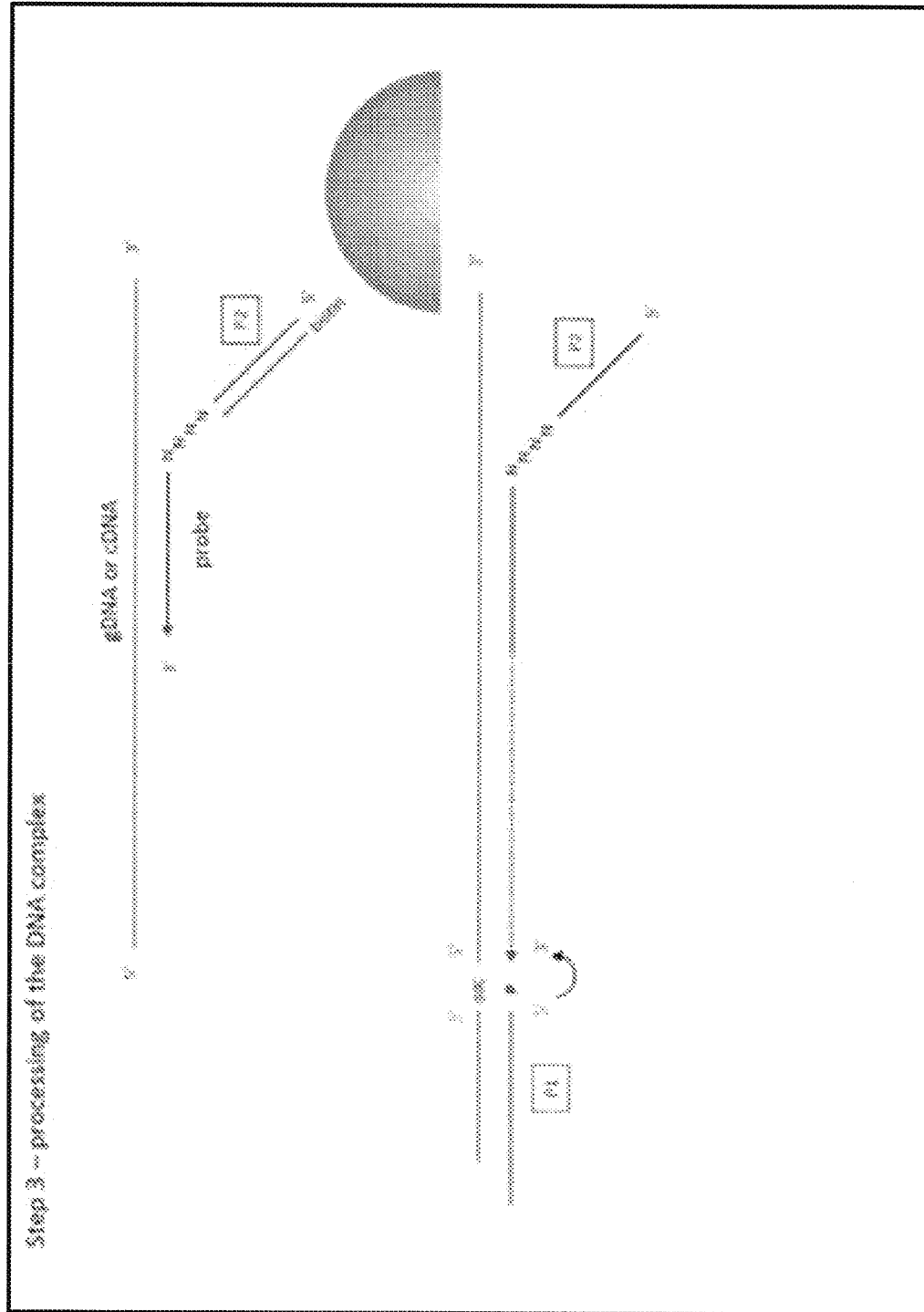


FIG. 34

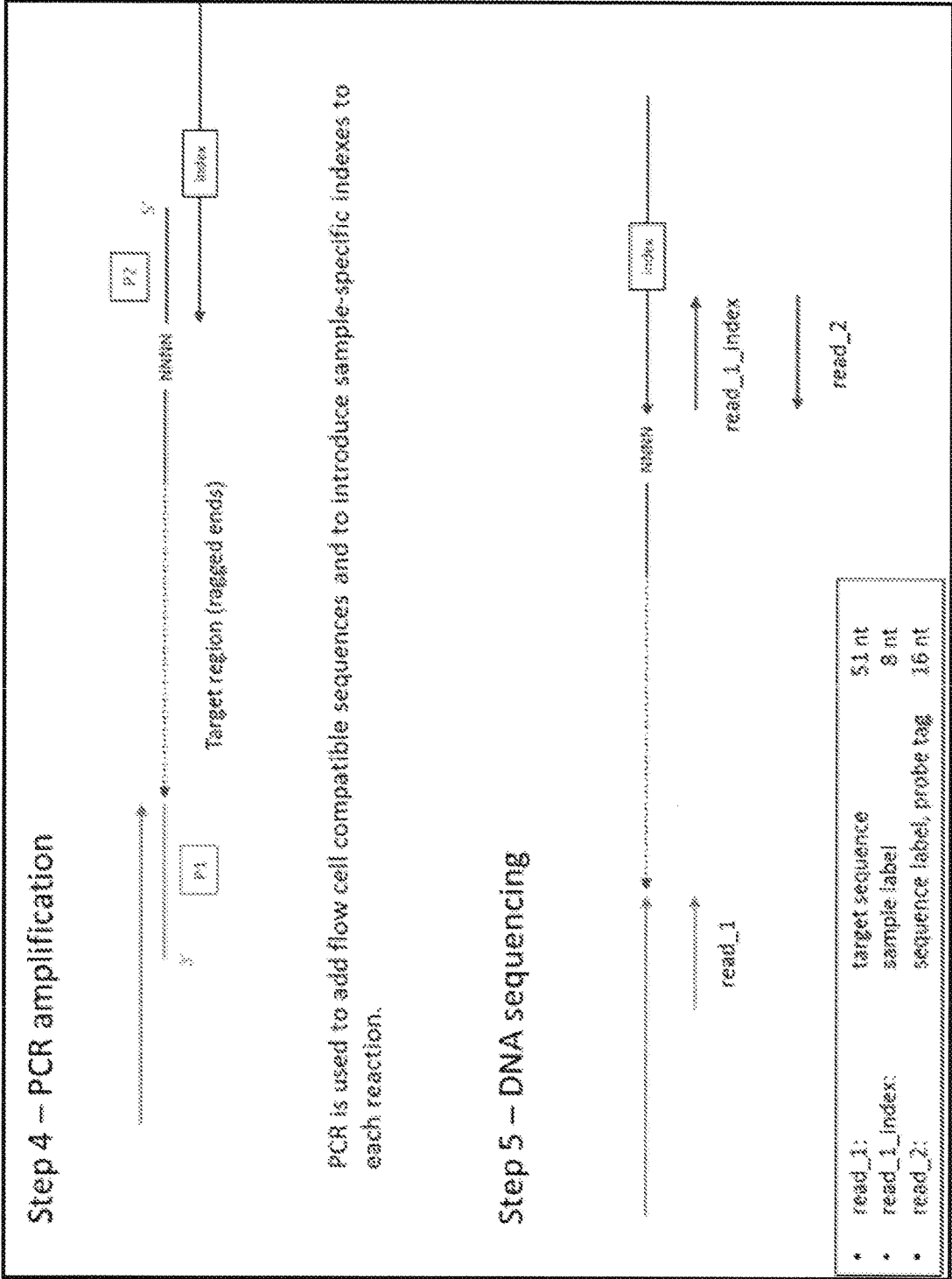


FIG. 35

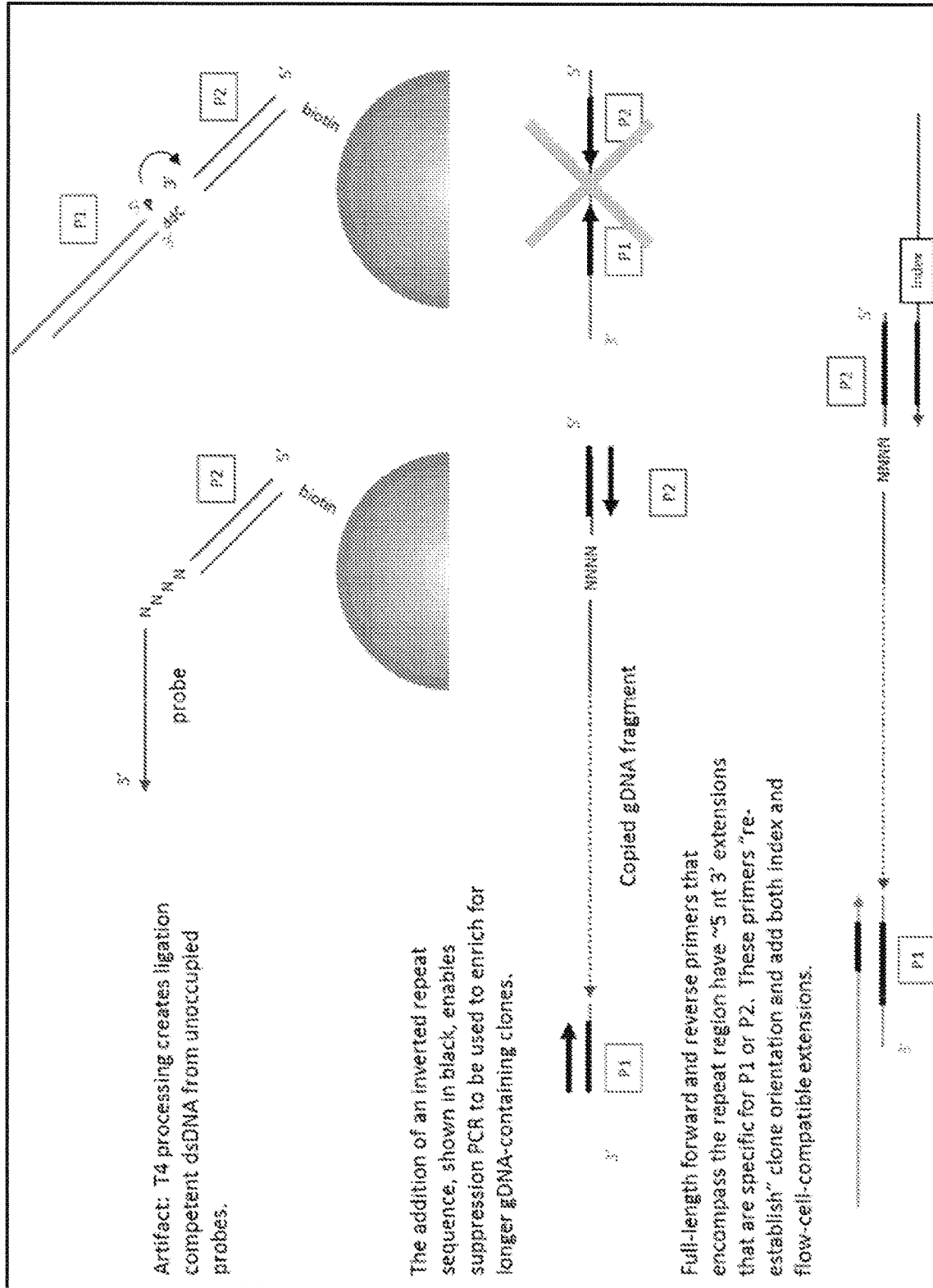
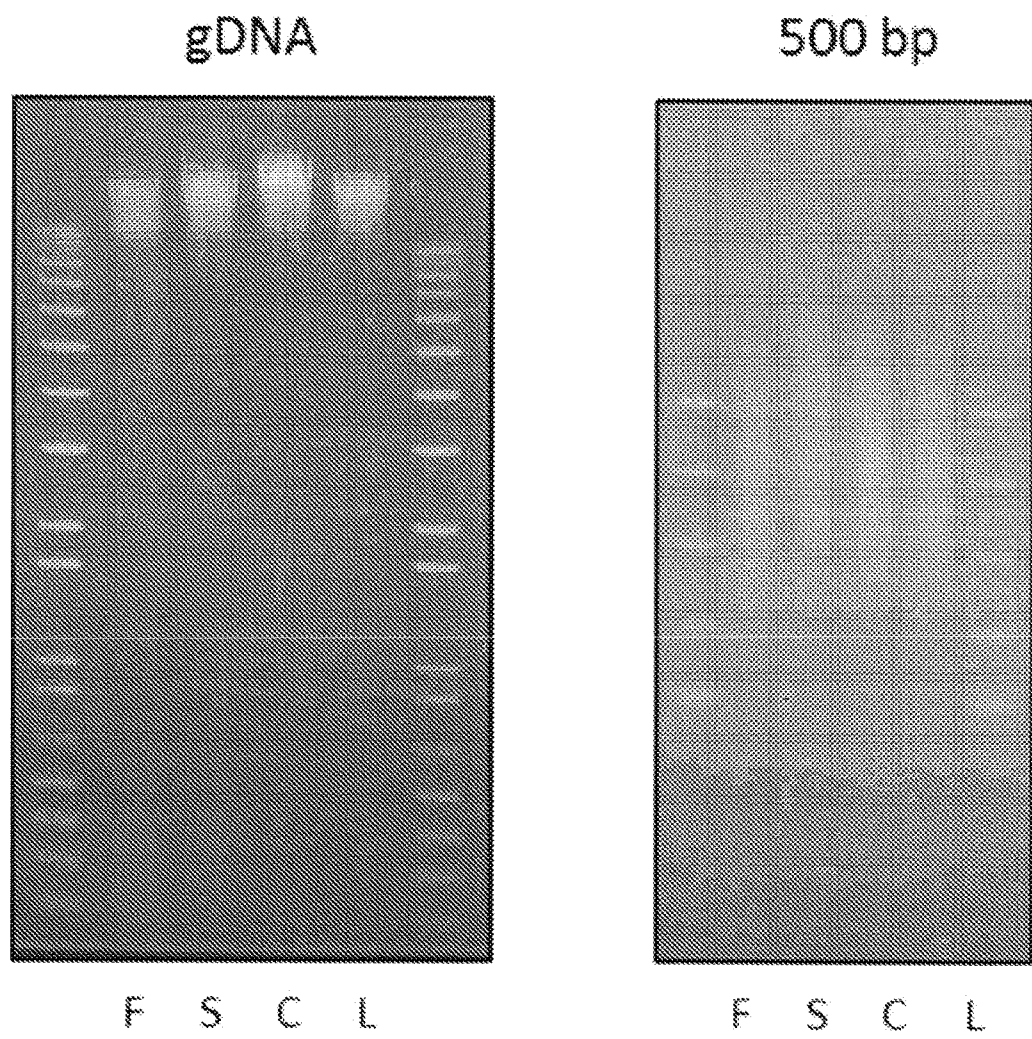


FIG. 36

*FIG. 37*

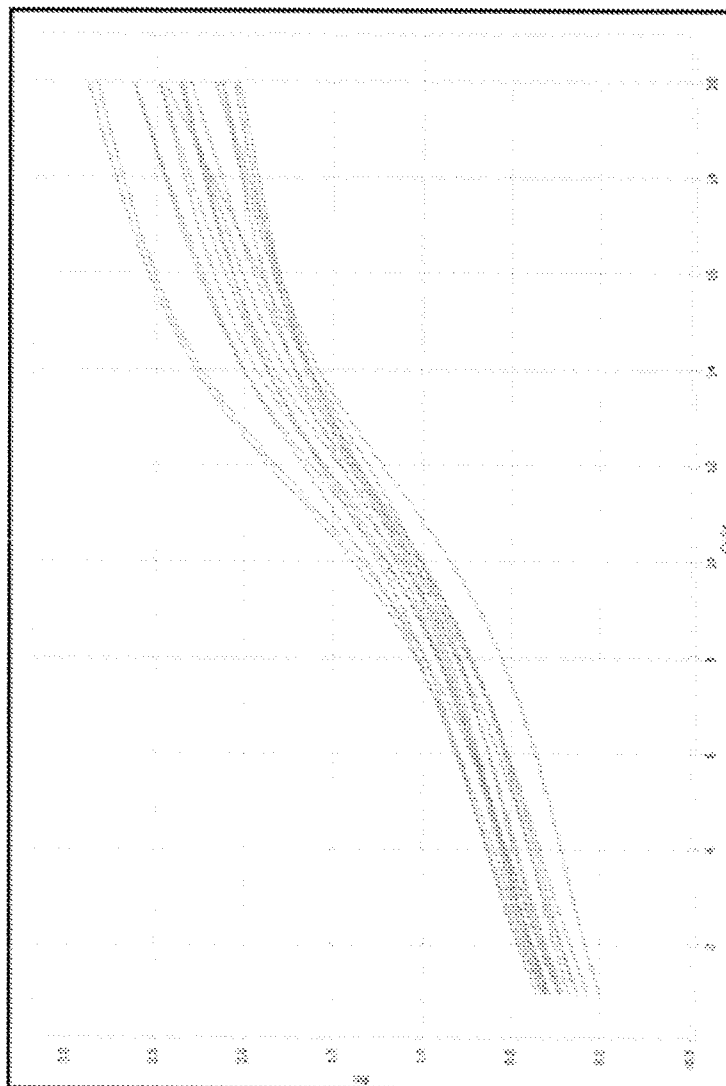


FIG. 38

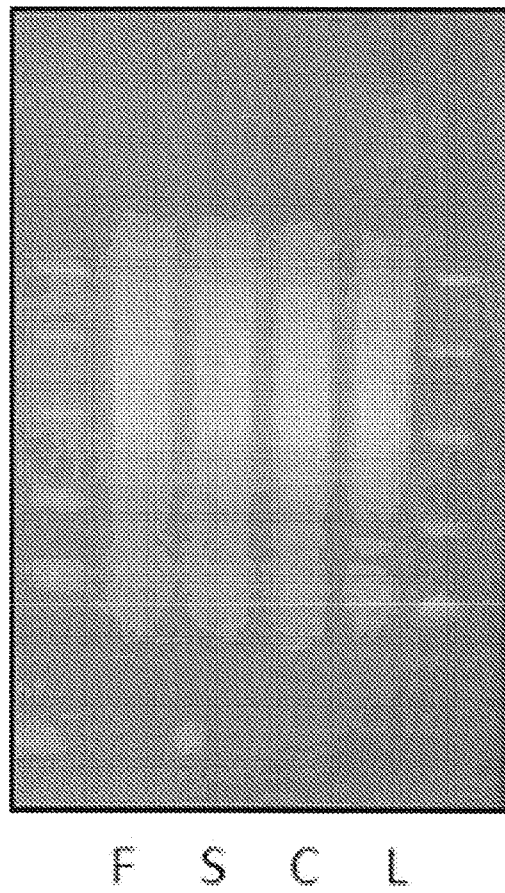
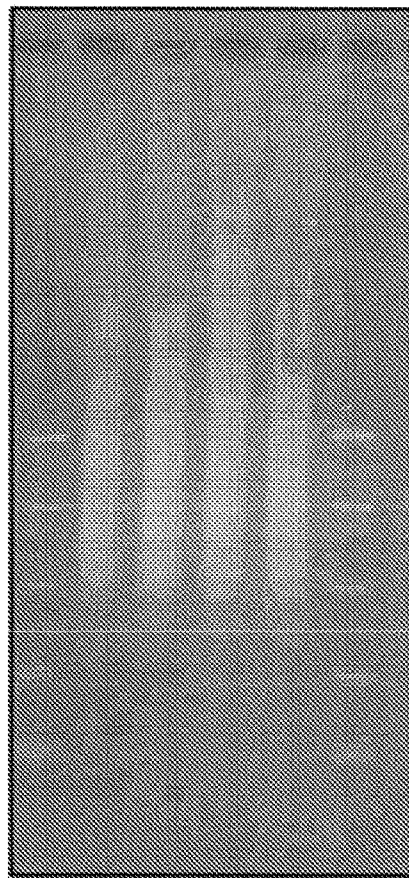


*FIG. 39*



F S C L

*FIG. 40*

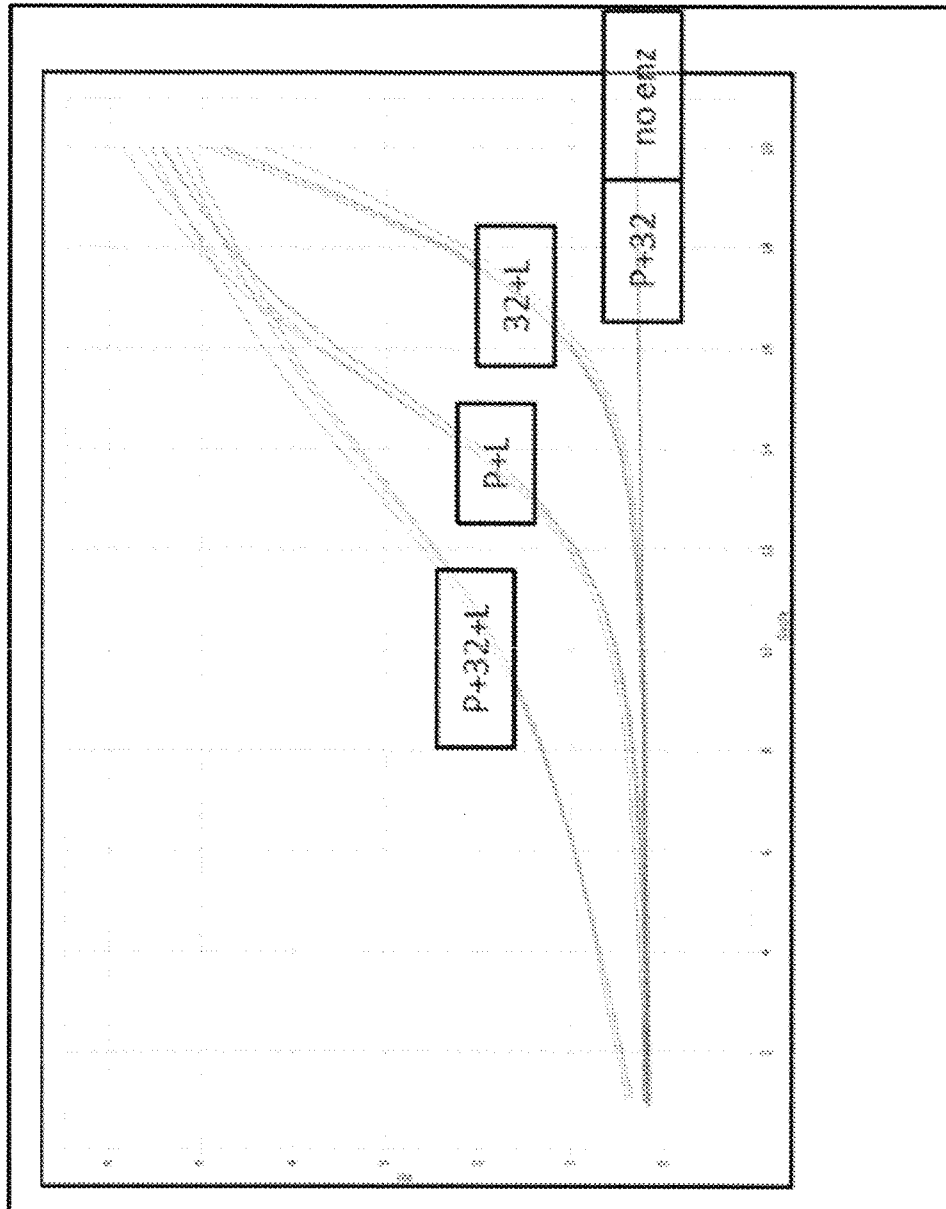


FIG. 41

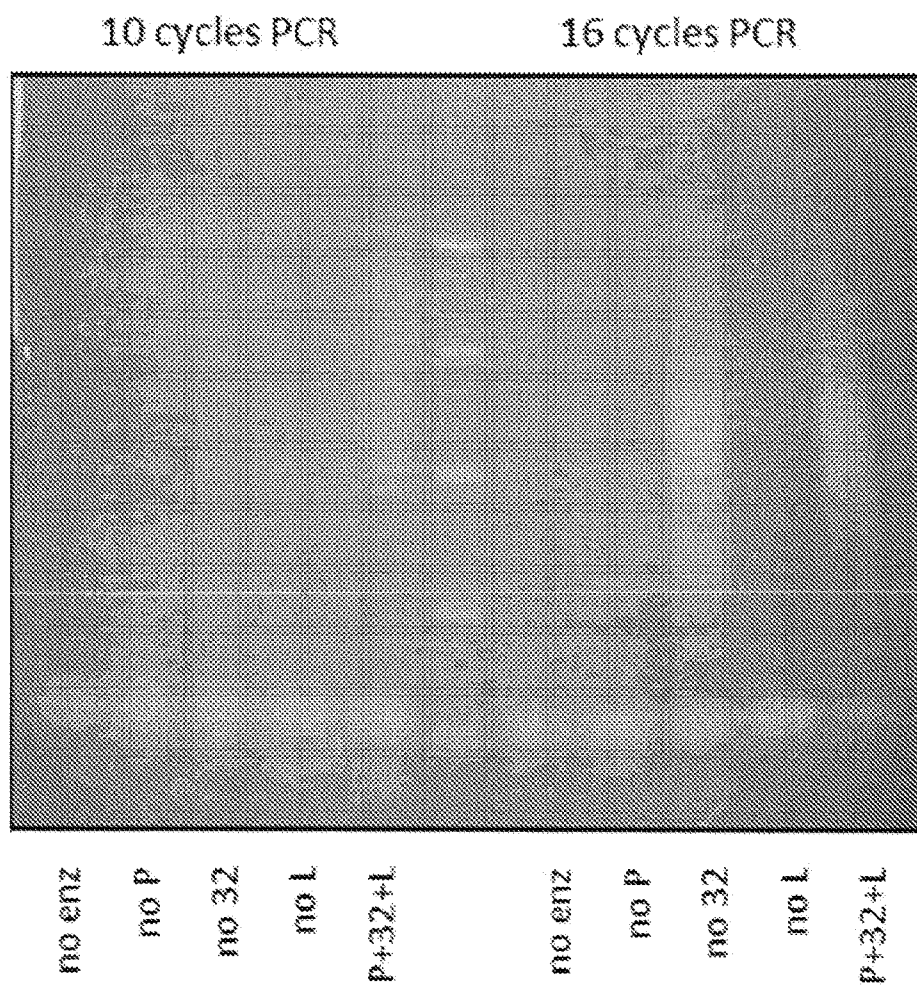
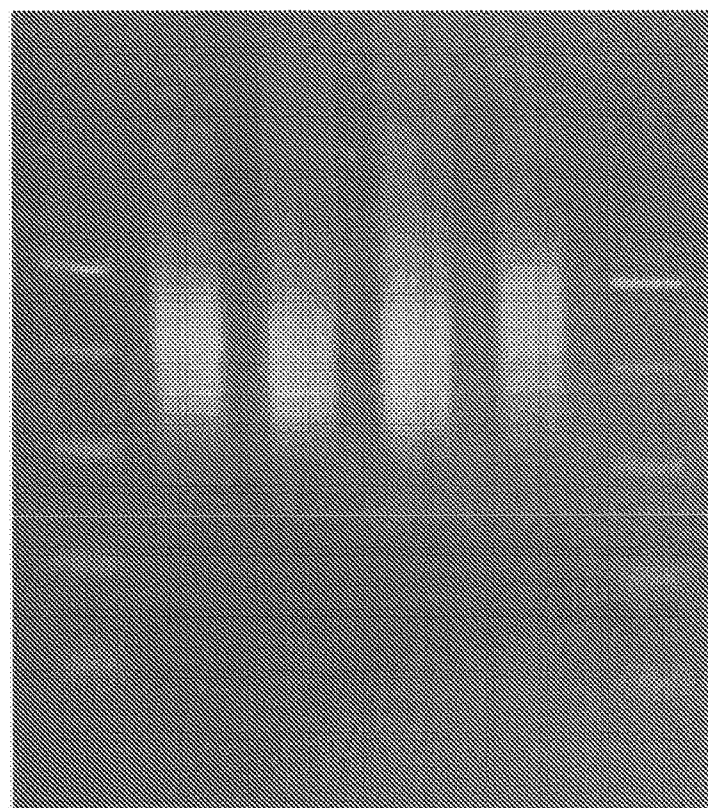
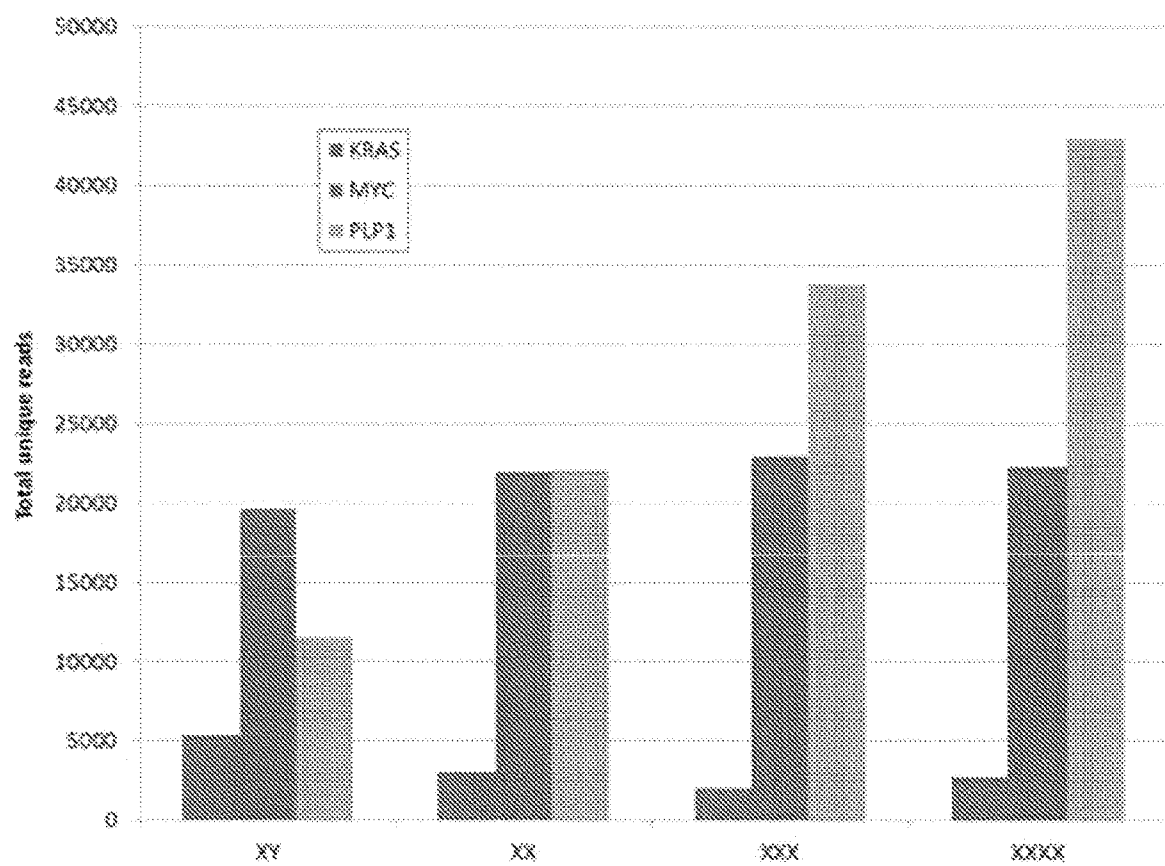


FIG. 42



11 13 15 16

*FIG. 43*

*FIG. 44*

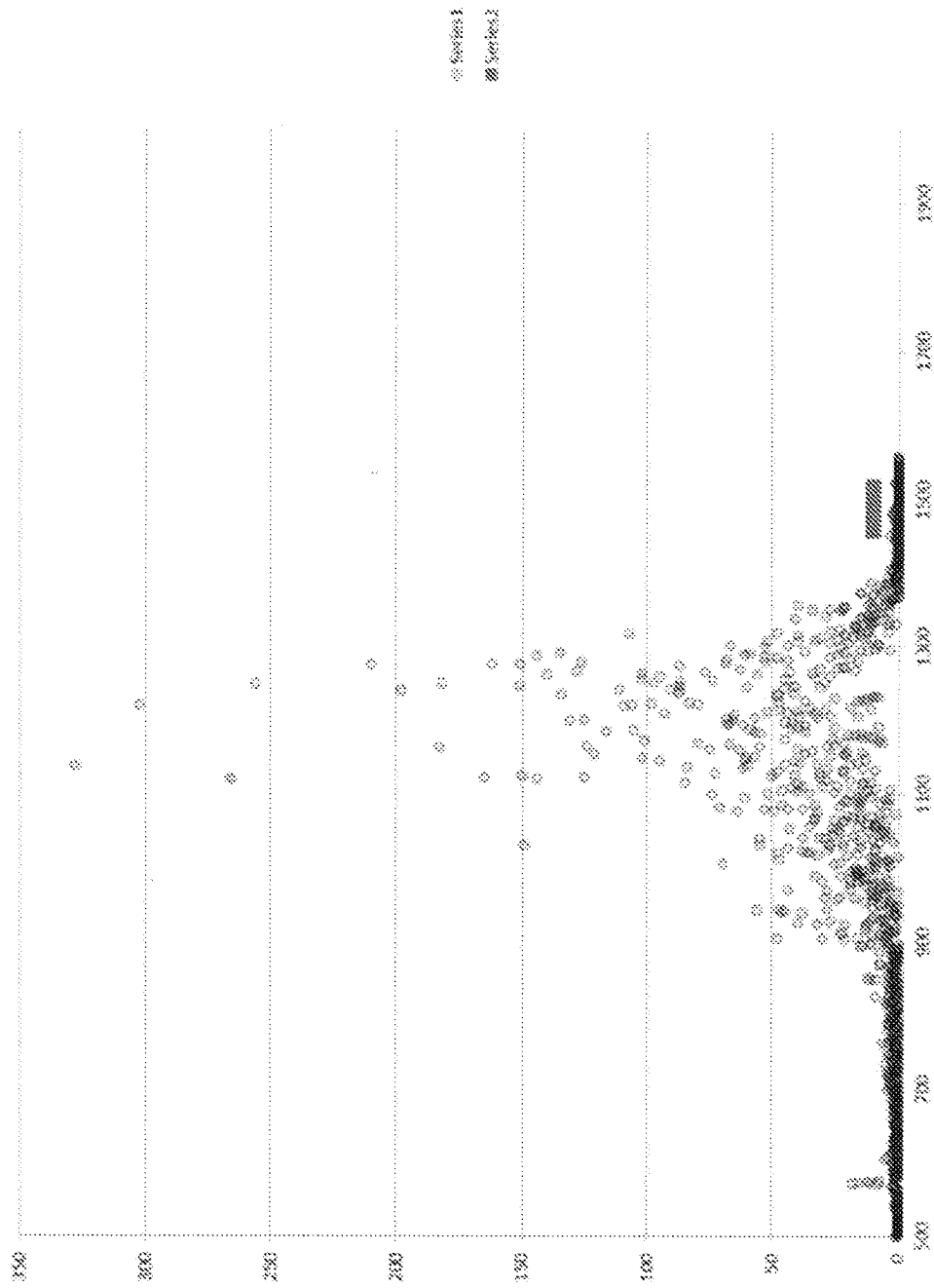


FIG. 45

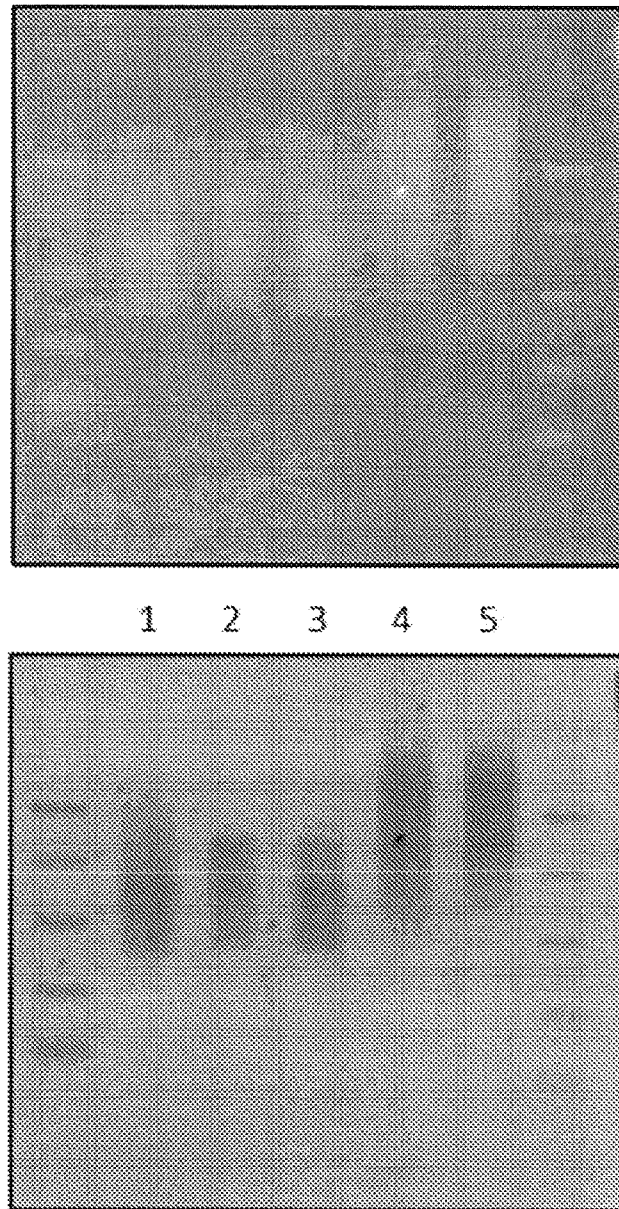


Figure 1. DNA gel of RNA-seq samples.

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*

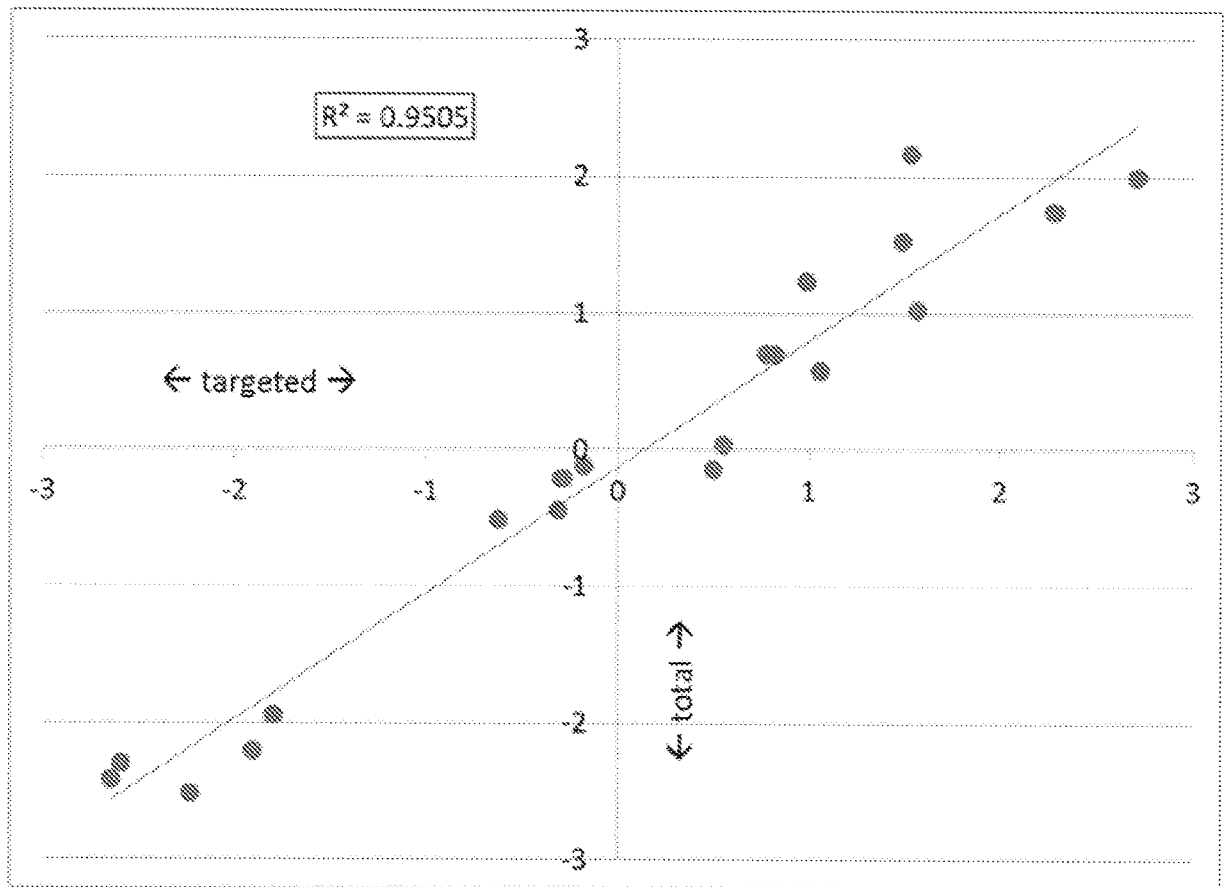


FIG. 47

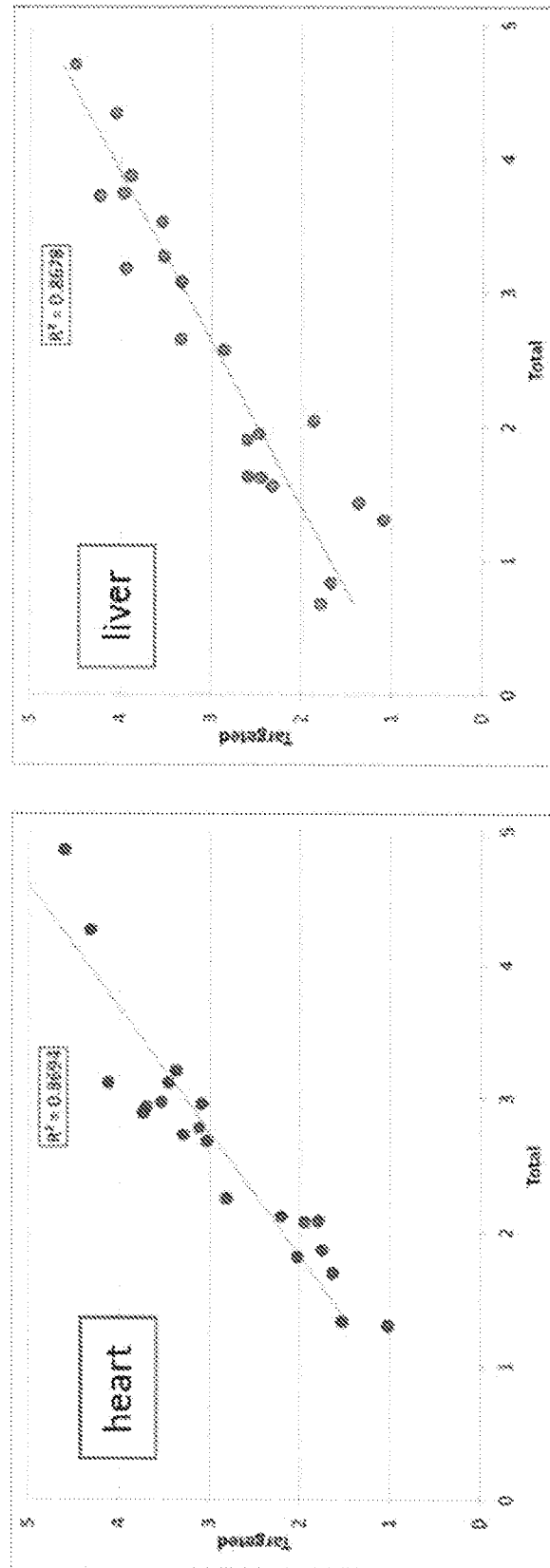


FIG. 48