

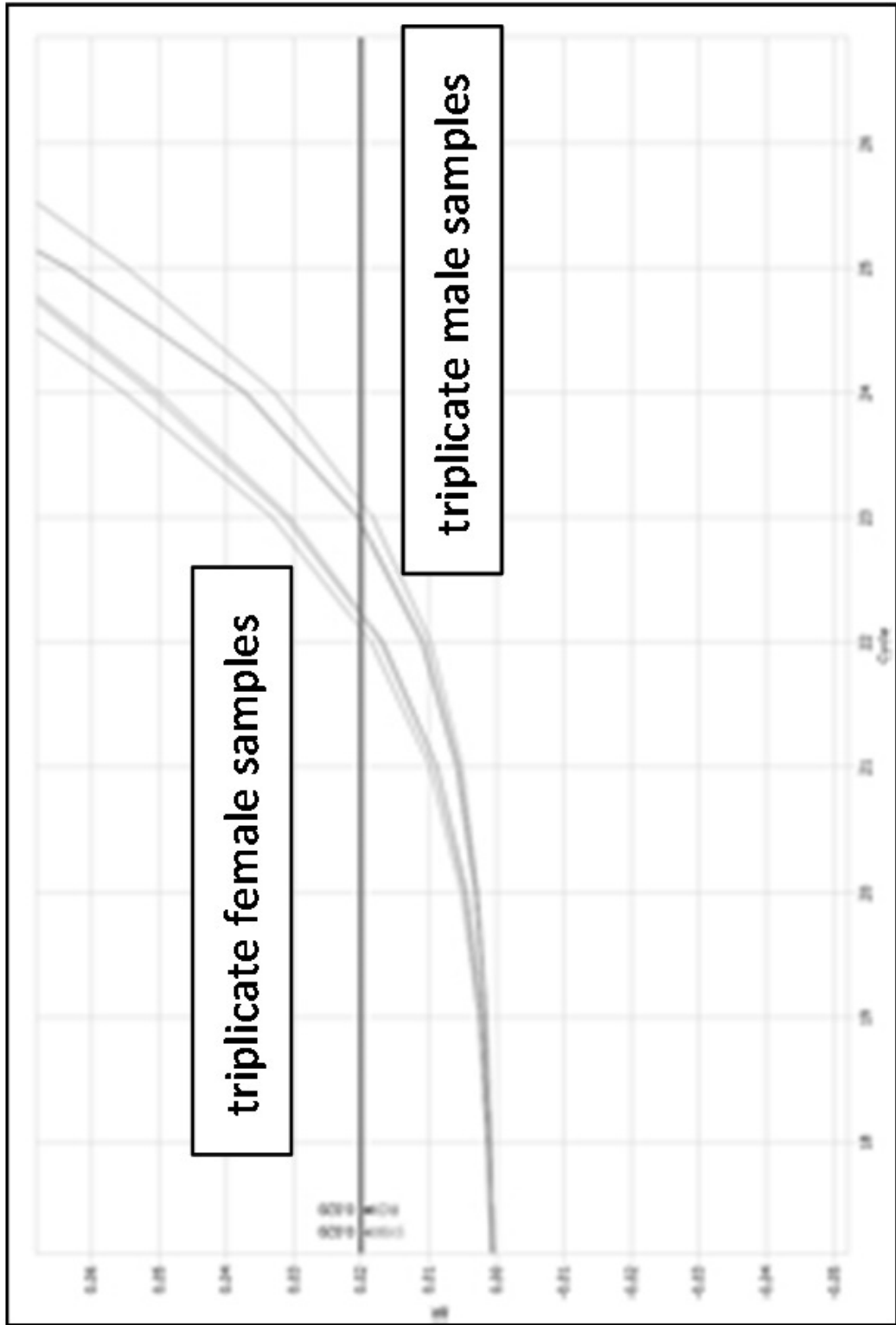
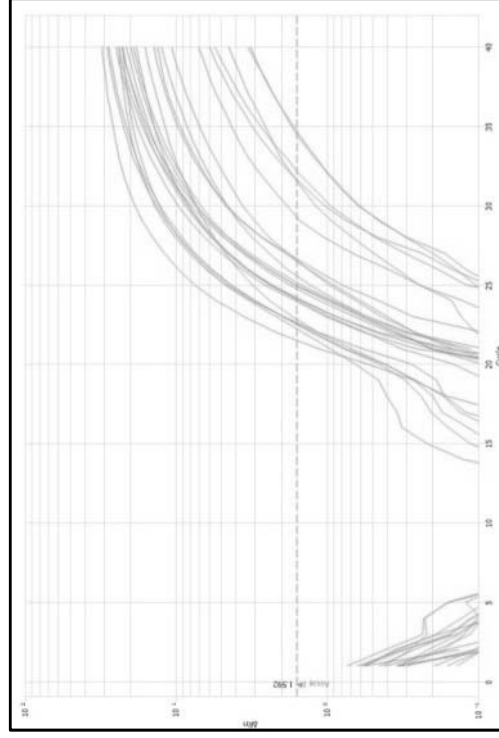


FIG. 9

B



A

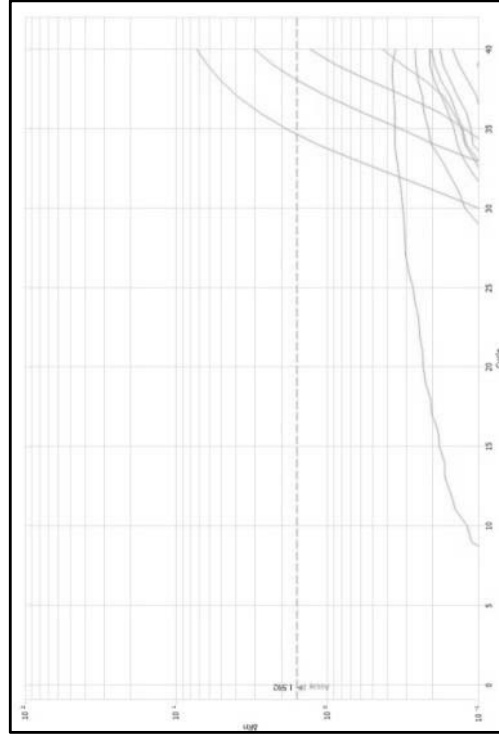


FIG. 11

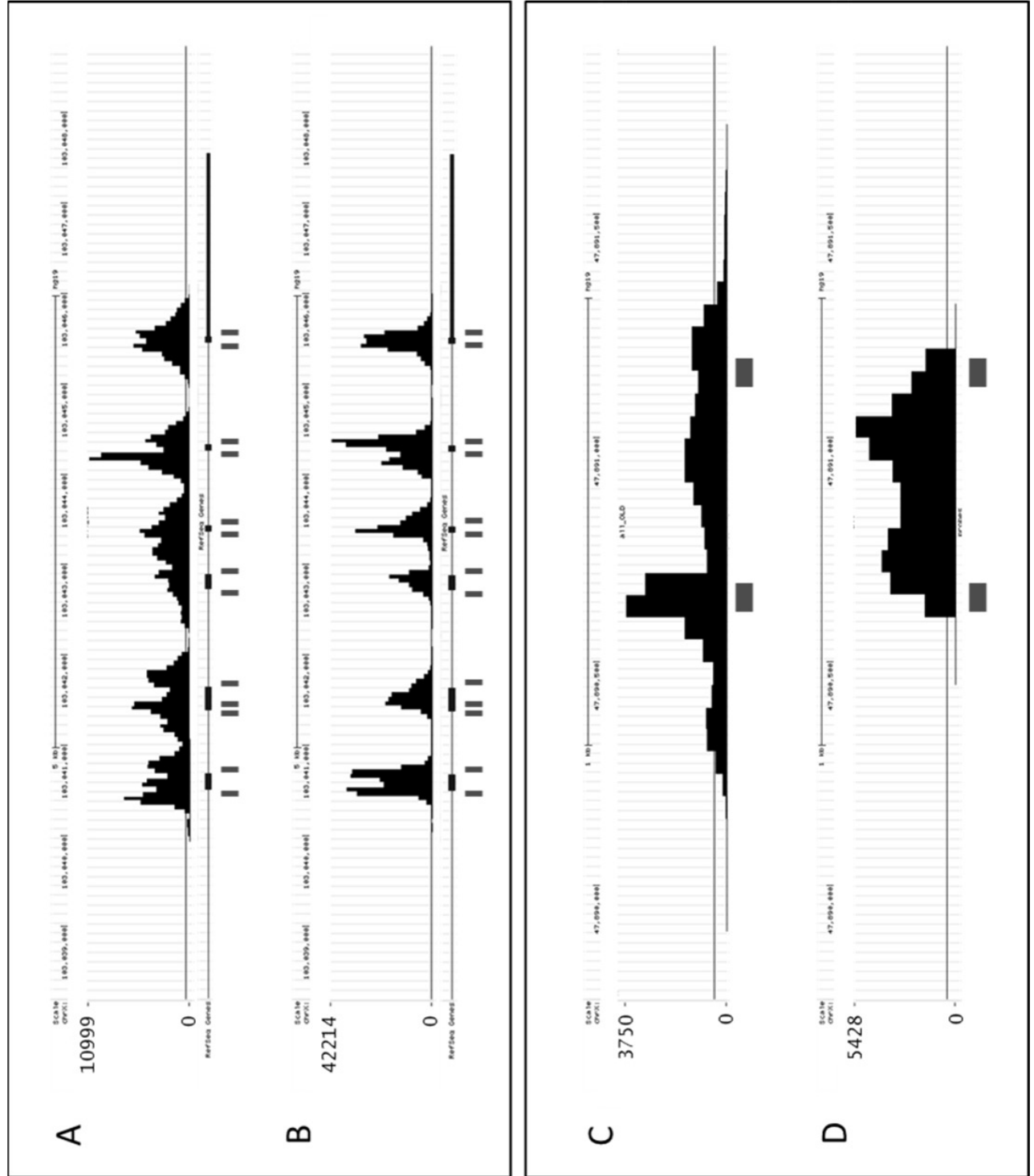
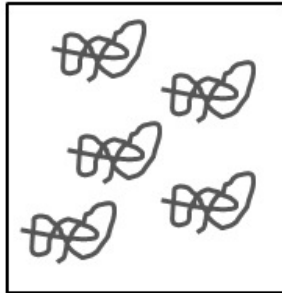


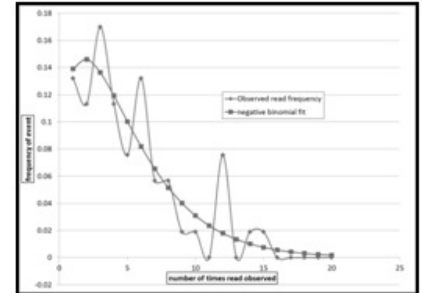
FIG. 18



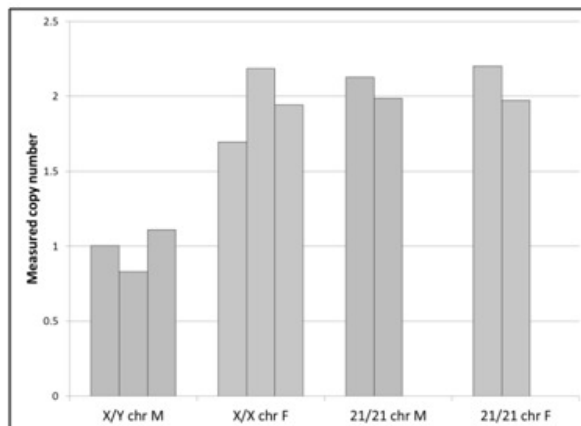
Low input genome libraries (e. g. ~50-100 genome equivalents) created with molecular annotation and subsequently captured with targeting probes.



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.

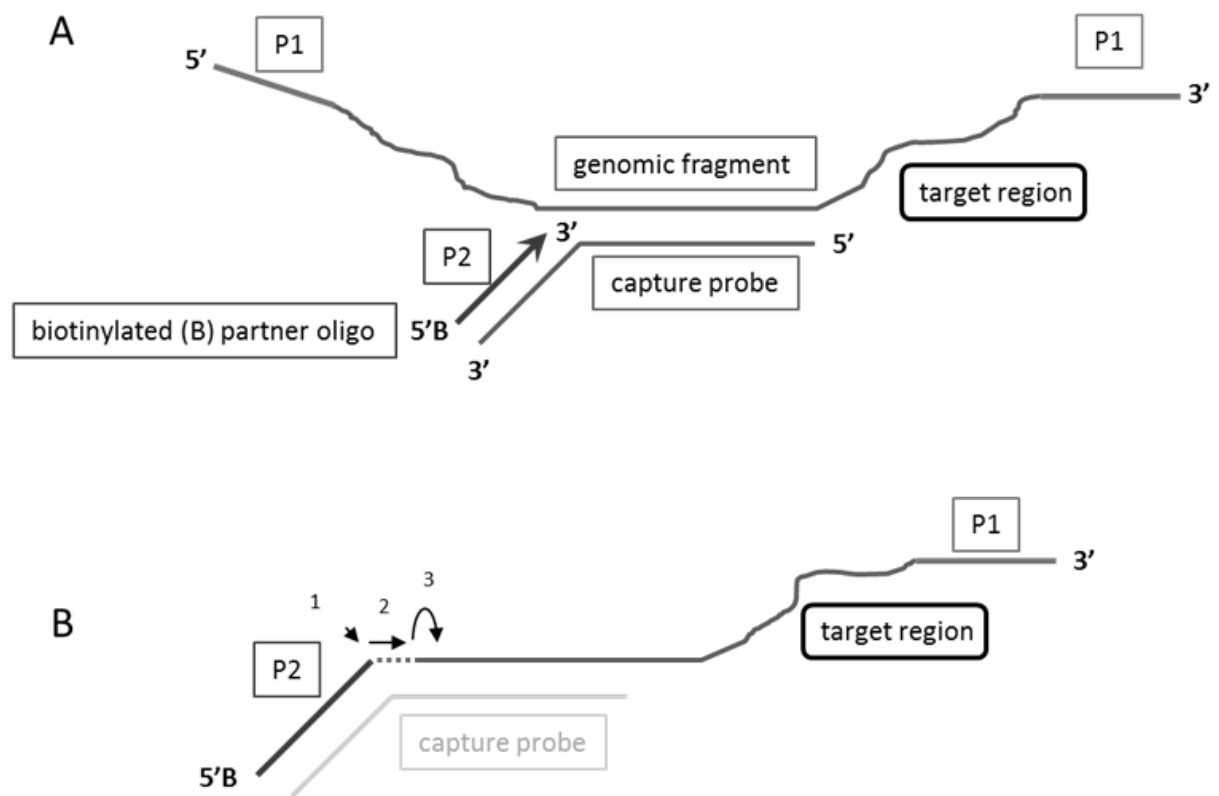


Observed read frequencies are fitted to a negative binomial distribution, which allows the “inferred class” of unobserved reads to be estimated. The number of observed reads + inferred reads = copy number.



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. 31*

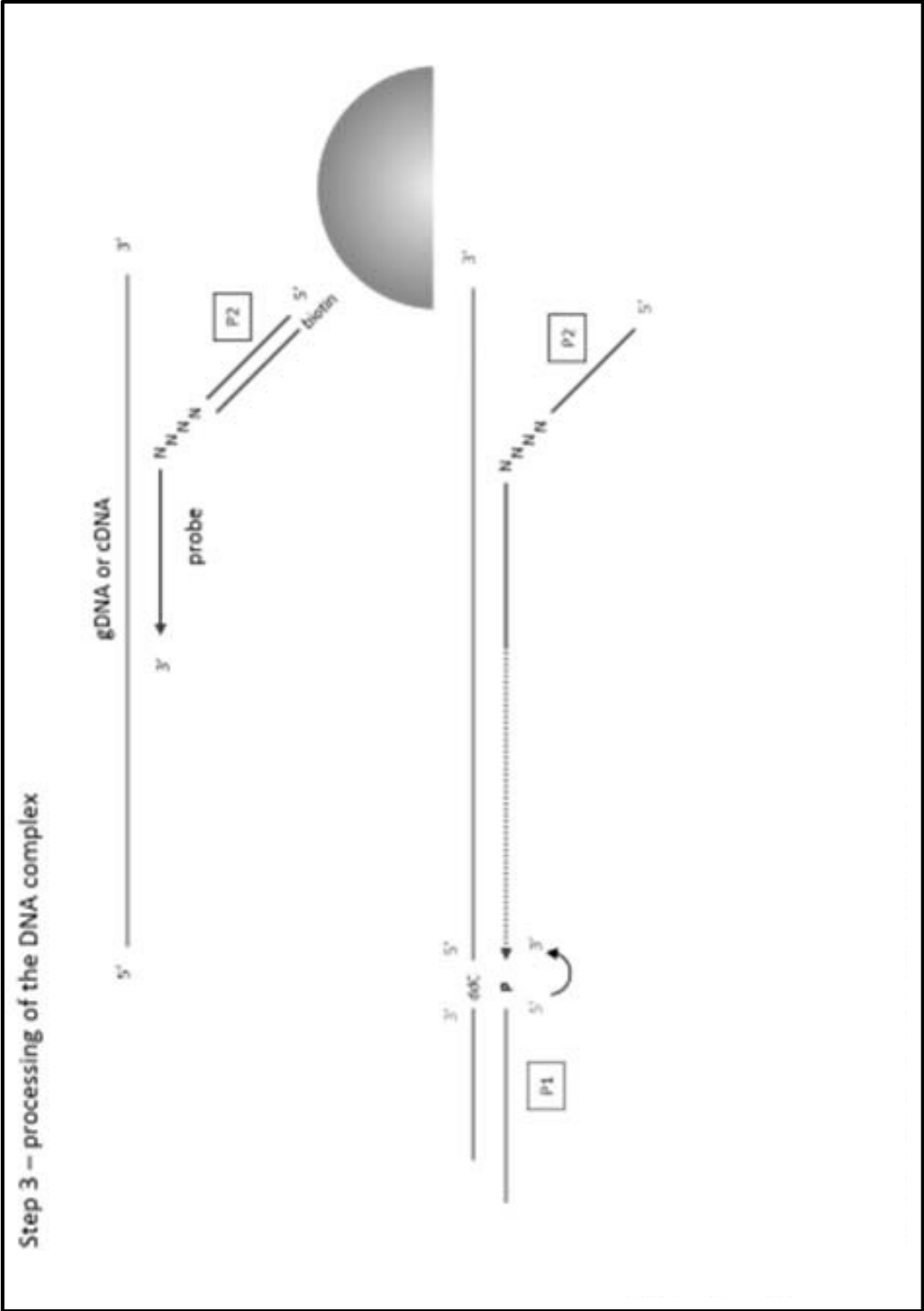


FIG. 34

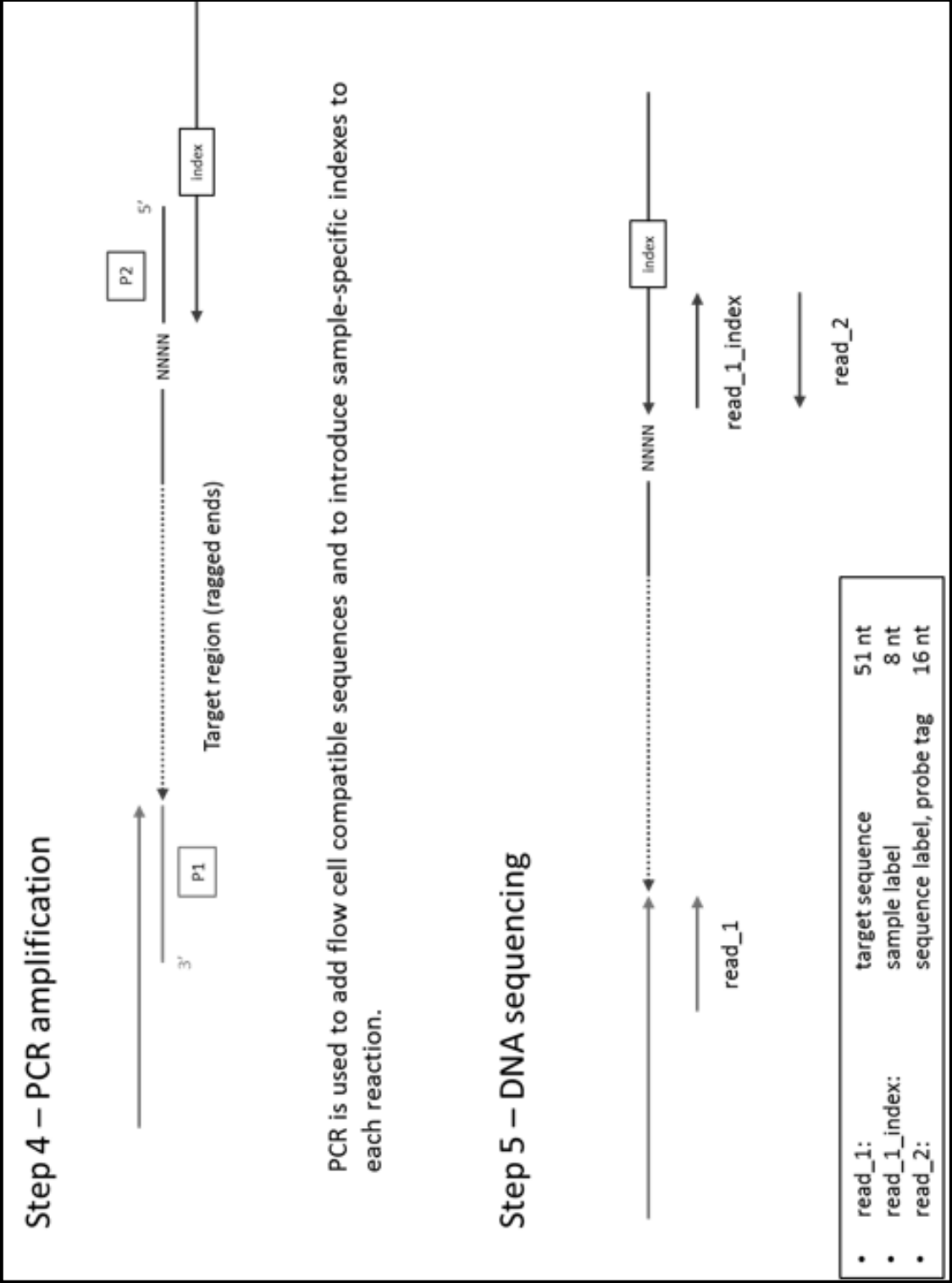


FIG. 35

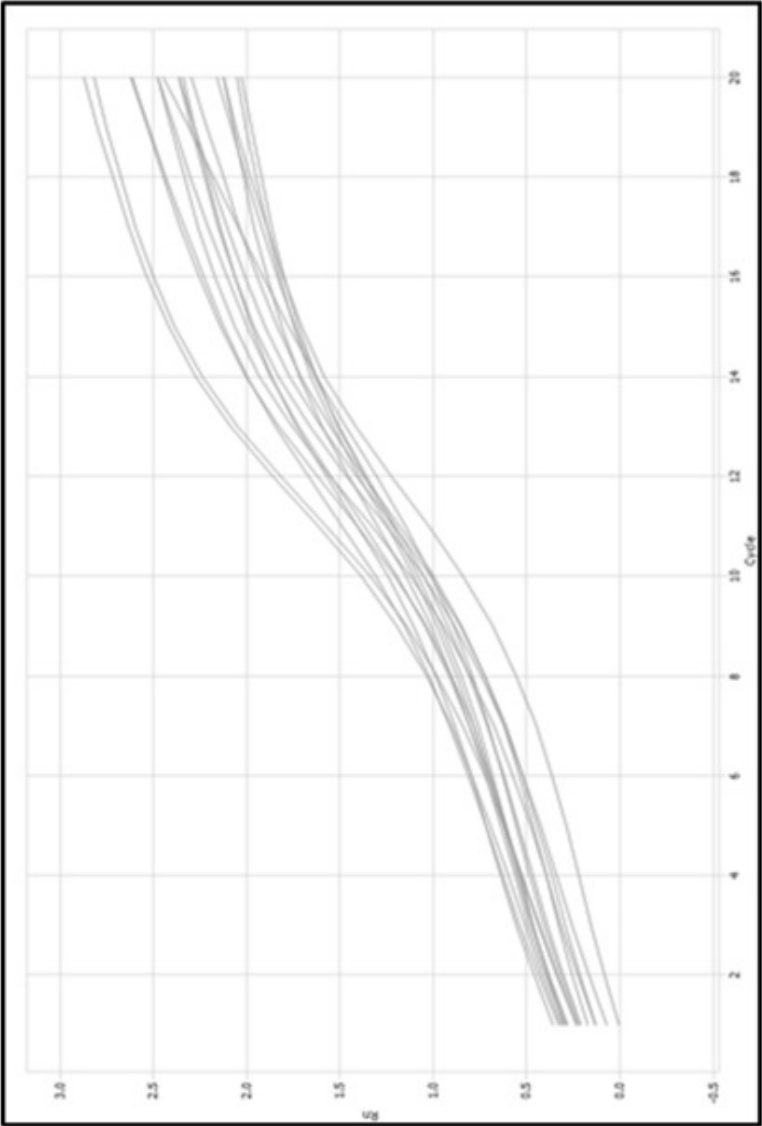


FIG. 38

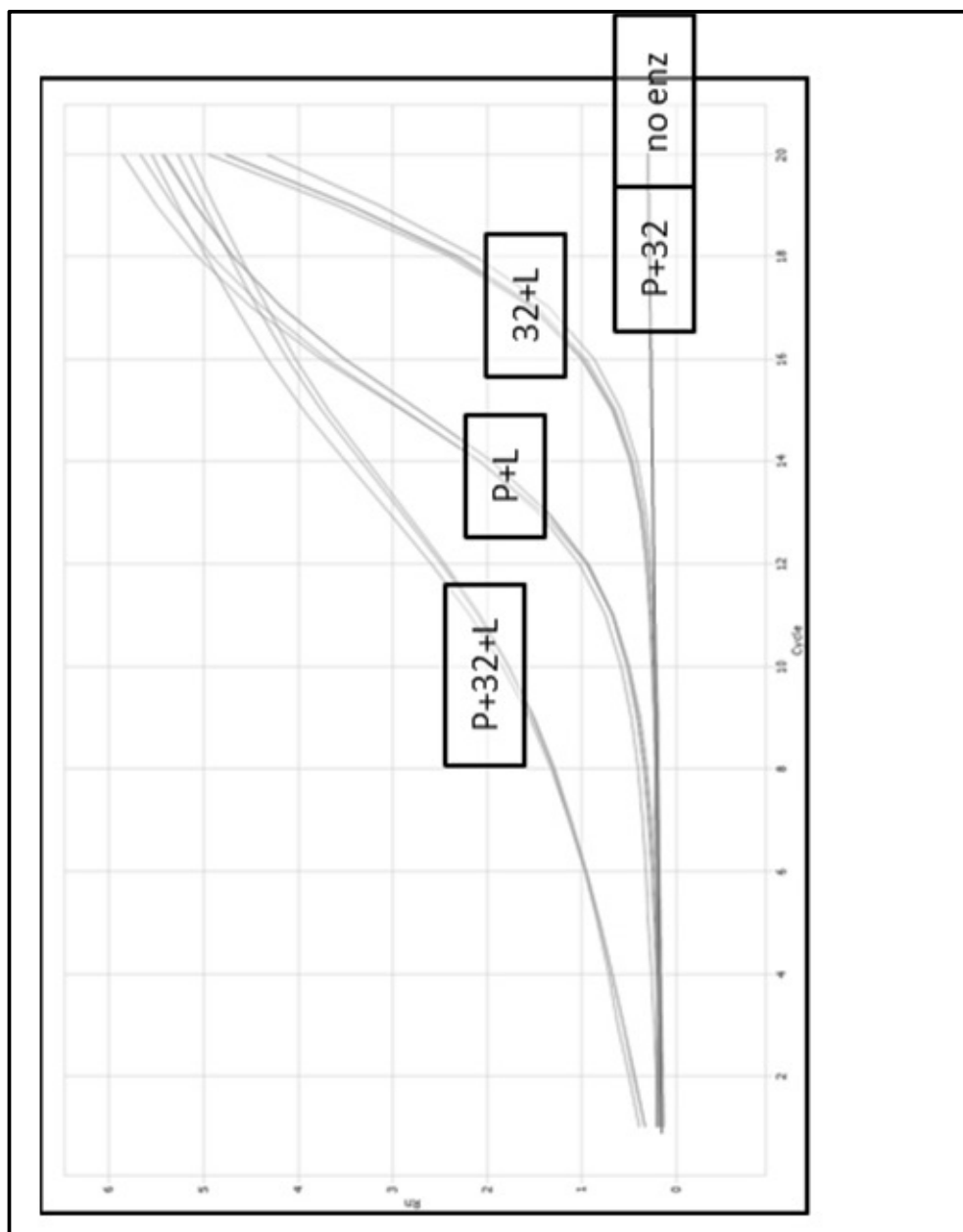


FIG. 41

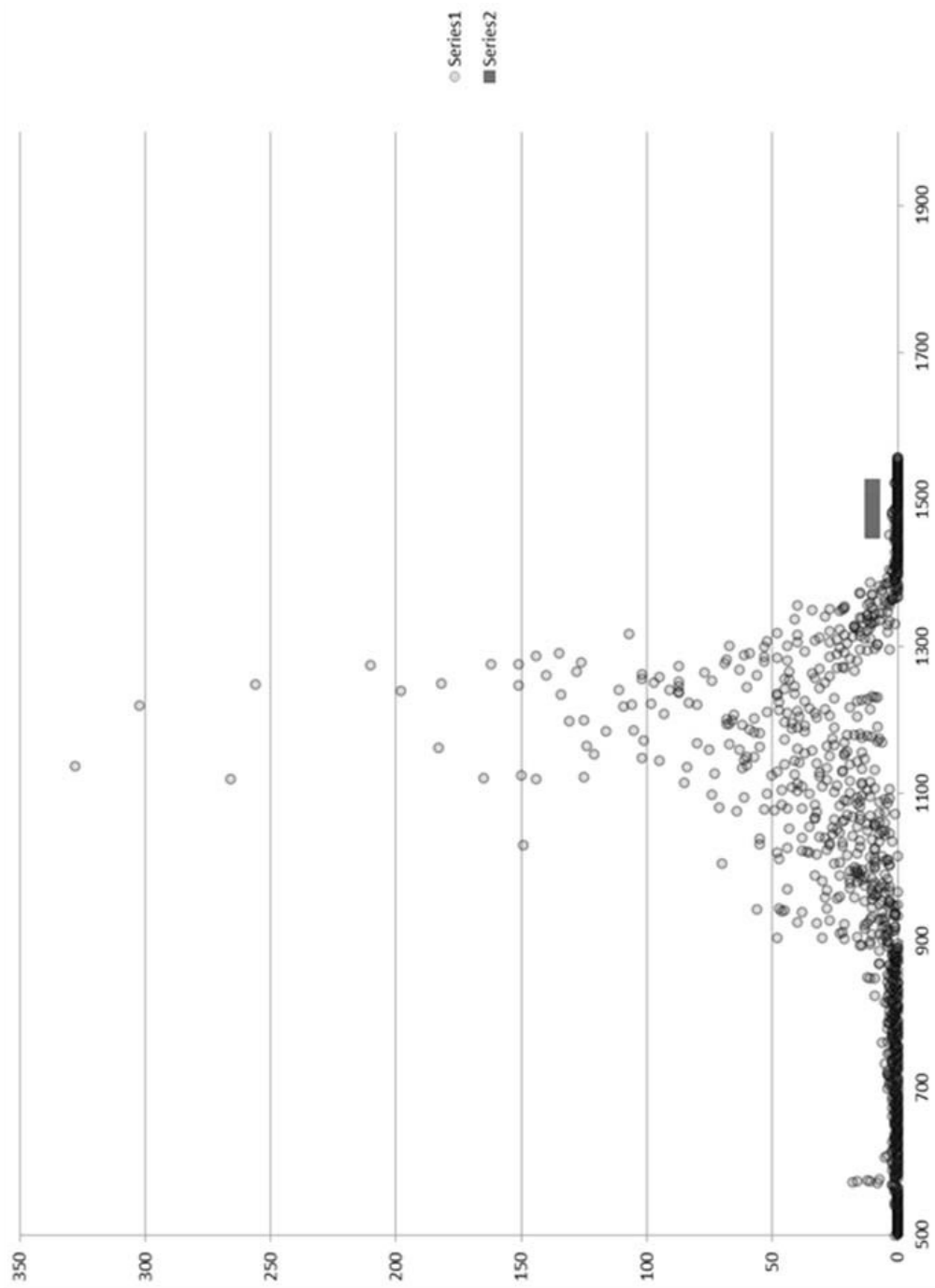


FIG. 45

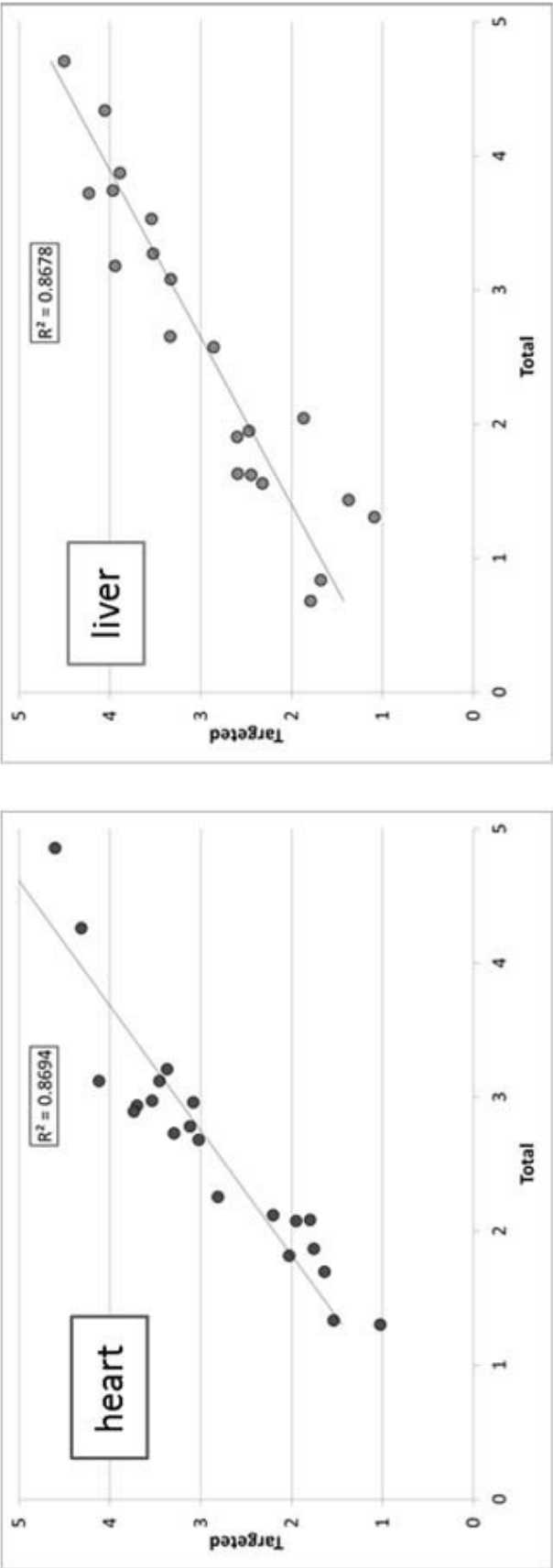


FIG. 48