

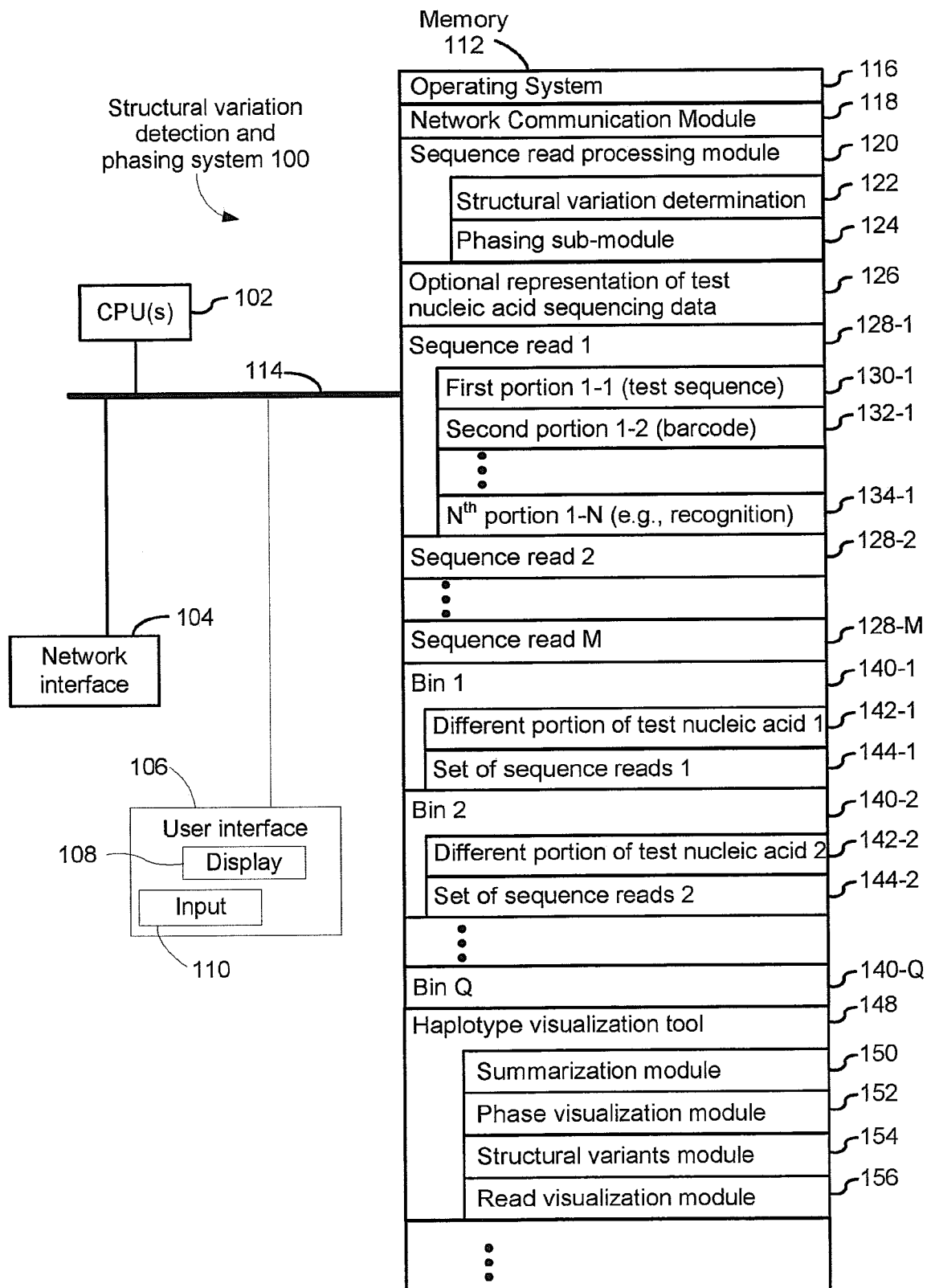


Figure 1

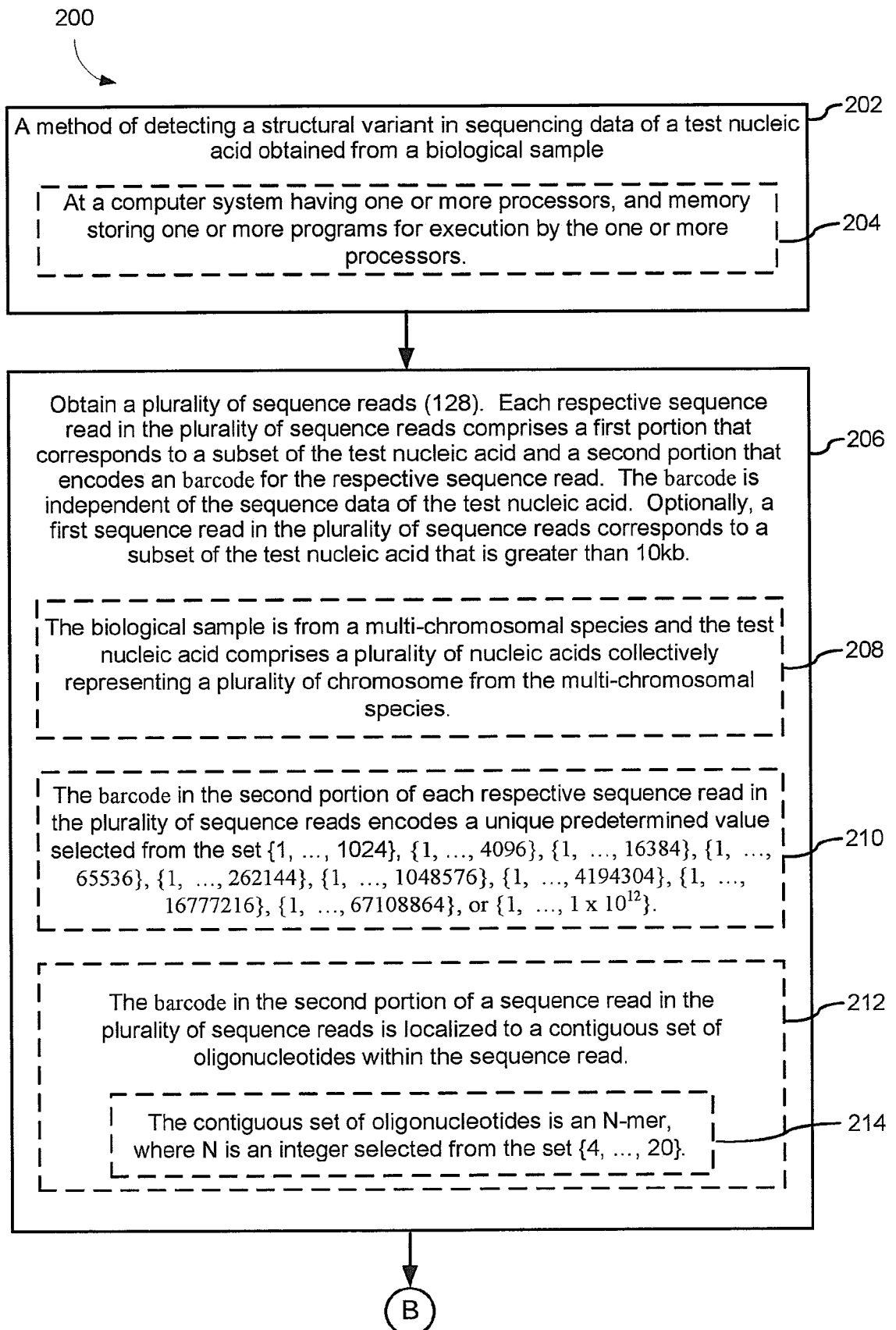


Figure 2A

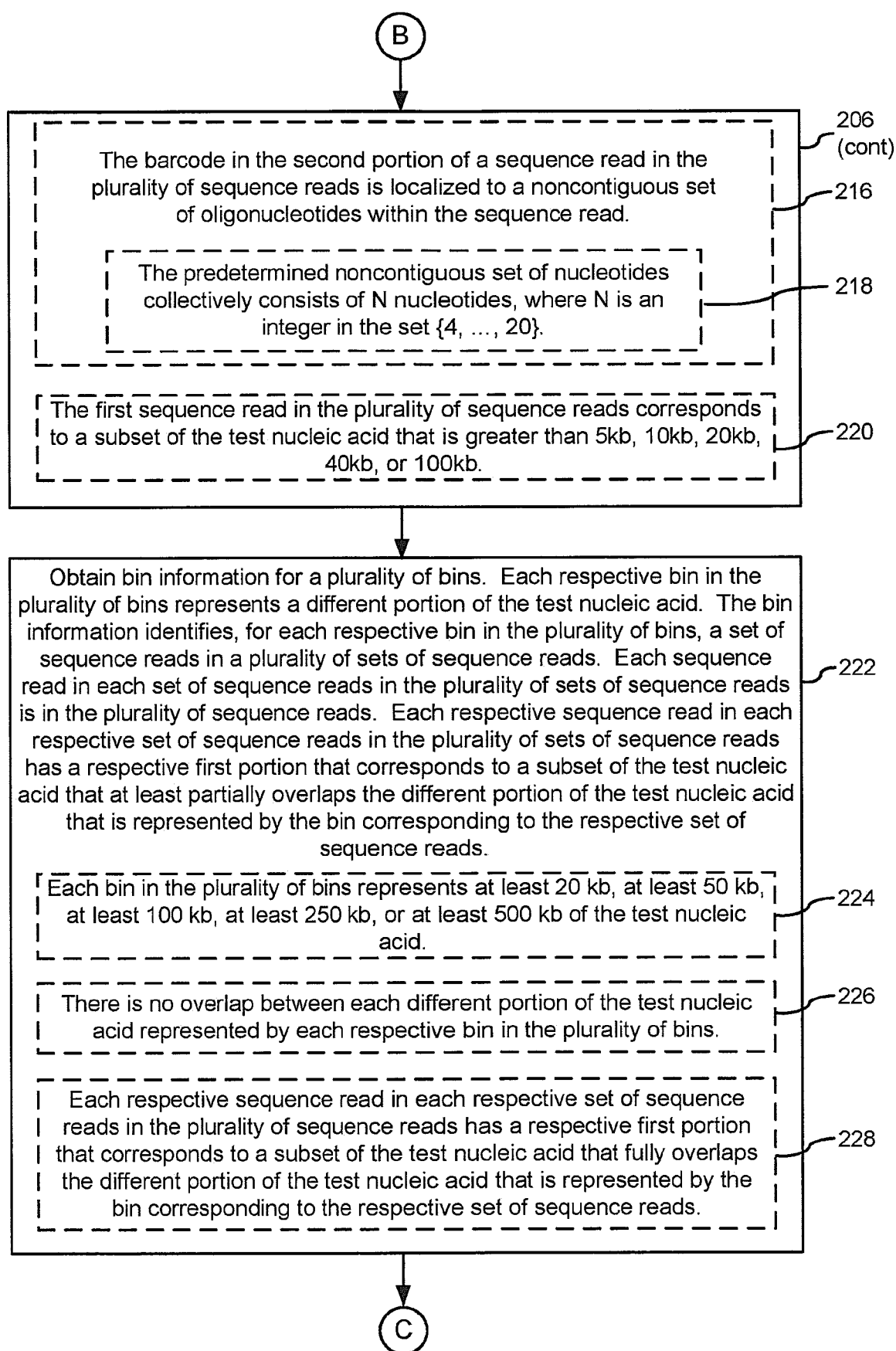


Figure 2B

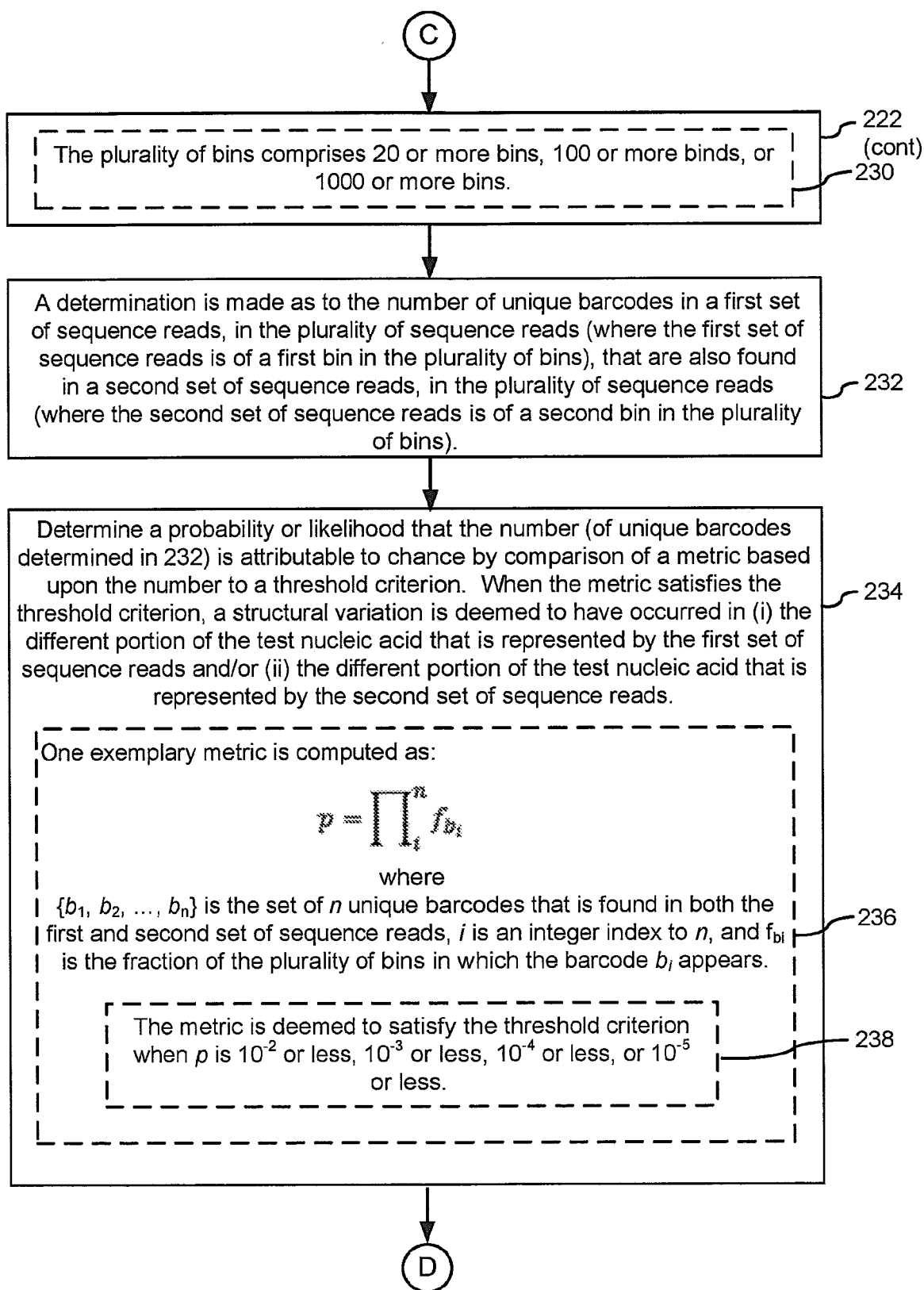


Figure 2C

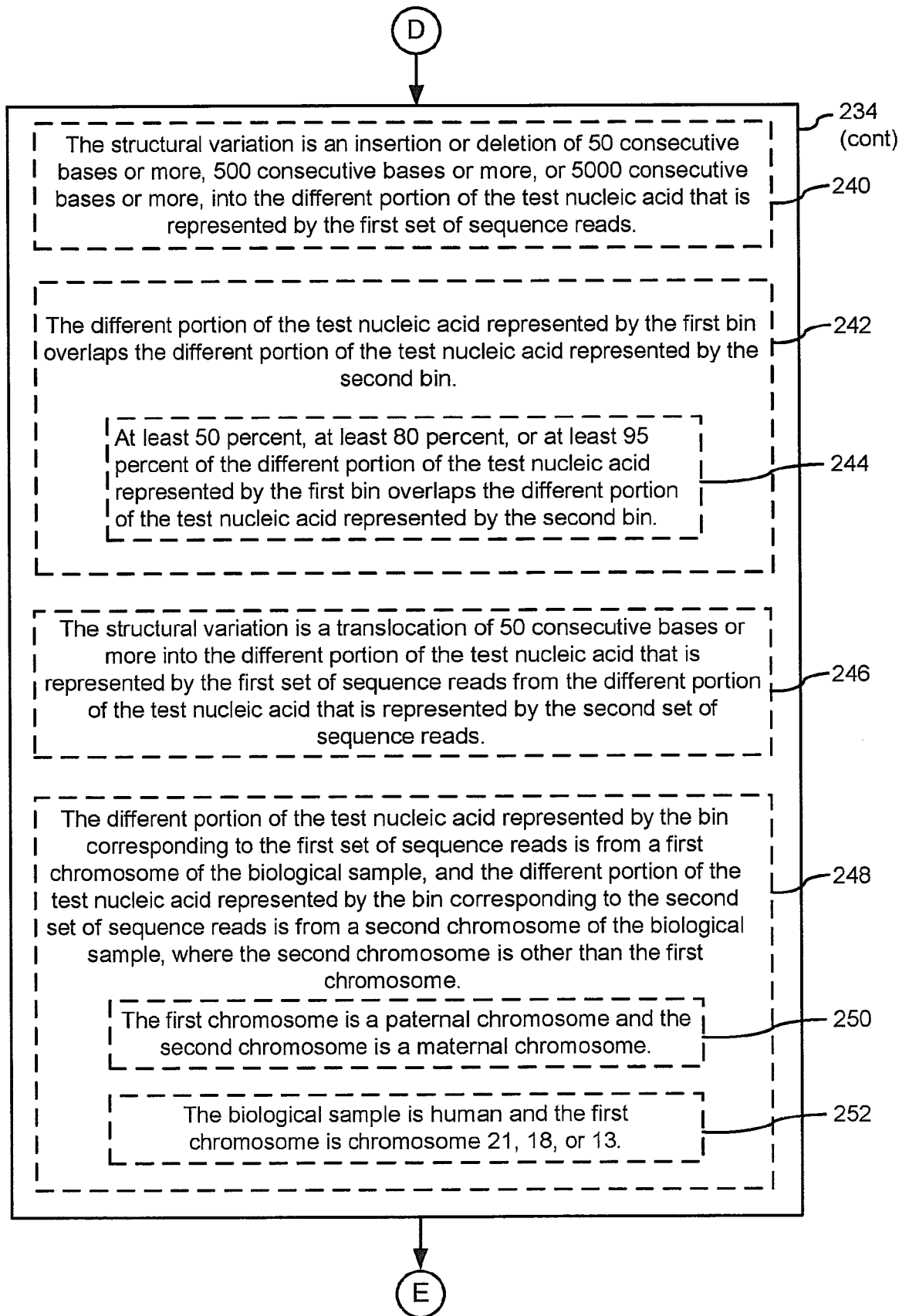


Figure 2D

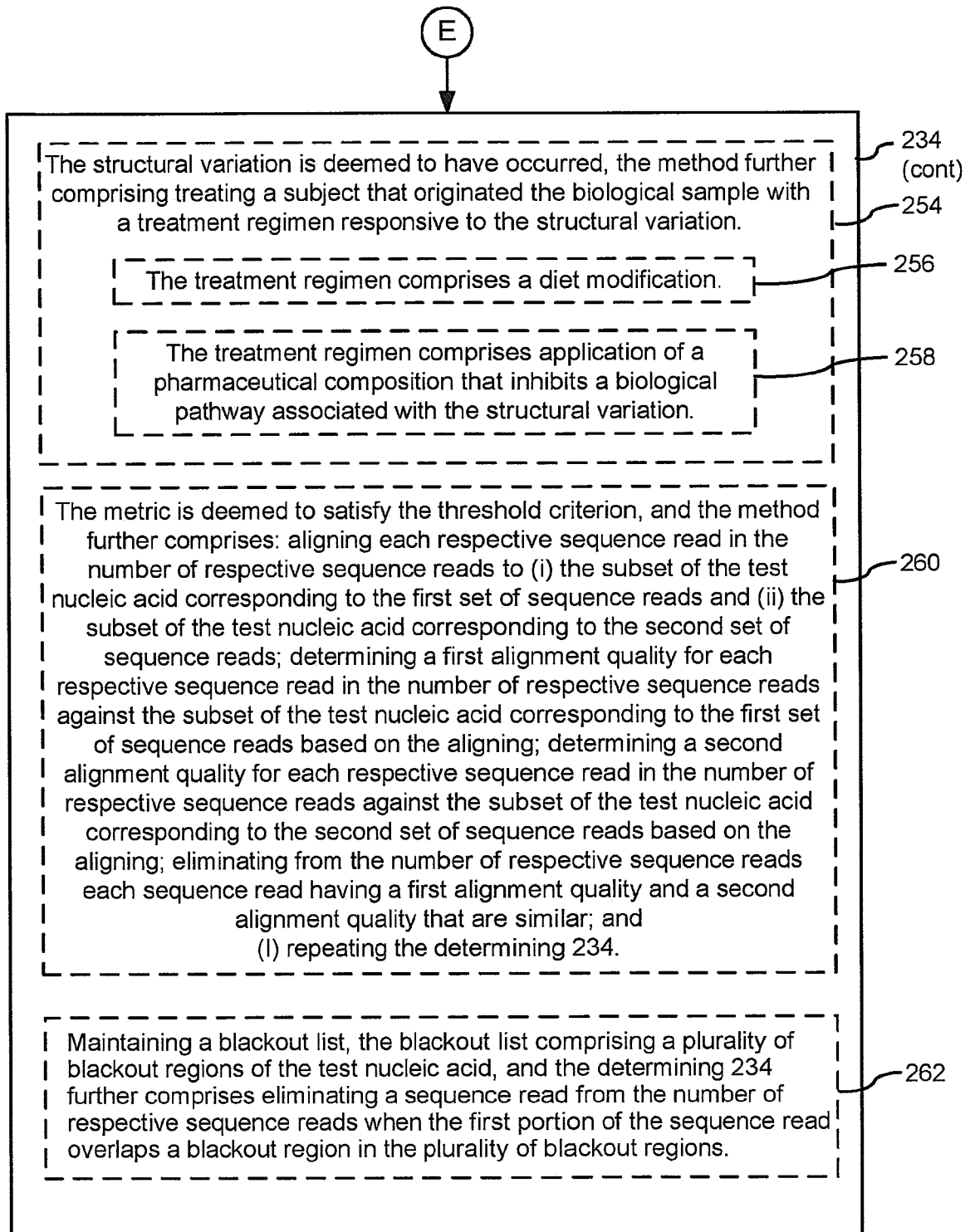


Figure 2E

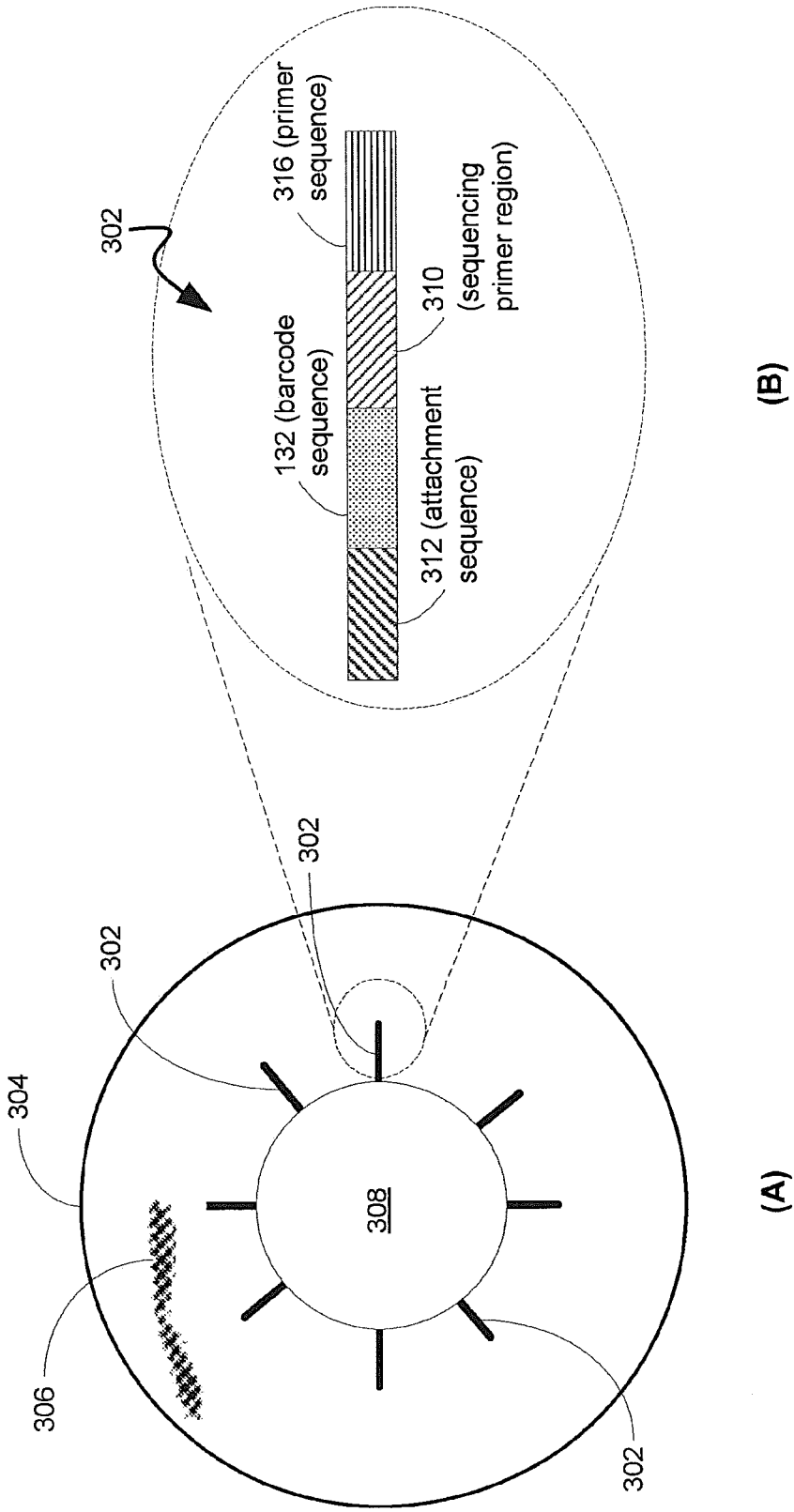


Figure 3A

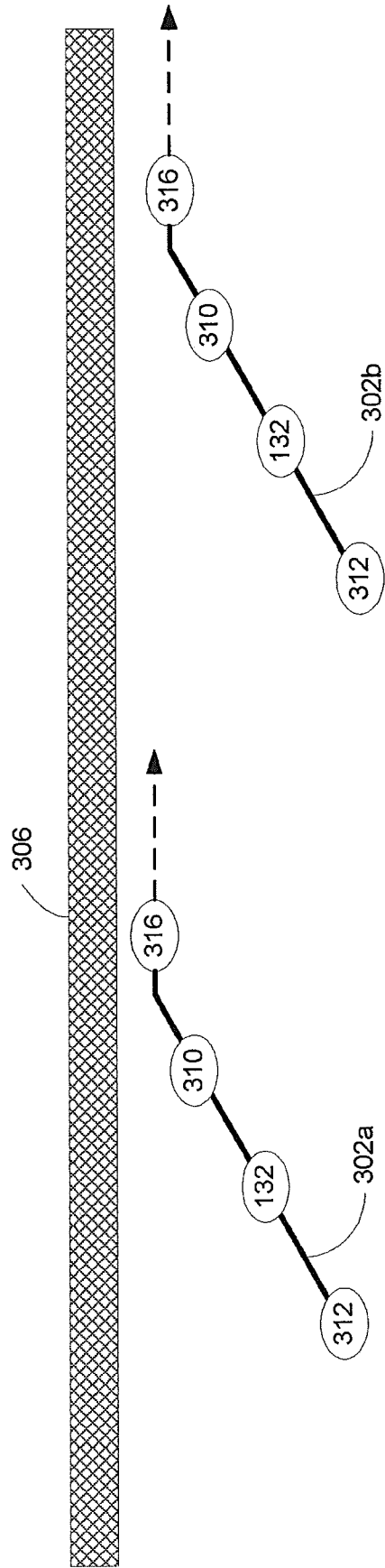


Figure 3B

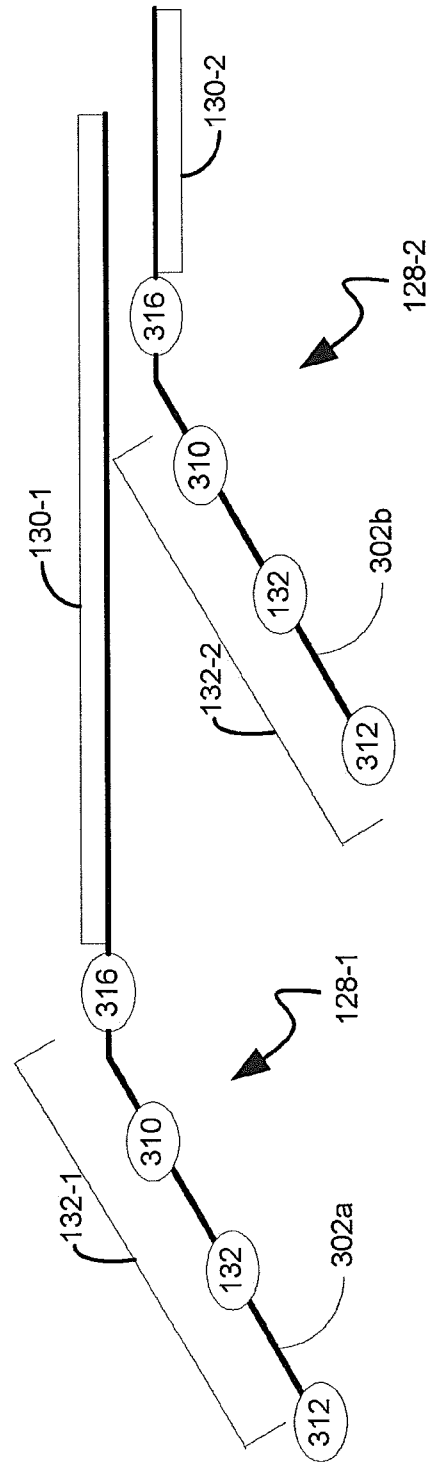


Figure 3C

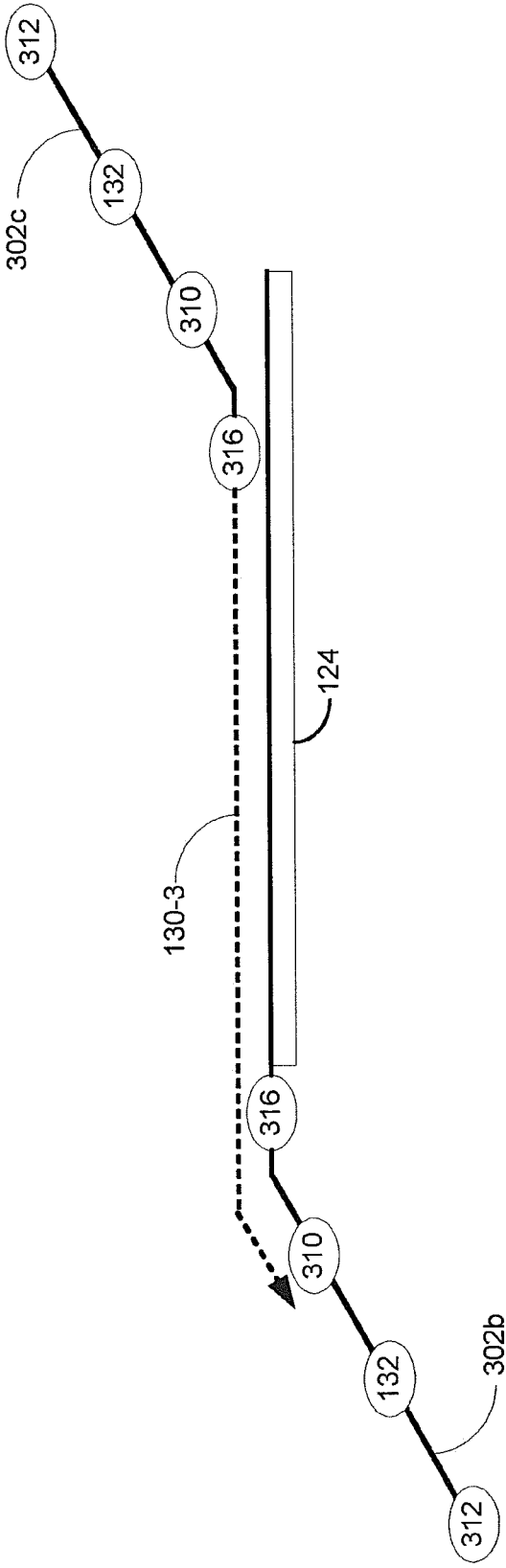


Figure 3D

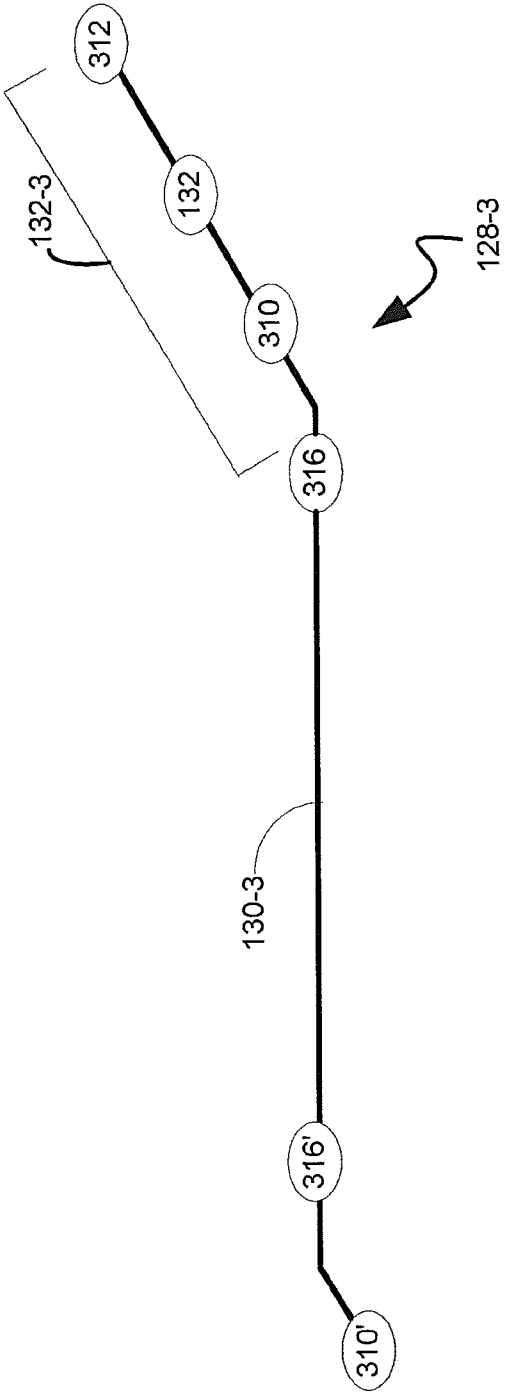
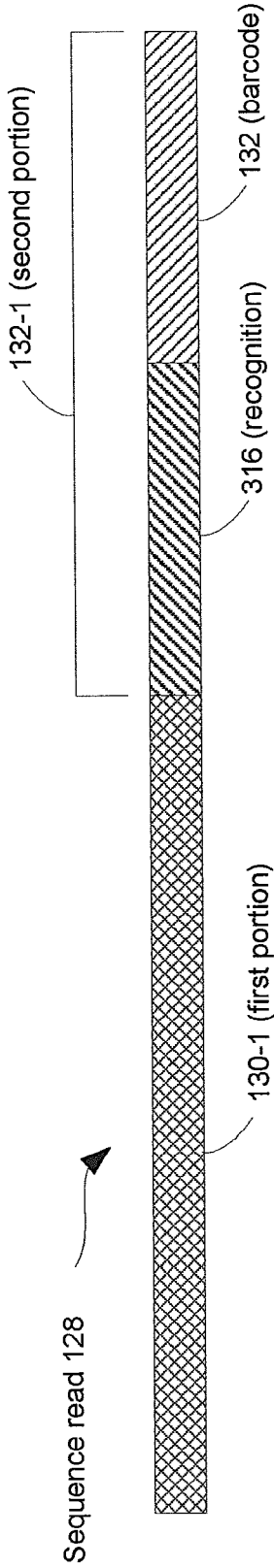
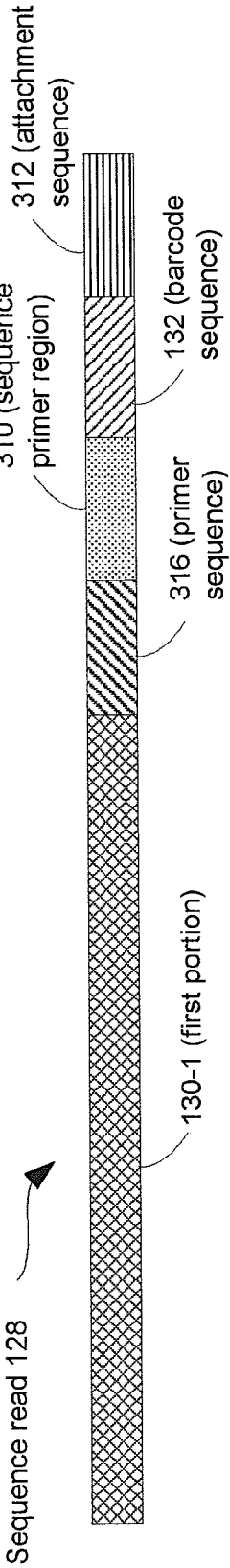
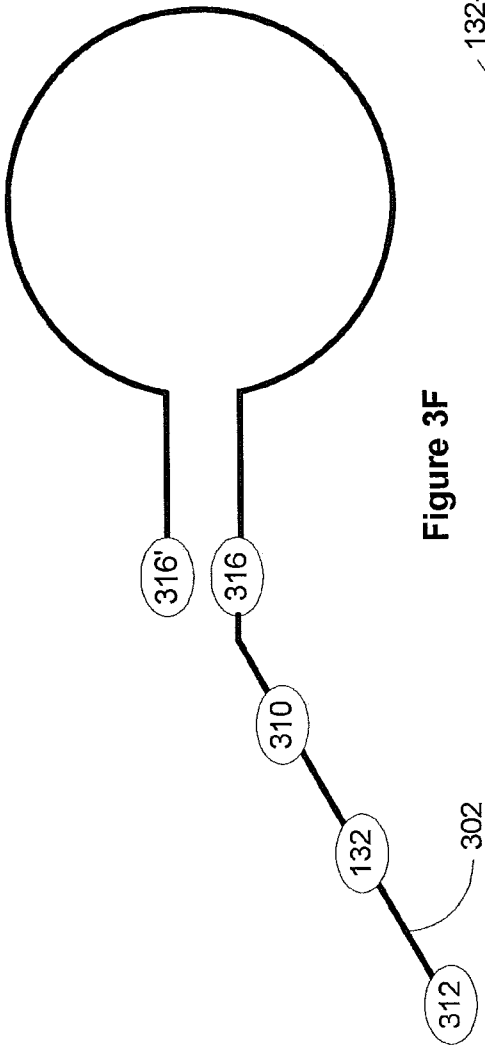


Figure 3E



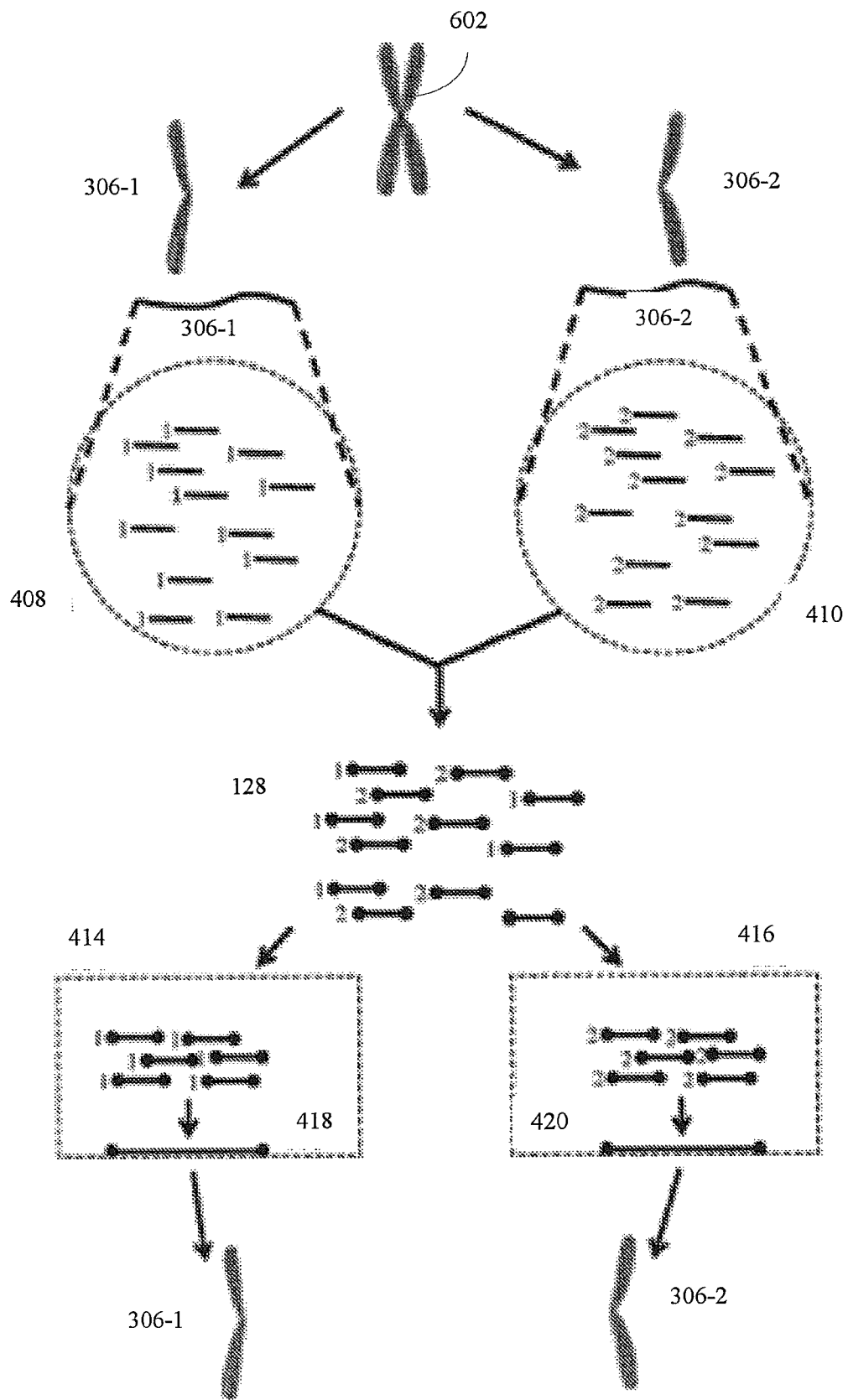


Figure 4

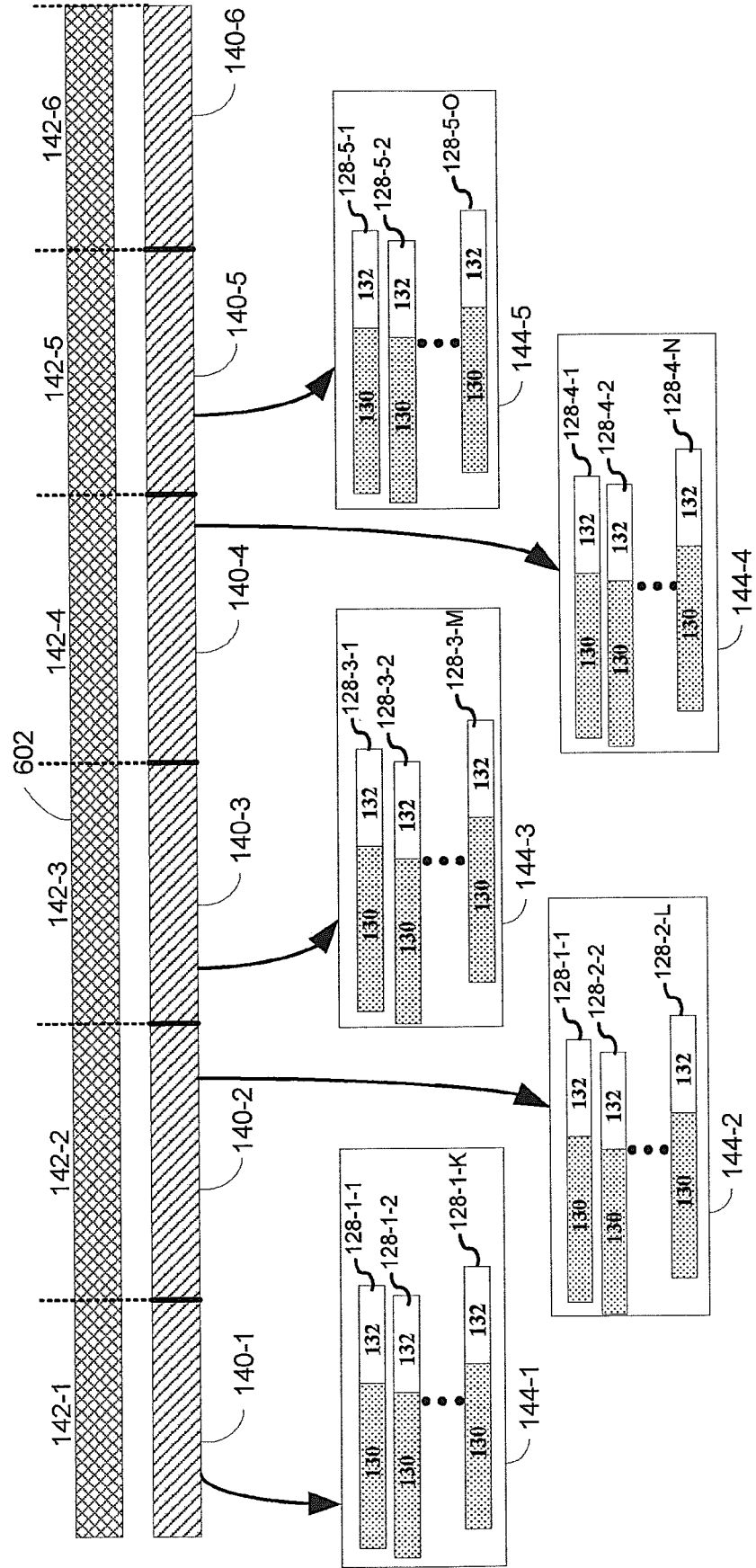


Figure 5

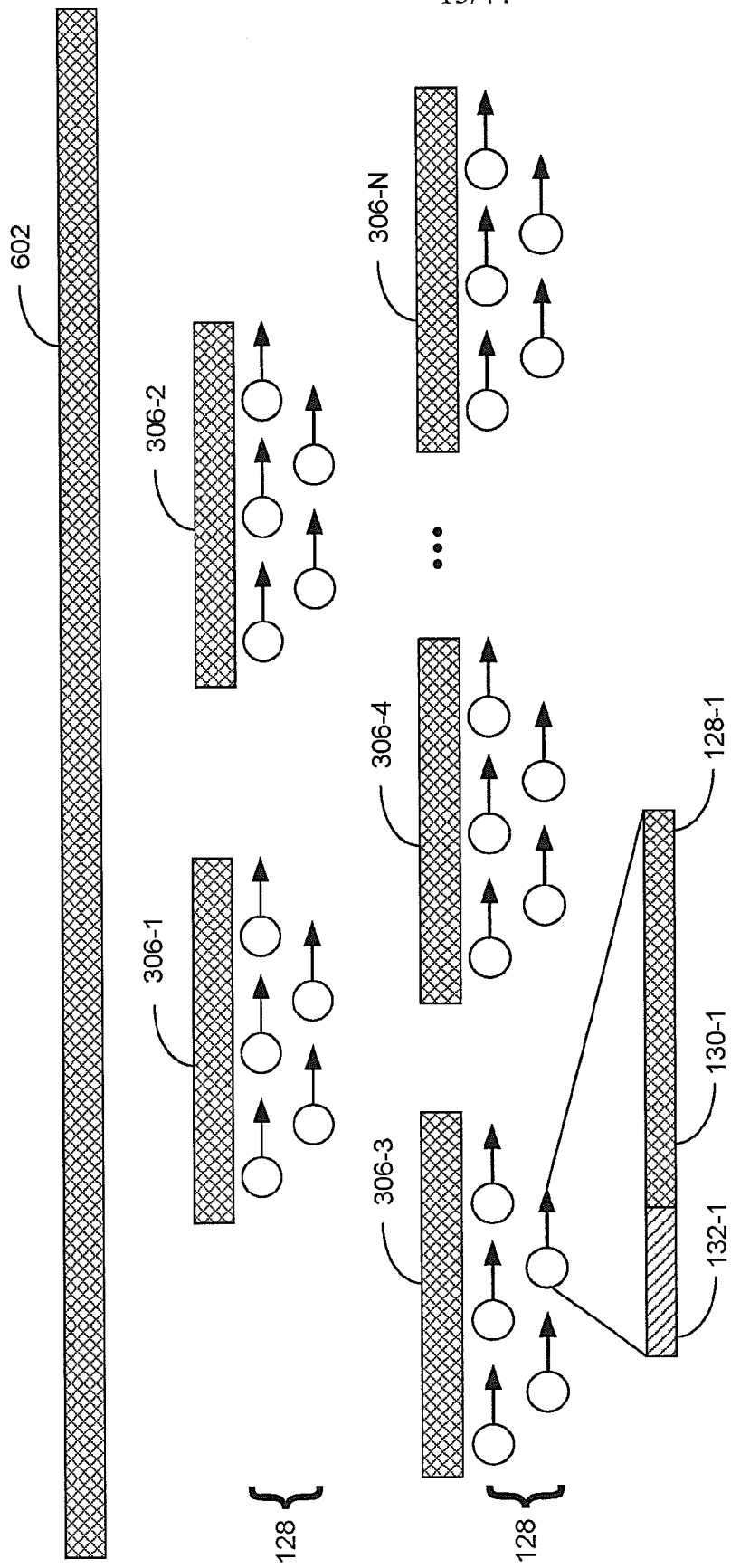


Figure 6

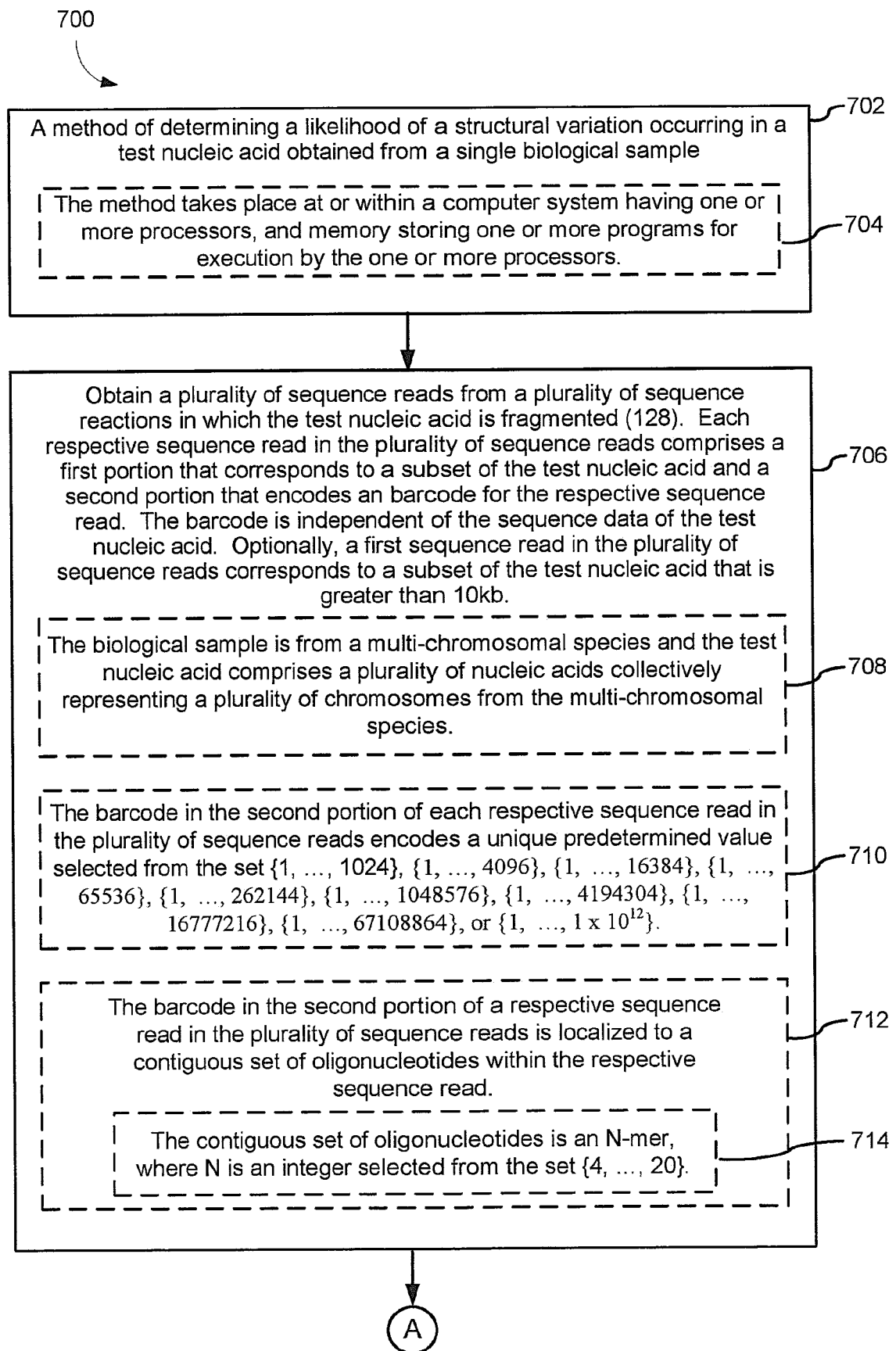


Figure 7A

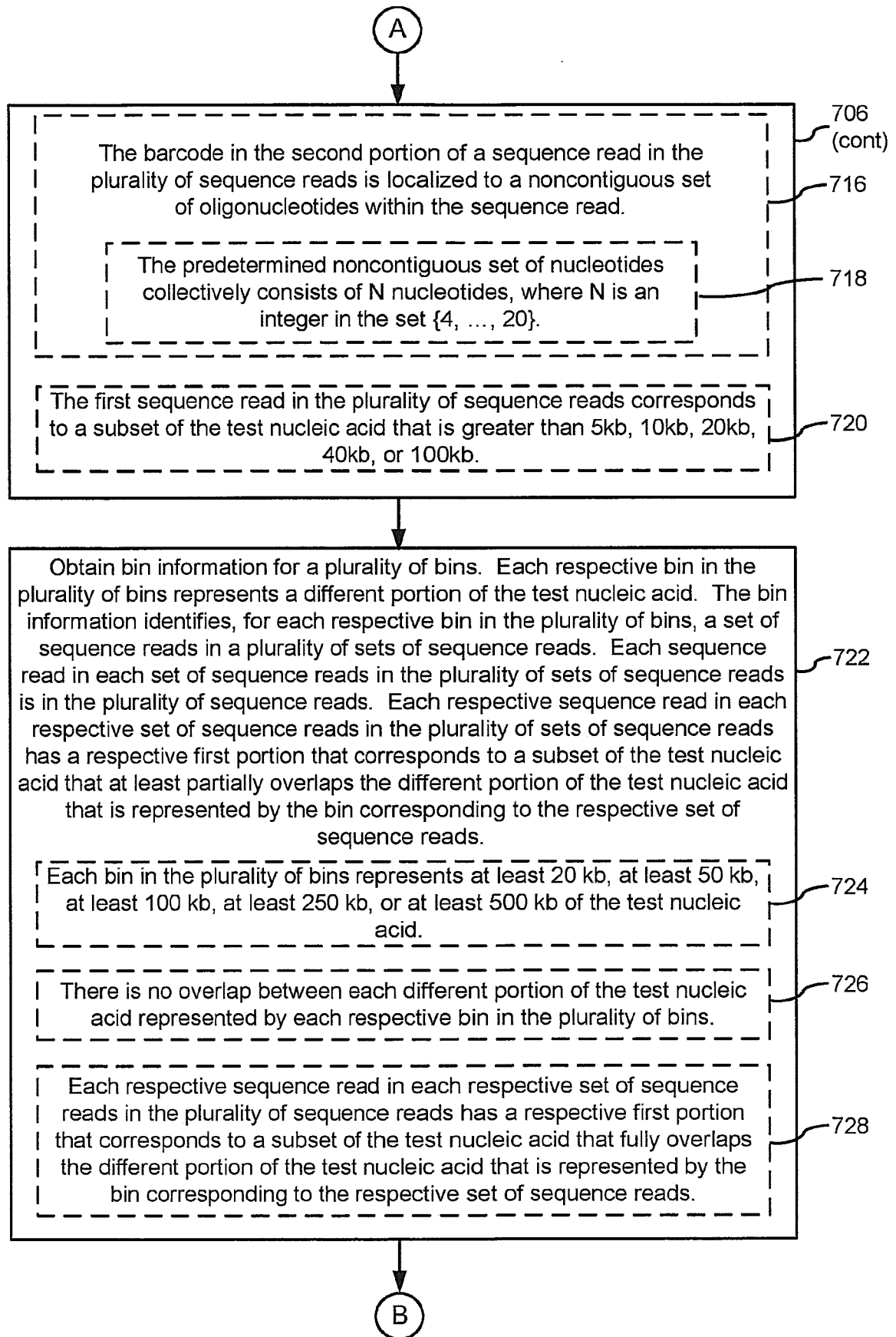


Figure 7B

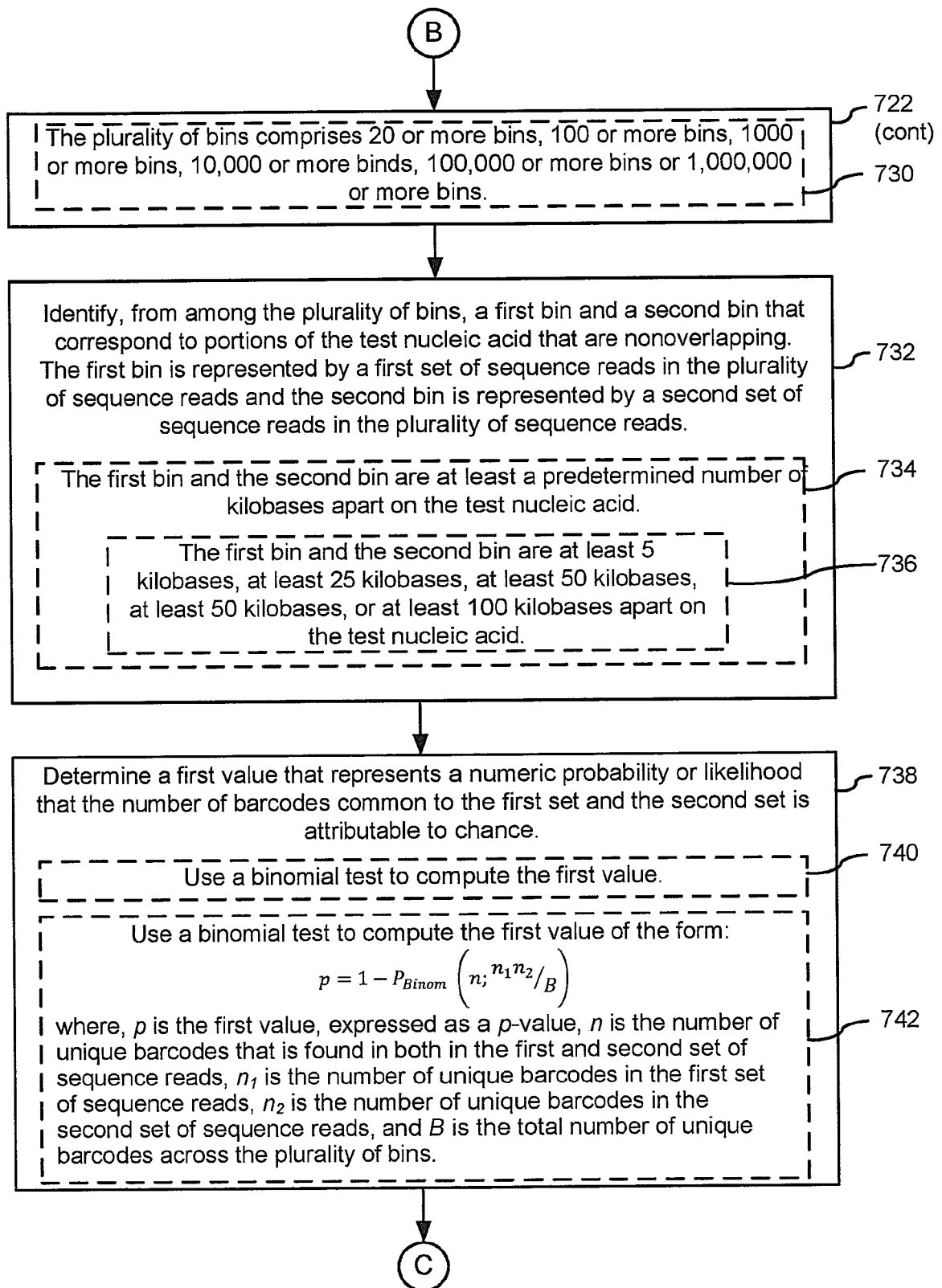


Figure 7C

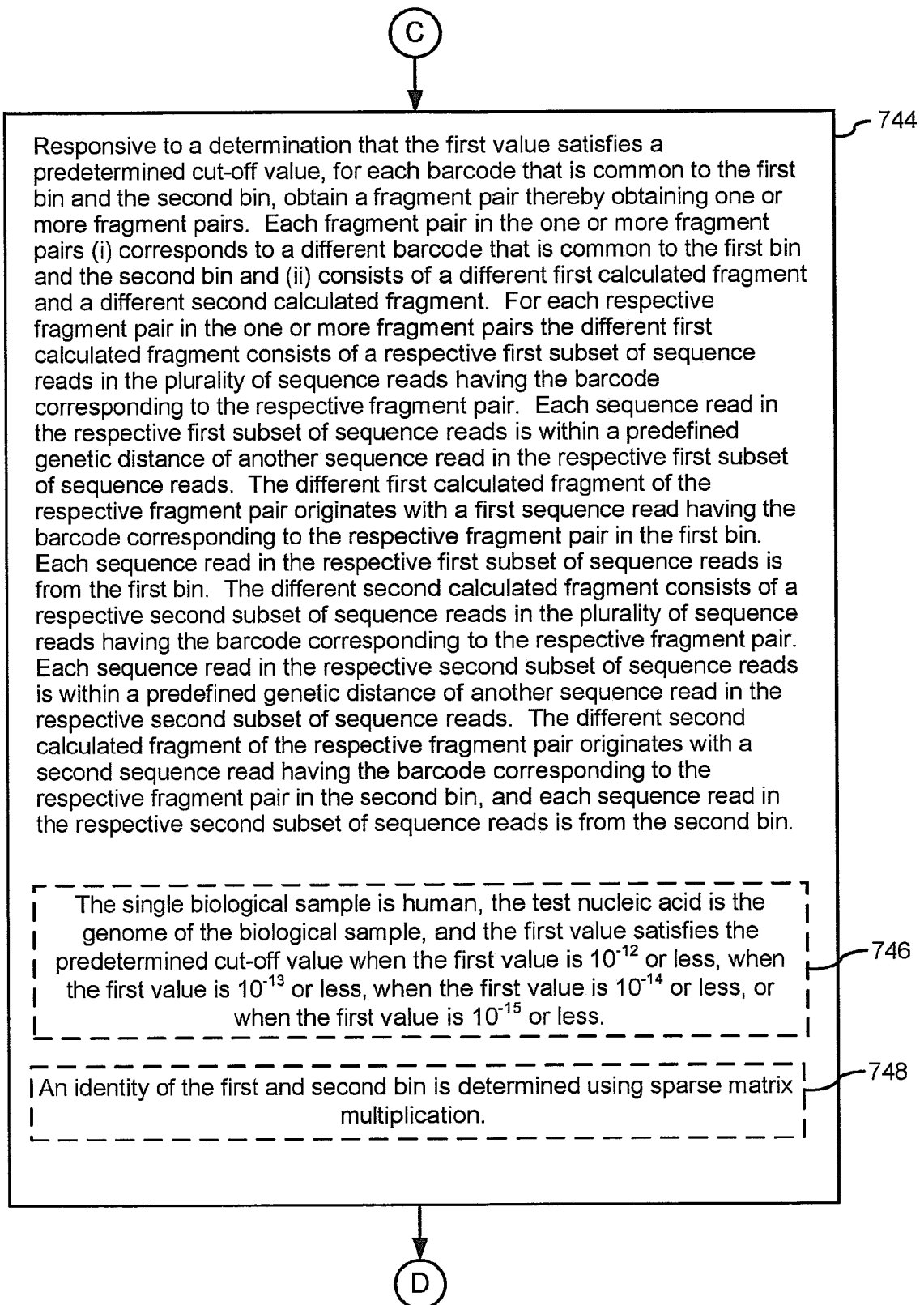


Figure 7D

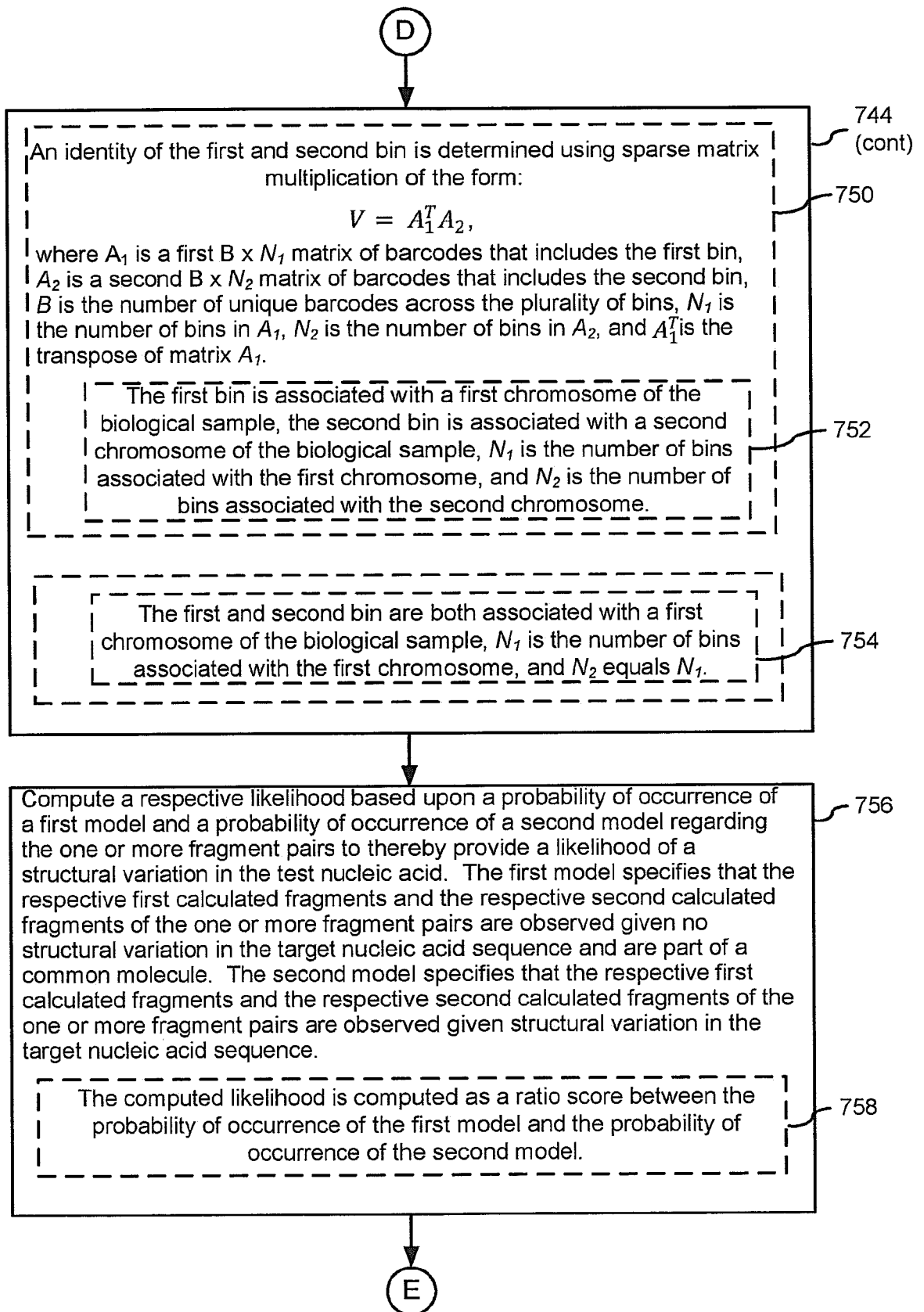


Figure 7E

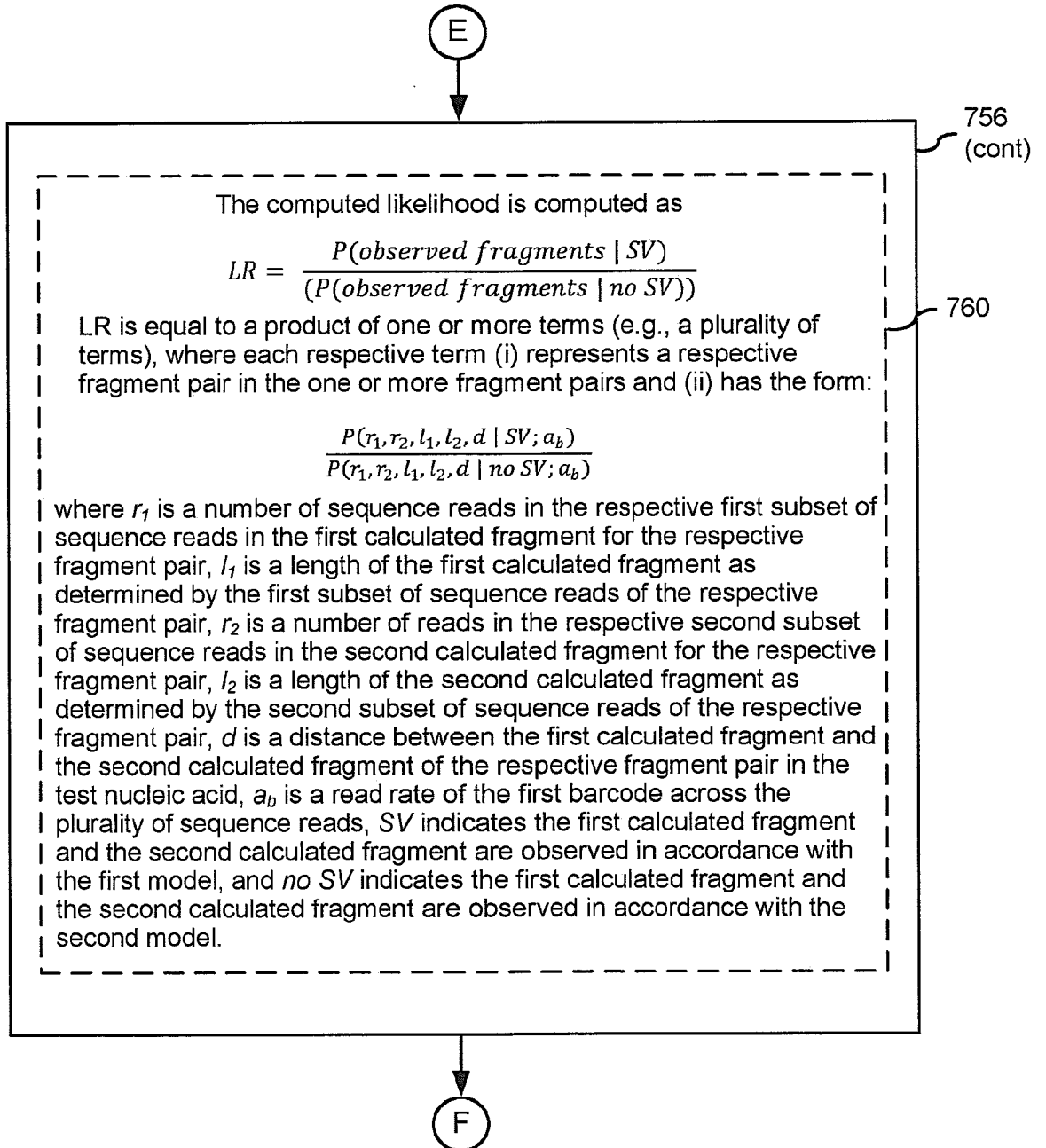


Figure 7F

(F)

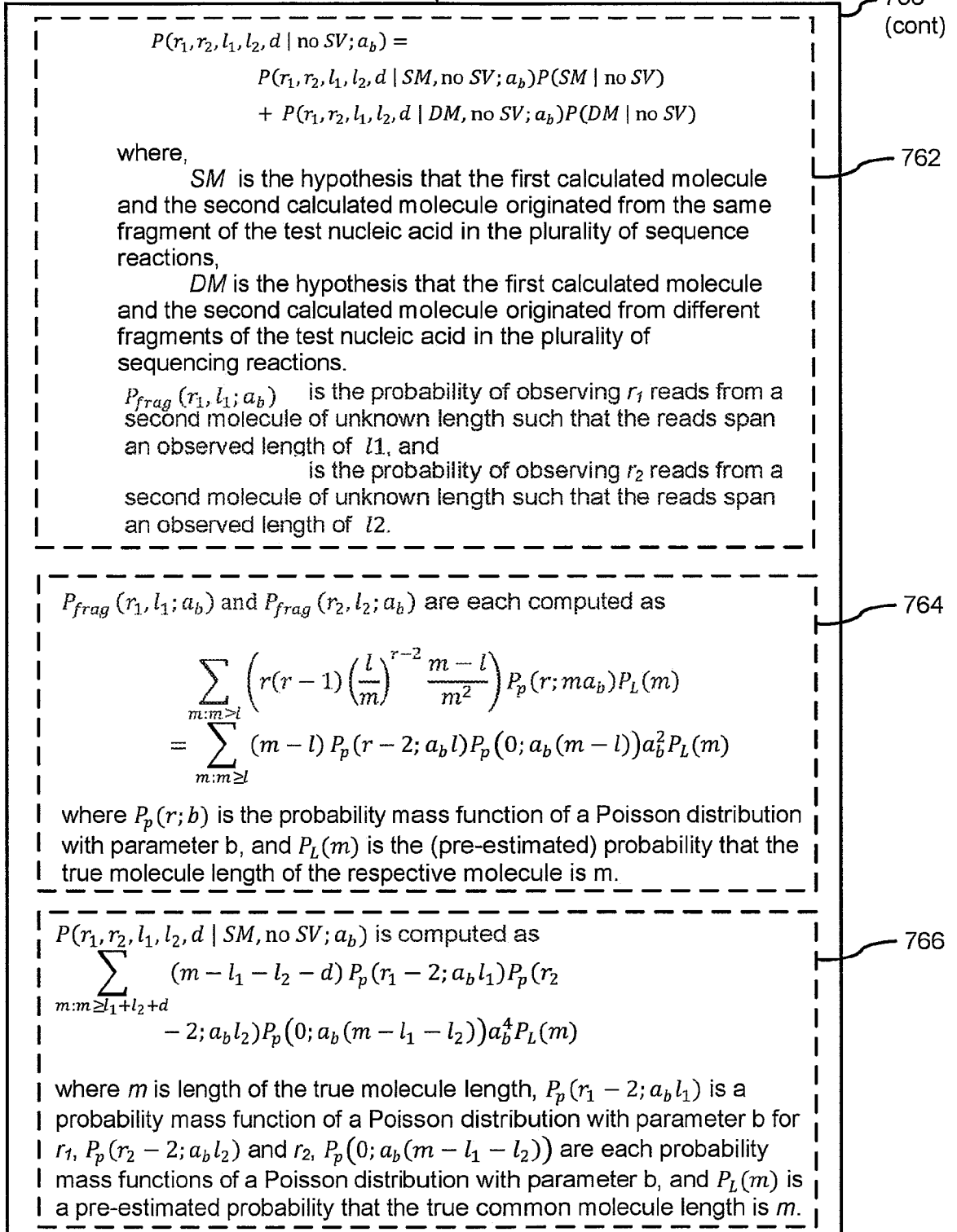


Figure 7G

G

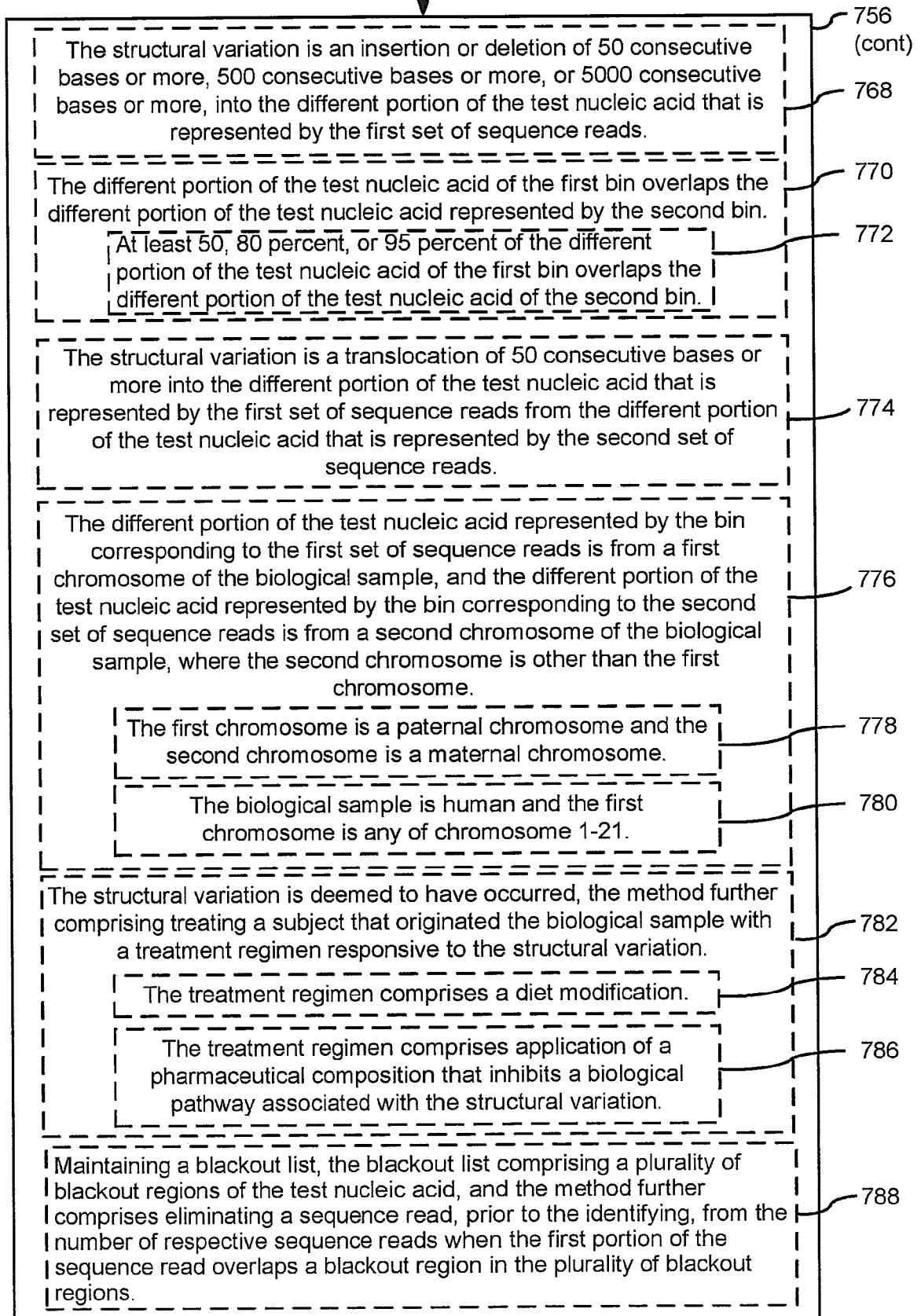


Figure 7H

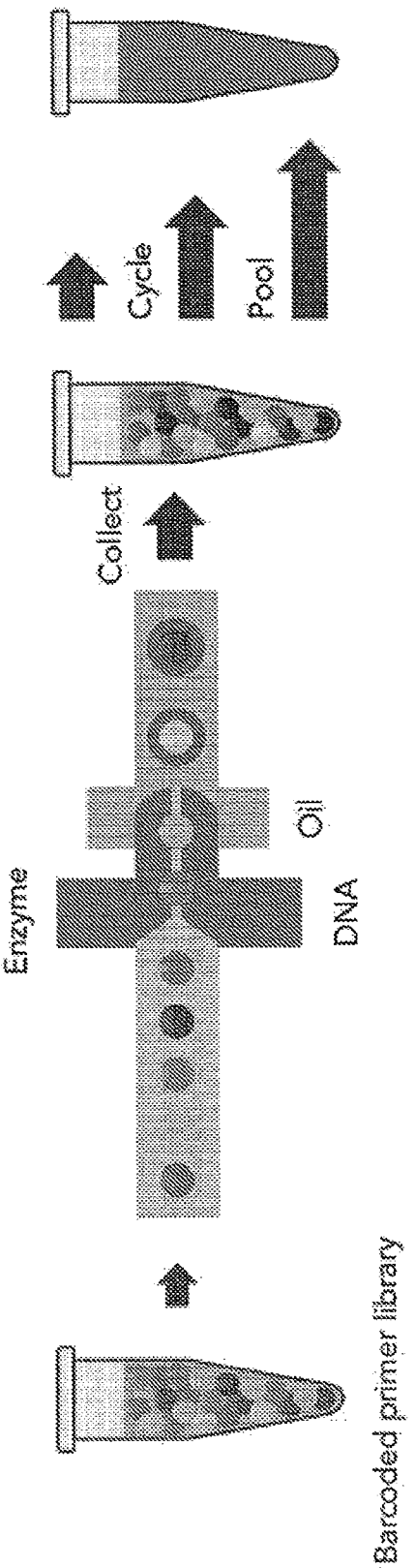


Figure 8

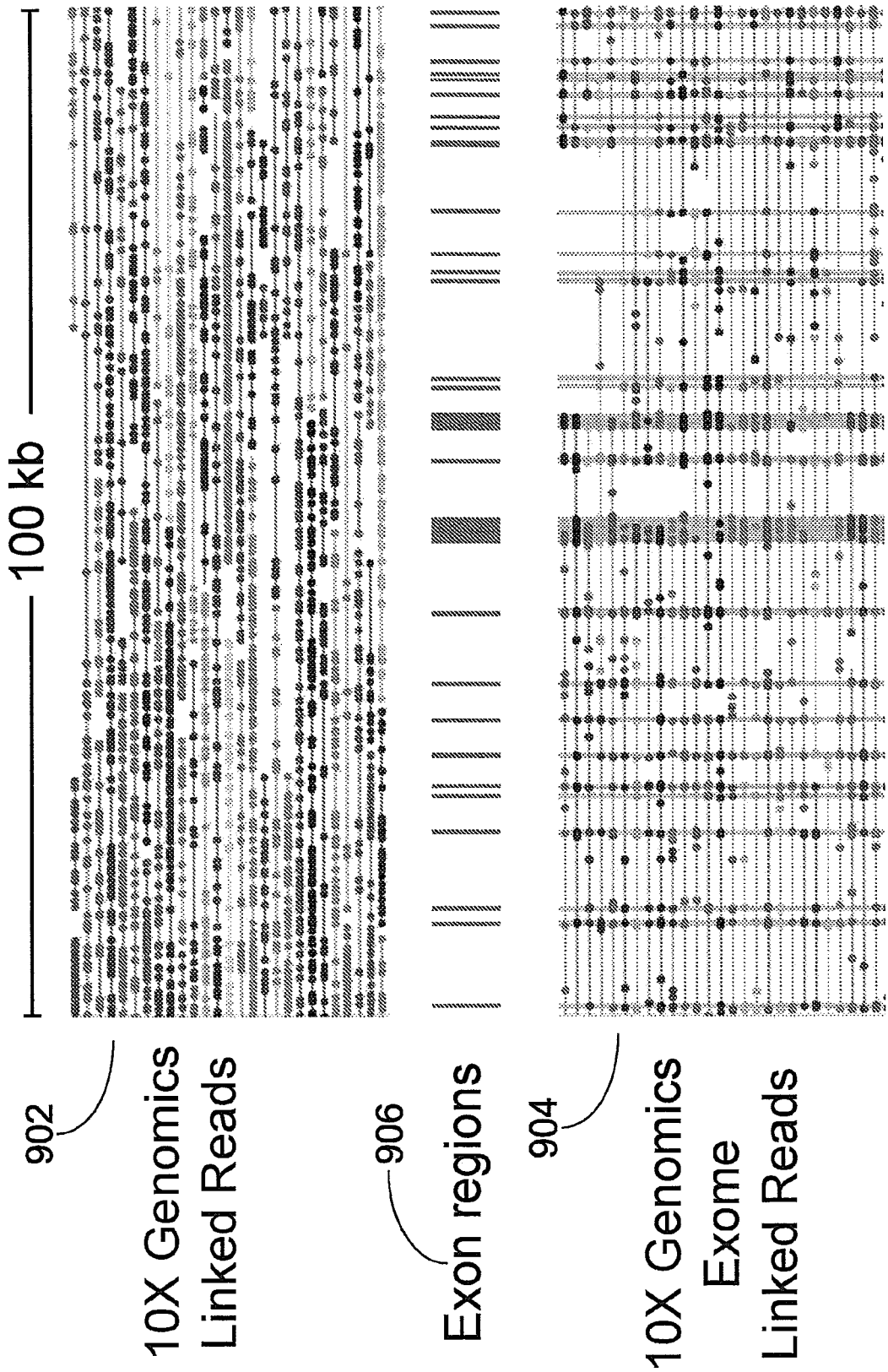
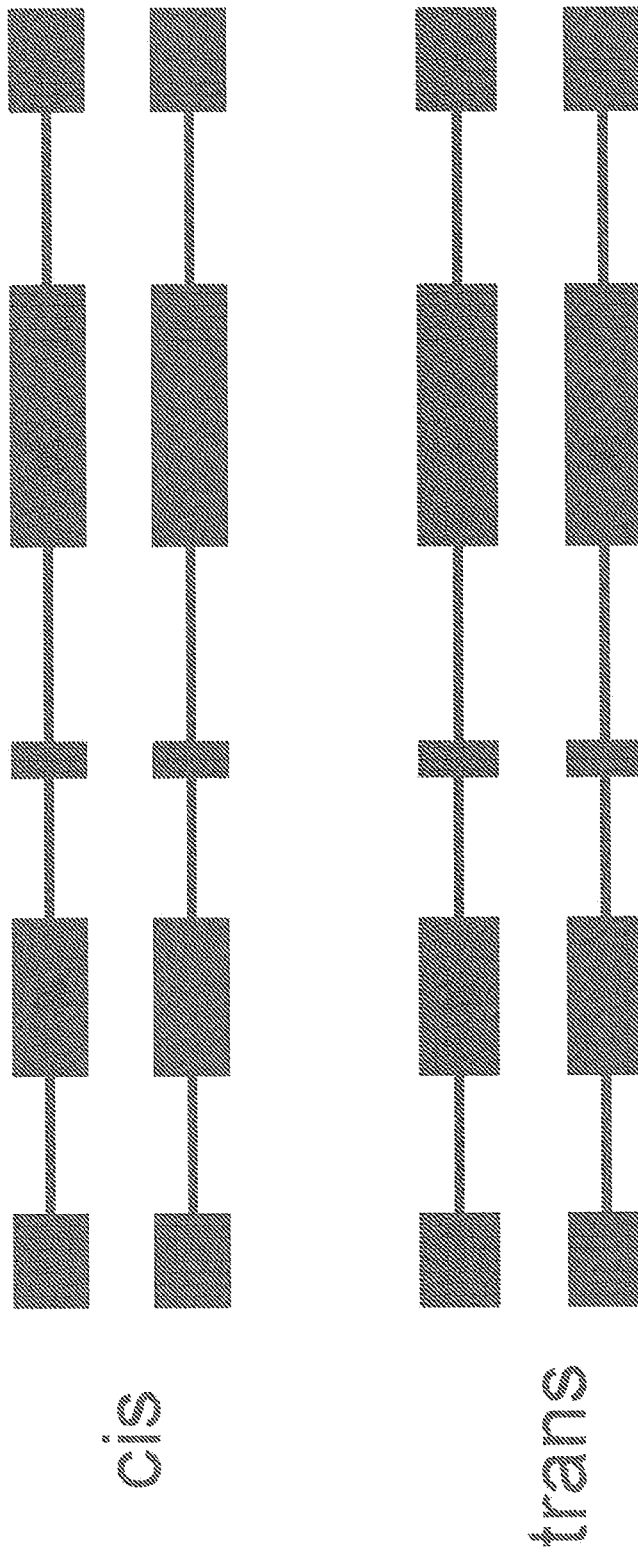


Figure 9

Metric	NA12878 WGS	NA12878 WES
Input Material	1 ng	1 ng
Mean Coverage	23x	139x
Mapped Read Fraction	97%	99%
PCR Duplication Rate	0.8%	4.7%
Barcode Diversity	>100k	>100k
N50 Linked Reads / Molecule	66	37
Mean Molecule Length (Length Weighted)	61.3 kb	64.7 kb

Figure 10



(Prior Art)

Figure 11

$$\log P(O_{i,f}|A_{i,p}) = \sum_r \mathbf{1}(S_r = A_{i,p})(1 - 10^{-Q_r/10}) + \mathbf{1}(S_r \neq A_{i,p})(10^{-Q_r/10})$$

$$P(O_{1,f},...O_{N,f}|\mathbf{X}) = \frac{(1-\alpha)}{2} (\prod_i P(O_{i,f}|A_{i,x_i}) + \prod_i P(O_{i,f}|A_{i,1...x_i})) + \alpha \prod_i^{0.5}$$

$$P(\mathbf{O}|\mathbf{X}) = \prod_f P(O_{1,f},...O_{N,f}|\mathbf{X})$$

Figure 12

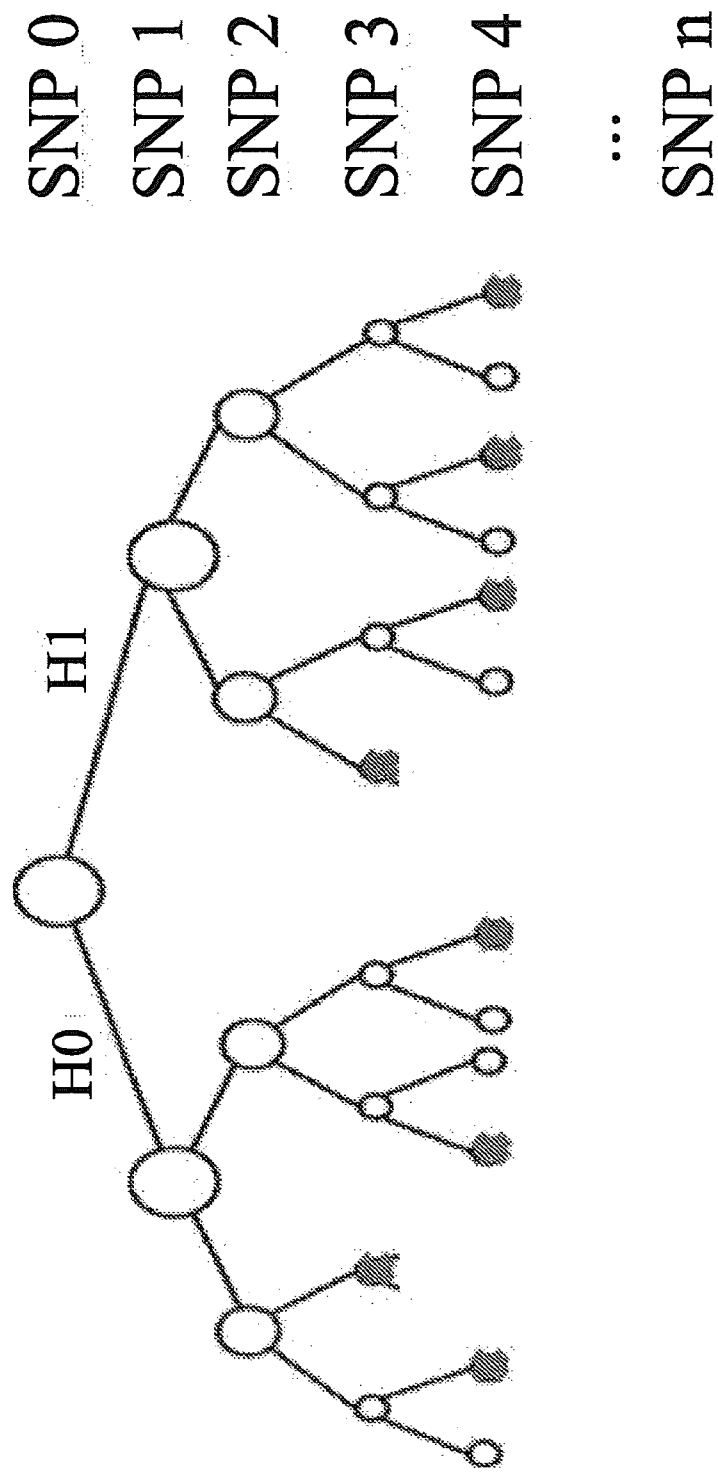


Figure 13

Metric	NA12878 WGS	HuRef 1 WGS	NA20847 WGS	NA12878 WES
Mean Coverage	23x	23x	23x	139x
N50 Phase Block	2.3MB	1.2MB	1.6MB	306KB
Short Switch	0.25%	0.4%	1.4%	0.24%
Long Switch	0.016%	0.017%	0.08%	0.03%
% SNPs Phased	97.2%	94.6%	97.4%	95.5%
% Genes Fully Phased (<100kb)	95.9%	90.0%	92.3%	96.1%

Figure 14

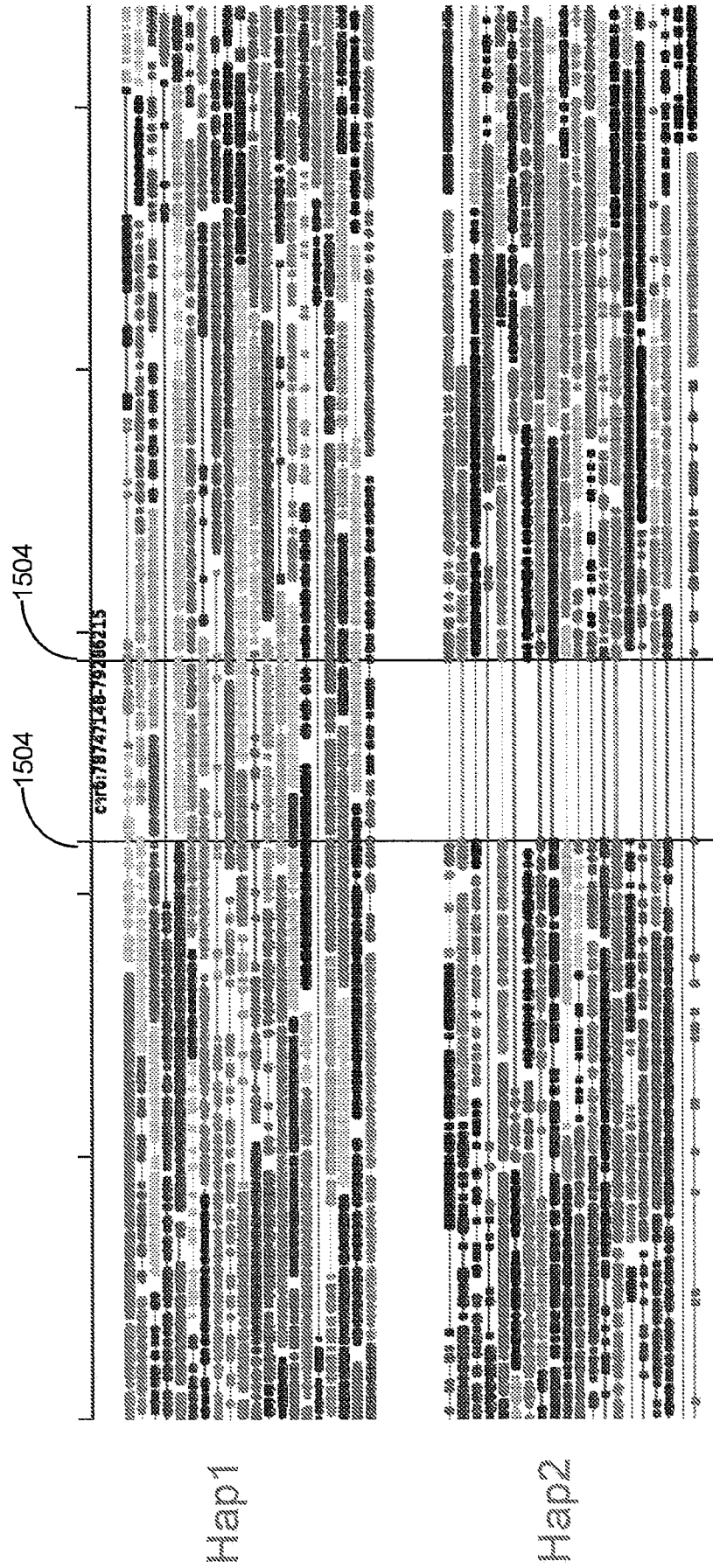
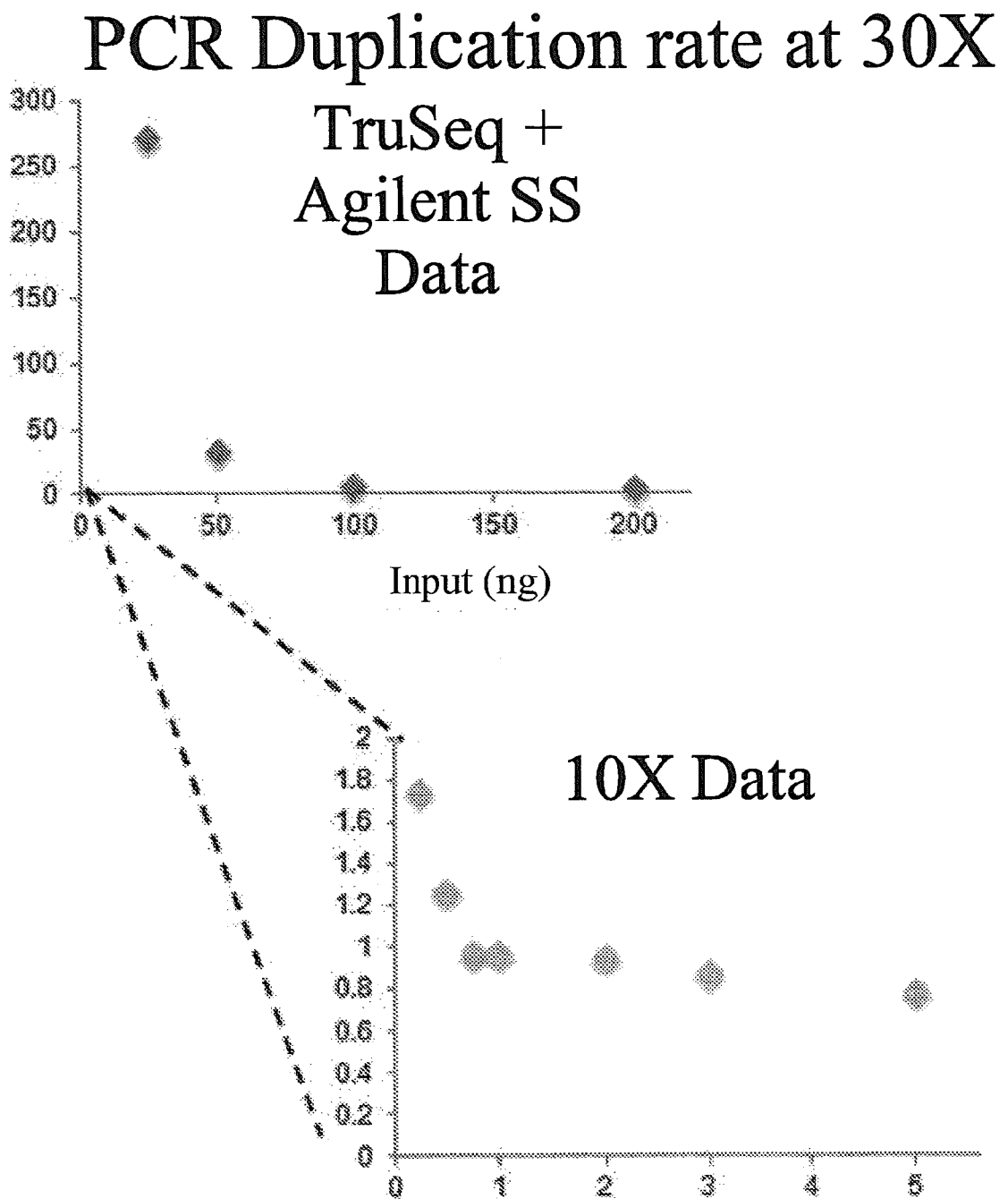


Figure 15

Chrom1	Start1	Chrom2	Start2	Phase Set	BCs Intersecting Hap1	BCs Intersecting Hap2	P	Deleted Haplotype	Deletion Inherited by NA12882	Phasing Consistent with Inheritance
chr1	189704495	chr1	189783155	50188502070	57	4	4.90E-13	1	No	Yes
chr3	162512117	chr3	162626153	190161712492	2	57	6.10E-15	2	Yes	Yes
chr5	104432082	chr5	104503476	27098341819	5	28	6.60E-05	2	Yes	Yes
chr6	78967148	chr6	79036215	31076466627	45	7	7.00E-08	1	No	Yes
chr6	78967148	chr6	79036215	31078967164	50	2	6.10E-13	1	No	Yes
chr8	39232044	chr8	39386994	39030957010	45	0	5.70E-14	1	Yes	Yes
chr8	39232044	chr8	39386994	39039370800	46	0	2.80E-14	1	Yes	Yes
chr5	99400840	chr5	99714778	27098341819	97	87	0.51	FP	No	N/A
chr14	37631599	chr14	37771213	62033781205	2	8	0.11	FP	No	N/A
chr14	106932612	chr14	107174772	640106692870	10	13	0.68	FP	No	N/A

Figure 16



Sequencing Metrics

Figure 17

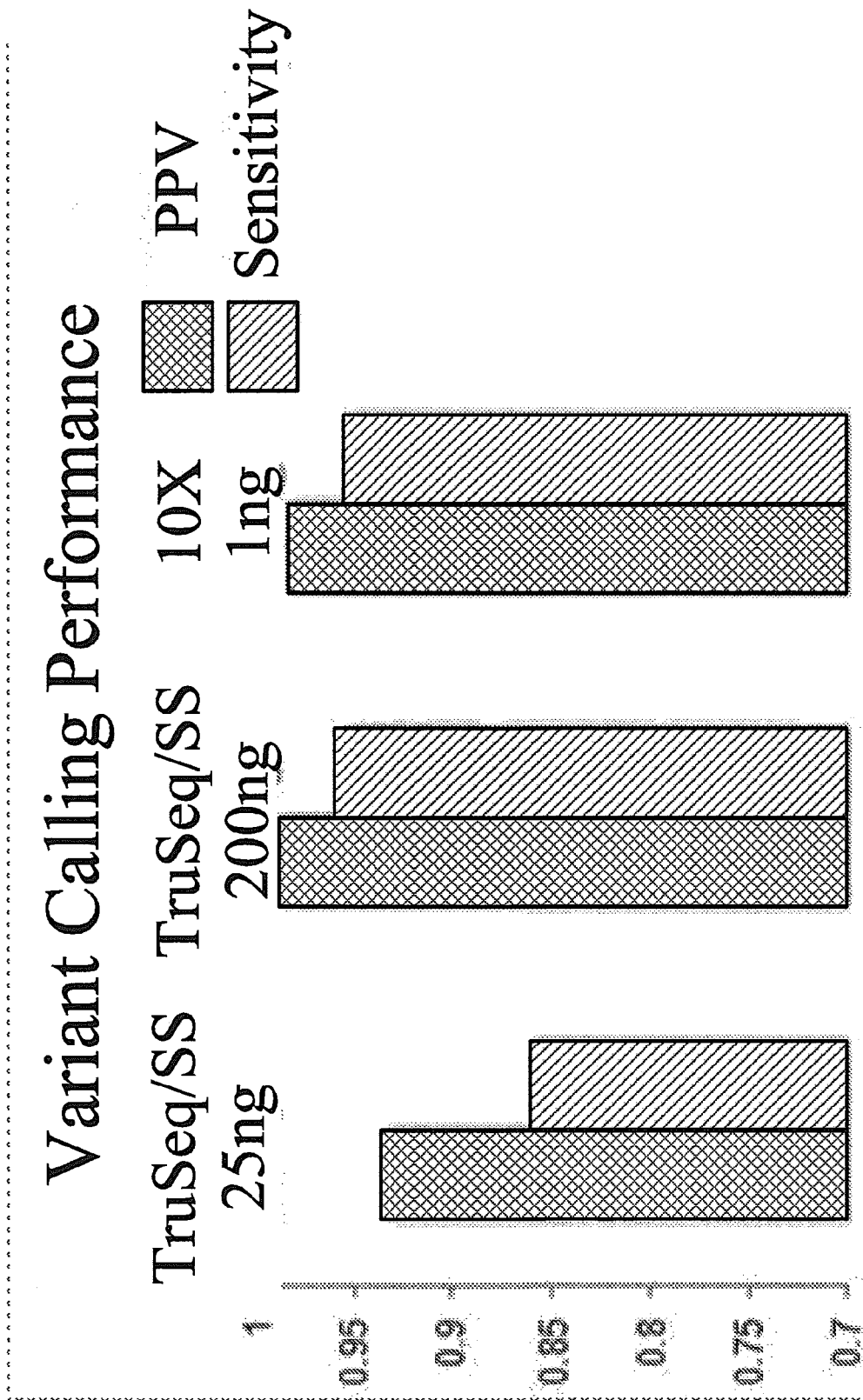


Figure 18

Sample Description	Sample name	Input mass (ng)	Mean Dup Rate	Mean Depth	% genes phased (<100kb)	Longest Phase Block (MB)
Acute Promyelocytic Leukemia	ACC207	1	1.09	178	91.4	1.2
Triple negative breast cancer	HCC38	1	1.13	173	93.7	1.5
Matched normal	HCC38BL	1	1.08	81	90.7	0.8
Triple negative breast cancer	HCC1143	1	1.13	176	95.5	1.1
Matched normal	HCC1143BL	1	1.07	100	93.8	0.6
Colorectal adenocarcinoma	HTC-15	1	1.09	140	91.9	1.8
Non-small cell lung cancer	NCH2228	1	1.05	159	91.3	0.6

Figure 19

Sample Description	Sample name	Input mass (ng)	Mean Dup Rate	Mean Depth	Longest phase block (Mb)	Gene Fusion(s) Annotated	Detected
Acute promyelocytic leukemia	ACC207	1	1.07	132	1.2	PML-RARA	Yes
Triple negative breast cancer	HCC38	1	1.1	123	1.5	LDHC-SERGF	Yes
						MBOAT2-PRKCE	Yes
						CUTA-SLC22A1	Yes
						SLC26A6-PRKAR2A	Yes
						ABCD6-RRP15	No
Triple negative breast cancer	HCC1143	1	1.08	99	1.1	FAM65B-MIPEP	Yes
						GCNT2-C6ORF52	Yes
						MCF2L2-NRG2	No
Non-small cell lung cancer	NCI-H2228	1	1.05	159	0.6	EML4-ALK	Yes
						ALK-PTPN3	Yes

Figure 20

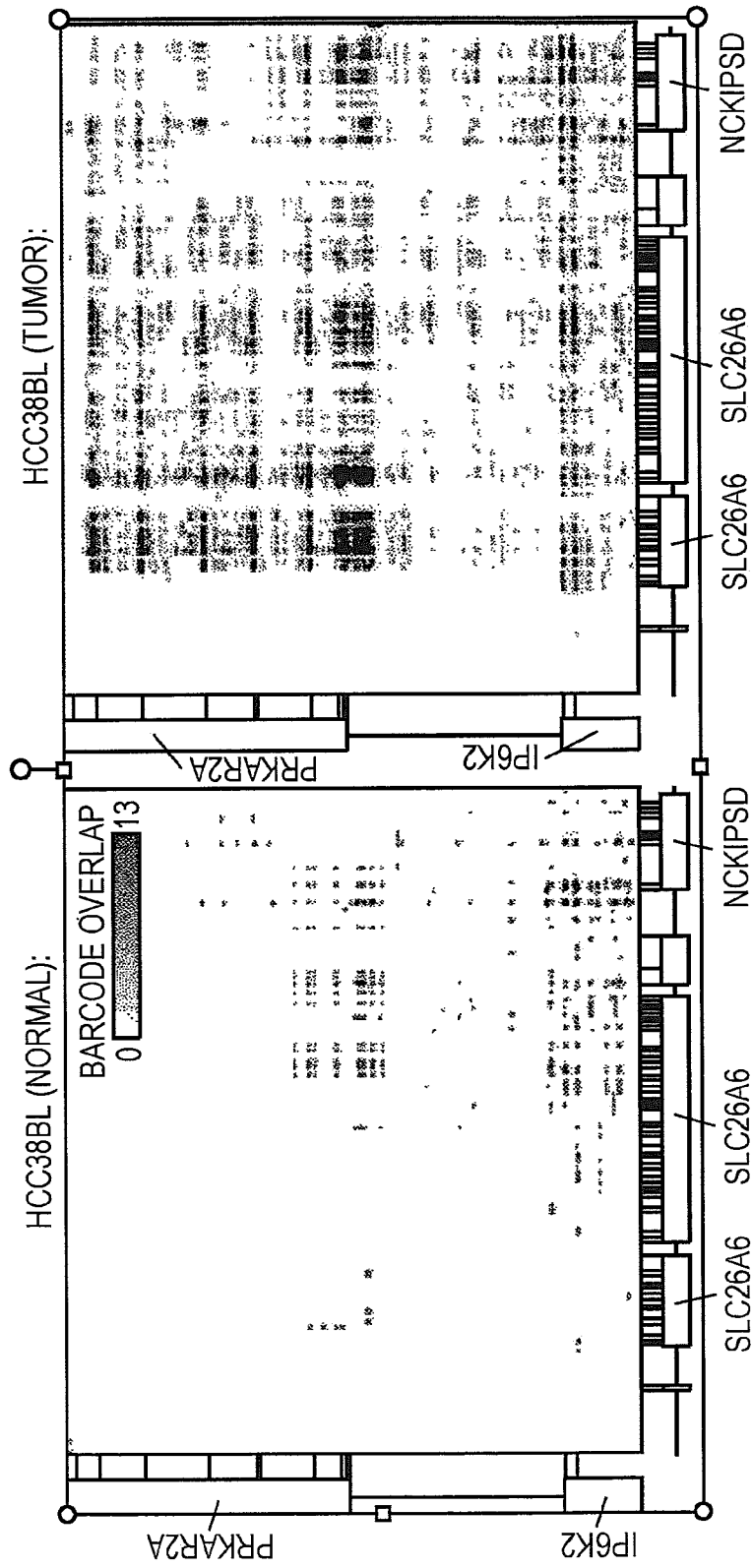


Figure 21

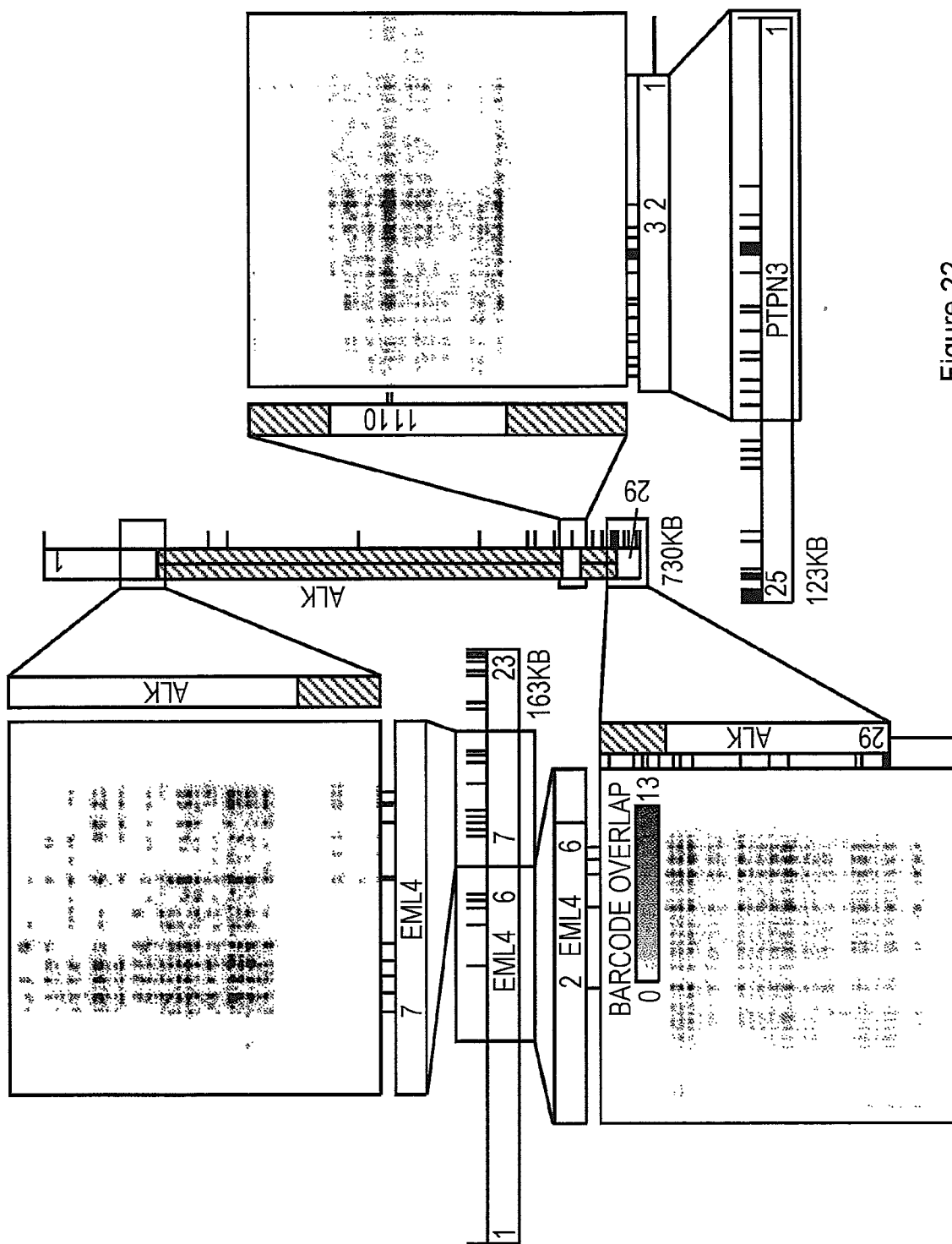


Figure 22

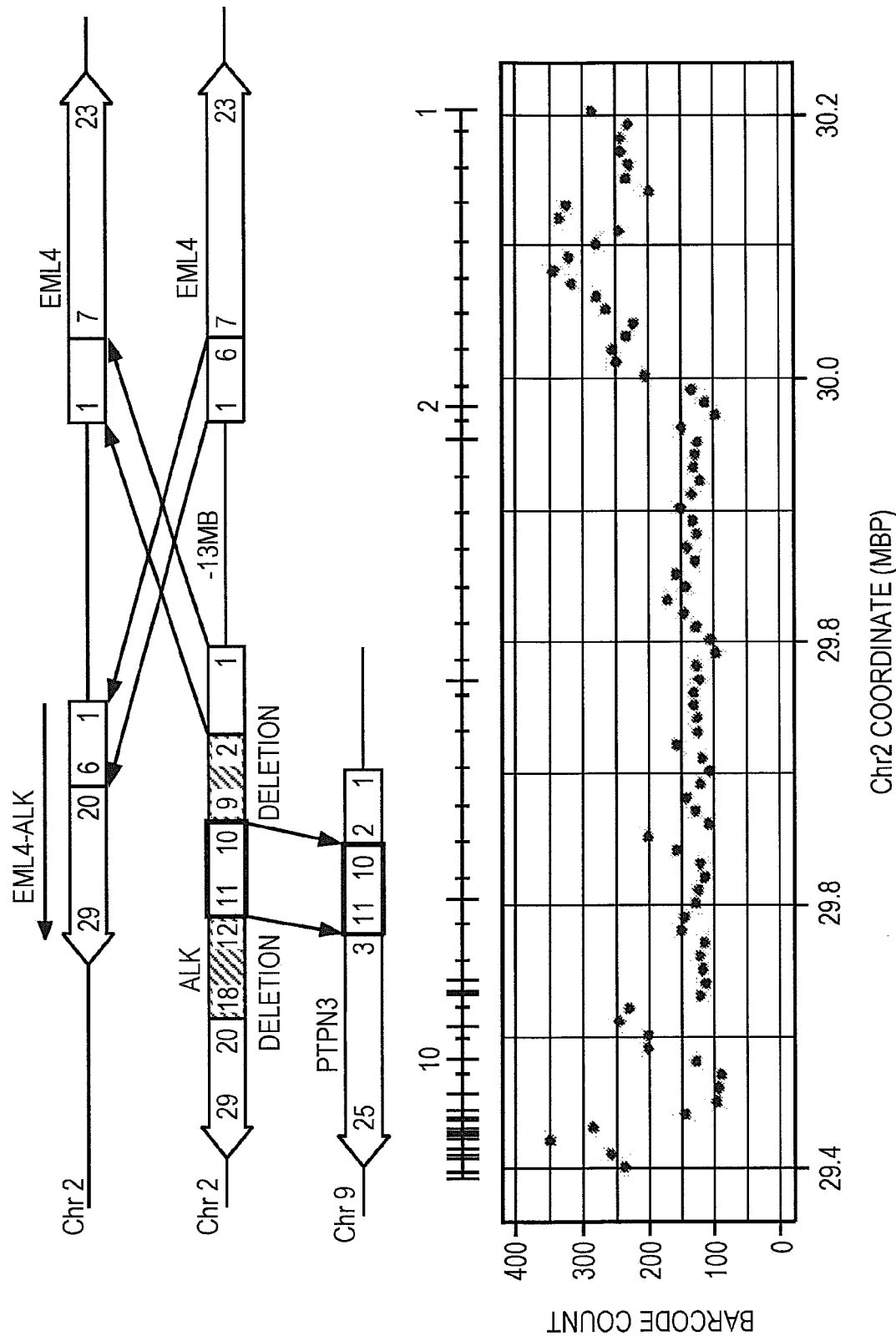


Figure 23

Fusion	Pos1	Pos2	Phase Block	# BC's in SV	# BC's, hap1	# BC's, hap2	<i>p</i>
EML4/ALK	Chr2: 29,447,360	Chr2: 42,499,30	Chr2:29,225,502- 29,446,202	25	0	23	2.40E-07
ALK/PTP N3	Chr2: 29,498,180	Chr9: 112,204,017	Chr9:112,184,972- 112,225,774	30	0	22	4.80E-07

Figure 24

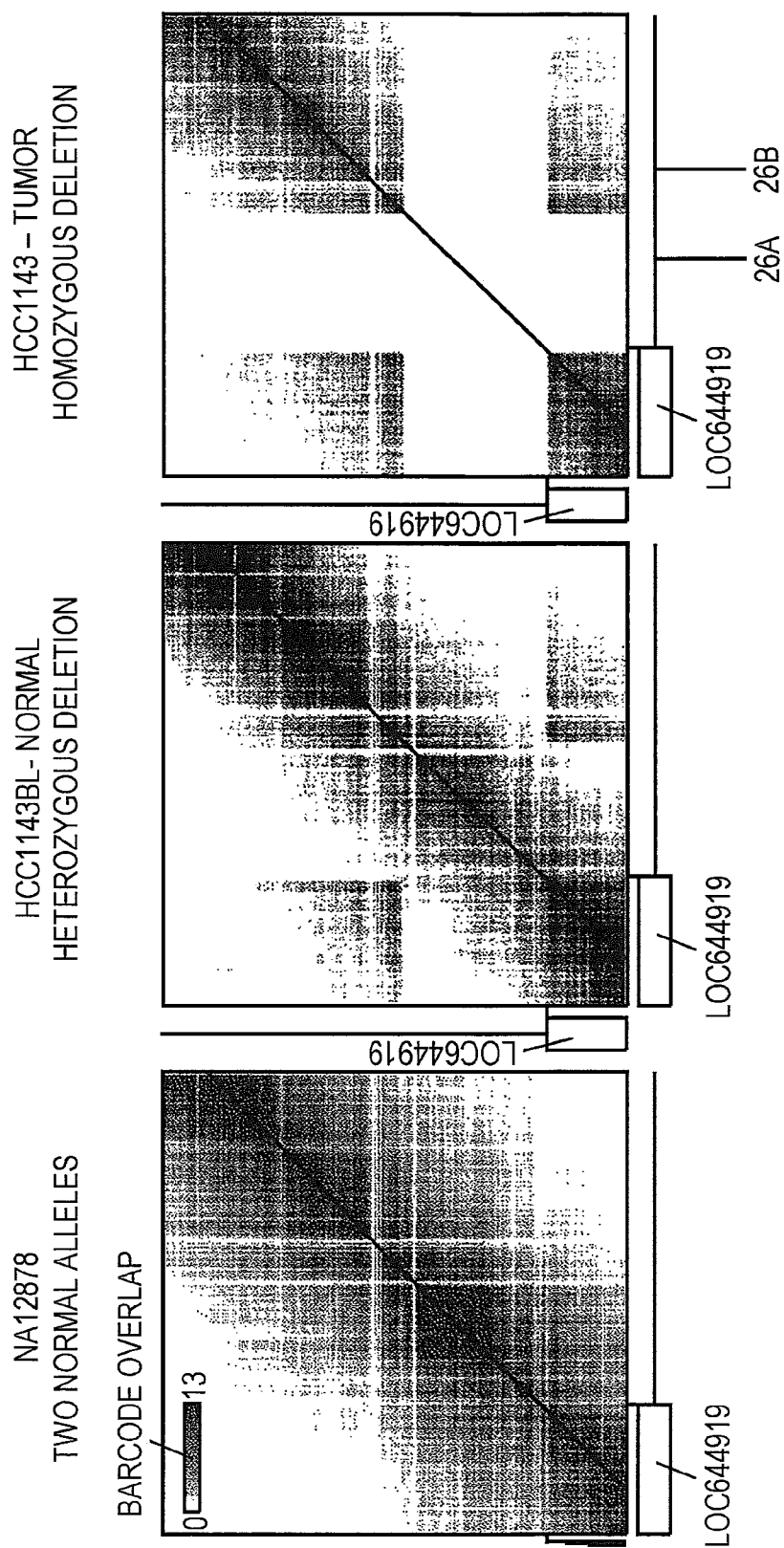


Figure 25

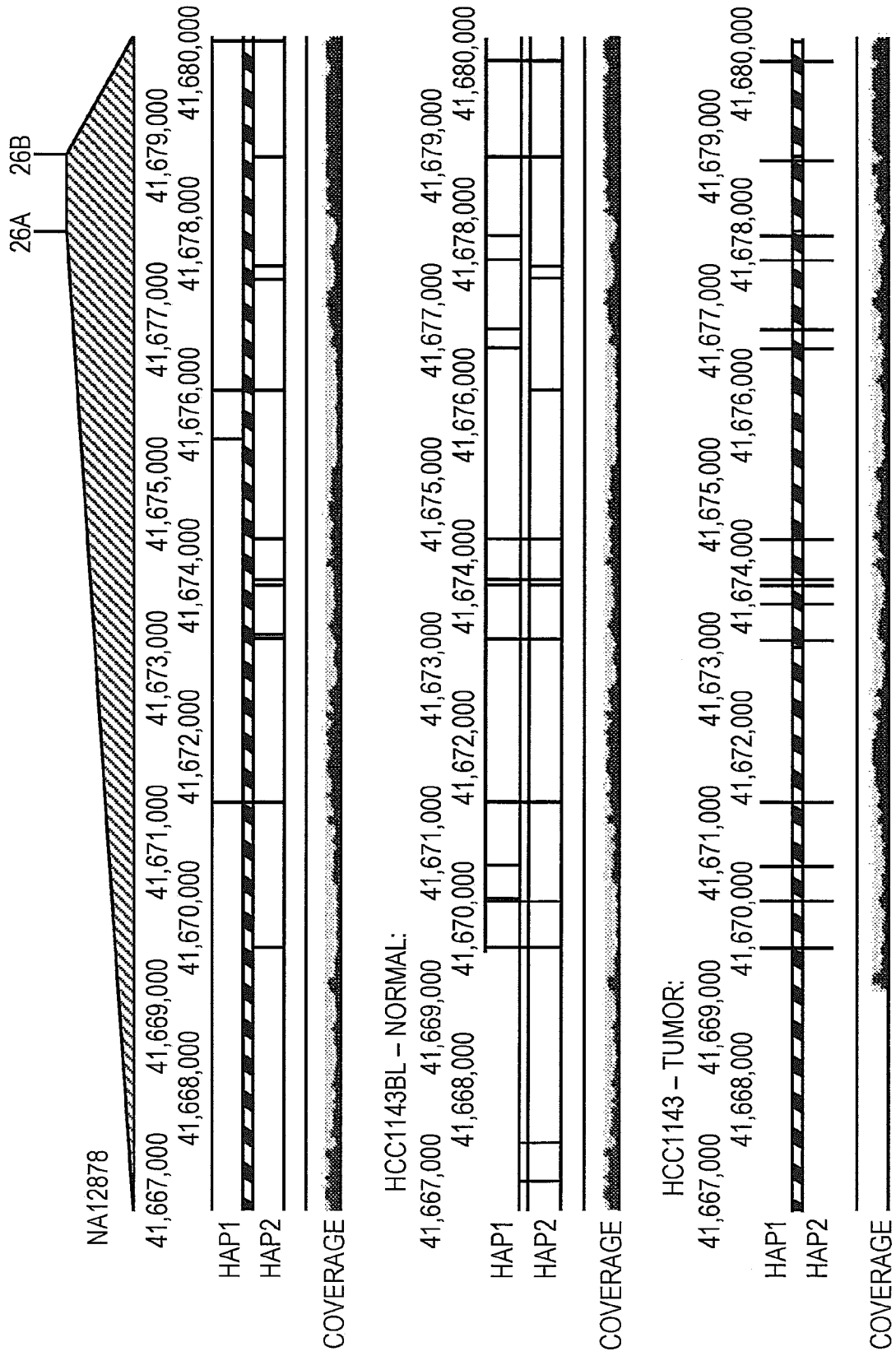


Figure 26

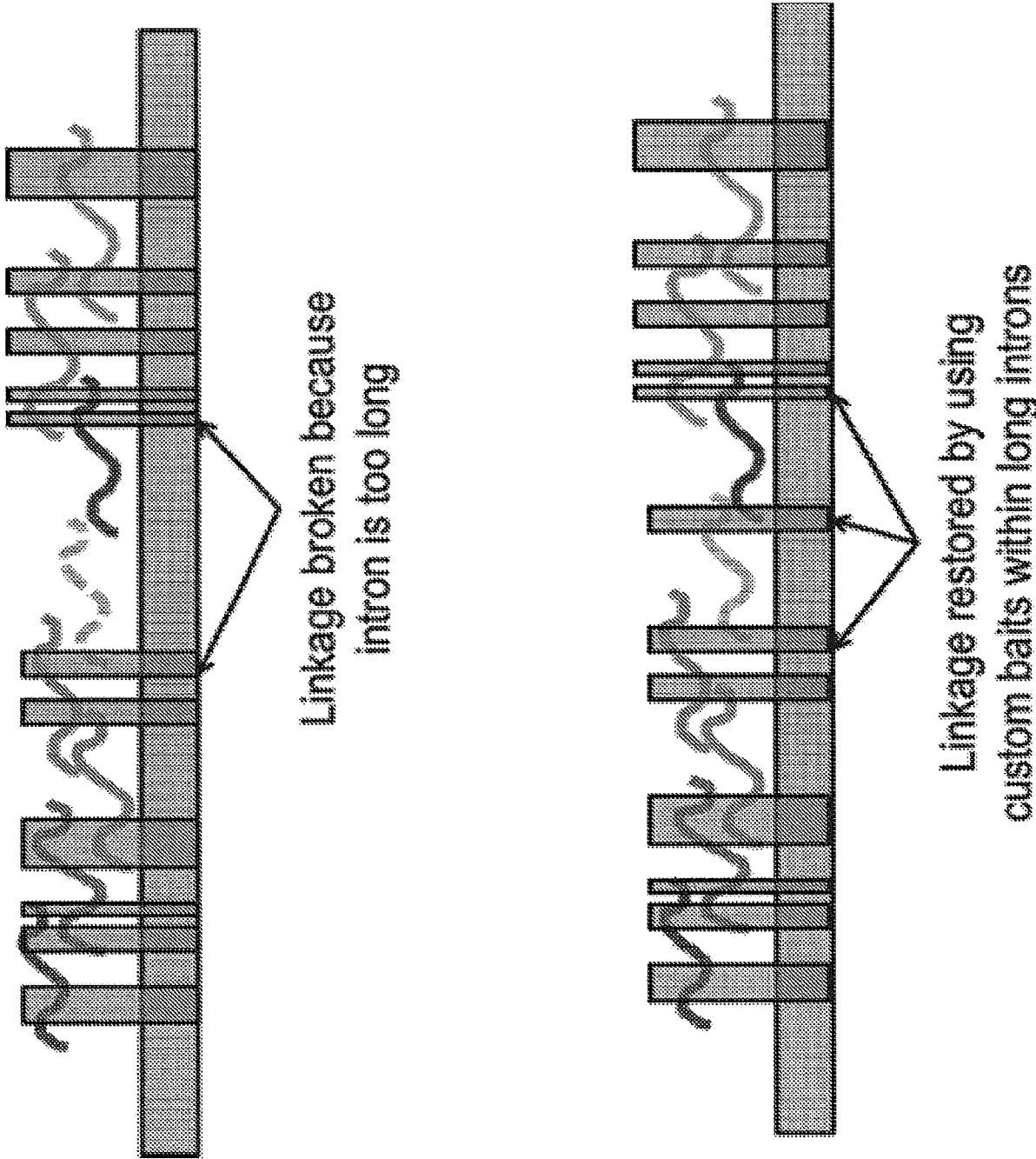


Figure 27

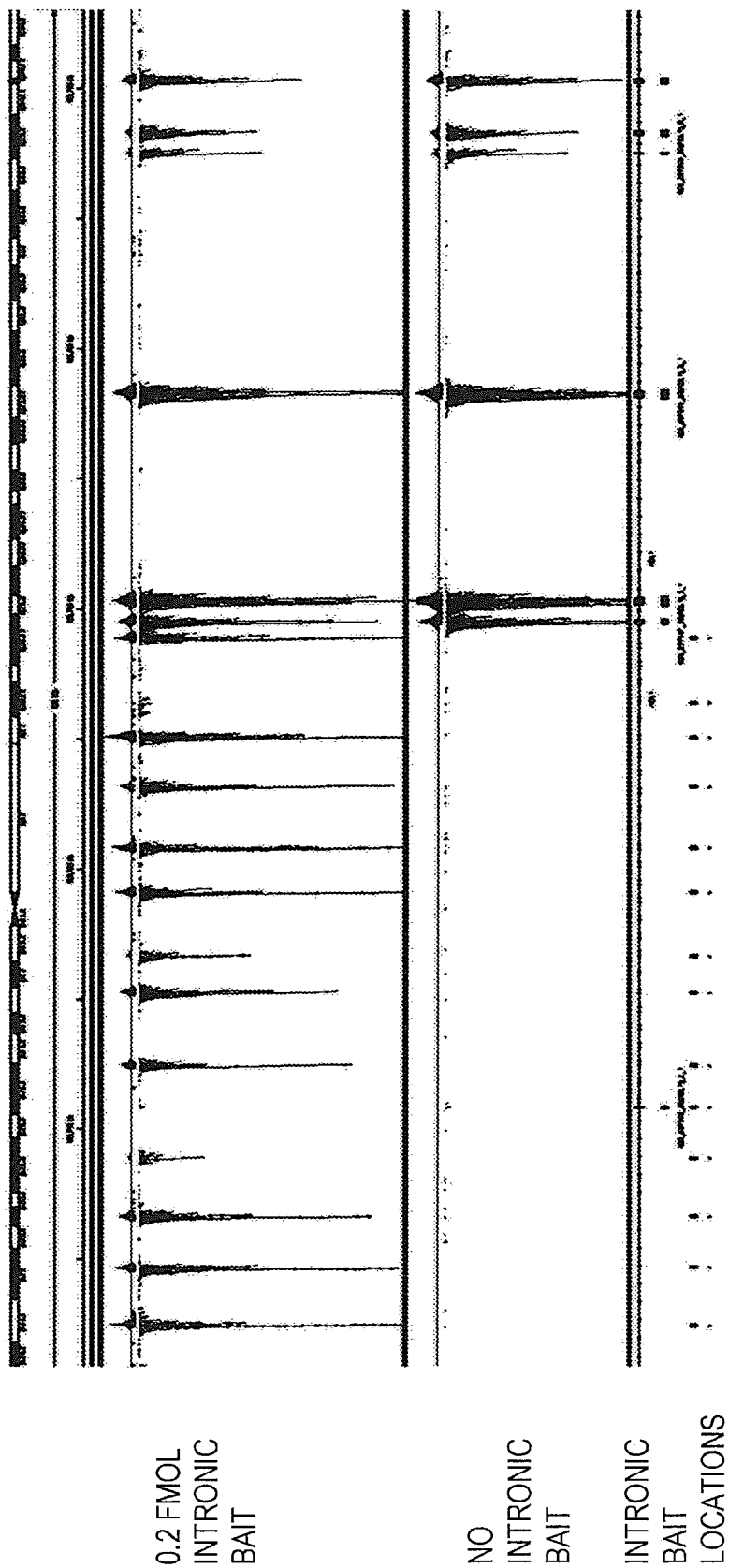


Figure 28

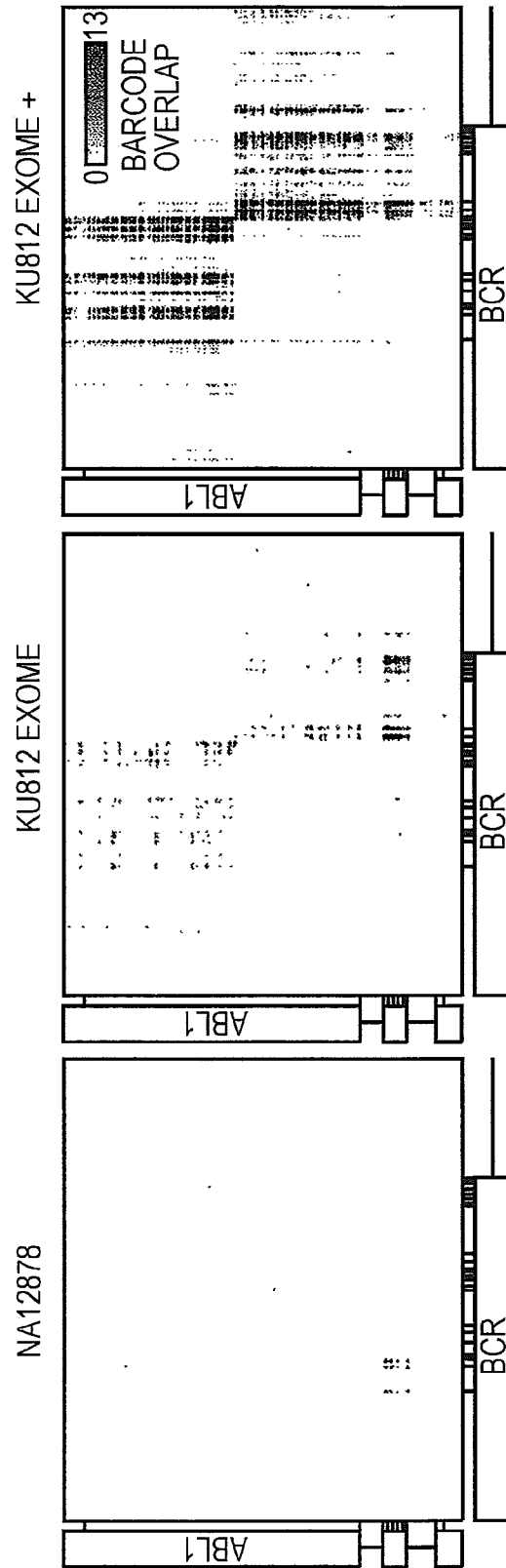


Figure 29

	Bases on Target	Fragments on Target	5' gene	3' gene	SV Quality Score
0.2 fmol intronic bait	0.55	0.94	BCR	ABL1	35
No intronic bait	0.56	0.95	BCR	ABL1	0

Figure 30