

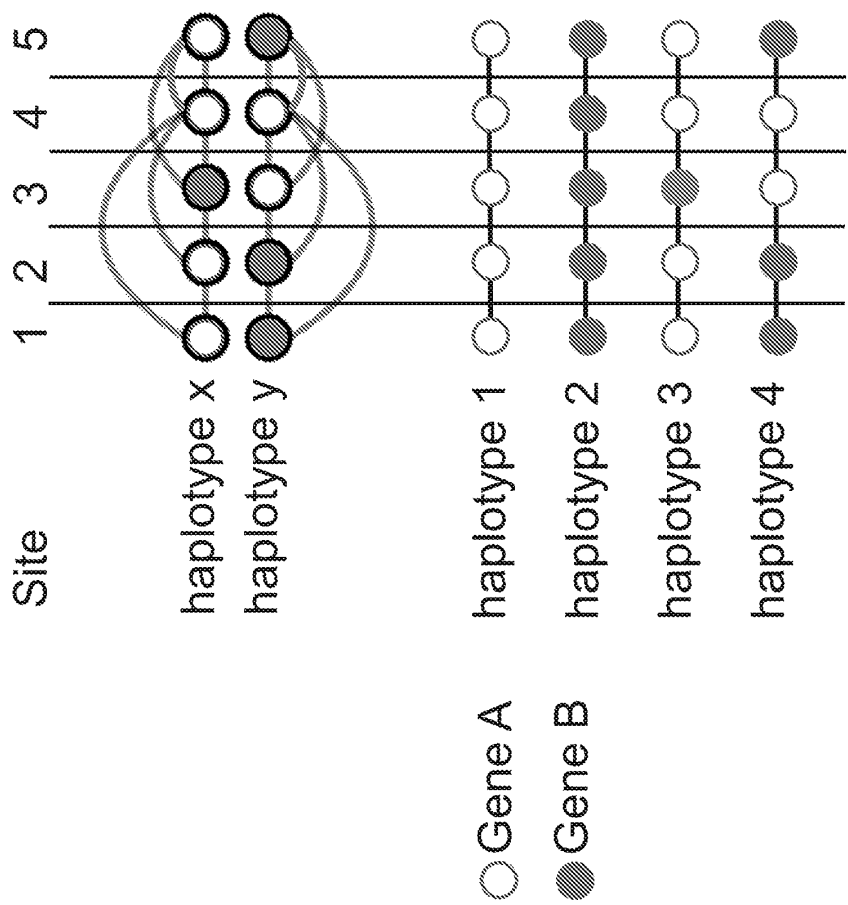


FIG. 1

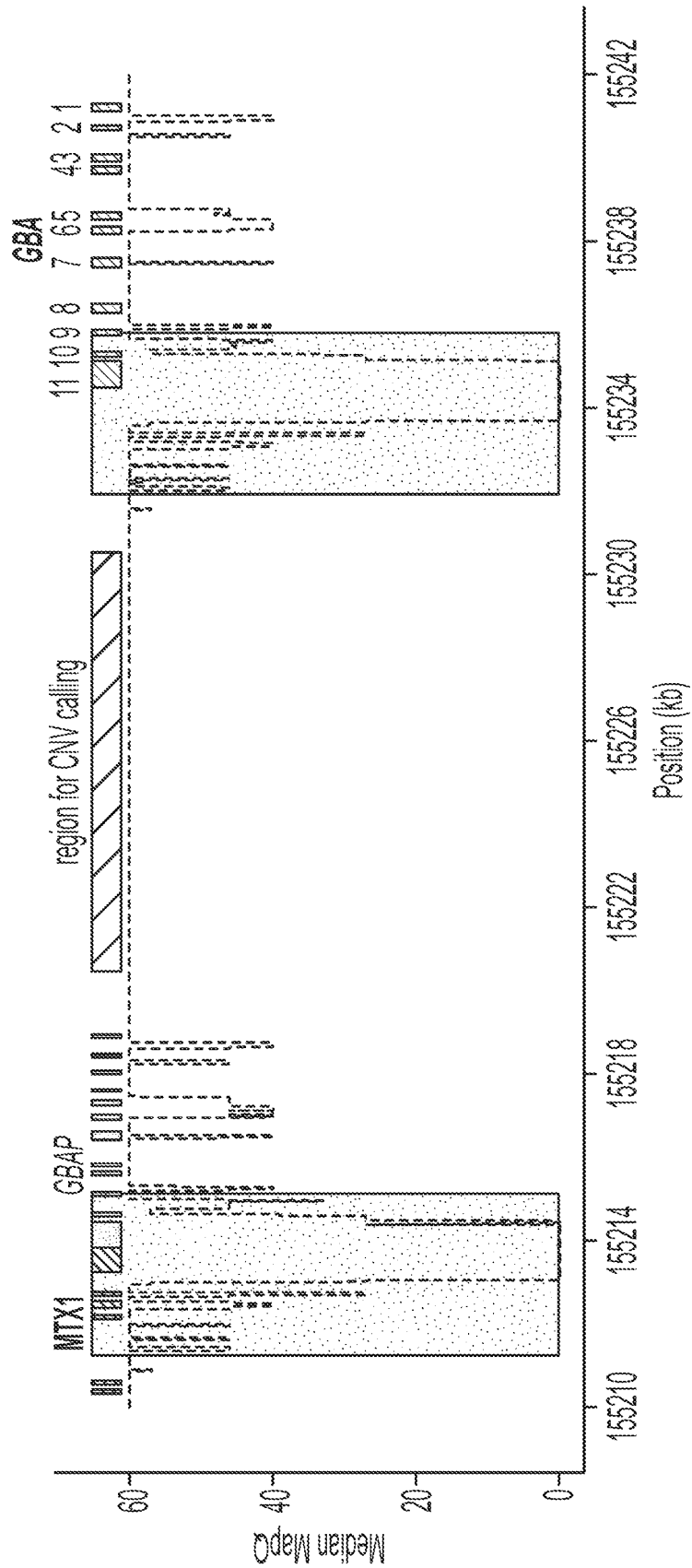
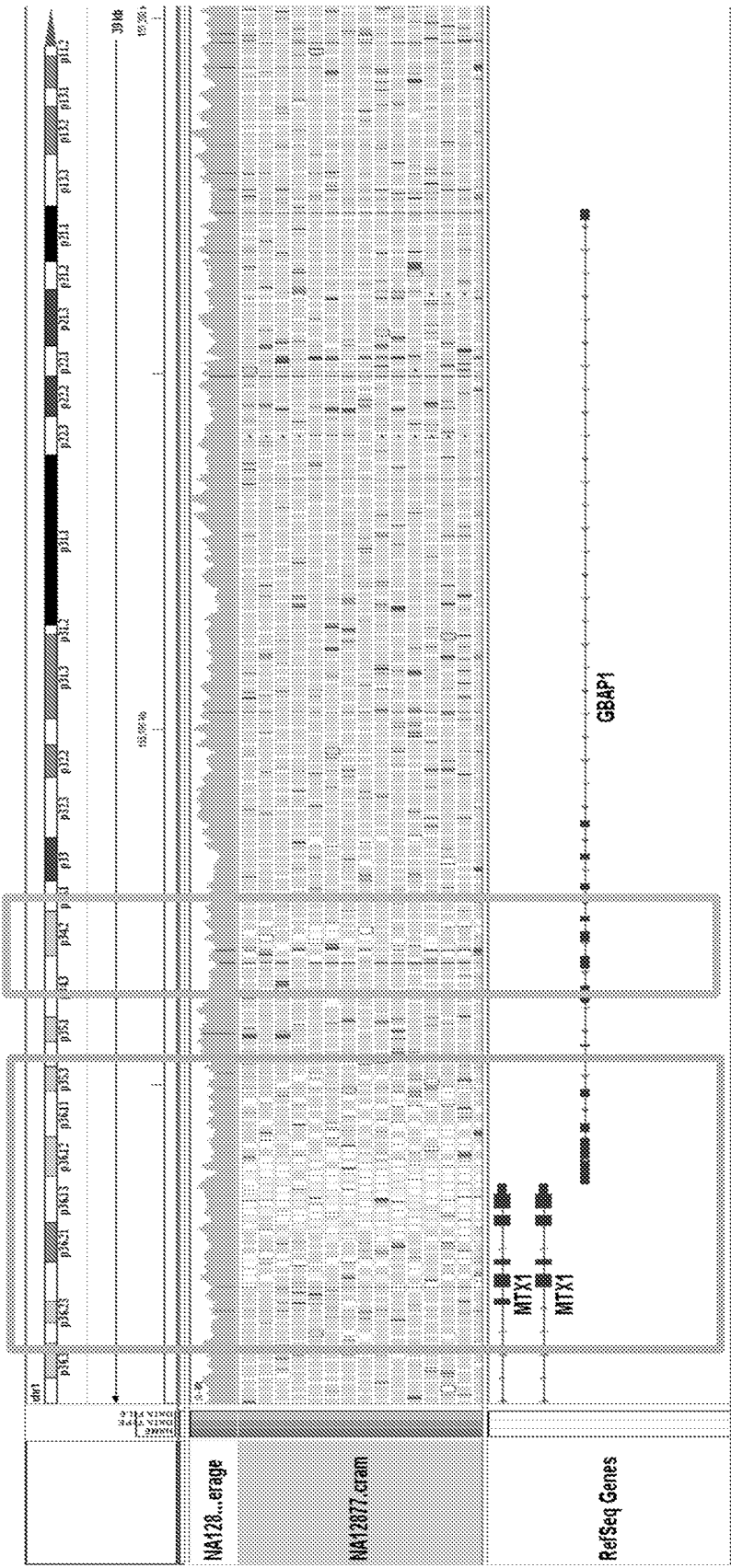


FIG. 2A1



1. Targeted CNV calling in this 10kb region without sequence homology to assess the presence of 20.6kb CNV
2. Identify deletion breakpoints by examining 82 GBA/GBAP differentiating SNP sites in the homology regions
3. Use depth to determine if wildtype GBA haplotype is present at one copy
4. Phase haplotypes to call gene recombination or conversion variants

FIG. 2A2

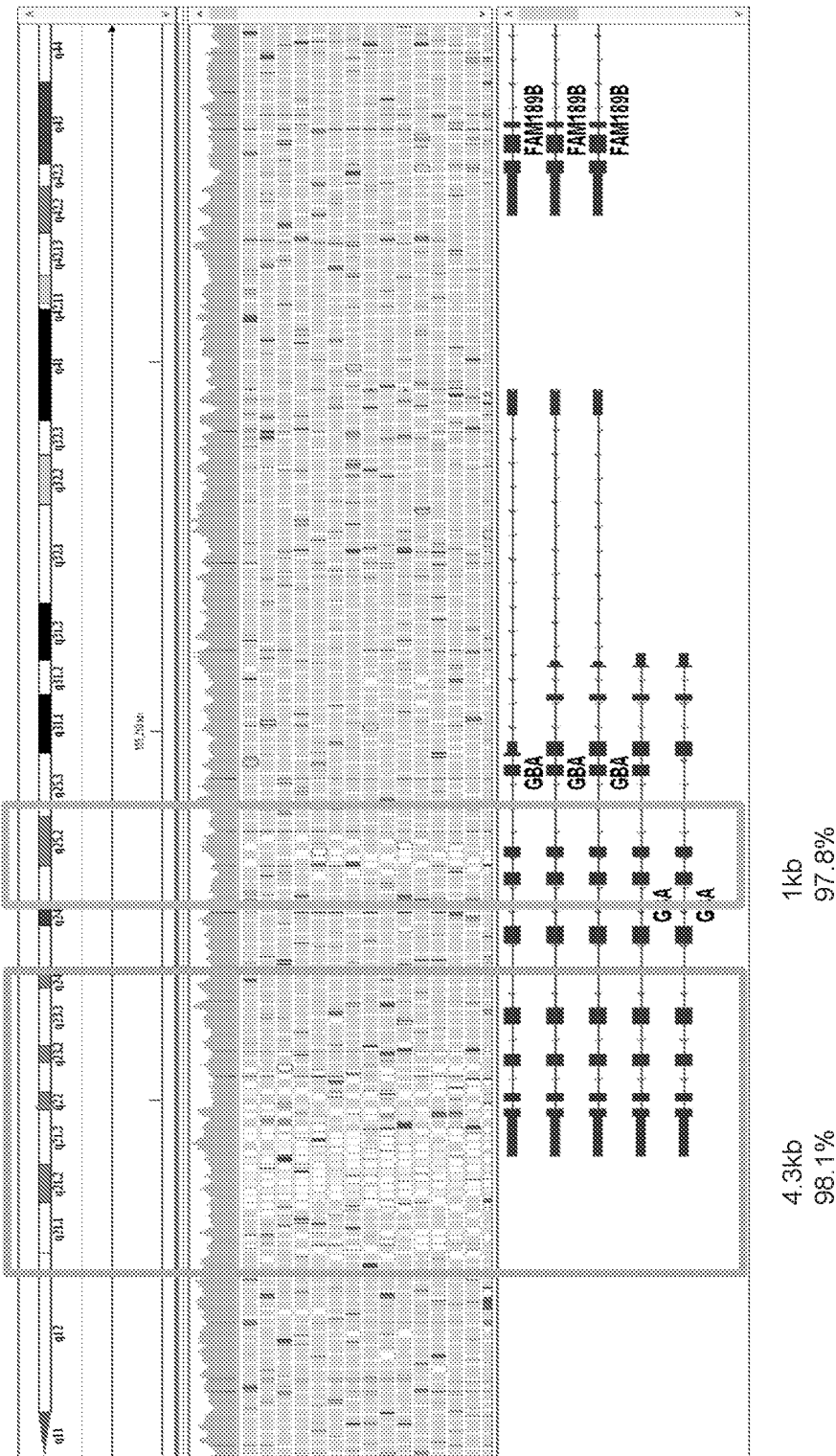


FIG. 2A2 (Cont'd)

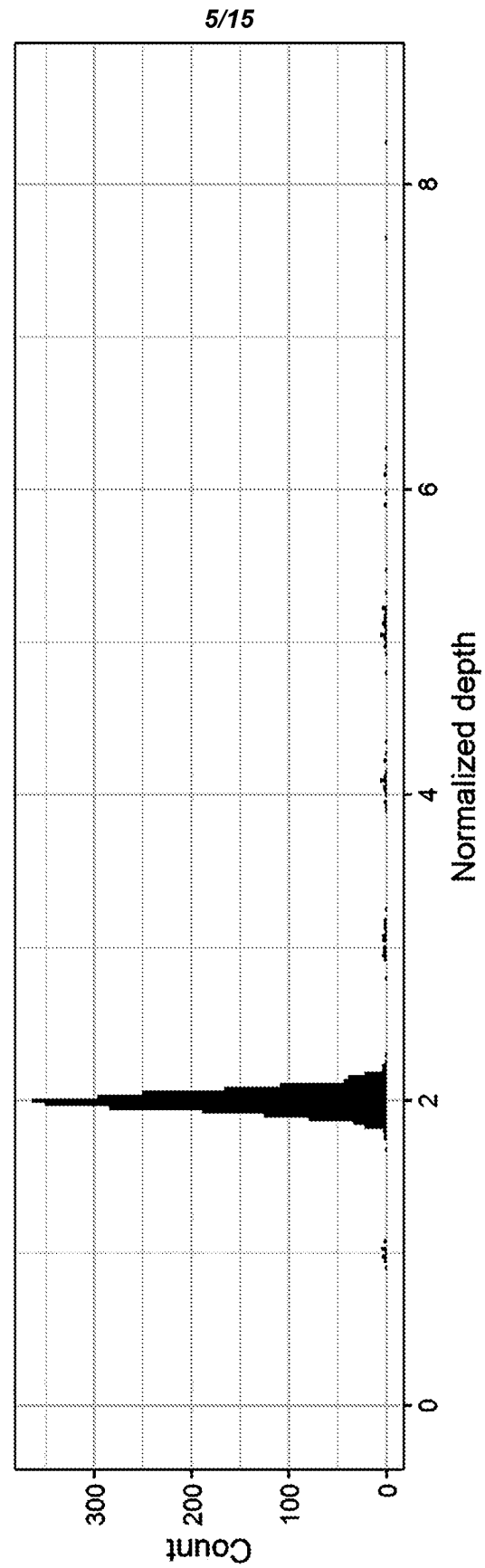


FIG. 2B1

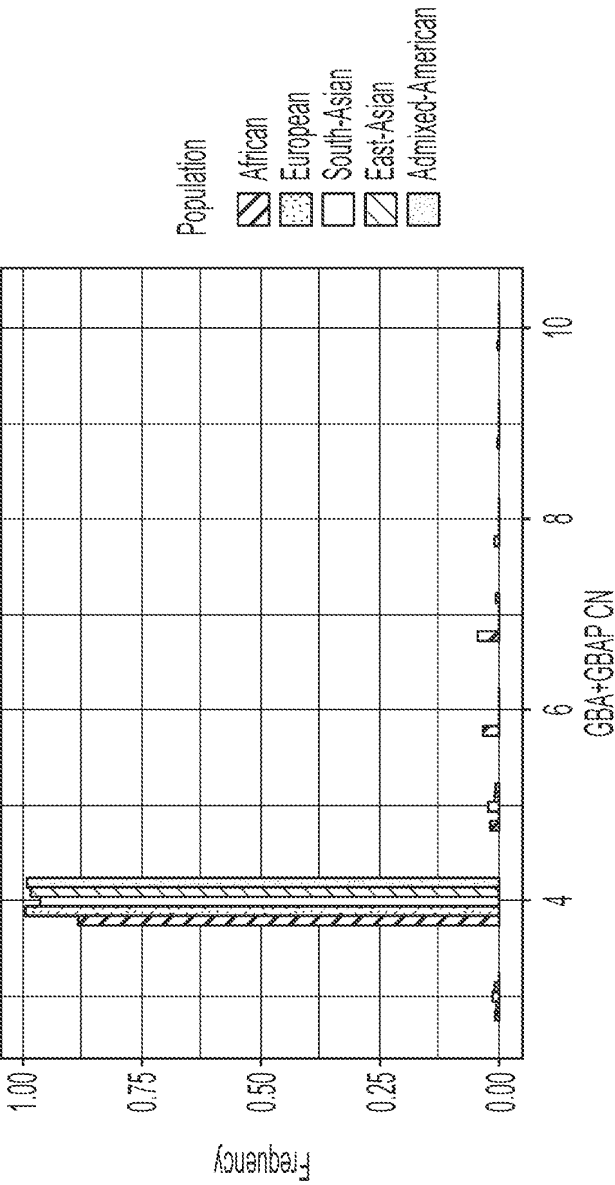
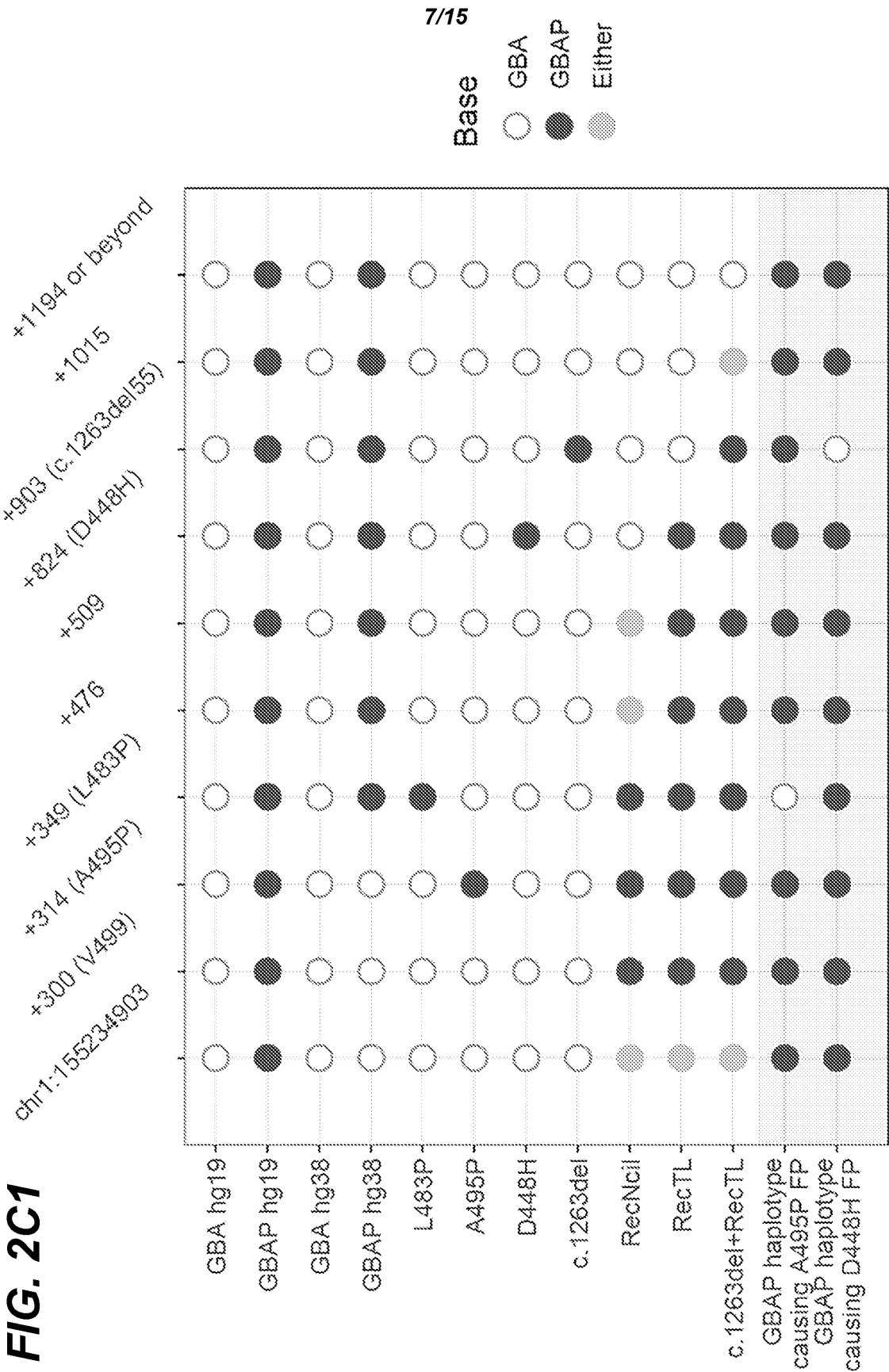


FIG. 2B2



1: GBA 2: GBAP	RecNciI									
	V460V				A456P					
	L444P				L444P					
	D409H				RecΔ55					
chr1:										
155234903	+300	+314	+349	+476	+509	+824	+845	+1015		
NA20815	1	1	2	1	1	1	1	1	L444P	
NA18991	1	1	2	1	1	1	1	1	L444P	
NA20513	1	1	2	1	1	1	1	1	L444P	
HG00310	1	1	2	1	1	1	1	1	L444P	
HG03967	1	1	2	1	1	1	1	1	L444P	
12 cases and 6 controls in AMP-PD										
NA20815	1	2	1	1	1	1	1	1	A456P	
3 cases in AMP-PD										
HG00119	2	2	2	2	2	2	2	1	RecNciI-RecΔ55	
HG00115	2	2	2	2	2	2	2	1	RecNciI-RecΔ55	
PD-POLR087WTH	2	2	2	2	2	2	2	1	RecNciI-RecΔ55	
HB-PD_INVEN86DWE	2	2	2	2	2	2	2	1	RecNciI-RecΔ55	
HG02439	1	1	1	1	1	1	2	x	RecΔ55	
HG02151	1	1	1	1	1	1	2	1	RecΔ55	
HG00422	2	2	2	2	2	1	1	1	RecNciI	
HB-PD_INVW835UXR	2	2	2	1	1	1	1	1	RecNciI	
HB-PD_INVWU757PGL	2	2	2	2	2	1	1	1	RecNciI	
PD-PD1205TP0	2	2	2	2	1	1	1	1	RecNciI	

FIG. 2C2

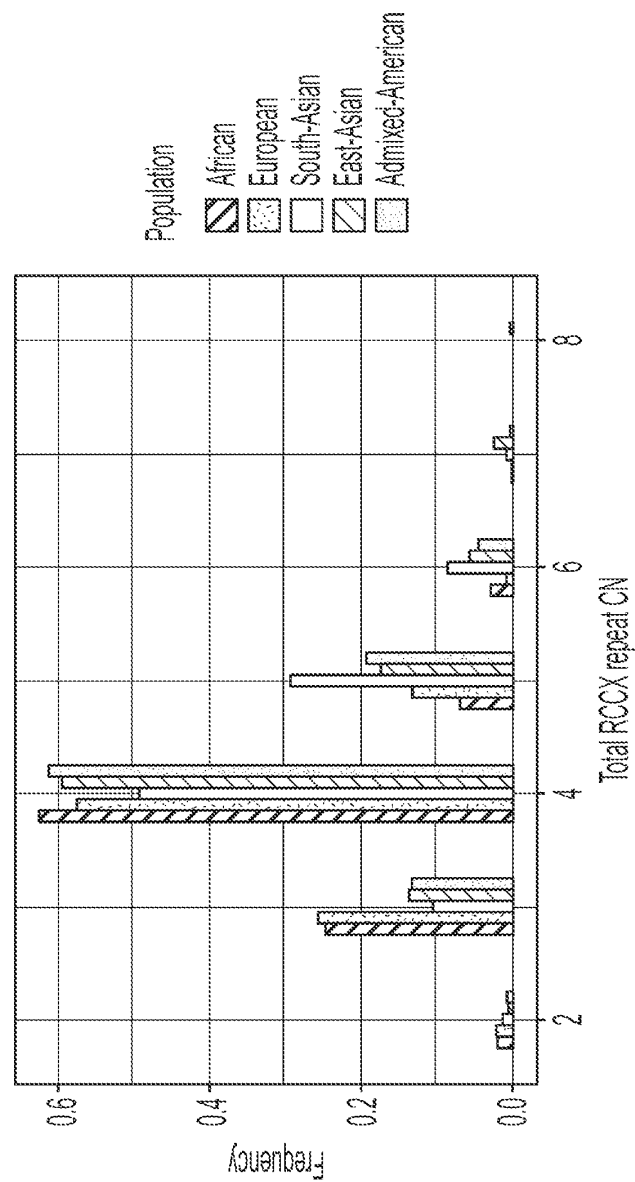


FIG. 3A

	CYP21A2	CYP21A2	CYP21A1P	CYP21A1P	
chr	position	base	position	base	Protein change
chr6	32039081	C/A	32006353	G	Splice variant
chr6	32039128	C	32006400	G	-
chr6	32039132	GGAGACTAC	32006404	G	Gly111ValfsTer21
chr6	32039143	C	32006407	T	-
chr6	32039426	T	32006690	A	Ile173Asn
chr6	32039548	C	32006812	G	-
chr6	32039802	T	32007066	C	-
chr6	32039807	T	32007071	A	Ile237Asn
chr6	32039810	T	32007074	A	Val238Glu
chr6	32039816	T	32007080	A	Met240Lys
chr6	32040182	GTGTTTTTT	32007446	GTGTTTTTT	Leu308PhefsTer6
chr6	32040216	G	32007481	C	-
chr6	32040421	C	32007686	T	Gln319Ter
chr6	32040535	C	32007800	T	Arg357Trp

FIG. 3B

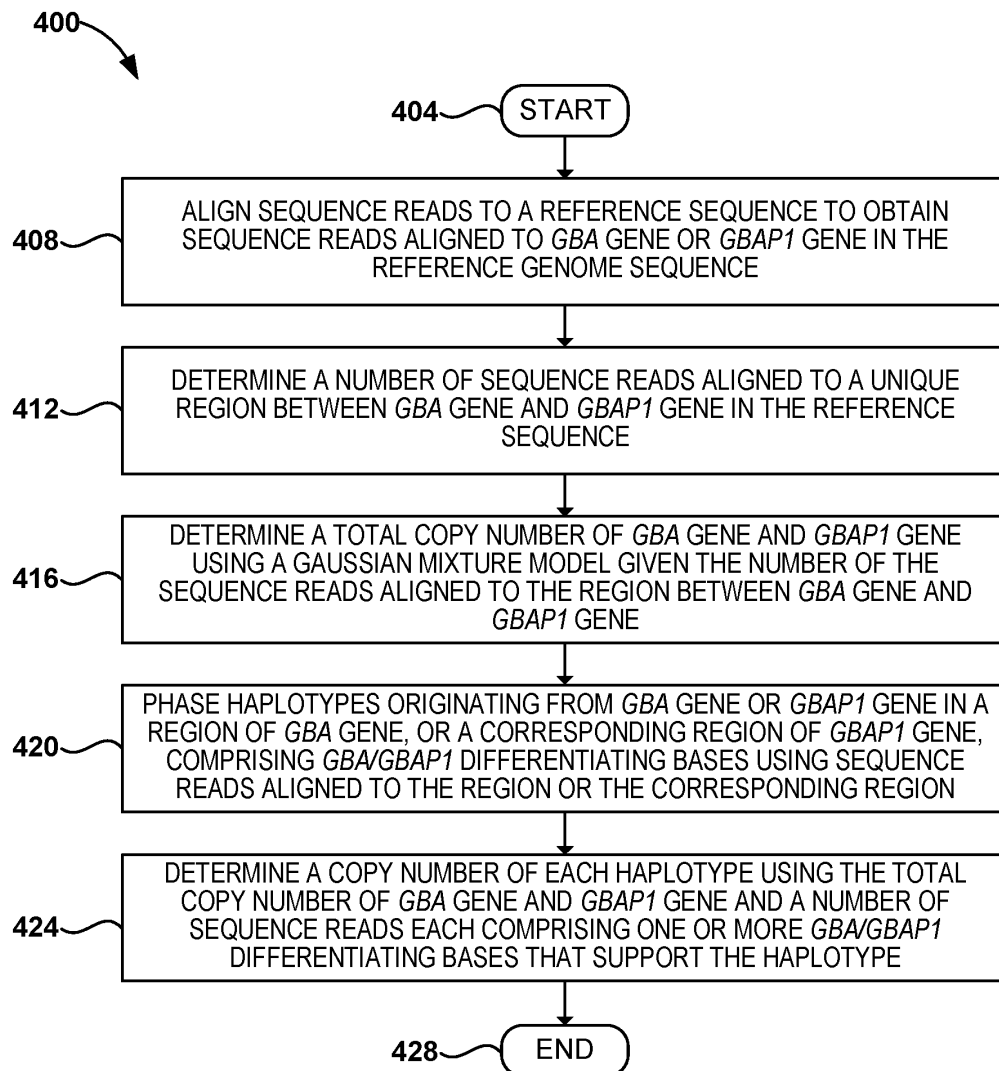
1: CYP21A2
2: CYP21A1P

Haplotype	Variants	# samples
11111111111121	Gln319Ter	1
111111111112211	Leu308PhefsTer6	1
11111111121111	Met240Lys	11
11111122221111	Ile237Asn, Val238Glu, Met240Lys	2
11111122222222	Ile237Asn, Val238Glu, Met240Lys, Leu308PhefsTer6, Gln319Ter, Arg357Trp	1
11112111111111	Ile173Asn	6
21111111111111	Splice variant	3
22221111111111	Splice variant, Gly111ValfsTer21	8
2222222222211	Splice variant, Gly111ValfsTer21, Ile173Asn, Ile237Asn, Val238Glu, Met240Lys, Leu308PhefsTer6	1
22222222222221	Splice variant, Gly111ValfsTer21, Ile173Asn, Ile237Asn, Val238Glu, Met240Lys, Leu308PhefsTer6, Gln319Ter	2

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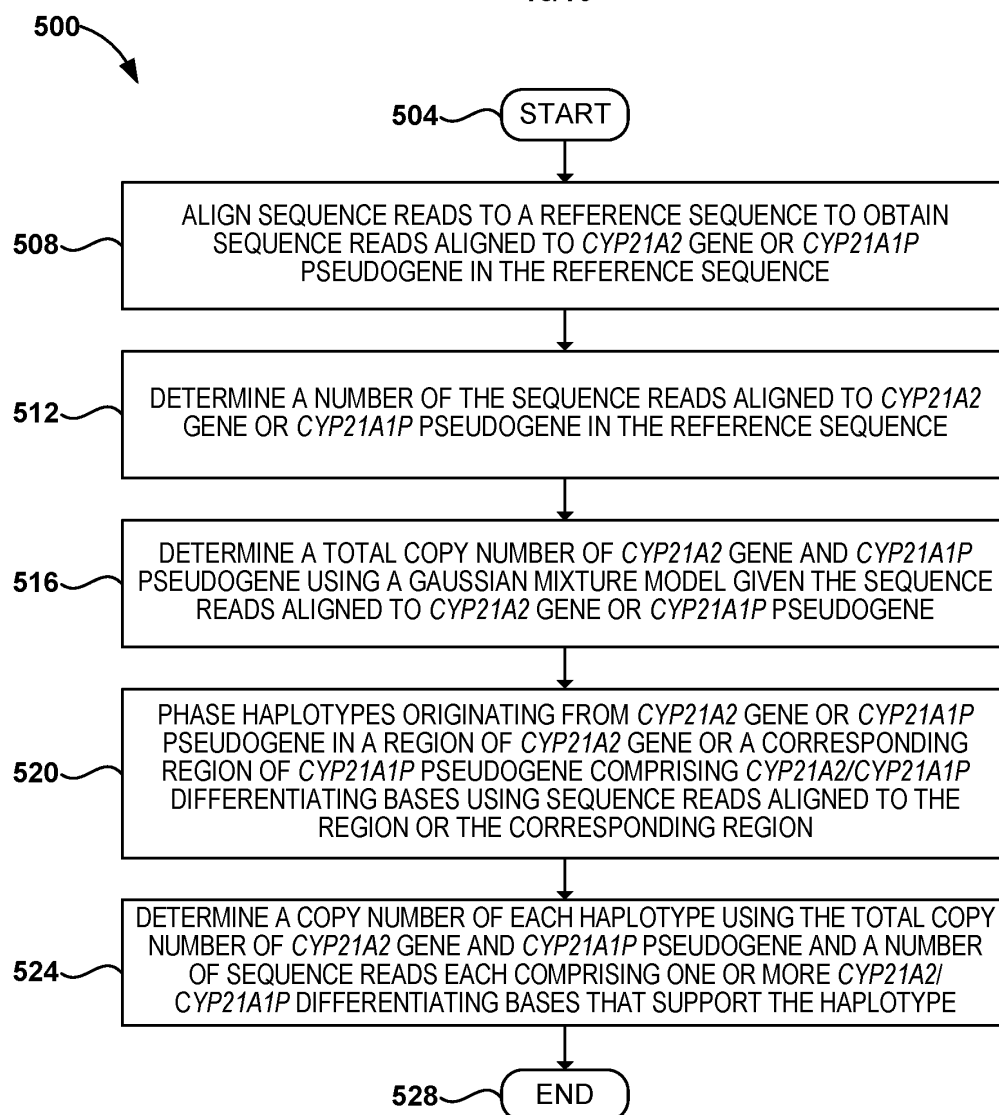
FIG. 3C

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**FIG. 4**

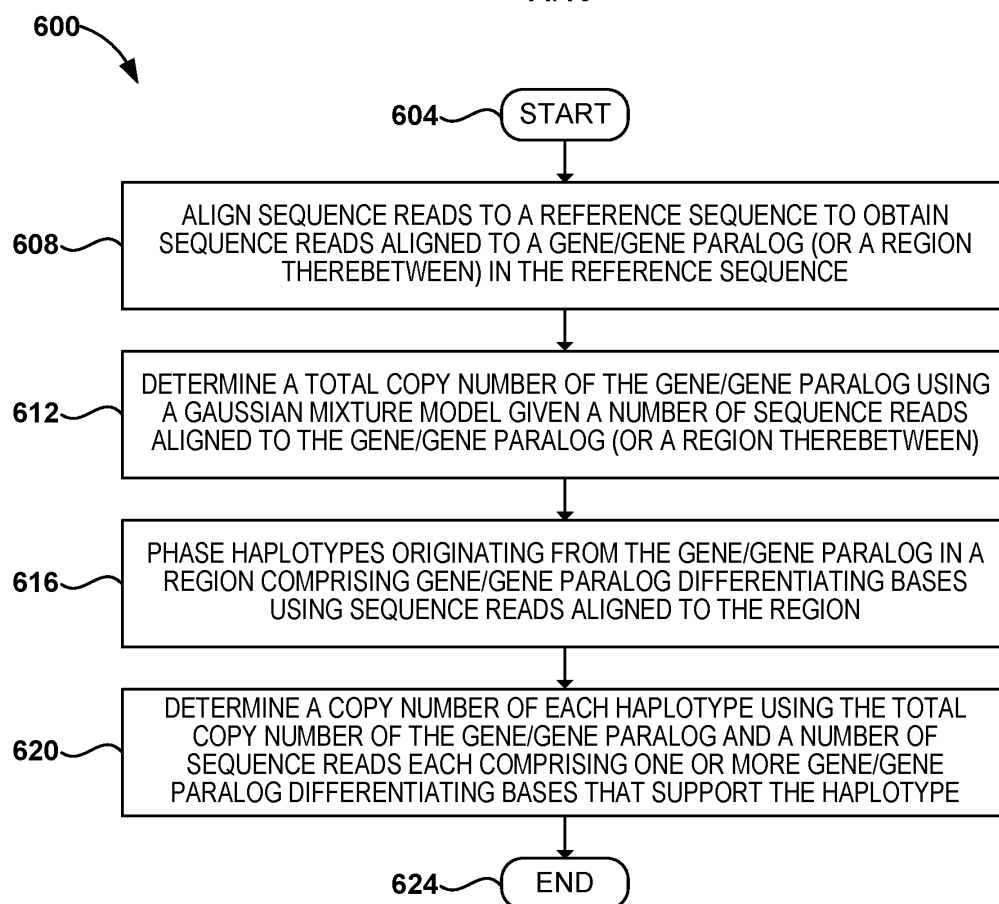
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**FIG. 5**

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**FIG. 6**

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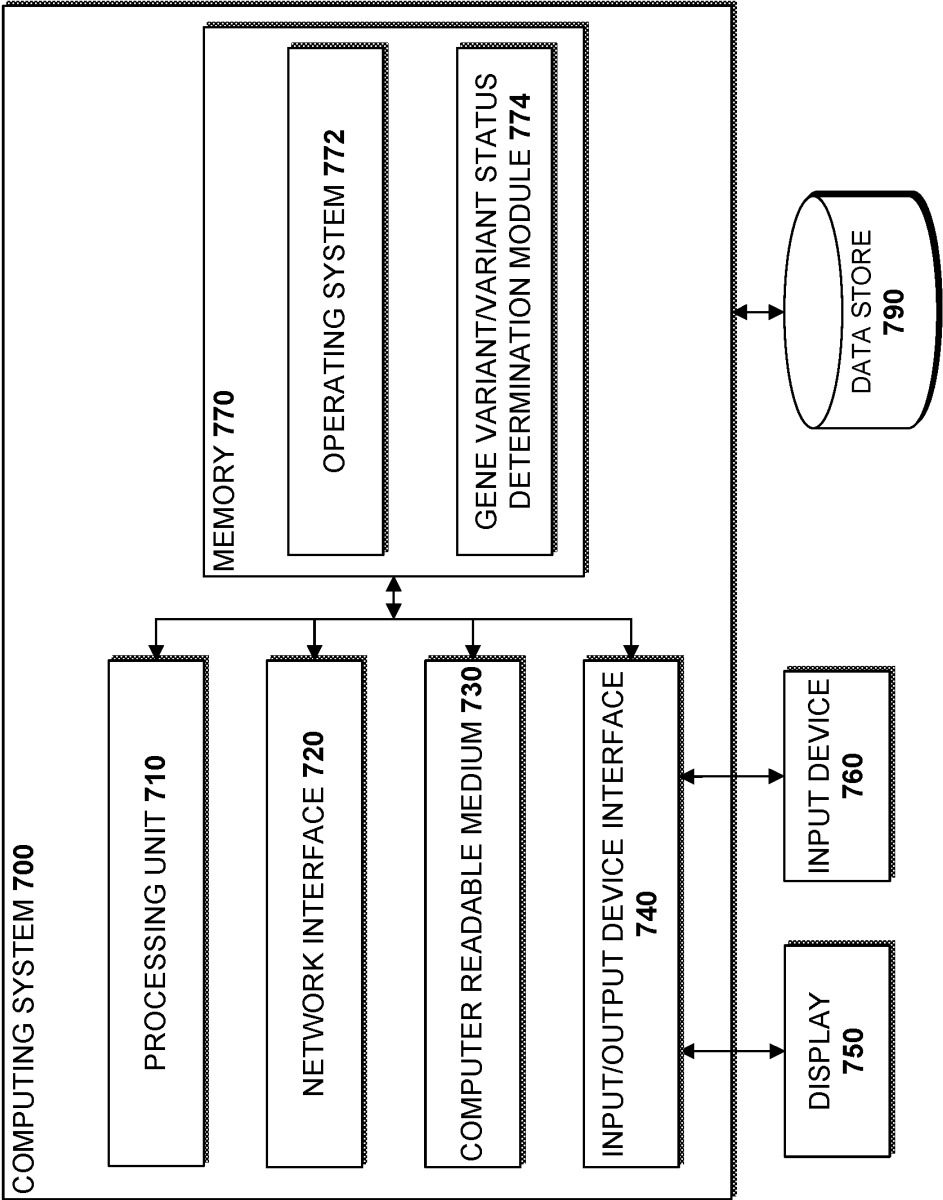


FIG. 7