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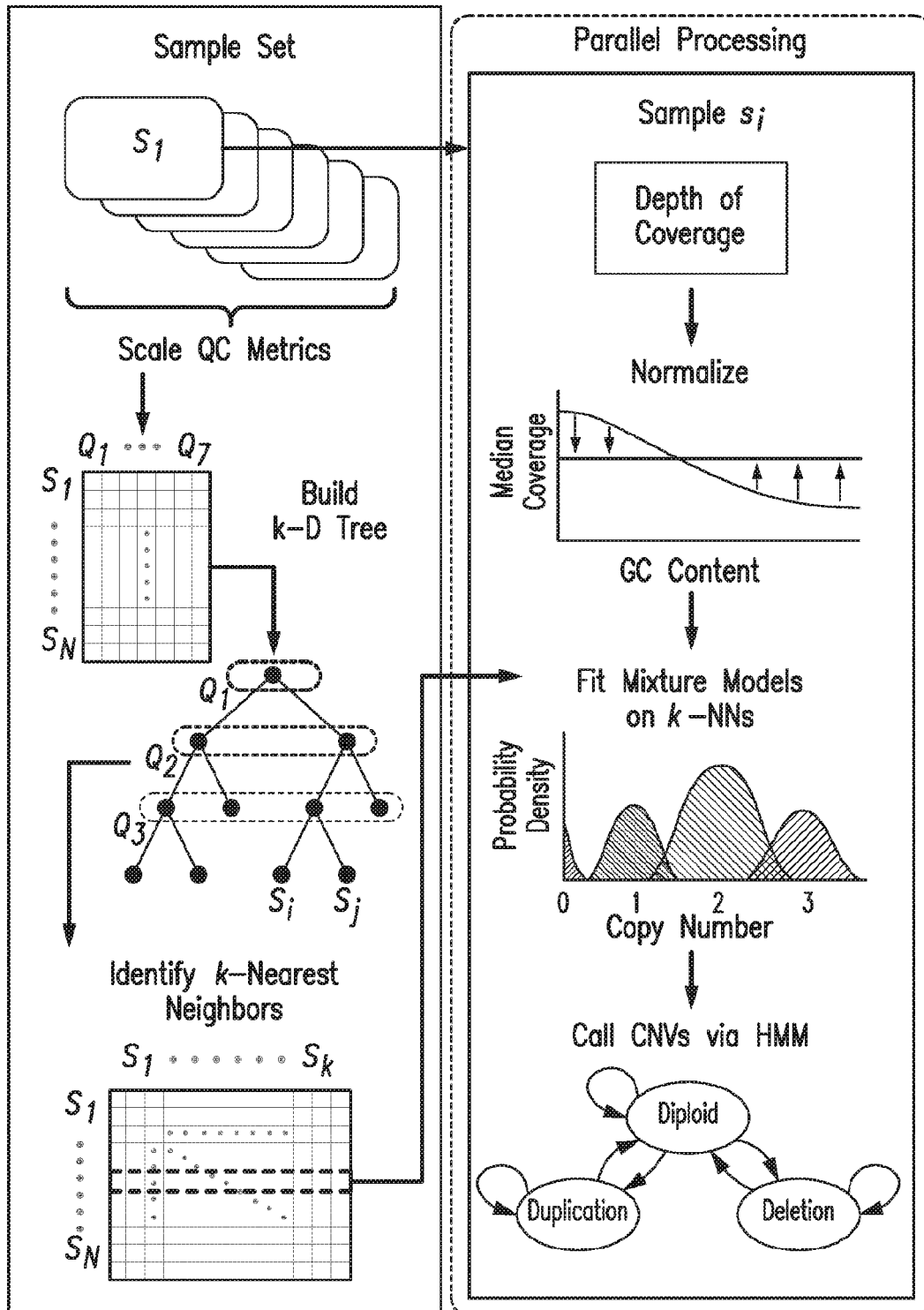


FIG. 1

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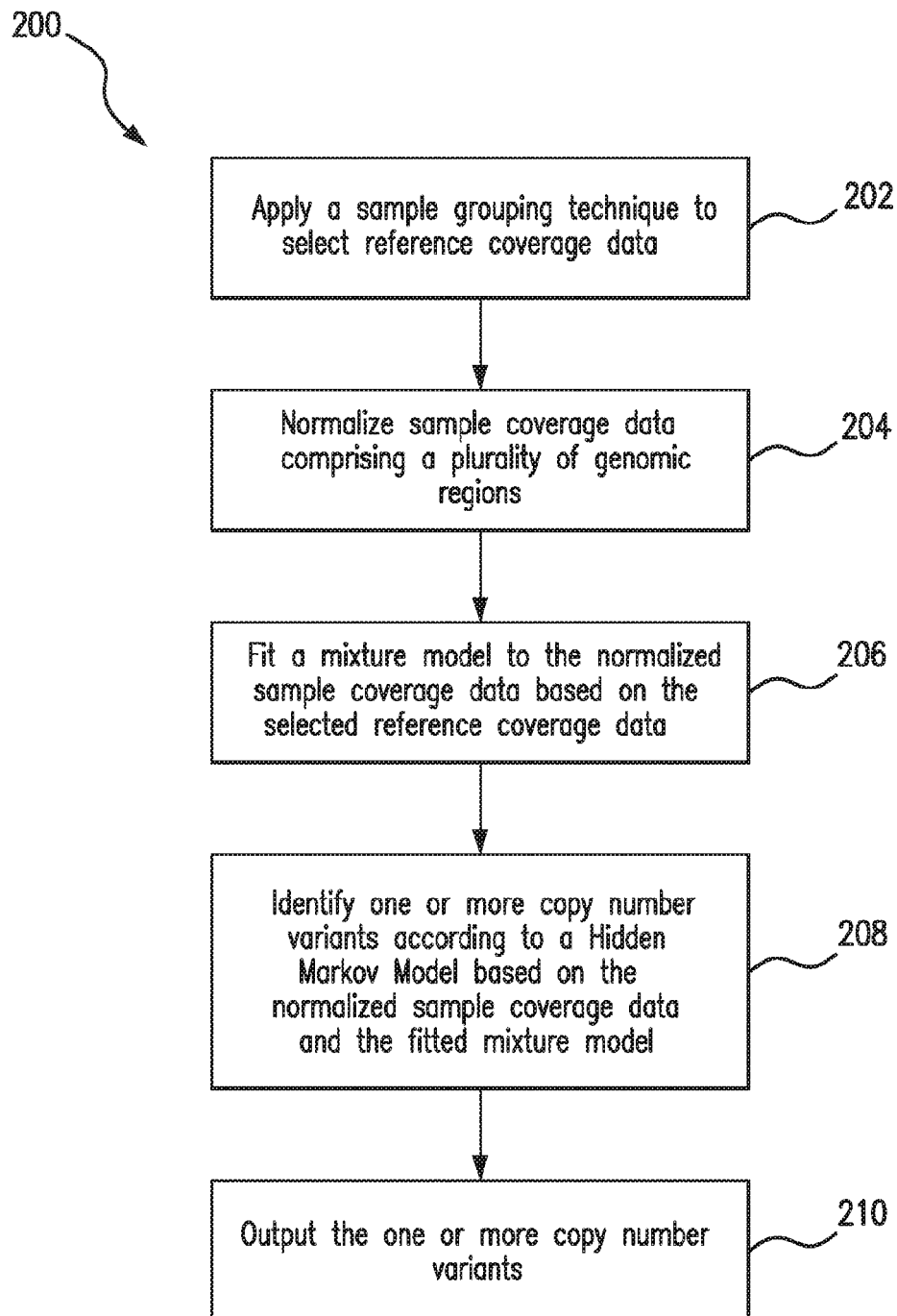


FIG. 2

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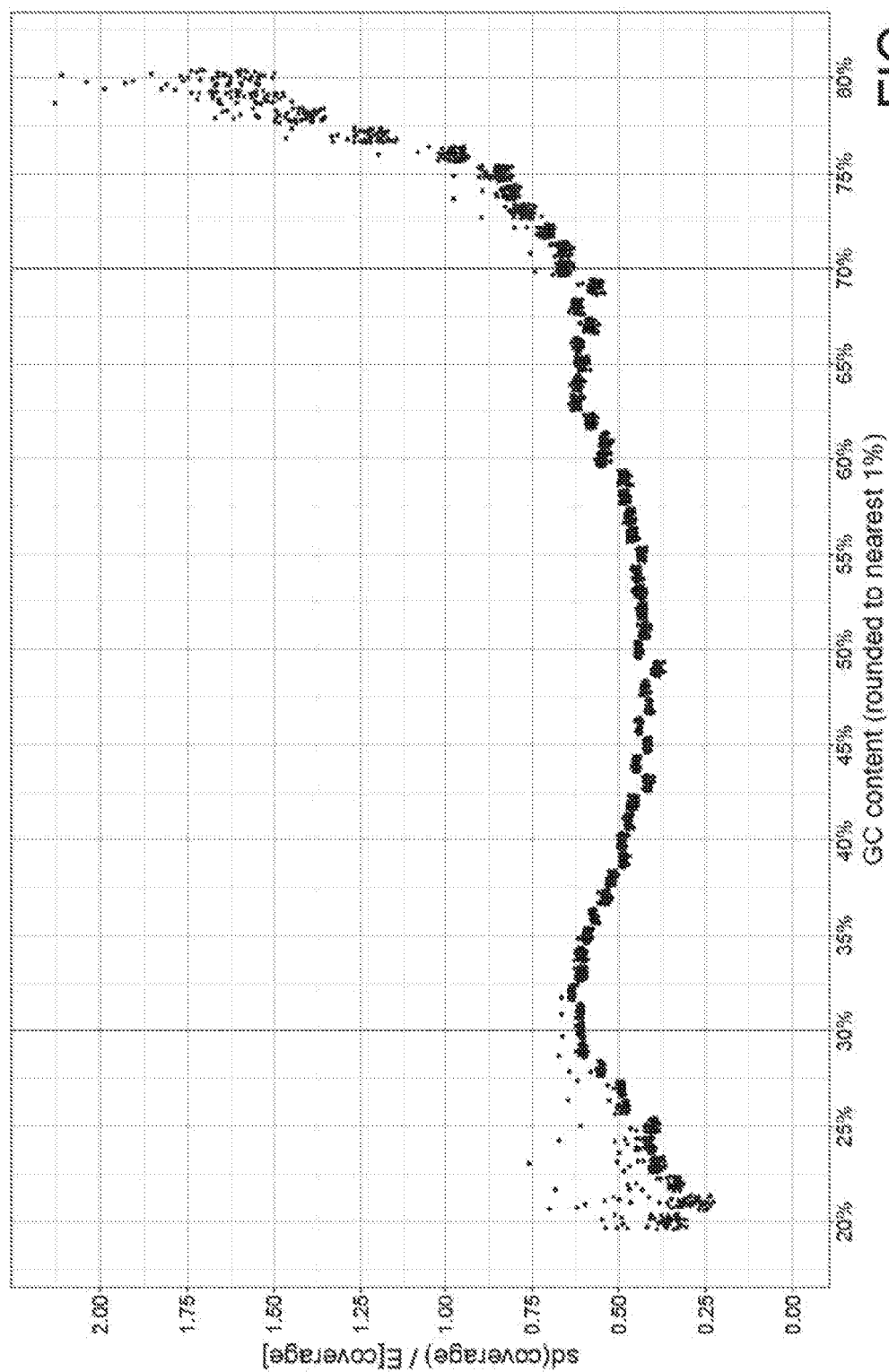
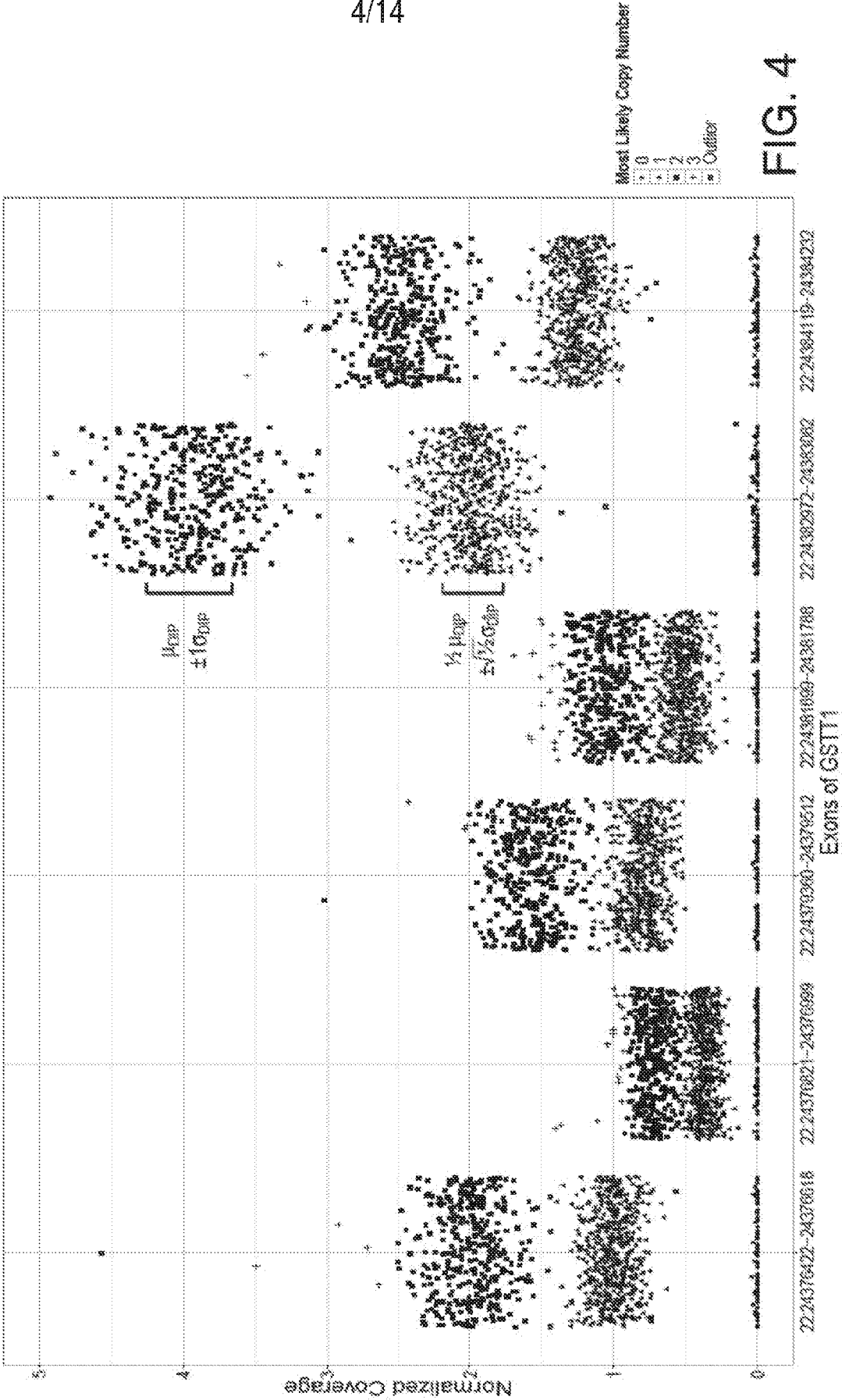


FIG. 3



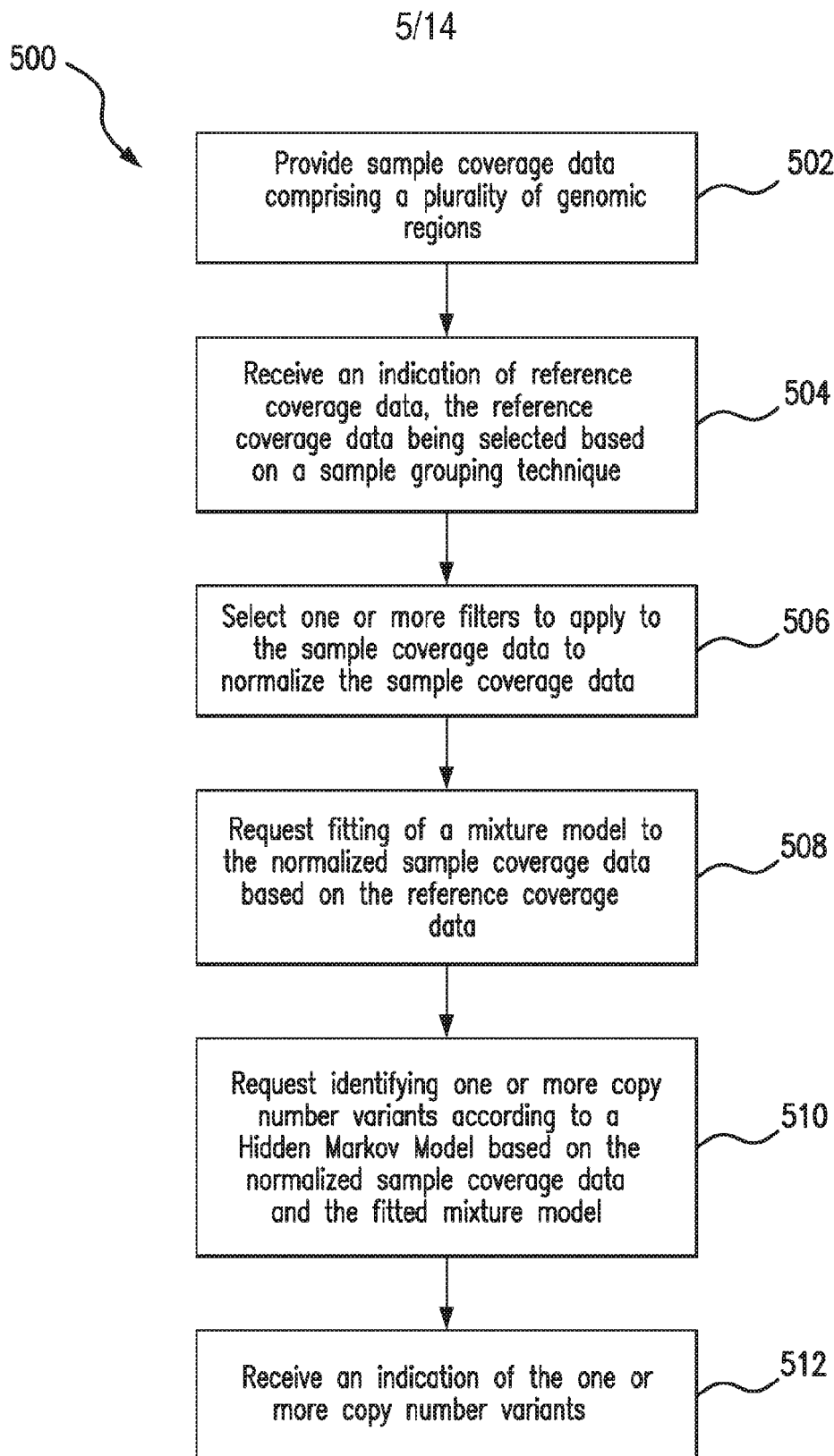


FIG. 5

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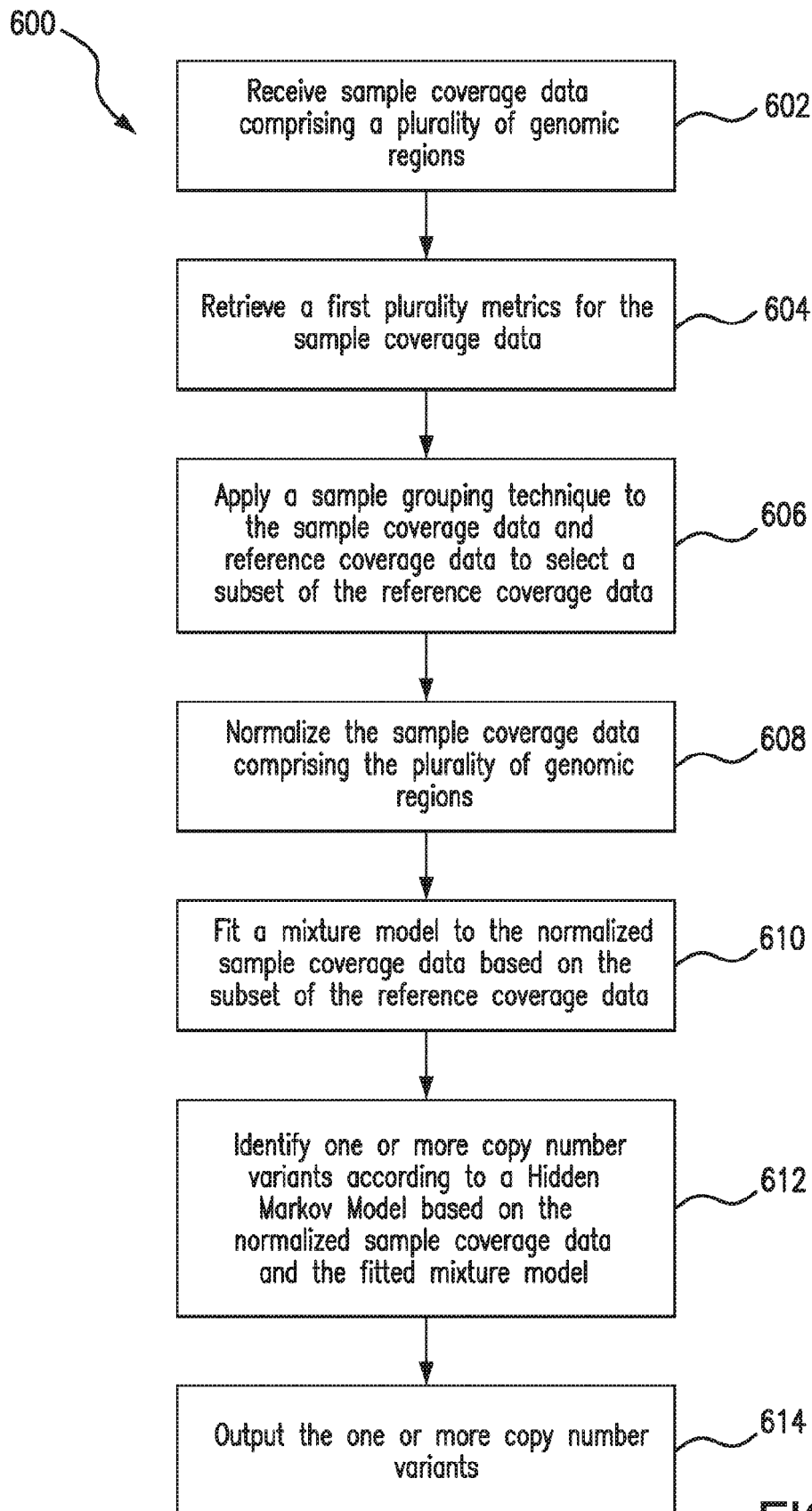


FIG. 6

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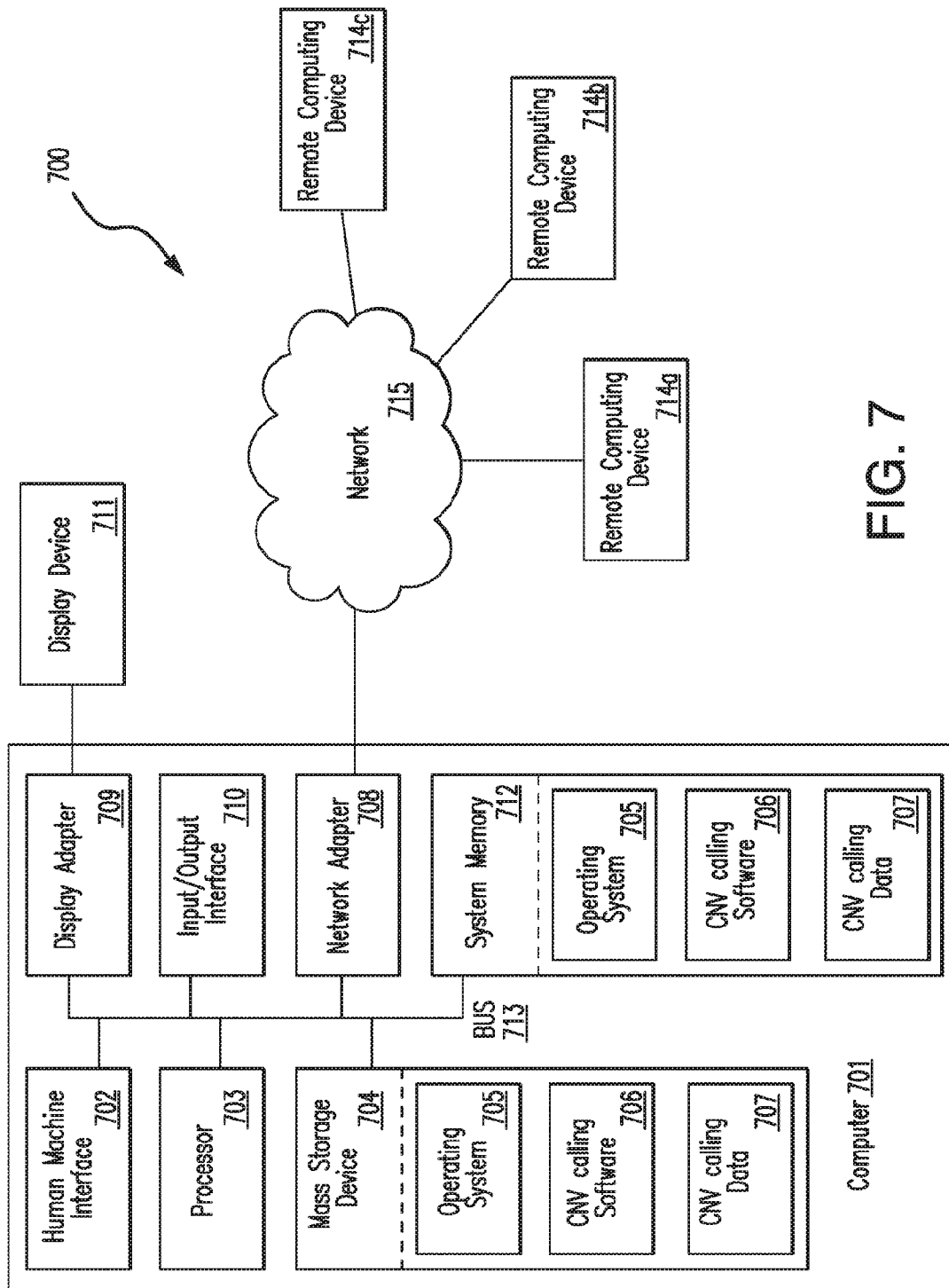


FIG. 7

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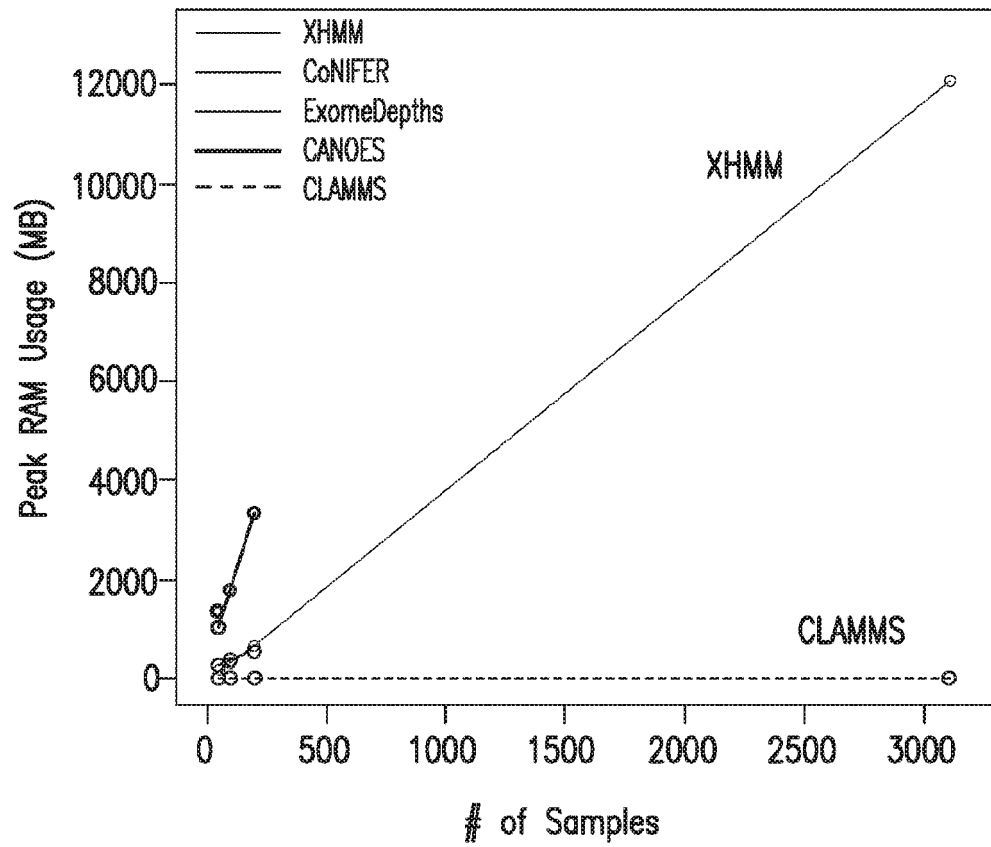


FIG. 8

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Algorithm	# Calls			Call Statistics (%)		
	Total	Common	Rare	Consistent	Transmitted	Inherited
CLAMMS	253	205	48	93.7	61.1	97.8
XHMM	86	25	61	38.5	10.6	23.3
CoNIFER	40	14	26	68.1	35.5	75.6
CANOES	26	15	11	92.6	0.0	0.0
ExomeDepth	279	202	77	69.9	30.6	64.7

FIG. 9

Metric	Algorithm	Any Overlap	33% Overlap	50% Overlap
Precision	CLAMMS	78.4	71.9	67.2
	XHMM (Q30)	66.4	60.2	55.4
	XHMM (Q60)	71.2	64.5	59.2
Recall	CLAMMS	65.4	49.7	41.9
	XHMM (Q30)	64.1	51.7	44.3
	XHMM (Q60)	59.9	49.7	42.5
F-score	CLAMMS	71.3	58.8	51.6
	XHMM (Q30)	65.2	55.6	49.2
	XHMM (Q60)	65.1	56.1	49.5

FIG. 10

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CNV	Size	Type	Gene [†]	Validated?
chr1:230371759-230415205	43,446	DUP	<i>GALNT2</i>	YES
chr10:113913307-114136207	222,900	DUP	<i>GPAM</i>	YES
chr11:104815479-105009807	194,328	DUP	<i>CARD16</i>	YES
chr12:21007961-21392124	384,163	DEL	<i>SLCO1B1</i>	YES
chr13:114514707-114538608	23,901	DUP	<i>GAS6</i>	YES
chr14:74753163-74991929	238,766	DUP	<i>NPC2</i>	YES
chr15:85147158-85681135	533,977	DEL	<i>NMB</i>	YES
chr16:21152620-21289573	136,953	DUP	<i>CRYM</i>	YES
chr17:12798256-12920439	122,183	DUP	<i>ELAC2</i>	YES
chr18:2544651-2707644	162,993	DUP	<i>SMCHD1</i>	YES
chr18:64176231-64239442	63,211	DEL	<i>CDH19</i>	YES
chr19:52271911-52588055	316,144	DUP	<i>FPR3</i>	YES
chr2:55910919-55920959	10,040	DUP	<i>PNPT1</i>	YES
chr3:124390506-124492760	102,254	DUP	<i>UMPS</i>	YES
chr4:10560030-10567775	26,550	DEL	<i>CLNK</i>	YES
chr7:82595086-82595804	718	DEL	<i>PCLO</i>	NO
chr7:121738503-121773781	35,278	DEL	<i>AASS</i>	YES
chrX:53560269-53622364	62,095	DUP	<i>HUWE1</i>	YES
chrX:105137824-105571052	433,228	DUP	<i>SERPINA7</i>	YES
chrX:120181538-120183935	2397	DEL	<i>GLUD2</i>	YES

[†] gene that led CNV to be selected (because of a disease association in HGMD)

FIG. 11

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CNV Locus	Size	Type	Gene [†]	# CNV [‡]	False +	False -
chr1:1634914-1663963	29,049	Both	<i>CDK11A</i>	27	0	5
chr1:16370987-16390132	19,145	DUP	<i>CLCNKB</i>	NA*	NA	NA
chr1:152573207-152586575	13,368	DEL	<i>LCE3B</i>	130	0	38
chr1:206317576-206331229	13,653	DEL	<i>CTSE</i>	10	0	0
chr10:27687222-27703180	15,958	DEL	<i>PTCHD3</i>	10	0	0
chr10:135340899-135379034	38,135	DUP	<i>SYCE1</i>	13	0	1
chr11:8959162-8959721	559	DEL	<i>ASCL3</i>	8	0	1
chr11:134151918-134214350	62,432	Both	<i>GLB1L3</i>	NA*	NA	NA
chr19:46623586-46627907	4,321	DEL	<i>IGFL3</i>	10	0	1
chr19:54801926-54804222	2,296	DEL	<i>LILRA3</i>	69	0	0
chr2:110881367-110962546	81,179	Both	<i>NPHP1</i>	7	0	0
chr21:37510122-37518976	108,854	DUP	<i>CBR3</i>	8	0	0
chr22:24373137-24384232	11,095	DEL	<i>GSTT1</i>	111	0	14
chr2:241627221-241710522	83,301	DUP	<i>KIF1A</i>	6	0	0
chr3:151531950-151545961	14,011	DEL	<i>AADAC</i>	11	0	1
chr4:3446037-3478270	32,233	DEL	<i>HGFAC</i>	6	0	0
chr4:69403342-69434293	30,861	DEL	<i>UGT2B17</i>	100	0	1
chr5:138651764-138658657	6,893	DUP	<i>MATR3</i>	NA*	NA	NA
chr7:142829209-142881529	52,320	DEL	<i>PIP</i>	9	0	0
chr9:215201-464220	249,019	DUP	<i>DOCK8</i>	6	1	0

* The variance of the TaqMan data for these loci was too high to determine copy number state

† gene that led CNV to be selected (because of a disease association in HGMD)

‡ Not representative of CNV allele frequency. The 165 samples tested for common CNV loci

FIG. 12

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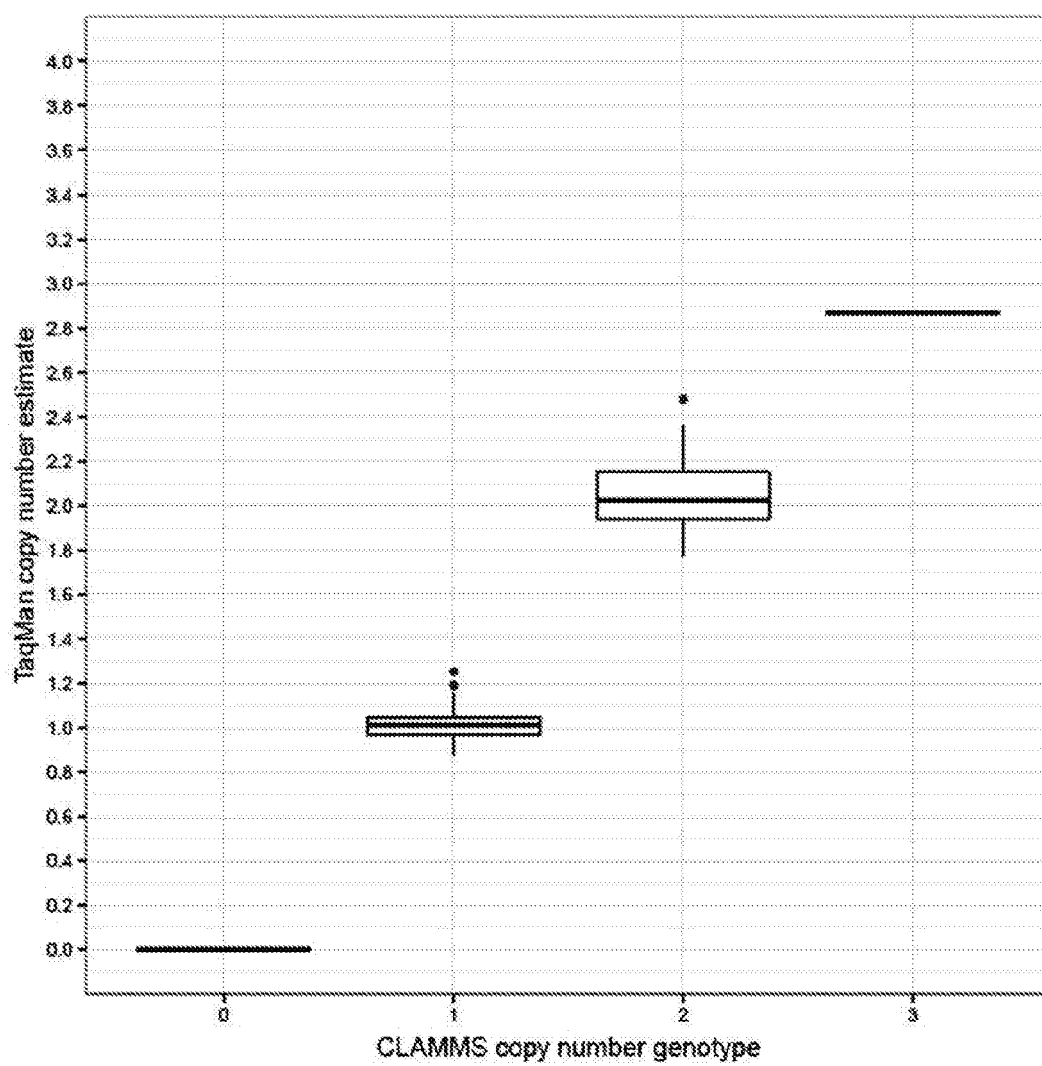


FIG. 13

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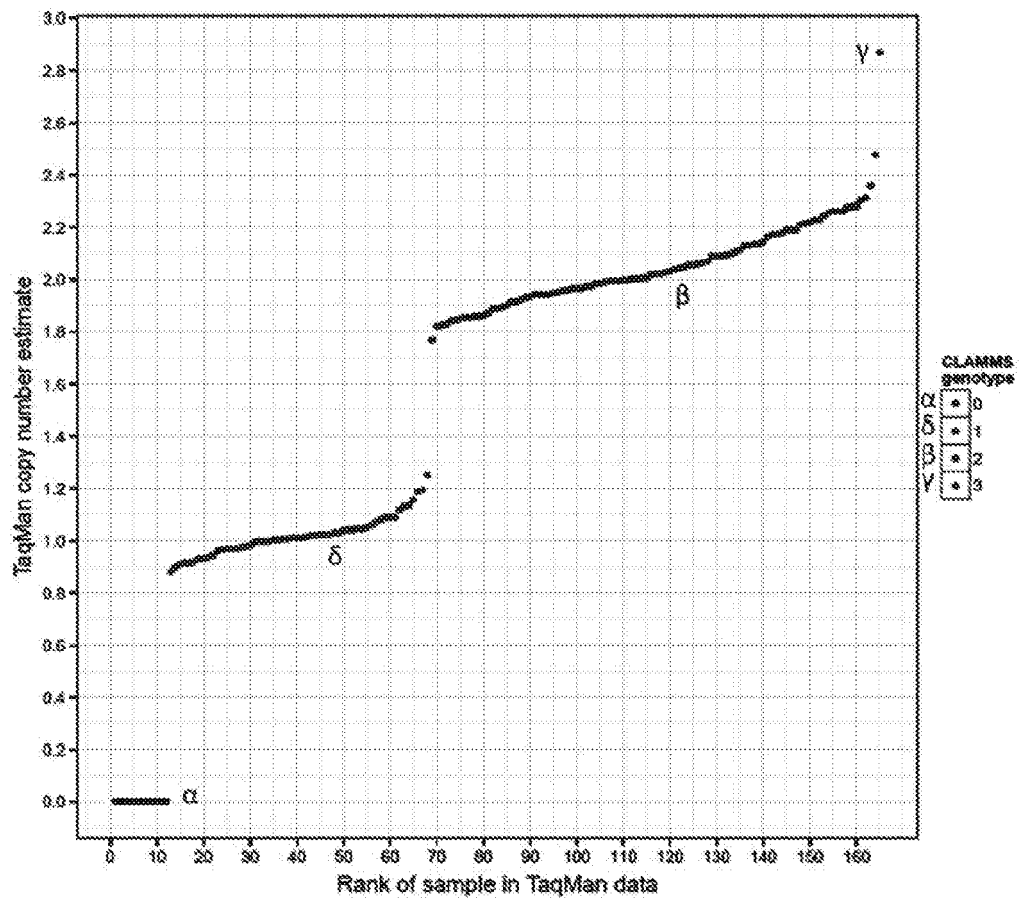


FIG. 14

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1	152573207	152586575	1: 152573207-152586575	sample_normalized_bed_coverage_files-0	DEL 1	2	69	67	-85348	50	9005	99	13079	37	-13012
1	248756130	248790430	1: 248756130-248790430	sample_normalized_bed_coverage_files-0	DEL 1	2	99	70	-71183	69	55176	99	33348	99	-33360
11	5798889	5799865	11: 5798889-5799865	sample_normalized_bed_coverage_files-0	DEL 1	1	79	66	-22919	7	10182	NA	NA	NA	NA
13	21729241	21742539	13: 21729241-21742539	sample_normalized_bed_coverage_files-0	DUP 3	6	99	99	-7917	99	9058	17	590	99	-6524
19	46623586	46627907	19: 46623586-46627907	sample_normalized_bed_coverage_files-0	DEL 1	4	99	26	-79385	63	23648	22	3556	27	-328
2	158974335	158978131	2: 158974335-158978131	sample_normalized_bed_coverage_files-0	DEL 1	2	99	40	-2723	55	2277	99	3585	99	-3680
22	24376821	24383082	22: 24376821-24383082	sample_normalized_bed_coverage_files-0	DEL 0	3	99	19	-63082	52	48887	99	4878	58	-1294
5	42629139	42629206	5: 42629139-42629206	sample_normalized_bed_coverage_files-0	DEL 1	1	24	59	-63163	70	59916	NA	NA	NA	NA
5	176516603	176563032	5: 176516603-176563032	sample_normalized_bed_coverage_files-0	DUP 3	17	99	19	-25091	44	55989	15	787	99	-38354
5	180375354	180429786	5: 180375354-180429786	sample_normalized_bed_coverage_files-0	DEL 0	7	99	62	-843	18	1091	99	884	99	-5297
7	72285958	72298655	7: 72285958-72298655	sample_normalized_bed_coverage_files-0	DEL 1	2	25	76	-417727	99	447125	70	12692	27	-12594

FIG. 15