

Supplement:

Table S1. The number of different variation types among the chromosomes

CH R	Summar y	Copy number allele									INS	INV
		0	0/2	0/2/ 3	0/2/3/ 4	234 5	2	23	23 4	*		
1	4671	2433	148	5	1	0	344	18	1	4	372	134 5
2	4894	2728	203	11	0	0	399	2	0	0	148 8	63
3	4180	2322	153	9	0	0	383	1	1	1	125 2	58
4	4068	2173	162	8	1	0	392	7	0	0	127 1	54
5	3811	2029	167	3	2	3	414	3	1	1	113 7	51
6	3622	1955	159	2	0	0	336	4	4	1	110 8	53
7	3658	2031	190	5	1	0	396	4	3	0	967	61
8	3151	1758	155	1	3	0	309	7	0	1	864	53
9	2623	1478	168	9	1	0	228	7	0	0	690	42
10	2755	1473	129	7	1	0	297	8	5	0	794	41
11	2912	1614	129	3	2	2	297	4	0	1	825	35
12	2898	1548	114	5	2	0	334	10	0	0	843	42
13	2138	1081	96	2	0	1	232	1	0	0	695	30
14	1829	1001	64	2	0	0	186	4	1	2	546	23
15	1612	862	78	1	0	7	161	1	0	7	483	16
16	1806	1103	121	5	8	6	211	4	3	4	325	16
17	1680	995	85	1	0	0	181	6	1	0	389	22
18	1720	980	101	0	3	0	155	0	0	0	465	16
19	1437	973	102	2	0	0	136	3	0	0	212	9
20	1290	732	55	1	0	0	151	0	0	0	334	17
21	761	415	22	0	2	0	67	3	0	0	241	11
22	732	466	41	4	7	1	76	4	1	0	127	5
X	1803	974	326	5	0	3	28	0	0	2	465	
Su m	60051	3312 4	296 8	91	34	23	571 3	10 1	21	2 4	17956	

* represents the value of the copy number allele is larger than 4

Table S2. The performance of different copy number allele on the same framework

Random Forest						
	Accuracy	Precision	Recall	F-Measure	ROC Area	PRC Area
CN0	0.833	0.852	0.833	0.808	0.975	0.920
CN012	0.8841	0.892	0.884	0.885	0.965	0.889
CN02	0.9003	0.900	0.900	0.947	0.914	0.924
CN023	0.5897	0.590	0.590	0.742	0.675	0.680
CN0234	0.7273	0.710	0.727	0.830	0.575	0.621
CN4+	0.5455	0.624	0.545	0.484	0.742	0.650
Multilayer Perceptron						
	Accuracy	Precision	Recall	F-Measure	ROC Area	PRC Area
CN0	0.841	0.842	0.841	0.840	0.928	0.826
CN012	0.6667	0.667	0.667	0.800	0.298	0.452
CN02	0.7563	0.688	0.756	0.797	0.840	0.762
CN023	0.7395	0.671	0.739	0.774	0.848	0.768
CN0234	0.7576	0.733	0.758	0.545	0.522	0.583
CN4+	0.2121	0.398	0.212	0.150	0.526	0.561
SVM						
	Accuracy	Precision	Recall	F-Measure	ROC Area	PRC Area
CN0	0.855	0.850	0.855	0.851	0.908	0.790
CN012	0.820	0.743	0.819	0.577	0.840	0.760
CN02	0.748	0.671	0.748	0.786	0.790	0.650
CN023	0.641	0.622	0.641	0.286	0.563	0.507
CN0234	0.758	0.733	0.758	0.846	0.636	0.625
CN4+	0.515	0.550	0.515	0.498	0.617	0.433
Naive Bayes						
	Accuracy	Precision	Recall	F-Measure	ROC Area	PRC Area
CN0	0.870	0.878	0.870	0.872	0.955	0.869
CN012	0.752	0.740	0.750	0.577	0.806	0.828
CN02	0.530	0.757	0.529	0.474	0.843	0.752
CN023	0.308	0.570	0.308	0.322	0.699	0.607
CN0234	0.212	0.398	0.212	0.150	0.526	0.561
CN4+	0.364	0.413	0.364	0.325	0.641	0.506
Decision Tree						
	Accuracy	Precision	Recall	F-Measure	ROC Area	PRC Area
CN0	0.790	0.597	0.790	0.748	0.905	0.740
CN012	0.789	0.570	0.790	0.752	0.864	0.581
CN02	0.622	0.567	0.622	0.594	0.647	0.518
CN023	0.744	0.611	0.783	0.733	0.639	0.584
CN0234	0.697	0.716	0.697	0.607	0.401	0.503
CN4+	0.394	0.302	0.394	0.330	0.486	0.345

Table S3. The performance of different copy number allele on the same framework

Feature Importance	AC	AF	NS	AN	EAS_AF	EUR_AF	AFR_AF	AMR_AF	SAS_AF
Data 1	0.15	0.15	0.00	0.00	0.11	0.16	0.15	0.15	0.00
Data 2	0.18	0.00	0.00	0.16	0.15	0.10	0.18	0.00	0.07
Data 3	0.18	0.00	0.05	0.16	0.15	0.10	0.18	0.00	0.00
Data 4	0.16	0.16	0.00	0.00	0.10	0.24	0.16	0.16	0.00
Data 5	0.18	0.00	0.00	0.16	0.15	0.10	0.18	0.00	0.10

Precision: Precision=TP/(TP+FP)

Recall: Recall=TP/(TP+FN)

F-measure: F-measure=2×Precision×Recall/(Precision+Recall)

Accuracy: Accuracy=(TP+TN)/(TP+TN+FP+FN)

$$AUC = \frac{\sum_{i \in \text{positiveClass}} \text{rank}_i - \frac{M(1+M)}{2}}{M \times N}$$