

## **Supplemental Materials**

- Supplemental figures (10)
- Supplemental tables (11)

## Supplementary figures

Figure S1

*Schematic diagram for the study design of GWAS on ABO blood types in a Chinese population*

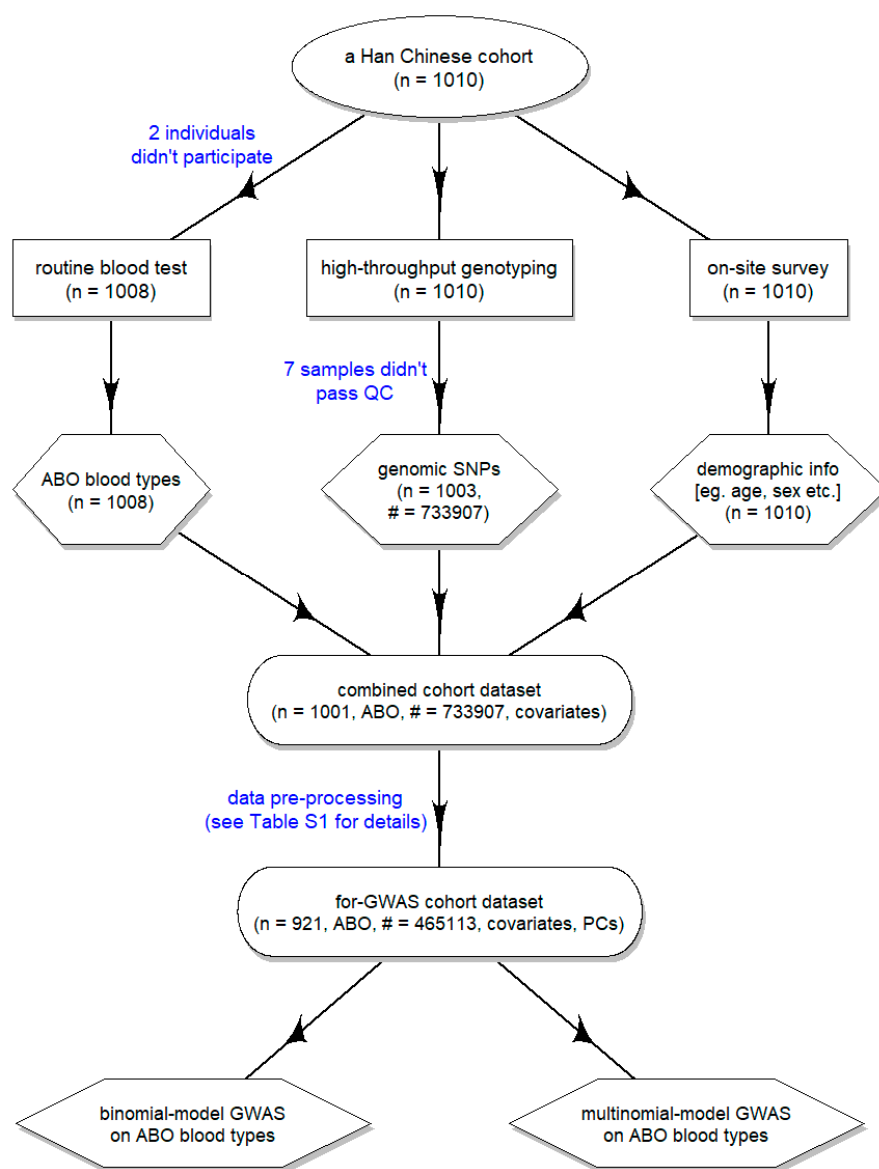
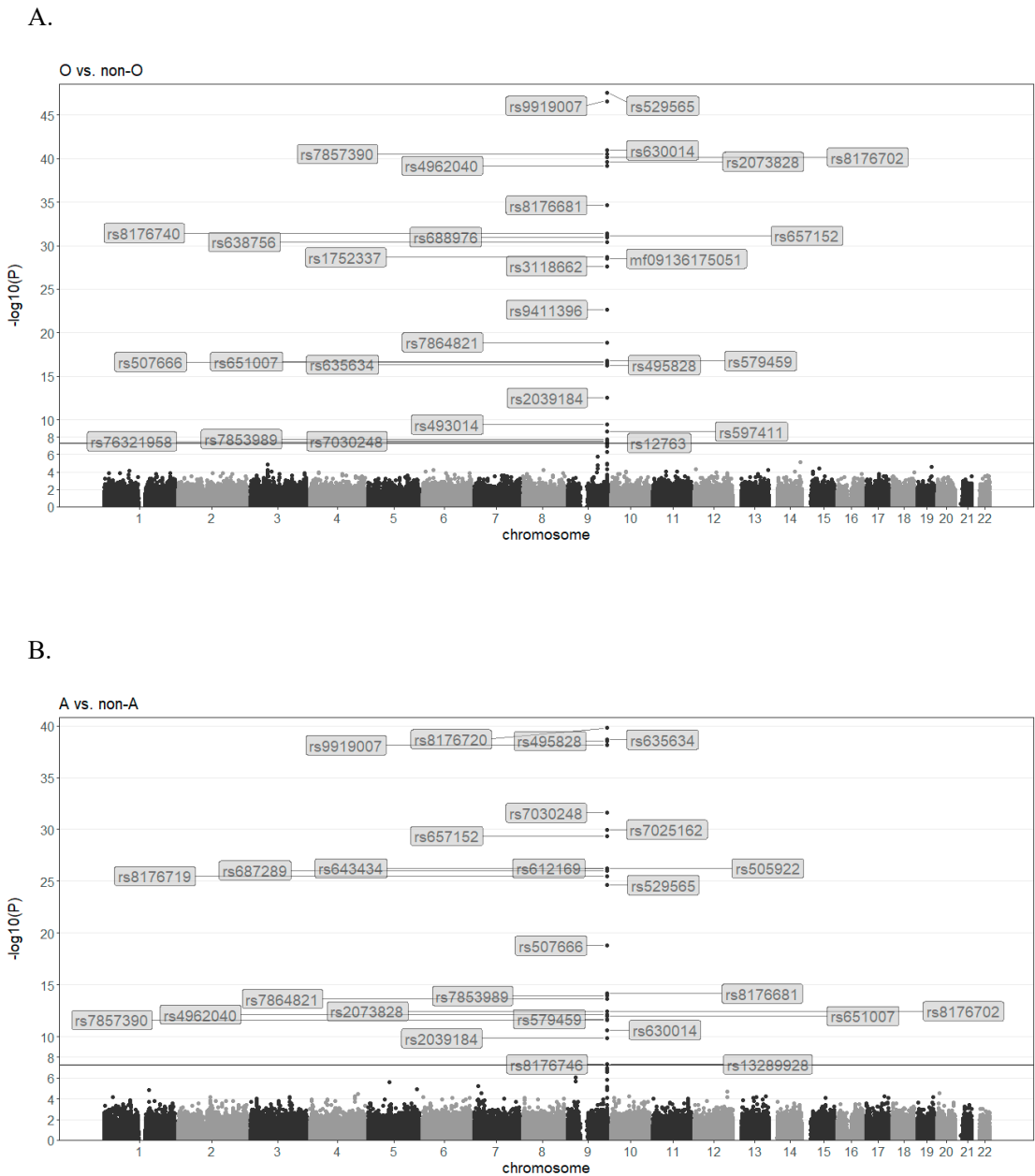
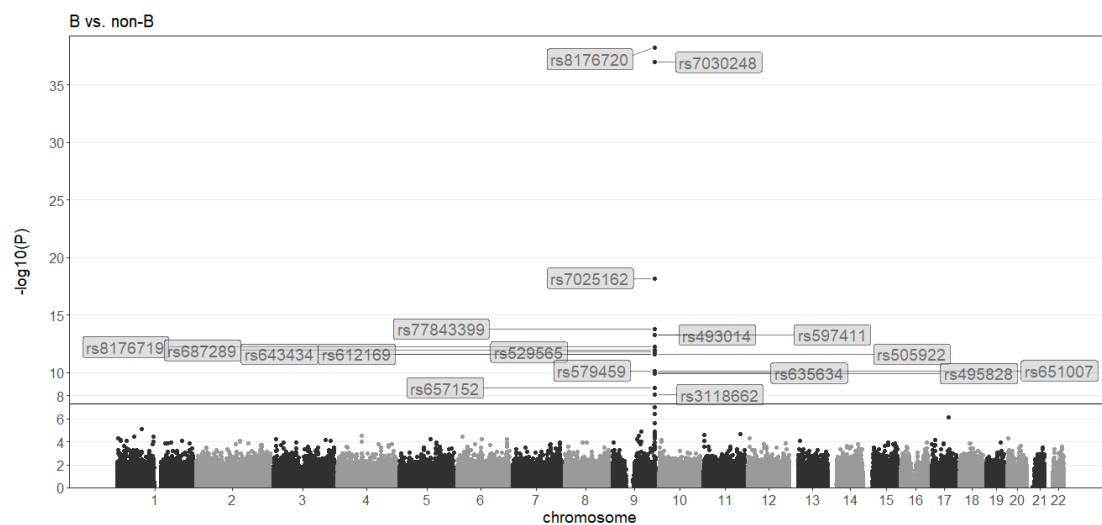


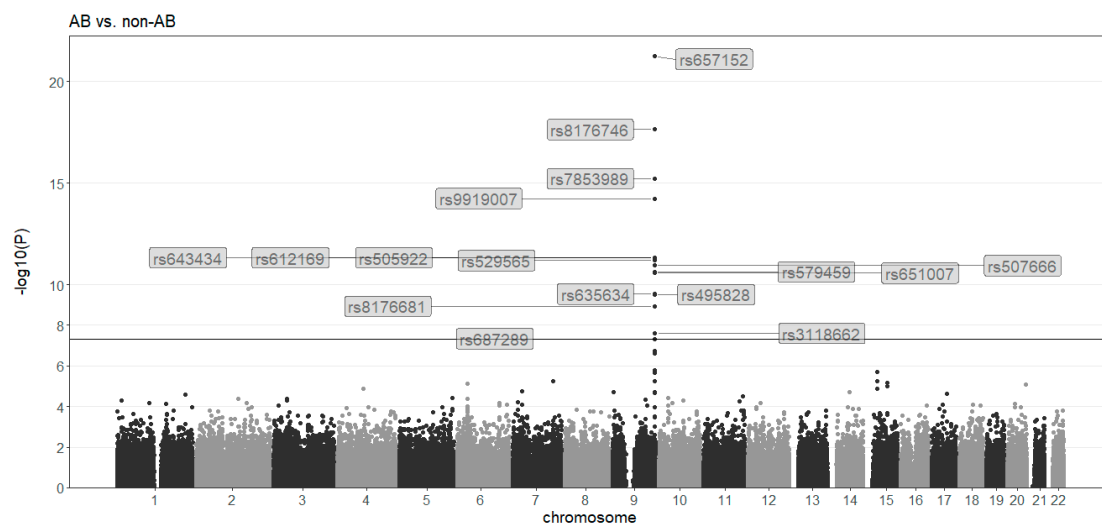
Figure S2



C.

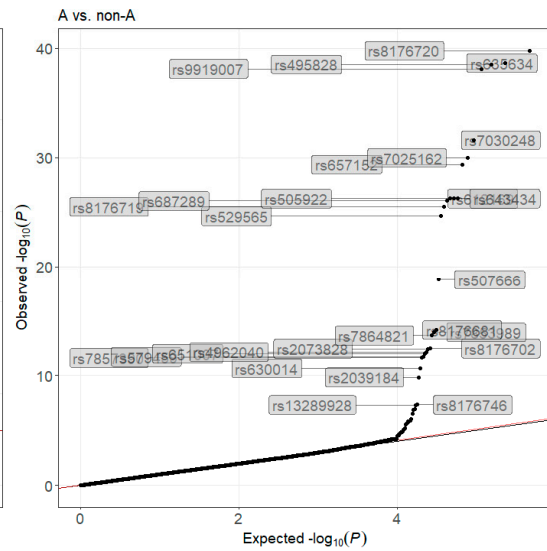


D.



A.

B.



D.

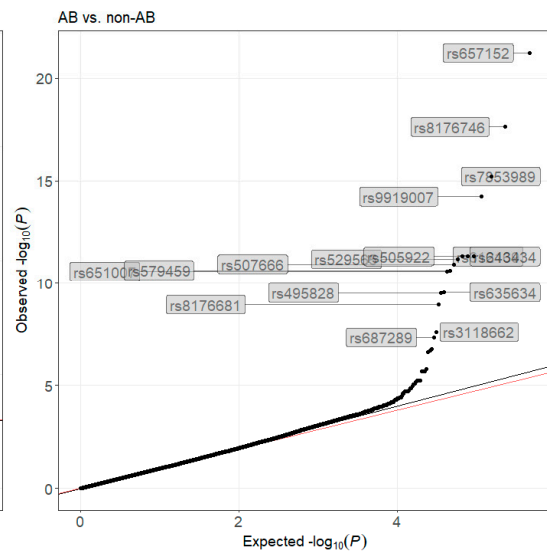
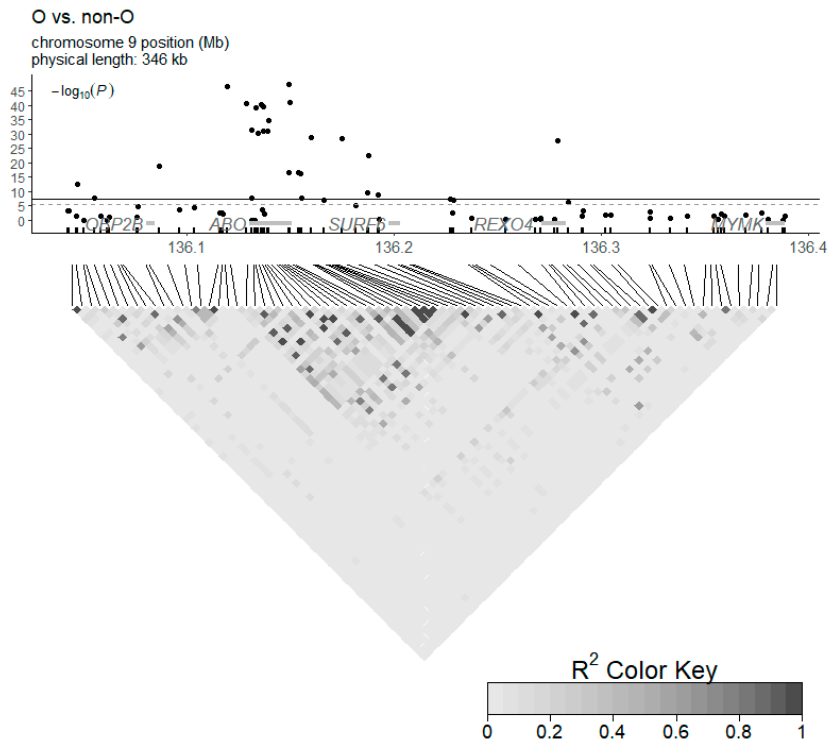
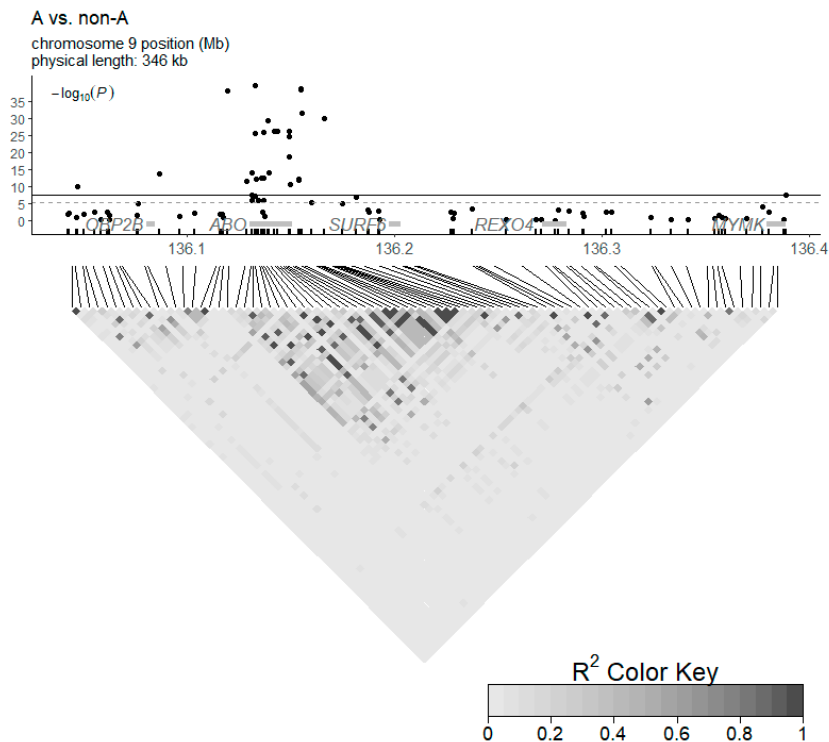


Figure S4

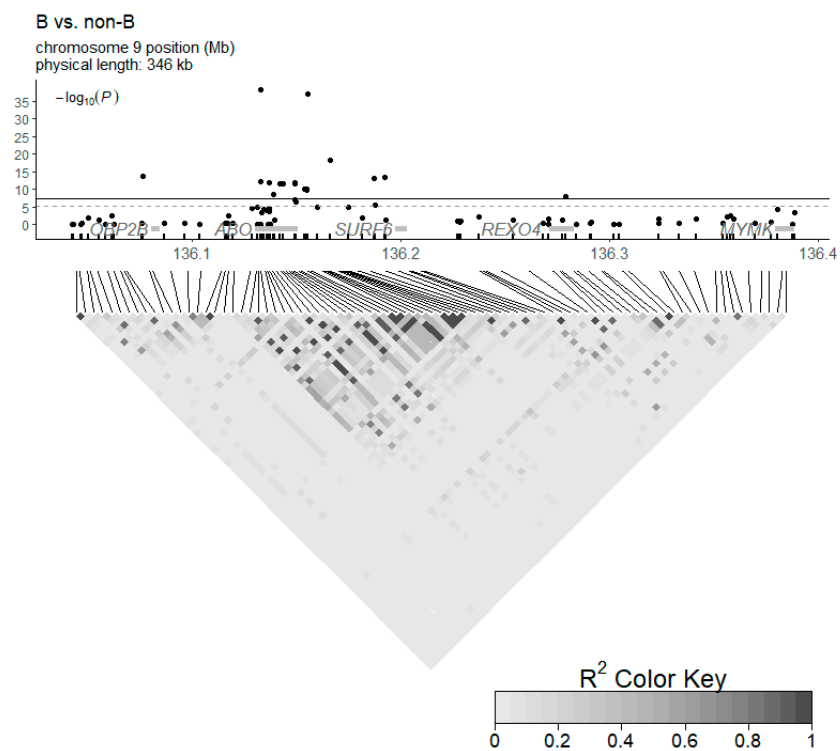
A.



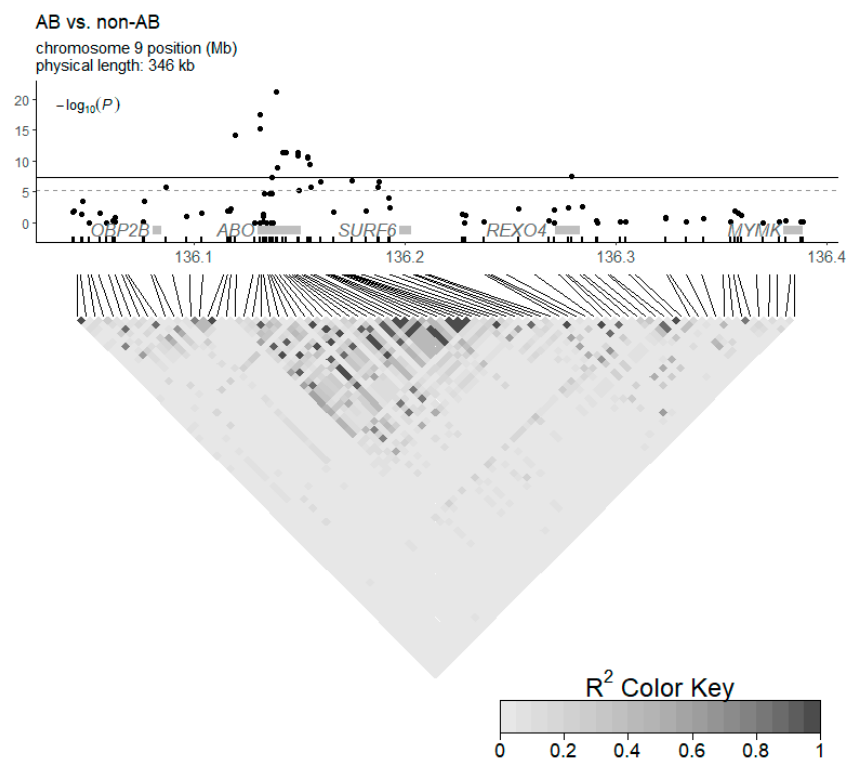
B.



C.

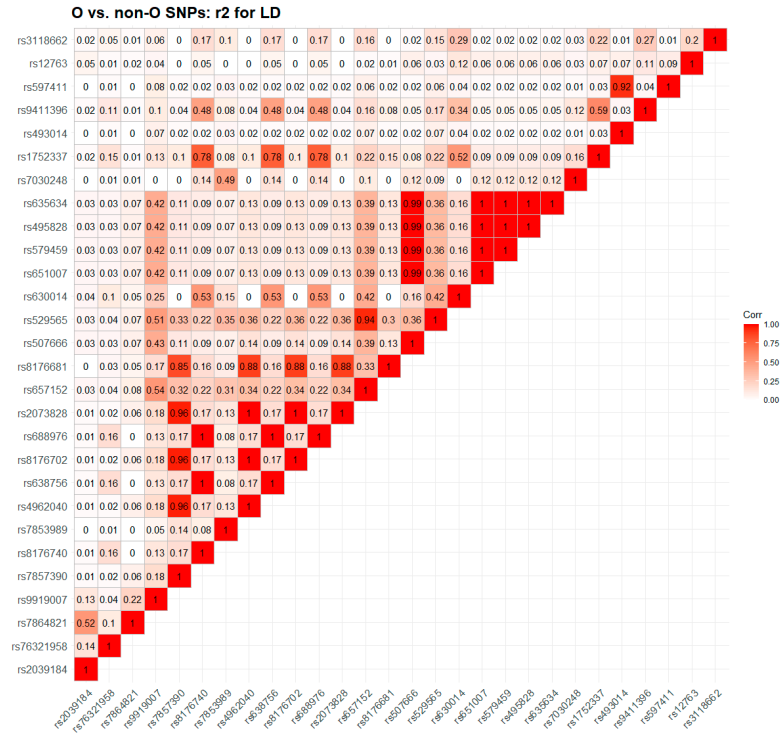


D.

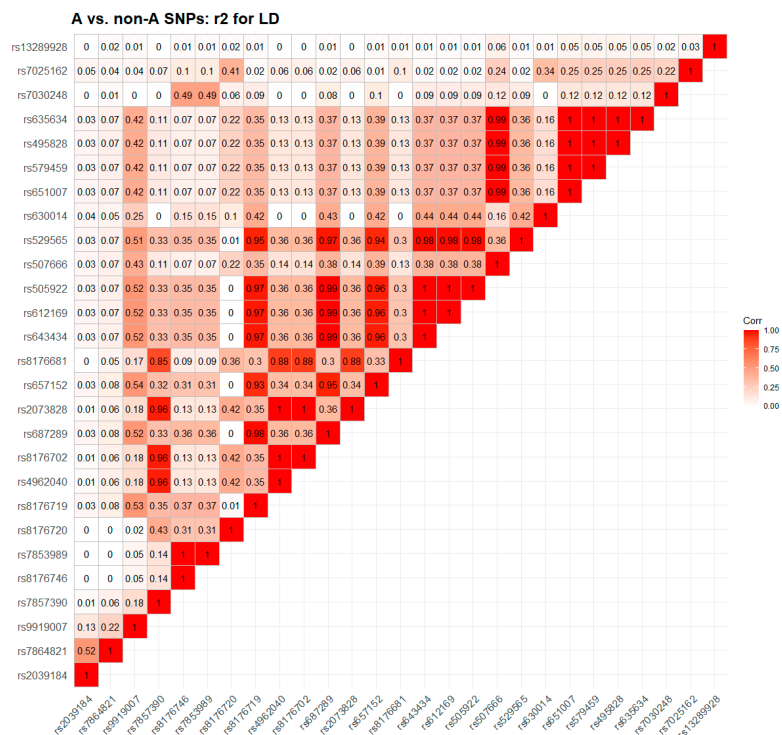


**Figure S5**

**A.**

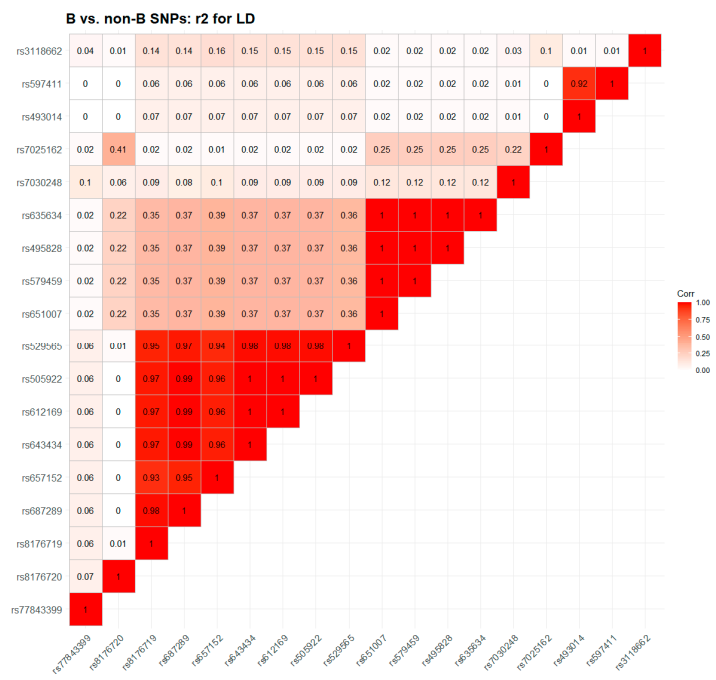


**B.**





C.



D.

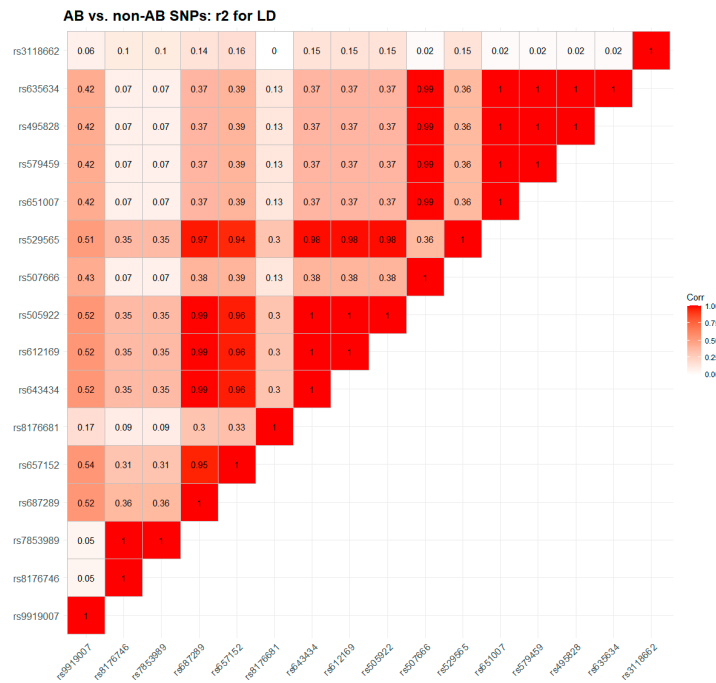


Figure S6

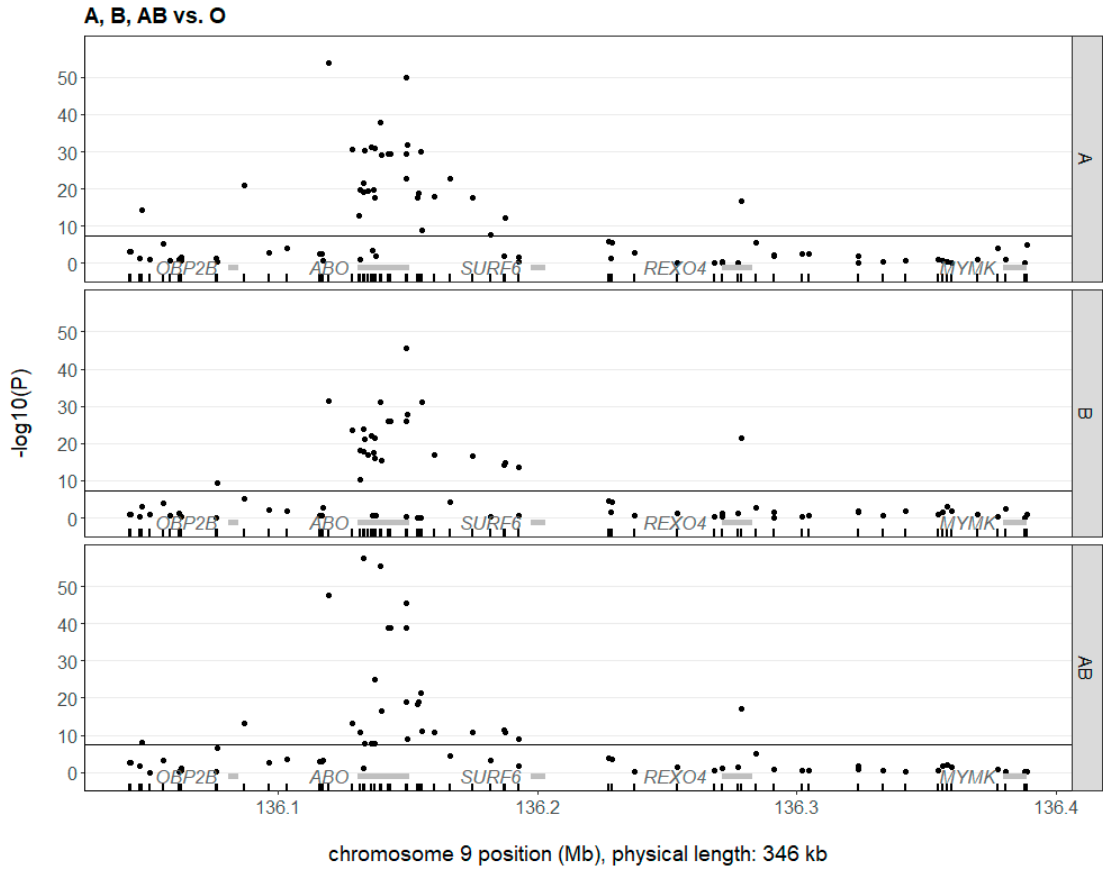
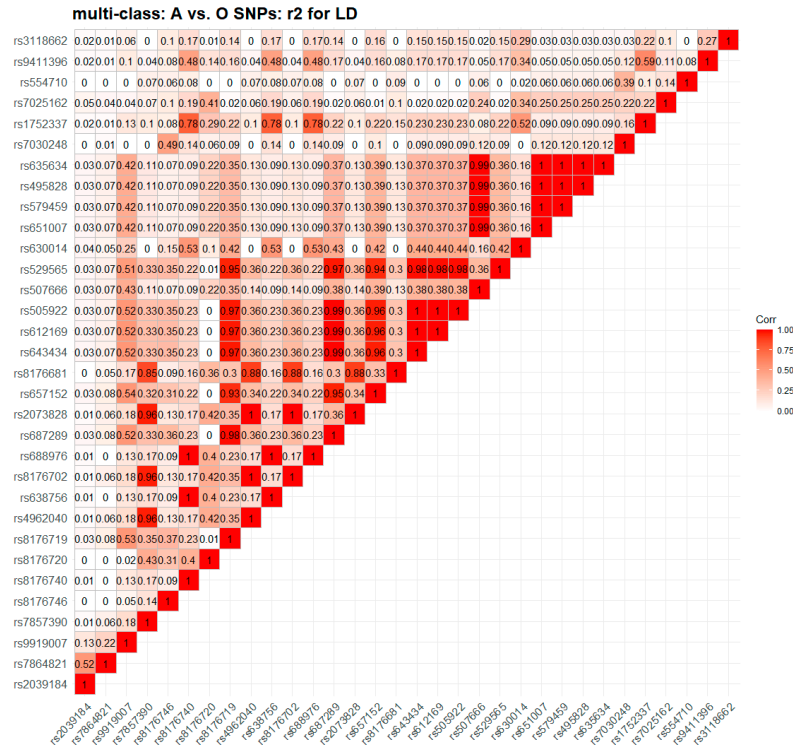
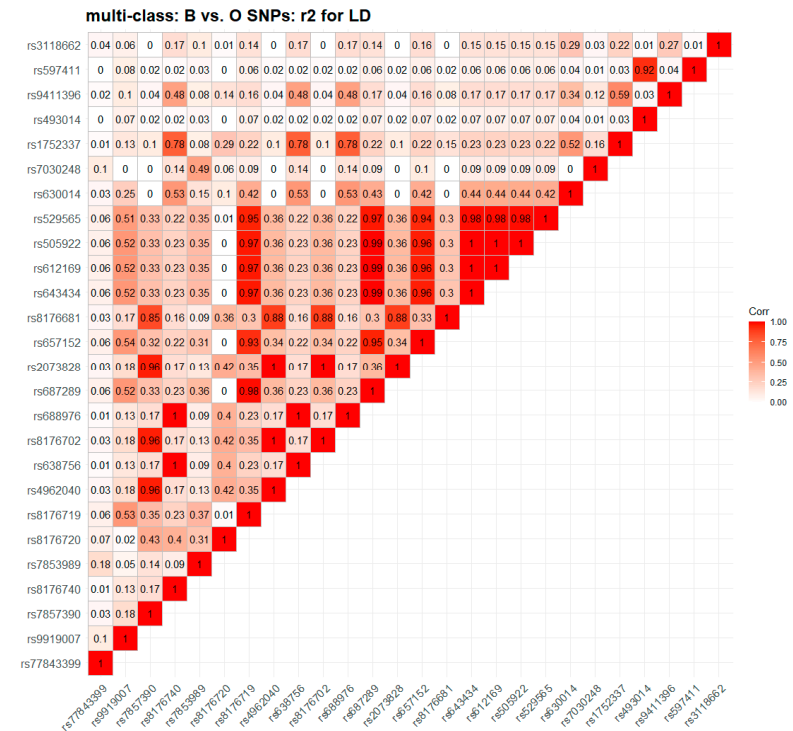


Figure S7

A.



B.



C.

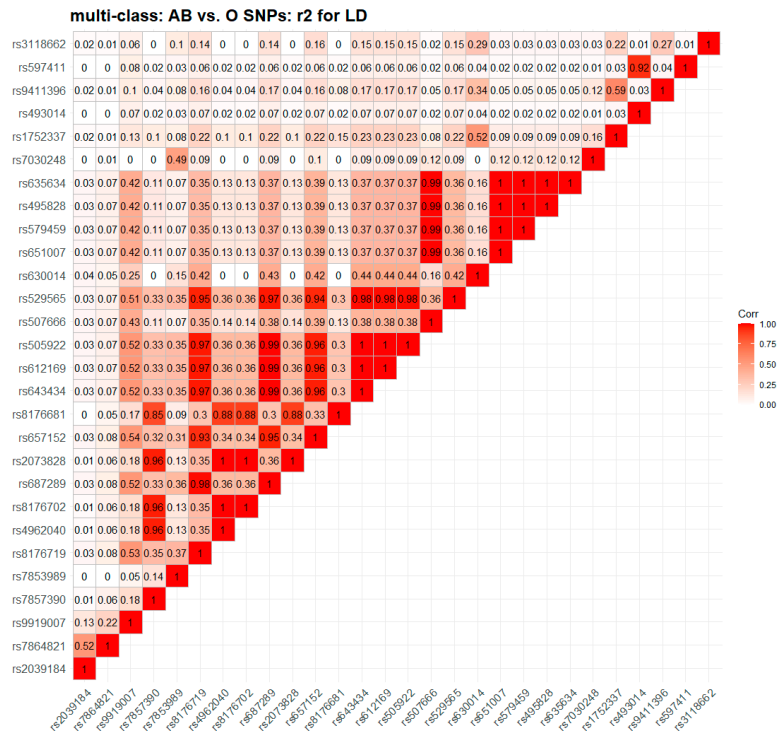
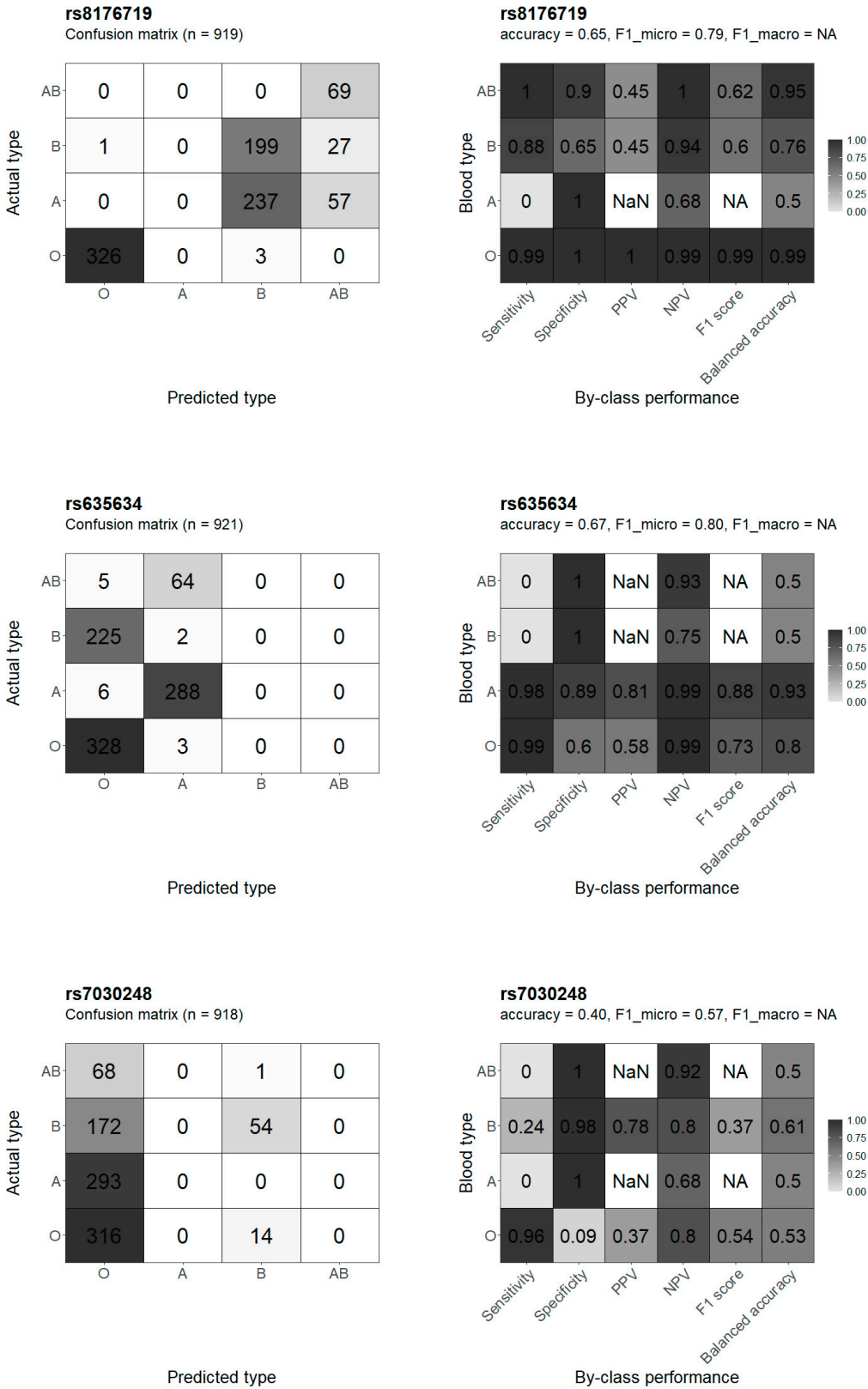


Figure S8

A.



B.

**rs8176719 + rs635634**

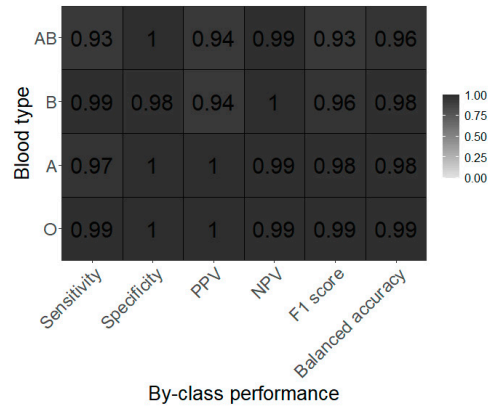
Confusion matrix (n = 919)

Actual type	AB	B	A	O
	0	0	5	64
	1	1	224	1
	0	285	6	3
	326	0	3	0
	O	A	B	AB

Predicted type

**rs8176719 + rs635634**

accuracy = 0.98, F1\_micro = 0.99, F1\_macro = 0.97



By-class performance

**rs8176719 + rs7030248**

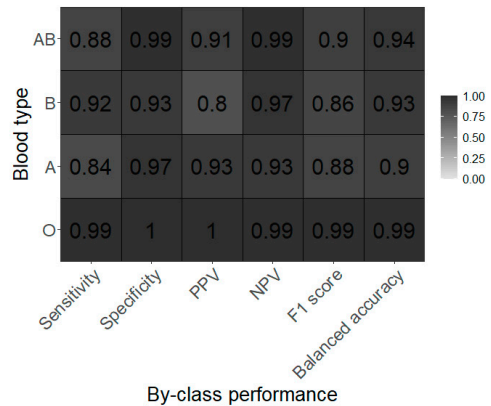
Confusion matrix (n = 916)

Actual type	AB	B	A	O
	0	7	1	61
	1	11	209	5
	0	245	47	1
	325	0	3	0
	O	A	B	AB

Predicted type

**rs8176719 + rs7030248**

accuracy = 0.92, F1\_micro = 0.96, F1\_macro = 0.91



By-class performance

**rs635634 + rs7030248**

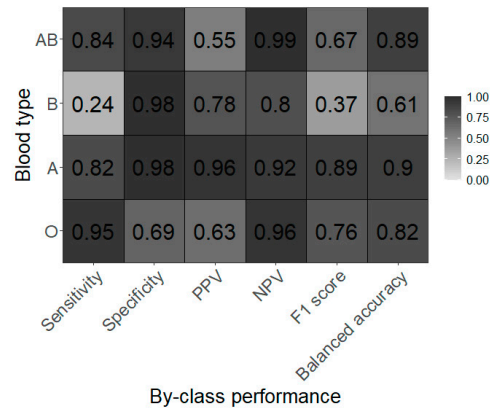
Confusion matrix (n = 918)

Actual type	AB	B	A	O
	4	6	1	58
	170	1	54	1
	6	241	0	46
	313	3	14	0
	O	A	B	AB

Predicted type

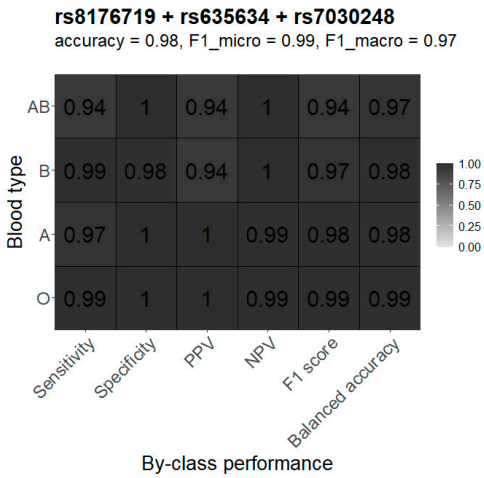
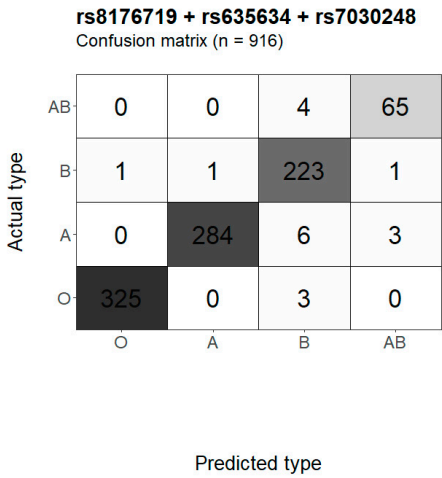
**rs635634 + rs7030248**

accuracy = 0.73, F1\_micro = 0.84, F1\_macro = 0.72

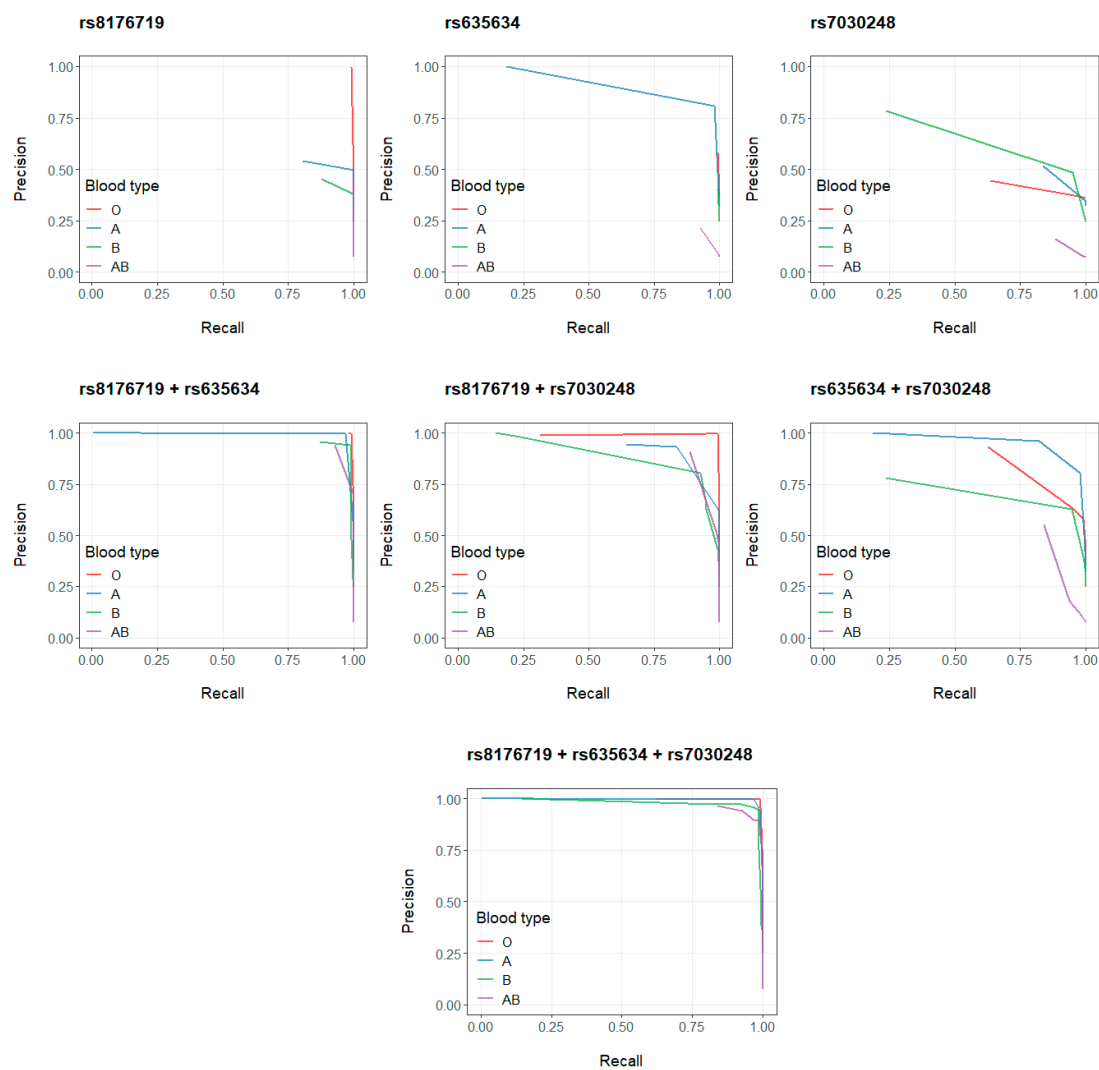


By-class performance

C.



**Figure S9**





**Figure S10**



## Supplementary figure legends

Figure S1. Schematic diagram for the study design of GWAS on ABO blood types in a Chinese population. QC: quality control; PCs: principal components.

Figure S2. Manhattan plots for the binary (2-level) outcomes: O vs. non-O (A), A vs. non-A (B), B vs. non-B (C) and AB vs. non-AB (D) in the cohort. Genome-wide hit SNPs (above the horizontal line) are annotated.

Figure S3. Quantile-quantile plots for the binary (2-level) outcomes: O vs. non-O (A), A vs. non-A (B), B vs. non-B (C) and AB vs. non-AB (D) in the cohort. Genome-wide hit SNPs are annotated. A line of  $y = x$  (black) and a line of  $y = \lambda_{median}x$  (red, the inflation factor  $\lambda_{median}$  are 1.008, 1.023, 1.013 and 0.952 respectively) are displayed.

Figure S4. Regional association plots for the binary (2-level) outcomes: O vs. non-O (A), A vs. non-A (B), B vs. non-B (C) and AB vs. non-AB (D) in the cohort. Genes are annotated as grey rectangles.

Figure S5. Linkage analysis ( $r^2$ ) between ABO-associated hit SNPs for the binary (2-level) outcomes: O vs. non-O (A), A vs. non-A (B), B vs. non-B (C) and AB vs. non-AB (D) in the Chinese (CHB + CHS) from the 1000 Genomes (<https://www.internationalgenome.org/>).

Figure S6. Regional association plots for the quaternary (4-level, reference level of type O) outcomes of A vs. O (upper), B vs. O (middle) and AB vs. O (lower) in the cohort. Genes are annotated as grey rectangles.

Figure S7. Linkage analysis ( $r^2$ ) between ABO-associated hit SNPs for the quaternary (4-level, reference level of type O) outcomes: A vs. O (A), B vs. O (B) and AB vs. O (C) in the Chinese (CHB + CHS) from the 1000 Genomes (<https://www.internationalgenome.org/>).

Figure S8. Confusion matrix (left) and classification performance parameters (right) for the prediction of ABO blood types based on *rs8176719*, *rs635634*, *rs7030248* (A), and their combinations (B and C). Accuracy,  $F1_{micro}$ , and  $F1_{macro}$  evaluate the overall multi-class performances; the color shaded tiles evaluate the by-blood-type binary performances.  $F1_{micro}$  and  $F1_{macro}$  are the F1

scores designed for multi-class classification obtained by micro-averaging (weighted by class frequency) or macro-averaging (weighting all classes equally).

Figure S9. By-blood-type precision-recall plots for *rs8176719*, *rs635634*, *rs7030248*, and their combinations. Note: for precision-recall plots, scenario in which all individuals are predicted for the “negative” class results in 0/0 for precision and 0 for recall, thus unable to plot such data points (leading to the incomplete portion of plot); consequently, AUC (area under the curve) is not calculated for the precision-recall plots.

Figure S10. Allele frequency of common hit SNPs in the multinomial models from this study among the globe and five super-ancestries from the 1000 Genomes (<https://www.internationalgenome.org/>).

## Supplementary tables

**Table S1. Summary of the general and by-type baseline characteristics**

	<b>All</b>	<b>O</b>	<b>A</b>	<b>B</b>	<b>AB</b>
<i>n</i>	1008	356	322	253	77
%	100	35.3	31.9	25.1	7.7
sex <sub>female</sub> (%)	569(56.4)	194(54.5)	187(58.1)	142(56.1)	46(59.7)
age (years)					
- median	46	46	47	47	44
- IQR	39-52	39-52	39-53	39-52	36-52
- range	30-79	30-72	30-78	30-79	30-77
height (cm)					
- median	158	159	158	158	158
- IQR	153-164	154-164	153-164	154-164	152-166
- range	124-182	140-180	124-178	138-182	146-178
weight (kg)					
- median	59.8	60.0	59.8	59.2	58.6
- IQR	53.1-67.6	53.8-67.0	54.4-67.5	52.3-67.9	51.8-67.6
- range	35.5-111.2	37.2-111.2	39.5-101.4	35.5-106.1	43.7-90.0

Note: IQR - inter-quartile range.

**Table S2. Distribution of ABO blood types across Asian ethnicities**

Ethnicity	O (%)	A (%)	B (%)	AB (%)
Chinese (this study)	36	32	25	7
Chinese (Canton)	46	23	25	6
Chinese (Peking)	29	27	32	13
Japanese	30	38	22	10
Korean	28	32	31	10
Thai	37	22	33	8
Asian (in U.S.A - general)	40	28	27	5

Note: frequency of ABO blood types in ethnicities other than this study were retrieved from Wikipedia ([https://en.wikipedia.org/wiki/Blood\\_type\\_distribution\\_by\\_country#cite\\_note-47](https://en.wikipedia.org/wiki/Blood_type_distribution_by_country#cite_note-47), last accessed on November 2, 2020), which was referenced to “Racial and ethnic distribution of ABO blood types” curated by Bloodbook.com (<http://www.bloodbook.com/world-abo.html>, last accessed on November 2, 2020).

**Table S10. Frequency summary of individuals with the defined set of SNPs ( $n = 916$ )**

count	percentage (%)	rs8176719	rs635634	rs7030248
205	22.4	0	0	0
104	11.4	0	0	1
14	1.5	0	0	2
3	0.3	0	1	0
14	1.5	1	0	0
160	17.5	1	0	1
33	3.6	1	0	2
185	20.2	1	1	0
45	4.9	1	1	1
1	0.1	1	2	0
1	0.1	2	0	0
7	0.8	2	0	1
22	2.4	2	0	2
8	0.9	2	1	0
60	6.6	2	1	1
54	5.9	2	2	0

Note: 916 out of the 921 individuals subjected to GWAS analysis have all these three SNPs successfully genotyped.

**Table S11. Summary of individuals with wrongly predicted ABO blood types ( $n = 19$ )**

count	rs8176719	rs635634	rs7030248	blood-typed	predicted
4	1	0	0	A	B
3	2	0	1	AB	B
3	1	0	1	O	B
2	1	0	1	A	B
2	2	1	0	A	AB
1	0	0	1	B	O
1	2	1	1	B	AB
1	1	1	0	B	A
1	2	0	2	AB	B
1	2	1	1	A	AB

**Table S3. Summary of GWAS on the binary trait of O vs. non-O blood types**

SNP	Chr.(position)	genomic region	ra(%)	ma(%)	allele zygosity (%)	-log <sub>10</sub> (P)	OR
<b><i>O vs. non-O</i></b>							
<i>rs529565</i> <sup>#</sup>	9(136149500)	ABO(intron)	C(59.4)	T(40.6)	CC(35.2),CT(48.4),TT(16.4)	47.5	0.0004
<i>rs9919007</i> <sup>#</sup>	9(136119527)	OBP2B*ABO	T(68.0)	C(32.0)	TT(46.8),TC(42.5),CC(10.7)	46.6	0.01
<i>rs630014</i>	9(136149722)	ABO(intron)	A(62.2)	G(37.8)	AA(37.9),AG(48.6),GG(13.5)	40.9	10.4
<i>rs7857390</i>	9(136128546)	ABO(3'-UTR)	A(66.4)	G(33.6)	AA(43.8),AG(45.0),GG(11.1)	40.5	7.5
<i>rs8176702</i>	9(136136146)	ABO(intron)	A(66.8)	G(33.2)	AA(44.7),AG(44.2),GG(11.1)	40.2	7.1
<i>rs2073828</i>	9(136137140)	ABO(intron)	A(66.4)	G(33.6)	AA(44.0),AG(45.0),GG(11.1)	39.6	7.0
<i>rs4962040</i> <sup>#</sup>	9(136133531)	ABO(intron)	G(66.9)	A(33.1)	GG(44.3),GA(45.2),AA(10.5)	39.2	7.0
<i>rs8176681</i>	9(136139754)	ABO(intron)	C(66.3)	T(33.7)	CC(43.8),CT(44.8),TT(11.4)	34.6	5.2
<i>rs8176740</i> <sup>#</sup>	9(136131472)	ABO(exon, F216I)	T(74.3)	A(25.7)	TT(54.6),TA(39.5),AA(5.9)	31.4	4.9
<i>rs657152</i>	9(136139265)	ABO(intron)	A(61.0)	C(39.0)	AA(37.1),AC(47.8),CC(15.1)	31.1	0.0003
<i>rs688976</i> <sup>#</sup>	9(136136770)	ABO(exon, V36F)	A(74.0)	C(25.9)	AA(54.3),AC(39.5),CC(6.2)	31.0	4.8
<i>rs638756</i>	9(136134472)	ABO(intron)	C(73.6)	A(26.4)	CC(53.5),CA(40.2),AA(6.3)	30.4	4.7
<i>rs1752337</i> <sup>#</sup>	9(136160228)	ABO*SURF6	G(72.3)	T(27.7)	GG(52.2),GT(40.2),TT(7.6)	28.7	4.2
<i>mf09136175051</i>	9(136175051)	ABO*SURF6	T(72.4)	C(27.6)	TT(52.2),TC(40.4),CC(7.4)	28.5	4.2
<i>rs3118662</i> <sup>#</sup>	9(136278870)	REXO4(intron)	G(51.6)	A(48.4)	GG(26.3),GA(50.7),AA(23)	27.6	3.8
<i>rs9411396</i> <sup>#</sup>	9(136187652)	ABO*SURF6	G(73.7)	T(26.3)	GG(53.8),GT(39.8),TT(6.4)	22.6	3.4
<i>rs7864821</i> <sup>#</sup>	9(136086916)	OBP2B(intron)	C(60.6)	T(39.4)	CC(36.8),CT(47.7),TT(15.5)	18.9	0.4
<i>rs579459</i>	9(136154168)	ABO*SURF6	C(77.6)	T(22.4)	CC(60.8),CT(33.7),TT(5.5)	16.8	0.007
<i>rs651007</i> <sup>#</sup>	9(136153875)	ABO*SURF6	T(77.4)	C(22.6)	TT(60.7),TC(33.5),CC(5.8)	16.7	0.007
<i>rs507666</i>	9(136149399)	ABO(intron)	A(77.8)	G(22.2)	AA(61.0),AG(33.6),GG(5.4)	16.6	0.007



<i>rs495828</i>	9(136154867)	ABO*SURF6	T(77.9)	G(22.1)	TT(61.5),TG(32.8),GG(5.7)	16.3	0.007
<i>rs635634</i>	9(136155000)	ABO*SURF6	T(77.9)	C(22.1)	TT(61.4),TC(32.9),CC(5.7)	16.3	0.007
<i>rs2039184<sup>#</sup></i>	9(136047393)	RALGDS*OBP2B	G(59.2)	A(40.8)	GG(35.3),GA(48.0),AA(16.7)	12.5	0.4
<i>rs493014<sup>#</sup></i>	9(136187163)	ABO*SURF6	G(90.7)	T(9.3)	GG(81.9),GT(17.5),TT(0.6)	9.4	0.2
<i>rs597411</i>	9(136192624)	ABO*SURF6	T(90.5)	G(9.5)	TT(81.7),TG(17.7),GG(0.6)	8.7	0.2
<i>rs7853989<sup>#</sup></i>	9(136131592)	ABO(exon, R176G)	C(82.6)	G(17.4)	CC(66.4),CG(32.4),GG(1.2)	7.8	0.003
<i>rs7030248<sup>#</sup></i>	9(136155359)	ABO*SURF6	A(71.9)	G(28.1)	AA(51.0),AG(41.8),GG(7.2)	7.6	0.5
<i>rs76321958<sup>#</sup></i>	9(136055528)	RALGDS*OBP2B	C(84.6)	G(15.4)	CC(71.7),CG(25.8),GG(2.5)	7.5	2.1
<i>rs12763<sup>#</sup></i>	9(136227260)	SURF2(exon,G213S)	A(67.7)	G(32.3)	AA(46.1),AG(43.2),GG(10.7)	7.3	0.5

Note: 1. coordinate position on chromosomes is based on GRCh37.

2. \* in “genomic region” annotates intergenic region, e.g. OBP2B\*ABO annotates an intergenic locus downstream of OBP2B and upstream of ABO.

3. ra - reference allele (major allele); ma - minor allele; OR, odds ratio.

4. the above notes also apply to Table S4 to S9.

5. <sup>#</sup> suffix in SNP annotates the subset of hit variants selected after LD screening ( $r^2 < 0.8$ ) and to be evaluated for binary classification performances in Table 1.

For example, *rs529565* is the top ranked SNP for O vs. non-O and selected for binary classification evaluation (thus annotated with the suffix as *rs529565<sup>#</sup>*). With linkage analysis from Figure S5A, those hit variants with high linkage ( $r^2 \geq 0.8$ ) to *rs529565* would not be selected for binary classification evaluation (thus no <sup>#</sup> suffix: *rs657152* with  $r^2 = 0.94$ ). Iterating the above analysis for all hit SNPs leads to the selection of <sup>#</sup> suffix genetic variants to be evaluated in Table 1. This note also applies to Table S4 to S6.

**Table S4. Summary of GWAS on the binary trait of A vs. non-A blood types**

SNP	Chr.(position)	genomic region	ra(%)	ma(%)	allele zygotity (%)	-log <sub>10</sub> (P)	OR
<b><u>A vs. non-A</u></b>							
<i>rs8176720</i> <sup>#</sup>	9(136132873)	ABO(exon, silent)	C(56.1)	T(43.9)	CC(30.4),CT(51.5),TT(18.1)	39.8	0.1
<i>rs635634</i> <sup>#</sup>	9(136155000)	ABO*SURF6	T(77.9)	C(22.1)	TT(61.5),TC(32.8),CC(5.7)	38.7	363.4
<i>rs495828</i>	9(136154867)	ABO*SURF6	T(77.9)	G(22.1)	TT(61.5),TG(32.8),GG(5.7)	38.5	358.6
<i>rs9919007</i> <sup>#</sup>	9(136119527)	OBP2B*ABO	T(68.0)	C(32.0)	TT(46.8),TC(42.6),CC(10.7)	38.1	6.4
<i>rs7030248</i> <sup>#</sup>	9(136155359)	ABO*SURF6	A(71.9)	G(28.1)	AA(51.0),AG(41.8),GG(7.2)	31.6	0.1
<i>rs7025162</i> <sup>#</sup>	9(136166346)	ABO*SURF6	T(60.2)	C(39.8)	TT(35.4),TC(49.8),CC(14.8)	30.0	4.6
<i>rs657152</i>	9(136139265)	ABO(intron)	A(61.0)	C(39.0)	AA(37.1),AC(47.8),CC(15.1)	29.4	4.2
<i>rs505922</i>	9(136149229)	ABO(intron)	C(59.3)	T(40.7)	CC(35),CT(48.7),TT(16.4)	26.2	3.6
<i>rs612169</i>	9(136143442)	ABO(intron)	G(59.2)	A(40.8)	GG(34.9),GA(48.8),AA(16.4)	26.2	3.6
<i>rs643434</i>	9(136142355)	ABO(intron)	A(59.2)	G(40.8)	AA(34.9),AG(48.8),GG(16.4)	26.2	3.6
<i>rs687289</i>	9(136137106)	ABO(intron)	A(59.5)	G(40.5)	AA(35.4),AG(48.2),GG(16.4)	26.0	3.6
<i>rs8176719</i> <sup>#</sup>	9(136132908)	ABO(exon, insert)	TC(59.4)	T(40.6)	II(35.0),ID(48.6),DD(16.4)	25.5	3.6
<i>rs529565</i>	9(136149500)	ABO(intron)	C(59.4)	T(40.6)	CC(35.2),CT(48.4),TT(16.4)	24.6	3.4
<i>rs507666</i>	9(136149399)	ABO(intron)	A(77.8)	G(22.2)	AA(61),AG(33.6),GG(5.4)	18.8	1302.9
<i>rs8176681</i>	9(136139754)	ABO(intron)	C(66.3)	T(33.7)	CC(43.8),CT(44.8),TT(11.4)	14.1	0.4
<i>rs7853989</i> <sup>#</sup>	9(136131592)	ABO(exon, R176G)	C(82.6)	G(17.4)	CC(66.4),CG(32.4),GG(1.2)	13.9	0.02
<i>rs7864821</i> <sup>#</sup>	9(136086916)	OBP2B(intron)	C(60.6)	T(39.4)	CC(36.8),CT(47.7),TT(15.5)	13.6	2.3
<i>rs8176702</i>	9(136136146)	ABO(intron)	A(66.8)	G(33.2)	AA(44.7),AG(44.2),GG(11.1)	12.5	0.4
<i>rs2073828</i>	9(136137140)	ABO(intron)	A(66.4)	G(33.6)	AA(44.0),AG(45.0),GG(11.0)	12.4	0.4
<i>rs4962040</i>	9(136133531)	ABO(intron)	G(66.9)	A(33.1)	GG(44.3),GA(45.2),AA(10.5)	12.1	0.4
<i>rs651007</i>	9(136153875)	ABO*SURF6	T(77.4)	C(22.6)	TT(60.7),TC(33.5),CC(5.8)	12.0	2330.0

<i>rs579459</i>	9(136154168)	ABO*SURF6	C(77.6)	T(22.4)	CC(60.8),CT(33.7),TT(5.5)	11.7	2471.6
<i>rs7857390</i> <sup>#</sup>	9(136128546)	ABO(3'-UTR)	A(66.4)	G(33.6)	AA(43.8),AG(45.1),GG(11.1)	11.6	0.4
<i>rs630014</i> <sup>#</sup>	9(136149722)	ABO(intron)	A(62.2)	G(37.8)	AA(37.9),AG(48.6),GG(13.5)	10.7	0.4
<i>rs2039184</i> <sup>#</sup>	9(136047393)	RALGDS*OBP2B	G(59.2)	A(40.8)	GG(35.3),GA(48.0),AA(16.8)	9.8	2.0
<i>rs13289928</i> <sup>#</sup>	9(136388702)	MYMK(intron)	G(72.0)	A(28.0)	GG(52.2),GA(39.6),AA(8.2)	7.3	1.8
<i>rs8176746</i>	9(136131322)	ABO(exon, L266M)	T(82.4)	G(17.6)	TT(67.3),TG(30.3),GG(2.4)	7.3	0.004

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**Table S5. Summary of GWAS on the binary trait of B vs. non-B blood types**

SNP	Chr.(position)	genomic region	ra(%)	ma(%)	allele zygosity (%)	-log <sub>10</sub> (P)	OR
<b><i>B vs. non-B</i></b>							
<i>rs8176720</i> <sup>#</sup>	9(136132873)	ABO(exon, silent)	C(56.1)	T(43.9)	CC(30.4),CT(51.5),TT(18.1)	38.3	8.5
<i>rs7030248</i> <sup>#</sup>	9(136155359)	ABO*SURF6	A(71.9)	G(28.1)	AA(51.0),AG(41.8),GG(7.2)	37.0	16.1
<i>rs7025162</i> <sup>#</sup>	9(136166346)	ABO*SURF6	T(60.2)	C(39.8)	TT(35.4),TC(49.8),CC(14.8)	18.2	0.3
<i>rs77843399</i> <sup>#</sup>	9(136076392)	RALGDS*OBP2B	T(95.8)	C(4.2)	TT(91.7),TC(8.2),CC(0.1)	13.8	7.3
<i>rs493014</i> <sup>#</sup>	9(136187163)	ABO*SURF6	G(90.7)	T(9.3)	GG(81.9),GT(17.5),TT(0.6)	13.3	3.8
<i>rs597411</i>	9(136192624)	ABO*SURF6	T(90.5)	G(9.5)	TT(81.7),TG(17.7),GG(0.6)	13.3	3.8
<i>rs8176719</i> <sup>#</sup>	9(136132908)	ABO(exon, insert)	TC(59.4)	T(40.6)	II(35.1),ID(48.6),DD(16.3)	12.2	2.3
<i>rs687289</i>	9(136137106)	ABO(intron)	A(59.5)	G(40.5)	AA(35.4),AG(48.2),GG(16.4)	11.9	2.3
<i>rs529565</i>	9(136149500)	ABO(intron)	C(59.4)	T(40.6)	CC(35.2),CT(48.4),TT(16.4)	11.8	2.3
<i>rs505922</i>	9(136149229)	ABO(intron)	C(59.3)	T(40.7)	CC(35.0),CT(48.6),TT(16.4)	11.6	2.2
<i>rs612169</i>	9(136143442)	ABO(intron)	G(59.2)	A(40.8)	GG(34.8),GA(48.8),AA(16.4)	11.6	2.2
<i>rs643434</i>	9(136142355)	ABO(intron)	A(59.2)	G(40.8)	AA(34.8),AG(48.8),GG(16.4)	11.6	2.2
<i>rs579459</i>	9(136154168)	ABO*SURF6	C(77.6)	T(22.4)	CC(60.8),CT(33.7),TT(5.5)	10.2	0.009
<i>rs651007</i>	9(136153875)	ABO*SURF6	T(77.4)	C(22.6)	TT(60.7),TC(33.5),CC(5.8)	10.2	0.009
<i>rs635634</i> <sup>#</sup>	9(136155000)	ABO*SURF6	T(77.9)	C(22.1)	TT(61.4),TC(32.9),CC(5.7)	10.0	0.01
<i>rs495828</i>	9(136154867)	ABO*SURF6	T(77.9)	G(22.1)	TT(61.5),TG(32.8),GG(5.7)	9.9	0.01
<i>rs657152</i>	9(136139265)	ABO(intron)	A(61.0)	C(39.0)	AA(37.1),AC(47.8),CC(15.1)	8.7	2.0
<i>rs3118662</i> <sup>#</sup>	9(136278870)	REXO4(intron)	G(51.6)	A(48.4)	GG(26.3),GA(50.7),AA(23.0)	8.1	0.5

**Table S6. Summary of GWAS on the binary trait of AB vs. non-AB blood types**

SNP	Chr.(position)	genomic region	ra(%)	ma(%)	allele zygosity (%)	-log <sub>10</sub> (P)	OR
<b><u>AB vs. non-AB</u></b>							
<i>rs657152</i>	9(136139265)	ABO(intron)	A(61.0)	C(39.0)	AA(37.1),AC(47.8),CC(15.1)	21.2	48.9
<i>rs8176746</i> <sup>#</sup>	9(136131322)	ABO(exon, L266M)	T(82.4)	G(17.6)	TT(67.3),TG(30.3),GG(2.4)	17.6	8.3
<i>rs7853989</i>	9(136131592)	ABO(exon, R176G)	C(82.6)	G(17.4)	CC(66.4),CG(32.4),GG(1.2)	15.2	13.0
<i>rs9919007</i> <sup>#</sup>	9(136119527)	OBP2B*ABO	T(68.0)	C(32.0)	TT(46.8),TC(42.6),CC(10.6)	14.2	4.8
<i>rs505922</i>	9(136149229)	ABO(intron)	C(59.3)	T(40.7)	CC(35.0),CT(48.7),TT(16.4)	11.3	199.9
<i>rs612169</i>	9(136143442)	ABO(intron)	G(59.2)	A(40.8)	GG(34.9),GA(48.8),AA(16.4)	11.3	199.9
<i>rs643434</i>	9(136142355)	ABO(intron)	A(59.2)	G(40.8)	AA(34.9),AG(48.8),GG(16.4)	11.3	199.9
<i>rs529565</i>	9(136149500)	ABO(intron)	C(59.4)	T(40.6)	CC(35.2),CT(48.4),TT(16.4)	11.2	208.1
<i>rs507666</i> <sup>#</sup>	9(136149399)	ABO(intron)	A(77.8)	G(22.2)	AA(61),AG(33.6),GG(5.4)	10.9	3.7
<i>rs579459</i>	9(136154168)	ABO*SURF6	C(77.6)	T(22.4)	CC(60.8),CT(33.7),TT(5.5)	10.6	3.5
<i>rs651007</i>	9(136153875)	ABO*SURF6	T(77.4)	C(22.6)	TT(60.7),TC(33.5),CC(5.8)	10.6	3.5
<i>rs495828</i>	9(136154867)	ABO*SURF6	T(77.9)	G(22.1)	TT(61.5),TG(32.8),GG(5.7)	9.5	3.2
<i>rs635634</i>	9(136155000)	ABO*SURF6	T(77.9)	C(22.1)	TT(61.4),TC(32.9),CC(5.7)	9.5	3.2
<i>rs8176681</i> <sup>#</sup>	9(136139754)	ABO(intron)	C(66.3)	T(33.7)	CC(43.8),CT(44.8),TT(11.4)	8.9	0.09
<i>rs3118662</i> <sup>#</sup>	9(136278870)	REXO4(intron)	G(51.6)	A(48.4)	GG(26.3),GA(50.7),AA(23.0)	7.6	0.3
<i>rs687289</i> <sup>#</sup>	9(136137106)	ABO(intron)	A(59.5)	G(40.5)	AA(35.4),AG(48.2),GG(16.4)	7.3	475.5

**Table S7. Summary of GWAS on the quaternary trait of A vs. O blood types**

SNP	Chr.(position)	genomic region	ra(%)	ma(%)	allele zygosity (%)	-log <sub>10</sub> (P)	OR
<b><u>A vs. O</u></b>							
<i>rs9919007</i>	9(136119527)	OBP2B*ABO	T(68.0)	C(32.0)	TT(46.8),TC(42.6),CC(10.7)	53.9	182.6
<i>rs529565</i>	9(136149500)	ABO(intron)	C(59.4)	T(40.6)	CC(35.2),CT(48.4),TT(16.4)	50.0	3.0×10 <sup>3</sup>
<i>rs657152</i>	9(136139265)	ABO(intron)	A(61.0)	C(39.0)	AA(37.1),AC(47.8),CC(15.1)	37.8	6.5×10 <sup>3</sup>
<i>rs630014</i>	9(136149722)	ABO(intron)	A(62.2)	G(37.8)	AA(37.9),AG(48.6),GG(13.5)	31.8	0.1
<i>rs8176702</i>	9(136136146)	ABO(intron)	A(66.8)	G(33.2)	AA(44.7),AG(44.2),GG(11.1)	31.1	0.1
<i>rs2073828</i>	9(136137140)	ABO(intron)	A(66.4)	G(33.6)	AA(44.0),AG(45.0),GG(11.0)	30.8	0.1
<i>rs7857390</i>	9(136128546)	ABO(3'-UTR)	A(66.4)	G(33.6)	AA(44.0),AG(45.0),GG(11.0)	30.7	0.1
<i>rs4962040</i>	9(136133531)	ABO(intron)	G(66.9)	A(33.1)	GG(44.3),GA(45.2),AA(10.5)	30.3	0.1
<i>rs635634</i>	9(136155000)	ABO*SURF6	T(77.9)	C(22.1)	TT(61.4),TC(32.9),CC(5.7)	30.1	1.1×10 <sup>4</sup>
<i>rs495828</i>	9(136154867)	ABO*SURF6	T(77.9)	G(22.1)	TT(61.4),TG(32.9),GG(5.7)	30.0	1.1×10 <sup>4</sup>
<i>rs505922</i>	9(136149229)	ABO(intron)	C(59.3)	T(40.7)	CC(35.0),CT(48.7),TT(16.4)	29.3	1.8×10 <sup>4</sup>
<i>rs612169</i>	9(136143442)	ABO(intron)	G(59.2)	A(40.8)	GG(34.9),GA(48.8),AA(16.4)	29.3	1.8×10 <sup>4</sup>
<i>rs643434</i>	9(136142355)	ABO(intron)	A(59.2)	G(40.8)	AA(34.9),AG(48.8),GG(16.4)	29.3	1.8×10 <sup>4</sup>
<i>rs8176681</i>	9(136139754)	ABO(intron)	C(66.3)	T(33.7)	CC(43.8),CT(44.8),TT(11.4)	29.2	0.2
<i>rs507666</i>	9(136149399)	ABO(intron)	A(77.8)	G(22.2)	AA(61.0),AG(33.6),GG(5.4)	22.9	4.8×10 <sup>4</sup>
<i>rs7025162</i>	9(136166346)	ABO*SURF6	T(60.2)	C(39.8)	TT(35.4),TC(49.8),CC(14.8)	22.7	4.3
<i>rs8176720</i>	9(136132873)	ABO(exon, silent)	C(56.1)	T(43.9)	CC(30.4),CT(51.5),TT(18.1)	21.5	0.2
<i>rs7864821</i>	9(136086916)	OBP2B(intron)	C(60.6)	T(39.4)	CC(36.8),CT(47.7),TT(15.5)	21.0	3.7
<i>rs8176740</i>	9(136131472)	ABO(exon, F216I)	T(74.3)	A(25.7)	TT(54.6),TA(39.5),AA(5.9)	19.9	0.2
<i>rs688976</i>	9(136136770)	ABO(exon, V36F)	A(74.1)	C(25.9)	AA(54.3),AC(39.5),CC(6.2)	19.6	0.2

<i>rs638756</i>	9(136134472)	ABO(intron)	C(73.6)	A(26.4)	CC(53.5),CA(40.2),AA(6.3)	19.4	0.2
<i>rs8176719</i>	9(136132908)	ABO(exon, insert)	TC(59.4)	T(40.6)	II(35.0),ID(48.6),DD(16.3)	19.1	$2.4 \times 10^7$
<i>rs579459</i>	9(136154168)	ABO*SURF6	C(77.6)	T(22.4)	CC(60.8),CT(33.7),TT(5.5)	18.8	$9.4 \times 10^4$
<i>rs1752337</i>	9(136160228)	ABO*SURF6	G(72.3)	T(27.7)	GG(52.2),GT(40.2),TT(7.6)	17.9	0.3
<i>mf09136175051</i>	9(136175051)	ABO*SURF6	T(72.4)	C(27.6)	TT(52.2),TC(40.4),CC(7.4)	17.7	0.3
<i>rs651007</i>	9(136153875)	ABO*SURF6	T(77.4)	C(22.6)	TT(60.7),TC(33.5),CC(5.8)	17.7	$1.2 \times 10^5$
<i>rs687289</i>	9(136137106)	ABO(intron)	A(59.5)	G(40.5)	AA(35.4),AG(48.2),GG(16.4)	17.6	$1.1 \times 10^5$
<i>rs3118662</i>	9(136278870)	REXO4(intron)	G(51.6)	A(48.4)	GG(26.3),GA(50.7),AA(23)	16.7	0.3
<i>rs2039184</i>	9(136047393)	RALGDS*OBP2B	G(59.2)	A(40.8)	GG(35.3),GA(48.0),AA(16.8)	14.4	2.7
<i>rs8176746</i>	9(136131322)	ABO(exon, L266M)	T(82.4)	G(17.6)	TT(67.3),TG(30.3),GG(2.4)	12.9	$9.7 \times 10^5$
<i>rs9411396</i>	9(136187652)	ABO*SURF6	G(73.7)	T(26.3)	GG(53.8),GT(39.8),TT(6.4)	12.2	0.4
<i>rs7030248</i>	9(136155359)	ABO*SURF6	A(71.9)	G(28.1)	AA(51.0),AG(41.8),GG(7.2)	9.0	0.3
<i>rs554710</i>	9(136181848)	ABO*SURF6	C(83.3)	T(16.7)	CC(69.5),CT(27.6),TT(2.9)	7.6	0.4

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**Table S8. Summary of GWAS on the quaternary trait of B vs. O blood types**

SNP	Chr.(position)	genomic region	ra(%)	ma(%)	allele zygosity (%)	-log <sub>10</sub> (P)	OR
<b><u>B vs. O</u></b>							
<i>rs529565</i>	9(136149500)	ABO(intron)	C(59.4)	T(40.6)	CC(35.2),CT(48.4),TT(16.4)	45.7	1.8×10 <sup>3</sup>
<i>rs9919007</i>	9(136119527)	OBP2B*ABO	T(68.0)	C(32.0)	TT(46.8),TC(42.6),CC(10.7)	31.5	41.1
<i>rs657152</i>	9(136139265)	ABO(intron)	A(61.0)	C(39.0)	AA(37.1),AC(47.8),CC(15.1)	31.1	2.2×10 <sup>3</sup>
<i>rs7030248</i>	9(136155359)	ABO*SURF6	A(71.9)	G(28.1)	AA(51.0),AG(41.8),GG(7.2)	31.0	17.3
<i>rs630014</i>	9(136149722)	ABO(intron)	A(62.2)	G(37.8)	AA(37.9),AG(48.6),GG(13.5)	27.9	0.1
<i>rs505922</i>	9(136149229)	ABO(intron)	C(59.3)	T(40.7)	CC(34.9),CT(48.8),TT(16.4)	26.1	8.9×10 <sup>3</sup>
<i>rs612169</i>	9(136143442)	ABO(intron)	G(59.2)	A(40.8)	GG(34.9),GA(48.8),AA(16.4)	26.1	8.9×10 <sup>3</sup>
<i>rs643434</i>	9(136142355)	ABO(intron)	A(59.2)	G(40.8)	AA(34.9),AG(48.8),GG(16.4)	26.1	8.9×10 <sup>3</sup>
<i>rs8176720</i>	9(136132873)	ABO(exon, silent)	C(56.1)	T(43.9)	CC(30.4),CT(51.5),TT(18.1)	23.7	6.2
<i>rs7857390</i>	9(136128546)	ABO(3'-UTR)	A(66.4)	G(33.6)	AA(43.8),AG(45),GG(11.1)	23.5	0.2
<i>rs8176702</i>	9(136136146)	ABO(intron)	A(66.8)	G(33.2)	AA(44.7),AG(44.2),GG(11.1)	22.2	0.2
<i>rs2073828</i>	9(136137140)	ABO(intron)	A(66.4)	G(33.6)	AA(44.0),AG(45.0),GG(11.0)	21.6	0.2
<i>rs3118662</i>	9(136278870)	REXO4(intron)	G(51.6)	A(48.4)	GG(26.3),GA(50.7),AA(23)	21.4	0.2
<i>rs4962040</i>	9(136133531)	ABO(intron)	G(66.9)	A(33.1)	GG(44.3),GA(45.2),AA(10.5)	21.2	0.2
<i>rs8176740</i>	9(136131472)	ABO(exon, F216I)	T(74.3)	A(25.7)	TT(54.6),TA(39.5),AA(5.9)	18.0	0.2
<i>rs8176719</i>	9(136132908)	ABO(exon, insert)	TC(59.4)	T(40.6)	II(35.1),ID(48.6),DD(16.3)	17.9	1.3×10 <sup>7</sup>
<i>rs688976</i>	9(136136770)	ABO(exon, V36F)	A(74.0)	C(25.9)	AA(54.3),AC(39.5),CC(6.2)	17.6	0.2
<i>rs638756</i>	9(136134472)	ABO(intron)	C(73.6)	A(26.4)	CC(53.5),CA(40.2),AA(6.3)	17.0	0.2
<i>rs1752337</i>	9(136160228)	ABO*SURF6	G(72.3)	T(27.7)	GG(52.2),GT(40.2),TT(7.6)	16.8	0.3
<i>mf09136175051</i>	9(136175051)	ABO*SURF6	T(72.4)	C(27.6)	TT(52.2),TC(40.4),CC(7.4)	16.7	0.3



<i>rs687289</i>	9(136137106)	ABO(intron)	A(59.5)	G(40.5)	AA(35.4),AG(48.2),GG(16.4)	15.9	6.0×10 <sup>4</sup>
<i>rs8176681</i>	9(136139754)	ABO(intron)	C(66.3)	T(33.7)	CC(43.8),CT(44.8),TT(11.4)	15.3	0.3
<i>rs9411396</i>	9(136187652)	ABO*SURF6	G(73.7)	T(26.3)	GG(53.8),GT(39.8),TT(6.4)	14.8	0.3
<i>rs493014</i>	9(136187163)	ABO*SURF6	G(90.7)	T(9.3)	GG(81.9),GT(17.5),TT(0.6)	14.2	7.8
<i>rs597411</i>	9(136192624)	ABO*SURF6	T(90.5)	G(9.5)	TT(81.7),TG(17.7),GG(0.6)	13.7	6.9
<i>rs7853989</i>	9(136131592)	ABO(exon, R176G)	C(82.6)	G(17.4)	CC(66.4),CG(32.4),GG(1.2)	10.3	3.9×10 <sup>11</sup>
<i>rs77843399</i>	9(136076392)	RALGDS*OBP2B	T(95.8)	C(4.2)	TT(91.7),TC(8.2),CC(0.1)	9.3	12.3

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**Table S9. Summary of GWAS on the quaternary trait of AB vs. O blood types**

SNP	Chr.(position)	genomic region	ra(%)	ma(%)	allele zygosity (%)	-log <sub>10</sub> (P)	OR
<b><u>AB vs. O</u></b>							
<i>rs8176719</i>	9(136132908)	ABO(exon, insert)	TC(59.4)	T(40.6)	II(35.1),ID(48.6),DD(16.3)	57.7	1.4×10 <sup>14</sup>
<i>rs657152</i>	9(136139265)	ABO(intron)	A(61.0)	C(39.0)	AA(37.1),AC(47.8),CC(15.1)	55.4	2.2×10 <sup>5</sup>
<i>rs9919007</i>	9(136119527)	OBP2B*ABO	T(68.0)	C(32.0)	TT(46.8),TC(42.6),CC(10.7)	47.7	3.5×10 <sup>2</sup>
<i>rs529565</i>	9(136149500)	ABO(intron)	C(59.4)	T(40.6)	CC(35.2),CT(48.4),TT(16.4)	45.6	5.3×10 <sup>5</sup>
<i>rs505922</i>	9(136149229)	ABO(intron)	C(59.3)	T(40.7)	CC(35.0),CT(48.6),TT(16.4)	38.9	2.7×10 <sup>6</sup>
<i>rs612169</i>	9(136143442)	ABO(intron)	G(59.2)	A(40.8)	GG(34.9),GA(48.8),AA(16.4)	38.9	2.7×10 <sup>6</sup>
<i>rs643434</i>	9(136142355)	ABO(intron)	A(59.2)	G(40.8)	AA(34.9),AG(48.8),GG(16.4)	38.9	2.7×10 <sup>6</sup>
<i>rs687289</i>	9(136137106)	ABO(intron)	A(59.5)	G(40.5)	AA(35.4),AG(48.2),GG(16.4)	25.0	4.1×10 <sup>7</sup>
<i>rs495828</i>	9(136154867)	ABO*SURF6	T(77.9)	G(22.1)	TT(61.5),TG(32.8),GG(5.7)	21.2	1.4×10 <sup>3</sup>
<i>rs635634</i>	9(136155000)	ABO*SURF6	T(77.9)	C(22.1)	TT(61.4),TC(32.9),CC(5.7)	21.2	1.4×10 <sup>3</sup>
<i>rs579459</i>	9(136154168)	ABO*SURF6	C(77.6)	T(22.4)	CC(60.8),CT(33.7),TT(5.5)	18.9	3.7×10 <sup>3</sup>
<i>rs507666</i>	9(136149399)	ABO(intron)	A(77.8)	G(22.2)	AA(61.0),AG(33.6),GG(5.4)	18.8	3.2×10 <sup>3</sup>
<i>rs651007</i>	9(136153875)	ABO*SURF6	T(77.4)	C(22.6)	TT(60.7),TC(33.5),CC(5.8)	18.3	4.0×10 <sup>3</sup>
<i>rs3118662</i>	9(136278870)	REXO4(intron)	G(51.6)	A(48.4)	GG(26.3),GA(50.7),AA(23.0)	17.2	0.1
<i>rs8176681</i>	9(136139754)	ABO(intron)	C(66.3)	T(33.7)	CC(43.8),CT(44.8),TT(11.4)	16.5	0.03
<i>rs7857390</i>	9(136128546)	ABO(3'-UTR)	A(66.4)	G(33.6)	AA(43.8),AG(45),GG(11.1)	13.2	2.0×10 <sup>-12</sup>
<i>rs7864821</i>	9(136086916)	OBP2B(intron)	C(60.6)	T(39.4)	CC(36.8),CT(47.7),TT(15.5)	13.2	5.0
<i>rs493014</i>	9(136187163)	ABO*SURF6	G(90.7)	T(9.3)	GG(81.9),GT(17.5),TT(0.6)	11.4	9.6
<i>rs7030248</i>	9(136155359)	ABO*SURF6	A(71.9)	G(28.1)	AA(51.0),AG(41.8),GG(7.2)	11.0	6.3
<i>rs9411396</i>	9(136187652)	ABO*SURF6	G(73.7)	T(26.3)	GG(53.8),GT(39.8),TT(6.4)	10.8	0.06

<i>mf09136175051</i>	9(136175051)	ABO*SURF6	T(72.4)	C(27.6)	TT(52.2),TC(40.4),CC(7.4)	10.7	0.03
<i>rs1752337</i>	9(136160228)	ABO*SURF6	G(72.3)	T(27.7)	GG(52.2),GT(40.2),TT(7.6)	10.7	0.03
<i>rs7853989</i>	9(136131592)	ABO(exon, R176G)	C(82.6)	G(17.4)	CC(66.4),CG(32.4),GG(1.2)	10.7	1.2×10 <sup>5</sup>
<i>rs597411</i>	9(136192624)	ABO*SURF6	T(90.5)	G(9.5)	TT(81.7),TG(17.7),GG(0.6)	8.9	7.0
<i>rs630014</i>	9(136149722)	ABO(intron)	A(62.2)	G(37.8)	AA(37.9),AG(48.6),GG(13.5)	8.9	0.001
<i>rs2039184</i>	9(136047393)	RALGDS*OBP2B	G(59.2)	A(40.8)	GG(35.4),GA(48.0),AA(16.8)	8.2	3.2
<i>rs4962040</i>	9(136133531)	ABO(intron)	G(66.9)	A(33.1)	GG(44.3),GA(45.2),AA(10.5)	7.8	0.003
<i>rs8176702</i>	9(136136146)	ABO(intron)	A(66.8)	G(33.2)	AA(44.7),AG(44.2),GG(11.1)	7.8	0.003
<i>rs2073828</i>	9(136137140)	ABO(intron)	A(66.4)	G(33.6)	AA(44.0),AG(45.0),GG(11.0)	7.7	0.003

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