Supplementary material for "African ancestry is associated with Glaucoma in the Women's Health Initiative genome-wide association study"

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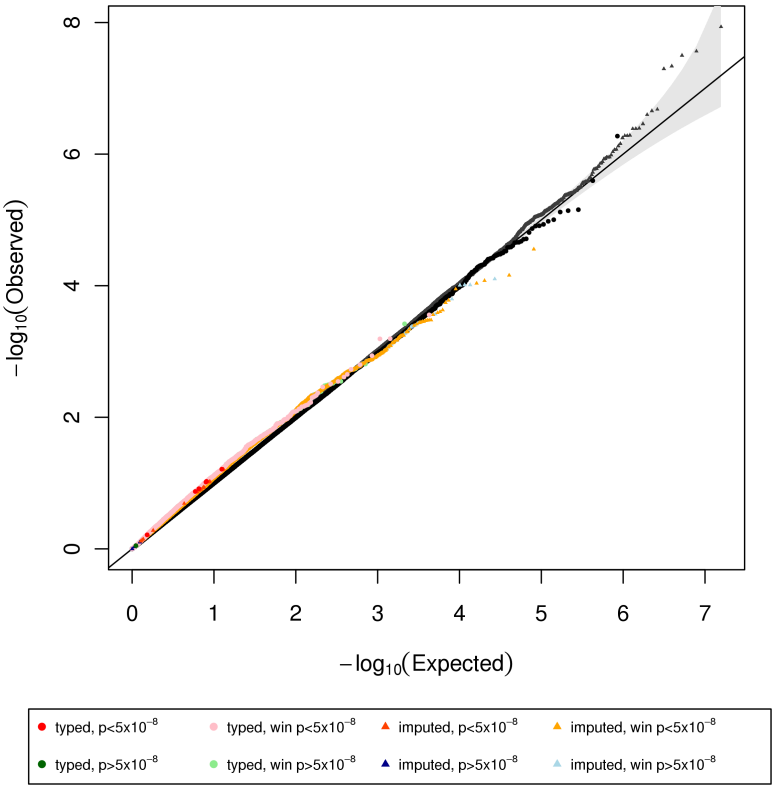
Table S1: Correlation (r^2) of markers in the CDKN2B/CDKN2BAS genes in the 1000 Genomes populations ASW/AMR (http://1000genomes.org), respectively.

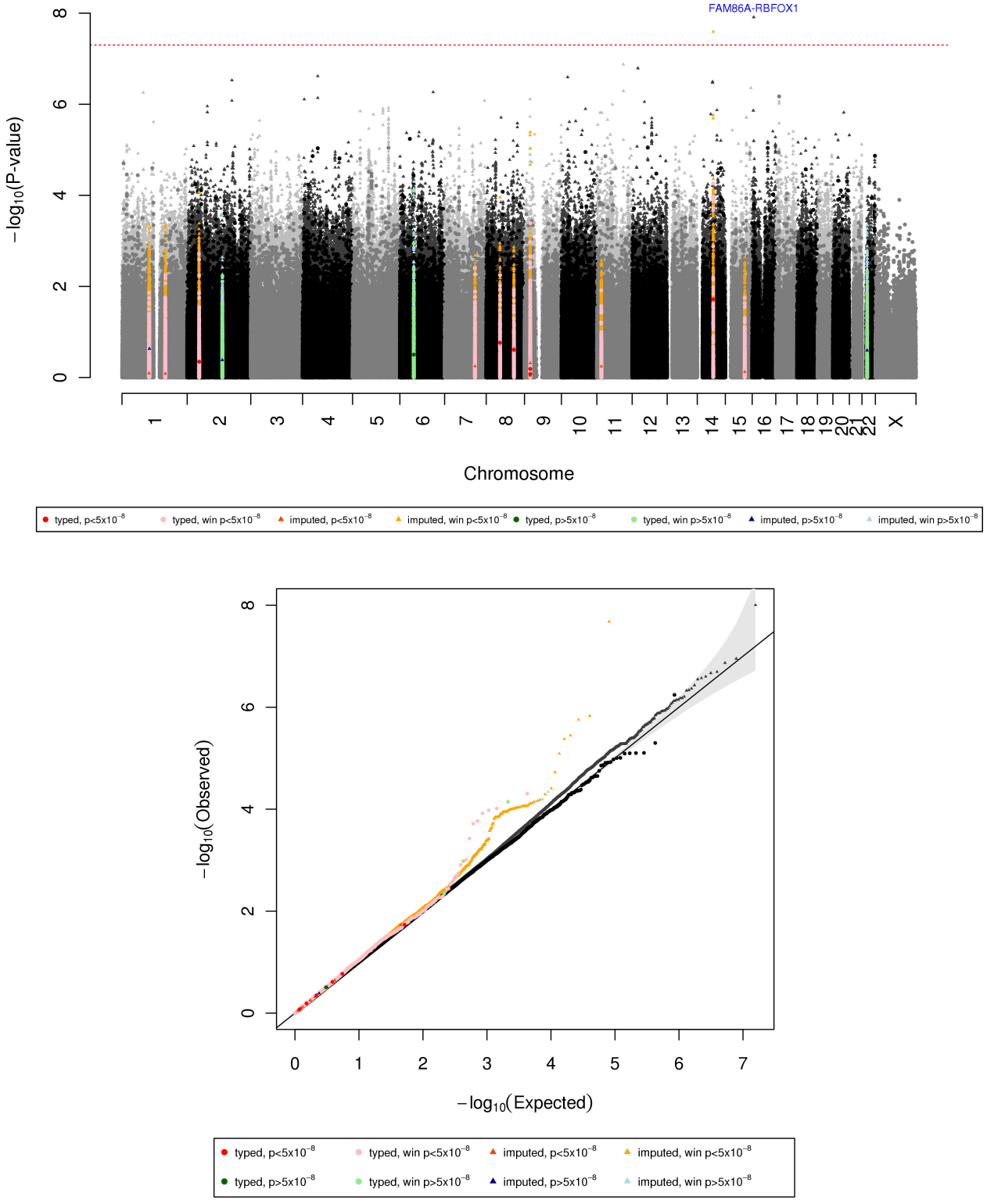
	rs7865618	rs2157719	rs4977756	rs1063192
rs523096	0.79/0.88	0.79/0.88	0.01/0.66	0.79/0.86
rs7865618	—	1/1	0.03/0.74	1/0.95
rs2157719	—	—	0.03/0.74	1/0.95
rs4977756	—	—	—	0.03/0.7

Figure S1: Manhattan and Q-Q plots. In the plot/legend, the color code of the p-values corresponds to the p-value in previously reported studies; for example "typed, p $< 5\times10^{-8}$" indicates that a previous study had a p-value $< 5\times10^{-8}$, i.e., genome-wide significant, and "typed, win p $< 5\times10^{-8}$" indicates that it is within 0.5 megabases (1 megabase window) of such a SNP.

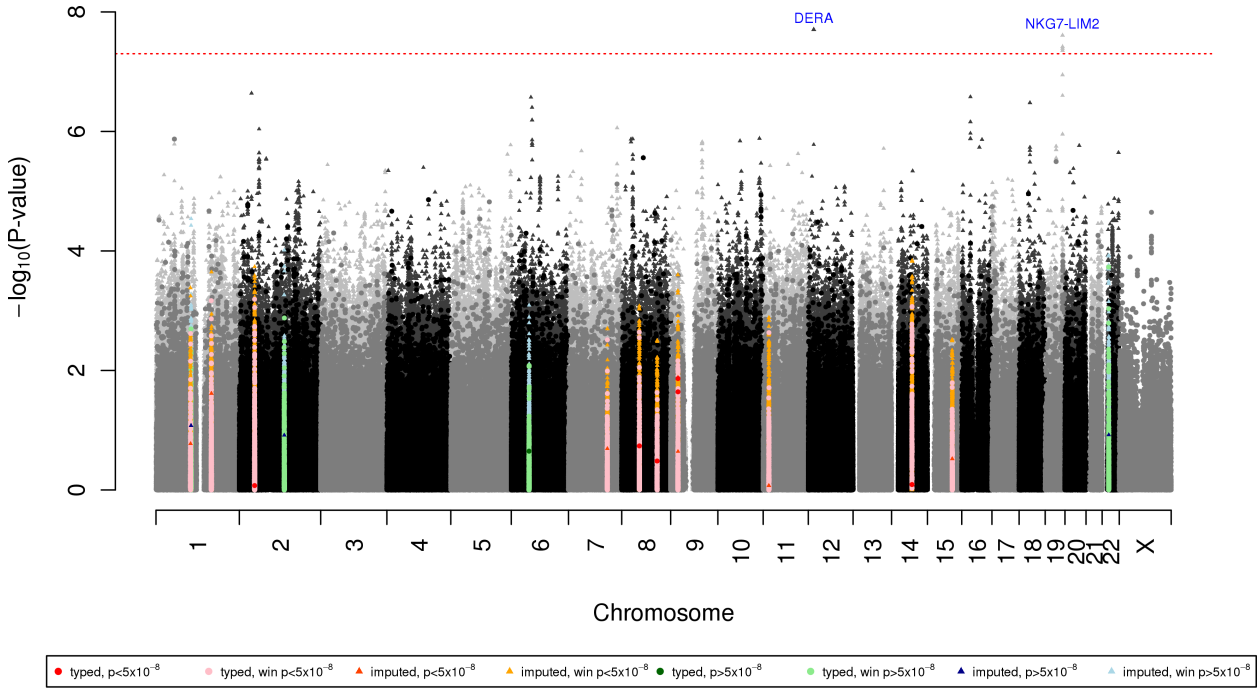
(a) African American incident + prevalent (combined via inverse variance method). $\lambda = 1.022$. The Manhattan plot is in the main manuscript (Figure 1), the Q-Q plot is shown below.

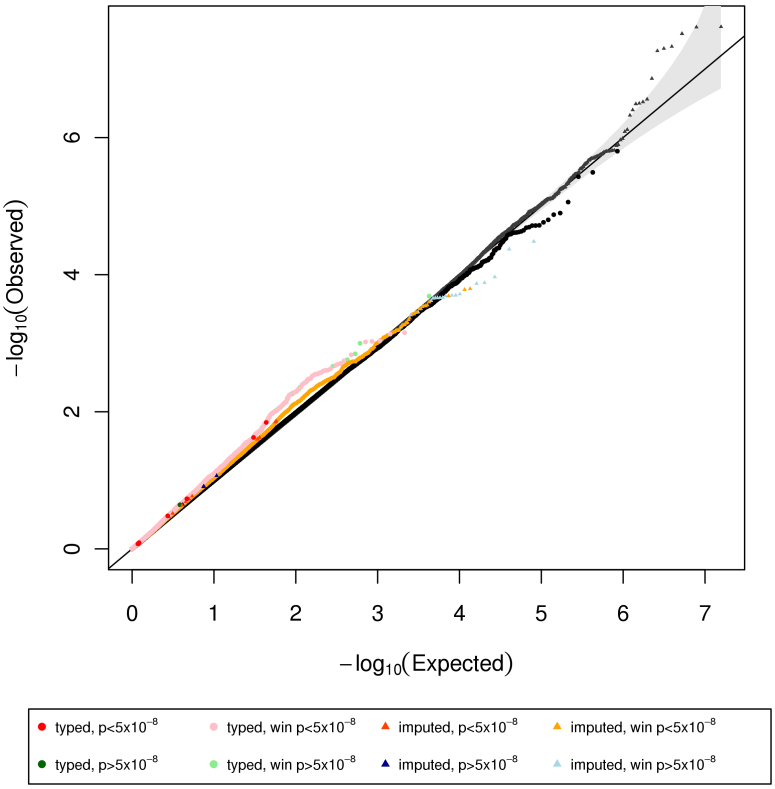


(b) African American incident. $\lambda = 0.988$.

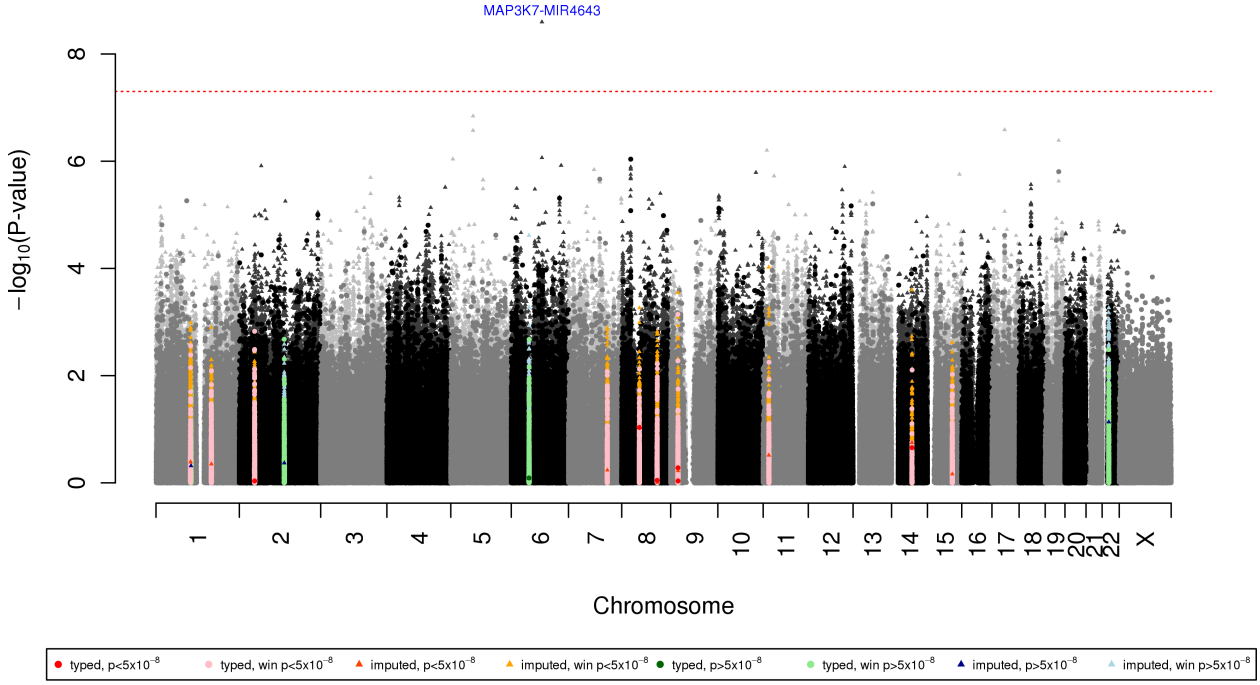


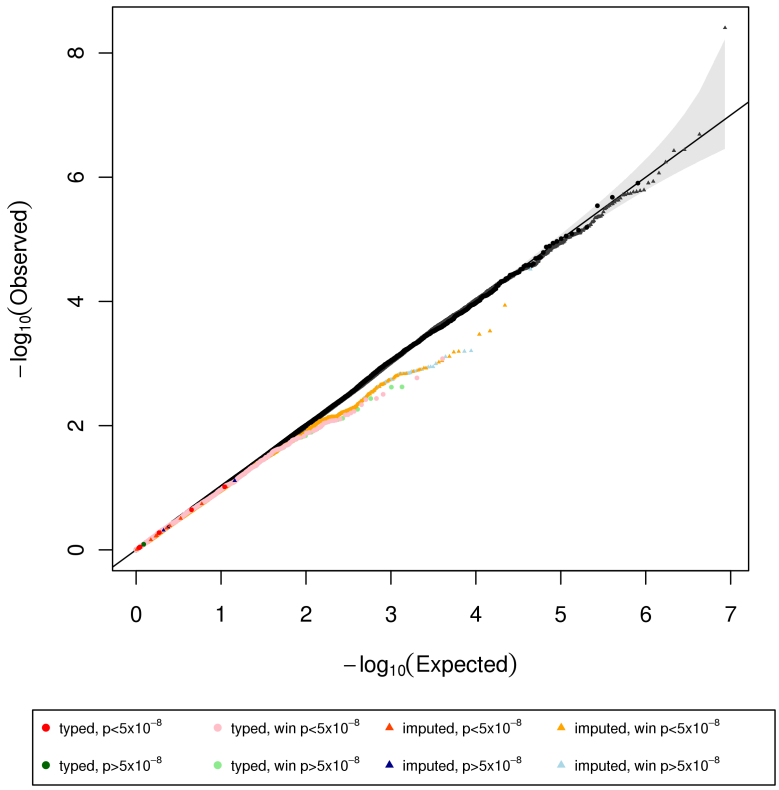
(c) African American prevalent. $\lambda = 1.014$.



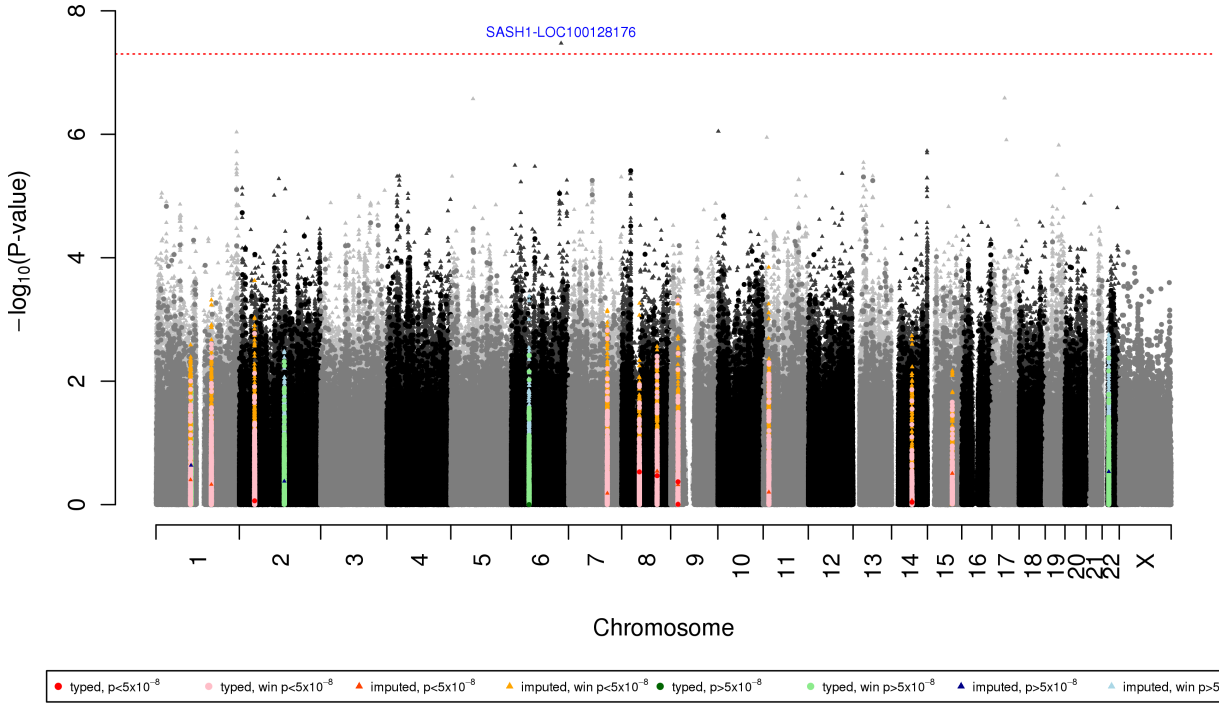


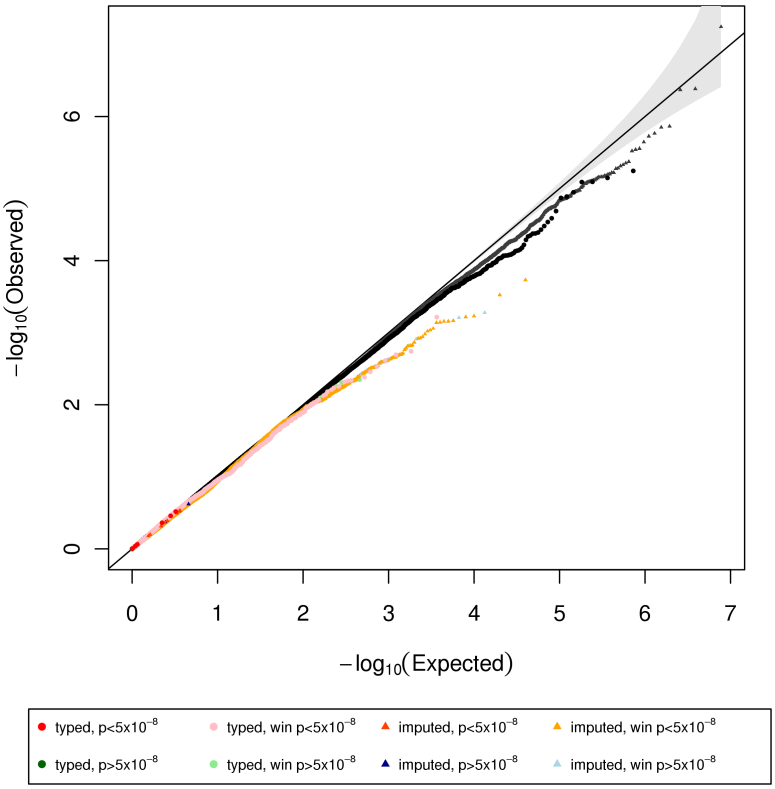
(d) Hispanic incident + prevalent (combined via inverse variance method). $\lambda = 1.025$.



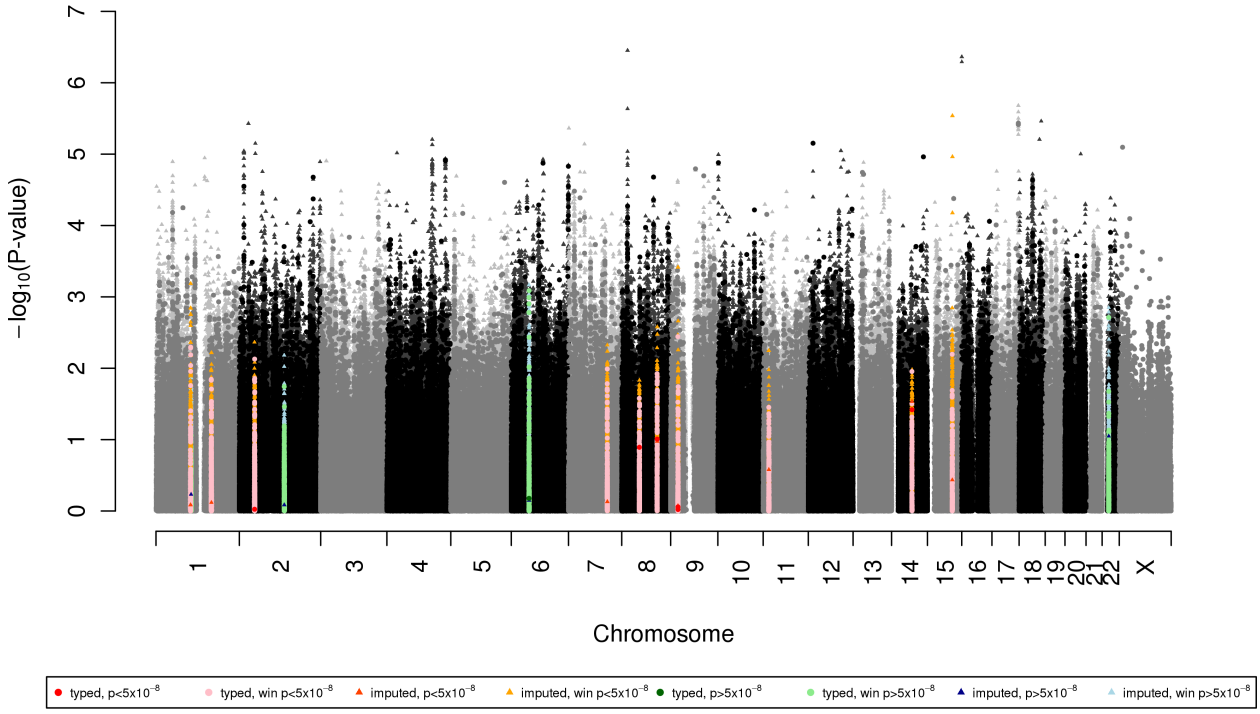


(e) Hispanic incident. $\lambda = 1.036$.





(f) Hispanic prevalent. $\lambda = 0.994$.



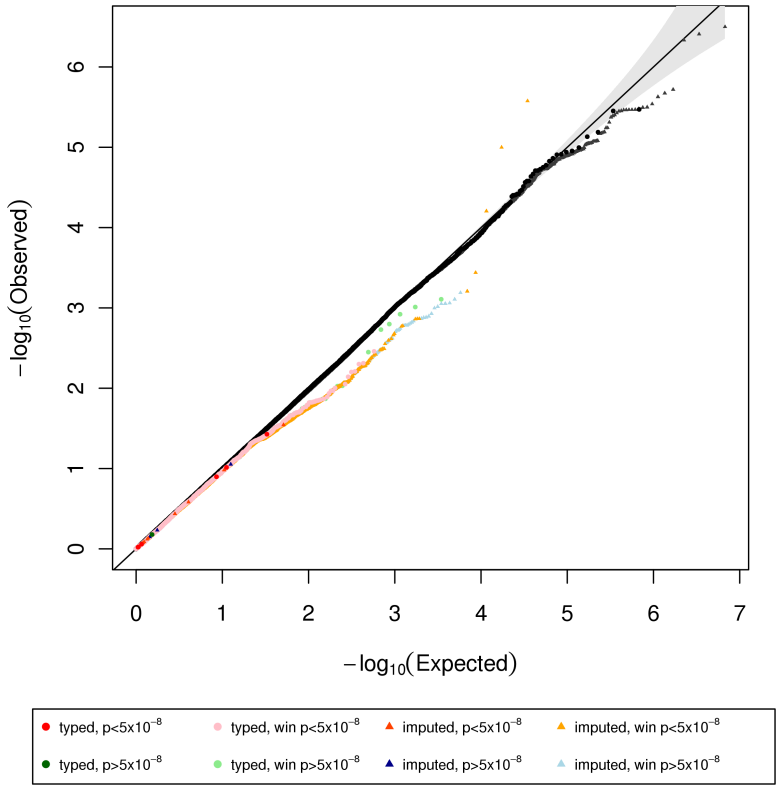
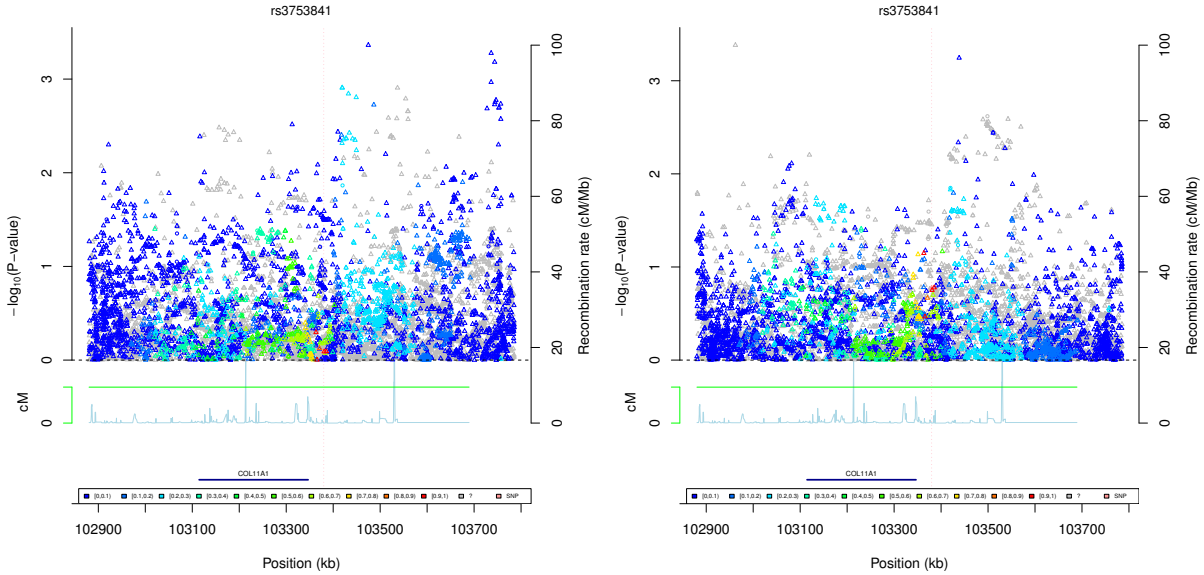


Table S2: P-values of SNPs with $p < 5 \times 10^{-8}$ in either African American incident, African American prevalent, Hispanic incident, or Hispanic prevalent. No hit replicates. Chr, chromosome; A1F, frequency of A1 allele; HR, hazards ratio (incident); OR, odds ratio (prevalent); P, p-value; Prev OR, odds ratio previously reported for SNP; Effect, going across the columns for African American incident, African American prevalent, Hispanic incident, and Hispanic prevalent, a "+" indicates the A1 allele was estimated deleterious and a "-" indicates the A2 allele was deleterious.

						African American Incident				African American Prevalent				African American Comb.	
SNP	Chr	Pos	A1	A2	r^2	Case	Cont	HR/OR	P	Case	Cont	HR/OR	P	HR/OR	P
rs151326733	12	16189478	C	G	.942	.027	.026	1.019 (.78,1.33)	.89	.053	.026	2.2 (1.67,2.89)	1.9e-08	1.48 (1.22,1.8)	5.7e-05
rs116769055	12	16191530	C	T	.944	.026	.026	1.018 (.78,1.33)	.9	.053	.026	2.2 (1.67,2.89)	2e-08	1.48 (1.22,1.8)	5.7e-05
chr14:60847430:I	14	60847430	AAAGG	A	.859	.45	.392	1.3 (1.19,1.43)	2.5e-08	.388	.401	.95 (.84,1.078)	.42	1.17 (1.083,1.26)	5.8e-05
rs3116139	19	51879034	T	C	.986	.936	.934	1.022 (.86,1.22)	.81	.894	.934	.57 (.47,.7)	2.5e-08	.79 (.69,.9)	.00037
rs2547318	19	51883727	A	G	.973	.934	.932	1.012 (.85,1.2)	.9	.892	.932	.58 (.48,.7)	4.1e-08	.79 (.69,.9)	.00036
rs2547316	19	51884597	G	A	.968	.934	.931	1.012 (.85,1.2)	.9	.892	.932	.58 (.48,.7)	3.8e-08	.79 (.69,.9)	.00035
rs1978456	19	51886715	G	T	.963	.933	.931	1.01 (.85,1.2)	.91	.892	.932	.58 (.48,.71)	4.4e-08	.79 (.69,.9)	.00036
rs190298731	6	149059291	T	A	.631	.158	.16	.98 (.85,1.14)	.83	.164	.159	1.053 (.87,1.28)	.6	1.0087 (.9,1.13)	.88

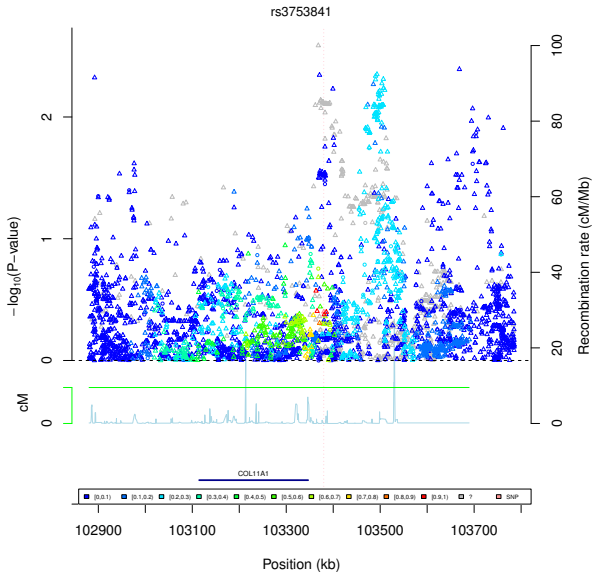
	Hispanic Incident					Hispanic Prevalent				Hispanic Comb.		
SNP	Hisp r^2	Hisp I Case	Hisp I Cont	Hisp I HR/OR	Hisp I P	Hisp P Case	Hisp P Cont	Hisp P HR/OR	Hisp P P	Hisp C HR/OR	Hisp C P	Effect
rs151326733	.896	.002	.002	— —	—	0	.002	— —	—	—	—	++(+)??(?)
rs116769055	.899	.002	.002	— —	—	0	.002	— —	—	—	—	++(+)??(?)
chr14:60847430:I	.87	.254	.244	1.094 (.91,1.32)	.35	.265	.245	1.15 (.86,1.52)	.34	1.11 (.95,1.3)	.19	+-(+)++(+)
rs3116139	.987	.74	.73	1.041 (.88,1.24)	.65	.739	.731	.995 (.76,1.3)	.97	1.027 (.89,1.19)	.72	+-(-)+-(+)
rs2547318	.991	.739	.729	1.034 (.87,1.23)	.7	.739	.73	1.0036 (.77,1.31)	.98	1.025 (.89,1.18)	.74	+-(-)++(+)
rs2547316	.985	.739	.727	1.04 (.88,1.24)	.66	.738	.729	1.0042 (.77,1.31)	.98	1.029 (.89,1.19)	.7	+-(-)++(+)
rs1978456	.983	.739	.727	1.042 (.88,1.24)	.64	.739	.729	1.013 (.78,1.32)	.92	1.033 (.89,1.19)	.66	+-(-)++(+)
rs190298731	.526	.128	.082	2.3 (1.71,3.09)	3.3e-08	.085	.087	.96 (.54,1.72)	.9	1.92 (1.47,2.49)	1.2e-06	--(-)--(-)

Figure S2: Local LD plots for SNPs identified as associated with Glaucoma in previous GWAS, and what population the SNP was originally found in. The LD reported is that in the population it was originally found in. AA, African American; Hisp, Hispanic; P, prevalent; I, incident; EUR, European race/ethnicity 1000 Genomes individuals; ASN, Asian race/ethnicity 1000 Genomes individuals.
(a) rs3753841 (Originally found in Asian race/ethnicity, replicated in both Asian and White race/ethnicity [8])

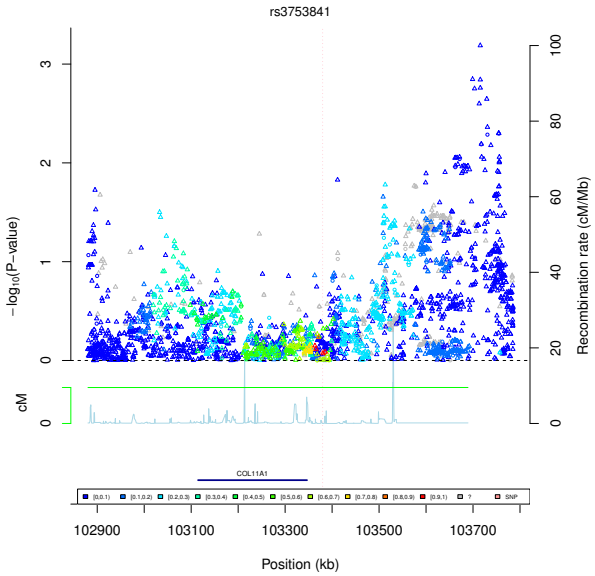


(i) AA I, ASN LD

(ii) AA P, ASN LD

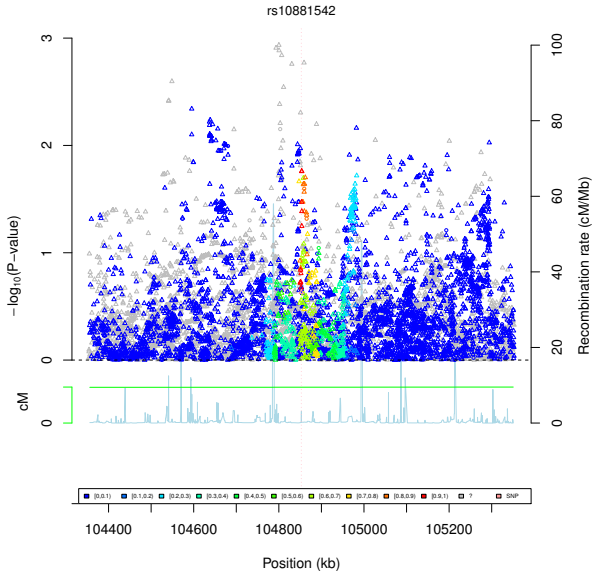


(iii) Hisp I, ASN LD

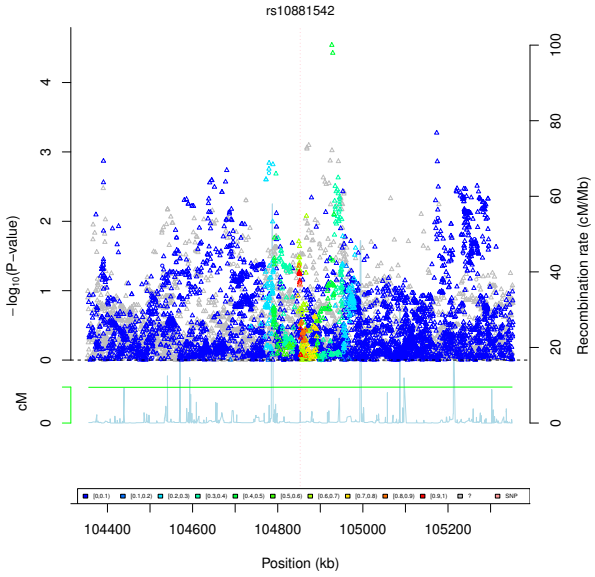


(iv) Hisp P, ASN LD

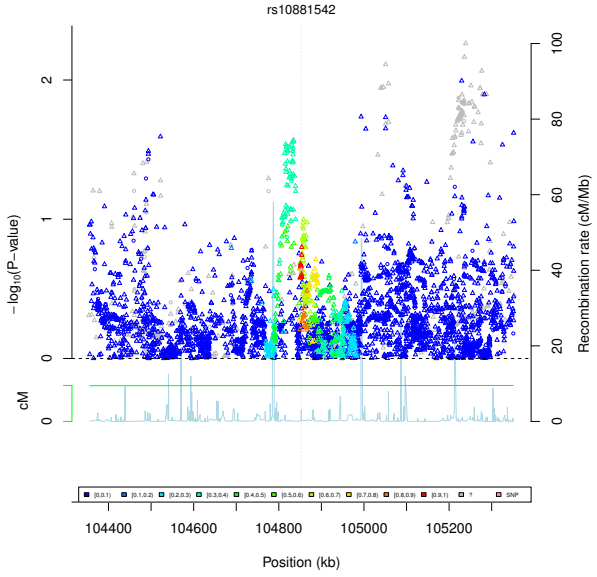
(b) rs10881542 (Originally found in Asian race/ethnicity, replicated in both Asian and White race/ethnicity [8])



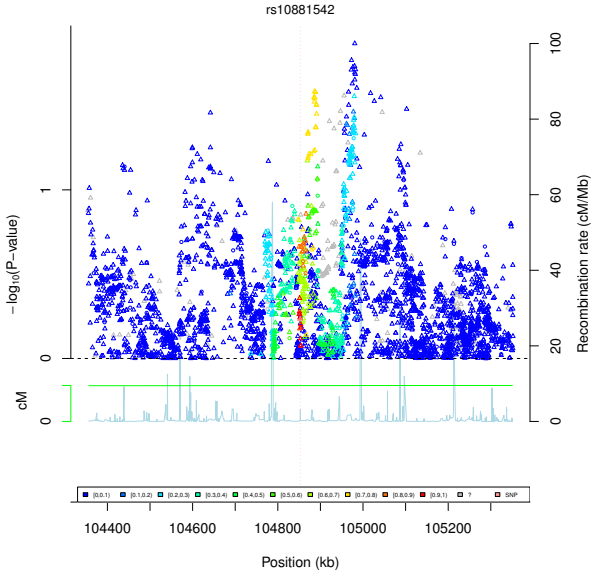
(i) AA I, ASN LD



(ii) AA P, ASN LD

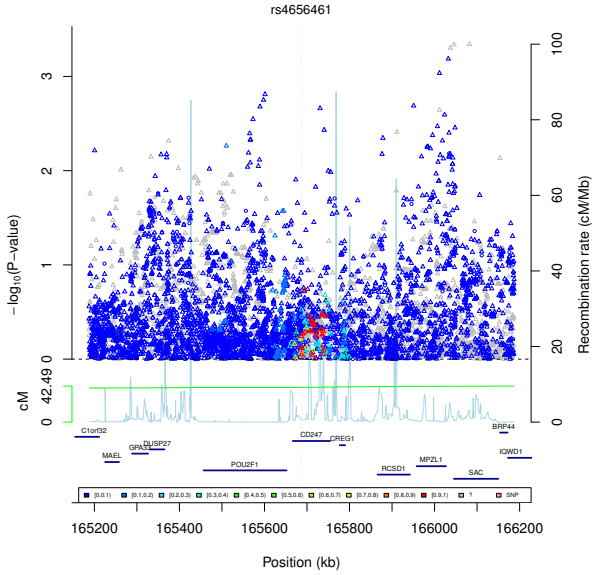


(iii) Hisp I, ASN LD

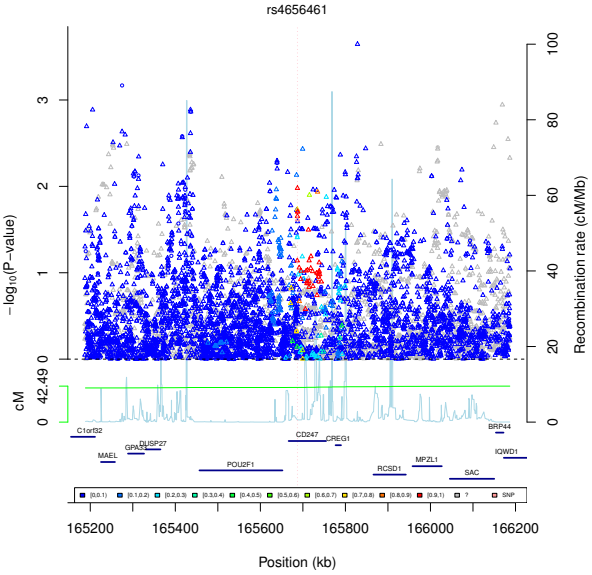


(iv) Hisp P, ASN LD

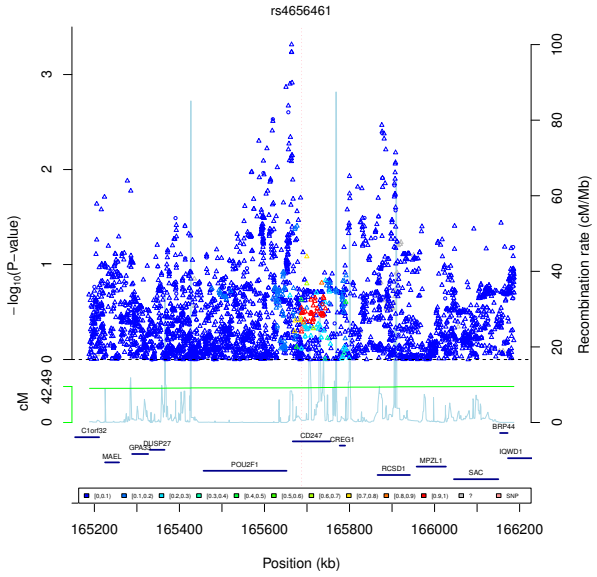
(c) rs4656461 (European, European replication [1])



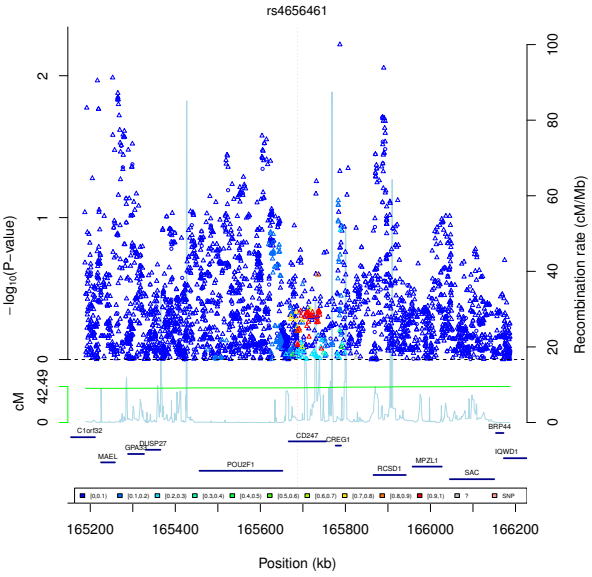
(i) AA I, EUR LD



(ii) AA P, EUR LD

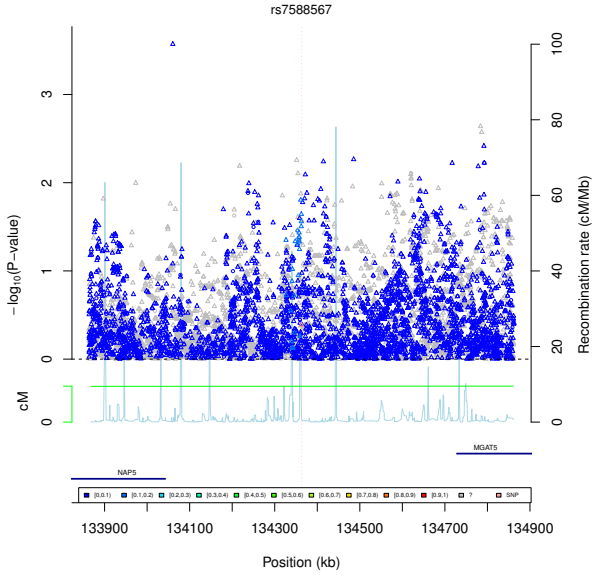


(iii) Hisp I, EUR LD

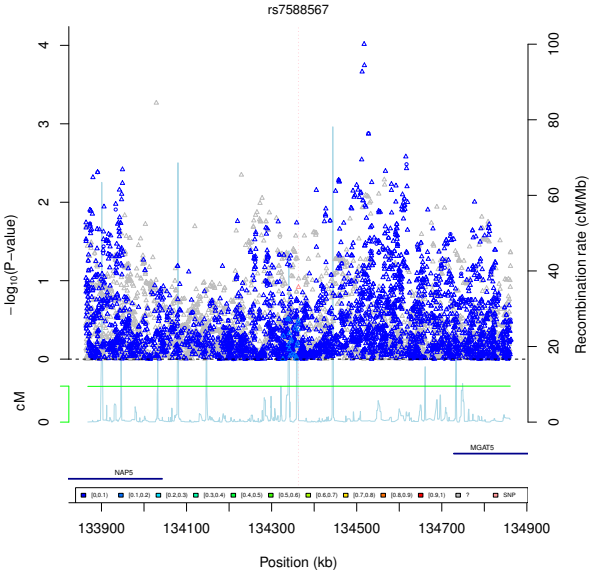


(iv) Hisp P, EUR LD

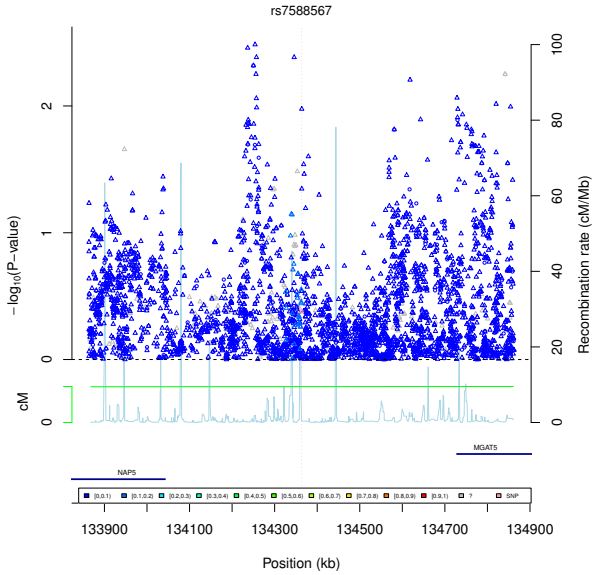
(d) rs7588567 (Japanese, Japanese replication [4])



(i) AA I, ASN LD



(ii) AA P, ASN LD



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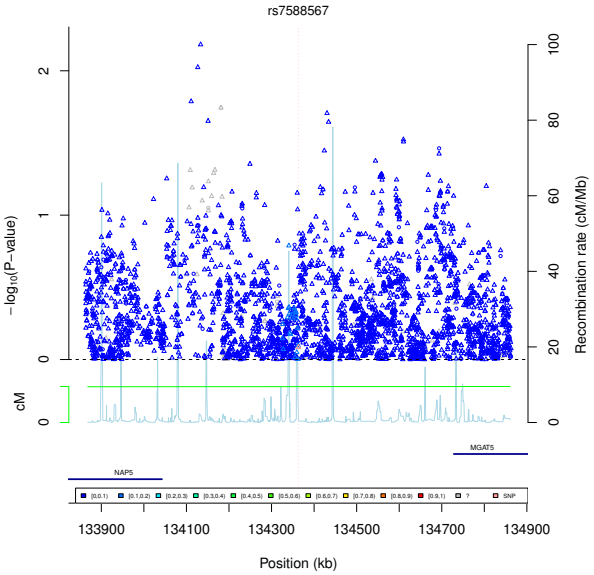
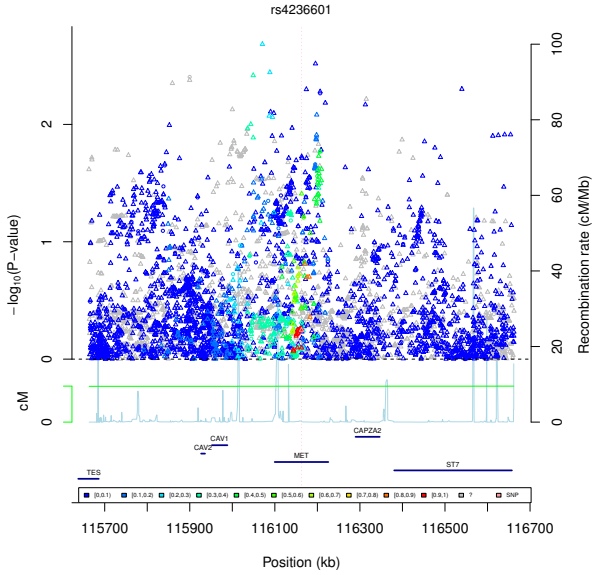
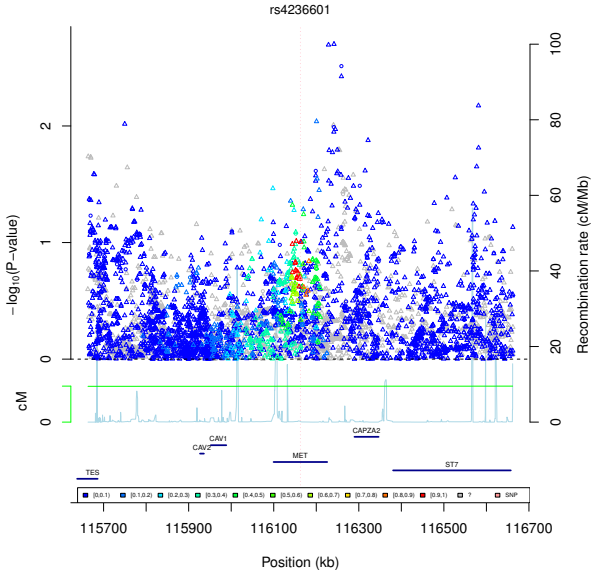


(iv) Hisp P, ASN LD

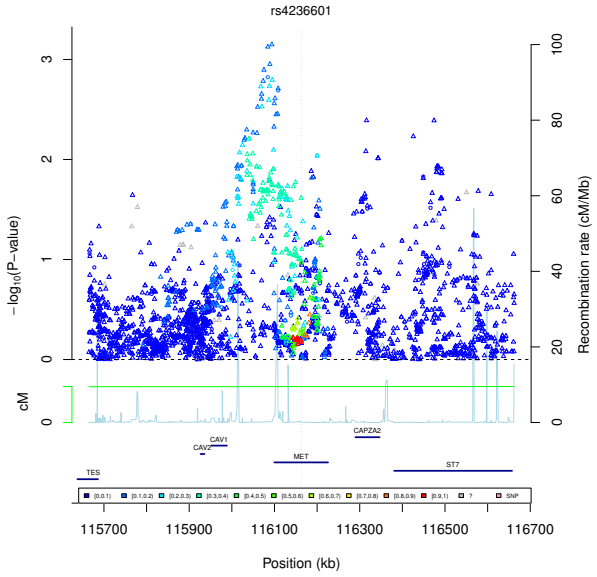
(e) rs4236601 (Icelandic, European replication [7])



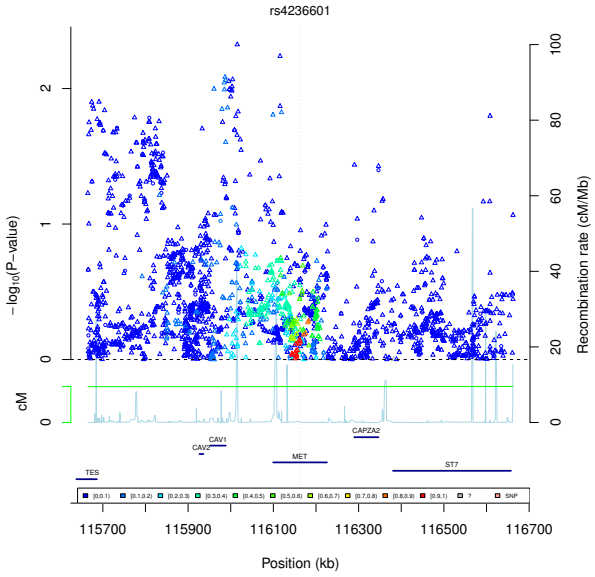
(i) AA I, EUR LD



(ii) AA P, EUR LD

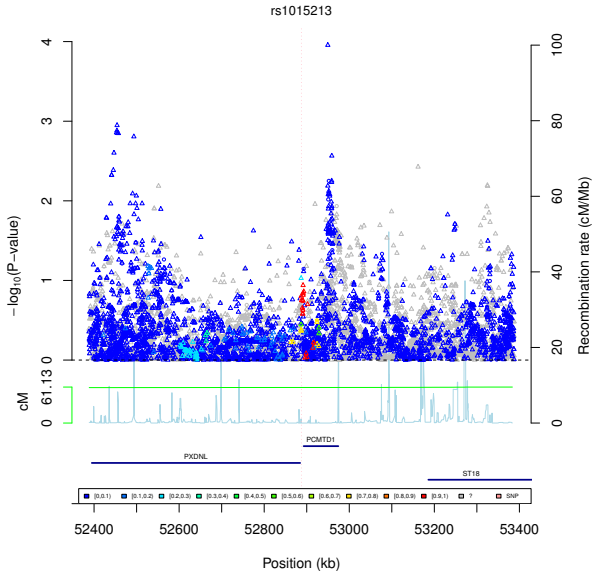


(iii) Hisp I, EUR LD



(iv) Hisp P, EUR LD

(f) rs1015213 (Originally found in Asian race/ethnicity, replicated in both Asian and White race/ethnicity [8])



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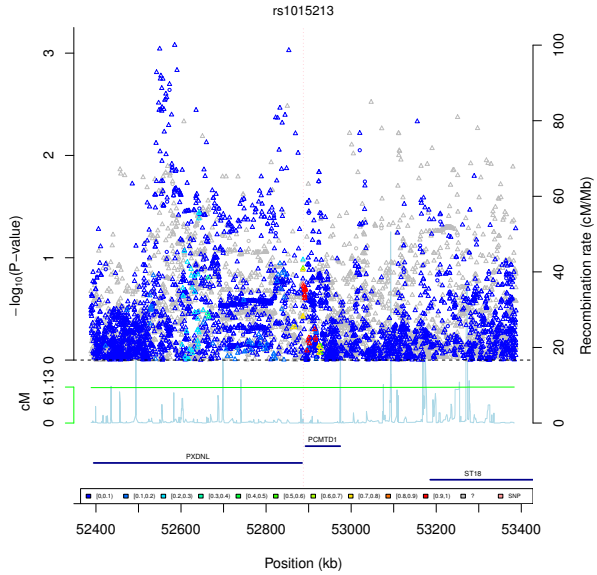
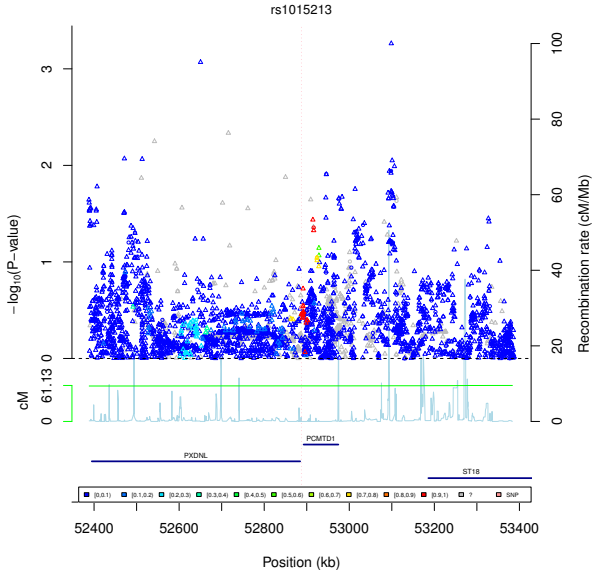


(ii) AA P, ASN LD



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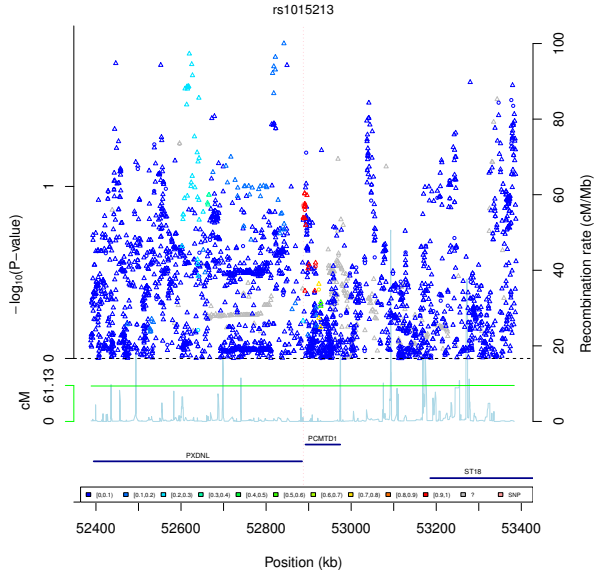
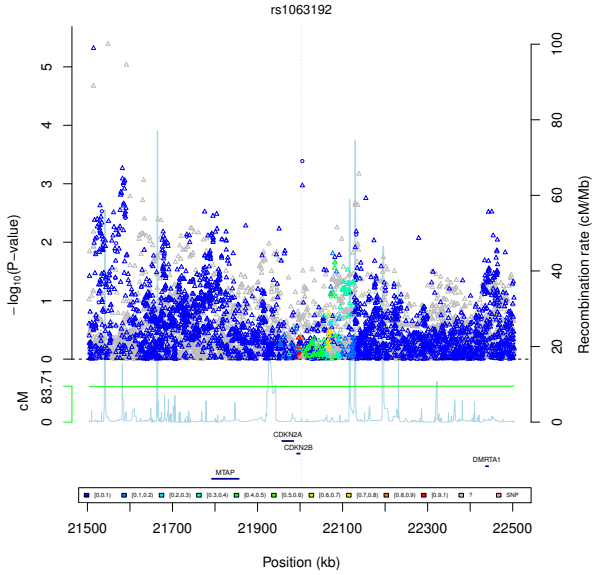
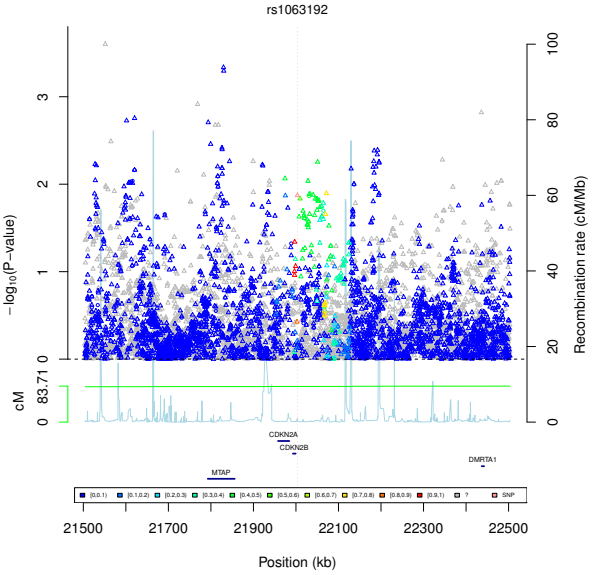


(iv) Hisp P, ASN LD

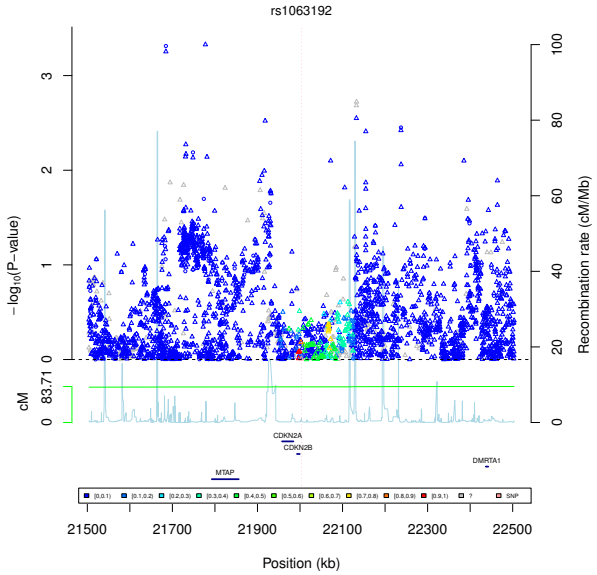
(g) rs1063192 (Japanese, Japanese replication [4])



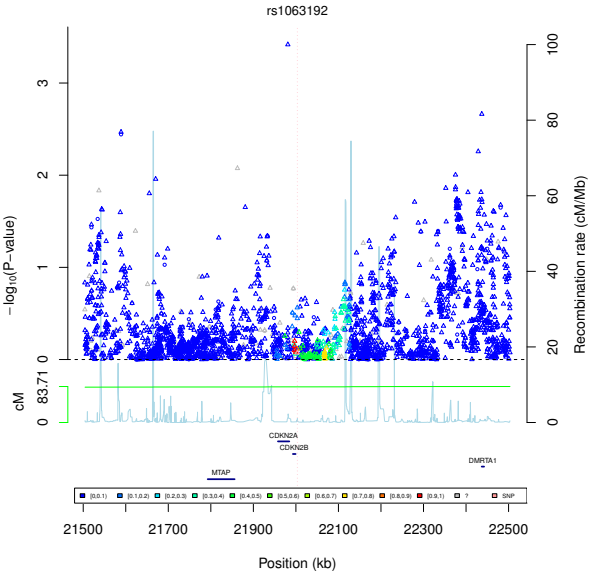
(i) AA I, ASN LD



(ii) AA P, ASN LD

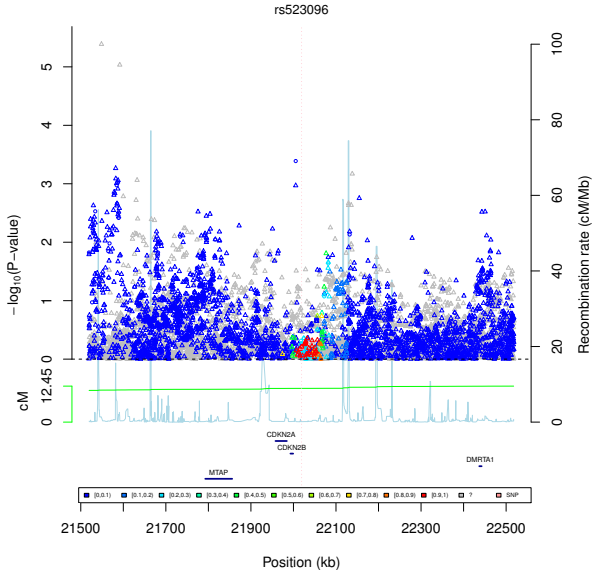


(iii) Hisp I, ASN LD

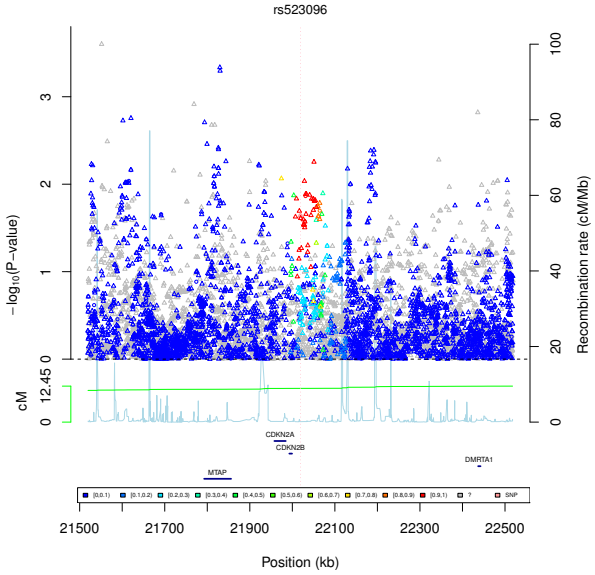


(iv) Hisp P, ASN LD

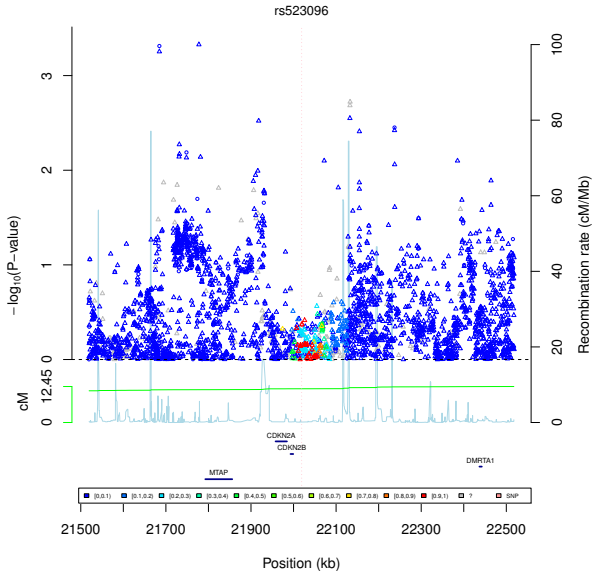
(h) rs523096 (Japanese, Japanese replication [5])



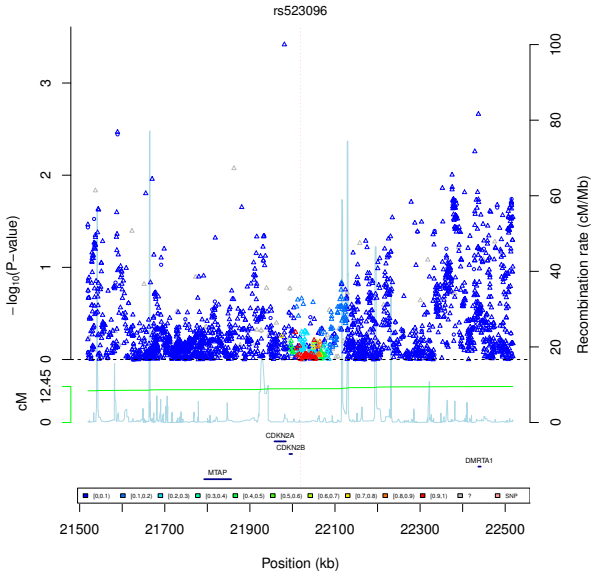
(i) AA I, ASN LD



(ii) AA P, ASN LD

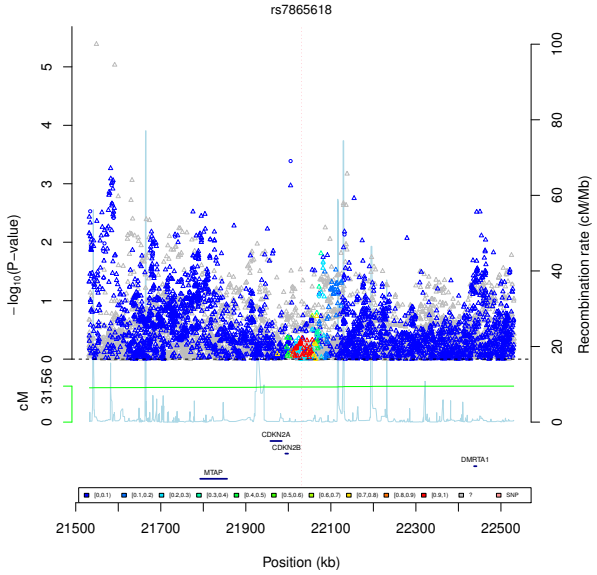


(iii) Hisp I, ASN LD

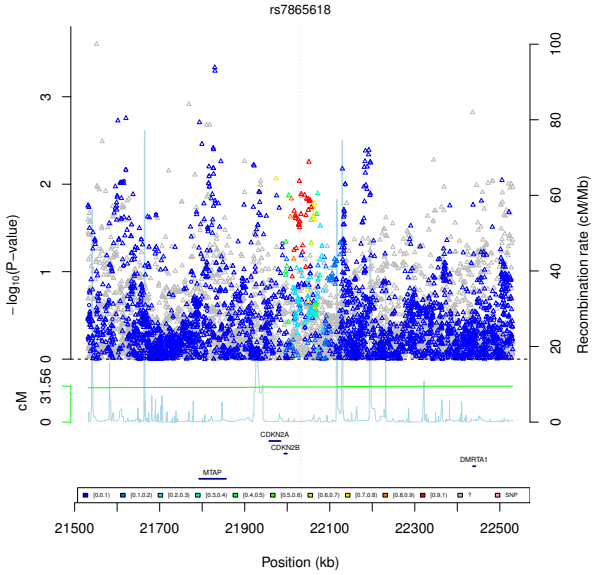


(iv) Hisp P, ASN LD

(i) rs7865618 (Japanese, Japanese replication [3])



(i) AA I, ASN LD



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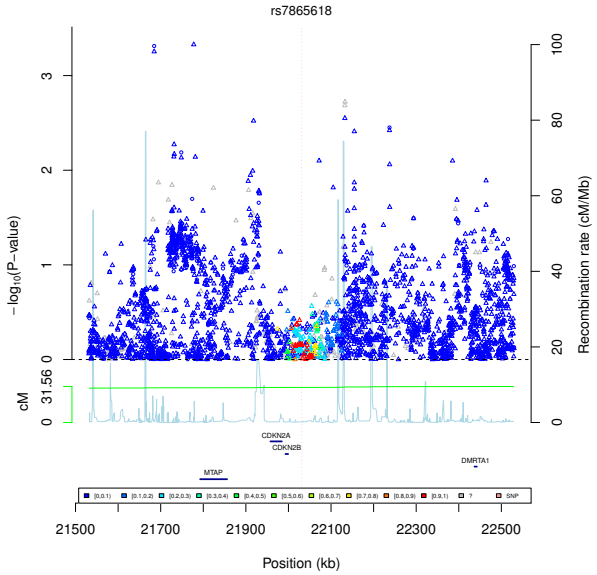
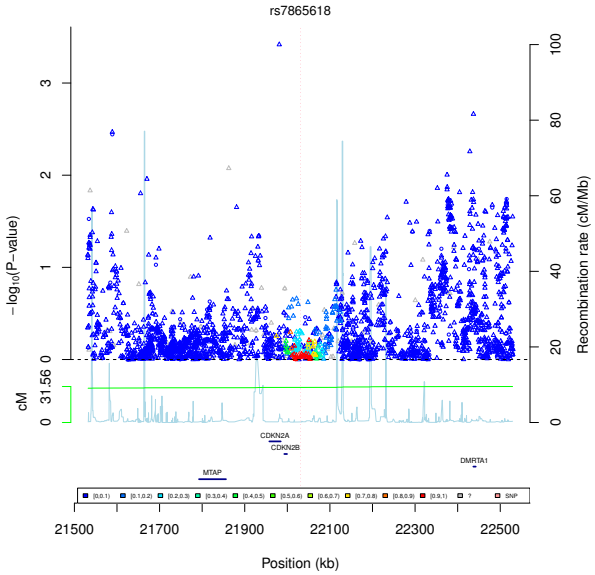
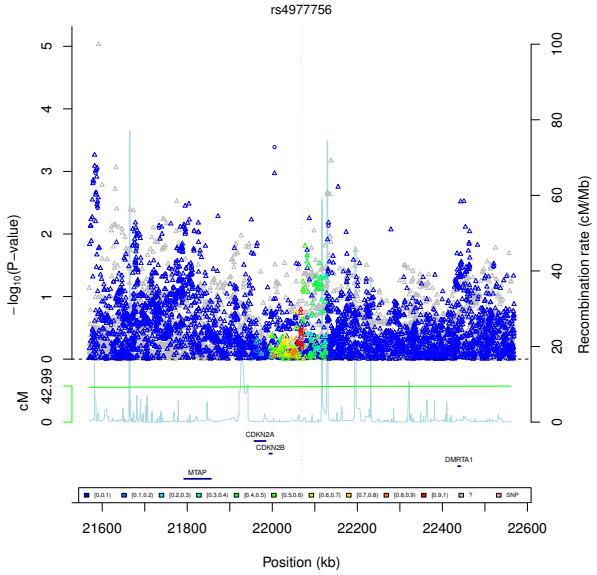


(iii) Hisp I, ASN LD



(iv) Hisp P, ASN LD

(j) rs4977756 (European, European replication [1])



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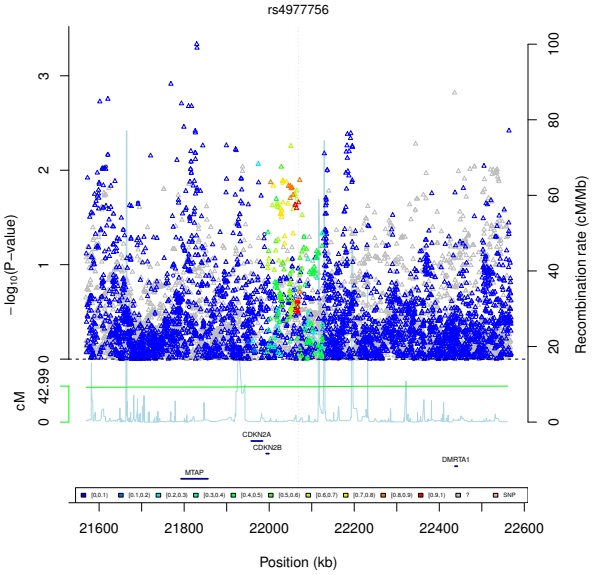
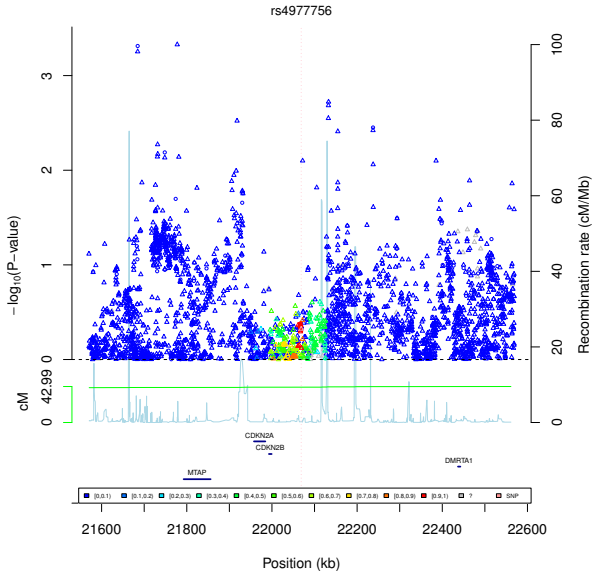
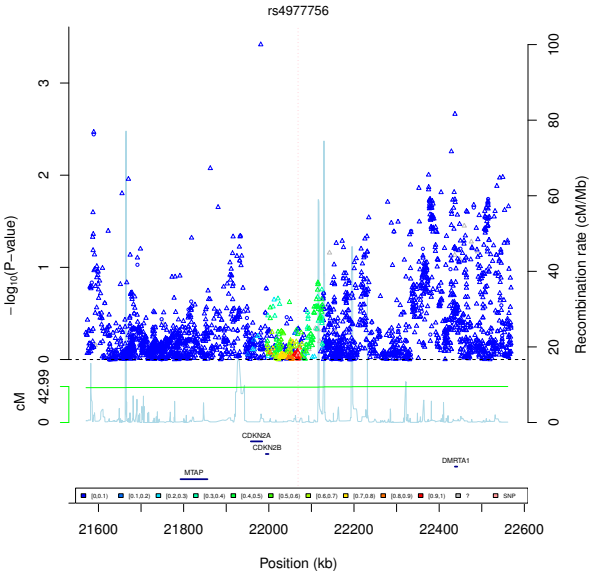


(ii) AA P, EUR LD

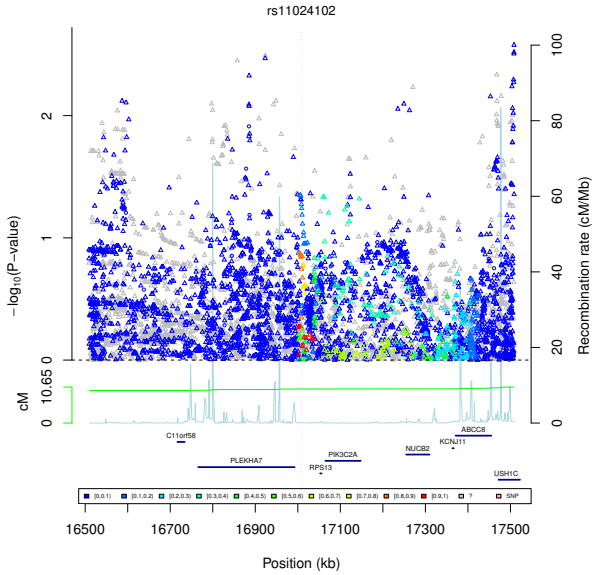


(iii) Hisp I, EUR LD



(iv) Hisp P, EUR LD

(k) rs11024102, (Originally found in Asian race/ethnicity, replicated in both Asian and White race/ethnicity [8])



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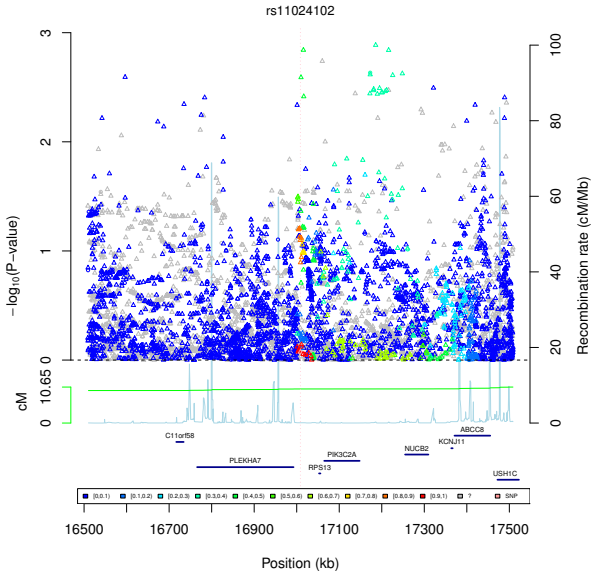

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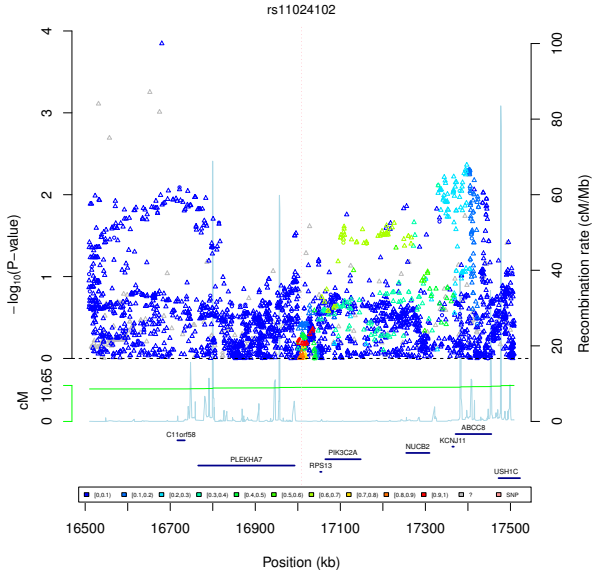
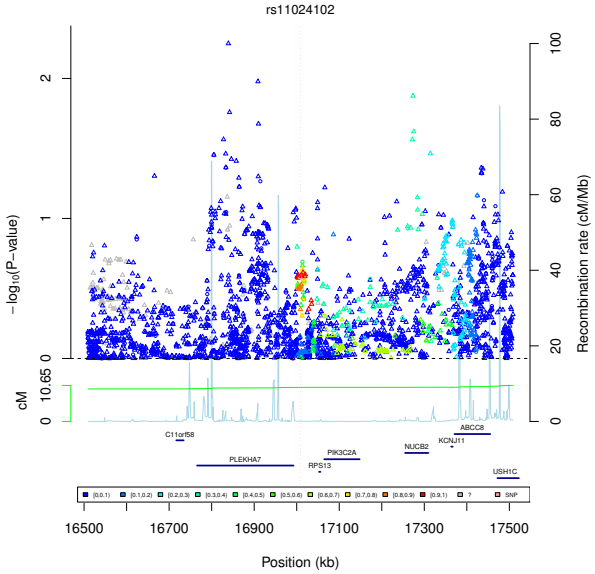
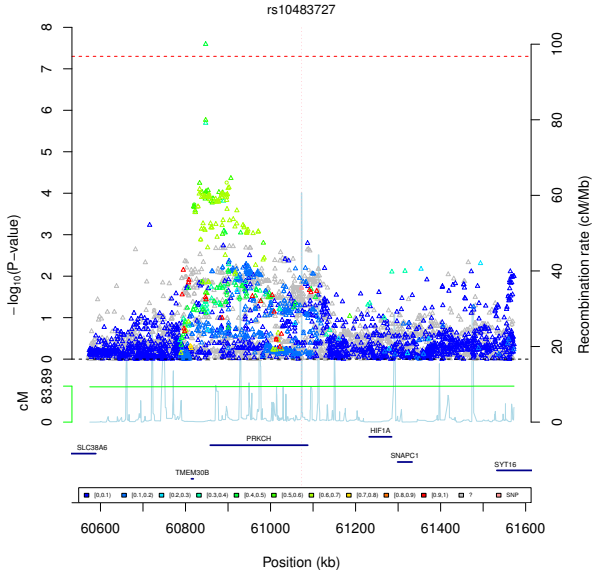


(iii) Hisp I, ASN LD

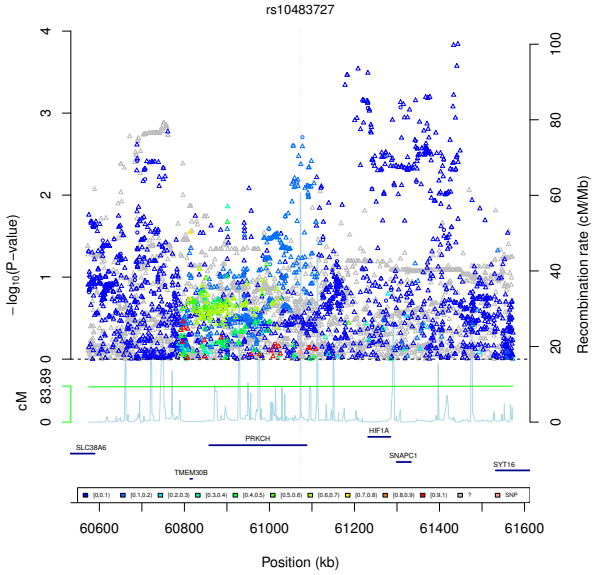


(iv) Hisp P, ASN LD

(l) rs10483727 (Meta-analysis of White ancestry [9]; Japanese, Japanese replication [4])



(i) AA I, EUR LD



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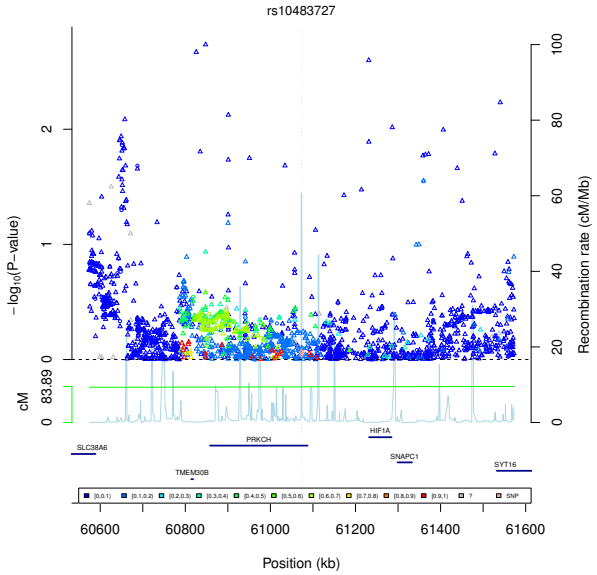
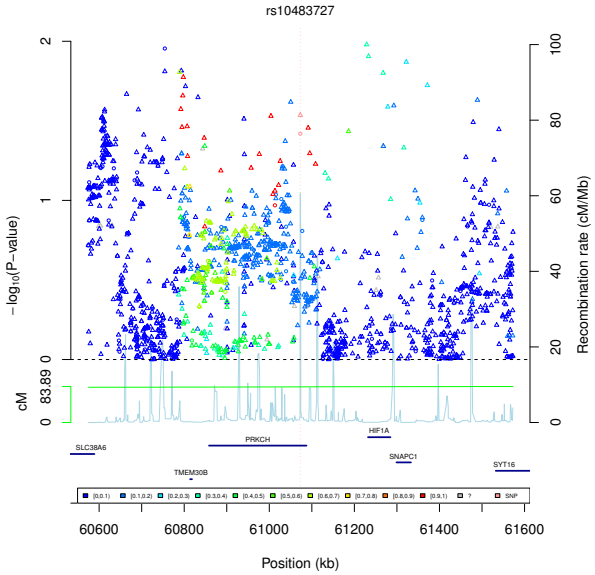
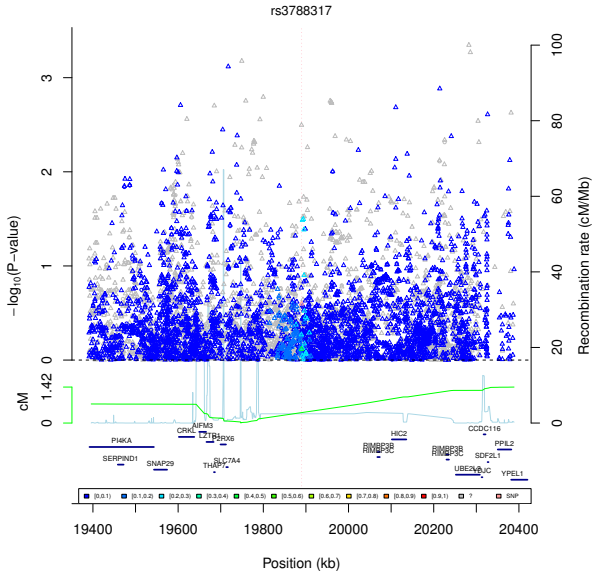


(iii) Hisp I, EUR LD

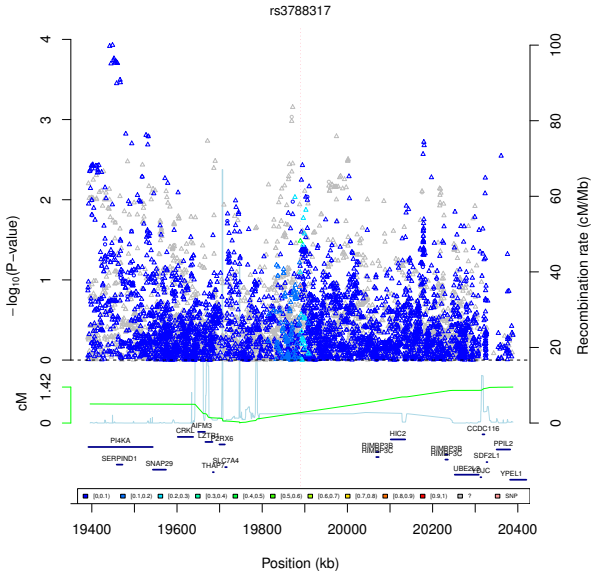


(iv) Hisp P, EUR LD

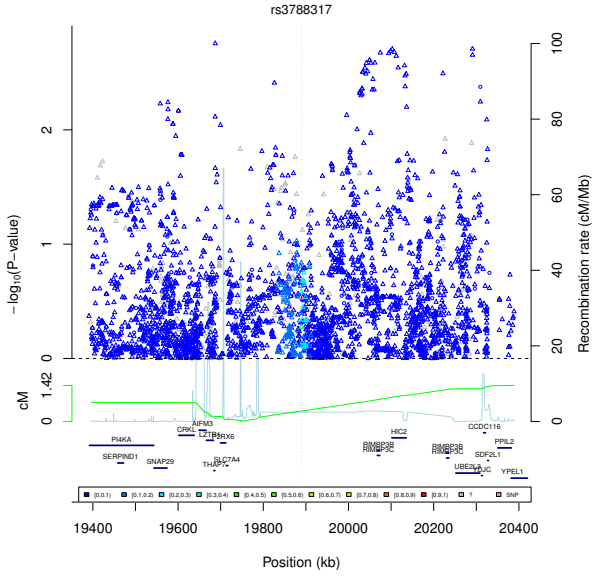
(m) rs3788317 (Originally found in Asian race/ethnicity, replicated in both Asian and White race/ethnicity [8])



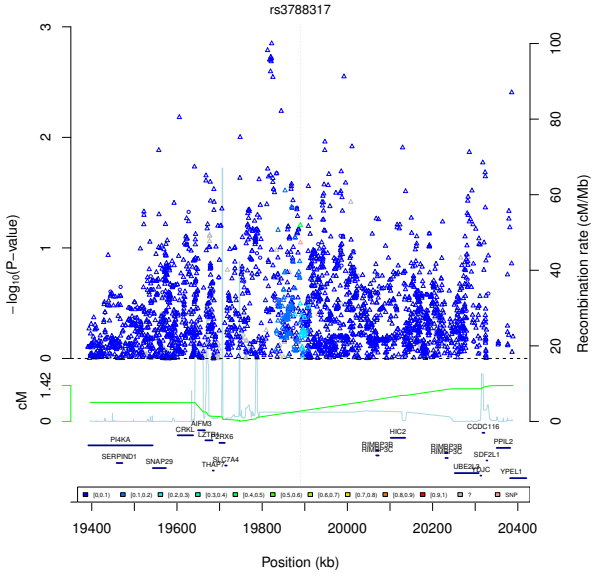
(i) AA I, ASN LD



(ii) AA P, ASN LD

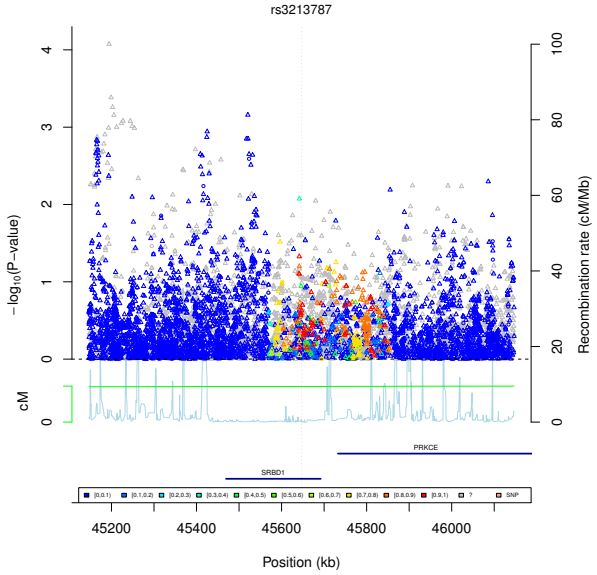


(iii) Hisp I, ASN LD



(iv) Hisp P, ASN LD

(n) rs3213787 (Japanese, not replicated [2])



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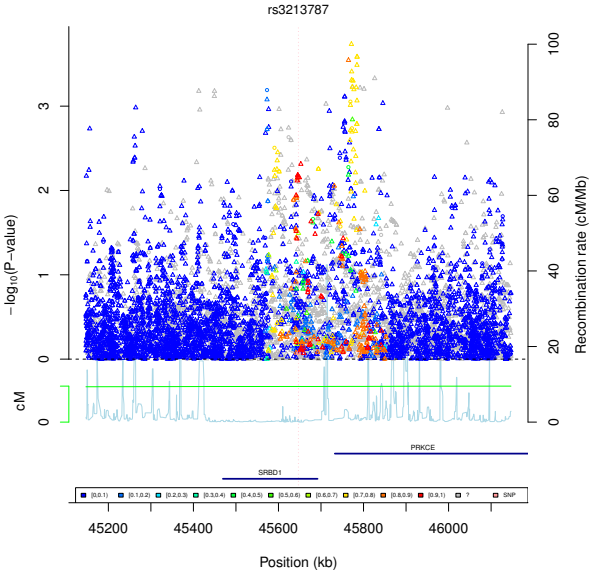

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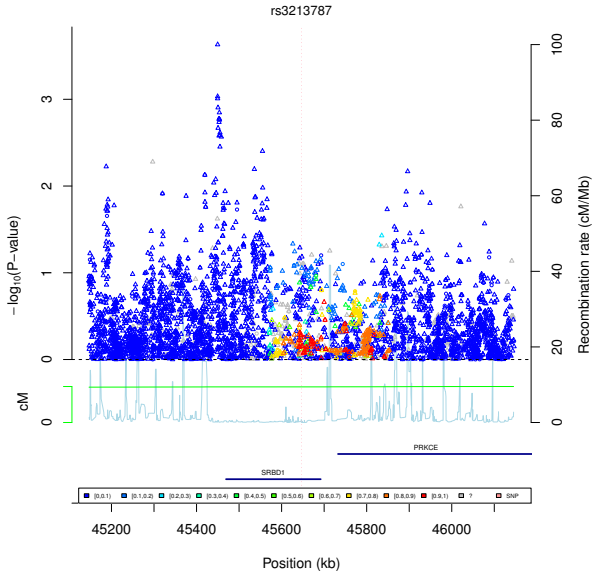
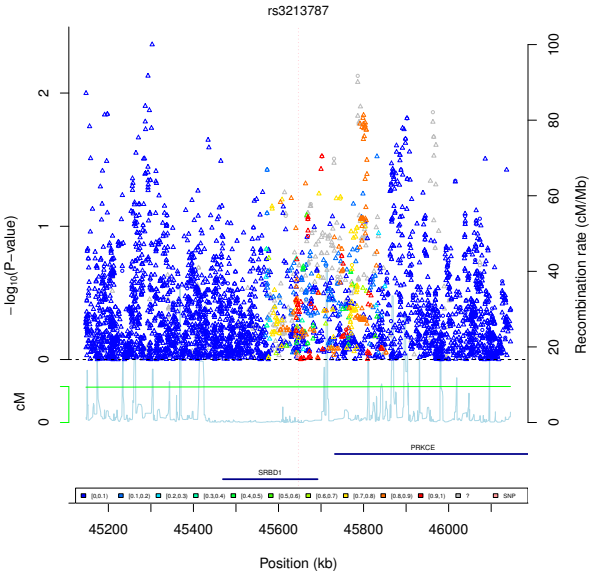
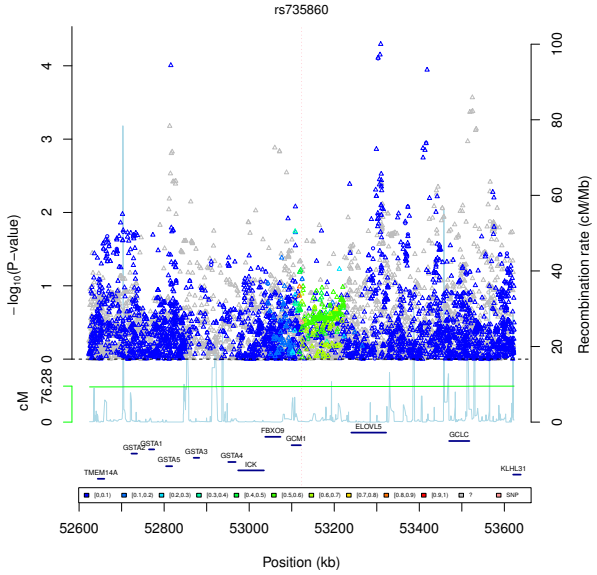


(iii) Hisp I, ASN LD



(iv) Hisp P, ASN LD

(o) rs735860 (Japanese, not replicated [2])



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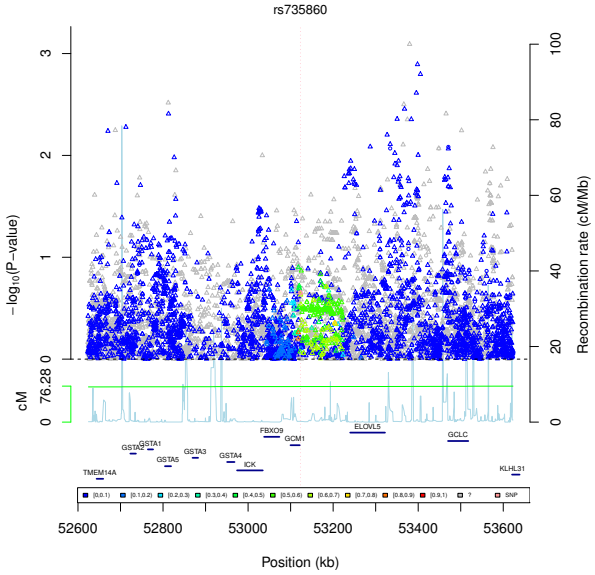

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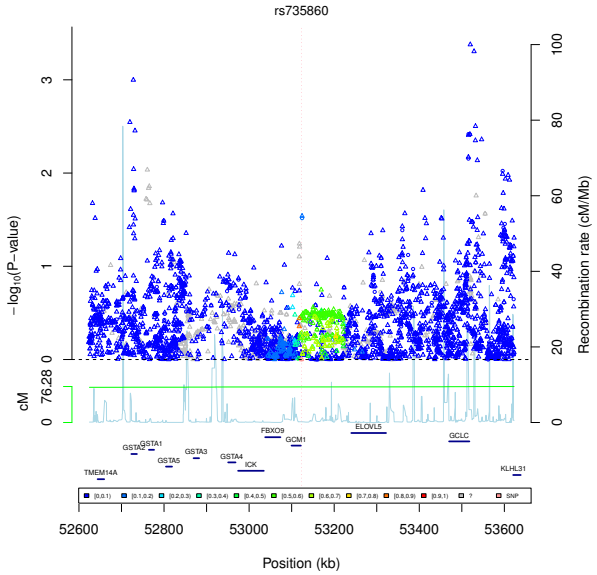

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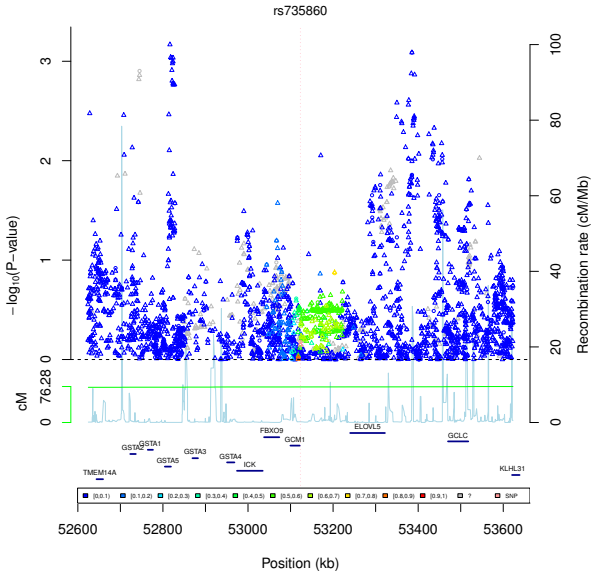
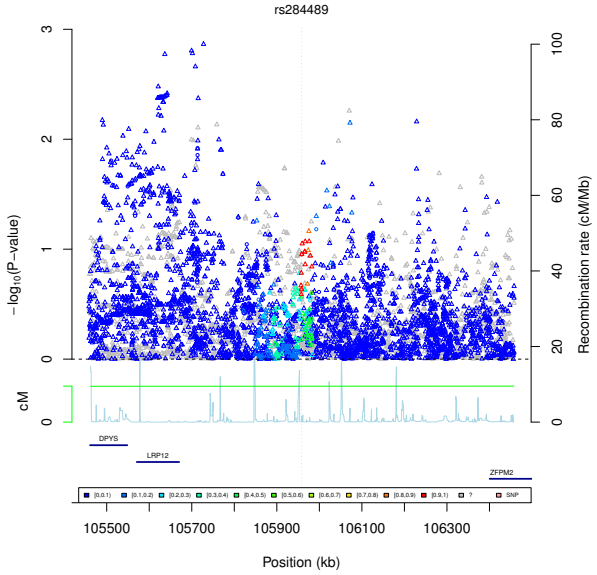
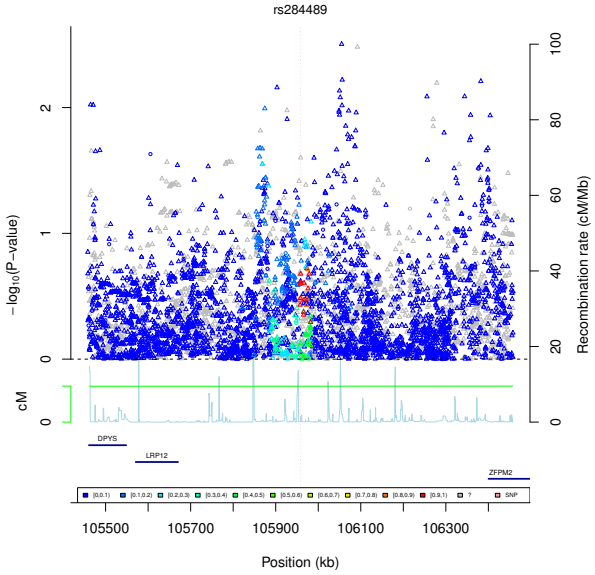


(iv) Hisp P, ASN LD

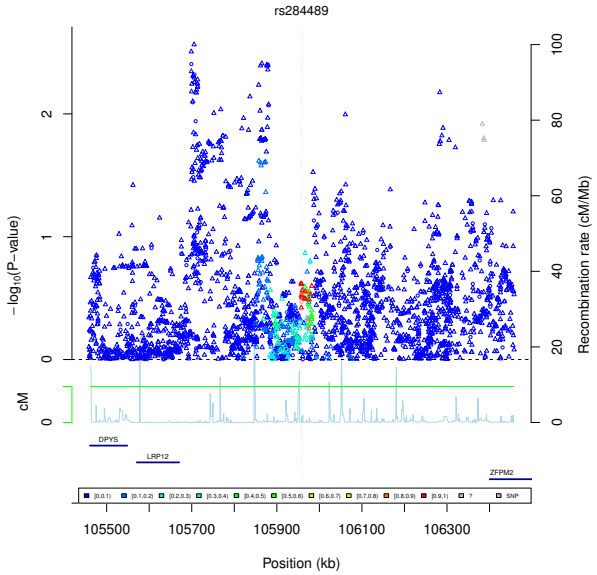
(p) rs284489 (Meta-analysis of White race/ethnicity [9])



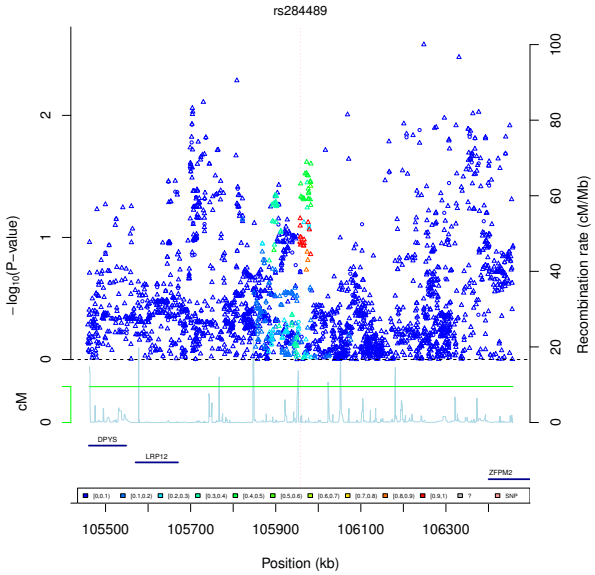
(i) AA I, EUR LD



(ii) AA P, EUR LD

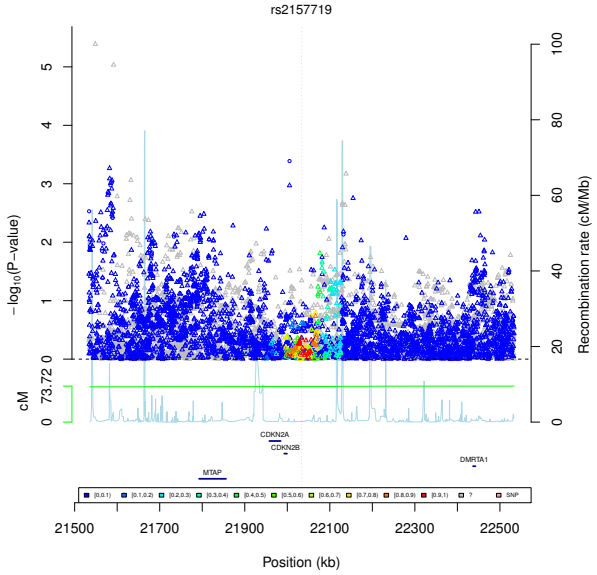


(iii) Hisp I, EUR LD



(iv) Hisp P, EUR LD

(q) rs2157719 (Meta-analysis of White ancestry [9])



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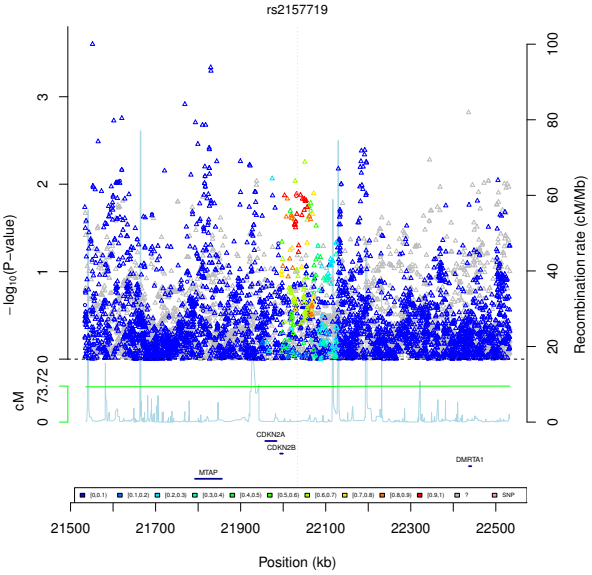
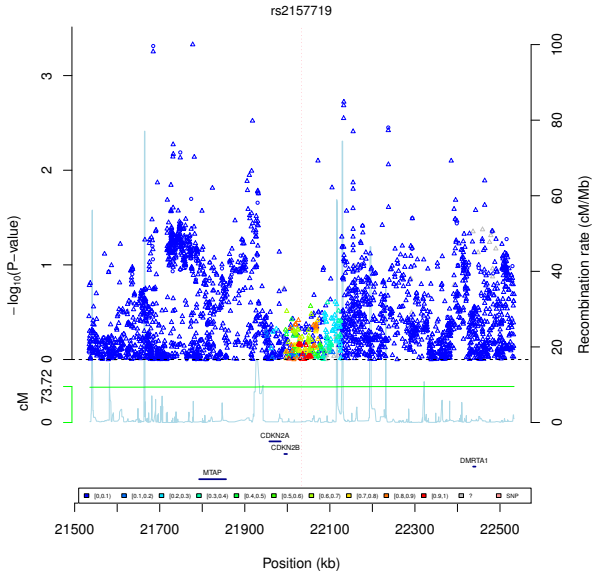
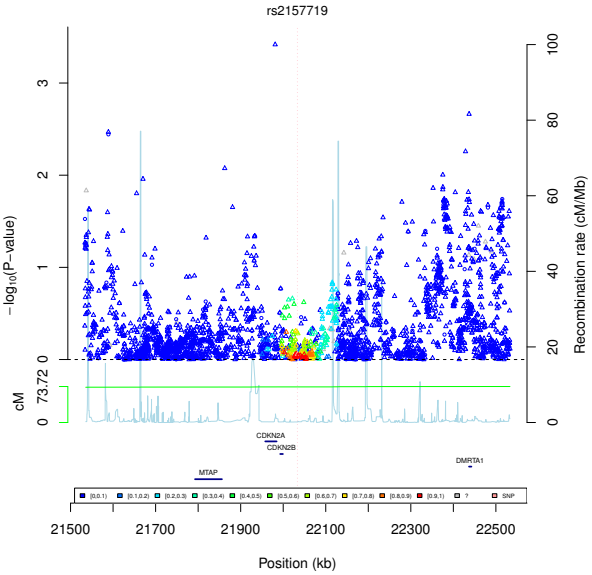


(ii) AA P, EUR LD

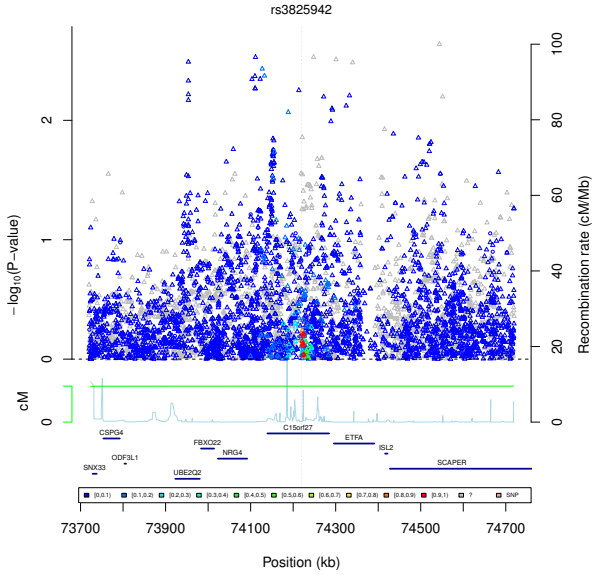


(iii) Hisp I, EUR LD



(iv) Hisp P, EUR LD

(r) rs3825942 (Glaucoma (Exfoliation) [6])



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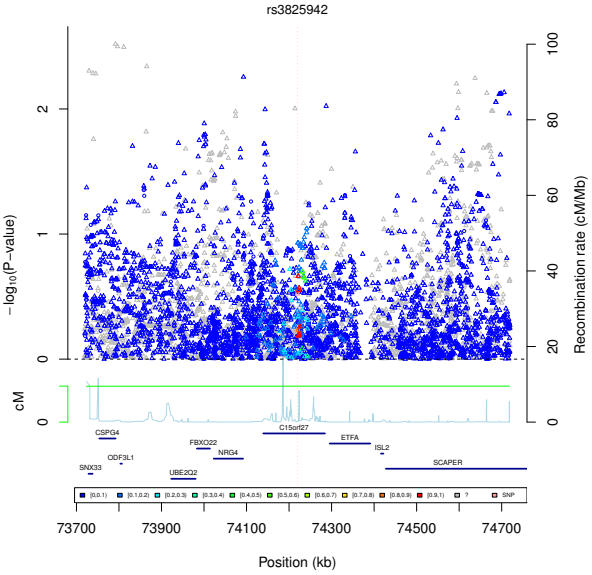

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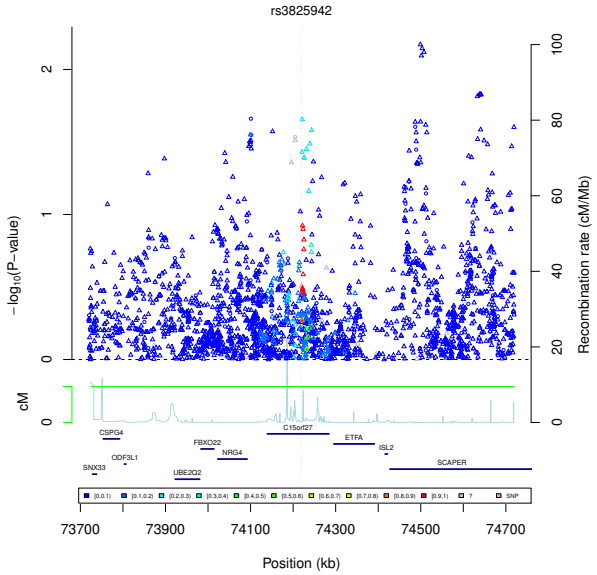
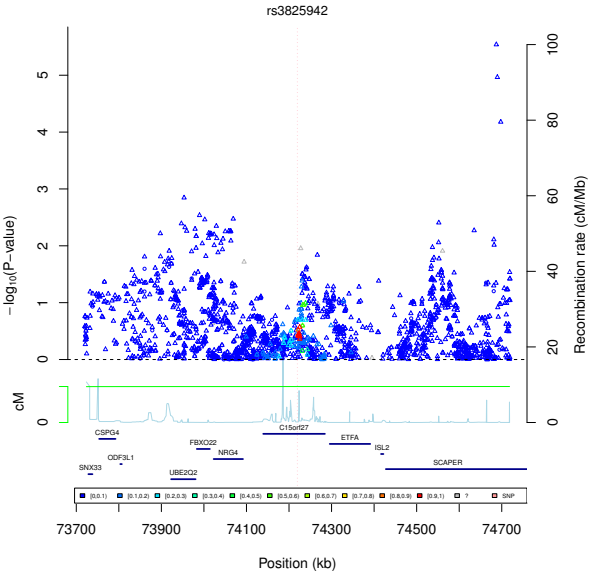


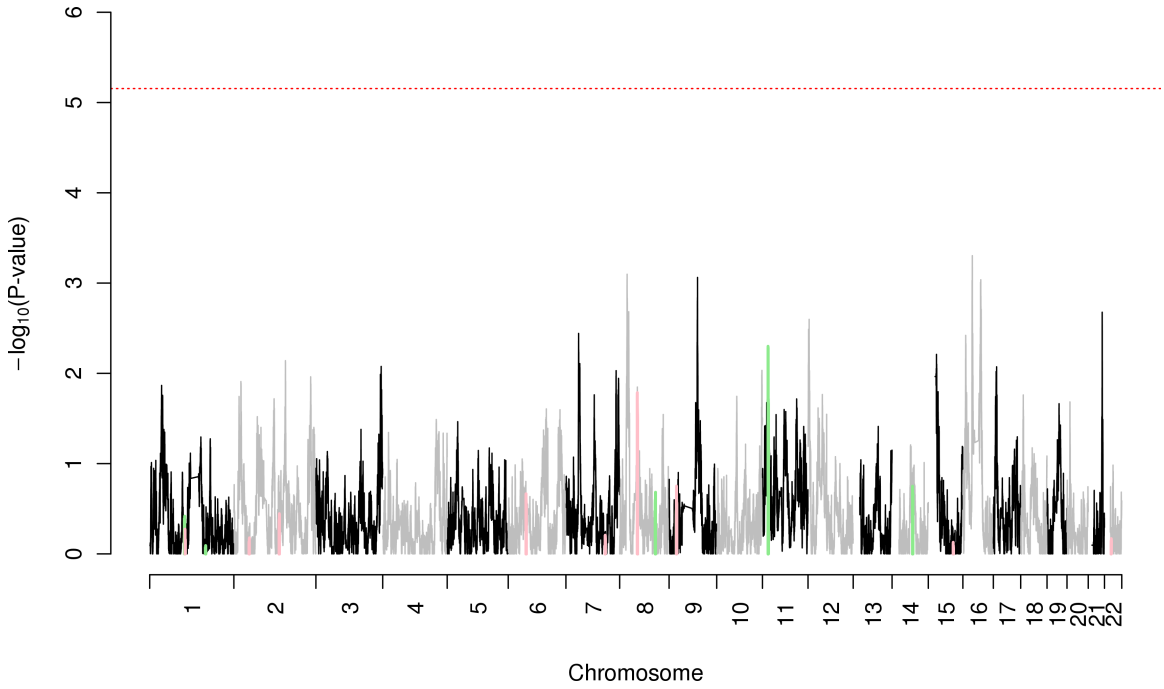
(iii) Hisp I, EUR LD



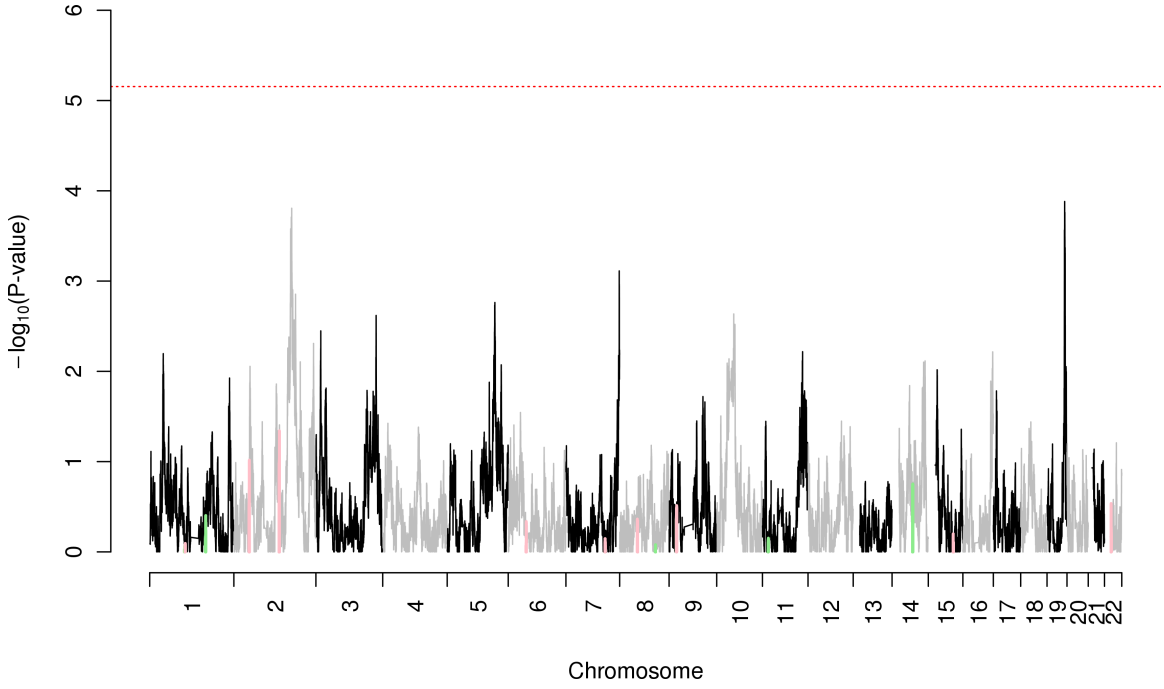
(iv) Hisp P, EUR LD

Figure S3: African American local ancestry plots. Pink lines indicate where a previous GWAS found an association with $p < 5 \times 10^{-8}$, whereas light green lines indicate where a previous GWAS found an association with $p > 5 \times 10^{-8}$. None of the points reach genome-wide significance of 7×10^{-6}.

(a) African American prevalent



(b) African American incident



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