

# SUPPLEMENTARY MATERIAL

## Identification of a novel locus associated with skin colour in African-admixed populations

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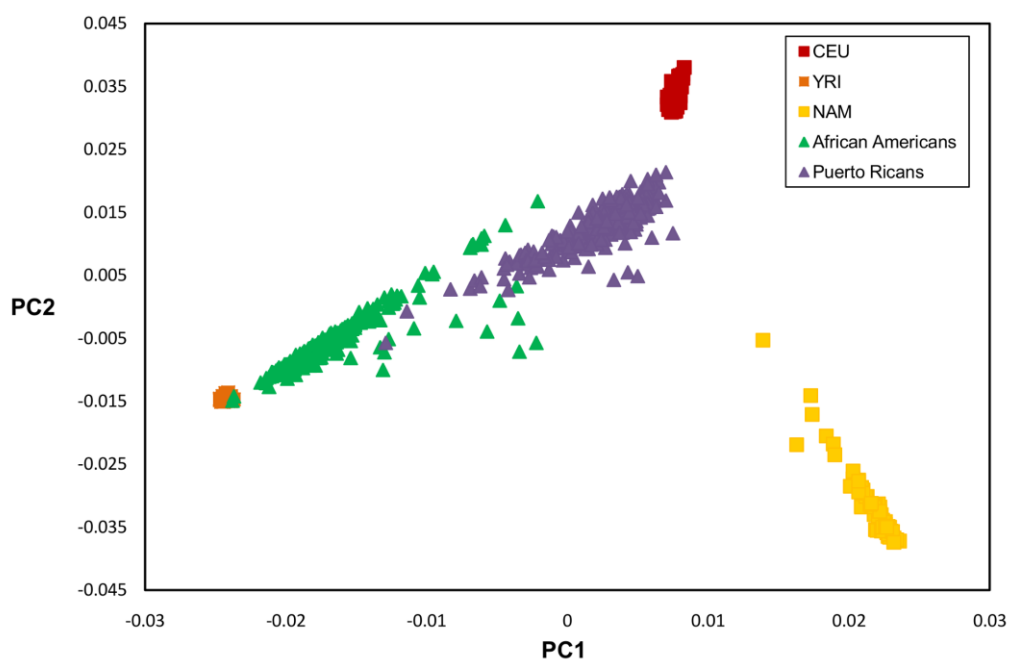
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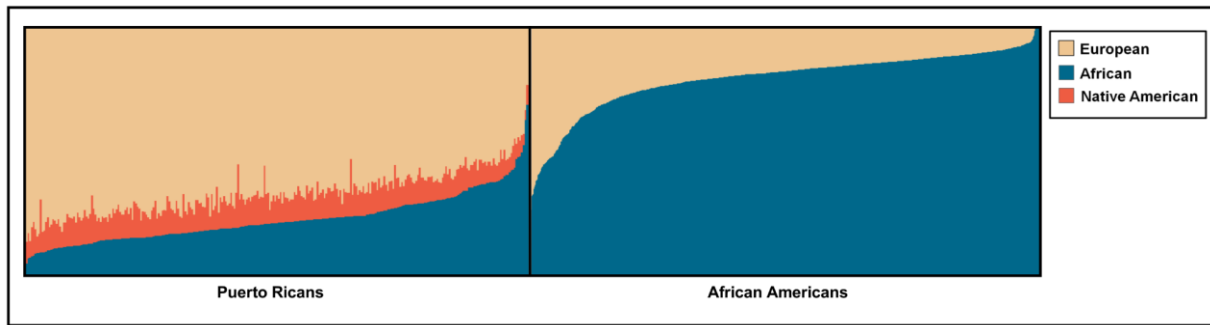
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<sup>16</sup>Centro de Neumología Pediátrica, San Juan, Puerto Rico.

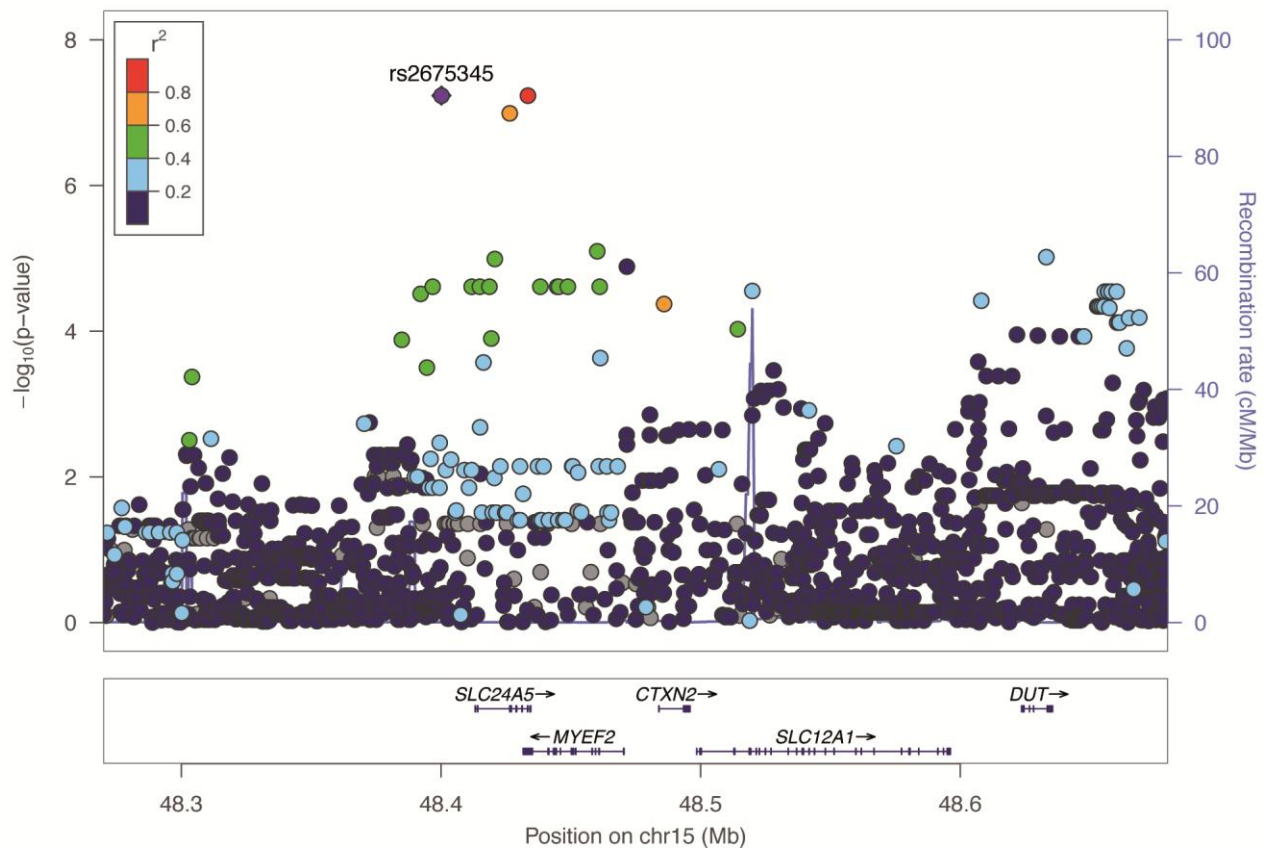
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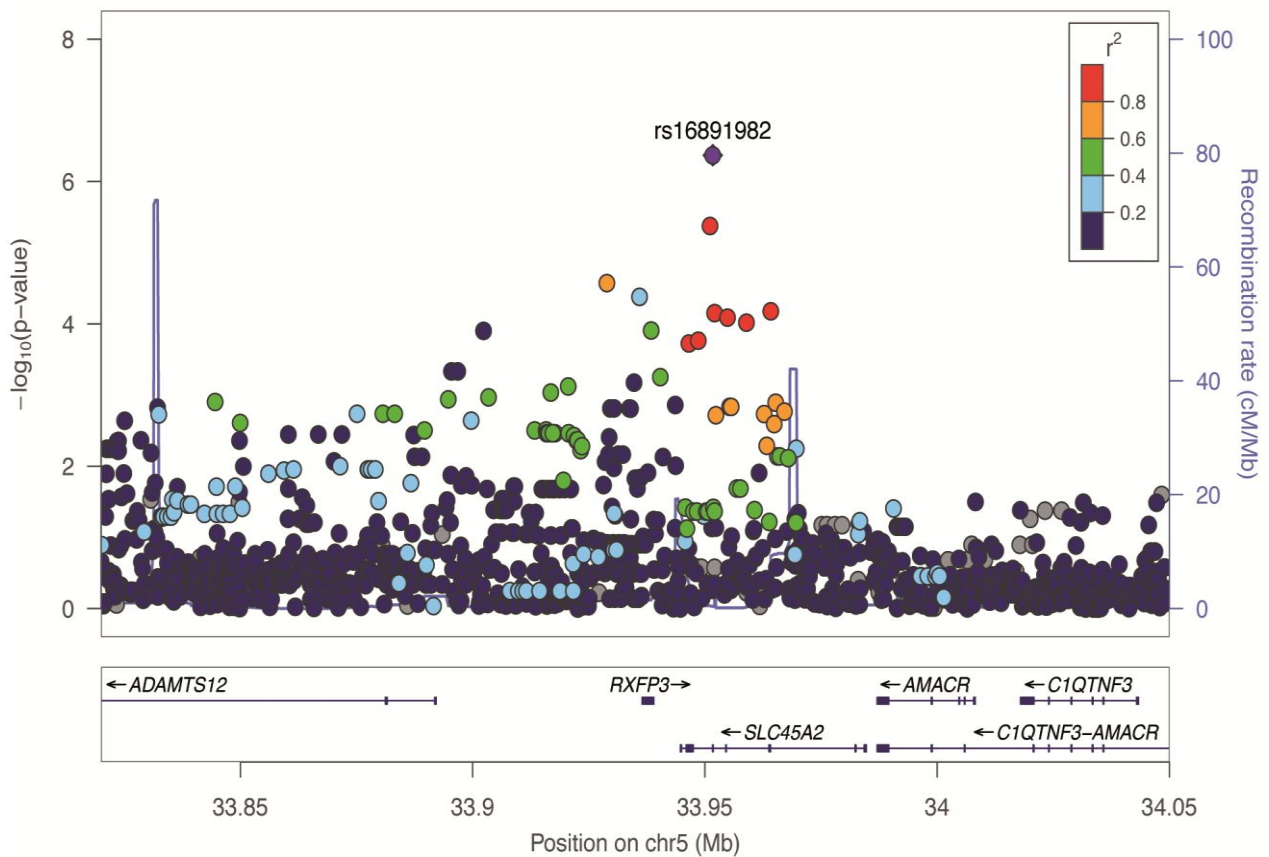
**Supplementary Figure S1. PCA plot showing the ancestral composition of the individuals included in the current study, represented by the two first principal components (PC1 and PC2).** Reference populations of European (CEU), African (YRI) and Native American (NAM) descent are shown along with the individuals included in discovery (Hispanics/Latinos from Puerto Rico) and replication (African Americans) stages.



**Supplementary Figure S2. Distribution of admixture proportions among Hispanics/Latinos from Puerto Rico and African American individuals.** Each subject is represented by one vertical bar and colours show ancestry components. The individuals have been sorted by their proportion of genetic ancestries.



**Supplementary Figure S3. Regional plot of association results for *SLC24A5* and surrounding genes in the discovery stage.** The transformed probability of association ( $-\log_{10} p\text{-value}$ ) is represented for each SNP as a dot by chromosome position. Top hit (rs2675345) is represented with a diamond and the remaining SNPs are colour coded based on their LD with this SNP, indicated by pairwise  $r^2$  values for the Admixed American populations (AMR) from the 1,000 Genomes Project.



**Supplementary Figure S4. Regional plot of association results for *SLC45A2* in the discovery stage.** The transformed probability of association results ( $-\log_{10} p\text{-value}$ ) is represented for each SNP as a dot by chromosome position. The top hit (rs16891982) is depicted by a diamond, and the remaining SNPs are colour coded based on their LD with this SNP, indicated by pairwise  $r^2$  values for the AMR populations of the 1000 Genomes Project.

**Supplementary Table S1.** Summary of SNPs with  $p$ -values  $\leq 1 \times 10^{-5}$  for association with melanin index.

SNP	Chromosome band	Position	A1/A2	Freq.(A1)	$\beta$ (95% CI)	$p$ -value
rs1432433	1q31.1	187918615	G/A	0.023	8.02 (4.64 to 11.41)	$5.24 \times 10^{-6}$
rs77443641	1q31.1	188000243	A/C	0.012	10.94 (6.42 to 15.46)	$3.29 \times 10^{-6}$
rs115105970	1q31.1	196942660	T/C	0.03	6.84 (4.07 to 9.61)	$2.20 \times 10^{-6}$
rs112725747	1q32.1	204344757	T/C	0.018	9.46 (5.68 to 13.25)	$1.65 \times 10^{-6}$
rs6659601	1p36.12	22124820	G/A	0.872	1.69 (0.18 to 3.20)	$2.58 \times 10^{-6}$
rs76327975	1q44	244364879	G/C	0.023	7.68 (4.35 to 11.01)	$9.24 \times 10^{-6}$
rs831984	2q12.3	108378250	A/C	0.298	2.59 (1.46 to 3.72)	$9.94 \times 10^{-6}$
rs831980	2q12.3	108380107	C/G	0.298	2.59 (1.46 to 3.72)	$9.94 \times 10^{-6}$
rs62170035	2q23.3	151111030	A/C	0.033	6.28 (3.64 to 8.93)	$5.07 \times 10^{-6}$
rs55767876	2p14	66820715	G/T	0.079	4.51 (2.6 to 6.42)	$5.68 \times 10^{-6}$
rs66871203	2p14	66821130	G/T	0.079	4.51 (2.6 to 6.42)	$5.68 \times 10^{-6}$
rs79592764	3q27.3	187398936	T/C	0.035	6.38 (3.74 to 9.02)	$3.47 \times 10^{-6}$
rs6788400	3p25.3	9886352	G/A	0.089	3.86 (2.19 to 5.52)	$8.89 \times 10^{-6}$
rs28479566	4p15.33	14776694	T/C	0.035	-6.35 (-9.09 to -3.61)	$8.05 \times 10^{-6}$
rs340417	5q35.3	178762064	A/C	0.111	-3.86 (-5.48 to -2.24)	$4.46 \times 10^{-6}$
rs153816	5q35.3	178763605	T/C	0.109	-3.85 (-5.48 to -2.23)	$5.24 \times 10^{-6}$
rs35397	5p13.2	33951116	T/G	0.511	-2.49 (-3.53 to -1.45)	$4.19 \times 10^{-6}$
rs16891982	5p13.2	33951693	G/C	0.523	-2.66 (-3.67 to -1.65)	$4.27 \times 10^{-7}$
rs77437330	6q21	111993747	A/G	0.072	4.45 (2.52 to 6.38)	$9.26 \times 10^{-6}$
rs73753762	6q15	92184362	G/A	0.014	10.3 (6.08 to 14.52)	$2.76 \times 10^{-6}$
rs77462788	7q31.32	122042978	C/T	0.021	8.08 (4.63 to 11.54)	$6.90 \times 10^{-6}$
rs116908038	7q31.32	122168537	C/A	0.021	8.08 (4.63 to 11.54)	$6.90 \times 10^{-6}$
rs7790204	7p14.2	36684481	C/T	0.549	-2.23 (-3.18 to -1.28)	$6.16 \times 10^{-6}$
rs7794780	7p14.2	36685144	C/T	0.542	-2.34 (-3.29 to -1.39)	$2.28 \times 10^{-6}$
rs12541402	8p22	15500967	C/T	0.172	-2.9 (-4.13 to -1.67)	$5.93 \times 10^{-6}$
rs6602665	10p13	13605982	C/T	0.079	4.72 (2.89 to 6.54)	$7.27 \times 10^{-7}$
rs6602666	10p13	13606490	G/A	0.079	4.72 (2.89 to 6.54)	$7.27 \times 10^{-7}$
rs151165649	10p12.1	25207241	A/G	0.019	9.42 (5.84 to 13.00)	$4.75 \times 10^{-7}$
rs141664730	10p12.1	25338228	C/G	0.016	9.83 (5.87 to 13.79)	$1.91 \times 10^{-6}$

rs149207584	10p12.1	25339373	C/T	0.016	9.83 (5.87 to 13.79)	1.91x10 <sup>-6</sup>
rs111256285	10q11.22	47632167	G/A	0.016	-8.43 (-12.08 to -4.79)	8.61x10 <sup>-6</sup>
rs7477798	10q11.22	47632478	C/A	0.016	-8.43 (-12.08 to -4.79)	8.61x10 <sup>-6</sup>
rs115019323	11q21	95895552	A/G	0.025	7.12 (4.13 to 10.12)	4.85x10 <sup>-6</sup>
rs144848699	11q21	95895953	C/T	0.025	7.12 (4.13 to 10.12)	4.85x10 <sup>-6</sup>
rs1902910	12q12	41778982	G/A	0.104	3.88 (2.3 to 5.46)	2.45x10 <sup>-6</sup>
rs17129378	12q12	41861386	C/T	0.104	3.99 (2.35 to 5.63)	3.05x10 <sup>-6</sup>
rs17578886	12q12	41862536	G/A	0.084	4.33 (2.57 to 6.09)	2.40x10 <sup>-6</sup>
rs10849455	12p13.33	785468	G/T	0.811	2.94 (1.68 to 4.21)	7.29x10 <sup>-6</sup>
rs138891280	13q33.2	106722485	G/A	0.021	8.06 (4.59 to 11.52)	7.85x10 <sup>-6</sup>
rs188019015	13q33.2	106743244	T/C	0.016	9.33 (5.35 to 13.31)	6.51x10 <sup>-6</sup>
rs146078872	13q33.2	106745894	G/A	0.016	9.33 (5.35 to 13.31)	6.51x10 <sup>-6</sup>
rs114396339	13q33.2	106746964	T/A	0.016	9.33 (5.35 to 13.31)	6.51x10 <sup>-6</sup>
rs727752	13q33.2	106754594	A/C	0.016	9.33 (5.35 to 13.31)	6.51x10 <sup>-6</sup>
rs2016565	13q33.2	106754788	C/T	0.016	9.33 (5.35 to 13.31)	6.51x10 <sup>-6</sup>
rs115940594	13q33.2	106754945	T/C	0.016	9.33 (5.35 to 13.31)	6.51x10 <sup>-6</sup>
rs3024737	13q34	113819785	G/A	0.012	11.26 (6.79 to 15.73)	1.37x10 <sup>-6</sup>
rs7326155	13q34	114460362	C/T	0.014	9.78 (5.58 to 13.99)	7.59x10 <sup>-6</sup>
rs10133804	14q32.12	92866905	C/T	0.011	1.44 (-3.58 to 6.45)	1.81x10 <sup>-6</sup>
rs11858919	15q12	26599133	T/C	0.025	7.59 (4.35 to 10.83)	6.48x10 <sup>-6</sup>
rs2675345	15q21.1	48400199	G/A	0.277	3.31 (2.14 to 4.47)	5.83x10 <sup>-6</sup>
rs1426654	15q21.1	48426484	G/A	0.312	3.06 (1.96 to 4.16)	1.02x10 <sup>-7</sup>
rs2470102	15q21.1	48433494	G/A	0.277	3.31 (2.14 to 4.47)	5.83x10 <sup>-8</sup>
rs8028919	15q21.1	48460188	A/G	0.793	-2.95 (-4.22 to -1.68)	7.99x10 <sup>-6</sup>
rs11637235	15q21.1	48633153	T/C	0.509	-2.26 (-3.24 to -1.28)	9.61x10 <sup>-6</sup>
rs2899446	15q21.2	50307416	G/A	0.544	2.28 (1.34 to 3.21)	2.73x10 <sup>-6</sup>
rs8033655	15q21.2	50308950	G/A	0.544	2.28 (1.34 to 3.21)	2.73x10 <sup>-6</sup>
rs7180182	15q21.2	50310295	G/A	0.544	2.28 (1.34 to 3.21)	2.73x10 <sup>-6</sup>
rs4580097	15q21.2	50315253	A/G	0.544	2.28 (1.34 to 3.21)	2.73x10 <sup>-6</sup>
rs28753701	15q22.1	59175467	T/C	0.963	-6.16 (-8.83 to -3.49)	9.23x10 <sup>-6</sup>
rs77045588	15q22.2	61136327	A/C	0.012	10.52 (6.01 to 15.02)	7.23x10 <sup>-6</sup>
rs78604138	15q22.2	61144845	G/A	0.012	10.52 (6.01 to 15.02)	7.23x10 <sup>-6</sup>

rs79617268	15q22.2	61145173	G/C	0.012	10.52 (6.01 to 15.02)	7.23x10 <sup>-6</sup>
rs532282237	15q22.2	61817211	C/T	0.011	12.51 (7.7 to 17.31)	6.14x10 <sup>-7</sup>
rs61310892	15q22.31	66319806	A/G	0.014	10.02 (5.8 to 14.25)	5.04x10 <sup>-6</sup>
rs7200304	16p13.12	13844197	C/T	0.916	4.2 (2.37 to 6.03)	9.99x10 <sup>-6</sup>
rs7193564	16p13.12	13844299	A/G	0.916	4.2 (2.37 to 6.03)	9.99x10 <sup>-6</sup>
rs7200773	16p13.12	13845690	A/G	0.916	4.2 (2.37 to 6.03)	9.99x10 <sup>-6</sup>
rs4141382	16p13.12	13845726	A/T	0.916	4.2 (2.37 to 6.03)	9.99x10 <sup>-6</sup>
rs7185574	16p13.12	13846274	T/A	0.916	4.2 (2.37 to 6.03)	9.99x10 <sup>-6</sup>
rs9926165	16p12.1	26958800	T/C	0.602	2.32 (1.32 to 3.32)	8.62x10 <sup>-6</sup>
rs9926268	16p12.1	26958967	T/C	0.600	2.32 (1.32 to 3.33)	8.48x10 <sup>-6</sup>
rs62029775	16p12.1	26959698	T/C	0.609	2.48 (1.48 to 3.48)	1.99x10 <sup>-6</sup>
rs117307642	17q12	33823098	T/C	0.042	6.42 (3.93 to 8.91)	7.94x10 <sup>-7</sup>
rs62143248	19q13.42	54364168	T/C	0.079	-4.72 (-6.63 to -2.81)	2.04x10 <sup>-6</sup>
rs62143250	19q13.42	54365667	A/G	0.079	-4.72 (-6.63 to -2.81)	2.04x10 <sup>-6</sup>
rs8102993	19q13.42	54366811	T/C	0.079	-4.68 (-6.59 to -2.78)	2.35x10 <sup>-6</sup>
rs11882947	19q13.42	54368893	A/G	0.074	-4.77 (-6.73 to -2.81)	3.00x10 <sup>-6</sup>
rs16985221	19q13.42	54374041	T/C	0.1	-3.92 (-5.6 to -2.24)	6.98x10 <sup>-6</sup>
rs62143251	19q13.42	54374699	A/G	0.096	-4.14 (-5.83 to -2.45)	2.55x10 <sup>-6</sup>
rs6142102	20q11.22	32704627	G/C	0.616	2.24 (1.28 to 3.2)	7.28x10 <sup>-6</sup>
rs755107	20q11.23	36662831	G/A	0.039	-5.5 (-7.88 to -3.11)	9.36x10 <sup>-6</sup>
rs201429679	22q12.2	31113081	T/G	0.032	6.91 (4.04 to 9.79)	3.92x10 <sup>-6</sup>



**Supplementary Table S2.** Results of the regression models in Hispanics/Latinos from Puerto Rico conditioned on the top hit of each chromosome region with genome-wide significant signals in the meta-analysis.

<b>Genomic region</b>	<b>SNP</b>	<b>Regression conditioned on</b>	<b>Original <i>p</i>-value of the SNP</b>	<b><i>p</i>-value conditional regression</b>
	rs1426654		1.02x10 <sup>-7</sup>	0.463
<i>SLC24A5, MYEF2, DUT, ATP8B4</i>	rs2470102	rs2675345	5.83x10 <sup>-8</sup>	NA <sup>a</sup>
	rs8028919		7.99x10 <sup>-6</sup>	0.826
	rs11637235		9.61x10 <sup>-6</sup>	0.112
<i>SLC45A2</i>	rs35397	rs16891982	4.19x10 <sup>-6</sup>	0.945
<i>BEND7-PRPF18</i>	rs6602666	rs6602665	7.27x10 <sup>-7</sup>	0.788

<sup>a</sup>The regression was not applicable due to the existence of co-linearity among rs2470102 and rs2675345 and high LD ( $R^2=1$ ).