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Supplemental Data

Characterization of Exome Variants and Their Metabolic Impact in 6,716 American Indians from the Southwest US

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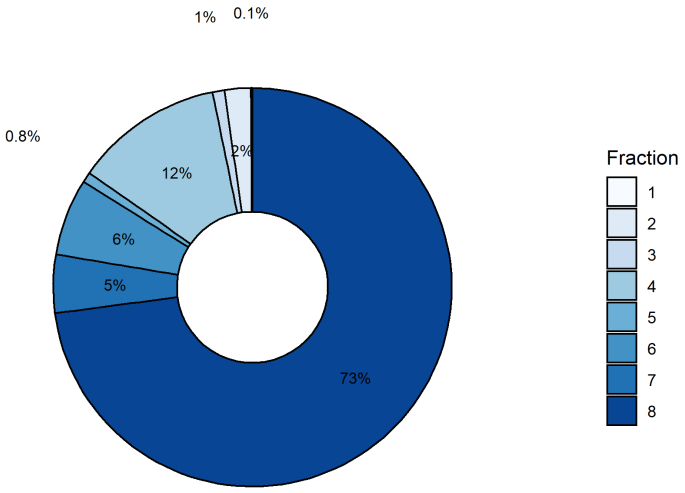
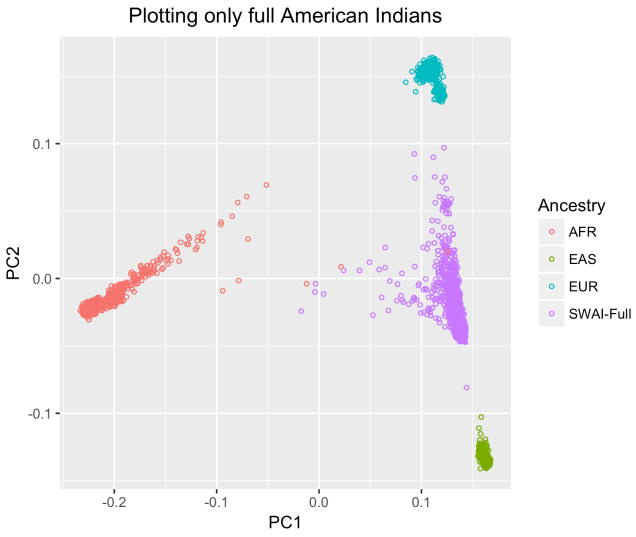
Figure S1. Population structure in the SWAI study

A

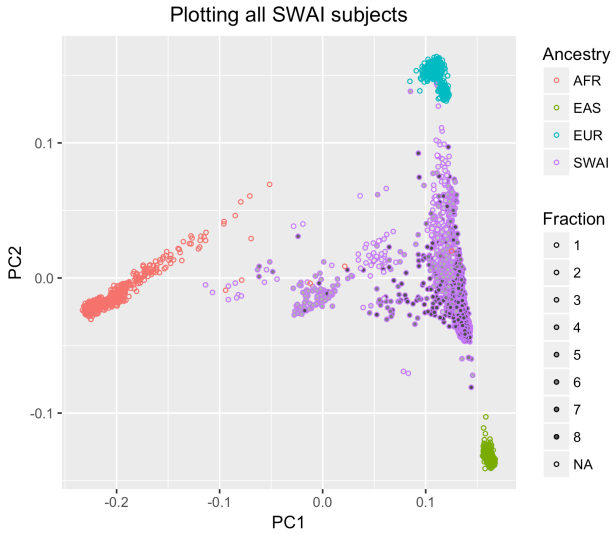
Fraction	Number	%
1	8	0.1
2	144	2.1
3	65	1.0
4	809	12.0
5	57	0.8
6	419	6.2
7	317	4.7
8	4897	72.9
Total	6716	100.0



B

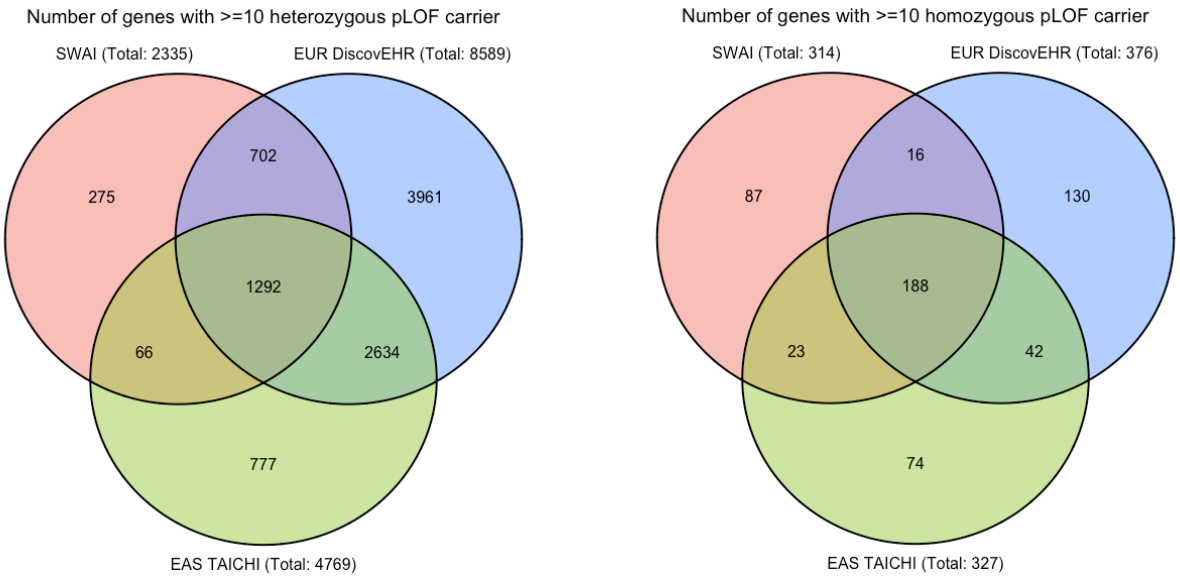


C



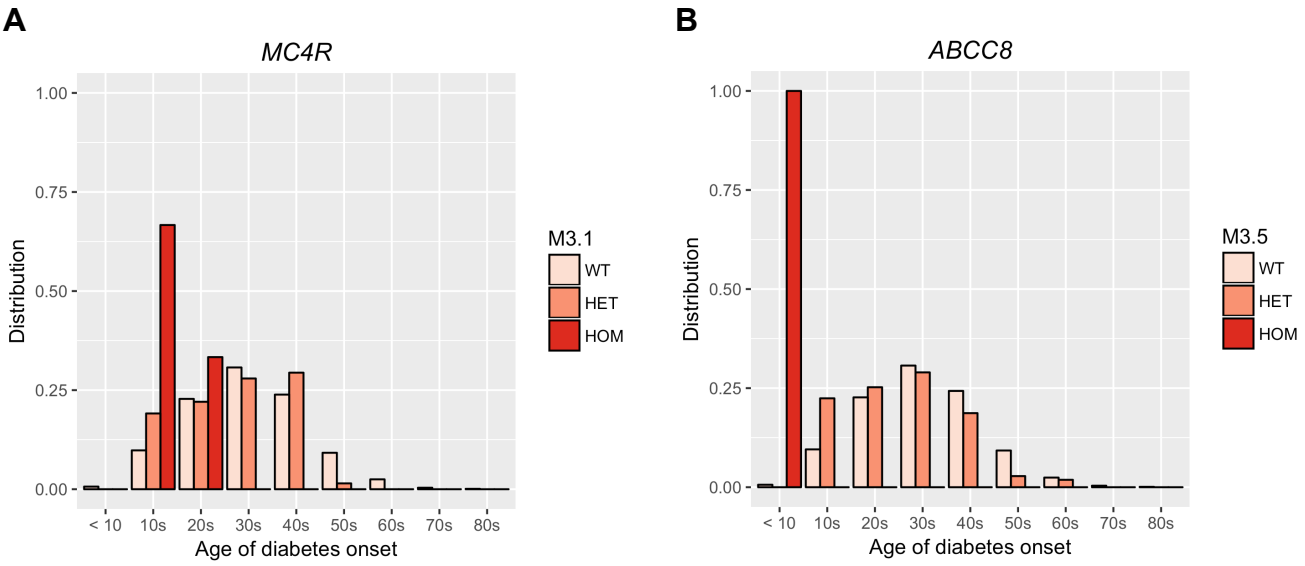
(A) The number and percentage of individuals according to the self-reported admixture (fraction of eight great grandparents that were American Indian). (B) Individuals with full self-reported American Indian ancestry in the SWAI study were projected onto the PC space calculated from African, East Asian, and European ancestries from the 1000 genomes project as reference. (C) All individuals from the SWAI study were projected onto the PC space with the fill intensity indicating the self-reported admixture.

Figure S2. Overlap of genes with at least 10 heterozygous or homozygous pLOF carriers among SWAI, European, and East Asian exomes



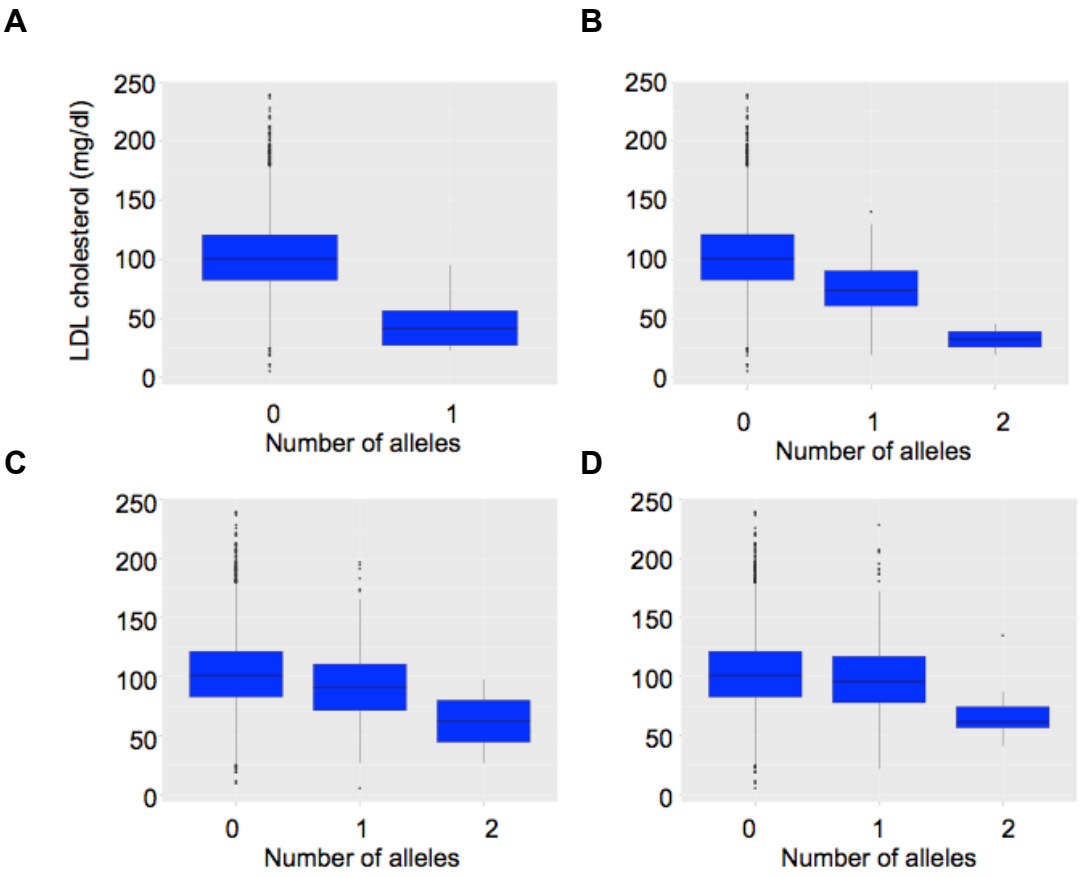
The Venn diagrams represent the overlap of the genes with at least 10 heterozygous (left) and homozygous (right) pLOF carriers in 6,716 SWAI, 29,575 European, and 13,947 East Asian exomes.

Figure S3. The distribution of T2D age of onset per genotype



The distribution of age of T2D onset per the genotype of (A) *MC4R* M3.1 mask and (B) *ABCC8* M3.5 mask.

Figure S4. Plasma LDL cholesterol levels per the genotype of SWAI-enriched variants in *APOB*, *APOE*, *PCSK9*, and *TM6SF2*



Variant	Genotypic counts	Genotypic means (stdev)	Genotypic medians
APOB p.Ala3175fs	5043 / 13 / 0	102.6 (30.34) / 44.9 (21.4) / -	100.4 / 41.6 / -
APOE p.Ala184Asp	4984 / 70 / 2	102.9 (30.4) / 76.4 (25.2) / 32.4 (18.7)	100.6 / 73.9 / 32.4
PCSK9 p.Gly244Asp	4823 / 231 / 2	103.0 (30.4) / 92.5 (30.5) / 62.1 (50.2)	100.8 / 90.6 / 62.1
TM6SF2 p.Arg138Trp	4591 / 457 / 8	103.0 (30.4) / 98.1 (31.0) / 70.5 (29.2)	100.8 / 95.6 / 61.4

Plasma LDL cholesterol levels per the genotype of (A) *APOB* p.Ala3175fs, (B) *APOE* p.Ala184Asp, (C) *PCSK9* p.Gly244Asp, and (D) *TM6SF2* p.Arg138Trp variants. The genotypic counts, means (with standard deviations), and medians are indicated in the table.

Table S2. Association of the individual variants in *MC4R* M3.1 mask with maximum body mass index

Gene Mask	Variant[a]	Variant effect	Amino acid change	Beta[b]	P-value	AAF[c]	Allele frequency ratio (AAF in full American Indians / AAF in respective populations)						
							GHS EUR	TAICHI EAS	gnomAD ALL	gnomAD AMR	gnomAD EUR	gnomAD EAS	gnomAD AFR
MC4R M3.1	18:60371857:G:C	missense	p.Arg165Gly	0.666	9.8E-04	0.0025	Inf	NC	Inf	Inf	Inf	Inf	Inf
	18:60372250:C:CT	frameshift	p.Gly34fs	0.622	1.0E-03	0.0023	Inf	Inf	Inf	Inf	Inf	Inf	Inf
	18:60371443:C:G	missense	p.Ala303Pro	0.881	1.0E-02	0.0008	Inf	Inf	Inf	Inf	Inf	Inf	Inf
	18:60371856:C:T	missense	p.Arg165Gln	0.353	2.0E-02	0.0044	132.9	125.3	188.2	Inf	102.1	82.6	Inf

[a] Variant is designated as chromosome:position:reference allele:alternate allele on GRCh38 reference.

[b] The unit is standard deviation of normalized traits.

[c] AAF: alternate allele frequency

The individual variants included in *MC4R* M3.1 mask with ≥10 alternate allele counts were tested for association with maximum body mass index in SWAI. The allele frequency ratios were derived by dividing the allele frequency in the full American Indians from the SWAI study by the allele frequencies in the European and East Asian individuals from DiscovEHR and TAICHI studies, and in the ancestral populations from the gnomAD exome database. When variants were not detected and fall outside of the consistently covered regions in a study, they are indicated as “not captured (NC)”.

Table S3. Association of the individual variants in *MC4R* M3.1 and *ABCC8* M3.5 masks with T2D

Gene Mask	Variant[a]	Variant effect	Amino acid change	OR[b]	P-value	AAF[c]	Allele frequency ratio (AAF in full American Indians / AAF in respective populations)						
							GHS EUR	TAICHI EAS	gnomAD ALL	gnomAD AMR	gnomAD EUR	gnomAD EAS	gnomAD AFR
MC4R M3.1	18:60372250:C:CT	frameshift	p.Gly34fs	3.04	7.90E-03	0.0021	Inf	Inf	Inf	Inf	Inf	Inf	Inf
	18:60371857:G:C	missense	p.Arg165Gly	2.92	1.61E-02	0.0024	Inf	NC	Inf	Inf	Inf	Inf	Inf
	18:60371856:C:T	missense	p.Arg165Gln	2.08	3.91E-02	0.0043	132.9	125.3	188.2	Inf	102.1	82.6	Inf
	18:60371443:C:G	missense	p.Ala303Pro	2.55	2.62E-01	0.0008	Inf	Inf	Inf	Inf	Inf	Inf	Inf
ABCC8 M3.5	11:17395658:C:T	missense	p.Arg1420His	2.23	1.53E-05	0.0162	489.2	115.3	701.8	107.0	Inf	Inf	Inf

[a] Variant is designated as chromosome:position:reference allele:alternate allele on GRCh38 reference.

[b] OR: odds ratio

[c] AAF: alternate allele frequency

The individual variants included in *MC4R* M3.1 and *ABCC8* M3.5 masks with ≥10 alternate allele counts were tested for association with T2D in SWAI. The allele frequency ratios were derived by dividing the allele frequency in the full American Indians from the SWAI study by the allele frequencies in the European and East Asian individuals from DiscovEHR and TAICHI studies, and in the ancestral populations from the gnomAD exome database. When variants were not detected and fall outside of the consistently covered regions in a study, they are indicated as “not captured (NC)”.

Table S4. Association of the individual variants in the masks of *APOB*, *APOE*, *PCSK9*, *TM6SF2*, *GPAM*, *LIPC*, and *CPT1A*, *GRB14*, and *APOC3* with plasma lipid levels

Gene Mask	Variant[a]	Variant effect	Amino acid change	TC		LDLC		HDLC		TG		AAF[c]	Allele frequency ratio (AAF in full American Indians / AAF in respective populations)						
				Beta[b]	P-value	Beta[b]	P-value	Beta[b]	P-value	Beta[b]	P-value		GHS EUR	TAICHI EAS	gnomAD ALL	gnomAD AMR	gnomAD EUR	gnomAD EAS	gnomAD AFR
APOB M4.5	2:21007344:GC:G	frameshift	p.Ala3175fs	-1.78	6.7E-10	-2.30	1.8E-13	0.50	1.5E-01	-1.03	4.2E-04	0.0012	Inf	Inf	Inf	Inf	Inf	Inf	Inf
	2:21006782:A:C	missense	p.Ile3362Met	-0.18	4.3E-03	-0.27	8.7E-05	0.06	4.4E-01	0.10	1.2E-01	0.0262	Inf	Inf	2324.7	320.2	Inf	Inf	Inf
	2:21011100:T:C	missense	p.His1923Arg	-0.51	5.2E-03	-0.66	1.1E-03	-0.04	8.4E-01	-0.12	5.1E-01	0.0031	0.0	4.3	0.0	0.1	0.0	4.2	0.1
	2:21026844:C:T	missense	p.Val730Ile	-0.28	6.5E-02	-0.27	1.1E-01	0.18	3.2E-01	-0.52	9.7E-04	0.0046	0.0	25.6	0.0	0.0	0.0	Inf	0.2
	2:21013525:C:T	missense	p.Arg1284Gln	-0.53	9.2E-02	NA	NA	-0.85	2.1E-02	NA	NA	0.0010	78.5	37.0	111.1	Inf	150.7	Inf	Inf
	2:21037212:G:A	missense	p.Thr194Met	-0.31	1.8E-01	-0.34	1.8E-01	0.23	4.1E-01	-0.55	2.4E-02	0.0019	21.1	0.0	0.4	0.6	11.6	0.0	4.4
	2:21015541:C:G	missense	p.Asp1113His	-0.30	2.1E-01	-0.40	1.2E-01	-0.25	3.7E-01	0.17	4.9E-01	0.0018	0.0	4.3	0.0	0.1	0.0	Inf	0.2
	2:21006931:G:T	missense	p.Leu3313Ile	-0.24	3.0E-01	-0.23	3.7E-01	0.21	4.4E-01	-0.36	1.3E-01	0.0019	Inf	Inf	8.7	13.2	Inf	Inf	0.6
	2:21010635:T:C	missense	p.Gln2078Arg	-0.24	3.0E-01	-0.23	3.7E-01	0.21	4.4E-01	-0.36	1.3E-01	0.0019	Inf	Inf	Inf	Inf	Inf	Inf	Inf
	2:21011704:C:T	missense	p.Asp1722Asn	0.07	5.7E-01	0.11	4.0E-01	0.03	8.3E-01	-0.18	1.2E-01	0.0074	93.0	54.8	329.4	90.6	446.8	Inf	Inf
	2:21007773:G:T	missense	p.Thr3032Asn	-0.14	5.9E-01	-0.37	1.8E-01	0.38	2.3E-01	-0.18	5.0E-01	0.0015	36.2	Inf	30.7	10.6	69.5	Inf	9.9
	2:21009973:C:G	missense	p.Asp2299His	-0.14	6.8E-01	-0.31	3.8E-01	0.25	5.2E-01	0.23	4.9E-01	0.0010	1.0	Inf	0.1	0.1	0.5	Inf	0.0
	2:21038062:G:A	missense	p.Pro145Ser	-0.08	7.3E-01	-0.09	7.1E-01	0.06	8.2E-01	-0.01	9.7E-01	0.0021	0.2	Inf	0.0	0.0	0.2	Inf	0.0
	2:21006988:A:G	missense	p.Ser3294Pro	-0.04	8.4E-01	-0.20	4.1E-01	0.00	9.9E-01	0.05	8.2E-01	0.0022	1.7	Inf	0.1	0.1	1.0	Inf	0.0
	2:21009501:G:T	missense	p.Ala2456Asp	-0.04	8.4E-01	-0.20	4.1E-01	0.00	9.9E-01	0.05	8.2E-01	0.0022	1.7	Inf	0.1	0.1	1.0	Inf	0.0
	2:21015135:G:T	missense	p.Leu1212Met	-0.04	8.6E-01	-0.04	8.7E-01	0.07	8.1E-01	-0.03	9.2E-01	0.0017	0.3	Inf	0.0	0.0	0.3	Inf	0.0
APOE M4.1	19:44908847:C:A	missense	p.Ala184Asp	-0.86	1.3E-13	-1.18	2.3E-20	0.02	8.7E-01	-0.20	9.0E-02	0.0073	Inf	NC	Inf	Inf	Inf	Inf	Inf
	19:44908822:C:T	missense	p.Arg176Cys	-0.46	1.5E-03	-0.69	2.4E-05	-0.05	7.8E-01	-0.04	7.8E-01	0.0049	0.0	0.1	0.0	0.1	0.0	0.0	0.0
	19:44908969:G:A	missense	p.Ala225Thr	0.08	7.7E-01	0.13	6.6E-01	-0.15	6.4E-01	0.08	7.8E-01	0.0011	Variant not present in full American Indians						
PCSK9 M2.5	1:55052723:G:A	missense	p.Gly244Asp	-0.25	2.0E-04	-0.46	4.7E-10	0.09	2.4E-01	0.18	1.0E-02	0.0237	Inf	Inf	2408.4	Inf	Inf	Inf	Inf
	1:55043951:G:A	missense	p.Gly106Arg	-0.59	6.3E-03	-0.91	1.6E-04	0.12	6.4E-01	-0.13	5.6E-01	0.0022	Inf	Inf	Inf	Inf	Inf	Inf	Inf

	1:55039995:C:T	missense	p.Ala53Val	-0.22	5.0E-02	-0.25	4.1E-02	0.12	3.5E-01	-0.14	2.1E-01	0.0083	0.0	0.0	0.0	0.1	0.0	0.0	0.1
	1:55043922:G:A	missense	p.Arg96His	-0.16	8.6E-02	-0.22	4.4E-02	0.05	6.7E-01	-0.11	2.6E-01	0.0114	Inf	Inf	1602.8	Inf	Inf	234.7	Inf
	1:55039974:G:T	missense	p.Arg46Leu	-0.45	1.3E-01	-0.34	3.0E-01	-0.67	5.7E-02	0.20	5.2E-01	0.0011	0.0	Inf	0.0	0.1	0.0	Inf	0.2
	1:55058129:A:G	missense	p.Asn425Ser	-0.41	1.6E-01	-0.45	1.6E-01	0.08	8.0E-01	0.06	8.3E-01	0.0012	Variant not present in full American Indians						
	1:55052746:G:A	missense	p.Val252Met	0.38	1.7E-01	0.66	4.2E-02	-0.42	2.0E-01	0.17	5.8E-01	0.0014	0.8	Inf	68.2	Inf	76.4	Inf	11.0
	1:55058549:C:T	missense	p.Arg469Trp	0.28	3.0E-01	0.40	1.9E-01	-0.66	4.2E-02	0.48	9.2E-02	0.0013	24.2	Inf	1.4	3.5	30.9	Inf	0.1
	1:55061388:C:G	missense	p.His565Gln	0.21	3.5E-01	0.44	8.8E-02	0.00	9.9E-01	-0.17	4.9E-01	0.0020	1.1	Inf	Variant failed QC				
	1:55058550:G:A	missense	p.Arg469Gln	0.11	6.8E-01	0.37	2.0E-01	-0.04	9.0E-01	0.11	6.8E-01	0.0014	Inf	Inf	436.3	Inf	Inf	31.9	Inf
	1:55058182:G:A	missense	p.Ala443Thr	-0.05	7.9E-01	-0.15	4.9E-01	0.16	4.9E-01	0.04	8.5E-01	0.0026	0.3	0.7	0.0	0.0	0.4	0.1	0.0
TM6SF2 M4.5	19:19269759:G:A	missense	p.Arg138Trp	-0.27	3.3E-08	-0.20	1.2E-04	0.10	7.0E-02	-0.33	2.5E-11	0.0461	2843.9	Inf	31.6	4.4	Inf	432.3	Inf
	19:19268740:C:T	missense	p.Glu167Lys	-0.23	1.4E-03	-0.27	6.4E-04	-0.06	4.7E-01	-0.15	4.3E-02	0.0201	0.2	0.2	0.3	0.4	0.2	0.2	0.5
	19:19269704:A:G	missense	p.Leu156Pro	-0.08	7.5E-01	0.12	6.4E-01	-0.09	7.6E-01	-0.21	4.1E-01	0.0017	0.2	10.7	0.1	0.3	0.1	Inf	0.9
GPAM M3.5	10:112159980:G:T	missense	p.Ser611Arg	0.19	3.2E-03	0.11	1.4E-01	0.57	3.8E-14	-0.17	9.0E-03	0.0250	Inf	383.1	Inf	Inf	Inf	Inf	Inf
LIPC M4.1	15:58548359:G:A	missense	p.Glu280Lys	0.01	9.5E-01	-0.19	1.6E-01	0.38	9.4E-03	0.14	2.9E-01	0.0068	386.5	Inf	410.0	226.0	370.3	120.1	Inf
	15:58541878:A:G	missense	p.Ile123Val	-0.35	7.6E-02	-0.35	1.1E-01	0.60	1.1E-02	-0.62	2.3E-03	0.0026	Inf	NC	Inf	Inf	Inf	Inf	Inf
	15:58548541:C:G	missense	p.Phe340Leu	-0.07	7.6E-01	-0.05	8.3E-01	0.24	3.5E-01	-0.25	2.6E-01	0.0021	0.8	Inf	Inf	Inf	Inf	Inf	Inf
CPT1A M2.1	11:68773378:C:T	missense	p.Asp543Asn	0.16	2.4E-01	0.23	1.2E-01	-0.54	6.6E-04	0.07	5.9E-01	0.0058	Inf	Inf	Inf	Inf	Inf	Inf	Inf
	11:68794860:C:T	missense	p.Ala275Thr	-0.05	7.6E-01	0.00	1.0E+00	-0.62	1.1E-03	0.24	1.5E-01	0.0039	0.0	3.4	0.0	0.1	0.0	22.5	0.1
	11:68780662:G:A	missense	p.Pro479Leu	-0.05	8.6E-01	-0.25	4.4E-01	-0.54	9.7E-02	-0.23	4.5E-01	0.0014	Inf	Inf	51.4	28.3	Inf	Inf	Inf
	11:68807515:C:G	missense	p.Trp135Cys	0.21	4.8E-01	0.62	5.5E-02	-0.44	2.1E-01	-0.01	9.9E-01	0.0011	Inf	Inf	Inf	Inf	Inf	Inf	Inf
GRB14 M2.1	2:164525023:G:T	missense	p.Ser220Tyr	0.05	7.1E-01	0.17	2.2E-01	-0.76	2.2E-07	0.21	1.1E-01	0.0068	Inf	Inf	1375.3	185.0	Inf	Inf	Inf
	2:164525032:C:A	missense	p.Gly217Val	-0.23	3.1E-01	-0.18	4.7E-01	-0.03	9.2E-01	-0.21	3.7E-01	0.0020	0.0	Inf	0.0	0.1	0.0	Inf	0.2
APOC3 M4.5	11:116830844:G:A	missense	p.Ala43Thr	-0.35	2.7E-08	-0.24	6.3E-04	0.74	8.5E-23	-1.17	1.1E-71	0.0259	122.1	403.1	33.3	33.3	155.2	265.2	11.1

[a] Variant is designated as chromosome:position:reference allele:alternate allele on GRCh38 reference.

[b] The unit is standard deviation of normalized traits.

[c] AAF: alternate allele frequency

The individual variants included in the masks with ≥10 alternate allele counts were tested for association with plasma lipid levels in SWAI. The allele frequency ratios were derived by dividing the allele frequency in the full American Indians from the SWAI study by the allele frequencies in the European and East Asian individuals from DiscovEHR and TAICHI studies, and in the ancestral populations from the gnomAD exome database. When variants were not detected and fall outside of the consistently covered regions in a study, they are indicated as “not captured (NC)”.

Table S5. Gene-burden associations of genes within ±250kb window of GWAS sentinel variants when the genes with significant gene-burden associations in SWAI were not the closest genes to the GWAS sentinel variants

Associations from European GWAS							Gene-burden associations in SWAI				
Sentinel variant	Closest gene	Variant effect	AAF[a]	Trait	Beta[b]	P-value	Gene	Top mask	Freq	Beta[b]	P-value
rs622082	*IGHMBP2*	missense	0.31	HDLC	-0.02	5.9E-10	***CPT1A***	**M2.1**	**0.014**	**-0.50**	**1.3E-06**
							GAL	M2.1	0.004	-0.41	2.3E-02
							MRPL21	M4.1	0.006	-0.27	8.7E-02
							MRGPRD	M4.5	0.045	0.08	1.5E-01
							TPCN2	M2.5	0.021	0.11	1.8E-01
							MRGPRF	M2.1	0.001	0.40	3.0E-01
							IGHMBP2	M4.1	0.004	-0.13	4.7E-01
							TESMIN	M2.1	0.001	0.17	6.4E-01
rs12328675	*COBLL1*	3' UTR	0.12	HDLC	0.05	3.1E-37	***GRB14***	**M2.1**	**0.013**	**-0.44**	**1.8E-05**
							COBLL1	M4.1	0.006	0.22	1.5E-01
							SLC38A11	M2.1	0.010	0.07	5.6E-01
rs964184	*ZPR1*	3' UTR	0.85	HDLC	0.11	2.6E-217	***APOC3***	**M4.5**	**0.026**	**0.74**	**6.0E-23**
							BUD13	M2.1	0.005	-0.38	2.1E-02
							APOA4	M2.5	0.052	-0.11	5.1E-02
							APOA5	M2.1	0.002	-0.40	1.6E-01
							SIK3	M2.1	0.006	0.13	4.1E-01
							ZPR1	M4.1	0.003	0.08	7.2E-01
							APOA1	M2.1	0.003	0.05	8.2E-01

[a] AAF: alternate allele frequency

[b] The unit is standard deviation of normalized traits.

Significant associations (P-value < 2.4×10^{-5}) are bolded.

Table S6. The frequency and trait association of the GWAS sentinel variants in SWAI and gene-burden associations before and after conditioning on the GWAS sentinel variants

Sentinel variant	Closest gene	Variant effect	Trait	Association of sentinel variants in original GWAS			Association of sentinel variants in SWAI			Association of gene-burden in SWAI						
				AAF[a]	Effect[b]	P-value	AAF[a]	Effect[b]	P-value	Gene	Top mask	Freq	Without conditioning		Upon conditioning on sentinel variant	
													Effect[b]	P-value	Effect[b]	P-value
rs6567160	*MC4R*	intergenic	BMI MaxBMI	0.230	0.06	1.8E-178	0.016	-0.04	6.3E-01	*MC4R*	M3.1	0.011	0.56	5.2E-09		
rs523288	*MC4R*	intergenic	T2D	0.238	1.05	7.6E-13	0.024	0.27	4.2E-02	*MC4R*	M3.1	0.010	2.62	1.2E-05		
rs67254669	*ABCC8*	missense	T2D	0.001	1.89	1.1E-08	Variant not detected			*ABCC8*	M3.5	0.018	2.21	9.3E-06		
rs541041	*APOB*	intergenic	TC	0.810	0.11	5.3E-237	0.950	0.22	2.5E-06	*APOB*	M4.5	0.062	-0.21	1.4E-06	-0.24	2.1E-07
			LDLC		0.12	1.3E-287		0.23	8.4E-07				-0.29	1.6E-09	-0.28	8.1E-10
rs445925	*APOE*	downstream	TC	0.110	-0.21	0	0.002	-0.57	8.2E-03	*APOE*	M4.1	0.014	-0.63	9.4E-14		
			LDLC		-0.32	0	0.002	-0.62	5.9E-03				-0.86	4.7E-20		
rs11591147	*PCSK9*	missense	TC	0.015	-0.41	0	0.001	-0.45	1.3E-01	*PCSK9*	M2.5	0.057	-0.20	6.2E-06		
			LDLC		-0.48	0		-0.34	3.0E-01		M3.5	0.028	-0.44	9.1E-11		
rs58542926	*TM6SF2*	missense	TC	0.074	-0.13	7.0E-155	0.020	-0.23	1.4E-03	*TM6SF2*	M4.5	0.067	-0.26	4.6E-10		
			LDLC		-0.10	6.5E-93		-0.27	6.4E-04				-0.22	7.9E-07	-0.14	5.4E-03
			TG		-0.12	3.7E-125		-0.15	4.3E-02				-0.28	2.5E-11		
rs2792751	*GPAM*	missense	HDLC	0.730	-0.03	3.8E-21	0.660	-0.13	1.2E-07	*GPAM*	M3.5	0.026	0.58	5.1E-15	0.33	1.1E-06
rs1800588	*LIPC*	upstream	HDLC	0.240	0.12	0	0.810	0.08	4.1E-03	*LIPC*	M4.1	0.013	0.47	9.8E-06	0.33	4.8E-04
rs622082	*IGHMBP2*	missense	HDLC	0.31	-0.02	5.90E-10	0.022	-0.01	9.2E-01	*CPT1A*	M2.1	0.014	-0.50	1.3E-06		
rs12328675	*COBLL1*	3' UTR	HDLC	0.12	0.05	3.1E-37	Variant not detected			*GRB14*	M2.1	0.013	-0.46	1.8E-05		
rs964184	*ZPR1*	3' UTR	TC	0.85	-0.09	4.7E-135	Variant not detected			*APOC3*	M4.5	0.026	-0.35	3.1E-08		
			HDLC		0.11	2.6E-217							0.74	6.0E-23		
			TG		-0.25	0							-1.16	7.5E-72		

[a] AAF: alternate allele frequency

[b] For quantitative traits: beta coefficients in standard deviation unit of normalized traits. For binary traits: odds ratio (OR).