Supplementary file

Splicing QTL of human adipose-related traits

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Supplementary Table S1. HaploReg information of 11 top SNPs of adipose GWAS (PMID: 27918534)

Chr.	SNP ID	Pos. (bp, hg38)	Pos. (bp, hg19)	Ref. allele	Alt. allele	Minor allele frequency				RefSeq gene
						AFR	AMR	ASN	EUR	
chr1	rs6587515	150636412	150608888	G	A	0	0.04	0.18	0.08	6.8kb 5' of *ENSA*
chr1	rs6689335	219455340	219628682	T	C	0.03	0.32	0.30	0.49	242kb 3' of *LYPLAL1*
chr2	rs10198628	12824371	12964497	A	G	0.6	0.51	0.84	0.55	82kb 3' of *TRIB2*
chr3	rs7374732	23161963	23203454	C	T	0.93	0.72	0.65	0.65	41kb 5' of *UBE2E2*
chr5	rs10060123	126348218	125683910	C	A	0.12	0.20	0.20	0.26	12kb 5' of *GRAMD3*
chr5	rs1650505	158602726	158029734	G	A	0.22	0.28	0.24	0.20	93kb 3' of *EBF1*
chr6	rs912056	6735964	6736197	A	T	0.82	0.56	0.25	0.64	81kb 3' of *LY86*
chr6	rs2842895	7106083	7106316	G	C	0.04	0.46	0	0.56	1.5kb 5' of *RREB1*
chr6	rs2237199	16429790	16430021	G	A	0.18	0.17	0.37	0.12	*ATXN1*
chr16	rs7185735	53788739	53822651	A	G	0.54	0.28	0.16	0.42	*FTO*
chr17	rs2123685	39897636	38053889	T	C	0.01	0.03	0	0.04	7kb 3' of *GSDMB*

AFR: African. AMR: Ad Mixed American. ASN: Asians. EUR: European.

Supplementary Table S2. Splicing QTL for adipose traits

SNP ID	Position (bp)	GWAS_pval	Splicing feature									Distance (bp)
			Trait-Tissue	Gene	Exon x1	Exon x2	Junction Coordinate	Direction	Junction	sQTL_pval	RefGene ID	
rs17817288	53807764	9.50E-09	SAT_OVERALL-AdiposeSubcutaneous	FTO	Exon 5/9	Exon 8/9	53907777-53967942	fwd	Novel	6.92E-04	NM_001080432	100,013
rs17817288	53807764	9.50E-09	SAT_OVERALL-ArteryTibial	AKTIP	Truncated Exon 5/10	Exon 10/10	53526431-53529017	fwd	Novel	5.52E-05	NM_001012398	281,333
rs7206790	53797908	4.20E-09	SAT_OVERALL-Thyroid	AKTIP	Exon 9/10	Truncated Exon 6/10	53526665-53528483	rev	Novel	7.31E-04	NM_001012398	271,243
rs7206790	53797908	4.20E-09	SAT_OVERALL-Thyroid	AKTIP	Exon 9/10	Exon 6/10	53526665-53528429	rev	Novel	5.73E-05	NM_001012398	271,243
rs11710420	23221931	3.20E-08	VATSAT_OVERALL-WholeBlood	UBE2E1	Exon 3/6	Exon 4/6	23853001-23929058	rev	Known	9.19E-04	NM_003341	631,070
rs11710420	23221931	3.20E-08	VATSAT_OVERALL-WholeBlood	UBE2E1	Exon 2/6	Exon 4/6	23848912-23929058	rev	Known	4.09E-04	NM_003341	626,981
rs2237199	16430271	1.40E-08	SATHU_MEN-ArteryTibial	DTNBP1	Exon4/10	Exon6/10	15615630-15637975	fwd	Novel	8.30E-04	NM_001271667	814,641

SAT, subcutaneous adipose tissue volume; VATSAT, ratio of visceral-to-subcutaneous adipose tissue volume; SATHU, Subcutaneous adipose tissue attenuation.

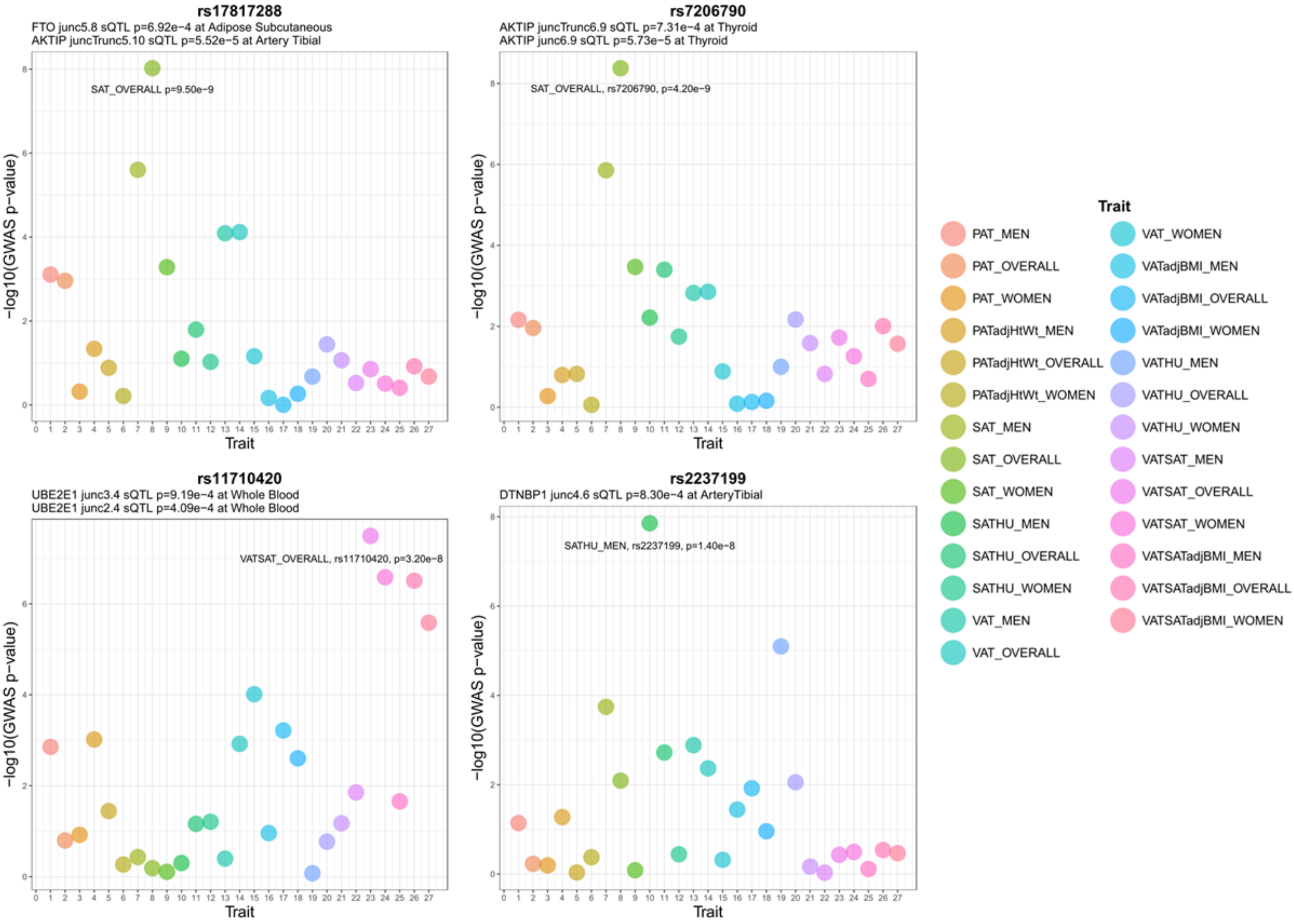
Distance: base pair from SNP to junction.

Supplementary Table S3. Four splicing QTL GWAS p-values for body fat distribution traits

Trait	Adipose GWAS p-value			
	rs17817288	rs7206790	rs11710420	rs2237199
PAT_MEN	7.80E-04	0.007	0.001	0.072
PAT_OVERALL	0.001	0.011	0.160	0.590
PAT_WOMEN	0.480	0.530	0.120	0.640
PATadjHtWt_MEN	0.046	0.160	9.60E-04	0.053
PATadjHtWt_OVERALL	0.130	0.150	0.036	0.920
PATadjHtWt_WOMEN	0.610	0.870	0.540	0.420
SAT_MEN	2.50E-06	1.40E-06	0.370	1.80E-04
SAT_OVERALL	9.50E-09	4.20E-09	0.650	0.008
SAT_WOMEN	5.20E-04	3.40E-04	0.780	0.820
SATHU_MEN	0.079	0.006	0.500	1.40E-08
SATHU_OVERALL	0.016	4.00E-04	0.069	0.002
SATHU_WOMEN	0.094	0.018	0.062	0.360
VAT_MEN	8.10E-05	0.002	0.400	0.001
VAT_OVERALL	7.60E-05	0.001	0.001	0.004
VAT_WOMEN	0.069	0.130	9.70E-05	0.480
VATadjBMI_MEN	0.680	0.820	0.110	0.036
VATadjBMI_OVERALL	0.990	0.740	6.10E-04	0.012
VATadjBMI_WOMEN	0.540	0.690	0.003	0.110
VATHU_MEN	0.210	0.100	0.840	8.00E-06
VATHU_OVERALL	0.036	0.007	0.170	0.009
VATHU_WOMEN	0.086	0.026	0.067	0.680
VATSAT_MEN	0.300	0.150	0.014	0.930
VATSAT_OVERALL	0.140	0.019	3.20E-08	0.370
VATSAT_WOMEN	0.310	0.055	2.60E-07	0.320
VATSATadjBMI_MEN	0.390	0.200	0.022	0.770
VATSATadjBMI_OVERALL	0.120	0.010	3.10E-07	0.290
VATSATadjBMI_WOMEN	0.210	0.027	2.60E-06	0.340

Supplementary Table S4. GWAS p-values of the four splicing QTLs for the adipose-related traits

PMID	Trait	GWAS p-value				Sample size	Ancestry
		rs17817288	rs7206790	rs11710420	rs2237199		
27918534	Adipose	9.50E-09	4.20E-09	3.20E-08	1.40E-08	18,332 (9,594 women/8,738 men)	European, African, Hispanic, and Chinese
27398621	Type 2 diabetes	2.88E-10	9.22E-12	0.7031	NA	12,940 (11,645 T2D cases/32,769 controls)	European, African, Hispanic, and Asian
26343387	Coronary artery disease	0.0232	0.0667	0.4568	0.4289	184,305 (60,801 cases/123,504 controls)	European, African Americans, Hispanic, South Asian, and East Asian
24097068	HDL	0.3028	3.92E-06	2.80E-05	0.9663	188,577 Europeans/7,898 non-Europeans	European, African, East Asian, and South Asian
	LDL	0.7714	0.9233	0.4601	0.6172		
	Total cholesterol	0.7378	0.4435	0.2637	0.6275		
	Triglycerides	0.3297	7.98E-05	4.05E-04	0.9626		



Supplementary Figure S1. The p-value distributions of 4 splicing junction SNPs in 27 GWAS including 9 traits in 3 groups (male, female and overall). Note: each dot represents a combo of trait and group. SAT, subcutaneous adipose tissue volume; VAT, visceral adipose tissue volume; VATadjBMI, visceral adipose tissue volume adjusted for BMI; PAT, pericardial adipose tissue volume; PATadjHtWt, pericardial adipose tissue volume adjusted for height and weight; SATHU, subcutaneous adipose tissue attenuation; VATHU, visceral adipose tissue attenuation; VATSAT, ratio of visceral-to-subcutaneous adipose tissue volume; and VATSATadjBMI, ratio of visceral-to-subcutaneous adipose tissue volume adjusted for BMI.