

Association analyses of 249,796 individuals reveal eighteen new loci associated with body mass index

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SUPPLEMENTARY INFORMATION

1. SUPPLEMENTARY TABLES AND FIGURES	P. 2
1.1. Supplementary Tables	p. 3
1.2. Supplementary Figures	p. 14
2. SUPPLEMENTARY NOTE	P. 28
2.1. Description of secondary analyses	p. 29
2.2. Supplementary note – tables	p. 38
2.3. Supplementary note – figure	p. 95
2.4. Authors contribution	p. 97
2.5. Acknowledgements	p. 101
2.6. Competing interests statement	p. 111

1. SUPPLEMENTARY TABLES AND FIGURES

1.1. SUPPLEMENTARY TABLES

Supplementary Table 1 The 42 SNPs, associated with BMI at $P < 5.10^{-6}$ at stage 1, that were taken forward for replication in stage 2.	p. 3
Supplementary Table 2 Association of 32 replicated SNPs with other anthropometric traits.	P. 4
Supplementary Table 3 Association between 32 replicated SNPs with risk of extreme obesity in children and adults, and with BMI in population-based childhood studies.	P. 5
Supplementary Table 4 Association of the 32 confirmed BMI SNPs with metabolic traits.	P. 6
Supplementary Table 5 Gene set enrichment analysis (MAGENTA) of biological pathways with one or more genes from the 32 confirmed BMI loci, using the BMI meta-analysis.	p. 7
Supplementary Table 6 Non-synonymous or splice-site variants in linkage disequilibrium ($r^2 > 0.75$) with lead SNPs.	P. 10
Supplementary Table 7 Significant associations between BMI SNPs and <i>cis</i> gene expression (<i>cis</i> -eQTLs) in lymphocyte, blood, adipose and brain tissues.	P. 11
Supplementary Table 8 Estimated number of BMI loci for each of the effect sizes observed in Stage 2 for the SNPs that reached a genome-wide significance of 5×10^{-8} in the joint analysis of stage 1 and stage 2, given the power to detect the association in the joint analysis of stage 1 and stage 2.	p. 13

Supplementary Table 1 - The 42 SNPs, associated with BMI at $P < 5 \times 10^{-6}$ at stage 1, that were taken forward for replication in stage 2.

Best Ranking SNP Results										Lead SNP Results															
Nearest Gene	SNP	Chr	Position (bp)	Alleles		Imputation quality		Stage 1		SNP	Position (kb)	SNP	Imputation quality		Alleles		Frequency effect allele (%)	Heterogeneity between Stage 1 samples		Stage 1		Stage 2		Stage 1 + Stage 2	
				Effect	Other	r.sq-hat	Proper-info	n	P value				r ² to best	r.sq-hat	Proper-info	Effect		Other	between-study (P-value)	between-gender (P-value)	n	P-value	n	P-value	n
FTO	rs1558902	16	52,361,075	a	t	0.96	0.98	123,846	2.05E-62	rs1558902	52,361,075	-	0.96	0.98	a	t	42%	0.03	0.58	123,846	2.05E-62	68,498	1.01E-60	192,344	4.75E-120
TMEM18	rs2860323	2	604,210	g	a	0.99	0.99	116,780	2.42E-22	rs2867125	612,827	1.00	1.00	1.00	c	t	83%	0.97	0.21	123,833	4.93E-22	73,973	4.42E-30	197,806	2.77E-49
MC4R	rs6567160	18	55,980,115	c	t	0.95	0.98	123,864	1.82E-22	rs571312	55,990,749	1.00	0.96	0.98	a	c	24%	0.04	0.35	123,812	2.14E-22	79,788	3.19E-21	203,600	6.43E-42
GNPDA2	rs10938397	4	44,877,284	g	a	0.88	0.91	123,849	4.35E-17	rs10938397	44,877,284	-	0.88	0.91	g	a	43%	0.46	0.89	123,849	4.35E-17	73,160	1.45E-15	197,008	3.78E-31
BDNF	rs10767664	11	27,682,562	a	t	0.99	0.99	123,865	5.53E-13	rs10767664	27,682,562	-	0.99	0.99	a	t	78%	0.73	0.66	123,865	5.53E-13	80,293	1.17E-14	204,158	4.69E-26
SEC16B	rs543874	1	176,156,103	g	a	0.98	0.99	123,863	1.66E-13	rs543874	176,156,103	-	0.98	0.99	g	a	19%	0.77	0.01	123,863	1.66E-13	55,551	2.41E-11	179,414	3.56E-23
NEGR1	rs2815752	1	72,585,028	a	g	0.97	0.97	123,849	1.17E-14	rs2815752	72,585,028	-	0.97	0.97	a	g	61%	0.03	0.26	123,849	1.17E-14	74,531	2.29E-09	198,380	1.61E-22
RBJ	rs10182181	2	25,003,800	g	a	0.97	0.97	123,745	1.80E-07	rs713586	25,011,512	0.94	0.95	0.96	c	t	47%	0.57	0.02	123,718	2.51E-07	107,030	1.44E-16	230,748	6.17E-22
GPRC5B	rs12444979	16	19,841,101	c	t	0.97	0.98	123,865	4.20E-11	rs12444979	19,841,101	-	0.97	0.98	c	t	87%	0.44	0.78	123,865	4.20E-11	115,850	8.13E-12	239,715	2.91E-21
SH2B1	rs7498665	16	28,790,742	g	a	0.96	0.99	123,620	1.75E-10	rs7359397	28,793,160	1.00	0.96	0.99	t	c	40%	0.85	0.30	123,864	2.41E-10	80,445	7.89E-12	204,309	1.88E-20
TFAP2B	rs987237	6	50,911,009	g	a	0.98	0.98	123,860	5.97E-16	rs987237	50,911,009	-	0.98	0.98	g	a	18%	0.29	0.25	123,860	5.97E-16	71,916	2.40E-06	195,776	2.90E-20
MAP2K5	rs2241423	15	65,873,892	g	a	1.00	1.00	123,835	1.15E-10	rs2241423	65,873,892	-	1.00	1.00	g	a	78%	0.68	0.76	123,835	1.15E-10	104,115	1.59E-09	227,950	1.19E-18
ETV5	rs9816226	3	187,317,193	t	a	0.96	0.98	123,859	7.61E-14	rs9816226	187,317,193	-	0.96	0.98	t	a	82%	0.78	0.42	123,859	7.61E-14	72,362	1.15E-06	196,221	1.69E-18
FAIM2	rs7138803	12	48,533,735	a	g	0.96	0.99	123,799	3.96E-11	rs7138803	48,533,735	-	0.96	0.99	a	g	38%	0.94	0.66	123,799	3.96E-11	76,265	7.82E-08	200,064	1.82E-17
QPCTL	rs2287019	19	50,894,012	c	t	0.78	0.89	100,473	3.18E-07	rs2287019	50,894,012	-	0.78	0.89	c	t	80%	0.95	0.22	100,473	3.18E-07	94,091	1.40E-10	194,564	1.88E-16
TNNI3K	rs1514177	1	74,763,990	c	g	0.98	0.99	123,852	1.36E-09	rs1514175	74,764,232	1.00	0.99	1.00	a	g	43%	0.002	0.11	123,835	1.41E-09	104,065	7.04E-06	227,900	8.16E-14
SLC39A8	rs13107325	4	103,407,732	t	c	0.83	0.88	123,348	1.37E-07	rs13107325	103,407,732	-	0.83	0.88	t	c	7%	0.67	0.19	123,348	1.37E-07	122,030	1.93E-07	239,378	1.50E-13
FLJ35779	rs2112347	5	75,050,998	t	g	0.97	0.99	123,863	4.76E-08	rs2112347	75,050,998	-	0.97	0.99	t	g	63%	0.71	0.22	123,863	4.76E-08	107,866	8.29E-07	231,729	2.17E-13
LRRN6C	rs10968576	9	28,404,339	g	a	0.98	0.99	123,861	1.88E-08	rs10968576	28,404,339	-	0.98	0.99	g	a	31%	0.23	0.65	123,861	1.88E-08	93,055	3.19E-06	216,916	2.65E-13
MTCH2	rs3817334	11	47,607,569	t	c	0.99	1.00	123,815	4.79E-11	rs3817334	47,607,569	-	0.99	1.00	t	c	41%	0.04	0.52	123,815	4.79E-11	68,128	1.10E-03	191,943	1.59E-12
TMEM160	rs3810291	19	52,260,843	a	g	0.78	0.87	119,531	1.04E-07	rs3810291	52,260,843	-	0.78	0.87	a	g	67%	0.86	0.13	119,531	1.04E-07	113,981	1.59E-06	233,512	1.64E-12
FANCL	rs887912	2	59,156,381	t	c	0.99	0.99	123,855	2.69E-06	rs887912	59,156,381	-	0.99	0.99	t	c	29%	0.32	0.99	123,855	2.69E-06	118,952	1.72E-07	242,807	1.79E-12
NRXN3	rs10150332	14	79,006,717	c	t	0.99	0.99	123,865	2.03E-07	rs10150332	79,006,717	-	0.99	0.99	c	t	21%	0.19	0.44	123,865	2.03E-07	59,157	2.86E-05	183,022	2.75E-11
CADM2	rs7640855	3	85,956,854	a	g	0.98	0.99	123,866	9.81E-08	rs13078807	85,966,840	1.00	0.97	0.99	g	a	20%	0.009	0.12	123,808	1.06E-07	113,596	5.32E-05	237,404	3.94E-11
PRKD1	rs11847697	14	29,584,863	t	c	0.85	0.95	119,851	1.11E-08	rs11847697	29,584,863	-	0.85	0.95	t	c	4%	0.82	0.69	119,851	1.11E-08	121,816	2.25E-04	241,667	5.76E-11
LRP1B	rs2890652	2	142,676,401	c	t	0.94	0.97	123,855	2.38E-07	rs2890652	142,676,401	-	0.94	0.97	c	t	18%	0.90	0.81	123,855	2.38E-07	85,213	9.47E-05	209,068	1.35E-10
PTBP2	rs11165643	1	96,696,685	t	c	0.98	0.99	123,861	7.65E-07	rs1555543	96,717,385	1.00	0.99	0.99	c	a	59%	0.37	0.45	123,856	7.81E-07	119,157	4.48E-05	243,013	3.68E-10
MTIF3	rs4771122	13	26,918,180	g	a	0.92	0.94	123,861	1.20E-07	rs4771122	26,918,180	-	0.92	0.94	g	a	24%	0.84	0.24	123,861	1.20E-07	74,716	8.24E-04	198,577	9.48E-10
ZNF608	rs4836133	5	124,360,002	a	c	0.90	0.92	123,266	7.04E-07	rs4836133	124,360,002	-	0.90	0.92	a	c	48%	0.55	0.17	123,266	7.04E-07	118,733	1.88E-04	241,999	1.97E-09
RPL27A	rs4929949	11	8,561,169	c	t	0.95	0.96	123,860	7.57E-08	rs4929949	8,561,169	-	0.95	0.96	c	t	52%	0.35	0.09	123,860	7.57E-08	125,931	1.00E-03	249,791	2.80E-09
KCTD15	rs29938	19	39,003,321	c	t	0.96	0.97	123,855	1.31E-09	rs29941	39,001,372	0.96	0.99	0.98	g	a	67%	0.97	0.98	123,842	2.42E-09	69,030	2.40E-02	192,872	3.01E-09
NUDT3	rs9296115	6	34,542,693	c	a	0.92	0.95	123,743	2.81E-06	rs206936	34,410,847	NA	0.99	0.99	g	a	21%	0.19	0.98	123,865	4.56E-06	125,912	7.39E-04	249,777	3.02E-08
HNF4G	rs2922763	8	76,736,266	t	g	0.93	0.95	123,861	2.13E-07	rs2922763	76,736,266	-	0.93	0.95	t	g	71%	0.47	0.26	123,861	2.13E-07	83,946	1.54E-02	207,807	6.46E-08
ADCY9	rs2444217	16	3,978,388	a	g	0.88	0.93	123,855	3.44E-06	rs2444217	3,978,388	-	0.88	0.93	a	g	57%	0.98	0.32	123,855	3.44E-06	74,931	4.25E-03	198,786	9.49E-08
LMXB1B	rs867559	9	128,505,146	g	a	0.91	0.95	123,779	2.24E-06	rs867559	128,505,146	-	0.91	0.95	g	a	22%	0.85	0.07	123,779	2.24E-06	102,704	4.26E-03	226,483	1.39E-07
CBX1	rs3764400	17	43,478,931	c	t	0.79	0.86	123,736	2.41E-06	rs3764400	43,478,931	-	0.79	0.86	c	t	14%	0.11	0.71	123,736	2.41E-06	74,847	1.73E-02	198,583	4.13E-07
HTR1A	rs255414	5	62,853,656	a	g	0.97	0.99	123,865	8.64E-07	rs255414	62,853,656	-	0.97	0.99	a	g	81%	0.76	0.05	123,865	8.64E-07	75,323	6.54E-02	199,188	1.05E-06
KIAA1505	rs6955651	7	76,477,807	t	c	0.52	0.55	82,760	1.68E-06	rs6955651	76,477,807	-	0.52	0.55	t	c	18%	0.51	0.10	82,760	1.68E-06	42,912	4.21E-02	125,672	1.92E-06
REG3A	rs17016663	2	79,347,442	c	g	0.96	0.98	123,857	3.52E-07	rs12622013	79,354,870	1.00	0.96	0.97	g	a	11%	0.76	0.11	123,856	3.68E-07	121,428	1.97E-01	245,284	2.13E-05
C9orf4	rs6477694	9	110,972,163	c	t	1.00	1.00	123,801	5.36E-06	rs6477694	110,972,163	-	1.00	1.00	c	t	36%	0.83	0.66	123,801	5.36E-06	110,357	1.89E-01	234,158	4.84E-05
SCG3	rs2652594	15	49,794,102	t	c	0.88	0.88	123,848	3.81E-06	rs2652594	49,794,102	-													

Supplementary Table 2

Supplementary Table 2 - Association of 32 replicated SNPs with other anthropometric traits.

SNP	Nearest Gene	Alleles		BMI			Height			Weight			% Body fat			Risk of overweight (BMI ≥ 25 kg/m ²)				Risk of obesity (BMI ≥ 30 kg/m ²)					
		Effect	Other	n	dir	P-value	n	dir	P-value	n	dir	P-value	n	dir	P-value	N	OR	95%CI	P-value	N	OR	95%CI	P-value		
Previous BMI loci																									
rs1558902	<i>FTO</i>	a	t	68,498	+	1.01E-60	69,709	-	9.62E-02	61,918	+	2.06E-41	14,935	+	8.79E-11	63,948	1.138	1.114	1.164	3.43E-31	42,591	1.203	1.169	1.237	4.71E-38
rs2867125	<i>TMEM18</i>	c	t	73,973	+	4.42E-30	75,182	+	4.06E-01	67,390	+	2.16E-22	20,481	+	5.29E-03	69,421	1.119	1.089	1.150	3.03E-16	45,424	1.134	1.095	1.174	2.53E-12
rs571312	<i>MC4R</i>	a	c	79,788	+	3.19E-21	80,997	+	5.50E-09	73,265	+	1.56E-28	16,973	+	2.96E-03	72,870	1.094	1.068	1.121	4.24E-13	49,825	1.108	1.074	1.143	1.44E-10
rs10938397	<i>GNPDA2</i>	g	a	73,160	+	1.45E-15	74,370	-	2.05E-03	66,577	+	4.04E-08	19,709	+	1.01E-05	68,608	1.061	1.039	1.084	4.57E-08	44,976	1.075	1.047	1.105	1.48E-07
rs10767664	<i>BDNF</i>	a	t	80,293	+	1.17E-14	81,502	-	8.16E-01	73,713	+	3.74E-11	17,263	+	2.86E-04	73,318	1.057	1.031	1.084	1.45E-05	50,052	1.079	1.044	1.115	5.21E-06
rs2815752	<i>NEGR1</i>	a	g	74,531	+	2.29E-09	75,743	-	4.57E-01	67,951	+	3.68E-06	20,632	+	4.10E-03	69,981	1.045	1.023	1.067	4.17E-05	45,696	1.061	1.033	1.090	1.73E-05
rs7359397	<i>SH2B1</i>	t	c	80,445	+	7.89E-12	81,656	+	1.95E-01	73,867	+	3.32E-10	17,330	+	3.94E-02	73,476	1.041	1.020	1.063	1.20E-04	50,141	1.049	1.022	1.077	3.84E-04
rs9816226	<i>ETV5</i>	t	a	72,362	+	1.15E-06	73,573	-	8.28E-01	65,780	+	3.75E-04	17,147	+	6.44E-01	69,401	1.029	1.001	1.057	3.93E-02	45,597	1.066	1.028	1.105	5.02E-04
rs3817334	<i>MTCH2</i>	t	c	68,128	+	1.10E-03	69,337	-	3.95E-05	61,548	+	2.41E-01	14,586	+	3.60E-01	63,579	1.016	0.995	1.039	1.43E-01	42,342	1.016	0.988	1.045	2.53E-01
rs29941	<i>KCTD15</i>	g	a	69,030	+	2.40E-02	70,237	-	9.42E-01	62,447	+	1.45E-01	15,444	+	8.47E-01	64,478	1.023	1.000	1.046	4.79E-02	42,827	1.017	0.988	1.046	2.65E-01
Previous waist & weight loci																									
rs543874	<i>SEC16B</i>	g	a	55,551	+	2.41E-11	60,009	+	1.50E-01	52,184	+	1.28E-11	6,696	+	4.70E-01	51,861	1.072	1.041	1.104	4.02E-06	38,577	1.098	1.059	1.138	3.43E-07
rs987237	<i>TFAP2B</i>	g	a	71,916	+	2.40E-06	76,384	+	2.91E-01	68,594	+	2.62E-06	21,465	+	2.49E-03	70,623	1.050	1.023	1.078	2.44E-04	46,043	1.085	1.049	1.122	1.60E-06
rs7138803	<i>FAIM2</i>	a	g	76,265	+	7.82E-08	77,474	+	1.58E-04	69,687	+	2.23E-10	15,124	+	5.95E-03	67,839	1.044	1.022	1.066	8.74E-05	44,452	1.069	1.040	1.098	1.47E-06
rs10150332	<i>NRXN3</i>	c	t	59,157	+	2.86E-05	63,614	+	6.60E-04	55,827	+	4.03E-09	6,673	+	3.43E-02	55,467	1.054	1.025	1.083	1.83E-04	40,487	1.085	1.049	1.123	2.06E-06
Newly identified BMI loci																									
rs713586	<i>RBJ; POMC</i>	c	t	115,850	+	8.13E-12	105,088	-	1.59E-12	97,305	+	2.91E-04	25,244	+	8.26E-05	105,415	1.052	1.034	1.071	6.62E-09	71,063	1.069	1.045	1.093	6.45E-09
rs12444979	<i>GPRC5B</i>	c	t	107,030	+	1.44E-16	117,062	-	1.87E-01	109,272	+	1.23E-08	28,335	+	1.62E-02	108,562	1.085	1.058	1.112	2.05E-10	72,346	1.078	1.044	1.114	6.30E-06
rs2241423	<i>MAP2K5</i>	g	a	104,115	+	1.59E-09	98,921	-	6.78E-01	91,132	+	6.30E-09	17,540	-	3.23E-01	96,812	1.045	1.022	1.069	9.74E-05	68,968	1.065	1.035	1.096	1.93E-05
rs2287019	<i>QPCTL; GIPR</i>	c	t	94,091	+	1.40E-10	88,905	-	2.21E-03	81,114	+	3.92E-05	16,800	+	6.01E-01	90,803	1.055	1.031	1.079	3.95E-06	63,153	1.086	1.054	1.119	7.46E-08
rs1514175	<i>TNNI3K</i>	a	g	118,952	+	1.72E-07	98,882	-	9.60E-01	91,093	+	3.19E-05	17,561	+	3.41E-01	95,783	1.043	1.024	1.063	6.49E-06	68,946	1.044	1.020	1.069	2.76E-04
rs13107325	<i>SLC39A8</i>	t	c	104,065	+	7.04E-06	116,812	-	1.53E-03	109,023	+	4.59E-03	28,089	+	4.72E-04	114,749	1.062	1.025	1.101	1.05E-03	78,427	1.098	1.048	1.150	9.03E-05
rs2112347	<i>FLJ35779; HMGCR</i>	t	g	93,055	+	3.19E-06	102,743	+	4.95E-01	94,954	+	9.95E-08	17,553	+	5.23E-01	100,925	1.042	1.023	1.061	1.30E-05	71,491	1.052	1.030	1.075	2.38E-06
rs10968576	<i>LRRNGC</i>	g	a	107,866	+	8.29E-07	94,267	+	2.91E-01	86,437	+	4.01E-06	16,283	+	1.79E-01	85,765	1.035	1.015	1.056	6.96E-04	60,107	1.040	1.016	1.064	1.13E-03
rs3810291	<i>TMEM160</i>	a	g	122,030	+	1.93E-07	115,191	+	8.60E-01	107,400	+	1.77E-06	24,854	+	9.52E-02	106,457	1.024	1.005	1.045	1.41E-02	71,161	1.055	1.029	1.083	3.27E-05
rs887912	<i>FANCL</i>	t	c	113,981	+	1.59E-06	120,162	+	5.48E-02	112,372	+	3.52E-07	28,089	+	4.14E-01	111,655	1.020	1.006	1.034	5.70E-03	74,506	1.051	1.026	1.077	5.98E-05
rs13078807	<i>CADM2</i>	g	a	113,596	+	5.32E-05	108,364	+	9.95E-01	100,572	+	2.26E-04	27,895	+	1.96E-03	110,328	1.028	1.006	1.051	1.07E-02	73,896	1.027	0.998	1.056	6.85E-02
rs11847697	<i>PRKD1</i>	t	c	85,213	+	9.47E-05	116,616	-	7.34E-01	108,827	+	3.91E-03	28,184	+	8.36E-02	114,493	1.073	1.025	1.124	2.61E-03	78,441	1.102	1.037	1.170	1.58E-03
rs2890652	<i>LRP1B</i>	c	t	121,816	+	2.25E-04	86,415	+	8.40E-01	78,623	+	4.38E-04	17,096	+	5.16E-01	81,945	1.027	1.003	1.053	3.06E-02	55,070	1.048	1.015	1.082	3.98E-03
rs1555543	<i>PTBP2</i>	c	a	74,716	+	8.24E-04	113,938	+	2.69E-03	106,149	+	4.04E-06	28,186	+	1.71E-02	112,176	1.016	0.999	1.033	7.07E-02	76,880	1.016	0.994	1.039	1.62E-01
rs4771122	<i>MTIF3</i>	g	a	118,733	+	1.88E-04	75,917	+	3.78E-02	68,128	+	6.41E-05	5,359	+	7.73E-02	67,423	1.030	1.004	1.055	2.22E-02	48,566	1.045	1.013	1.079	6.31E-03
rs4836133	<i>ZNF608</i>	a	c	119,157	+	4.48E-05	113,530	-	9.70E-01	105,701	+	6.64E-04	28,413	+	2.52E-01	112,955	1.031	1.014	1.049	3.81E-04	76,231	1.029	1.006	1.052	1.19E-02
rs4929949	<i>RPL27A</i>	c	t	125,931	+	1.00E-03	120,718	+	9.55E-01	112,929	+	5.80E-03	28,425	+	1.60E-01	118,650	1.013	0.996	1.029	1.37E-01	81,228	1.027	1.005	1.049	1.49E-02
rs206936	<i>NUDT3</i>	g	a	125,912	+	7.39E-04	120,688	+	3.08E-02	116,162	+	3.75E-04	28,423	+	3.33E-01	118,697	1.034	1.013	1.056	1.36E-03	81,206	1.032	1.005	1.060	1.99E-02

All results were obtained from stage 2 (replication) samples

Supplementary Table 3 - Association between 32 replicated SNPs with risk of extreme obesity in children and adults, with BMI in population-based childhood studies, and with extreme obesity in family-based studies.

Nearest gene	SNP	Case-control studies with extreme obese adults and children/adolescents								Case-control studies with extreme obese children/adolescents (adults not included)					Population-based studies in children and adolescents				TDT on families with one extremely obese child/adolescent					
		Allele		Frequency effect	95%CI		P-value	N	Frequency effect	95%CI		P-value	N	Frequency effect allele (%)	Beta	SE	P-value	N	Frequency effect allele (%)	Effect direction	P-value	N (one parent + child)		
		Effect	Other	allele (%)*	OR	Lower limit			Upper limit	allele (%)*	OR			Lower limit					Upper limit				(%)	(%)
Previous BMI loci																								
<i>FTO</i>	rs1558902	a	t	42%	1.476	1.338	1.629	8.42E-15	4,142	43%	1.435	1.268	1.623	8.91E-09	2,765	42%	-0.023	0.064	7.16E-01	355	49%	+	9.18E-04	848
<i>TMEM18</i>	rs2867125	c	t	83%	1.463	1.326	1.614	3.58E-14	9,954	83%	1.445	1.294	1.614	6.31E-11	8,574	85%	0.056	0.094	5.53E-01	355	86%	+	8.42E-03	848
<i>MC4R</i>	rs571312	a	c	24%	1.370	1.267	1.480	2.28E-15	10,507	24%	1.376	1.263	1.499	2.44E-13	9,127	28%	0.001	0.075	9.90E-01	355	29%	+	4.46E-02	848
<i>GNPDA2</i>	rs10938397	g	a	42%	1.182	1.067	1.309	1.34E-03	3,741	43%	1.135	1.000	1.289	5.06E-02	2,363	37%	-0.112	0.072	1.22E-01	355	44%	+	2.24E-01	848
<i>BDNF</i>	rs10767664	a	t	77%	1.252	1.112	1.408	1.94E-04	4,015	77%	1.224	1.056	1.420	7.33E-03	2,635	76%	-0.005	0.077	9.52E-01	355	80%	+	6.78E-01	848
<i>NEGR1</i>	rs2815752	a	g	62%	1.249	1.161	1.342	2.01E-09	9,901	62%	1.241	1.145	1.346	1.49E-07	8,521	62%	0.012	0.070	8.65E-01	355	63%	+	1.30E-01	848
<i>SH2B1</i>	rs7359397	t	c	38%	1.111	1.034	1.194	4.00E-03	10,461	38%	1.096	1.013	1.186	2.31E-02	9,081	40%	-0.096	0.067	1.49E-01	355	43%	+	3.08E-03	848
<i>ETV5</i>	rs9816226	t	a	82%	1.178	1.036	1.339	1.22E-02	3,733	81%	1.142	0.974	1.339	1.01E-01	2,353	81%	0.091	0.086	2.90E-01	355	84%	+	1.41E-02	848
<i>MTCH2</i>	rs3817334	t	c	41%	1.050	0.978	1.126	1.77E-01	10,337	41%	1.030	0.953	1.114	4.51E-01	8,957	39%	0.089	0.070	2.02E-01	355	42%	+	5.18E-01	848
<i>KCTD15</i>	rs29941	g	a	68%	1.084	1.006	1.168	3.44E-02	10,523	68%	1.080	0.994	1.173	7.02E-02	9,143	68%	0.185	0.069	7.23E-03	355	69%	+	1.08E-01	848
Previous waist & weight loci																								
<i>SEC16B</i>	rs543874	g	a	20%	1.278	1.174	1.392	1.69E-08	9,933	21%	1.257	1.145	1.379	1.42E-06	8,553	17%	0.103	0.084	2.23E-01	355	21%	+	7.06E-03	848
<i>TFAP2B</i>	rs987237	g	a	18%	1.154	1.057	1.259	1.33E-03	10,184	18%	1.185	1.076	1.304	5.43E-04	8,804	17%	0.027	0.091	7.62E-01	355	21%	+	1.12E-01	848
<i>FAIM2</i>	rs7138803	a	g	37%	1.222	1.138	1.312	3.50E-08	10,364	37%	1.224	1.131	1.324	4.38E-07	8,984	38%	-0.003	0.069	9.63E-01	354	41%	+	1.99E-01	848
<i>NRXN3</i>	rs10150332	c	t	19%	1.096	0.971	1.237	1.39E-01	4,103	20%	1.081	0.930	1.258	3.10E-01	2,723	22%	0.162	0.089	6.81E-02	355	23%	+	5.68E-01	848
Newly identified BMI loci																								
<i>RBJ; POMC</i>	rs713586	c	t	47%	1.085	1.029	1.143	2.38E-03	14,774	47%	1.100	1.037	1.166	1.63E-03	12,433	46%	0.087	0.025	6.06E-04	3,166	49%	+	4.34E-01	1,456
<i>GPRC5B</i>	rs12444979	c	t	87%	1.004	0.850	1.187	9.59E-01	2,681	88%	0.881	0.689	1.125	3.10E-01	1,301	87%	0.031	0.028	2.57E-01	5,706	88%	+	2.37E-02	848
<i>MAP2K5</i>	rs2241423	g	a	76%	1.105	1.037	1.178	1.99E-03	15,250	76%	1.111	1.033	1.194	4.40E-03	12,891	81%	0.014	0.020	4.68E-01	8,506	79%	+	3.25E-02	1,460
<i>QPCTL; GIPR</i>	rs2287019	c	t	83%	1.165	1.015	1.338	2.98E-02	4,209	84%	1.152	0.963	1.378	1.22E-01	2,829						91%	+	9.35E-01	848
<i>TNNI3K</i>	rs1514175	a	g	42%	1.149	1.090	1.211	2.71E-07	14,733	43%	1.172	1.104	1.243	1.49E-07	12,374	46%	0.066	0.015	1.60E-05	8,508	44%	-	5.78E-01	1,454
<i>SLC39A8</i>	rs13107325	t	c	7%	1.217	1.075	1.379	2.01E-03	7,536	7%	1.248	1.072	1.451	4.17E-03	5,174	5%	-0.026	0.053	6.17E-01	3,178	8%	-	6.53E-01	1,462
<i>FLJ35779; HMGCR</i>	rs2112347	t	g	64%	1.044	0.979	1.113	1.93E-01	8,547	63%	1.047	0.970	1.130	2.39E-01	6,199	62%	0.016	0.016	2.98E-01	8,494	66%	+	5.79E-01	1,464
<i>LRRN6C</i>	rs109688576	g	a	30%	1.032	0.953	1.118	4.39E-01	10,216	31%	1.033	0.945	1.129	4.78E-01	8,836	39%	0.014	0.019	4.72E-01	5,704	44%	+	5.61E-01	848
<i>TMEM160</i>	rs3810291	a	g	66%	0.980	0.904	1.062	6.17E-01	9,878	66%	0.983	0.899	1.075	7.07E-01	8,501	61%	0.105	0.079	1.85E-01	355	72%	+	2.52E-02	848
<i>FANCL</i>	rs887912	t	c	27%	1.180	1.060	1.315	2.58E-03	3,656	27%	1.100	0.961	1.258	1.66E-01	2,276	27%	0.011	0.021	6.13E-01	5,700	29%	+	7.44E-01	848
<i>CADM2</i>	rs13078807	g	a	19%	1.053	0.984	1.126	1.36E-01	14,752	19%	1.047	0.971	1.130	2.32E-01	12,396	17%	0.083	0.020	4.54E-05	8,540	19%	-	7.22E-01	1,458
<i>PRKD1</i>	rs11847697	t	c	5%	1.377	1.190	1.594	1.80E-05	8,934	5%	1.470	1.242	1.742	7.94E-06	6,570	4%	-0.003	0.049	9.44E-01	8,529	5%	-	3.54E-01	1,462
<i>LRP1B</i>	rs2890652	c	t	16%	1.004	0.875	1.152	9.52E-01	3,665	17%	0.963	0.814	1.140	6.62E-01	2,286	26%	0.001	0.021	9.68E-01	5,561	17%	+	9.48E-01	848
<i>PTBP2</i>	rs1555543	c	a	57%	1.066	1.010	1.125	2.01E-02	14,738	57%	1.046	0.984	1.110	1.47E-01	12,374	59%	0.037	0.016	1.73E-02	8,515	60%	+	1.40E-01	1,456
<i>MTIF3</i>	rs4771122	g	a	22%	1.121	0.973	1.291	1.14E-01	2,680	22%	1.239	1.013	1.515	3.66E-02	1,301	34%	0.017	0.020	3.88E-01	5,700	4%	-	8.98E-01	848
<i>ZNF608 (triallelic)</i>	rs4836133	a	c	50%	1.087	1.017	1.161	1.42E-02	7,518	50%	1.108	1.022	1.201	1.25E-02	5,162	54%	-0.009	0.015	5.82E-01	8,498	51%	-	8.09E-01	608
<i>RPL27A</i>	rs4929949	c	t	49%	0.996	0.935	1.060	9.01E-01	8,852	48%	0.962	0.892	1.037	3.12E-01	6,489	52%	0.035	0.015	2.04E-02	8,495	49%	-	1.86E-01	1,454
<i>NUDT3</i>	rs206936	g	a	21%	1.076	1.009	1.147	2.67E-02	15,008	21%	1.071	0.996	1.152	6.33E-02	12,648	23%	0.024	0.018	1.90E-01	8,502	21%	-	5.78E-01	1,462

* Frequency effect allele from controls only

Supplementary Table 4

Supplementary Table 4 - Association of the 32 confirmed BMI SNPs with metabolic traits.

SNP	Nearest Gene	Alleles		HDL-cholesterol n~19,000		LDL-cholesterol n~19,000		Triglycerides n~19,000		Fasting Glucose n=8,684		Fasting Insulin n=8,684		2 hour Glucose (BMI adjusted) n=8,684		2 hour Glucose n=8,684		HOMA IR n=8,684		HOMA B n=8,684		Type 2 diabetes n _{cases} =4,549 vs. n _{controls} = 5,579	
		Effect	Other	dir	P-value	dir	P-value	dir	P-value	dir	P-value	dir	P-value	dir	P-value	dir	P-value	dir	P-value	dir	P-value	dir	P-value
Previous BMI loci																							
rs1558902	<i>FTO</i>	a	t	-	1.20E-03	+	7.39E-02	+	1.34E-01	+	5.64E-02	+	7.60E-05	-	9.67E-01	+	2.15E-01	+	9.50E-05	+	2.06E-02	+	1.91E-01
rs2867125	<i>TMEM18</i>	c	t	-	2.28E-01	-	2.01E-01	+	9.32E-02	+	2.35E-01	+	3.22E-02	+	4.49E-01	+	1.81E-01	+	3.87E-02	+	3.39E-02	+	1.39E-01
rs571312	<i>MC4R</i>	a	c	-	1.58E-01	-	7.18E-01	+	4.20E-01	+	3.57E-01	+	3.10E-02	+	1.91E-01	+	5.06E-02	+	2.30E-02	+	3.56E-02	+	6.67E-01
rs10938397	<i>GNPDA2</i>	g	a	-	2.40E-03	+	2.23E-01	+	3.51E-01	-	6.21E-01	+	5.20E-05	-	5.26E-01	-	8.01E-01	+	7.60E-04	+	1.10E-03	+	3.90E-04
rs10767664	<i>BDNF</i>	a	t	-	4.65E-01	+	2.10E-01	+	4.08E-01	+	2.64E-01	+	2.28E-01	-	2.02E-01	-	5.21E-01	+	1.69E-01	+	6.32E-01	-	8.62E-01
rs2815752	<i>NEGR1</i>	a	g	-	7.85E-01	-	4.87E-01	+	1.90E-01	-	3.52E-01	+	4.08E-01	+	1.43E-01	+	1.14E-01	+	3.55E-01	+	1.11E-01	-	4.04E-01
rs7359397	<i>SH2B1</i>	t	c	-	9.94E-02	-	2.43E-01	+	2.46E-01	+	4.97E-02	+	1.70E-03	-	6.55E-02	-	2.57E-01	+	3.90E-03	+	2.87E-02	+	3.06E-01
rs9816226	<i>ETV5</i>	t	a	+	6.34E-01	-	6.34E-01	+	9.41E-01	+	6.10E-01	+	5.76E-01	+	6.91E-01	+	3.14E-01	+	5.12E-01	-	7.45E-01	-	4.73E-01
rs3817334	<i>MTCH2</i>	t	c	-	3.01E-02	-	3.47E-01	+	1.12E-01	+	2.65E-01	+	1.77E-01	-	9.00E-01	+	7.24E-01	+	1.34E-01	-	9.76E-01	+	6.08E-01
rs29941	<i>KCTD15</i>	g	a	-	4.61E-01	-	8.03E-01	+	5.35E-01	+	8.06E-01	+	8.92E-01	-	2.20E-01	-	3.16E-01	+	7.44E-01	-	6.61E-01	-	5.24E-01
Previous waist & weight loci																							
rs543874	<i>SEC16B</i>	g	a	+	9.92E-01	-	9.36E-02	-	9.09E-01	+	4.49E-01	+	1.23E-01	-	8.92E-01	+	6.93E-01	+	1.28E-01	+	9.20E-01	+	7.01E-01
rs987237	<i>TFAP2B</i>	g	a	-	9.94E-01	+	9.08E-02	+	9.44E-01	+	2.13E-01	+	6.89E-02	+	7.56E-01	+	6.50E-01	+	2.58E-02	+	3.29E-01	+	9.24E-02
rs7138803	<i>FAIM2</i>	a	g	-	4.15E-01	+	2.12E-01	+	2.13E-01	-	4.90E-01	+	1.73E-01	-	5.07E-01	-	7.97E-01	+	2.09E-01	+	4.58E-02	+	2.02E-01
rs10150332	<i>NRXN3</i>	c	t	-	3.46E-01	+	8.86E-02	+	9.65E-01	+	7.20E-01	+	3.10E-04	-	8.38E-01	+	8.22E-01	+	1.90E-04	+	3.30E-03	+	5.60E-03
Newly identified BMI loci																							
rs713586	<i>RB1; POMC</i>	c	t	-	4.45E-01	-	5.98E-01	-	5.35E-01	+	4.93E-01	+	4.76E-01	+	7.81E-01	+	5.18E-01	+	4.58E-01	+	6.92E-01	-	8.33E-01
rs12444979	<i>GPRC5B</i>	c	t	-	5.33E-02	-	6.90E-01	+	4.26E-01	+	6.14E-01	+	4.20E-03	+	3.84E-01	+	2.89E-01	+	4.50E-03	+	1.24E-02	+	2.33E-01
rs2241423	<i>MAP2K5</i>	g	a	-	4.43E-01	+	8.44E-01	-	1.99E-01	+	3.00E-01	-	2.46E-01	-	2.78E-01	-	5.07E-01	-	4.74E-01	-	7.25E-02	+	1.12E-02
rs2287019	<i>QPCTL; GIPR</i>	c	t	+	6.79E-02	+	2.81E-02	+	9.11E-01	+	3.99E-02	+	5.57E-01	-	1.00E-04	-	5.00E-04	+	4.83E-01	-	9.10E-01	+	NA
rs1514175	<i>TNNI3K</i>	a	g	+	4.84E-01	+	9.75E-01	+	8.68E-01	+	1.08E-01	-	6.07E-01	-	1.36E-01	-	2.12E-01	+	8.00E-01	-	5.75E-01	+	6.73E-01
rs13107325	<i>SLC39A8</i>	t	c	-	1.20E-06	+	7.13E-01	+	6.30E-03	-	6.66E-01	+	3.99E-01	+	5.37E-01	+	4.72E-01	+	5.52E-01	+	3.15E-01	+	NA
rs2112347	<i>FLJ35779; HMGCR</i>	t	g	+	8.18E-01	-	6.70E-04	-	4.25E-01	-	3.02E-01	+	7.03E-01	+	8.14E-02	+	2.73E-02	+	7.05E-01	+	6.08E-01	+	8.45E-01
rs10968576	<i>LRRN6C</i>	g	a	-	4.13E-02	+	8.42E-01	+	4.87E-01	+	1.16E-01	+	6.50E-03	+	5.95E-01	+	3.02E-01	+	1.66E-02	+	1.81E-02	+	5.92E-01
rs3810291	<i>TMEM160</i>	a	g	-	9.24E-02	+	4.42E-01	+	5.81E-02	+	6.90E-01	-	4.90E-01	+	4.52E-01	+	2.80E-01	-	6.95E-01	-	9.75E-01	-	9.87E-01
rs887912	<i>FANCL</i>	t	c	-	3.18E-01	+	3.64E-01	+	6.15E-02	-	6.73E-01	+	2.26E-01	-	7.27E-01	-	9.85E-01	+	2.82E-01	+	1.67E-01	+	1.81E-01
rs13078807	<i>CADM2</i>	g	a	-	8.55E-01	+	8.02E-01	-	9.60E-01	+	7.62E-01	+	3.22E-01	-	6.15E-01	-	6.31E-01	+	2.82E-01	+	2.78E-01	-	5.67E-01
rs11847697	<i>PRKD1</i>	t	c	-	5.39E-01	+	2.36E-01	+	8.04E-01	-	6.20E-01	+	1.27E-01	+	2.73E-01	+	1.97E-01	+	6.13E-02	+	5.67E-02	+	7.07E-02
rs2890652	<i>LRP1B</i>	c	t	+	3.51E-01	-	5.28E-01	-	8.38E-01	+	4.70E-02	-	1.25E-01	-	1.93E-01	-	2.12E-01	-	4.36E-01	-	1.22E-01	+	5.73E-02
rs1555543	<i>PTBP2</i>	c	a	-	9.80E-02	-	8.34E-01	+	3.51E-01	-	7.74E-01	+	2.13E-01	-	4.25E-02	-	8.51E-02	+	1.83E-01	+	8.83E-02	+	2.50E-01
rs4771122	<i>MTIF3</i>	g	a	-	4.91E-01	-	5.64E-01	+	4.00E-01	+	8.88E-01	+	1.76E-01	-	9.78E-01	+	8.16E-01	+	3.61E-01	+	3.27E-01	+	5.51E-01
rs4836133	<i>ZNF608</i>	a	c	-	5.65E-01	-	8.62E-02	+	1.34E-01	+	7.12E-01	+	1.14E-01	-	1.43E-01	-	2.27E-01	+	3.13E-02	+	1.12E-01	+	6.44E-01
rs4929949	<i>RPL27A</i>	c	t	+	4.54E-01	+	9.14E-01	-	1.00E-01	+	2.60E-01	+	1.67E-02	+	1.25E-01	+	6.62E-02	+	2.28E-02	+	2.43E-01	+	4.54E-01
rs206936	<i>NUDT3</i>	g	a	-	8.40E-01	+	2.07E-01	+	1.52E-01	+	6.78E-01	-	8.72E-01	-	1.92E-01	-	2.68E-01	-	7.14E-01	-	6.02E-01	-	1.28E-01

Association results for HDL-, LDL- Cholesterol levels were obtained from publicly available data of the GLOBAL Lipids Genetics Consortium (<http://www.sph.umich.edu/csg/abecasis/public/lipids2008/>)

Association results for fasting insulin and glucose, 2hr-glucose, HOMA-IR and HOMA-B were obtained from the MAGIC (Dupuis et al Nature Genetics 2010; Saxena et al Nature Genetics 2010)

Association results for risk of type 2 diabetes were obtained from the DIAGRAM consortium (Zeggini et al Nature Genetics 2010)

Supplementary Table 5. Gene set enrichment analysis (MAGENTA) of biological pathways with one or more genes from the 32 confirmed BMI loci, using the BMI meta-analysis.

Database	Gene set	Original # genes in gene set	# genes in gene set analyzed by MAGENTA	Nominal GSEA P value	FDR q-value	# genes expected above 75 th percentile cutoff	# genes observed above 75 th percentile cutoff	Genes within 300 kb from the 32 confirmed BMI SNPs
KEGG	ADIPOCYTOKINE SIGNALING PATHWAY	70	67	0.0095	0.2049	17	26	NFKB1; POMC
KEGG	GAP JUNCTION	92	87	0.0161	0.2312	22	31	ADCY3; MAP2K5
KEGG	FOCAL ADHESION	206	194	0.021	0.2529	49	61	GRLF1; VASP
KEGG	MAPK SIGNALING PATHWAY	269	246	0.0254	0.2011	62	75	BDNF; NFKB1; MAP2K5
KEGG	T CELL RECEPTOR SIGNALING PATHWAY	97	91	0.0315	0.1720	23	31	NFKB1; LAT
KEGG	PHOSPHATIDYLINOSITOL SIGNALING SYSTEM	92	91	0.0328	0.1504	23	31	DGKG; DMPK
KEGG	INOSITOL PHOSPHATE METABOLISM	64	63	0.0494	0.1891	16	22	DMPK
Ingenuity	T CELL RECEPTOR SIGNALING	34	33	0.009	0.1299	8	15	LAT
Ingenuity	NFKB SIGNALING	43	40	0.0515	0.4614	10	15	NFKB1
Panther	PDGF SIGNALING PATHWAY	52	43	0.0008	0.0061	11	21	SPDEF
Panther	AXON GUIDANCE MEDIATED BY NETRIN	16	16	0.0064	0.0321	4	9	VASP
Panther	P53 PATHWAY	59	55	0.041	0.2195	14	20	NFATC2IP
PANTHER, BP	PROTEIN PHOSPHORYLATION	660	562	0.0001	0.0453	141	173	DMPK; PRKD1; MAP2K5; COL4A3BP; PACSIN1; TNNI3K; STK33; FLJ40125
PANTHER, BP	PRE-MRNA PROCESSING	39	27	0.0016	0.0411	7	14	SNRPD2
PANTHER, BP	RECEPTOR PROTEIN TYROSINE KINASE SIGNALING PATHWAY	211	182	0.014	0.3222	46	59	BDNF
PANTHER, BP	INTRACELLULAR SIGNALING CASCADE	261	225	0.0184	0.3723	56	70	RACGAP1; RBJ
PANTHER, BP	DNA REPAIR	169	138	0.0236	0.3657	35	45	ERCC1; POLK
PANTHER, BP	OTHER INTRACELLULAR SIGNALING CASCADE	225	192	0.0266	0.3623	48	60	DGKG; PACSIN1; FLJ40125
PANTHER, BP	NEUROGENESIS	587	477	0.0326	0.3476	119	136	BDNF; FOXA3; RTN2; MADD; ZNF608; IRX3; NEGR1; LBXCOR1
PANTHER, BP	CELL STRUCTURE	687	475	0.0373	0.3454	119	135	DMPK; FOXA3; LIN7C
PANTHER, BP	JNK CASCADE	61	58	0.0382	0.3785	15	21	MADD
PANTHER, BP	LIPID AND FATTY ACID TRANSPORT	131	98	0.046	0.3127	25	32	COL4A3BP
PANTHER, BP	LIPID METABOLISM	151	129	0.0489	0.3391	32	41	DGKG; LASS5
PANTHER, MF	HOMEBOX TRANSCRIPTION FACTOR	249	155	0.0001	0.0110	39	59	MEIS3; IRX3; SIX5
PANTHER, MF	TRANSLATION ELONGATION FACTOR	33	15	0.0008	0.0066	4	10	TUFM
PANTHER, MF	RNA HELICASE	83	63	0.0135	0.1787	16	24	DHX34
PANTHER, MF	KINASE	30	29	0.0166	0.2281	7	13	DGKG
PANTHER, MF	PROTEIN KINASE	191	163	0.0179	0.1748	41	53	MAP2K5; COL4A3BP; TNNI3K
PANTHER, MF	DNA METHYLTRANSFERASE	16	16	0.0259	0.1842	4	8	DNMT3A
PANTHER, MF	G-PROTEIN MODULATOR	35	25	0.0292	0.1722	6	11	RACGAP1
PANTHER, MF	VOLTAGE-GATED POTASSIUM CHANNEL	91	82	0.0388	0.2595	21	28	KCNH3
Gene Ontology, BP	NEUROGENESIS	93	90	0.0001	0.0214	23	39	NRXN3; RACGAP1
Gene Ontology, BP	NEURON DIFFERENTIATION	76	73	0.0001	0.0324	18	32	NRXN3
Gene Ontology, BP	GENERATION OF NEURONS	83	80	0.0002	0.0335	20	34	NRXN3; RACGAP1
Gene Ontology, BP	REGULATION OF CELLULAR METABOLIC PROCESS	782	714	0.0002	0.0308	179	214	ERCC1; FOSB; GRLF1; HMGA1; SMARCD1; SPI1; SPN; TFAP2B; IGF2BP2; AKTIP; MTIF3
Gene Ontology, BP	BIOPOLYMER METABOLIC PROCESS	1674	1458	0.0003	0.0349	365	408	DMPK; DNMT3A; ERCC1; FOSB; GTF3A; HMGA1; NFKB1; PRKD1; SFRS10; SMARCD1; SNRPD2; SPI1; TFAP2B; TUFM; SAE1; COL4A3BP; MCRS1; CUGBP1; CD3EAP; POLK; AKTIP
Gene Ontology, BP	GROWTH	76	74	0.0003	0.0256	19	32	NDUFS3; BBC3
Gene Ontology, BP	TRANSMISSION OF NERVE IMPULSE	188	171	0.0006	0.0311	43	61	GRM4; RAPSN
Gene Ontology, BP	REGULATION OF METABOLIC PROCESS	794	723	0.0007	0.0298	181	216	ERCC1; FOSB; GRLF1; HMGA1; SMARCD1; SPI1; SPN; TFAP2B; IGF2BP2; AKTIP; MTIF3
Gene Ontology, BP	NEURON DEVELOPMENT	61	59	0.001	0.0291	15	26	NRXN3
Gene Ontology, BP	NERVOUS SYSTEM DEVELOPMENT	379	333	0.0013	0.0353	83	107	BDNF; NPAS1; TFAP2B; NRXN3; RACGAP1
Gene Ontology, BP	NUCLEOBASE NUCLEOSIDE NUCLEOTIDE AND NUCLEIC ACID METABOLIC PROCESS	1235	1095	0.0014	0.0348	274	310	DNMT3A; ERCC1; FOSB; GRLF1; GTF3A; HMGA1; NFKB1; SFRS10; SMARCD1; SNRPD2; SPI1; TFAP2B; CUGBP1; CD3EAP; NUDT3; POLK
Gene Ontology, BP	NEURITE DEVELOPMENT	53	51	0.0016	0.0297	13	23	NRXN3
Gene Ontology, BP	REGULATION OF GROWTH	57	55	0.0016	0.0335	14	24	NDUFS3; BBC3
Gene Ontology, BP	REGULATION OF GENE EXPRESSION	669	617	0.0018	0.0461	154	183	DNMT3A; FOSB; GRLF1; HMGA1; SMARCD1; SPI1; SPN; TFAP2B; IGF2BP2; MTIF3
Gene Ontology, BP	AXONOGENESIS	43	41	0.0021	0.0362	10	19	NRXN3
Gene Ontology, BP	CELL DEVELOPMENT	575	541	0.0022	0.0411	135	163	GRM4; NDUFS3; NFKB1; MADD; NRXN3; BBC3; RACGAP1

Supplementary Table 5

Database	Gene set	Original # genes in gene set	# genes in gene set analyzed by MAGENTA	Nominal GSEA P value	FDR q-value	# genes expected above 75 th percentile cutoff	# genes observed above 75 th percentile cutoff	Genes within 300 kb from the 32 confirmed BMI SNPs
Gene Ontology, BP	PROTEIN METABOLIC PROCESS	1220	1079	0.0022	0.0470	270	304	DMPK; HMGA1; PRKD1; RPL21; RPL27A; RPS10; SPN; TUFM; SAE1; COL4A3BP; MCRS1; IGF2BP2; CHST8; AKTIP; MTIF3
Gene Ontology, BP	CELLULAR COMPONENT ASSEMBLY	295	277	0.0025	0.0473	69	89	AP2S1; HMGA1; SNRPD2; CUGBP1; MTIF3
Gene Ontology, BP	MACROMOLECULAR COMPLEX ASSEMBLY	277	260	0.0025	0.0475	65	85	HMGA1; SNRPD2; CUGBP1; MTIF3
Gene Ontology, BP	T CELL ACTIVATION	44	40	0.0026	0.0400	10	15	LAT
Gene Ontology, BP	NEGATIVE REGULATION OF CELL ADHESION	17	17	0.0037	0.0342	4	10	SPN
Gene Ontology, BP	CELL ACTIVATION	75	70	0.0048	0.0483	18	28	LAT
Gene Ontology, BP	CELLULAR MACROMOLECULE METABOLIC PROCESS	1121	996	0.0048	0.0666	249	278	DMPK; HMGA1; PRKD1; RPL21; RPL27A; RPS10; SPN; TUFM; SAE1; COL4A3BP; MCRS1; IGF2BP2; CHST8; AKTIP; MTIF3
Gene Ontology, BP	REGULATION OF MOLECULAR FUNCTION	324	302	0.0048	0.0525	76	96	C5AR1; GIPR; GRM4; MADD; BBC3; AKTIP
Gene Ontology, BP	REGULATION OF MULTICELLULAR ORGANISMAL PROCESS	150	140	0.0052	0.0548	35	49	ATP2A1; DMPK; MC4R; MYBPC3; LAT
Gene Ontology, BP	REGULATION OF DEVELOPMENTAL PROCESS	437	408	0.0056	0.0628	102	124	GRM4; NDUFS3; NFKB1; SPI1; MADD; BBC3
Gene Ontology, BP	REGULATION OF NUCLEOBASE NUCLEOSIDE NUCLEOTIDE AND NUCLEIC ACID METABOLIC PROCESS	615	567	0.0061	0.0640	142	166	ERCC1; FOSB; GRLF1; HMGA1; SMARCD1; SPI1; TFAP2B
Gene Ontology, BP	CELLULAR PROTEIN METABOLIC PROCESS	1107	986	0.0064	0.0622	247	275	DMPK; HMGA1; PRKD1; RPL21; RPL27A; RPS10; SPN; TUFM; SAE1; COL4A3BP; MCRS1; IGF2BP2; CHST8; AKTIP; MTIF3
Gene Ontology, BP	NEGATIVE REGULATION OF GROWTH	39	38	0.0066	0.0522	10	17	NDUFS3; BBC3
Gene Ontology, BP	POSITIVE REGULATION OF BIOLOGICAL PROCESS	707	657	0.0066	0.0653	164	190	ATP2A1; GRM4; HMGA1; MC4R; NDUFS3; SMARCD1; SPI1; SPN; TFAP2B; BBC3; AKTIP
Gene Ontology, BP	REGULATION OF APOPTOSIS	339	319	0.0066	0.0683	80	99	GRM4; NDUFS3; NFKB1; MADD; BBC3
Gene Ontology, BP	REGULATION OF PROGRAMMED CELL DEATH	340	320	0.0066	0.0645	80	99	GRM4; NDUFS3; NFKB1; MADD; BBC3
Gene Ontology, BP	CELLULAR MORPHOGENESIS DURING DIFFERENTIATION	49	47	0.0071	0.0545	12	20	NRXN3
Gene Ontology, BP	REGULATION OF CELLULAR PROTEIN METABOLIC PROCESS	161	150	0.0071	0.0632	38	51	SPN; IGF2BP2; AKTIP; MTIF3
Gene Ontology, BP	REGULATION OF TRANSCRIPTION	563	519	0.0083	0.0720	130	152	FOSB; GRLF1; HMGA1; SMARCD1; SPI1; TFAP2B
Gene Ontology, BP	LYMPHOCYTE ACTIVATION	60	55	0.0086	0.0637	14	22	LAT
Gene Ontology, BP	REGULATION OF LYMPHOCYTE ACTIVATION	35	34	0.0092	0.0650	9	15	LAT
Gene Ontology, BP	MAPK KINASE CASCADE GO 0000165	103	97	0.0096	0.0664	24	35	C5AR1; GRM4; MADD
Gene Ontology, BP	REGULATION OF CELL ADHESION	36	36	0.0098	0.0634	9	16	SPN
Gene Ontology, BP	REGULATION OF BIOLOGICAL QUALITY	416	372	0.0104	0.0786	93	112	C5AR1; GRM4; MC4R; NDUFS3; SPI1
Gene Ontology, BP	REGULATION OF IMMUNE SYSTEM PROCESS	67	62	0.0104	0.0690	16	24	LAT
Gene Ontology, BP	POSITIVE REGULATION OF CELLULAR METABOLIC PROCESS	227	215	0.0106	0.0728	54	69	HMGA1; SMARCD1; SPI1; SPN; TFAP2B; AKTIP
Gene Ontology, BP	POSITIVE REGULATION OF METABOLIC PROCESS	234	220	0.0108	0.0787	55	70	HMGA1; SMARCD1; SPI1; SPN; TFAP2B; AKTIP
Gene Ontology, BP	NEGATIVE REGULATION OF CATALYTIC ACTIVITY	68	65	0.0111	0.0650	16	25	GRM4
Gene Ontology, BP	APOPTOSIS GO	429	403	0.0115	0.0791	101	120	GRM4; NDUFS3; NFKB1; MADD; BBC3
Gene Ontology, BP	POSITIVE REGULATION OF CELLULAR PROCESS	666	621	0.0129	0.0826	155	178	GRM4; HMGA1; NDUFS3; SMARCD1; SPI1; SPN; TFAP2B; BBC3; AKTIP
Gene Ontology, BP	REGULATION OF CATALYTIC ACTIVITY	276	261	0.0129	0.0804	65	81	C5AR1; GIPR; GRM4; MADD; BBC3
Gene Ontology, BP	NUCLEOTIDE EXCISION REPAIR	20	20	0.0131	0.0695	5	10	ERCC1
Gene Ontology, BP	REGULATION OF PROTEIN METABOLIC PROCESS	172	159	0.0133	0.0801	40	52	SPN; IGF2BP2; AKTIP; MTIF3
Gene Ontology, BP	PROGRAMMED CELL DEATH	430	404	0.014	0.0808	101	120	GRM4; NDUFS3; NFKB1; MADD; BBC3
Gene Ontology, BP	POST TRANSLATIONAL PROTEIN MODIFICATION	474	443	0.0152	0.0880	111	130	DMPK; PRKD1; SAE1; COL4A3BP; AKTIP
Gene Ontology, BP	LEUKOCYTE ACTIVATION	68	63	0.0157	0.0803	16	24	LAT
Gene Ontology, BP	REGULATION OF KINASE ACTIVITY	156	150	0.0163	0.0875	38	49	C5AR1; GRM4; MADD
Gene Ontology, BP	PHOSPHORYLATION	311	290	0.0168	0.0932	73	88	DMPK; PRKD1; COL4A3BP; AKTIP
Gene Ontology, BP	ESTABLISHMENT AND OR MAINTENANCE OF CHROMATIN ARCHITECTURE	76	70	0.0187	0.0857	18	26	HMGA1; SMARCD1
Gene Ontology, BP	RNA METABOLIC PROCESS	836	759	0.0194	0.1008	190	212	FOSB; GTF3A; HMGA1; NFKB1; SFRS10; SMARCD1; SNRPD2; SPI1; TFAP2B; CUGBP1; CD3EAP
Gene Ontology, BP	TRANSCRIPTION	748	687	0.0199	0.1005	172	193	FOSB; GRLF1; GTF3A; NFKB1; SMARCD1; SPI1; TFAP2B; CD3EAP
Gene Ontology, BP	REGULATION OF PROTEIN KINASE ACTIVITY	154	148	0.0206	0.1020	37	48	C5AR1; GRM4; MADD
Gene Ontology, BP	POTASSIUM ION TRANSPORT	58	52	0.0218	0.0879	13	20	KCNH3
Gene Ontology, BP	RESPONSE TO NUTRIENT LEVELS	28	27	0.0226	0.0820	7	12	GIPR
Gene Ontology, BP	DNA REPAIR	123	117	0.0234	0.1025	29	39	ERCC1
Gene Ontology, BP	REGULATION OF CELL GROWTH	45	43	0.0237	0.0942	11	17	NDUFS3
Gene Ontology, BP	REGULATION OF TRANSCRIPTION DNA DEPENDENT	459	428	0.0237	0.1114	107	124	FOSB; HMGA1; SMARCD1; SPI1; TFAP2B
Gene Ontology, BP	POSITIVE REGULATION OF CELLULAR PROTEIN METABOLIC PROCESS	73	69	0.0255	0.1032	17	25	SPN; AKTIP
Gene Ontology, BP	HOMEOSTASIS OF NUMBER OF CELLS	20	19	0.0271	0.0886	5	9	SPI1
Gene Ontology, BP	POSITIVE REGULATION OF CATALYTIC ACTIVITY	165	157	0.0281	0.1125	39	50	C5AR1; GIPR; GRM4; MADD; BBC3
Gene Ontology, BP	UBIQUITIN CYCLE	48	47	0.0292	0.1027	12	18	SAE1
Gene Ontology, BP	POSITIVE REGULATION OF NUCLEOBASE NUCLEOSIDE NUCLEOTIDE AND NUCLEIC ACID METABOLIC PROCESS	153	147	0.0299	0.1133	37	47	HMGA1; SMARCD1; SPI1; TFAP2B
Gene Ontology, BP	BIOPOLYMER MODIFICATION	648	599	0.0304	0.1186	150	168	DMPK; DNMT3A; PRKD1; SAE1; COL4A3BP; MCRS1; AKTIP
Gene Ontology, BP	POSITIVE REGULATION OF TRANSFERASE ACTIVITY	86	84	0.0315	0.1131	21	29	C5AR1; GRM4; MADD
Gene Ontology, BP	REGULATION OF TRANSLATION	92	87	0.0323	0.1132	22	30	SPN; IGF2BP2; MTIF3

Supplementary Table 5

Database	Gene set	Original # genes in gene set	# genes in gene set analyzed by MAGENTA	Nominal GSEA <i>P</i> value	FDR <i>q</i> -value	# genes expected above 75 th percentile cutoff	# genes observed above 75 th percentile cutoff	Genes within 300 kb from the 32 confirmed BMI SNPs
Gene Ontology, BP	REGULATION OF CELLULAR COMPONENT ORGANIZATION AND BIOGENESIS	124	119	0.0324	0.1146	30	39	AP2S1; ERCC1; MTF3
Gene Ontology, BP	CHROMATIN MODIFICATION	54	51	0.0327	0.1108	13	19	HMGA1; SMARCD1
Gene Ontology, BP	ACTIVATION OF MAPK ACTIVITY	41	41	0.0328	0.1088	10	16	CSAR1; GRM4; MADD
Gene Ontology, BP	POSITIVE REGULATION OF PROTEIN MODIFICATION PROCESS	29	29	0.033	0.1083	7	12	AKTIP
Gene Ontology, BP	POSITIVE REGULATION OF TRANSCRIPTION	143	137	0.0332	0.1188	34	44	HMGA1; SMARCD1; SPI1; TFAP2B
Gene Ontology, BP	REGULATION OF TRANSFERASE ACTIVITY	160	154	0.0333	0.1146	39	49	CSAR1; GRM4; MADD
Gene Ontology, BP	CELL CYCLE PROCESS	192	183	0.0334	0.1200	46	57	RACGAP1
Gene Ontology, BP	POSITIVE REGULATION OF PROTEIN METABOLIC PROCESS	75	71	0.0340	0.1130	18	25	SPN; AKTIP
Gene Ontology, BP	POSITIVE REGULATION OF DEVELOPMENTAL PROCESS	218	209	0.0343	0.1174	52	64	NDUFS3; BBC3
Gene Ontology, BP	GENERATION OF PRECURSOR METABOLITES AND ENERGY	122	113	0.0350	0.1178	28	37	GIPR; POMC
Gene Ontology, BP	NEGATIVE REGULATION OF CELLULAR COMPONENT ORGANIZATION AND BIOGENESIS	28	26	0.0379	0.1118	7	11	ERCC1
Gene Ontology, BP	REGULATION OF RNA METABOLIC PROCESS	469	437	0.0381	0.1294	109	125	FOSB; HMGA1; SMARCD1; SPI1; TFAP2B
Gene Ontology, BP	OXYGEN AND REACTIVE OXYGEN SPECIES METABOLIC PROCESS	20	20	0.0405	0.1116	5	9	NDUFS3
Gene Ontology, BP	NEURON APOPTOSIS	17	17	0.0408	0.1087	4	8	GRM4
Gene Ontology, BP	POSITIVE REGULATION OF BINDING	28	26	0.0435	0.1138	7	11	AKTIP
Gene Ontology, BP	PROTEIN UBIQUITINATION	40	39	0.0443	0.1209	10	15	SAE1
Gene Ontology, BP	PROTEIN MODIFICATION PROCESS	629	582	0.0446	0.1481	146	162	DMPK; PRKD1; SAE1; COL4A3BP; MCRS1; AKTIP
Gene Ontology, BP	PROTEIN AMINO ACID PHOSPHORYLATION	277	260	0.0462	0.1518	65	77	DMPK; PRKD1; COL4A3BP; AKTIP
Gene Ontology, BP	CHROMOSOME ORGANIZATION AND BIOGENESIS	123	115	0.0476	0.1437	29	37	ERCC1; HMGA1; SMARCD1
Gene Ontology, BP	CELL CELL SIGNALING	403	362	0.0483	0.1524	91	104	GRM4; POMC; RAPSN; NUDT3
Gene Ontology, BP	PROTEIN COMPLEX ASSEMBLY	166	158	0.0483	0.1458	40	49	HMGA1
Gene Ontology, BP	SYNAPTIC TRANSMISSION	173	156	0.0487	0.1668	39	55	GRM4; RAPSN
Gene Ontology, BP	POSITIVE REGULATION OF MAPKKK CASCADE	11	9	0.049	0.1015	2	5	GRM4
Gene Ontology, MF	HORMONE RECEPTOR BINDING	29	27	0.0002	0.0082	7	16	HMGA1
Gene Ontology, MF	NUCLEAR HORMONE RECEPTOR BINDING	28	26	0.0005	0.0085	7	15	HMGA1
Gene Ontology, MF	KINASE ACTIVITY	366	338	0.0030	0.1568	85	106	DGKG; DMPK; PRKD1; COL4A3BP
Gene Ontology, MF	IDENTICAL PROTEIN BINDING	304	278	0.0043	0.1243	70	89	DMPK
Gene Ontology, MF	NUCLEOTIDE BINDING	223	207	0.0074	0.1485	52	67	DMPK
Gene Ontology, MF	TRANSFERASE ACTIVITY TRANSFERRING PHOSPHORUS CONTAINING GROUPS	421	385	0.0084	0.1805	96	116	DGKG; DMPK; PRKD1; COL4A3BP
Gene Ontology, MF	SMALL GTPASE REGULATOR ACTIVITY	67	64	0.0099	0.1559	16	25	MADD; RASAL2
Gene Ontology, MF	PURINE RIBONUCLEOTIDE BINDING	204	190	0.0144	0.2105	48	61	DMPK
Gene Ontology, MF	PHOSPHOTRANSFERASE ACTIVITY ALCOHOL GROUP AS ACCEPTOR	332	311	0.0169	0.1775	78	94	DGKG; DMPK; PRKD1; COL4A3BP
Gene Ontology, MF	PURINE NUCLEOTIDE BINDING	210	195	0.0169	0.2039	49	62	DMPK
Gene Ontology, MF	GTPASE REGULATOR ACTIVITY	124	120	0.0214	0.1882	30	40	MADD; RASAL2; RACGAP1
Gene Ontology, MF	ATP BINDING	155	145	0.0257	0.1912	36	47	DMPK
Gene Ontology, MF	SUBSTRATE SPECIFIC CHANNEL ACTIVITY	156	138	0.0264	0.2050	35	45	AQP2; AQP5; AQP6; KCNH3
Gene Ontology, MF	DNA BINDING	596	548	0.0265	0.2003	137	156	DNMT3A; ERCC1; ETV5; GRLF1; HMGA1; FOXA3; NFKB1; NPAS1; SPI1; TFAP2B
Gene Ontology, MF	ION CHANNEL ACTIVITY	149	132	0.0271	0.1918	33	43	KCNH3
Gene Ontology, MF	RAS GUANYL NUCLEOTIDE EXCHANGE FACTOR ACTIVITY	19	19	0.0288	0.1914	5	9	MADD
Gene Ontology, MF	PROTEIN KINASE ACTIVITY	283	265	0.0294	0.1937	66	80	DMPK; PRKD1; COL4A3BP
Gene Ontology, MF	RNA BINDING	255	236	0.0358	0.2123	59	71	RPL21; RPL27A; RPS10; SFRS10; IGF2BP2; CUGBP1
Gene Ontology, MF	ADENYL RIBONUCLEOTIDE BINDING	162	152	0.0366	0.2084	38	48	DMPK
Gene Ontology, MF	ADENYL NUCLEOTIDE BINDING	168	157	0.0444	0.2183	39	49	DMPK

Nominal gene set enrichment analysis (GSEA) *p*-values and false discovery rates were computed for biological gene-sets that contain at least one gene from the 32 confirmed Body Index Mass (BMI) loci, using MAGENTA. The biological gene-sets were taken from four different resources. Results are presented for the nominally significant pathways ($p < 0.05$). Bonferroni correction was done separately for each database due to considerable overlap of gene sets between databases. The Bonferroni corrected *p*-value cutoffs for the different databases are: KEGG (36 pathways): $p < 0.0014$, Ingenuity (24 pathways): $p < 0.0021$, PANTHER (21 metabolic and signaling pathways): $p < 0.0024$, PANTHER, BP (Biological Processes; 90 gene sets): $p < 5.6E-04$, PANTHER, MF Molecular Functions; 71 gene sets): $p < 7.0E-04$, Gene Ontology (GO) biological process (BP) terms (334 gene sets): $p < 1.50E-05$, and Gene Ontology (GO) molecular function (MF) terms (124 gene sets): $p < 4.0E-04$. In bold are gene sets that pass or are close to the Bonferroni cutoff.

Supplementary Table 6

Supplementary Table 6 - Non-synonymous or splice-site variants in linkage disequilibrium ($r^2 > 0.75$) with lead SNPs.

Chr	Position	lead SNP	Coding SNP	r^2	Gene	Coding change	Type	Information source (map build)
2	25,011,512	rs713586	rs11676272	0.90	<i>ADCY3</i>	S107P	Non-synonymous	HapMap_r21_CEU
4	103,407,732	rs13107325	rs13107325	1.00	<i>SLC39A8</i>	A391T	Non-synonymous	HapMap_r21_CEU
5	75,050,998	rs2112347	rs2307111	0.81	<i>FLJ35779</i>	H11R	Non-synonymous	HapMap_r21_CEU
11	27,682,562	rs10767664	rs6265	0.76	<i>BDNF</i>	V66M	Non-synonymous	HapMap_r21_CEU
11	47,607,569	rs3817334	rs1064608	0.84	<i>MTCH2</i>	P290A	Non-synonymous	HapMap_r21_CEU
15	65,873,892	rs2241423	rs7170185	0.94	<i>LBXCOR1</i>	W200R	Non-synonymous	1000G_0904_CEU
16	28,793,160	rs7359397	rs180743	0.85	<i>APOB48R</i>	P419A	Non-synonymous	HapMap_r21_CEU
16	28,793,160	rs7359397	rs7498665	1.00	<i>SH2B1</i>	T484A	Non-synonymous	HapMap_r21_CEU
16	28,793,160	rs7359397	rs1059491	0.75	<i>SULT1A2</i>	N235T	Non-synonymous	1000G_0904_CEU
16	28,793,160	rs7359397	rs180743	0.75	<i>AC138894.2</i>	P419A	Non-synonymous	1000G_0904_CEU
16	28,793,160	rs7359397	rs55719896	1.00	<i>ATXN2L</i>	-	splice site alteration	1000G_0908_CEU
19	50,894,012	rs2287019	rs1800437	0.83	<i>GIPR</i>	E354Q	Non-synonymous	HapMap_r21_CEU

r^2 is the linkage disequilibrium (LD) between the lead SNP, examined in the current study, and the nearby coding SNPs.

Supplementary Table 7 - Significant associations between BMI SNPs and *cis* gene expression (*cis*-eQTLs) in lymphocyte, blood, liver, adipose tissue*, and brain tissues.

BMI SNP	Chr	Position	BMI increasing allele	Tissue	Gene	Effect ^a	P-value	P _{adj} ^b	Peak SNP ^c	r ² ^d	P-value	P _{adj} ^e
rs2815752	1	72,585,028	A	Blood	<i>NEGR1</i>	+	3.60E-27	0.44	rs2568958	1.00	1.30E-27	0.13
rs713586	2	25,011,512	C	Lymphocyte	<i>ADCY3</i>	-	1.04E-19	0.75	rs6737082	0.78	1.79E-21	0.17
				Omental fat	<i>ADCY3</i>	+	5.40E-06	0.23	rs7608976	0.66	1.19E-11	0.004
				Blood	<i>ADCY3</i>	-	1.70E-08	0.04	rs478222	0.53	3.60E-09	0.007
				Adipose	<i>POMC</i>	-	4.00E-09	3.50E-04	rs6734859	0.03	6.20E-15	8.10E-10
rs13078807	3	85,966,840	G	Subcutaneous fat	<i>Contig17333_RC</i>	+	2.74E-17	0.002	rs9309982	0.29	6.51E-24	5.27E-08
				Omental fat	<i>Contig17333_RC</i>	+	1.00E-10	0.007	rs4364177	0.22	5.15E-19	1.85E-09
				Adipose	<i>Contig17333_RC</i>	+	3.00E-08	0.001	rs17518584	0.13	5.50E-11	2.30E-06
rs13107325	4	103,407,732	T	Liver	<i>SLC39A8</i>	-	1.36E-16	1.00	rs13107325	1.00	1.36E-16	1.00
rs2112347	5	75,050,998	T	Blood	<i>C5orf37</i>	-	4.20E-07	2.30E-08	rs888788	0.50	1.80E-36	6.70E-38
rs206936	6	34,410,847	G	Liver	<i>NUDT3</i>	+	2.00E-07	0.15	rs6911272	0.54	5.24E-13	9.73E-04
rs4929949	11	8,561,169	C	Omental fat	<i>Contig39122_RC</i>	-	6.14E-16	0.88	rs10840111	0.17	1.77E-109	4.94E-74
				Subcutaneous fat	<i>Contig39122_RC</i>	-	3.39E-14	0.04	rs10840111	0.16	6.88E-82	6.17E-56
				Liver	<i>Contig39122_RC</i>	-	2.21E-10	0.34	rs10840111	0.20	4.12E-51	1.32E-32
				Adipose	<i>RPL27A</i>	-	1.70E-12	0.001	rs11042036	0.13	4.50E-47	4.20E-36
				Blood	<i>STK33</i>	-	8.90E-16	0.003	rs7934876	0.12	3.90E-36	2.60E-22
				Adipose	<i>TRIM66</i>	+	2.20E-10	0.002	rs10840106	0.14	2.50E-82	1.10E-71
				Blood	<i>TRIM66</i>	+	9.80E-08	0.04	rs10840106	0.14	4.90E-86	8.20E-78
				Omental fat	<i>TRIM66</i>	+	1.50E-07	0.54	rs10840106	0.12	1.54E-45	5.49E-34
				Liver	<i>TRIM66</i>	+	1.02E-06	0.001	rs10840106	0.11	4.77E-08	1.06E-04
rs10767664	11	27,682,562	A	Blood	<i>LGR4</i>	-	4.50E-07	0.36	rs11030024	0.44	6.60E-09	0.003
				Lymphocyte	<i>LGR4</i>	-	1.01E-06	0.006	rs7937671	0.10	3.97E-11	4.40E-07
rs3817334	11	47,607,569	T	Blood	<i>ACP2</i>	+	1.50E-07	0.31	rs2013867	0.21	2.40E-30	1.40E-23
				Adipose	<i>CELF1</i>	+	2.00E-45	0.08	rs10838738	0.84	1.70E-56	2.40E-10
				Blood	<i>CELF1</i>	+	2.70E-13	0.83	rs12794570	0.82	4.30E-17	6.40E-05
				Omental fat	<i>CUGBP1</i>	+	2.67E-19	0.65	rs2290850	0.72	9.26E-25	0.02
				Lymphocyte	<i>FNBP4</i>	+	4.26E-18	0.71	rs2290851	0.65	2.76E-32	1.27E-09
				Omental fat	<i>MTCH2</i>	+	2.83E-14	0.72	rs10838738	0.78	1.50E-19	0.03
				Subcutaneous fat	<i>MTCH2</i>	+	6.79E-12	0.91	rs12794570	0.76	2.62E-14	0.25
				Brain	<i>MTCH2</i>	+	7.18E-08	NA	NA	NA	NA	NA
				Blood	<i>MTCH2</i>	+	3.10E-26	0.52	rs1317149	0.84	7.50E-39	1.90E-12
				Adipose	<i>MTCH2</i>	+	1.30E-08	0.61	rs11039433	0.84	1.20E-10	0.003
				Blood	<i>MYBPC3</i>	-	1.70E-18	0.86	rs4992357	0.70	3.50E-33	1.10E-14
				Adipose	<i>MYBPC3</i>	-	5.50E-14	0.81	rs755553	0.76	1.30E-22	2.60E-09
				Lymphocyte	<i>NDUFS3</i>	-	2.41E-07	0.99	rs4752845	0.85	3.86E-09	0.15
				Blood	<i>NR1H3</i>	+	8.30E-06	0.43	rs7395581	0.21	8.40E-12	2.50E-07
				Adipose	<i>NUP160</i>	+	7.90E-06	0.06	rs12787112	0.84	1.40E-10	8.40E-07
rs4771122	13	26,918,180	G	Lymphocyte	<i>GTF3A</i>	-	6.73E-12	0.77	rs7988412	0.85	5.57E-15	0.08
				Blood	<i>GTF3A</i>	-	9.50E-21	0.54	rs7988412	0.83	4.00E-22	0.02
				Lymphocyte	<i>POLR1D</i>	+	6.19E-05	5.38E-04	rs7097	0.00	5.70E-21	5.81E-21
rs2241423	15	65,873,892	G	Omental fat	<i>HSS00330420</i>	+	2.13E-16	0.53	rs3784700	0.63	5.48E-20	0.03
				Subcutaneous fat	<i>HSS00330420</i>	+	5.48E-10	0.91	rs3784700	0.64	1.59E-13	0.05
				Omental fat	<i>MAP2K5</i>	+	9.97E-12	0.91	rs12441823	0.55	3.27E-20	1.31E-04
				Adipose	<i>MAP2K5</i>	+	2.80E-11	0.81	rs4776361	0.45	5.50E-25	3.50E-14
				Subcutaneous fat	<i>MAP2K5</i>	+	7.49E-09	0.77	rs12902812	0.58	1.08E-17	1.90E-04
				Blood	<i>MAP2K5</i>	+	1.80E-20	0.54	rs4776361	0.45	2.00E-46	1.30E-24
				Adipose	<i>SMAD3</i>	-	9.50E-08	0.007	rs11071942	0.19	4.90E-14	4.30E-09
				Blood	<i>SMAD3</i>	+	1.10E-06	0.08	rs10152913	0.26	3.40E-17	2.60E-12
rs12444979	16	19,841,101	C	Omental fat	<i>C16orf88</i>	-	2.54E-36	0.05	rs11647001	0.42	4.93E-90	1.42E-30
				Adipose	<i>C16orf88</i>	-	1.20E-19	0.94	rs2074037	0.21	1.80E-41	1.50E-20
				Lymphocyte	<i>C16orf88</i>	-	8.88E-13	0.69	rs3829539	0.16	5.83E-33	1.35E-17
				Subcutaneous fat	<i>C16orf88</i>	-	1.17E-12	0.10	rs1016971	0.45	8.82E-47	4.21E-19
				Blood	<i>C16orf88</i>	-	1.00E-06	0.60	rs8045761	0.14	1.60E-14	5.50E-09
				Subcutaneous fat	<i>GPRC5B</i>	+	1.08E-09	0.11	rs8045761	0.44	3.48E-10	0.06
				Omental fat	<i>IQCK</i>	+	1.71E-09	0.91	rs11865578	0.83	3.17E-12	0.33
				Subcutaneous fat	<i>IQCK</i>	+	8.79E-07	1.00	rs12444979	1.00	8.79E-07	1.00
rs7359397	16	28,793,160	T	Blood	<i>APOB48R</i>	+	8.00E-08	0.70	rs2411453	0.82	4.70E-08	0.28
				Adipose	<i>EIF3C</i>	+	5.40E-19	0.72	rs7189927	0.73	1.20E-22	1.30E-04
				Blood	<i>EIF3C</i>	+	7.60E-13	0.72	rs7189927	0.73	3.90E-15	0.002
				Lymphocyte	<i>EIF3S8</i>	+	9.88E-10	NA	rs7189927	0.79	1.50E-13	NA
				Liver	<i>NUPR1</i>	+	5.67E-14	0.68	rs3743963	0.49	1.14E-25	9.17E-07
				Adipose	<i>SH2B1</i>	-	1.50E-12	0.68	rs12928404	0.92	9.20E-13	0.30
				Omental fat	<i>SH2B1</i>	-	1.85E-06	1.00	rs7359397	1.00	1.85E-06	1.00
				Lymphocyte	<i>SPNS1</i>	+	9.66E-10	1.00	rs7500321	0.58	7.63E-16	3.19E-04
				Subcutaneous fat	<i>SULT1A1</i>	+	1.48E-13	1.00	rs7359397	1.00	1.48E-13	1.00
				Adipose	<i>SULT1A1</i>	+	7.70E-15	0.83	rs151181	0.75	1.80E-24	3.50E-10

Supplementary Table 7

BMI SNP	Chr	Position	BMI increasing allele	Tissue	Gene	Effect ^a	<i>P</i> -value	<i>P</i> _{adj} ^b	Peak SNP ^c	<i>r</i> ² ^d	<i>P</i> -value	<i>P</i> _{adj} ^e
				Blood	<i>SULT1A1</i>	+	1.20E-06	0.002	rs2291937	0.00	1.70E-10	2.10E-07
				Subcutaneous fat	<i>SULT1A2</i>	+	4.11E-20	0.75	rs1074631	0.80	4.97E-22	0.30
				Omental fat	<i>SULT1A2</i>	+	5.43E-11	0.93	rs151181	0.70	2.21E-14	0.06
				Adipose	<i>SULT1A2</i>	+	5.60E-19	0.66	rs151181	0.75	2.70E-31	2.50E-12
				Blood	<i>SULT1A2</i>	+	5.20E-10	0.45	rs12446550	0.86	7.80E-12	0.004
				Adipose	<i>SULT1A3</i>	+	4.40E-16	0.47	rs151181	0.75	1.50E-24	3.70E-09
				Lymphocyte	<i>TUFM</i>	+	1.19E-07	0.82	rs12928404	0.98	1.83E-08	0.66
				Blood	<i>TUFM</i>	+	9.10E-69	0.10	rs8049439	0.96	9.10E-71	0.003
rs3810291	19	52,260,843	A	Lymphocyte	<i>TMEM160</i>	-	3.09E-05	0.49	rs1862499	0.14	3.10E-06	0.11
				Blood	<i>ZC3H4</i>	-	2.60E-16	0.24	rs8101091	0.66	1.80E-20	1.60E-05
				Adipose	<i>ZC3H4</i>	-	9.00E-09	1.00	rs3810291	1.00	9.00E-09	1.00

* we distinguished 'adipose' tissue from a general population and 'omental fat' and subcutaneous fat' from patients who underwent bariatric surgery (see **Methods**)

^a Direction of effect for the BMI increasing allele

^b *P*-value for the BMI SNP after conditioning on the most significant SNP for the gene transcript.

^c Most significant SNP associated with the gene transcript

^d Correlation between the BMI SNP and the peak SNP

^e *P*-value for the peak SNP after conditioning on the BMI SNP

Supplementary Table 8

Supplementary Table 8 - Estimated number of BMI loci for each of the effect sizes observed in stage 2 for the SNPs that reached a genome-wide significance of 5×10^{-8} in the joint analysis of stage 1 and stage 2, given the power to detect the association in the joint analysis of stage 1 and stage 2.

	SNP	MAF*	Mean Difference	Standardized effect size	Power (%)	Estimated number of loci
1	rs29941	0.329	-0.013	7.35E-05	1%	50.2
2	rs4929949	0.486	-0.013	8.71E-05	2%	43.3
3	rs206936	0.206	0.017	9.11E-05	3%	41.4
4	rs4836133	0.49	0.016	1.20E-04	8%	27.8
5	rs11847697	0.039	0.042	1.30E-04	11%	23.2
6	rs1555543	0.415	-0.017	1.40E-04	14%	18.9
7	rs13078807	0.193	0.022	1.44E-04	15%	17.3
8	rs4771122	0.242	0.021	1.56E-04	19%	12.3
9	rs3817334	0.398	0.018	1.57E-04	20%	12
10	rs2890652	0.2	0.024	1.86E-04	32%	4.3
11	rs1514175	0.434	0.02	1.95E-04	36%	3.8
12	rs3810291	0.343	-0.022	2.16E-04	45%	2.9
13	rs2112347	0.381	-0.022	2.28E-04	51%	2.3
14	rs887912	0.275	0.024	2.30E-04	51%	2.3
15	rs10968576	0.331	0.023	2.34E-04	53%	2.2
16	rs13107325	0.067	0.048	2.82E-04	71%	1.6
17	rs10150332	0.223	0.029	2.95E-04	75%	1.5
18	rs987237	0.185	0.032	3.10E-04	79%	1.4
19	rs9816226	0.183	-0.034	3.36E-04	85%	1.3
20	rs2241423	0.208	-0.033	3.53E-04	88%	1.3
21	rs7138803	0.382	0.028	3.75E-04	91%	1.2
22	rs12444979	0.135	-0.042	4.04E-04	94%	1.1
23	rs2287019	0.204	-0.037	4.54E-04	97%	1.1
24	rs2815752	0.378	-0.032	4.81E-04	98%	1.1
25	rs7359397	0.4	0.035	5.81E-04	100%	1
26	rs713586	0.462	0.036	6.41E-04	100%	1
27	rs10767664	0.208	-0.048	7.50E-04	100%	1
28	rs543874	0.188	0.052	8.19E-04	100%	1
29	rs10938397	0.442	0.043	9.12E-04	100%	1
30	rs571312	0.226	0.057	1.14E-03	100%	1
31	rs2867125	0.172	-0.078	1.75E-03	100%	1
32	rs1558902	0.419	0.092	4.08E-03	100%	1
Estimated number of total loci						283.5 (132.3, 510.0)*
Total phenotypic variance explained (%)						4.50% (3.1, 6.8)*

† Numbers in parenthesis are for a 95% confidence interval by the percentile-based approach

1.2. SUPPLEMENTARY FIGURES

Supplementary Figure 1 Study design.	P. 15
Supplementary Figure 2 Regional plots of the 32 confirmed BMI loci with missense and CNV variants.	P. 17
Supplementary Figure 3 Quantile-quantile plot of SNPs at stage 1 GIANT meta-analysis.	p. 24
Supplementary Figure 4 Relationship between the predicted BMI, based on the 32 confirmed BMI loci combined, and the actual BMI in the ARIC Study (N=8,120).	p. 26

Supplementary Figure 1 Study design. Stage 1 - Meta analysis of genome-wide association data was performed in stage 1 across 46 studies of white European Ancestry. A total of 42 SNPs representing the best associating ($P < 5.10^{-6}$) loci (shown) were taken forward for replication. Nineteen of these SNPs (loci in bold) reached already genome-wide significance at stage 1. Stage 2 – The 42 SNPs were genotyped in 16 de novo replication studies and extracted from 18 *in silico* replication studies, all adults of European ancestry and were tested for association with BMI. In a joint analyses of stage 1 and stage 2 data, 32 SNPs (loci in bold) reached genome-wide significance ($P < 5 \times 10^{-8}$). Follow-up analyses – The 32 confirmed loci were taken forward for additional analyses.

STAGE 1

GWA meta-analyses of BMI

Meta-analysis of GWA analyses of 2.8M imputed and genotyped SNPs with BMI in 123,865 adults from 46 studies.

42 loci ($P < 5 \times 10^{-6}$) taken forward



STAGE 2

Follow-up of the most significant loci in additional samples

Association analyses of 42 SNPs with BMI in 125,931 adults.

de novo replication
(N = 79,561 from 16 studies)

In silico replication
(N = 46,370 from 18 studies)

32 loci ($P < 5 \times 10^{-8}$) were confirmed



Stage 1 + stage 2 meta-analyses of 42 SNPs

($n_{\max} = 249,796$).

Follow-up analyses

Additional analyses of 32 confirmed BMI loci

- Association with risk of extreme obesity
- Association with BMI-related traits
- eQTL analyses
- Pathway analyses
- CNV and functional variant analyses
- Explained variance and polygenes
- 2nd signal analyses
- Dominant, recessive analyses
- Gene-by-gene and Gene-by-sex

<i>Previous BMI loci</i>	FTO	TMEM18
	MC4R	GNPDA2
	BDNF	NEGR1
	SH2B1	ETV5
	MTCH2	KCTD15

<i>Previous weight & waist loci</i>	SEC16B	TFAP2B
	FAIM2	NRXN3

RBJ	GPRC5B
MAP2K5	QPCTL
TNNI3K	SLC39A8
FLJ35779	LRRN6C
TMEM160	FANCL
CADM2	PRKD1
LRP1B	PTBP2
MTIF3	ZNF608
RPL27A	NUDT3
HNF4G	ADCY9
LMX1B	CBX1
HTR1A	KIAA1505
C9orf4	REG3A
SCG3	RASA2

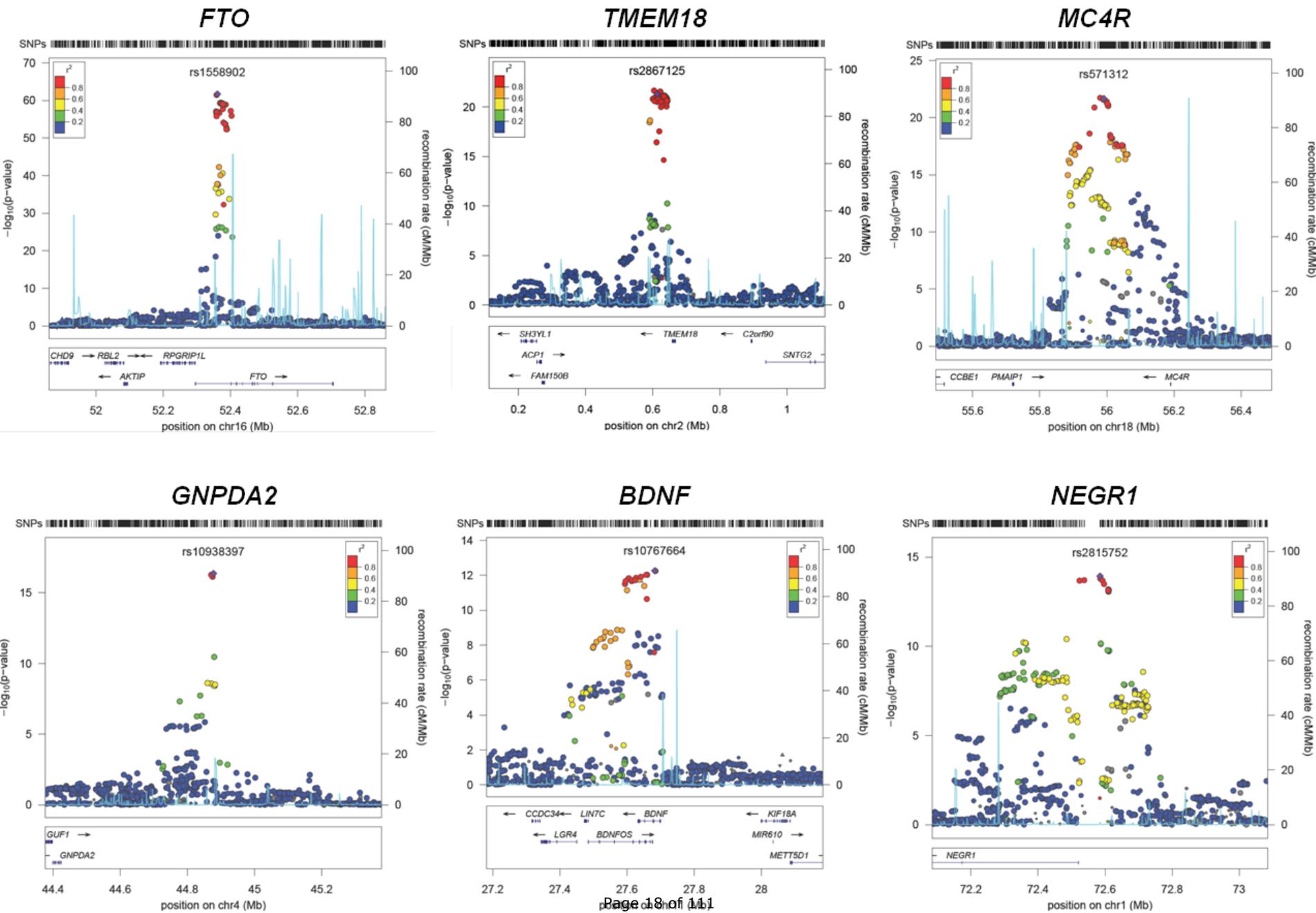
<i>Previous BMI loci</i>	FTO	TMEM18
	MC4R	GNPDA2
	BDNF	NEGR1
	SH2B1	ETV5
	MTCH2	KCTD15

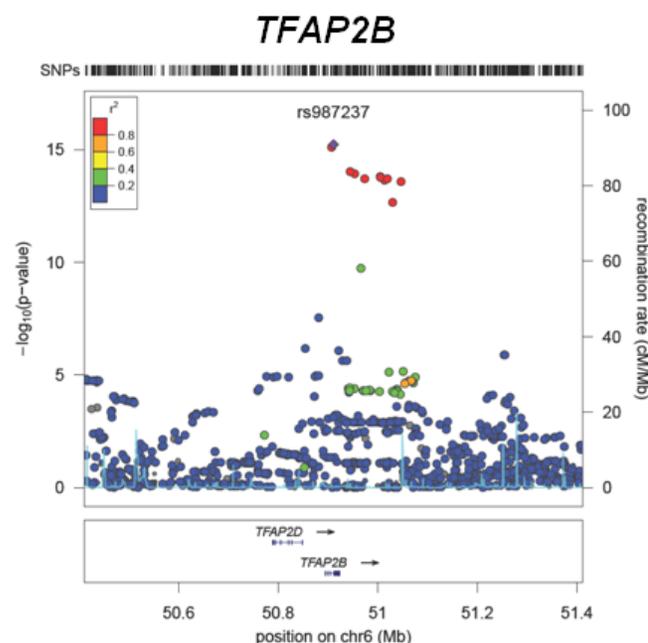
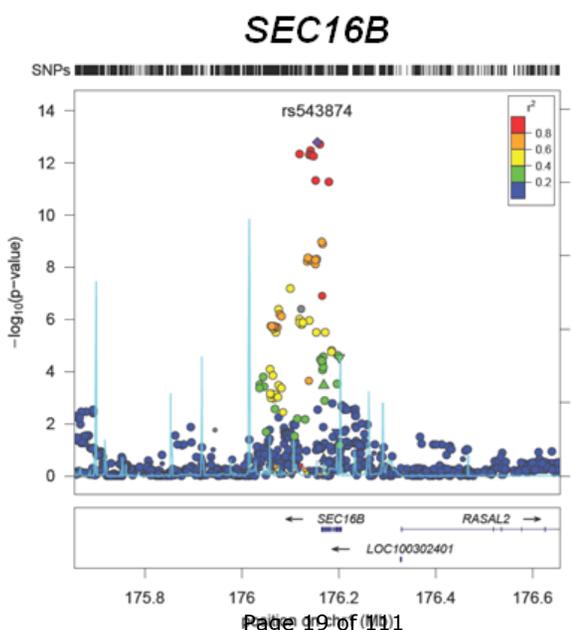
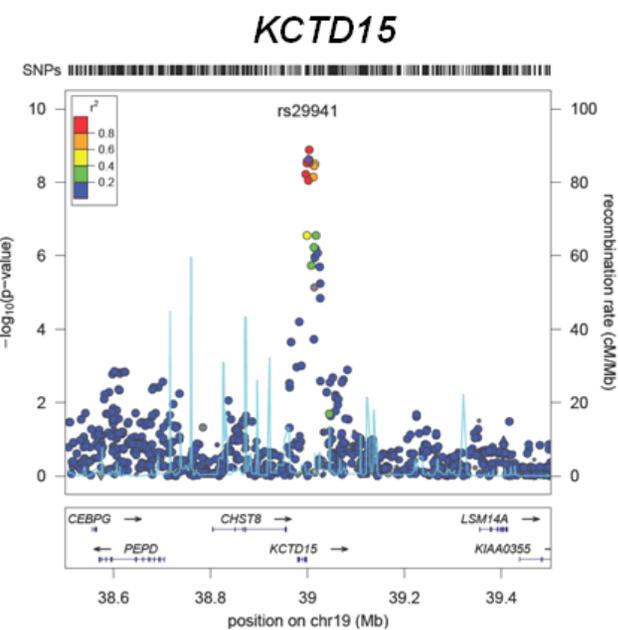
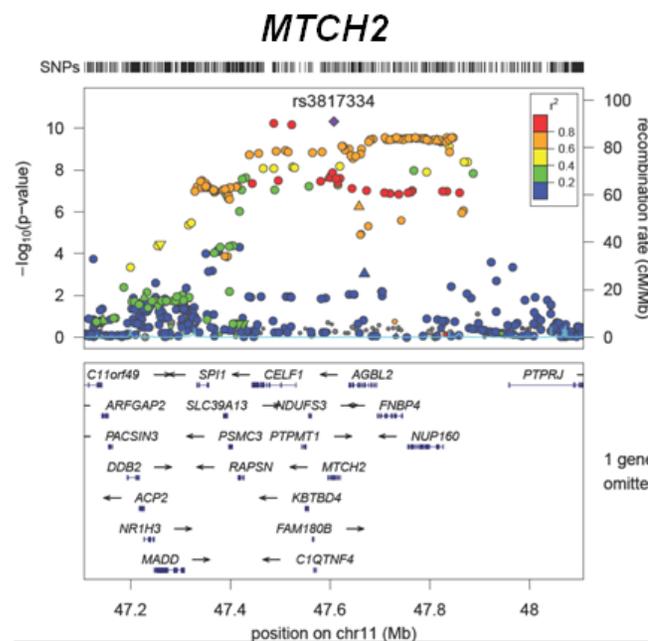
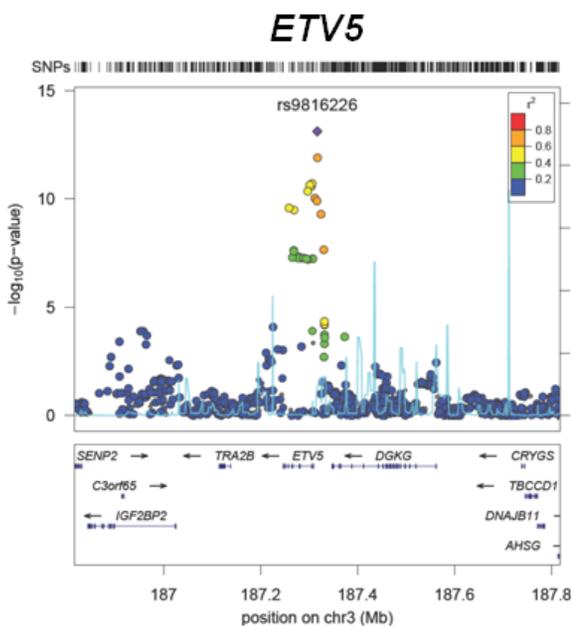
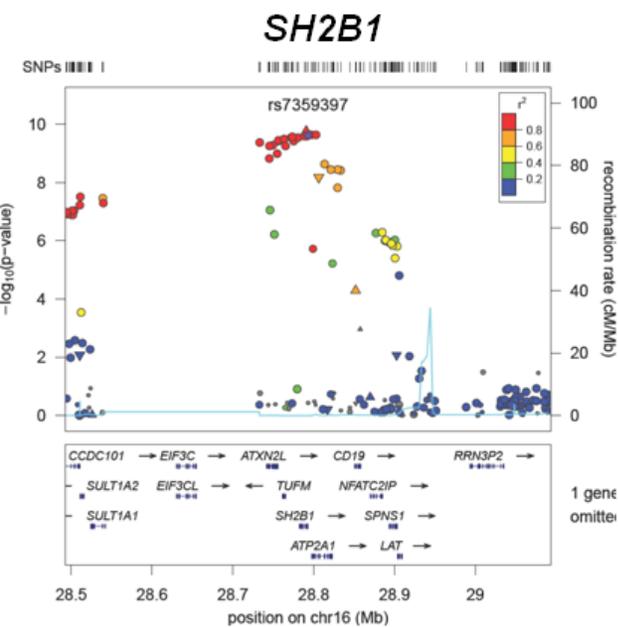
<i>Previous weight & waist loci</i>	SEC16B	TFAP2B
	FAIM2	NRXN3

<i>18 New BMI loci</i>	RBJ	GPRC5B
	MAP2K5	QPCTL
	TNNI3K	SLC39A8
	FLJ35779	LRRN6C
	TMEM160	FANCL
	CADM2	PRKD1
	LRP1B	PTBP2
	MTIF3	ZNF608
	RPL27A	NUDT3

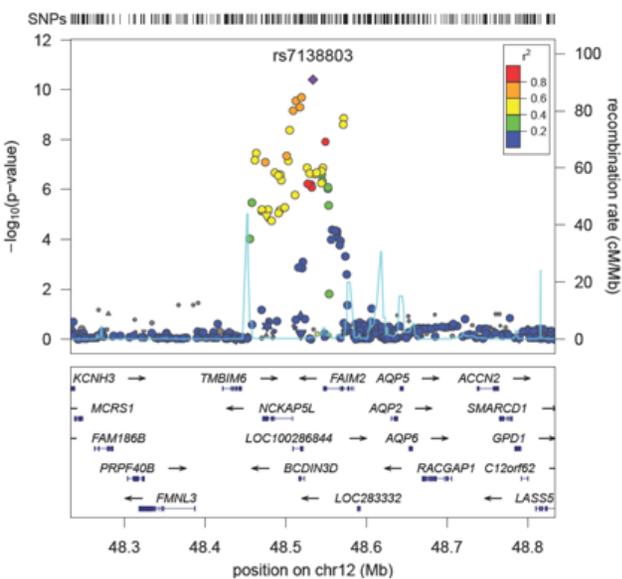
Supplementary Figure 2 Regional plots of the 32 confirmed BMI loci with missense and CNV variants. SNPs are plotted by position on chromosome against association with BMI ($-\log_{10} P$ -value). The SNP name shown on the plot was the most significant SNP after stage 1 meta-analysis. Estimated recombination rates (from HapMap) are plotted in cyan to reflect the local LD structure. The SNPs surrounding the most significant SNP are color-coded to reflect their LD with this SNP (taken from pairwise r^2 values from the HapMap CEU database, www.hapmap.org). Genes, position of exons, and direction of transcription from UCSC genome browser (genome.ucsc.edu) are noted. Hash-marks represent SNP positions available in the meta-analysis. Plots were generated using LocusZoom (<http://csg.sph.umich.edu/locuszoom>).

Supplementary Figure 2

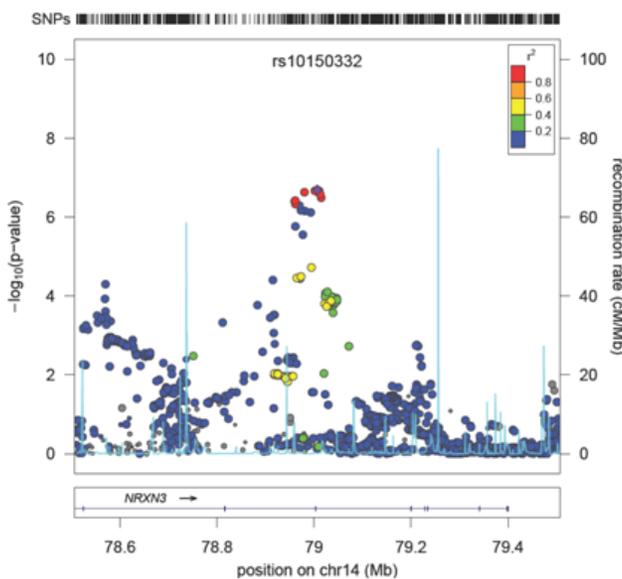




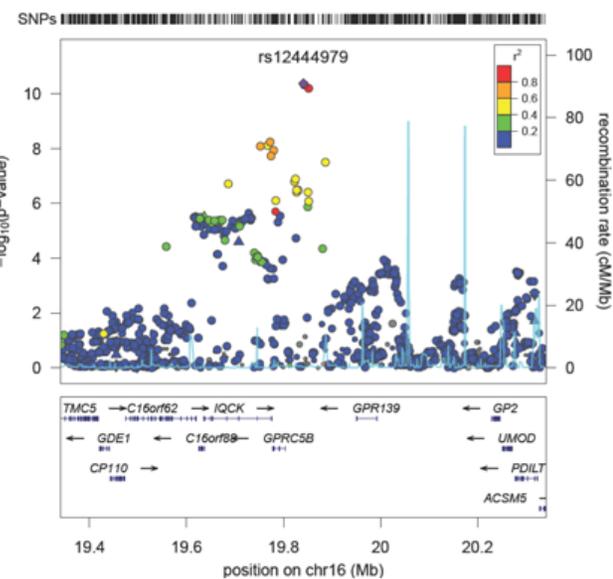
FAIM2



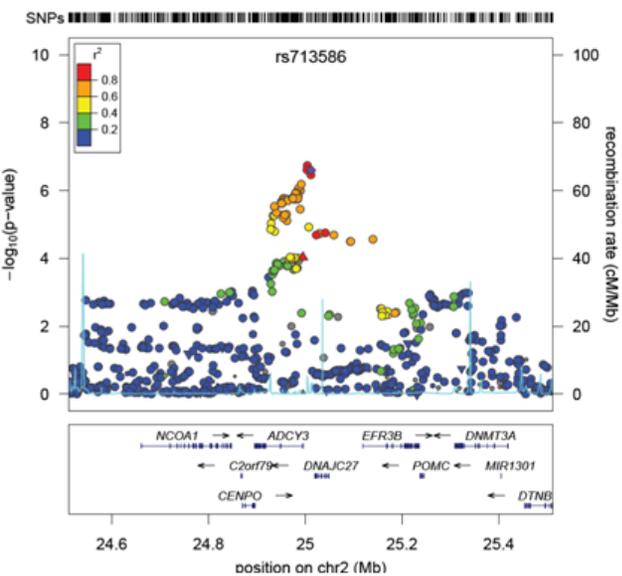
NRXN3



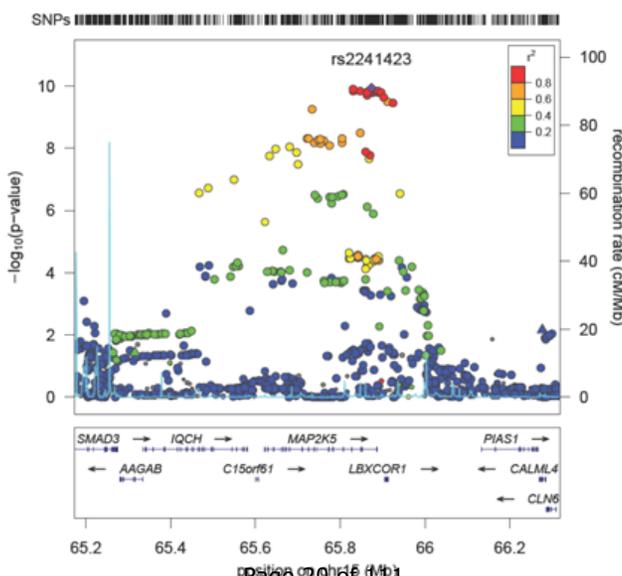
GPRC5B



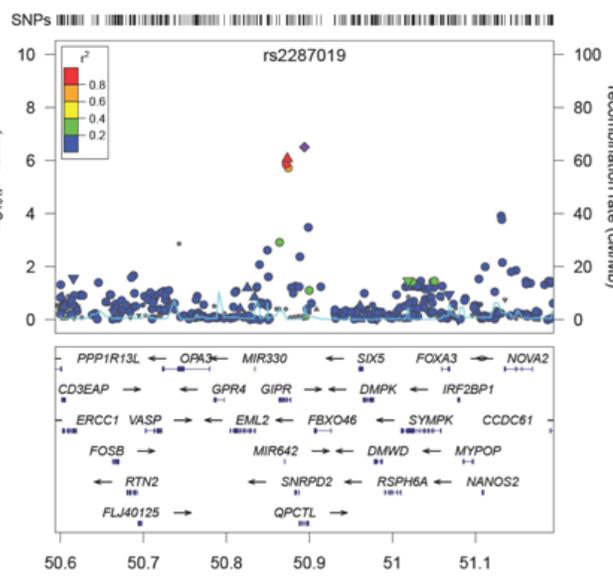
RBJ; POMC



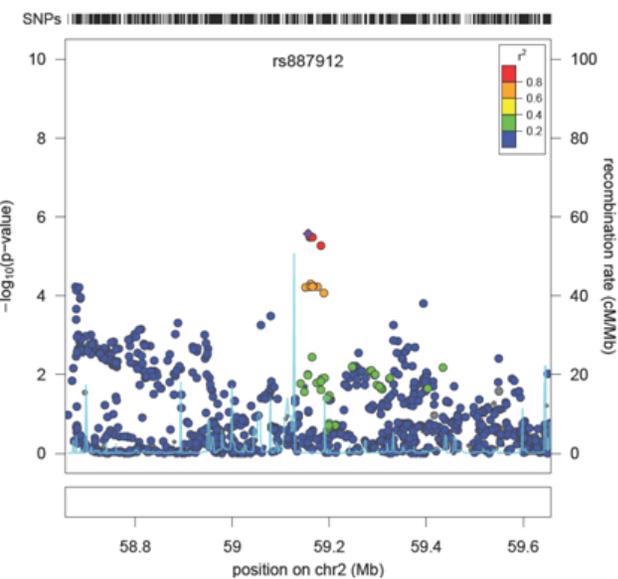
MAP2K5



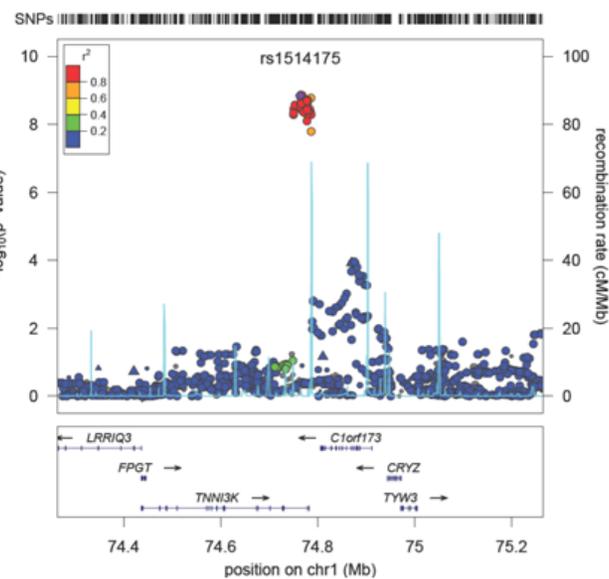
QPCTL; GIPR



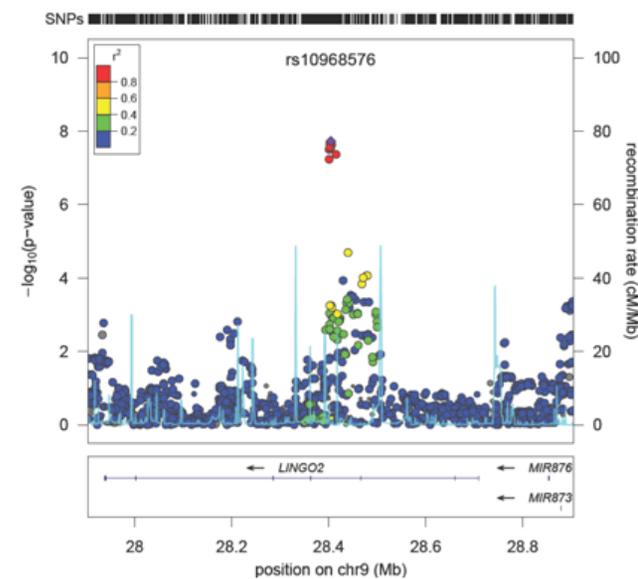
FANCL



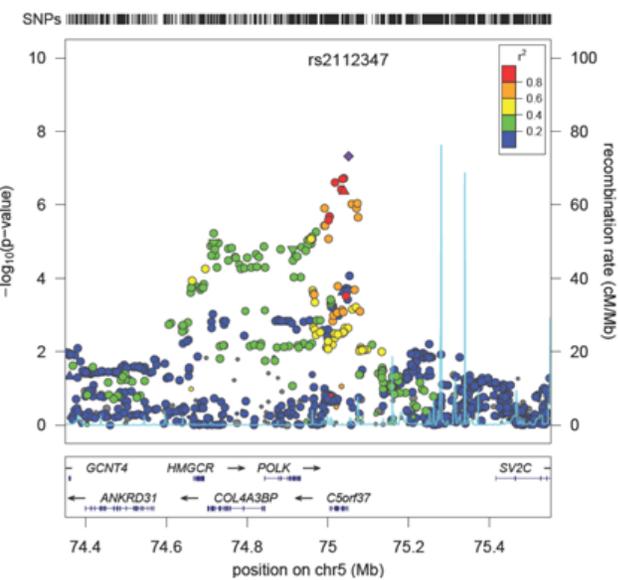
TNNIK3



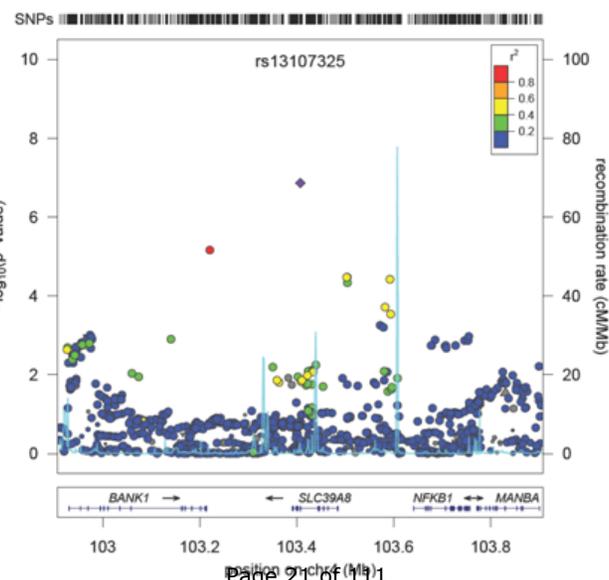
LRRN6C



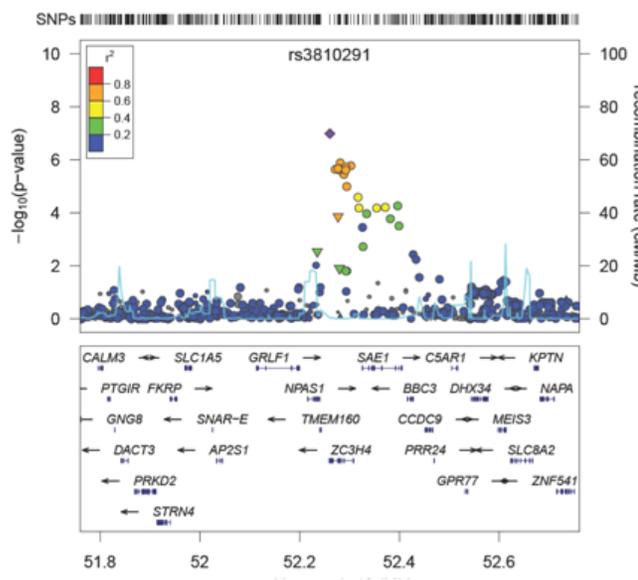
FLJ35779



SLC39A8

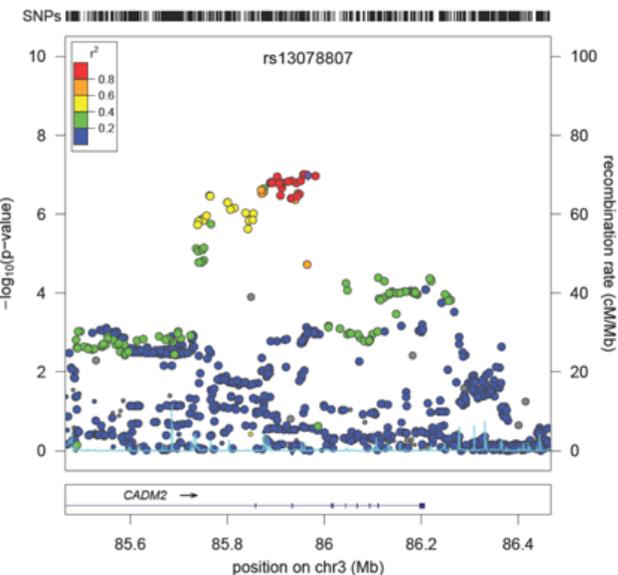


TMEM160

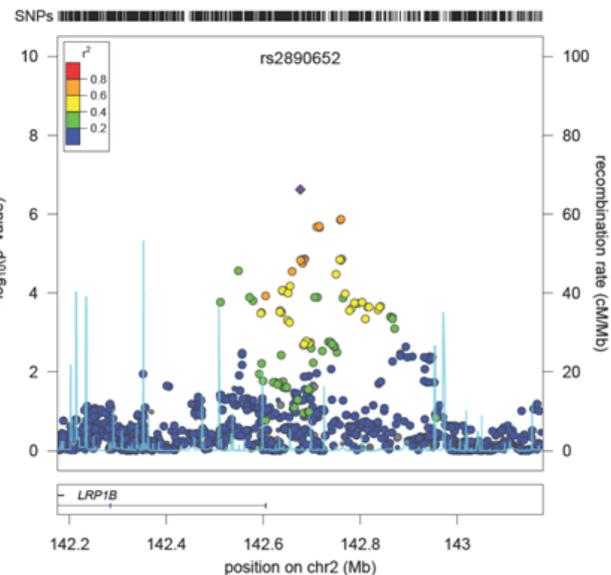


Supplementary Figure 2 (continued)

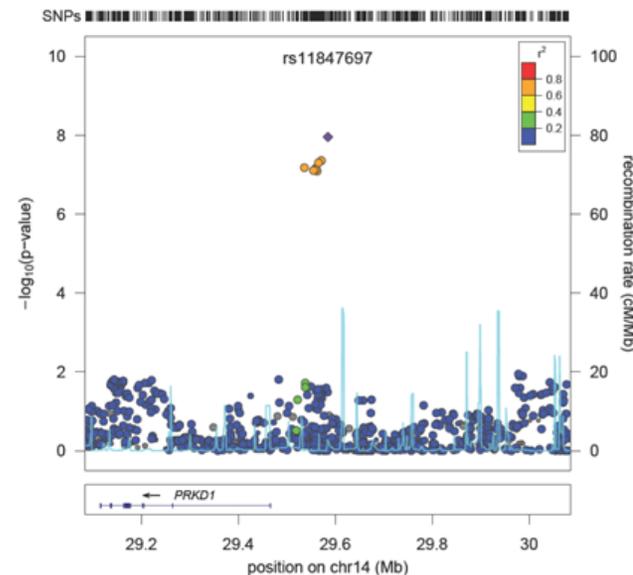
CADM2



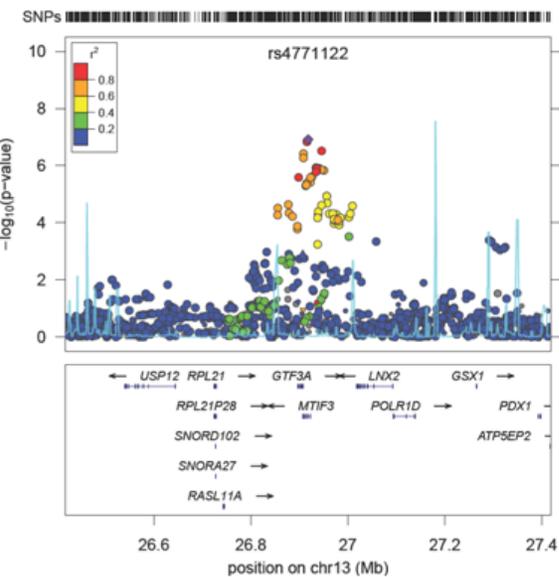
LRP1B



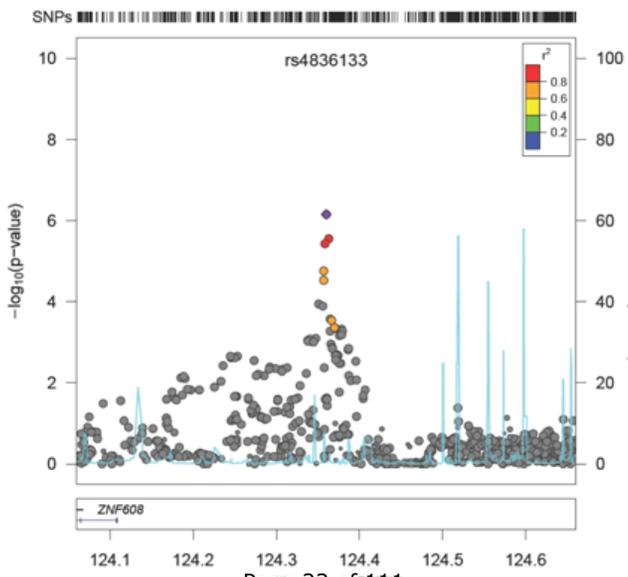
PRKD1



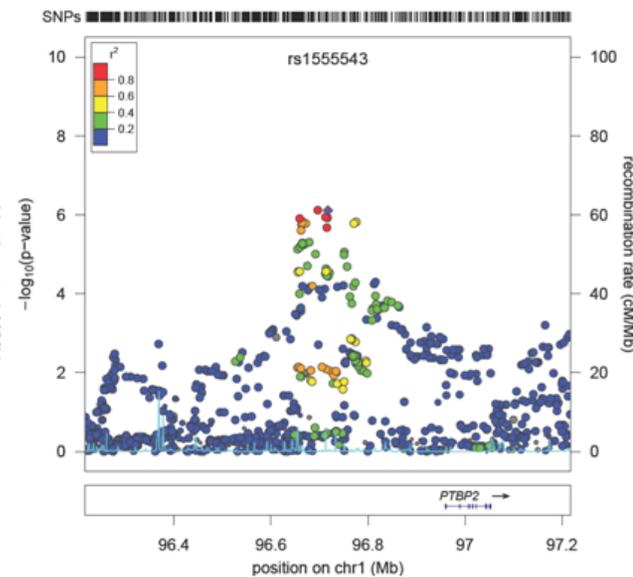
MTIF3



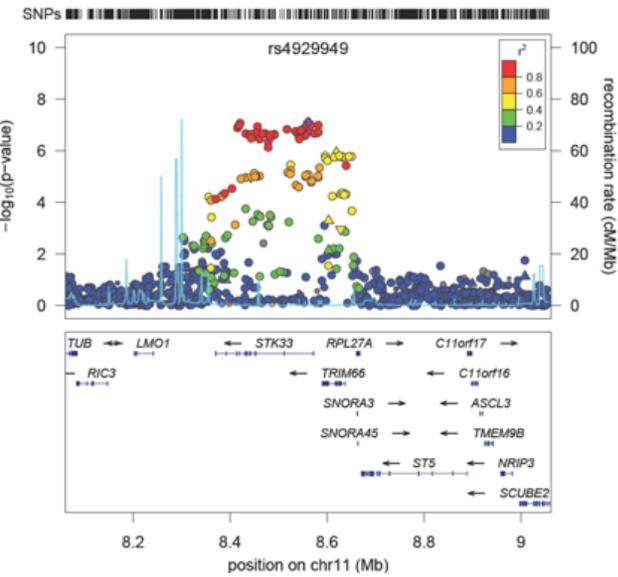
ZNF608



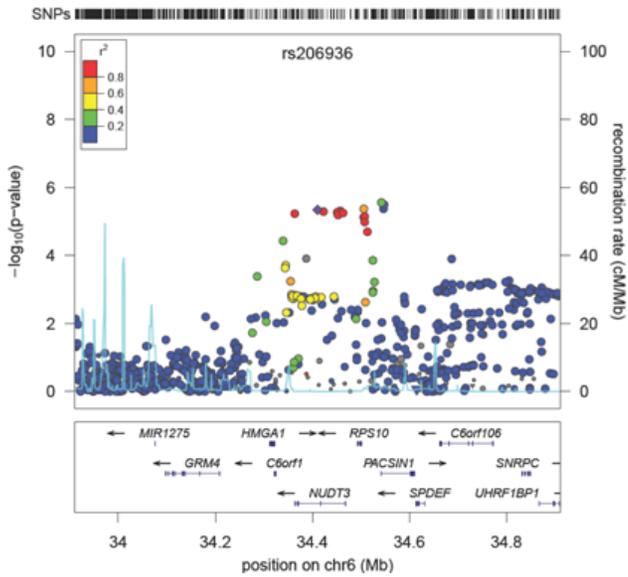
PTBP2



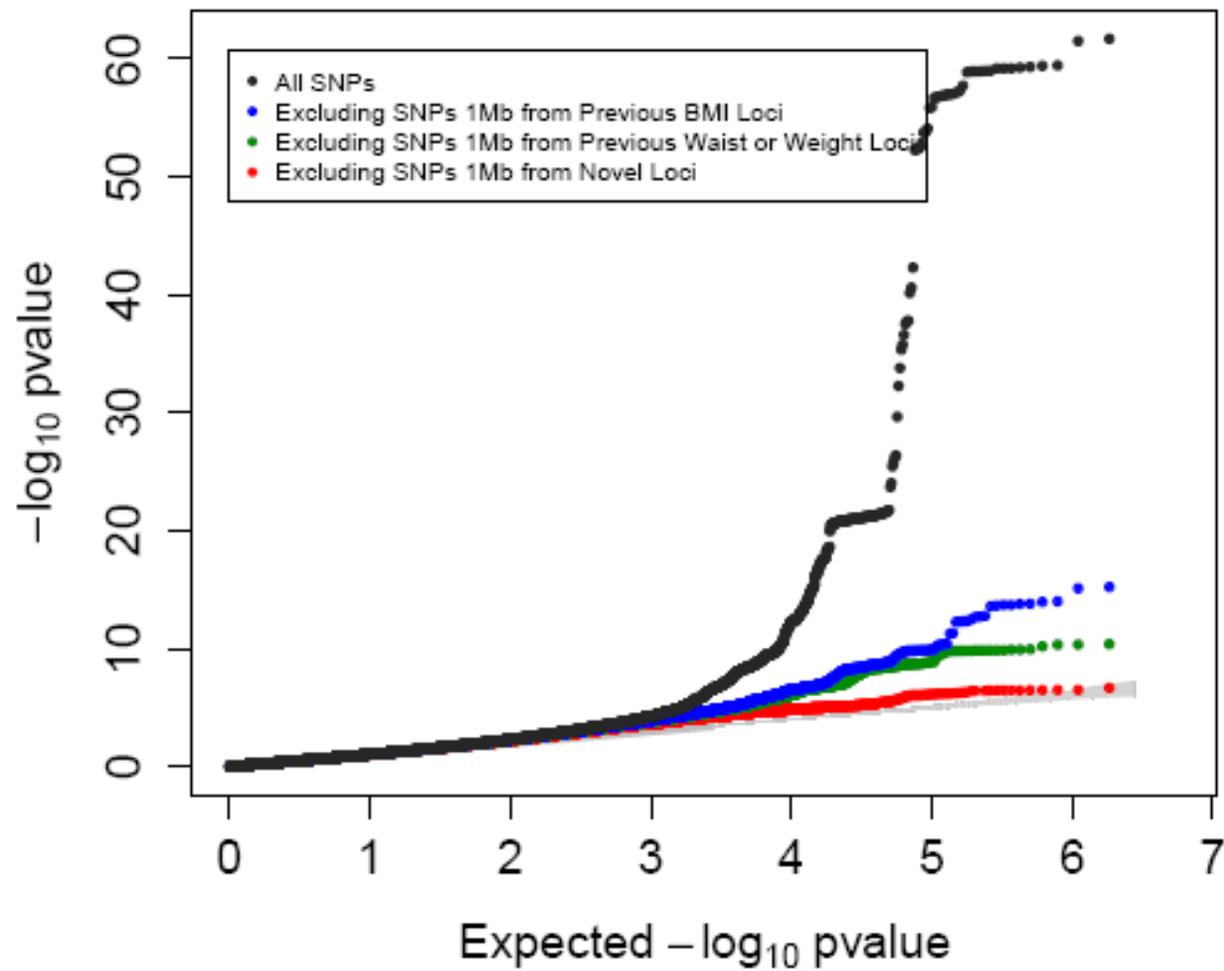
RPL27A



NUDT3

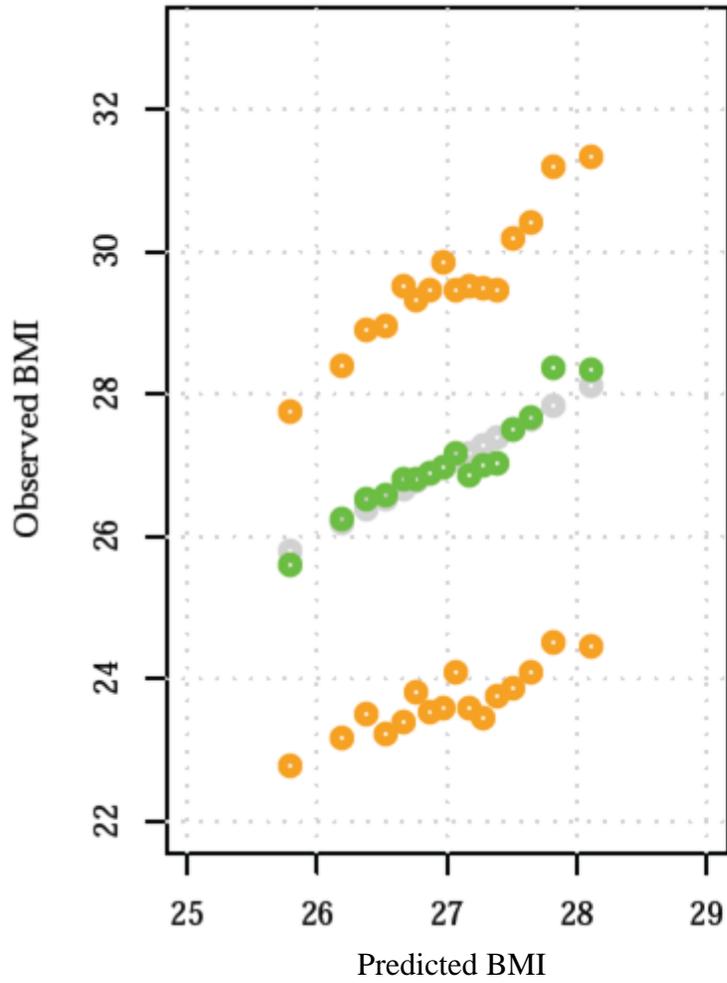


Supplementary Figure 3 Quantile-quantile plot of SNPs at stage 1 GIANT meta-analysis (black) and after removing any SNPs within 1Mb of the 10 previously reported genome-wide significant hits for BMI (blue), after additionally excluding the four loci for waist/weight (green) and after excluding all 32 confirmed loci (red).

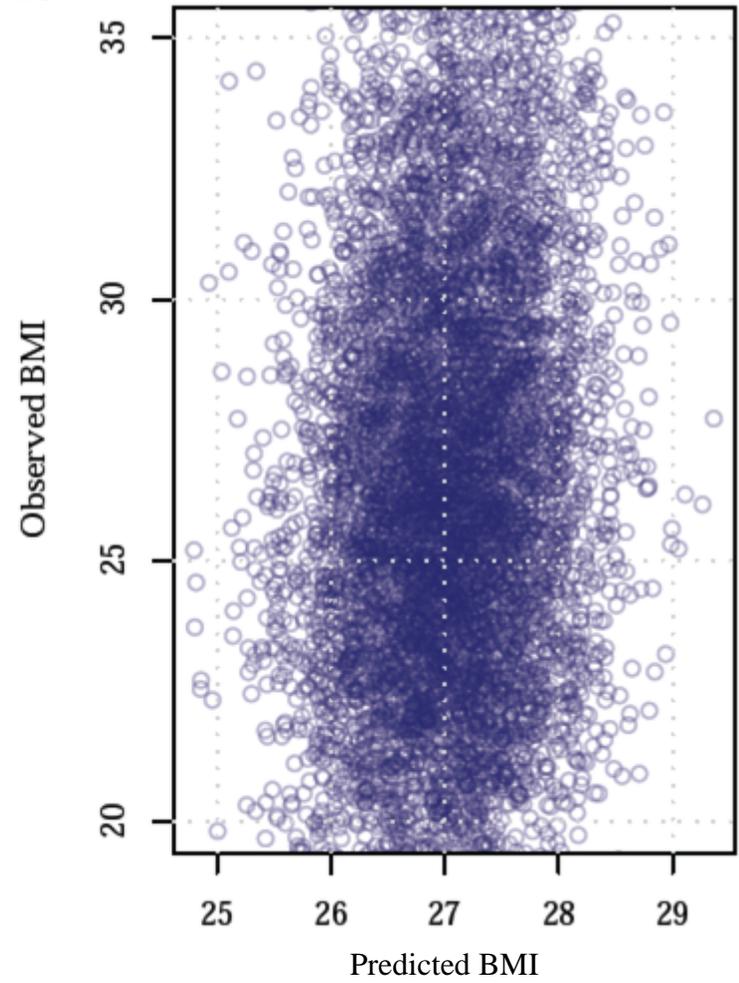


Supplementary Figure 4 Relationship between the predicted BMI, based on the 32 confirmed BMI loci combined, and the actual BMI in the ARIC Study (N=8,120). Panel (a) shows the comparison of the predicted BMI (grey), with the actual BMI (green) and quartile ranges (orange) in sets of 500 individuals, suggesting good average predictions. Panel (b) shows the comparison of the individual predicted and observed BMI values, suggesting poor individual predictions.

a.



b.



2. SUPPLEMENTARY NOTE	P. 28
2.1. Description of secondary analyses	p. 29
2.2. Supplementary note – tables	p. 38
2.3. Supplementary note – figure	p. 95
2.4. Author contributions	p. 97
2.5. Acknowledgements	p. 101
2.6. Competing interests statement	p. 111

2. SUPPLEMENTARY NOTE

2.1. DESCRIPTION OF SECONDARY ANALYSES

2.1.1. Predictive value and cumulative effects of confirmed loci

To estimate the cumulative effect of the 32 confirmed BMI loci combined, we calculated a genetic risk score in the Atherosclerosis Risk in Communities (ARIC) study (N = 8,120), one of our largest stage 1 samples. The genetic risk score was calculated by summing the number of risk alleles (0,1,2) carried by an individual or the number of “best guess” risk alleles from imputed data at each of the 32 confirmed SNPs, weighted by the relative effect sizes of that SNP estimated from stage 2 data. The genetic risk score was rounded to the nearest integer to represent the individual’s risk allele score (range 16-44). The mean BMI in each genetic risk score category was calculated.

To assess the predictive ability of the SNPs for obesity (BMI \geq 30 kg/m²), the area under the ROC curve was obtained from the c-statistic of the logistic regression model in which the 32 confirmed SNPs, represented by the weighted genetic risk score (see above), were predictors coded in an additive genetic model with obesity as the outcome, adjusted for age, age², and sex. We estimated the AUC for a model with and without inclusion of the 32 SNPs. The *P*-values for these analyses were obtained from the corresponding models.

Furthermore, we assessed the ability of the 32 confirmed SNPs to predict BMI in the ARIC study. Predicted BMI for each individual was calculated based on mean BMI in the study population plus the individual risk allele count multiplied by the effect on BMI per risk allele, summed over all 32 confirmed BMI loci. We subsequently compared the predicted BMI with the observed BMI using the average values of sets of 500 individuals as well as using the individual values.

2.1.2. Association analyses with obese and overweight

Association between the 32 confirmed BMI variants and overweight (defined as BMI \geq 25 kg/m²) or obese status (BMI \geq 30 kg/m²) was assessed in stage 2 samples using logistic regression analysis, adjusting for age and age². Tests assumed an additive genetic model and were stratified by sex and case status where appropriate. Summary statistics (betas and standard errors) were subsequently meta-analysed using the inverse variance method in METAL, and the meta-analysis results were converted back to ORs and 95% CIs.

2.1.3. Effects in specific populations – children and extreme obesity

We tested for association between the 32 confirmed SNPs with BMI in children and adolescents in four population-based studies (total N=8,540, **Supplementary Note – Tables 7-9**). In three of these studies (*de novo*), the 32 SNPs were directly genotyped,

and in one study (*in silico*), they were imputed. Before analyses, BMI was standardized by age and sex in each study and only SNPs that met the QC criteria described for stage 2 samples were included (**Supplementary Note – Tables 7-9**). Associations between SNPs and BMI assumed an additive genetic model and summary statistics were subsequently meta-analyzed using the inverse variance method in METAL.

Furthermore, association between the 32 SNPs and extreme or early onset obesity were tested in seven case-control studies (three with *de novo* genotyping, four with *in silico* data) of extremely obese adults (2 studies; N=1,686 cases) and extremely obese children or adolescents (5 studies; total N=5,136 cases) (**Supplementary Note – Tables 7-9**). The seven studies underwent QC as described before, and two meta-analyses were performed: [1] adults and children/adolescents combined, and [2] children/adolescents only, using the inverse variance method in METAL.

To ensure consistency of results, the meta-analyses described above were repeated using a weighted Z-score method. The *P*-values obtained by both methods were highly correlated ($r > 0.99$), and we present the results of the inverse variance meta-analysis.

In addition, we tested whether BMI-increasing alleles of the 32 confirmed SNPs were overtransmitted to extremely obese offspring using a transmission disequilibrium test (TDT) in two family-based studies (one *in silico* and one *de novo*). Summary statistics were meta-analysed using a weighted Z-score method weighed by the two-thirds of the sample size of each study (i.e offspring and one parent per trio).

2.1.4. Association analyses with BMI-related traits

Association between the 32 confirmed BMI SNPs and height and weight were extracted from stage 2 replication samples. The analyses were performed in a manner identical to BMI, except for height where age-adjusted Z-scores, calculated for men and women separately, were used.

Association between the 32 confirmed BMI SNPs and related traits were extracted from previously reported GWA meta-analyses for type 2 diabetes (DIAGRAM Consortium¹), lipid levels (the Global Lipids Genetics Consortium²), glycemic traits (MAGIC^{3,4}).

2.1.5. Enrichment analysis of BMI associations in biological pathways

To discover potentially new pathways associated with BMI, and to test whether the 32 confirmed BMI SNPs cluster near genes that constitute specific biological connections, we tested whether predefined biological processes or molecular functions that contain at least one gene within 300 kb of the 32 confirmed BMI SNPs were enriched for multiple modest BMI associations (**Supplementary Table 5**). Therefore, we applied an adaptation of the gene set enrichment analysis (GSEA) framework named MAGENTA

Supplementary Note

(Meta-Analysis Gene-set Enrichment of variaNT Associations)⁵ to the BMI meta-analysis SNP data. First, we calculated a corrected gene association P-value for each gene in the genome. The gene P -value is based on the most significant SNP BMI association P-value of all SNPs within 110 kb upstream and 40 kb downstream to each gene's most extreme transcript start and end sites, respectively, adjusted for potential confounders such as gene size, number of SNPs per gene and linkage-related properties. Subsequently, we grouped genes into pathways using annotations from the KEGG, PANTHER, Gene Ontology (GO), and Ingenuity databases (from the Molecular Signatures Database, MsigDB, <http://www.broad.mit.edu/gsea/msigdb/collections.jsp>). The PANTHER molecular function and biological process gene-sets were downloaded from the PANTHER website (<http://www.pantherdb.org/>). Finally, for each pathway, we evaluated potential enrichment of highly ranked gene scores by comparing the fraction of genes within each gene set whose corrected P-value is more significant than the 75th percentile of all gene P-values to that of 10,000 randomly sampled gene sets of identical size from the genome. The 75th percentile cut-off was chosen based on our polygenic model analysis that suggests there may be many small genetic effects for BMI (**Fig. 4a**). We adjusted for genes in a given gene set that are physically adjacent in the genome by keeping only one of each subset of genes assigned the same best SNP (the gene with the most significant gene score was retained).

2.1.6. Functional variants and candidate genes within the 32 confirmed loci

To identify SNPs in each of the 32 loci that may be causal, we selected all lead SNPs and identified any SNP in $r^2 > 0.7$ (from 1000 Genomes CEU or HapMap CEU) with either the lead SNP or a HapMap $r^2=1$ proxy, to account for lead SNPs that may have been poorly genotyped in the 1000 Genomes. From this list of SNPs and proxies from 1000 Genomes or HapMap, we identified SNPs that were likely non-synonymous, nonsense or occurred within 5bp of the exon/intron boundary by comparing the SNP alleles and position to the transcription start sites and initiation codons of known genes from UCSC.

To identify candidate genes in the regions of association, we identified any gene within 500kb of the 32 confirmed BMI SNPs and performed an automated search of PubMed (<http://www.pubmed.com>) using search terms "BMI" or "obesity" and each gene name. We also scanned the OMIM (<http://www.ncbi.nlm.nih.gov/omim>) entry for each gene to identify the presence of the search terms. We used a bioinformatics tool, Snipper (<http://csg.sph.umich.edu/boehnke/snipper>), to perform the search. To highlight results that point towards functional candidacy of genes in the region, we excluded any references to GWAS or other genetic association studies. Results are summarized in the **Supplementary Note - Table 12**.

2.1.7. Copy Number Variant (CNV) analyses

We previously typed 1,350 CNVs in the HapMap analysis panels, of which 360 were common (MAF > 5%) in individuals of European ancestry (HapMap CEU), and explained more than 80% of the copy-number differences between any two individuals. Of these common CNVs, 323 appeared to be biallelic and 258 of these were in strong LD with HapMap SNPs that are close to, but do not overlap, with the CNVs⁶. We first identified the SNPs in our meta-analysis that best captured each of these 258 common biallelic CNVs based on the LD reported in the HapMap CEU population. Next, we evaluated the significance of their association with BMI in our meta-analysis.

2.1.8. eQTL analyses

We examined the *cis*-associations (defined as genes within 1 Mb) between each of the 32 confirmed BMI SNPs and expression of nearby genes in adipose tissue^{7,8}, whole blood⁷, lymphocytes^{9,8}, and brain (cortical tissue)¹⁰.

Adipose tissue and whole blood - The eQTL analyses using in adipose tissue and in whole blood have been described in detail previously (GEO database: GSE7965 and GPL3991)^{7,11,12}. In brief, 603 individuals from Iceland with adipose tissue samples and 745 with blood samples were genotyped with the Illumina 317K or 370K chip. RNA samples were hybridized to a single custom-made human array containing 23,720 unique oligonucleotide probes. *Cis*-association was tested between SNPs and the mean logarithm (\log_{10}) expression ratio (MLR) adjusted for age, sex, and age x sex, as well as differential cell count in the blood analyses, assuming an additive genetic model and accounting for familial relatedness. Only *cis*-associations with a *P*-value < 1×10^{-5} corresponding to 5% false discovery rate (FDR) are presented.

Lymphocytes - The eQTL analyses in lymphocytes have been described in detail previously (GEO database: GSE8052)⁹. In brief, lymphoblastoid cell lines, derived from peripheral blood lymphocytes were available for 830 parents and offspring of 206 families of white European descent. RNA sample were hybridized to U133 Plus 2.0 GeneChips (Affymetrix) to assess the expression of 54,675 transcripts, representing 20,599 genes. All participants were genotyped using the Illumina Human1M Beadchip and Illumina HumanHap300K Beadchip and genotypes were imputed using the data from the Phase II HapMap CEU population. Before analyses, expression data was normalized together to remove any technical or spurious background variation and subsequently an inverse normal transformation was applied to avoid any outliers. SNPs were tested for *cis* associations, assuming an additive genetic model, adjusting for non-genetic effects in the gene expression value. A *P*-value threshold of < 6.8×10^{-6} , which corresponds to a 1% FDR determined by permutation testing in this dataset, was used for the *cis*-associations presented.

Liver and subcutaneous and omental fat tissue - The eQTL analyses in liver, subcutaneous and omental fat tissue have been described in detail previously⁸. In brief, liver (n=567), subcutaneous (n=610) and omental (n=742) fat tissue were obtained from patients who underwent bariatric surgery. RNA was extracted and hybridized to a custom Agilent 44,000 feature microarray composed of 39,280 oligonucleotide probes targeting transcripts representing 34,266 known and predicted genes. All patients were genotyped on the Illumina 650Y SNP genotyping arrays. Gene expression data was adjusted for age, race, gender, and surgery year using linear regression. *Cis*-associations between each SNP and the adjusted gene expression data were tested, and a nominal *P*-value cut-off of 6×10^{-6} corresponding to a 5% FDR was used.

Brain tissue - The eQTL analyses in cortical tissue have been described in detail previously (GEO database: GSE8919)¹⁰. In brief, DNA and RNA of neuropathologically normal cortical brain samples of 193 individuals (mean age 81 years, range 65-100 years) of European descent were isolated. DNA was genotyped using the Affymetrix Gene-Chip Human Mapping 500K Array Set and genotypes were imputed using the data from the Phase II HapMap CEU population. RNA expression was assessed with the Illumina Human Refseq-8 Expression BeadChip system. *Cis*-association analyses assumed an additive model and were adjusted for sex and age at death. Only *cis*-associations with a *P*-value $< 3.5 \times 10^{-5}$ are presented.

Cis-association analyses – SNPs were tested for *cis*-associations with transcripts within a 1Mb region, assuming an additive effect of the BMI allele or using an ANOVA test. Adjustments were specific for each of the tissue datasets. Only *cis*-associations with a $P < 1 \times 10^{-5}$ for blood and adipose tissue, $P < 6.8 \times 10^{-5}$ for lymphocytes, $P < 6.0 \times 10^{-6}$ for liver, omental, and subcutaneous fat, and $P < 3.5 \times 10^{-5}$ for brain tissue are presented in the table. These *P*-value thresholds correspond to a false discovery rate of 1% for the lymphocytes and 5% for the remaining tissues. Conditional analyses were performed by conditioning the BMI-associated SNP on the most significant *cis*-associated SNP for that particular gene transcript and vice versa. Conditional analyses were performed for all expression data, except for the cortical tissue sample because of the small sample size and thus low statistical power.

2.1.9. Explained variance and additional number of loci to be discovered

The variation in BMI explained by the 32 confirmed SNPs was estimated from stage 2 analyses using $2f(1-f)a^2$, where *f* is the frequency of the variant and *a* is its additive effect¹⁰. We subsequently estimated how much more of the variance can be explained if more SNPs of the current GWA meta-analyses are considered and how many more loci would have been uncovered if the stage 1 meta-analysis had been larger.

Estimation of variance explained and polygene analysis

We evaluated the amount of phenotypic variance explained by the 32 BMI loci in an Australian and two Dutch independent validation sets (the Queensland Institute of Medical Research (QIMR) study and The Rotterdam Study (RS) II & III) using a method proposed by the International Schizophrenia Consortium¹³. To avoid the influence of potential cryptic relatedness between discovery and validation sets, we performed analyses by excluding the four Dutch studies from the discovery when using the RSII and RSIII as validation sets. The twelve studies from the United Kingdom from the discovery were excluded for the QIMR validation study.

First, we re-ran the stage 1 meta-analysis excluding the Dutch and UK studies, respectively. A list of independent SNPs associated with BMI at various P -value thresholds (from $P < 5 \times 10^{-8}$ to $P < 0.05$) was computed (using the clumping procedure implemented in PLINK, with an LD-based threshold of $r^2 \geq 0.05$, and a physical distance of 1 Mb from the top hit).

Second, using this list of independent SNPs from the revised meta-analyses described above, we performed profile scoring for each individual of the two validation studies as implemented in PLINK, where:

$$\text{Score}_i = \sum_{j=1}^m b_j x_{ij}, \text{ where}$$

m = number of SNPs

b_j = effect of allele at locus j

x_{ij} = number of reference alleles of individual i at locus j

Third, the measure of variance explained (adjusted R^2) was estimated from a linear regression model incorporating the score as the predictor and the age-adjusted inverse normal transformed BMI residuals as outcome. We reported the average explained variance weighted by the sample size of each study, while **Fig 4.** shows the study specific explained variance for various P -values thresholds.

Prediction of number of susceptibility loci

Based on the distribution of effect sizes and minor allele frequencies observed for the established BMI loci and the power to detect those effects in the combined stage 1 and stage 2 analysis, we estimated the number of susceptibility loci that are likely to exist using a new method¹⁴. All loci that reached genome-wide significance ($P < 5 \times 10^{-8}$) assuming a GC correction and had a power of at least 1% in the combined stage 1 and stage 2 analysis were included in the prediction. The stage 2 replication data was used to estimate the effect sizes and minor allele frequencies for these loci. Power was calculated based on the combined stage 1 and stage 2 sample sizes accounting for the number of SNPs that could be identified with the particular effect size. The phenotypic variance explained was estimated by summing the product of each effect size and the

number of loci predicted with that particular effect size. A parametric bootstrap method was used to obtain an estimate of the variability of the estimated number of loci.

2.1.10. Conditional analyses

We performed a conditional genome-wide association analysis to examine whether any of the 32 confirmed BMI loci harbored additional independent signals. Therefore, each individual study repeated stage 1 GWAS analyses, but adjusted for the 32 SNPs in a regression framework. The quality control criteria used for the initial stage 1 analyses was applied here as well. For inclusion of a particular study, we required that each of the 32 conditioned SNPs was no longer associated with BMI (all $P > 0.2$) and that SNPs outside the 32 conditioned loci had similar P -values and effect sizes to the primary stage 1 analysis. Of the 46 studies that contributed to the primary stage 1 analyses, seven studies ($N = 18,445$) were excluded that appeared to have errors in their conditional analysis and four studies ($N = 5,753$) did not submit conditional analyses due to logistical constraints. Meta-analysis was performed in the same way as for the GWA studies in Stage 1. The threshold for significance was determined to be 5×10^{-6} for independent signals within 1Mb of the conditioned SNPs since we estimated we were examining approximately 1% of the genome.

2.1.11. Gene-by-gene (GxG) interaction

We examined gene-by-gene interactions among the 32 confirmed BMI loci. First, analyses were performed in each individual study using the extracted genotype imputation dosages for each of the 32 SNPs from the stage 1 meta-analysis (based on 2Mb distance pruning; $P < 5 \times 10^{-6}$). The alleles were coded such that the BMI-increasing allele was always the dosage increasing allele (*i.e.* the BMI-increasing allele was coded as allele 2). Dosages of the pairwise interaction ($Y = b_0 + b_1.A + b_2.B + b_3.AB + e$; Test of $b_3 = 0$) analyses were then regressed against residuals of inverse normal transformed BMI, adjusted for age, age² and appropriate covariates (e.g. principal components) to identically match the residuals used in the main effect analyses. Analyses were stratified by sex and case-status (for samples ascertained for other diseases). Each study excluded SNPs from where the minor allele count ($MAC = 2 * N * MAF$) < 20 and/or imputation quality was poor (see stage 1 analyses). We meta-analysed the study specific pairwise interaction terms using METAL.

2.1.12. Dominant and recessive analyses

Dominant and recessive analyses were performed for the 32 confirmed BMI SNPs by each individual study. To test for non-additive effects (*i.e.* dominant or recessive), we performed analyses for four models; *i.e.* an additive, a dominant, a recessive and a dominant deviation model. Deviation from additivity was inferred by testing for evidence of a dominance deviation model.

We used “best guess” genotypes based on genotype dosage, which were regressed against inverse normal transformed BMI, adjusted for age and appropriate covariates (e.g. principal components). Each study excluded SNPs with a minor allele count (MAC = 2*N*MAF) of < 20 and/or imputation quality was poor (see stage 1 analyses). The individual study results were meta-analysed using METAL for the additive, dominant, recessive, dominant deviation models. As deviation from additivity is harder to detect when imputation quality is relatively low we also re-ran the meta-analyses excluding SNPs with a minor allele count of < 60 and/or imputation quality < 0.9, but the results were essentially the same. As an additional quality check of the “best guess” genotypes, we repeated the additive model using additive dosage and compared the additive dosage and additive best guess results from this meta-analysis to that from obtained from the primary stage 1 meta-analysis, and the correlations between $-\log_{10} P$ -values of both analyses were very high ($r > 0.99$).

2.1.13. Sex-specific analyses

To examine whether the associations of the 32 confirmed BMI SNPs were different in men and women, we performed the meta-analyses of stage 1 and stage 2 data as described before, stratified by sex ($n_{\text{women}}=140,490$ and $n_{\text{men}}=107,795$). Subsequently, we tested for each SNP whether the beta of men only (b_m) was different from the beta of women only (b_w) using the T statistic

$$(b_m - b_w) / (se_m^2 + se_w^2 - 2 * \text{corr}(b_m, b_w) * se_m * se_w)$$

with se_m and se_w being the standard errors of b_m or b_w from which we estimated the P -value using the normal approximation.

2.1.14. References

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Supplementary Note

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2.2. SUPPLEMENTARY NOTE - TABLES

- Supplementary Note - Table 1** Study design, number of individuals and sample quality control for genome-wide association studies of stage 1. **p. 40**
- Supplementary Note - Table 2** Information on genotyping methods, quality control of SNPs, imputation, and statistical analysis for stage 1 genome-wide association studies. **P. 49**
- Supplementary Note - Table 3** Study-specific descriptive statistics genome-wide association studies of stage 1. **p. 54**
- Supplementary Note - Table 4** Study design, number of individuals and sample quality control for genome-wide association studies of stage 2 - *in silico* and *de novo* replication. **P. 59**
- Supplementary Note - Table 5** Information on genotyping methods, quality control of SNPs, imputation, and statistical analysis for stage 2 *in silico* and *de novo* replication studies. **P. 65**
- Supplementary Note - Table 6** Study-specific descriptive statistics for stage 2 *in silico* and *de novo* replication studies. **P. 68**
- Supplementary Note - Table 7** Study design, number of individuals and sample quality control for studies used in extreme obesity case-control analyses, childhood BMI analyses, family-based analyses and polygenes analyses. **P. 71**
- Supplementary Note - Table 8** Supplementary Note - Table 8 -- Information on genotyping methods, quality control of SNPs, imputation, and statistical analysis for studies used in extreme obesity case-control analyses, childhood BMI analyses, family-based analyses and polygenes analyses. **P. 74**
- Supplementary Note - Table 9** Study-specific descriptive statistics for studies used in extreme obesity case-control analyses, childhood BMI analyses, family-based analyses and polygenes analyses. **P. 76**
- Supplementary Note - Table 10** Nominally significant interactions ($P_{\text{interaction}} < 0.05$) for all pairwise tests between the lead SNPs at the 32 confirmed BMI loci. **p. 78**
- Supplementary Note - Table 11** Dominant, recessive and dominance deviation results for nominally significant (dominance deviation $P < 0.05$) lead SNPs at the 32 confirmed BMI loci. The effect allele is the BMI-increasing allele. Only SNPs with a dominance deviation $P < 0.05$ are

Supplementary Note

presented. The results are not significant at $P < 0.05$ after correcting for the number of tests performed. **P. 80**

Supplementary Note - Table 12 PubMed and OMIM hits for genes within 500kb of 32 BMI-associated SNPs, using terms "obesity" and "BMI". **P. 82**

Supplementary Note - Table 13 Assessment of population-stratification by comparison of effect sizes and directions of association between family-based data from Framingham and overall meta-analysis using the QFAM -- within procedure from PLINK. **P. 93**

Supplementary Note

Supplementary Note - Table 1 Study design, number of individuals and sample quality control for genome-wide association studies of stage 1.

Supplementary Note - Table 1

Supplementary Note - Table 1 -- Study design, number of individuals and sample quality control for genome-wide association studies of stage 1.

Study		Study design	Total sample size (N)	Call rate*	Sample QC Study specific QC criteria	Samples in analyses (N)**	Anthropometric assessment method	References
Short name	Full name							
ADVANCE	Atherosclerotic Disease, Vascular Function, and Genetic Epidemiology	Population-based case-control	599	>98.5%	1) duplicates 2) missing weight or height	586 (275 CAD cases, 311 controls)	measured	[PMID: 18443000] Assimes TL et al. Susceptibility locus for clinical and subclinical coronary artery disease at chromosome 9p21 in the multi-ethnic ADVANCE study. Hum Mol Genet. 2008 Aug 1;17(15):2320-8.
AGES	Age, Gene/Environment Susceptibility-Reykjavik Study	Population-based	3219	≥ 97%	1) mismatch with previous genotypes 2) remove A/T & G/C SNPs 3) remove SNPs not in HapMap	3207	measured	[PMID: 17351290] Harris et al. Age, Gene/Environment Susceptibility-Reykjavik Study: multidisciplinary applied phenomics. American Journal of Epidemiology (2007) vol. 165 (9) pp. 1076-87
Amish HAPI Heart Study	Amish Heredity and Phenotype Intervention Heart Study	Founder population	915	≥ 93%	1) Misidentified pedigree relationships 2) Misidentified sex	905	measured	[PMCID: PMC2443415] Mitchell BD, McArdle PF, Shen H, et al. The genetic response to short-term interventions affecting cardiovascular function: Rationale and design of the Heredity and Phenotype Intervention (HAPI) Heart Study. Am Heart J 823:828, 2008 [PMID: 18779467] Rampersaud E, Mitchell BD, Pollin TI, et al. Physical Activity and the Association of common FTO Gene Variants with Body Mass Index and
ARIC	Atherosclerosis Risk in Communities Study	Population-based	15792	≥ 90%	1) True sex/gender mismatch 2) Discordant genotype with earlier TaqMan genotyping. If >10/47 genotypes discordant -> exclude 3) First-degree relative 4) PC>8SD in Eigenstrat run (10 iterations with 10 PCs) 5) Outlier based on average IBS 6) missing	8108	measured	[PMID: 2646917] Atherosclerosis Risk in Communities (ARIC) Study: design and objectives. 1989. ARIC Investigators. Am. J. Epidemiol. 129: 687-702. [PMID: 19557197] Heard-Costa, NL et al. NRXN3 is a novel locus for waist circumference: a genome-wide association study from the CHARGE Consortium. 2009. Plos Genet. 5(6): e1000539.
B58C (T1DGC)	British 1958 birth cohort (Type 1 Diabetes Genetic Consortium controls)	Population-based	2,592	≥ 98%	1) contamination 2) non-European identity 3) Missing body weight and height.	2,587	measured	[PMID: 17255346] Strachan DP, et al. Lifecourse influences on health among British adults: effects of region of residence in childhood and adulthood. Int J Epidemiol 2007;36:522-531. [PMID: 19430480] Barrett JC, The Type 1 Diabetes Genetics Consortium, et al. Genome-wide association study and meta-analysis find that over 40 loci affect risk of type 1 diabetes. Nat Genet 2009 May 10. [Epub ahead of print]
B58C (WTCCC)	British 1958 birth cohort (Wellcome Trust Case Control Consortium controls)	Population-based	1,502	≥ 97%	1) contamination 2) non-European identity 3) Missing body weight and height.	1,479	measured	[PMID: 17554300] The Wellcome Trust Case Control Consortium Genome-wide association study of 14,000 cases of seven common diseases and 3,000 shared controls. Nature 2007;447: 661-678. [PMID: 16155052] Power,C. & Elliott,J. Cohort profile: 1958 British birth cohort (National Child Development Study). Int J Epidemiol 2006;35:34-41.
BRIGHT (WTCCC-HT) Cases	The British Genetics of Hypertension (BRIGHT) Study	Hypertension case series	2,000	≥ 97%	1) heterozygosity <23% or >30% 2) external discordance 3) non-European ancestry 4) duplicate/first/second degree relatives.	1,895	measured	[PMID: 17554300] The Wellcome Trust Case Control Consortium Genome-wide association study of 14,000 cases of seven common diseases and 3,000 shared controls. Nature 447, 661-678 (2007). [PMID: 12826435] Caulfield,M. et al. Genome-wide mapping of human loci for essential hypertension. Lancet 361, 2118-2123 (2003).

Supplementary Note - Table 1

Study			Total sample size (N)	Call rate*	Sample QC	Samples in analyses (N)**	Anthropometric assessment method	References
Short name	Full name	Study design			Study specific QC criteria			
CAD_WTCCC	WTCCC Coronary Heart Disease cases	case series	2000	≥ 97%	1) heterozygosity <23% or >30%; 2) discrepancy with external identifying information; 3) ethnic outliers; 4) related individuals and duplicates;	1876	self reported	[PMID: 17554300] The Wellcome Trust Case Control Consortium Genome-wide association study of 14,000 cases of seven common diseases and 3,000 shared controls. Nature 447, 661-678 (2007)
CAPS1 Cases	Cancer Prostate in Sweden 1	Case-control study of prostate cancer	505	> 95%	1) related individuals and duplicates; 2) ethnic outliers; 3) missing body weight and height.	505	self-reported	[PMID: 18073375] Duggan D, et al. Two genome-wide association studies of aggressive prostate cancer implicate putative prostate tumor suppressor gene DAB2IP. J Natl Cancer Inst 2007 Dec 19;99(24):1836-44 (2007).
CAPS1 Controls	Cancer Prostate in Sweden 1	Case-control study of prostate cancer	506	> 95%	1) related individuals and duplicates; 2) ethnic outliers; 3) missing body weight and height.	506	self-reported	[PMID: 18073375] Duggan D, et al. Two genome-wide association studies of aggressive prostate cancer implicate putative prostate tumor suppressor gene DAB2IP. J Natl Cancer Inst 2007 Dec 19;99(24):1836-44 (2007).
CAPS2 Cases	Cancer Prostate in Sweden 2	Case-control study of prostate cancer	1,483	> 95%	1) related individuals and duplicates; 2) ethnic outliers; 3) missing body weight and height.	1,483	self-reported	[PMID: 18073375] Duggan D, et al. Two genome-wide association studies of aggressive prostate cancer implicate putative prostate tumor suppressor gene DAB2IP. J Natl Cancer Inst 2007 Dec 19;99(24):1836-44 (2007).
CAPS2 Controls	Cancer Prostate in Sweden 2	Case-control study of prostate cancer	519	> 95%	1) related individuals and duplicates; 2) ethnic outliers; 3) missing body weight and height.	519	self-reported	[PMID: 18073375] Duggan D, et al. Two genome-wide association studies of aggressive prostate cancer implicate putative prostate tumor suppressor gene DAB2IP. J Natl Cancer Inst 2007 Dec 19;99(24):1836-44 (2007).
CHS	Cardiovascular Health Study	Population-based cohort study	3228	>95%	1) Prevalent clinical CVD 2) African-americans 3) Sex discordant 4) Missing body weight and height	3228	measured	[PMID: 1669507] Fried LP, Borhani NO, Enright P, Furberg CD, Gardin JM, Kronmal RA, Kuller LH, Manolio TA, Mittelmark MB, Newman A. The Cardiovascular Health Study: design and rationale. Ann Epidemiol. 1991; 1: 263-276.
CoLaus	Cohorte Lausannoise	Population-based	6188	>90%	1) ethnic outliers; 2) related individuals and duplicates; 3) Missing body weight and height	5408	measured	[PMID 18366642] Firmann et al. The CoLaus study: a population-based study to investigate the epidemiology and genetic determinants of cardiovascular risk factors and metabolic syndrome BMC Cardiovascular Disorders 2008, 8:6
deCODE	deCODE genetics sample set	Population-based	38,446	≥ 96%	Missing body weight and height.	26,799	measured	[PMID: 19079260] Thorleifsson, G. et al. Genome-wide association yields new sequence variants at seven loci that associate with measures of obesity. Nat Genet. 41, 18-24 (2009).
DGI (cases)	Diabetes Genetics Initiative of Broad Institute of Harvard and MIT, Lund University, and Novartis Institutes of BioMedical Research	Case control for diabetes	1658	≥ 95%	1) Related individuals 2. Sample contamination 3) Missing body weight and height.	1317	measured	[PMID: 17463246] Saxena et al. Genome-wide association analysis identifies loci for type 2 diabetes and triglyceride levels. Science 316, 1331-6 (2007).

Supplementary Note - Table 1

Study		Study design	Total sample size (N)	Call rate*	Sample QC	Samples in analyses (N)**	Anthropometric assessment method	References
Short name	Full name				Study specific QC criteria			
DGI (controls)	Diabetes Genetics Initiative of Broad Institute of Harvard and MIT, Lund University, and Novartis Institutes of BioMedical Research	Case control for diabetes	1595	≥ 95%	1) Related individuals 2) Sample contamination 3) Missing body weight and height.	1088	measured	[PMID: 17463246] Saxena et al. Genome-wide association analysis identifies loci for type 2 diabetes and triglyceride levels. <i>Science</i> 316, 1331-6 (2007).
EGCUT	Estonian Genome Center, University of Tartu	Population-based	1412	≥ 95%	1) Related individuals and duplicates 2) Sex mismatch 3) Phenotype missing	1412	measured	[PMID: 19424496] Nelis M, Esko T, Magi R, Zimprich F, Zimprich A, et al. (2009) Genetic Structure of Europeans: A View from the North–East. <i>PLoS ONE</i> 4(5): e5472. [PMID: 15133739] Metspalu, A. The Estonian Genome Project. <i>Drug Development Research</i> 62, 97-101 (2004).
EPIC-Obesity Study	European Prospective Investigation into Cancer and Nutrition - Obesity Study	Population-based	2,566	≥ 94%	1) heterozygosity <23% or >30%; 2) >5.0% discordance in SNP pairs with r ² = 1 in HapMap; 3) ethnic outliers; 4) related individuals and duplicates; 5) Missing body weight and height.	2,415	measured	[PMID: 10466767] Day, N.E. et al. EPIC-Norfolk: study design and characteristics of the cohort. <i>European Prospective Investigation of Cancer. British Journal of Cancer</i> 80, 95-103 (1999). [PMID: 18454148] Loos, R.J. et al. Common variants near MC4R are associated with fat mass, weight and risk of obesity. <i>Nat Genet</i> 40, 768-775 (2008).
ERF	Erasmus Rucphen Family (EUROSPAN)	family based	2300	> 95%	1) excess heterozygosity based on FDR 2) ethnic outliers 3) sex mismatch 4) missing phenotype	2090	measured	[PMID: 15054401] Aulchenko YS, Heutink P, Mackay I, Bertoli-Avella AM, Pullen J, et al. (2004) Linkage disequilibrium in young genetically isolated Dutch population. <i>Eur J Hum Genet</i> 12: 527-534 [PMID: 20049090] Pecioska S, Zillikens MC, Henneman P, Snijders PJ, Oostra BA, van Duijn CM, Aulchenko YS. Association between type 2 diabetes loci and measures of fatness. <i>PLoS One</i> 5, e8541 (2010).
Fenland	Fenland Study	Population-based	1,500	≥ 95%	1) heterozygosity <27.3% or >28.8%; 2) duplicate check; 3) relatedness check	1402	measured	[PMID: 19079261] Willer CJ, Speliotes EK, Loos RJ et al. (2009) Six new loci associated with body mass index highlight a neuronal influence on body weight regulation. <i>Nat Genet</i> , 41(1): 25-34.
FHS cases	Family Heart Study	Case-Controls	463	≥ 98%	1) technical errors 2) discrepancies between reported sex and sex-diagnostic markers	441	measured	[PMID: 8651220] M. Higgins et al., NHLBI Family Heart Study: objectives and design, <i>Am J Epidemiol</i> 143, 1219–1228 (1996).
FHS controls	Family Heart Study	Case-Controls	434	≥ 98%	1) technical errors 2) discrepancies between reported sex and sex-diagnostic markers	416	measured	[PMID: 8651220] M. Higgins et al., NHLBI Family Heart Study: objectives and design, <i>Am J Epidemiol</i> 143, 1219–1228 (1996).
FRAM	Framingham Heart Study	Population-based, multi-generational	9274	≥ 97%	1) pHWE<1e-6 call rate<97% 2) mishap p<1e-9 3) MAF<0.01 4) Mendelian errors>100 5) SNPs not in Hapmap or strandedness issues merging with Hapmap	8094	measured	[PMID 14025561] DAWBER TR, KANNEL WB, LYELL LP. An approach to longitudinal studies in a community: the Framingham Study. <i>Ann N Y Acad Sci.</i> 1963;107:539-556. [PMID 1208363] Feinleib M, KANNEL WB, Garrison RJ, McNamara PM, Castelli WP. The Framingham Offspring Study. Design and preliminary data. <i>Prev Med.</i> 1975;4:518-525. [PMID 17372189] Splansky GL, Corey D, Yang Q et al. The Third Generation Cohort of the National Heart, Lung, and Blood Institute's Framingham Heart Study: design, recruitment, and initial examination. <i>Am J Epidemiol.</i> 2007;165:1328-1335.

Supplementary Note - Table 1

Study		Study design	Total sample size (N)	Call rate*	Sample QC Study specific QC criteria	Samples in analyses (N)**	Anthropometric assessment method	References
Short name	Full name							
FTC	Finnish Twin Cohort	Monozygotic twins	152 pairs	≥ 95%	1) ethnic outliers; 2) related individuals and duplicates; 3) Missing body weight and height for both individuals of the pair.	138 pairs	measured	[PMID: 19060911] Aulchenko YS, Ripatti S, Lindqvist I et al. Loci influencing lipid levels and coronary heart disease risk in 16 European population cohorts, Nat Genet (2009) 41(1): 47-55.
FUSION cases	Finland-United States Investigation of NIDDM Genetics	Case-control	1161	> 97.5%	related individuals; missing BMI	857	measured	[PMID: 17463248] Scott,LJ et al. A genome-wide association study of type 2 diabetes in Finns detects multiple susceptibility variants. Science 316(5829):1341-5 (2007).
FUSION controls	Finland-United States Investigation of NIDDM Genetics	Case-control	1174	> 97.5%	related individuals; missing BMI	869	measured	[PMID: 17463248] Scott,LJ et al. A genome-wide association study of type 2 diabetes in Finns detects multiple susceptibility variants. Science 316(5829):1341-5 (2007).
GENMETS cases	Health 2000 / GENMETS substudy	Case-control study of Metabolic syndrome	932	≥ 95%	1) ethnic outliers; 2) related individuals and duplicates; 3) Missing body mass index	1092	measured	http://www.terveys2000.fi/indexe.html
GENMETS controls	Health 2000 / GENMETS substudy	Case-control study of Metabolic syndrome	948	≥ 95%	1) ethnic outliers; 2) related individuals and duplicates; 3) Missing body mass index	1171	measured	http://www.terveys2000.fi/indexe.html
GerMiFSI	German Myocard Infarct Family Study I	Case - Control (only Cases)	875	> 97%	1) related individuals and duplicates; 2)missin phenotypes 3) heterozygosity mean +- 3*sd outlier	600	measured	[PMID: 17634449] Samani NJ, Erdmann J, Hall AS et al. Genomewide association study of coronary artery disease. NEJM 2007; 357(5): 436-9.
GerMiFSII	German Myocard Infarct Family Study II	Case – Control (only Cases)	1222	> 97%	1) related individuals and duplicates; 2)missing phenotypes 3) heterozygosity mean +-3*sd outlier	1124	measured	[PMID: 19198612] Erdmann J, Grosshennig A, Braund PS et al. New susceptibility locus for coronary artery disease on chromosome 3q22.3. Nat Genet 2009; 41(3): 280-2.
KORA3	Cooperative Health Research in the Region of Augsburg, KOoperative Gesundheitsforschung in der Region Augsburg	Population-based	1644	>=93%	1) All with German passport	1644	measured	[PMID 20031538] Heid IM, Boes E, Müller M, Kollerits B, et al. Genome-wide association analysis of high-density lipoprotein cholesterol in the population-based KORA study sheds new light on intergenic regions. Circ Cardiovasc Genet, 2008; 1(10): 10-20.
KORA4	Cooperative Health Research in the Region of Augsburg, KOoperative Gesundheitsforschung in der Region Augsburg	Population-based	1814	>=93%	1) All with German passport	1814	measured	[PMID 16032514] Wichmann HE, Gieger C, Illig T; MONICA/KORA Study Group. KORA-gen--resource for population genetics, controls and a broad spectrum of disease phenotypes. Gesundheitswesens 2005; 67 Suppl 1: S26-30.

Supplementary Note - Table 1

Study		Study design	Total sample size (N)	Call rate*	Sample QC	Samples in analyses (N)**	Anthropometric assessment method	References
Short name	Full name				Study specific QC criteria			
MICROS	MICROS (EUROSPAN)	Population-based	1098	≥ 97%	1) ethnic outliers; 2) duplicates; 3) Missing body weight and height.	1079	measured	[PMID: 17550581] Pattaro C, Marroni F, Riegler A, et al. The genetic study of three population microisolates in South Tyrol (MICROS): study design and epidemiological perspectives. BMC Med Genet. 2007 Jun 5;8-29. [PMID: 18952825] Johansson A. et al. Common variants in the JAZF1 gene associated with height identified by linkage and genome-wide association analysis. Hum Mol Genet. 18(2), 373-80 (2009). [PMID: 19798445] Hicks, AA. et al. Genetic determinants of circulating sphingolipid concentrations in European populations. PLoS Genet. 5(10):e1000672 (2009)
Migen (cases)	Myocardial Infarction Genetics Consortium	Case control for early onset MI	1420	≥ 95%	1) Cohort deficient or excess heterozygosity 2) Population Stratification outliers 3) Close Relatives and Sample Duplicates 4) Sample Contamination (excess close low-level genome sharing) 5) missing body weight or height	1274	measured	[PMID: 19198609] Kathiresan et al. Genome-wide association of early-onset myocardial infarction with single nucleotide polymorphisms and copy number variants. Nature Genetics 41, 334-41 (2009).
Migen (controls)	Myocardial Infarction Genetics Consortium	Case control for early onset MI	1558	≥ 95%	1) Cohort deficient or excess heterozygosity 2) Population Stratification outliers 3) Close Relatives and Sample Duplicates 4) Sample Contamination (excess close low-level genome sharing) 5) Missing body weight or height	1407	measured	[PMID: 19198609] Kathiresan et al. Genome-wide association of early-onset myocardial infarction with single nucleotide polymorphisms and copy number variants. Nature Genetics 41, 334-41 (2009).
NBS_WTCCC	WTCCC National Blood Service donors	Population-based	1500	≥ 97%	1) heterozygosity <23% or >30%; 2) discrepancy with external identifying information; 3) ethnic outliers; 4) related individuals and duplicates;	1437	self reported	[PMID: 17554300] The Wellcome Trust Case Control Consortium Genome-wide association study of 14,000 cases of seven common diseases and 3,000 shared controls. Nature 447, 661-678 (2007)
NFBC-1966	Northern Finland Birth Cohorts 1966	Population-based	5654	≥ 95%	1) gender discrepancy with genetic data from X-linked markers; 2) withdrawn consent; 3) duplicates and first and second degree relatives; 4) contaminated samples	4773	measured	[PMID: 19060910] Sabatti, C. et al. Genome-wide association analysis of metabolic traits in a birth cohort from a founder population. Nat Genet (2008).

Supplementary Note - Table 1

Study		Study design	Total sample size (N)	Call rate*	Sample QC	Samples in analyses (N)**	Anthropometric assessment method	References
Short name	Full name				Study specific QC criteria			
NHS	The Nurses' Health Study	Nested case-control	2368	>90%	1) Low genotyping completion (<90%) 2) Unclear identity and admixed origin 3) Missing body weight and height;	2265	self-reported	[PMID: 17529973] Hunter D, et al. A genome-wide association study identifies alleles in FGFR2 associated with risk of sporadic postmenopausal breast cancer. Nat Genet. 2007 Jul;39(7):870-4.
NSPHS	Northern Sweden Population Health Study (EUROSPAN)	Population-based	720	≥ 97%	1) ethnic outliers; 2) duplicates; 3) Missing body weight and height.	647	measured	[PMID: 18952825] Johansson A. et al. Common variants in the JAZF1 gene associated with height identified by linkage and genome-wide association analysis. Hum Mol Genet. 18(2), 373-80 (2009). [PMID: 19798445] Hicks, AA. et al. Genetic determinants of circulating sphingolipid concentrations in European populations. PLoS Genet. 5(10):e1000672 (2009)
NTRNESDA cases	Netherlands Twin Register & the Netherlands Study of Depression and Anxiety	case-control	1860	≥ 95%	1) evidence of sample contamination (heterozygosity); 2) ethnic outliers; 3) related individuals and duplicates; 5) missing body weight and height.	1727	measured	Boomsma DI, de Geus EJ, Vink JM, Stubbe JH, Distel MA, Hottenga JJ et al. Netherlands Twin Register: from twins to twin families. Twin Res Hum Genet 2006; 9: 849–857. [PMID: 18763692] Penninx B, Beekman A, Smit J. The Netherlands Study of Depression and Anxiety (NESDA): rationales, objectives and methods. Int J Methods Psychiatr Res 2008; 17: 121–140. [PMID: 18197199] Boomsma DI, Willemsen G, Sullivan PF, Heutnik P, Meijer P, Sondervan D et al. Genome-wide association of major depression: Description of samples for the GAIN major depressive disorder study: NTR and NESDA Biobank Projects. Eur J Hum Genet 2008; 16: 335–342.
NTRNESDA controls	Netherlands Twin Register & the Netherlands Study of Depression and Anxiety	case-control	1860	≥ 95%	1) evidence of sample contamination (heterozygosity); 2) ethnic outliers; 3) related individuals and duplicates; 5) missing body weight and height.	1789	measured	Boomsma DI, de Geus EJ, Vink JM, Stubbe JH, Distel MA, Hottenga JJ et al. Netherlands Twin Register: from twins to twin families. Twin Res Hum Genet 2006; 9: 849–857. [PMID: 18763692] Penninx B, Beekman A, Smit J. The Netherlands Study of Depression and Anxiety (NESDA): rationales, objectives and methods. Int J Methods Psychiatr Res 2008; 17: 121–140. [PMID: 18197199] Boomsma DI, Willemsen G, Sullivan PF, Heutnik P, Meijer P, Sondervan D et al. Genome-wide association of major depression: Description of samples for the GAIN major depressive disorder study: NTR and NESDA Biobank Projects. Eur J Hum Genet 2008; 16: 335–342.
ORCADES	Orkney Complex Disease Study (EUROSPAN)	Population-based	719	≥ 97%	1) ethnic outliers; 2) duplicates; 3) Missing body weight and height.	697	measured	[PMID: 18952825] Johansson A. et al. Common variants in the JAZF1 gene associated with height identified by linkage and genome-wide association analysis. Hum Mol Genet. 18(2), 373-80 (2009). [PMID: 19798445] Hicks, AA. et al. Genetic determinants of circulating sphingolipid concentrations in European populations. PLoS Genet. 5(10):e1000672 (2009)
PLCO	Prostate, Lung, Colorectal, and Ovarian Cancer Screening Trial	Population-based	2,298	≥ 94%	1) Gender discordance 2) Non-European ancestry 3) Related individuals and duplicates; 4) Missing height.	2238	self-report	[PMID: 17401363] Yeager M, et al. Genome-wide association study of prostate cancer identifies a second risk locus at 8q24. Nat Genet 39, 645-9 (2007).

Supplementary Note - Table 1

Study			Total sample size (N)	Call rate*	Sample QC	Samples in analyses (N)**	Anthropometric assessment method	References
Short name	Full name	Study design			Study specific QC criteria			
Procardis	Precocious Coronary Artery Disease (ProCARDIS)	case-control	2573	> 95%	none	3,212	measured	Broadbent, H.M. et al. Susceptibility to coronary artery disease and diabetes is encoded by distinct, tightly linked SNPs in the ANRIL locus on chromosome 9p. Hum Mol Genet 17, 806-14 (2008).
RS-I	Rotterdam Study I	Population-based	7,983	≥ 97.5%	1) missing DNA; 2) gender mismatch with typed X-linked markers; 3) excess autosomal heterozygosity > 0.336~FDR>0.1%; 4) duplicates and/or 1st or 2nd degree relatives using IBS probabilities >97% from PLINK; 5) ethnic outliers using IBS distances > 3SD f	5,744	measured	[PMID: 11753597] Visscher, 2001 A comparison of body mass index, waist-hip ratio and waist circumference as predictors of all-cause mortality among the elderly: the Rotterdam study [PMID:19700477] Estrada, 2009 GRIMP: a web- and grid-based tool for high-speed analysis of large-scale genome-wide association using imputed data [PMID:19728115] Hofman, 2009 The Rotterdam Study: 2010 objectives and design update [PMID:1833235] Hofman, 1991 Determinants of disease and disability in the elderly: the Rotterdam Elderly Study
RUNMC	Nijmegen Bladder Cancer Study and Nijmegen Biomedical Study	Case-study and Population-based study	3,081	≥ 96%	Missing body weight and height.	2,873	measured	[PMID: 17568781] Wetzels, J.F. et al. Age- and gender-specific reference values of estimated GFR in Caucasians: the Nijmegen Biomedical Study. Kidney Int 72, 632-637 (2007). [PMID: 18794855] Kiemeny, L.A. et al. Sequence variant on 8q24 confers susceptibility to urinary bladder cancer. Nat Genet 40, 1307-1312 (2008).
SardiNIA	SardiNIA	Population-based	6,148	≥ 95%	1) Missing BMI	4,301	measured	[PMID: 16934002] Pilia, G. et al. Heritability of cardiovascular and personality traits in 6,148 Sardinians. PLoS Genet 2, e132 (2006). [PMID: 17658951] Scuteri et al Genome-wide association scan shows genetic variants in the FTO gene are associated with obesity-related traits. PLoS Genet. 2007 Jul;3(7):e115.
SASBAC Cases	Swedish And Singapore Breast Association Consortium	Case-control study of breast cancer	803	≥ 96%	1) related individuals and duplicates; 2) ethnic outliers; 3) missing body weight and height.	795	self-reported	[PMID: 10209946] Magnusson, C. et al. Breast-cancer risk following long-term oestrogen- and oestrogen-progestin-replacement therapy. Int J Cancer 81, 339-44 (1999). [PMID: 17132159] Einarsdóttir, K. et al. Comprehensive analysis of the ATM, CHEK2 and ERBB2 genes in relation to breast tumour characteristics and survival: a population-based case-control and follow-up study. Breast Cancer Res 8, R67 (2006).
SASBAC Controls	Swedish And Singapore Breast Association Consortium	Case-control study of breast cancer	764	≥ 96%	1) related individuals and duplicates; 2) ethnic outliers; 3) missing body weight and height.	764	self-reported	[PMID: 10209946] Magnusson, C. et al. Breast-cancer risk following long-term oestrogen- and oestrogen-progestin-replacement therapy. Int J Cancer 81, 339-44 (1999). [PMID: 17132159] Einarsdóttir, K. et al. Comprehensive analysis of the ATM, CHEK2 and ERBB2 genes in relation to breast tumour characteristics and survival: a population-based case-control and follow-up study. Breast Cancer Res 8, R67 (2006).
SEARCH/UKOPS	Studies of Epidemiology and Risk factors in Cancer Heredity / UK Ovarian Cancer Population Study	Population-based	1710	≥ 80%	1) ethnic outliers 2) duplicates 3) Missing body weight and height	1556	self-reported	[PMID: 19648919] H. Song, S. J. Ramus, J. Tyrer et al. A genome-wide association study identifies a new ovarian cancer susceptibility locus on 9p22.2. Nature Genetics, 41, p996-1000, (2009).
SHIP	Study of Health in Pomerania	Population-based	4310	≥ 92%	1) missing genotype or phenotype data	4092	measured	[PMID: 20167617] Völzke H, et al. Cohort Profile: The Study of Health in Pomerania. Int J Epidemiol. Online (2010).

Supplementary Note - Table 1

Study		Study design	Total sample size (N)	Call rate*	Sample QC	Samples in analyses (N)**	Anthropometric assessment method	References
Short name	Full name				Study specific QC criteria			
T2D_WTCCC	WTCCC Type 2 Diabetes cases	case series	1999	≥ 97%	1) heterozygosity <23% or >30%; 2) discrepancy with external identifying information; 3) ethnic outliers; 4) related individuals and duplicates;	1903	measured	The Wellcome Trust Case Control Consortium Genome-wide association study of 14,000 cases of seven common diseases and 3,000 shared controls. <i>Nature</i> 447, 661-678 (2007)
TwinsUK	TwinsUK	Twins pair	2226	≥ 95%	1) heterozygosity <33% or >37%; 2) ethnic outliers; 3) related individuals and duplicates; 4) Missing body weight and height.	1477	measured	[PMID: 17254428] Spector, T.D., Williams, F.M. The UK Adult Twin Registry (TwinsUK). <i>Twin Res Hum Genet</i> 9, 899-906 (2006). [PMID: 12537873] Spector, T.D., MacGregor, A.J. The St. Thomas' UK Adult Twin Registry. <i>Twin Res</i> 5, 440-443 (2002).
VIS	VIS (EUROSPAN) and KORCULA	Population-based	795	≥ 97%	1) ethnic outliers; 2) duplicates; 3) Missing body weight and height.	770	measured	[PMID: 18952825] Johansson A. et al. Common variants in the JAZF1 gene associated with height identified by linkage and genome-wide association analysis. <i>Hum Mol Genet.</i> 18(2), 373-80 (2009). [PMID: 19798445] Hicks, AA. et al. Genetic determinants of circulating sphingolipid concentrations in European populations. <i>PLoS Genet.</i> 5(10):e1000672 (2009)

*Sample genotyping success rate; i.e. percentage of successfully genotyped SNPs per sample

**Ns reported are numbers of individuals association data was provided for; Ns in paper may vary due to effective sample size used in meta-analyses

Supplementary Note

Supplementary Note - Table 2 Information on genotyping methods, quality control of SNPs, imputation, and statistical analysis for stage 1 genome-wide association studies.

Supplementary Note - Table 2

Supplementary Note - Table 2 -- Information on genotyping methods, quality control of SNPs, imputation, and statistical analysis for stage 1 genome-wide association studies.

Study	Platform	Genotyping				SNPs that met QC criteria	Imputation software	Imputation		SNPs in meta-analysis	combined	λ_{GC} men	women	Analysis software
		Genotype calling algorithm	MAF	Inclusion criteria Call rate*	p for HWE			Inclusion criteria Imputation quality*	MAF					
ADVANCE Cases	Illumina 550k	Beadstudio	none	$\geq 98.5\%$	$> 10^{-3}$	543985	BimBam	none	proper-info ≥ 0.40	2198192	--	1.041	1.046	SNPtest
ADVANCE Controls	Illumina 550k	Beadstudio	none	$\geq 98.5\%$	$> 10^{-3}$	543985	BimBam	none	proper-info ≥ 0.40	2212041	--	1.042	1.018	SNPtest
AGES	Illumina	BeadStudio	$\geq 1\%$	$\geq 95\%$	$> 10^{-6}$	308,340	MACH	none	$r2_hat \geq 0.30$	2458927	--	1.029	1.037	ProbABEL
Amish	Human370CNV Affymetrix GeneChip Human Mapping 500K	BRLMM	$\geq 1\%$	$\geq 95\%$	$> 10^{-6}$	338,598	MACH	none	$r2_hat \geq 0.30$	2291092	1.048	--	--	MMAP
ARIC	Affymetrix Genome-Wide Human SNP Array 6.0	Birdseed	$\geq 1\%$	$\geq 90\%$	$> 10^{-6}$	685,812	MACH v1.0.16	none	$r2_hat \geq 0.30$	2511301	--	1.013	1.03	ProbABEL
B58C (T1DGC)	Illumina HumanHap 550 V.1	ILLUMINUS	$> 0\%$	none	none	539,458	MACH	none	$r2_hat \geq 0.30$	2507904	--	1.007	1.01	ProbABEL
B58C (WTCCC)	Affymetrix GeneChip Human Mapping 500K	CHIAMO	$> 0\%$	none	none	490,032	IMPUTE	none	proper-info ≥ 0.40	2448867	--	1.008	0.992	SNPtest
BRIGHT	Affymetrix GeneChip Human Mapping 500K	CHIAMO	$\geq 5\%$	$\geq 95\%$	$> 10^{-6}$	387,666	IMPUTE	none	proper-info ≥ 0.40	2429493	--	1.007	1.017	SNPtest
CAD_WTCCC	Affymetrix GeneChip Human Mapping 500K	CHIAMO	$> 5\%$	$\geq 95\%$	$> 10^{-6}$	387,667	IMPUTE	none	proper-info ≥ 0.40	2430162	--	1.004	1.025	SNPTEST
CAPS1 cases	Affymetrix GeneChip Human Mapping 500K	BRLMM	$\geq 1\%$	$\geq 95\%$	$> 10^{-7}$	330,124	IMPUTE	none	proper-info ≥ 0.40	2388288	--	0.992	N/A	SNPtest
CAPS1 controls	Affymetrix GeneChip Human Mapping 500K	BRLMM	$\geq 1\%$	$\geq 95\%$	$> 10^{-7}$	330,124	IMPUTE	none	proper-info ≥ 0.40	2391197	--	1.001	N/A	SNPtest
CAPS2 cases	Affymetrix GeneChip Human Mapping 5.0K	BLRMM-P	$\geq 1\%$	$\geq 95\%$	$> 10^{-7}$	348,163	IMPUTE	none	proper-info ≥ 0.40	2416628	--	1.013	N/A	SNPtest
CAPS2 controls	Affymetrix GeneChip Human Mapping 5.0K	BLRMM-P	$\geq 1\%$	$\geq 95\%$	$> 10^{-7}$	348,163	IMPUTE	none	proper-info ≥ 0.40	2392236	--	1.013	N/A	SNPtest
CHS	Illumina 370-CNV	BeadStudio	-.**	$> 97\%$	$> 10^{-5}$	306,655	BimBam	$> 1\%$	$r2_hat \geq 0.30$	2191627	--	1.017	1.018	R

Supplementary Note - Table 2

Study	Platform	Genotyping				SNPs that met QC criteria	Imputation software	Imputation		SNPs in meta-analysis	combined	λ_{GC} men	women	Analysis software
		Genotype calling algorithm	MAF	Inclusion criteria Call rate*	p for HWE			MAF	Inclusion criteria Imputation quality*					
CoLaus	Affymetrix GeneChip Human Mapping 500K	BRLMM	$\geq 1\%$	$\geq 70\%$	$> 10^{-7}$	390631	IMPUTE	none	$r^2_{\text{hat}} \geq 0.30$	2479488	--	0.997	1.009	QUICKTEST
deCODE	Illumina HumanHap300 or HumanHapCNV370	BeadStudio	$\geq 1\%$	$\geq 96\%$	$> 10^{-6}$	290,447	IMPUTE	none	proper-info ≥ 0.40	2455981	0.993	--	--	SNPtest
DGI (cases)	Affymetrix 500K Array Set	BRLMM	$> 5\%$	$\geq 95\%$	$> 10^{-6}$	347,010	MACH	$\geq 1\%$	$r^2_{\text{hat}} \geq 0.30$	2,375,087	--	0.996	1.016	MACH2QTL
DGI (controls)	Affymetrix 500K Array Set	BRLMM	$> 5\%$	$\geq 95\%$	$> 10^{-6}$	347,010	MACH	$\geq 1\%$	$r^2_{\text{hat}} \geq 0.30$	2,375,087	--	0.993	1.007	MACH2QTL
EGCUT	Illumina Beadarray Human370CNV	Beadstudio	$\geq 1\%$	$\geq 98\%$	$> 10^{-6}$	299,484	IMPUTE	none	proper-info ≥ 0.40	2428764	--	1.01	1.009	SNPtest
EPIC-Obesity Study	Affymetrix GeneChip Human Mapping 500K	BRLMM	$\geq 1\%$	$\geq 90\%$	$> 10^{-6}$	397,438	IMPUTE	none	proper-info ≥ 0.40	2415364	--	1.009	1.008	SNPtest
ERF	Illumina 318K, 370K, Affymetrix 250K	Beadstudio, BRLMM	$> 0.5\%$	$> 95\%$	$> 10^{-6}$	NA	MACH	none	$r^2_{\text{hat}} \geq 0.30$	2463910	1.022	--	--	ProbABEL
Fenland	Affymetrix SNP5.0	BRLMM	$\geq 1\%$	$\geq 90\%$	$> 10^{-6}$	362,055	IMPUTE	none	proper-info ≥ 0.40	2473344	--	1.021	1.011	SNPtest
FHS cases	Illumina 1Million GeneChip	BeadStudio	$\geq 1\%$	$\geq 98\%$	$> 10^{-6}$	874,830	MACH	none	$r^2_{\text{hat}} \geq 0.30$	2473344	1.043	--	--	SAS
FHS controls	Illumina 1Million GeneChip	BeadStudio	$\geq 1\%$	$\geq 98\%$	$> 10^{-6}$	874,830	MACH	none	$r^2_{\text{hat}} \geq 0.30$	2470979	1.032	--	--	SAS
FRAM	Affymetrix 500K Affymetrix 50K supplemental	BRLMM	$\geq 1\%$	$\geq 97\%$	$> 10^{-6}$	378,163	MACH v1.0.15	none	$r^2_{\text{hat}} \geq 0.30$	2455455	1.076	--	--	R
FTC	Illumina HumanHap 318K	BeadStudio	$\geq 1\%$	$\geq 90\%$	$> 10^{-6}$	304,582	MACH	none	$r^2_{\text{hat}} \geq 0.30$	2298003	--	N/A	0.984	ProbABEL
FUSION cases	Illumina Infinium™ II HumanHap300 BeadChip	BeadStudio	$> 1\%$	$\geq 90\%$	$\geq 10^{-6}$	315,635	MACH	none	$r^2_{\text{hat}} \geq 0.30$	2452583	--	1.005	1.011	MACH2QTL
FUSION controls	Illumina Infinium™ II HumanHap300 BeadChip	BeadStudio	$> 1\%$	$\geq 90\%$	$\geq 10^{-6}$	315,635	MACH	none	$r^2_{\text{hat}} \geq 0.30$	2451292	--	0.998	1.012	MACH2QTL
GENMETS cases	Illumina HumanHap 610K	Illuminus	$\geq 1\%$	$\geq 95\%$	$> 10^{-6}$	555,388	MACH	none	$r^2_{\text{hat}} \geq 0.30$	2482250	--	0.998	0.992	ProbABEL
GENMETS controls	Illumina HumanHap 610K	Illuminus	$\geq 1\%$	$\geq 95\%$	$> 10^{-6}$	555,388	MACH	none	$r^2_{\text{hat}} \geq 0.30$	2482703	--	1.008	1.002	ProbABEL

Supplementary Note - Table 2

Study	Platform	Genotyping					SNPs that met QC criteria	Imputation			SNPs in meta-analysis	combined	λ_{GC} men	women	Analysis software
		Genotype calling algorithm	MAF	Inclusion criteria Call rate*	p for HWE			Imputation software	MAF	Inclusion criteria Imputation quality*					
GerMIFSI	Affymetrix NSP/STY	BRLMM	>1%	>97%	$> 10^{-5}$	282,215	MACH	none	$r2_hat \geq 0.30$	2333219	--	0.99	1.014	GenABEL	
GerMiFSII	Affymetrix 6.0	birdseed	>1%	>97%	$> 10^{-5}$	653,149	MACH	none	$r2_hat \geq 0.30$	2492326	--	1.01	1.016	GenABEL	
KORA3	Affymetrix GeneChip Human Mapping 500K	BRLMM	>5%	none	none	380,507	MACH1.0	none	$r2_hat \geq 0.30$	2415072	--	1.005	0.997	MACH2QTL	
KORA4	Affy 6.0	Birdseed	none	$\geq 93\%$	none	906,716	MACH1.16	none	$r2_hat \geq 0.30$	2480938	--	1.001	0.994	MACH2QTL	
MICROS	ILLUMINA318K	BeadStudio	$\geq 1\%$	$\geq 98\%$	$> 10^{-6}$	318237	MACH 1.0	none	$r2_hat \geq 0.30$	2435741	0.992	--	--	ProbABLE	
Migen (cases)	Affymetrix 6.0 Array	Birdseed	>0.01	>0.95	1×10^{-6}	618,475	MACH(1.0.15)	>0.01	$r2_hat \geq 0.30$	2271906	--	1.009 (avg)	1.017 (avg)	MACH2QTL	
Migen (controls)	Affymetrix 6.0 Array	Birdseed	>0.01	>0.95	1×10^{-6}	618,475	MACH(1.0.15)	>0.01	$r2_hat \geq 0.30$	2302578	--	1.002 (avg)	1.014 (avg)	MACH2QTL	
NBS_WTCCC	Affymetrix GeneChip Human Mapping 500K	CHIAMO	>5%	$\geq 95\%$	$> 10^{-6}$	387,667	IMPUTE	none	proper-info ≥ 0.40	2415681	--	1.021	1.01	SNPTEST	
NFBC-1966	Illumina HumanCNV-370DUO Analysis BeadChip	Standard Illumina BeadStudio	$\geq 5\%$	$\geq 95\%$	$> 10^{-4}$	328,007	IMPUTE	none	proper-info ≥ 0.40	2458713	--	1.016	1.018	SNPTEST	
NHS	Illumina HumanHap550	Standard Illumina BeadStudio	$\geq 1\%$	$\geq 90\%$	none	510,073	MACH	none	$r2_hat \geq 0.30$	2520492	--	N/A	0.992	MACH2QTL	
NSPHS	ILLUMINA318K	BeadStudio	$\geq 1\%$	$\geq 98\%$	$> 10^{-6}$	318236	MACH 1.0	none	$r2_hat \geq 0.30$	2385350	1.007			ProbABLE	
NTRNESDA cases	Perlegen - Affymetrix gene chip 600K	Proprietary Perlegen	>1%	$\geq 95\%$	none	435,291	IMPUTE	none	proper-info ≥ 0.40	2490078	--	1.016	1.02	SNPtest	
NTRNESDA controls	Perlegen - Affymetrix gene chip 600K	Proprietary Perlegen	>1%	$\geq 95\%$	none	435,291	IMPUTE	none	proper-info ≥ 0.40	2488884	--	1.021	1.016	SNPtest	
ORCADES	ILLUMINA318K	BeadStudio	$\geq 1\%$	$\geq 98\%$	$> 10^{-6}$	318235	MACH 1.0	none	$r2_hat \geq 0.30$	2434295	1	--	--	ProbABLE	
PLCO	Illumina HumanHap300 and Illumina HumanHap240	BeadStudio	none	$\geq 90\%$	none	523,231	MACH	none	$r2_hat \geq 0.30$	2519768	--	1.009	NA	MACH2QTL	
Procardis	HumanHap300 BeadChips	Illumina Beadstudio 2.0 software	> 5%	$\geq 95\%$	$>5 \times 10^{-7}$	~820k	IMPUTE	none	proper-info ≥ 0.40	2581539	--	1	1.02	SNPTEST	
RS-I	Illumina / HumanHap 550 V.3	Beadstudio GeneCall	$\geq 1\%$	$\geq 97.5\%$	$> 10^{-6}$	512,349	MACH	none	$r2_hat \geq 0.30$	2488215	--	1.029	1.039	MACH2QTL	
RUNMC	Illumina HumanHapCNV370	BeadStudio	$\geq 1\%$	$\geq 96\%$	$> 10^{-6}$	312,199	IMPUTE	none	proper-info ≥ 0.40	2466347	0.999	--	--	SNPtest	
SardiNIA	Affymetrix GeneChip Human Mapping 500K and 10K	BRLMM	$\geq 5\%$	$\geq 90\%$	$> 10^{-6}$	356,359	MACH	none	$r2_hat \geq 0.30$	2251689	1.104	--	--	Merlin -- fastassoc	

Supplementary Note - Table 2

Study	Platform	Genotyping				SNPs that met QC criteria	Imputation			SNPs in meta-analysis	combined	λ_{GC} men	women	Analysis software
		Genotype calling algorithm	MAF	Inclusion criteria Call rate*	p for HWE		Imputation software	MAF	Inclusion criteria Imputation quality*					
SASBAC cases	Illumina HumanHap300+240S	Standard Illumina BeadStudio (GenCall)	$\geq 3\%$	$\geq 90\%$	$> 10^{-7}$	510,578	IMPUTE	none	proper-info ≥ 0.40	2491837	--	N/A	0.998	SNPtest
SASBAC controls	Illumina HumanHap550	Standard Illumina BeadStudio (GenCall)	$\geq 3\%$	$\geq 90\%$	$> 10^{-7}$	512,223	IMPUTE	none	proper-info ≥ 0.40	2474796	--	N/A	1.003	SNPtest
SEARCH/UKOPS	Illumina HumanHap 610 Quad	Illuminus	$\geq 1\%$	$\geq 95\%$	$> 10^{-4}$	495,229	In-house method similar to IMPUTE	none	$r2_hat \geq 0.30$	2486401	--	N/A	1.018	C++
SHIP	Affymetrix Human SNP Array 6.0	Birdseed V2	$\geq 0\%$	$\geq 0\%$	$\geq 0\%$	869,224	IMPUTE v0.5.0	none	proper-info ≥ 0.40	2607463	--	1.014	1.001	SNPTEST v1.1.5 InforSense SNPTEST
T2D_WTCCC	Affymetrix GeneChip Human Mapping 500K	CHIAMO	$> 5\%$	$\geq 95\%$	$> 10^{-6}$	387,667	IMPUTE	none	proper-info ≥ 0.40	2425154	--	1.012	1.017	SNPTEST
TwinsUK	Illumina / HumanHap 300 & 550	Illuminus	$\geq 1\%$	$\geq 95\%$	$> 10^{-6}$	295,702	IMPUTE	none	proper-info ≥ 0.40	2460840	--	N/A	1.015	SNPtest
VIS	ILLUMINA318K	BeadStudio	$\geq 1\%$	$\geq 98\%$	$> 10^{-6}$	317465	MACH 1.0	none	$r2_hat \geq 0.30$	2423020	1.001	--	--	ProbABLE

*SNPtest calculates the 'proper_info' statistic as a measure of the relative statistical information about the additive genetic effect being estimated. The proper_info statistic has a direct relationship to the power of the test and ranges from 0 to 1, with 1 indicating perfect information. MACH calculates the 'rsq_hat', which is the r^2 between each imputed genotype and its true underlying genotype. Rsq_hat ranges from 0 to 1, with 1 indicating perfect imputation

Supplementary Note

Supplementary Note - Table 3 Study-specific descriptive statistics genome-wide association studies of stage 1.

Supplementary Note - Table 3

Supplementary Note - Table 3 -- Study-specific descriptive statistics genome-wide association studies of stage 1.

Study	Trait	Men						Women					
		n	mean	SD	median	min	max	n	mean	SD	median	min	max
ADVANCE Cases	Age (yrs)	114	40.42	3.98	41.20	20.40	45.10	161	49.46	4.68	50.50	34.00	55.00
	BMI (kg/m ²)	114	31.39	5.77	30.89	19.48	54.32	161	31.40	8.17	30.65	17.30	61.08
	Weight (kg)	114	99.03	21.16	97.59	64.05	181.44	161	83.98	21.78	81.74	48.58	153.00
	Height (m)	114	1.77	0.07	1.77	1.61	1.95	161	1.64	0.07	1.64	1.48	1.84
ADVANCE Controls	Age (yrs)	128	40.46	3.23	41.20	33.40	46.80	183	48.69	4.45	49.80	34.80	55.40
	BMI (kg/m ²)	128	27.00	4.48	26.21	17.86	49.38	181	26.08	6.36	24.65	15.76	54.12
	Weight (kg)	128	86.45	16.38	84.37	51.48	158.76	182	71.35	17.13	68.27	40.23	140.71
	Height (m)	128	1.79	0.07	1.78	1.58	1.96	181	1.66	0.06	1.66	1.45	1.80
AGES Midlife	Age (yrs)	1352	49.69	5.87	50.00	34.00	75.00	1867	52.00	6.54	52.00	34.00	77.00
	BMI (kg/m ²)	1351	25.62	3.09	25.48	16.94	38.61	1856	24.89	3.81	24.31	13.65	50.41
	Weight (kg)	1351	81.32	11.41	80.40	51.00	139.00	1856	67.13	10.51	66.00	32.80	140.60
	Height (m)	1352	1.78	0.06	1.78	1.56	1.98	1867	1.64	0.05	1.64	1.45	1.83
Amish	Age (yrs)	468	45.90	16.60	43.00	20.00	98.00	437	47.50	15.10	48.00	20.00	95.00
	BMI (kg/m ²)	468	26.30	3.50	26.00	18.60	39.00	437	28.50	5.70	28.30	16.90	47.10
	Weight (kg)	468	78.60	11.70	77.00	49.40	112.80	437	73.50	14.40	71.90	37.80	114.30
	Height (m)	468	1.73	0.07	1.73	1.48	1.93	437	1.61	0.06	1.61	1.39	1.75
ARIC	Age (yrs)	3822	54.69	5.70	55.00	44.00	66.00	4286	53.97	5.67	54.00	44.00	66.00
	BMI (kg/m ²)	3822	27.48	4.01	26.97	17.21	56.26	4286	26.63	5.52	25.45	14.38	55.20
	Weight (kg)	3822	85.54	13.76	84.10	44.55	182.27	4286	70.00	14.99	66.80	36.36	141.82
	Height (m)	3822	1.76	0.06	1.76	1.49	1.99	4286	1.62	0.06	1.62	1.37	1.87
B58C-T1DGC	Age (yrs)	1259	45.31	0.34	45.33	44.50	46.00	1328	45.27	0.34	45.25	44.50	46.00
	BMI (kg/m ²)	1259	28.02	4.19	27.56	16.84	51.63	1328	26.97	5.58	25.73	17.18	52.20
	Weight (kg)	1259	87.05	14.41	86.00	50.80	177.10	1328	71.63	15.45	68.40	43.00	155.30
	Height (m)	1261	1.76	0.07	1.76	1.55	1.99	1330	1.63	0.06	1.63	1.40	1.85
B58C-WTCCC	Age (yrs)	741	44.89	0.34	44.75	44.50	45.60	738	44.89	0.35	44.75	44.50	45.60
	BMI (kg/m ²)	741	27.84	4.29	27.23	15.93	48.41	738	26.92	5.44	25.56	17.34	56.55
	Weight (kg)	741	86.56	14.63	85.20	51.00	137.50	738	70.96	14.68	68.20	41.80	139.40
	Height (m)	741	1.76	0.07	1.76	1.52	2.02	738	1.62	0.06	1.63	1.42	1.80
BRIGHT	Age (yrs)	719	56.29	11.15	57.00	21.00	84.00	1087	57.43	11.23	58.00	21.00	85.00
	BMI (kg/m ²)	719	27.74	3.28	27.68	17.20	38.26	1087	27.36	4.04	27.03	16.85	41.66
	Weight (kg)	719	84.22	11.90	83.45	51.00	121.00	1087	71.19	11.55	69.90	41.70	122.80
	Height (m)	719	1.74	0.07	1.74	1.51	1.95	1087	1.61	0.06	1.61	1.39	1.81
CAD_WTCCC	Age (yrs)	1489	59.97	7.98	61.00	35.00	82.00	387	60.30	8.47	61.00	36.00	81.00
	BMI (kg/m ²)	1489	27.55	3.91	27.13	16.53	53.40	387	27.84	5.23	27.18	12.81	51.73
	Weight (kg)	1489	83.25	13.07	82.50	37.70	173.00	387	71.04	14.13	69.20	29.20	149.50
	Height (m)	1489	1.74	0.07	1.74	1.40	1.98	387	1.60	0.07	1.58	1.42	1.78
CAPS1 cases	Age (yrs)	505	68.15	7.38	67.90	49.50	81.10						
	BMI (kg/m ²)	484	26.42	3.48	26.01	18.36	41.77						
	Weight (kg)	485	82.50	12.26	82.00	47.00	135.00						
	Height (m)	489	1.77	0.07	1.77	1.58	1.97						
CAPS1 controls	Age (yrs)	506	66.36	7.50	65.90	44.90	79.80						
	BMI (kg/m ²)	483	26.49	3.58	26.25	16.60	58.36						
	Weight (kg)	485	82.75	13.10	82.00	53.00	187.00						
	Height (m)	491	1.77	0.07	1.76	1.58	2.01						
CAPS2 cases	Age (yrs)	1483	66.13	7.07	65.40	44.90	82.20						
	BMI (kg/m ²)	1423	26.34	3.37	25.95	15.74	55.24						
	Weight (kg)	1424	82.53	12.24	82.00	47.00	185.00						
	Height (m)	1452	1.77	0.06	1.77	1.54	2.00						
CAPS2 controls	Age (yrs)	519	67.24	7.35	66.90	49.10	80.10						
	BMI (kg/m ²)	500	26.03	3.32	25.75	17.56	45.20						
	Weight (kg)	504	80.80	11.38	80.00	55.00	140.00						
	Height (m)	511	1.76	0.06	1.76	1.59	1.98						
CHS	Age (yrs)	1281	73.00	5.66	72.00	65.00	95.00	1957	71.90	5.15	71.00	65.00	98.00
	BMI (kg/m ²)	1276	26.40	3.50	26.10	18.60	44.20	1952	26.40	4.78	25.80	18.50	48.30
	Weight (kg)	1276	79.70	11.90	79.00	50.00	145.00	1952	67.10	12.90	65.50	37.30	133.20
	Height (m)	1277	1.73	0.07	1.73	1.51	1.92	1955	1.59	0.06	1.59	1.24	1.78
CoLaus	Age (yrs)	2547	52.92	10.77	52.20	34.90	75.10	2862	53.88	10.72	53.70	35.00	75.40
	BMI (kg/m ²)	2547	26.64	4.19	26.20	11.70	81.10	2861	25.15	4.91	24.20	8.10	59.20
	Weight (kg)	2547	81.54	13.41	79.90	36.50	175.40	2861	66.43	12.98	64.00	21.40	171.00
	Height (m)	2547	1.75	0.07	1.75	1.33	1.98	2862	1.62	0.07	1.63	1.31	1.85
deCODE*	Age (yrs)	9213	64.74	15.93	78.00	18.00	103.00	17586	57.94	18.46	43.00	11.50	108.00
	BMI (kg/m ²)	9213	27.71	4.70	45.99	14.52	72.14	17586	26.83	5.49	18.56	13.67	73.51
	Weight (kg)	9213	87.89	16.57	149.00	40.00	216.00	17586	73.49	15.77	53.00	33.00	220.00
	Height (m)	9213	1.78	0.07	1.80	1.30	2.07	17586	1.65	0.06	1.69	1.34	1.99
DGI (cases)	Age (yrs)	688	63.42	9.91	64.37	37.10	84.96	629	65.20	10.12	66.31	36.79	84.97
	BMI (kg/m ²)	688	28.09	3.83	27.99	18.05	46.71	629	28.62	4.80	28.16	18.51	53.73
	Weight (kg)	688	85.64	13.26	84.50	53.40	148.00	629	74.47	13.31	73.30	43.80	141.00
	Height (m)	688	1.75	0.06	1.74	1.43	1.96	629	1.61	0.06	1.61	1.41	1.80

Supplementary Note - Table 3

Study	Trait	Men						Women					
		n	mean	SD	median	min	max	n	mean	SD	median	min	max
DGI (controls)	Age (yrs)	553	58.08	9.74	58.22	35.54	79.71	535	58.40	9.40	58.70	35.16	80.56
	BMI (kg/m ²)	553	26.76	3.22	26.53	16.95	43.89	535	26.76	4.11	26.17	17.96	42.77
	Weight (kg)	553	82.92	11.47	82.00	57.40	143.00	535	71.02	11.76	70.00	46.80	124.00
	Height (m)	553	1.76	0.06	1.76	1.59	2.00	535	1.63	0.06	1.63	1.42	1.87
EGCUT	Age (yrs)	697	40.62	16.78	38.00	18.00	90.00	720	42.88	15.93	42.00	18.00	92.00
	BMI (kg/m ²)	697	26.05	4.61	25.39	15.82	54.00	720	26.25	6.02	25.08	15.90	58.40
	Weight (kg)	697	83.32	15.27	82.00	49.00	191.00	720	71.41	16.36	68.00	39.00	160.00
	Height (m)	697	1.79	0.07	1.79	1.58	2.03	720	1.65	0.06	1.65	1.45	1.84
EPIC-Obesity Study	Age (yrs)	1131	59.82	9.02	60.00	40.00	77.00	1284	58.74	9.00	59.00	39.00	77.00
	BMI (kg/m ²)	1131	26.62	3.21	26.22	16.85	42.06	1284	26.19	4.39	25.50	16.09	47.57
	Weight (kg)	1131	80.43	11.09	79.40	42.80	134.00	1284	67.98	11.62	66.20	44.60	119.00
	Height (m)	1131	1.73	0.07	1.73	1.51	1.96	1284	1.61	0.06	1.60	1.37	1.82
ERF	Age (yrs)	890	50.14	14.98	50.67	18.00	88.60	1170	49.30	15.34	49.52	18.03	92.10
	BMI (kg/m ²)	890	27.14	3.98	26.78	15.85	42.44	1170	26.36	4.77	25.64	15.54	45.37
	Weight (kg)	890	82.70	13.52	81.40	48.00	133.30	1170	68.96	13.14	67.00	42.10	133.90
	Height (m)	890	1.75	0.07	1.75	1.52	1.96	1170	1.61	0.07	1.62	1.41	1.83
Fenland	Age (yrs)	615	44.48	7.32	45.00	30.00	57.00	787	45.34	7.18	46.00	30.00	57.00
	BMI (kg/m ²)	615	27.62	4.07	27.27	18.62	56.66	787	26.68	5.46	25.44	17.27	55.39
	Weight (kg)	615	86.76	13.87	85.50	49.40	155.70	787	71.48	15.25	68.30	42.40	142.50
	Height (m)	615	1.77	0.07	1.77	1.59	2.01	787	1.64	0.06	1.64	1.43	1.90
FHS cases	Age (yrs)	220	54.20	11.87	55.75	26.38	74.14	243	57.43	10.08	58.42	26.48	84.00
	BMI (kg/m ²)	208	28.51	4.68	28.15	15.96	45.72	233	28.27	6.51	26.75	18.43	50.18
	Weight (kg)	208	89.04	15.03	87.77	51.71	146.51	233	73.87	17.94	69.40	45.36	1.00
	Height (m)	208	1.77	0.07	1.77	1.58	1.96	233	1.62	0.06	1.62	1.42	1.79
FHS controls	Age (yrs)	218	52.09	12.20	54.19	26.99	76.86	216	58.25	8.57	59.10	27.33	81.09
	BMI (kg/m ²)	208	27.74	3.59	27.10	19.56	42.51	207	26.64	4.66	25.61	17.48	43.39
	Weight (kg)	208	87.22	13.09	84.80	57.61	131.09	207	70.34	13.59	67.59	43.09	122.05
	Height (m)	208	1.77	0.07	1.78	1.55	1.98	207	1.62	0.06	1.63	1.46	1.81
FRAM	Age (yrs)	3706	38.72	8.73	38.00	21.00	72.00	4388	38.22	8.64	38.00	21.00	70.00
	BMI (kg/m ²)	3706	27.08	4.18	26.61	16.91	56.54	4388	24.89	5.26	23.58	14.96	60.58
	Weight (kg)	3706	84.41	14.45	82.56	30.39	177.36	4388	65.85	14.64	62.60	38.10	170.10
	Height (m)	3706	1.76	0.07	1.77	1.23	2.00	4388	1.63	0.06	1.63	1.33	1.85
FTC	Age (yrs)							125	63.78	11.68	66.38	26.52	75.94
	BMI (kg/m ²)							125	25.07	3.41	24.65	18.69	35.04
	Weight (kg)							125	65.25	9.20	64.00	46.50	93.00
	Height (m)							125	1.61	0.06	1.61	1.46	1.77
FUSION cases	Age (yrs)	623	62.06	7.33	62.41	40.77	77.81	469	63.66	7.75	64.01	45.00	83.19
	BMI (kg/m ²)	623	29.44	4.02	29.14	18.19	43.14	469	31.20	5.25	30.71	16.00	47.59
	Weight (kg)	623	88.43	13.58	88.00	50.90	144.00	469	79.51	14.70	76.90	35.00	125.50
	Height (m)	617	1.73	0.06	1.73	1.52	1.97	465	1.60	0.06	1.59	1.40	1.76
FUSION controls	Age (yrs)	572	63.41	7.62	64.00	46.00	90.91	599	63.71	7.27	64.75	42.60	89.15
	BMI (kg/m ²)	572	27.02	3.53	26.78	19.22	51.07	599	27.24	4.15	26.80	17.50	45.90
	Weight (kg)	572	81.40	11.98	80.65	52.10	151.10	599	69.99	11.44	68.80	45.70	127.10
	Height (m)	569	1.74	0.06	1.74	1.56	1.91	598	1.60	0.06	1.60	1.44	1.79
GENMETS cases	Age (yrs)	425	49.19	10.42	49.00	30.00	75.00	432	52.36	11.71	51.00	30.00	75.00
	BMI (kg/m ²)	425	29.45	3.61	28.86	23.19	47.07	432	29.70	4.94	28.76	20.58	45.78
	Weight (kg)	410	91.16	12.56	89.00	65.00	151.00	414	76.60	13.40	75.00	49.00	123.00
	Height (m)	410	1.76	0.07	1.75	1.57	1.97	414	1.61	0.07	1.61	1.34	1.81
GENMETS controls	Age (yrs)	401	48.91	10.15	49.00	30.00	74.00	423	48.59	10.17	49.00	30.00	74.00
	BMI (kg/m ²)	401	25.41	3.08	24.94	17.09	39.04	423	25.35	3.15	24.92	17.09	39.04
	Weight (kg)	401	78.03	10.33	77.00	54.00	116.00	422	77.62	10.60	77.00	51.00	113.00
	Height (m)	401	1.75	0.07	1.75	1.54	1.96	422	1.75	0.07	1.75	1.54	1.96
GerMiFSI	Age (yrs)	394	57.27	8.57	59.00	32.00	82.00	206	60.39	8.67	61.00	36.00	82.00
	BMI (kg/m ²)	394	27.36	3.30	26.83	18.42	46.24	206	27.17	4.17	26.91	19.05	40.75
	Weight (kg)	394	83.92	11.67	83.00	60.00	140.00	206	72.29	12.24	71.00	48.00	115.00
	Height (cm)	394	1.75	0.06	1.75	1.59	1.97	206	1.63	0.06	1.63	1.44	1.79
GerMiFSII	Age (yrs)	901	60.14	12.17	59.00	29.00	88.00	223	62.80	12.76	61.00	34.00	90.00
	BMI (kg/m ²)	901	27.82	3.54	27.41	18.44	54.08	223	28.06	4.76	27.69	16.90	46.30
	Weight (kg)	901	83.00	12.49	83.00	50.20	160.00	223	73.55	12.62	72.10	47.00	130.00
	Height (cm)	901	1.74	0.07	1.73	1.52	2.00	223	1.62	0.06	1.61	1.49	1.79
KORA3	Age (yrs)	813	52.96	10.09	54.00	25.00	69.00	831	52.09	10.08	53.00	25.00	69.00
	BMI (kg/m ²)	813	27.69	3.45	27.29	18.73	40.67	829	26.98	4.64	26.40	16.71	45.43
	Weight (kg)	813	83.58	11.46	83.30	59.00	132.50	829	69.87	11.88	68.30	42.50	121.80
	Height (m)	813	1.74	0.07	1.74	1.51	1.96	830	1.61	0.06	1.61	1.44	1.80
KORA4	Age (yrs)	884	54.22	8.92	54.00	28.00	72.00	930	53.62	8.80	53.00	25.00	74.00
	BMI (kg/m ²)	883	27.99	3.91	27.59	18.31	55.11	928	27.49	5.07	26.78	18.21	51.22
	Weight (kg)	883	85.13	12.93	84.00	54.20	192.70	929	71.46	13.30	69.60	43.90	142.00
	Height (m)	883	1.74	0.07	1.74	1.56	1.94	928	1.61	0.06	1.61	1.43	1.82
MICROS	Age (yrs)	475	45.09	15.67	41.97	18.19	87.85	622	45.38	16.41	42.55	18.00	83.88
	BMI (kg/m ²)	475	26.07	3.96	25.62	18.13	42.75	622	25.28	5.32	24.27	14.03	71.26
	Weight (kg)	468	78.38	13.32	76.90	47.00	127.50	612	65.16	13.19	63.00	36.60	169.00

Supplementary Note - Table 3

Study	Trait	Men						Women					
		n	mean	SD	median	min	max	n	mean	SD	median	min	max
Migen (cases)	Height (m)	467	1.73	0.07	1.73	1.53	1.95	612	1.61	0.07	1.61	1.40	1.79
	Age (yrs)	800	44.53	4.77	45.97	19.38	51.00	474	49.66	7.28	51.22	23.00	61.00
	BMI (kg/m ²)	775	28.79	4.99	28.27	3.25	54.30	470	28.97	7.42	27.40	3.37	74.72
	Weight (kg)	779	88.72	17.91	86.18	52.16	181.60	470	77.01	21.43	72.57	43.09	205.02
Migen (controls)	Height (m)	782	177.02	26.13	175.26	153.70	525.78	474	164.91	29.76	162.56	110.00	525.78
	Age (yrs)	846	46.23	8.41	45.51	20.75	92.00	561	49.22	7.50	50.93	18.71	61.00
	BMI (kg/m ²)	844	27.12	4.08	26.74	17.99	50.21	558	27.07	6.65	25.70	17.99	78.41
	Weight (kg)	844	84.52	14.39	82.20	52.00	158.00	558	72.28	17.87	68.04	44.00	203.00
NBS_WTCCC*	Height (m)	844	176.42	7.48	176.20	153.00	208.28	559	163.52	6.71	163.00	136.00	184.00
	Age (yrs)	694	45.46	11.75	47.00	17.00	69.00	743	41.44	12.59	42.00	17.00	69.00
	BMI (kg/m ²)	694	26.76	4.12	26.30	18.13	53.19	743	25.75	4.46	24.86	18.08	47.22
	Weight (kg)	694	85.03	14.35	82.73	54.09	173.00	743	69.74	12.21	66.82	50.00	127.27
NFBC-1966	Height (m)	694	1.78	0.07	1.77	1.50	2.00	743	1.65	0.07	1.65	1.48	1.82
	Age (yrs)	2250	31.00	0.00	31.00	31.00	31.00	2247	31.00	0.00	31.00	31.00	31.00
	BMI (kg/m ²)	2250	25.18	3.62	24.86	15.32	47.58	2247	24.16	4.68	23.13	15.43	54.35
	Weight (kg)	2250	80.15	12.72	78.70	49.40	150.40	2247	65.52	13.24	63.00	29.20	165.40
NHS	Height (m)	2250	1.78	0.06	1.78	1.52	2.02	2247	1.65	0.06	1.64	1.05	1.86
	Age (yrs)							2265	54.46	6.45	55.00	39.00	66.00
	BMI (kg/m ²)							2265	25.13	4.53	24.13	16.40	53.14
	Weight (kg)							2265	149.16	27.78	144.00	84.00	310.00
NSPHS*	Height (m)							2265	1.64	0.06	1.63	1.45	1.98
	Age (yrs)	309	47.56	20.83	48.00	15.00	87.00	347	46.47	20.60	45.00	14.00	91.00
	BMI (kg/m ²)	307	26.75	4.54	26.23	17.78	46.49	340	25.97	5.07	24.98	16.44	46.68
	Weight (kg)	307	78.42	14.66	77.00	51.00	138.00	342	64.99	13.11	63.00	38.00	121.00
NTRNESDA cases	Height (m)	308	1.71	0.07	1.72	1.48	1.89	344	1.58	0.07	1.59	1.40	1.75
	Age (yrs)	527	44.65	11.84	46.00	18.00	74.00	1200	41.62	12.77	42.00	18.00	77.00
	BMI (kg/m ²)	527	26.42	4.43	26.01	15.95	50.21	1200	25.41	5.31	24.23	14.61	53.27
	Weight (kg)	527	87.07	15.22	85.50	51.00	170.00	1200	72.44	15.15	70.00	44.00	167.00
NTRNESDA controls	Height (m)	527	1.82	0.07	1.81	1.59	2.07	1200	1.69	0.06	1.69	1.51	1.96
	Age (yrs)	683	47.17	14.45	50.00	18.00	81.00	1106	43.72	13.63	42.00	18.00	78.00
	BMI (kg/m ²)	683	25.76	3.46	25.40	17.26	39.19	1106	24.86	4.22	24.08	15.88	47.32
	Weight (kg)	683	84.98	12.53	83.80	50.10	133.10	1106	71.16	12.75	68.80	44.30	150.00
ORCADES*	Height (m)	683	1.81	0.07	1.82	1.60	2.06	1106	1.69	0.06	1.69	1.50	1.92
	Age (yrs)	332	54.27	15.73	54.66	17.29	93.75	384	53.01	15.68	54.27	17.71	97.62
	BMI (kg/m ²)	332	28.08	4.27	27.67	16.97	47.10	384	27.48	5.18	26.60	18.47	47.63
	Weight (kg)	324	85.76	13.21	84.25	44.40	148.40	371	71.06	13.69	69.10	45.60	123.10
PLCO	Height (m)	324	1.75	0.07	1.75	1.59	1.99	371	1.61	0.06	1.61	1.38	1.78
	Age (yrs)	2238	64.2	5.1	64.0	55.0	74.0						
	BMI (kg/m ²)	2238	27.5	3.8	27.1	13.3	48.2						
	Weight (kg)	2238	87.4	13.6	86.2	38.6	176.9						
Procardis	Height (m)	2238	1.78	0.07	1.78	1.55	2.08						
	Age (yrs)	1700	59.29	7.08	60.00	34.00	82.00	612	61.21	6.72	62.00	33.00	81.00
	BMI (kg/m ²)	1700	27.60	3.80	27.14	18.34	48.23	612	26.71	5.00	25.94	15.43	51.37
	Weight (kg)	1700	84.51	12.91	83.50	51.00	159.00	612	71.21	13.30	69.00	42.00	145.00
RS-I	Height (m)	1700	1.75	0.07	1.75	1.51	2.06	612	1.63	0.07	1.64	1.44	1.85
	Age (yrs)	2427	68.13	8.16	67.05	55.01	97.81	3547	70.32	9.60	69.40	55.00	99.22
	BMI (kg/m ²)	2372	25.68	2.99	25.61	14.19	38.19	3372	26.74	4.10	26.31	15.43	59.50
	Weight (kg)	2375	78.58	10.74	77.80	41.00	122.30	3383	69.59	11.29	68.70	40.10	146.50
RUNMC	Height (m)	2372	1.75	0.07	1.74	1.51	1.98	3375	1.61	0.07	1.61	1.01	1.91
	Age (yrs)	1777	63.47	8.34	64.00	24.00	91.00	1096	55.41	11.14	64.00	25.00	91.00
	BMI (kg/m ²)	1777	25.98	3.66	21.90	16.10	61.30	1096	25.44	4.26	24.50	17.30	52.70
	Weight (kg)	1777	81.49	12.33	75.00	46.00	185.00	1096	70.30	12.16	75.00	46.00	150.00
SardiNIA*	Height (m)	1777	1.77	0.07	1.85	1.55	2.00	1096	1.66	0.06	1.75	1.38	1.85
	Age (yrs)	1886	44.1	18.1	42.9	14.0	93.9	2419	43.2	17.3	42.1	14.0	101.3
	BMI (kg/m ²)	1885	26.1	4.1	25.9	14.9	42.9	2416	24.7	5.0	23.8	13.9	53.3
	Weight (kg)	1883	72.3	11.7	72.0	34.0	135.0	2415	59.2	11.4	57.0	32.0	145.0
SASBAC cases	Height (m)	1883	1.66	0.07	1.66	1.44	1.96	2415	1.55	0.07	1.55	1.31	1.78
	Age (yrs)							795	62.64	6.26	63.00	50.00	75.00
	BMI (kg/m ²)							793	25.79	4.00	25.21	16.22	46.67
	Weight (kg)							794	69.68	11.18	68.00	40.00	117.00
SASBAC controls	Height (m)							794	1.64	0.06	1.65	1.47	1.82
	Age (yrs)							764	62.77	6.34	63.00	49.00	75.00
	BMI (kg/m ²)							755	25.52	4.10	25.22	16.94	59.52
	Weight (kg)							760	68.67	11.69	67.00	42.00	168.00
SEARCH/UKOPS	Height (m)							758	1.63	0.05	1.64	1.28	1.81
	Age (yrs)							1710	57.15	10.20	58.00	20.00	91.00
	BMI (kg/m ²)							1556	26.99	5.20	25.99	17.47	53.67
	Weight (kg)							1581	71.32	13.99	69.00	44.00	135.17
SHIP	Height (m)							1592	1.62	0.07	1.62	1.34	1.82
	Age (yrs)	2019	50.88	16.43	52.00	20.00	80.00	2073	48.58	16.02	48.00	20.00	81.00
	BMI (kg/m ²)	2019	27.68	4.04	27.41	18.06	48.07	2073	26.92	5.31	26.16	16.10	52.40

Supplementary Note - Table 3

Study	Trait	Men						Women					
		n	mean	SD	median	min	max	n	mean	SD	median	min	max
T2D_WTCCC	Weight (kg)	2019	85.06	13.56	83.80	49.90	156.40	2073	71.20	13.74	69.20	41.30	133.30
	Height (m)	2019	1.75	0.07	1.75	1.48	1.98	2073	1.63	0.07	1.63	1.42	1.94
	Age (yrs)	1105	58.95	9.91	59.00	29.00	96.00	798	57.94	10.45	59.00	27.00	85.00
	BMI (kg/m ²)	1105	30.29	5.36	29.71	18.02	55.91	798	32.56	6.87	31.52	17.91	62.37
	Weight (kg)	1105	93.37	17.86	91.17	47.63	161.94	798	85.04	19.29	82.56	43.00	155.70
TwinsUK*	Height (m)	1105	1.75	0.07	1.75	1.50	1.98	798	1.61	0.07	1.61	1.37	1.82
	Age (yrs)							1479	46.19	12.31	47.55	16.62	76.54
	BMI (kg/m ²)							1477	25.02	4.80	24.06	13.22	52.71
	Weight (kg)							1477	66.03	12.97	64.00	35.10	140.90
VIS	Height (m)							1479	1.62	0.06	1.63	1.42	1.80
	Age (yrs)	328	55.95	14.94	57.00	18.00	88.00	467	56.97	15.64	57.00	18.00	93.00
	BMI (kg/m ²)	328	27.55	3.69	27.49	18.36	40.69	467	27.18	4.50	27.08	17.01	52.02
	Weight (kg)	325	85.56	13.01	84.80	50.90	136.50	445	70.99	12.45	69.80	46.60	153.00
	Height (m)	325	1.76	0.07	1.76	1.58	2.04	459	1.62	0.07	1.62	1.43	1.91

*Includes individuals <18 years of age (deCODE: n=3; NBS_WTCCC: n=10 (17 years); NSPHS: n=55 (range 14-17 years); ORCADES: n=2 (17.3 and 17.7 years); SardiNIA: n=294 (range 14-17 years); TwinsUK: n=5)

Supplementary Note

Supplementary Note - Table 4 Study design, number of individuals and sample quality control for genome-wide association studies of stage 2 - *in silico* and *de novo* replication.

Supplementary Note - Table 4

Supplementary Note - Table 4 -- Study design, number of individuals and sample quality control for genome-wide association studies of stage 2 - in silico and de novo replication.

Study		Study design	Total sample size (N)	Call rate*	Sample QC	Samples in analyses (N)†	Anthropometric assessment method	References
Short name	Full name				Study specific QC criteria			
<i>In silico</i> follow-up studies								
BHS	Busselton Health Study	Population-based	1,366	≥ 75%	1) ethnic outliers; 2) related individuals and duplicates; 3) Missing body waist and hip.	1,327	measured	James AL, Palmer LJ, Kicic E, et al. Decline in lung function in the Busselton Health Study: the effects of asthma and cigarette smoking. <i>Am J Respir Crit Care Med</i> 2005;171:109-14. Hui J, Oka A, James A, et al. A genome-wide association scan for asthma in a general Australian population. <i>Hum Genet</i> 2008;123:297-306
Corogene	Genetic Predisposition of Coronary Heart Disease in Patients Verified with Coronary Angiogram	Population-based	4,130	≥ 95%	1) missing gender 2) related individuals and duplicates 3) (From this specific analysis) Missing body height and/or weight	3756	measured	[PMID: 19820697] Soranzo, N. et al. A genome-wide meta-analysis identifies 22 loci associated with eight hematological parameters in the HaemGen consortium. <i>Nat Genet.</i> 2009 Nov;41(11):1182-90. Epub 2009 Oct 11.
EGCUT	Estonian Genome Center, University of Tartu	Population-based	345	≥ 95%	1) Related individuals and duplicates 2) Sex mismatch 3) Phenotype missing	345	measured	Nelis M, Esko T, Magi R, Zimprich F, Zimprich A, et al. (2009) Genetic Structure of Europeans: A View from the North–East. <i>PLoS ONE</i> 4(5): e5472. Metspalu, A. The Estonian Genome Project. <i>Drug Development Research</i> 62, 97-101 (2004).
FHS	Family Heart Study	Case-Control	1808	≥ 98%	1) technical errors 2) discrepancies between reported sex and sex-diagnostic markers	1463	measured	[PMID: 8651220] M. Higgins <i>et al.</i> , NHLBI Family Heart Study: objectives and design, <i>Am J Epidemiol</i> 143, 1219–1228 (1996).
FinGesture cases	Finnish Genetic Study of Arrhythmic Disease cohort (MI cases only)	Disease cohort (MI cases only)	1,103	≥ 97%	1) PLINK heterozygosity F-value <-0.05 or >0.05; 2) ethnic outliers; 3) related individuals and duplicates; 4) Missing body weight and height.	943	measured	Kari S. Kaikkonen, Marja-Leena Kortelainen, Eeva Linna, Heikki V. Huikuri. Family History and the Risk of Sudden Cardiac Death as a Manifestation of an Acute Coronary Event <i>Circulation</i> 114, 1462-7 (2006).
GOOD	Gothenburg Osteoporosis and Obesity Determinants Study	Population-based	1056	≥ 97.5%	1) heterozygosity > 33%; 2) ethnic outliers; 3) related individuals and duplicates.	938	measured	[PMID: 16007330] Lorentzon, M. et al Free testosterone is a positive whereas free estradiol is a negative predictor of cortical bone size in young Swedish men-The GOOD Study. <i>J Bone Miner Res</i> 20, 1334-1341 (2005).
HBCS	Helsinki Birth Cohort Study	Population-based birth cohort	1,872	≥ 95%	1) related individuals and duplicates 2) (From this specific analysis) Missing body height and/or weight	1726	measured	[PMID: 18541567] Ylihärsilä H, Kajantie E, Osmond C, Forsén T, Barker DJ, Eriksson JG. Body mass index during childhood and adult body composition in men and women aged 56-70 y. <i>Am J Clin Nutr.</i> 2008 Jun;87(6):1769-75. [PMID: 15764690] Kajantie E, Osmond C, Barker DJ, Forsén T, Phillips DI, Eriksson JG: Size at birth as a predictor of mortality in adulthood: a follow-up of 350 000 person-years. <i>Int J Epidemiol</i> 2005;34:655-663.

Supplementary Note - Table 4

Study		Study design	Total sample size (N)	Call rate*	Sample QC	Samples in analyses (N)†	Anthropometric assessment method	References
Short name	Full name				Study specific QC criteria			
HYPERGENES - cases	HYPERGENES	Case-control	2124	>90%	1) ethnic outliers 2) Missing body weight and height.	1788	measured	http://www.hypergenes.eu/
HYPERGENES - controls	HYPERGENES	Case-control	1934	>90%	1) ethnic outliers 2) Missing body weight and height.	1838	measured	http://www.hypergenes.eu/
MGS	Molecular Genetics of Schizophrenia/NIMH Repository Control Sample	Population-based (survey research method)	2681	99.7%	1) call rate < 97% for samples, 95% for SNPs 2) heterozygosity <26% or >28.5% 3) excess duplicate discordancies or mendelian errors (SNPs) 4) ethnic outliers (principal component scores) 5) related individuals and	2597	self-report	[PMID: 19571809] Shi, J, et al. Common variants on chromosome 6p22.1 are associated with schizophrenia. <i>Nature</i> . 2009 Aug 6;460(7256):753-7. [PMID: 18198266] Sanders, A.R. No significant association of 14 candidate genes with schizophrenia in a large European ancestry sample: implications for psychiatric genetics. <i>Am J Psychiatry</i> . 2008 Apr;165(4):497-506.
NHS	The Nurses' Health Study	Nested case-control	3221	>98%	1) Low genotyping completion (<98%); 2) Unclear identity and admixed origin; 3) related individuals and duplicates; 4) DNA contamination; 5) Missing body weight and height;	2988	self-report	[PMID: 20147318] Qi L et al. Genetic variants in ABO blood group region, plasma soluble E-selectin levels, and risk of type 2 diabetes. <i>Hum Mol Genet</i> . 2010 Feb 10
RS-II	Rotterdam Study II	Population-based	3,011	≥ 97.5%	1) gender mismatch with typed X-linked markers; 2) excess autosomal heterozygosity ($F < -0.055$); 3) duplicates and/or 1st degree relatives using IBD PIHAT >40% from PLINK; 4) ethnic outliers IBS distances > 4SD mean HaMAP CEU cluster from PLINK; 5) Missin	2,122	measured	[PMID: 11753597] Visscher, 2001 A comparison of body mass index, waist-hip ratio and waist circumference as predictors of all-cause mortality among the elderly: the Rotterdam study [PMID:19700477] Estrada, 2009 GRIMP: a web- and grid-based tool for high-speed analysis of large-scale genome-wide association using imputed data [PMID:19728115] Hofman, 2009 The Rotterdam Study: 2010 objectives and design update [PMID:1833235] Hofman, 1991 Determinants of disease and disability in the elderly: the Rotterdam Elderly Study
Sorbs	Sorbs are self-contained population from Eastern Germany, European Descent	Population-based	1097	≥ 94%	1) gender mismatch; 2) ethnic outliers; 3) duplicates; 4) Missing body weight and height.	907	measured	[PMID: 19584900] Tönjes, A. et al. Association of FTO variants with BMI and fat mass in the self-contained population of Sorbs in Germany. <i>Eur J Hum Genet</i> . 2010 Jan;18(1):104-10.

Supplementary Note - Table 4

Study		Study design	Total sample size (N)	Call rate*	Sample QC	Samples in analyses (N)†	Anthropometric assessment method	References
Short name	Full name				Study specific QC criteria			
WGHS	Women's Genome Health Study	Population-based	23,294	>98%	1) includes only WGHS participants with confirmed, self-reported European ancestry; 2) all SNPs have HWE $p > 10E-6$; 3) all SNPs have genotype for >90% samples 4) only samples with biometric measures included in analysis	22,888	self-report	[PMID: 18070814] Ridker PM, Chasman DI, Zee RY, Parker A, Rose L, Cook NR, Buring JE; Women's Genome Health Study Working Group. Rationale, design, and methodology of the Women's Genome Health Study: a genome-wide association study of more than 25,000 initially healthy american women. Clin Chem. 2008 Feb;54(2):249-55. Epub 2007 Dec 10.
YFS	The Cardiovascular Risk in Young Finns Study	Population-based cohort	2,443	≥ 95%	1) missing gender 2) related individuals and duplicates 3) (From this specific analysis) Missing body height and/or weight	1989	measured	Raitakari OT et al. Cohort profile: The cardiovascular risk in Young Finns Study. Int J Epidemiol. 2008;37:1220-6
De novo follow-up studies								
B58C-WTCCC & B58C replication set (R58)	British 1958 birth cohort (Type 1 Diabetes Genetic Consortium controls & Wellcome Trust Case Control Consortium controls)	Population-based	1345	N/A	Missing phenotypes	1340	measured	[PMID 17255346] Strachan DP, Rudnicka AR, Power C, Shepherd P, Fuller E, Davis A, Gibb I, Kumari M, Rumley A, Macfarlane GJ, Rahi J, Rodgers B, Stansfeld S. Lifecourse influences on health among British adults: effects of region of residence in childhood and adulthood. Int J Epidemiol 2007;36:522-531. [PMID 16155052] Power C, Elliott J. Cohort profile: 1958 British birth cohort (National Child Development Study). Int J Epidemiol 2006;35:34-41
BCG	Barry Caerphilly Growth Study	Population based	659	N/A	1. Missing phenotypes 2. Missing DNA	634	measured	[PMID: 12791629] Martin, R. M., McCarthy, A., Smith, G. D., Davies, D. P., and Ben-Shlomo, Y. Infant nutrition and blood pressure in early adulthood: the Barry Caerphilly Growth study. American Journal of Clinical Nutrition, 77: 1489-97, 2003.
BPPP	Botnia Prevalence, Prediction and Prevention of Diabetes study	Population-based	3505	N/A	1. Missing phenotypes 2. Missing DNA	3471	measured	Isomaa B, Forsén B, Lahti K, Holmström N, Wadén J, Matintupa O, Almgren P, Eriksson JG, Lyssenko V, Taskinen M-R, Tuomi T, Groop LC. A family history of diabetes is associated with reduced physical fitness in the Prevalence, Prediction and Prevention of Diabetes (PPP)-Botnia study. Diabetologia 2010, in press.
BWHHS	British Women's Heart and Health Study	Population based	3757	N/A	1. Missing phenotypes 2. Missing DNA	3687	measured	Lawlor, D. A., Bedford, C., Taylor, M., and Ebrahim, S. Geographical variation in cardiovascular disease, risk factors, and their control in older women: British Women's Heart and Health Study. Journal of Epidemiology & Community Health. 57: 134-40. 2003.
DESIR	Data from the Epidemiological Study on the Insulin Resistance Syndrome	Population-based	5212	N/A	1. Missing DNA 2. Missing phenotypes 3. Non Europeans or born outside continental France	4535	measured	[PMID: 8927780] Balkau B. [An epidemiologic survey from a network of French Health Examination Centres, (D.E.S.I.R.): epidemiologic data on the insulin resistance syndrome]. Rev Epidemiol Sante Publique. 1996 Aug;44(4):373-5. French.

Supplementary Note - Table 4

Study		Study design	Total sample size (N)	Call rate*	Sample QC	Samples in analyses (N)†	Anthropometric assessment method	References
Short name	Full name				Study specific QC criteria			
DIAGEN	Diabetes Genetics	Population-based	1855	N/A	Missing phenotypes	1821	measured	[PMID: 16801592] Fischer S. et al. Hypoadiponectinemia is associated with progression toward type 2 diabetes and genetic variation in the ADIPOQ gene promoter. <i>Diabetes Care</i> 29, 1645-50 (2006).
EGCUT	Estonian Genome Center, University of Tartu	Population-based	3779	N/A	1. Missing phenotypes	3779	measured	Nelis M, Esko T, Magi R, Zimprich F, Zimprich A, et al. (2009) Genetic Structure of Europeans: A View from the North–East. <i>PLoS ONE</i> 4(5): e5472. Metspalu, A. The Estonian Genome Project. <i>Drug Development Research</i> 62, 97-101 (2004).
EPIC-Norfolk	European Prospective Investigation into Cancer and Nutrition - Norfolk	Population-based	19086	N/A	1. Missing phenotypes 2. Missing DNA	18484	measured	[PMID: 10466767] Day, N.E. et al. EPIC-Norfolk: study design and characteristics of the cohort. <i>European Prospective Investigation of Cancer. British Journal of Cancer</i> 80, 95-103 (1999).
EPIC-Potsdam	European Prospective Investigation into Cancer and Nutrition - Potsdam	Population-based	5,000	N/A	1. Missing DNA	4,996	measured	[PMID: 10592369] Boeing, H. et al. Recruitment procedures of EPIC-Germany. <i>European Investigation into Cancer and Nutrition. Ann Nutr Metab</i> 43, 205-215 (1999)
Finrisk	FINRISK 97	Population-based	8447	N/A	1. Missing phenotypes 2. Missing DNA	7818	measured	Vartiainen, E. et al. Cardiovascular risk factor changes in Finland, 1972-1997. <i>Int. J. Epidemiol.</i> 29, 49-56 (2000).
FUSION Stage 2 T2D - cases**	Finland-United States Investigation of NIDDM Genetics: T2D cases	Case-control	1233	N/A	Missing phenotypes	1226	measured	[PMID 17463248] Scott L.J. et al. A genome-wide association study of type 2 diabetes in Finns detects multiple susceptibility variants. <i>Science</i> 316, 1341-5 (2007).
FUSION Stage 2 T2D - controls***	Finland-United States Investigation of NIDDM Genetics: T2D controls	Case-control	3704	N/A	Missing phenotypes	3696	measured	[PMID 17463248] Scott L.J. et al. A genome-wide association study of type 2 diabetes in Finns detects multiple susceptibility variants. <i>Science</i> 316, 1341-5 (2007).
HUNT2	Nord-Trøndelag Health (HUNT) 2 Study	Population-based	6399	N/A	Missing phenotypes	4864	measured	[PMID 10546013] Midthjell, K. et al. Rapid changes in the prevalence of obesity and known diabetes mellitus in an adult Norwegian population. The Nord-Trøndelag Health Surveys: 1984-1986 and 1995-1997. <i>Diabetes Care</i> 22, 1813-1820 (1999). Holmen, J. et al. The Nord-Trøndelag Health Study 1995-97 (HUNT 2). Objectives, contents, methods and participation. <i>Norwegian Journal of Epidemiology</i> 13, 19-32 (2003).
Inter99	A population-based primary prevention study on cardiovascular disease and type 2 diabetes	Population-based	6,514	N/A	1. Missing phenotypes 2. Missing DNA	6,510	measured	[PMID: 14663300] Jørgensen T, Borch-Johnsen K, Thomsen TF, Ibsen H, Glümer C, Pisinger C. A randomized non-pharmacological intervention study for prevention of ischaemic heart disease: baseline results Inter99. <i>Eur J Cardiovasc Prev Rehabil.</i> 2003 Oct;10(5):377-86.
KORA2	Cooperative Health Research in the Region of Augsburg, KOoperative Gesundheitsforschung in der Region Augsburg	Population-based	3692	N/A	1. Missing phenotypes 2. Missing DNA	3691	measured	[PMID 20031538] Heid IM, Boes E, Müller M, Kollerits B et al. Genome-wide association analysis of high-density lipoprotein cholesterol in the population-based KORA study sheds new light on intergenic regions. <i>Circ Cardiovasc Genet.</i> 2008; 1(1):10-20.

Supplementary Note - Table 4

Study		Study design	Total sample size (N)	Call rate*	Sample QC	Samples in analyses (N)†	Anthropometric assessment method	References
Short name	Full name				Study specific QC criteria			
KORA3	Cooperative Health Research in the Region of Augsburg, KOoperative Gesundheitsforschung in der Region Augsburg	Population-based	1498	N/A	1. Missing phenotypes 2. Missing DNA	1324	measured	[PMID 16032514] Wichmann HE, Gieger C, Illig T; MONICA/KORA Study Group. KORA-gen--resource for population genetics, controls and a broad spectrum of disease phenotypes. Gesundheitswesen. 2005 Aug;67 Suppl 1:S26-30.
KORA4	Cooperative Health Research in the Region of Augsburg, KOoperative Gesundheitsforschung in der Region Augsburg	Population-based	1502	N/A	1. Missing phenotypes 2. Missing DNA	1502	measured	[PMID 16032514] Wichmann HE, Gieger C, Illig T; MONICA/KORA Study Group. KORA-gen--resource for population genetics, controls and a broad spectrum of disease phenotypes. Gesundheitswesen. 2005 Aug;67 Suppl 1:S26-30.
METSIM	Metabolic Syndrome in Men	Population-based	8182	N/A	Missing phenotypes	7127	measured	[PMID: 19223598] Stancakova A. et al. Changes in insulin sensitivity and insulin release in relation to glycemia and glucose tolerance in 6414 Finnish men. Diabetes 58, 1212-21 (2009).
OBB	Oxford Biobank	Population-based	1,469	N/A	1. Missing phenotypes 2. Missing DNA	1469	measured	Tan GD, Neville MJ, Liverani E, Humphreys SM, Currie JM, Dennis L, Fielding BA, Karpe F. The in vivo effects of the Pro12Ala PPARgamma2 polymorphism on adipose tissue NEFA metabolism: the first use of the Oxford Biobank. Diabetologia. 2006;49:158-68

*Sample genotyping success rate; i.e. percentage of successfully genotyped SNPs per sample

**Includes samples from Health 2000, Dehko2D 2004, Finrisk 1987 and 2002, Savitaipale Diabetes Study and Action LADA

***Includes samples from Health 2000, Dehko2D 2004, Finrisk 1987 and 2002, and Savitaipale Diabetes Study

†Ns reported are numbers of individuals association data was provided for; Ns in paper may vary due to effective sample size used in meta-analyses

Supplementary Note

Supplementary Note - Table 5 Information on genotyping methods, quality control of SNPs, imputation, and statistical analysis for stage 2 *in silico* and *de novo* replication studies.

Supplementary Note - Table 5

Supplementary Note - Table 5 -- Information on genotyping methods, quality control of SNPs, imputation, and statistical analysis for stage 2 in silico and de novo replication studies.

Study	Platform	Genotyping				SNPs that met QC criteria	Imputation software	Imputation		Association analyses	
		Genotype calling algorithm	MAF	Inclusion criteria Call rate*	p for HWE			MAF	Inclusion criteria Imputation quality**	SNPs in meta-analysis	Analysis software
In silico follow-up studies											
BHS	Illumina Human 610-Quad	Beadstudio	≥ 1%	≥ 95%	$> 10^{-6}$	549,284	MACH	≥1%	proper-info ≥ 0.30	1,977,680	R
Corogene	Illumina BeadChip Human 610-Quad	Illuminus	≥ 1%	≥ 95%	$> 10^{-6}$	554,988	MACH	≥1%	none	2,543,887	PLINK
EGCUT	Illumina Beadarray Human370CNV	Beadstudio	≥ 1%	≥ 98%	$> 10^{-6}$	316,924	IMPUTE	≥ 1%	proper-info ≥ 0.30	2,345,812	SNPtest
FHS	Illumina 1Million GeneChip	BeadStudio	≥1%	≥ 98%	$> 10^{-6}$	863,024	MACH-1.0.16	≥1%	r2-hat ≥ 0.30	2,453,887	SAS
FinGesture cases	Affymetrix Genome-Wide Human SNP Array 6.0	Birdseed	≥ 5%	≥ 95%	$> 10^{-6}$	606,717	MACH	0%	r2-hat ≥ 0.30	665	MACH2QTL
GOOD	Illumina Infinium HumanHap 610K	BeadStudio	≥ 1%	≥ 98%	$> 10^{-6}$	521,160	MACH	none	none	2,543,887	MACH2QTL
HBCS	Illumina custom made BeadChip Human 670-Quad	Illuminus	≥ 1%	≥ 95%	$> 10^{-6}$	533491	MACH	≥1%	none	2,543,887	PLINK
HYPERGENES - cases	Illumina Human1M-Duov3_B	GenCall, Beadstudio	≥ 1%	≥ 90%	$> 10^{-7}$	Center I: 861759, Center II: 872576	MACH	none	none	2,543,887	Matlab
HYPERGENES - controls	Illumina Human1M-Duov3_B	GenCall, Beadstudio	≥ 1%	≥ 90%	$> 10^{-7}$	Center I: 861759, Center II: 872576	MACH	none	none	2,543,887	Matlab
MGS	Affymetrix Genome-Wide Human SNP Array 6.0	Birdsuite 2.0	≥ 1%	≥ 95%	$> 10^{-6}$	696,492	MACH 1.0	≥1%	r2-hat ≥ 0.30	696,492	PLINK and local software
NHS	Affymetrix Genome-Wide Human 6.0 array	Birdseed calling algorithm v2	≥ 2%	≥ 98%	$> 10^{-4}$	704,409	MACH	≥ 2%	r2-hat ≥ 0.30	2,543,887	ProbABEL
RS-II	Illumina / HumanHap 550 V.3 DUO; Illumina / HumanHap 610 QUAD	Genomestudio	≥ 1%	≥ 97.5%	$> 10^{-6}$	466,389	MACH	≥1%	(O/E)σ2 ratio ≥ 0.1	667	MACH2QTL
Sorbs	500K Affymetrix GeneChip and Affymetrix Genome-Wide Human SNP Array 6.0	Genecall	≥ 1%	≥ 95%	$> 10^{-4}$	378,513	MACH V.1.000.16	>1%	proper-info ≥ 0.40	2,352,557	SNPTEST
WGHS	Illumina HumanHap300 Duo "+"	BRLMM (500K), Birdseed (6.0)	NA	≥ 90%	$> 10^{-6}$	339,596	IMPUTE	NA	none	2,608,508	R
YFS	Illumina custom made BeadChip Human 670-Quad	Beadstudio v 3.3	≥ 1%	≥ 95%	$> 10^{-6}$	546,674	MACH	≥1%	none	2,543,887	PLINK
De novo follow-up studies											
Study	Platform	Call rate	Concordance rate for duplicates	p for HWE	Analysis software						
B58C-WTCCC & B58C replication set (R58)	Custom TaqMan® SNP Genotyping Assays	≥ 90%	NA	$> 10^{-3}$	SNPTEST						
BCG	KASPAR (Kbiosciences)	> 90%	-	$> 10^{-3}$	STATA 10						

Supplementary Note - Table 5

Study	Platform	Call rate	Concordance rate for duplicates	p for HWE	Analysis software
BPPP	iPLEX™ Sequenom MassARRAY®	≥ 95%	≥ 99%	> 10 ⁻⁶	PLINK
BWHHS	KASPAR (Kbiosciences)	> 90%	-	> 10 ⁻³	STATA 10
DESIR	SNPkex, Applied biosystems, Foster City, CA, USA	≥ 95%	none	≥ 10 ⁻⁶	R
DIAGEN	Sequenom MassARRAY® system ((Sequenom, San Diego, USA)	≥ 88%	≥ 98.7%	> .005	Merlin
EGCUT	Custom TaqMan® SNP Genotyping Assays	≥ 90%	NA	> 10 ⁻³	PLINK
EPIC-Norfolk	1. Custom TaqMan® SNP Genotyping Assays 2. Sequenom MassARRAY® system	≥ 95%	≥ 97%	> 10 ⁻⁶	SAS
EPIC-Potsdam	KASPar assay system (KBiosciences, Herts, UK)	≥ 95%	not tested	> 0.01	SAS
Finrisk	iPLEX™ Sequenom MassARRAY®	≥ 95%	≥ 99%	> 10 ⁻⁶	PLINK
FUSION STAGE2 T2D cases	Sequenom MassARRAY® system	≥ 94%	100%	>.03	Merlin
FUSION STAGE2 controls	Sequenom MassARRAY® system	≥ 94%	100%	>.03	Merlin
HUNT	Sequenom MassARRAY® system	≥ 92%	100%	> .005	Merlin
Inter99	Kbioscience allele-specific PCR (KASPar)	≥ 95%	≥ 99%	> 10 ⁻⁶	R
KORA2	iPLEX™ Sequenom MassARRAY®	≥ 90%	-	≥ 0.001	R
KORA3	iPLEX™ Sequenom MassARRAY®	≥ 90%	-	≥ 0.001	R
KORA4	iPLEX™ Sequenom MassARRAY®	≥ 90%	-	≥ 0.001	R
METSIM	Sequenom MassARRAY® system	≥ 94%	≥ 99.5%	> .005	Merlin
OBB	Custom TaqMan® SNP Genotyping Assays	≥ 90%	NA	> 10 ⁻³	SNPTEST

* Sample genotyping success rate; i.e. percentage of successfully genotyped SNPs per sample

** SNPtest calculates the 'proper_info' statistic as a measure of the relative statistical information about the additive genetic effect being estimated. The proper_info statistic has a direct relationship to the power of the test and ranges from 0 to 1, with 1 indicating perfect information. MACH calculates the 'rsq_hat', which is the r2 between each imputed genotype and its true underlying genotype. Rsq_hat ranges from 0 to 1, with 1 indicating perfect imputation

Supplementary Note

Supplementary Note - Table 6 Study-specific descriptive statistics for stage 2 *in silico* and *de novo* replication studies.

Supplementary Note - Table 6 -- Study-specific descriptive statistics for stage 2 in silico and de novo replication studies

Study	Trait	Men						Women					
		n	mean	SD	median	min	max	n	mean	SD	median	min	max
<i>In silico</i> follow-up studies													
BHS*	Age (yrs)	558	53.47	17.15	53.65	17.60	91.40	769	53.72	17.08	53.10	17.30	90.50
	BMI (kg/m ²)	558	26.62	3.57	26.25	15.77	40.12	769	25.49	4.42	24.66	16.82	40.77
	Weight (kg)	558	81.80	12.31	80.25	46.40	127.00	769	67.06	12.02	65.00	34.80	109.00
	Height (m)	558	1.75	0.07	1.75	1.53	1.99	769	1.62	0.06	1.62	1.35	1.90
Corogene	Age (yrs)	2266	59.66	12.83	61.00	25.00	92.00	1490	62.61	13.47	65.00	25.00	94.00
	BMI (kg/m ²)	2265	27.39	4.23	26.79	15.95	54.88	1491	26.87	5.21	26.07	13.63	57.68
	Weight (kg)	2265	85.00	14.42	83.50	44.00	170.00	1491	70.14	13.88	68.30	36.00	144.00
	Height (m)	2267	1.76	0.07	1.76	1.34	2.03	1491	1.62	0.07	1.62	1.05	1.85
EGCUT	Age (yrs)	135	40.93	17.81	36.50	18.00	80.00	210	41.03	16.46	39.00	18.00	87.00
	BMI (kg/m ²)	135	26.03	4.95	25.11	17.30	43.65	210	25.63	6.09	24.02	17.00	48.24
	Weight (kg)	135	83.68	16.41	80.50	50.00	143.00	210	70.46	16.22	66.50	40.00	136.00
	Height (m)	135	1.79	0.07	1.80	1.58	2.04	210	1.66	0.07	1.66	1.44	1.84
FHS	Age (yrs)	662	48.20	13.70	46.30	25.60	85.70	880	47.50	13.00	45.00	25.70	85.80
	BMI (kg/m ²)	632	27.80	4.30	27.20	18.40	46.20	831	27.10	6.10	26.10	16.50	55.00
	Weight (kg)	632	87.10	14.60	85.30	55.30	140.60	831	72.30	16.60	68.90	41.70	144.20
	Height (m)	632	1.77	0.07	1.77	1.57	2.03	831	1.63	0.06	1.63	1.41	1.96
FinGesture cases	Age (yrs)	745	61.19	10.58	62.00	34.00	85.00	198	67.44	10.33	68.00	31.00	85.00
	BMI (kg/m ²)	739	27.22	3.93	27.02	16.20	44.80	196	28.14	5.17	27.98	16.67	46.09
	Weight (kg)	743	82.32	14.09	81.00	42.00	150.00	197	71.91	14.06	71.60	37.50	112.00
	Height (m)	745	1.74	0.07	1.74	1.55	1.97	198	1.60	0.06	1.60	1.46	1.76
GOOD	Age (yrs)	938	18.90	0.60	18.80	18.00	20.10						
	BMI (kg/m ²)	938	22.40	3.20	21.90	16.10	41.60						
	Weight (kg)	938	73.90	11.60	72.00	51.30	127.00						
	Height (m)	938	1.82	0.07	1.82	1.61	2.03						
HBCS	Age (yrs)	737	61.41	2.75	60.80	57.00	69.30	991	61.55	3.05	60.90	56.70	69.80
	BMI (kg/m ²)	736	27.56	4.30	27.01	18.75	68.39	990	27.75	5.06	26.98	14.79	50.10
	Weight (kg)	737	86.33	14.51	84.50	56.20	213.30	990	73.90	13.89	71.70	37.30	133.80
	Height (m)	736	1.77	0.06	1.77	1.59	1.97	990	1.63	0.06	1.63	1.46	1.83
HYPERGENES - cases*	Age (yrs)	1190	49.41	10.41	50.00	17.63	84.00	598	48.47	9.58	49.00	18.38	93.00
	BMI (kg/m ²)	1190	27.42	3.52	27.13	16.00	47.43	598	26.85	4.86	26.21	17.45	46.32
	Weight (kg)	1190	81.35	12.08	80.00	49.00	139.50	598	68.42	13.09	67.00	44.00	114.50
	Height (m)	1190	1.72	0.07	1.72	1.48	2.11	598	1.60	0.07	1.60	1.30	1.97
HYPERGENES - controls	Age (yrs)	1072	62.27	10.71	59.81	28.00	98.00	766	64.30	11.28	61.00	44.93	113.00
	BMI (kg/m ²)	1072	25.95	3.27	25.59	10.15	40.77	766	24.98	3.73	24.60	16.53	41.35
	Weight (kg)	1072	76.10	10.59	75.00	29.00	118.00	766	64.25	10.13	63.00	41.00	110.00
	Height (m)	1072	1.71	0.07	1.70	1.50	1.96	766	1.60	0.06	1.60	1.40	1.81
MGS	Age (yrs)	1,247	52.67	16.01	52.00	18.00	90.00	1,350	48.48	16.29	48.00	18.00	90.00
	BMI (kg/m ²)	1,247	30.85	6.45	29.84	15.83	72.56	1,350	31.92	8.55	30.32	16.34	69.09
	Weight (kg)	1,247	98.77	22.67	95.25	53.98	249.48	1,350	86.13	24.22	81.65	47.63	201.85
	Height (m)	1247	1.79	0.07	1.78	1.58	2.06	1350	1.64	0.07	1.65	1.35	2.01
NHS	Age (yrs)							2988	53.40	6.74	54.00	39.00	65.00
	BMI (kg/m ²)							2988	27.13	5.63	26.00	17.01	54.87
	Weight (kg)							2988	160.87	35.21	155.00	90.00	340.00
	Height (m)							2988	1.64	0.06	1.63	1.35	1.83
RS-II	Age (yrs)	973	64.48	7.59	61.89	55.14	93.95	1156	65.04	8.33	62.03	55.12	95.33
	BMI (kg/m ²)	971	26.92	3.36	26.72	16.78	40.52	1151	27.52	4.45	26.89	16.66	50.12
	Weight (kg)	972	83.32	11.58	82.20	54.00	126.80	1151	72.77	12.74	71.10	36.20	150.00
	Height (m)	971	1.76	0.06	1.75	1.56	2.03	1153	1.63	0.06	1.62	1.41	1.89
Sorbs	Age (yrs)	371	48.10	16.70	48.10	18.10	82.10	536	48.00	15.90	48.60	18.00	88.40
	BMI (kg/m ²)	371	27.20	4.00	26.80	19.00	43.90	536	26.90	5.50	26.20	15.40	47.40
	Weight (kg)	371	85.40	12.70	84.00	58.00	139.00	536	72.10	14.00	70.00	43.00	126.00
	Height (m)	371	1.77	0.07	1.77	1.58	1.95	536	1.64	0.07	1.64	1.44	1.82
WGHS	Age (yrs)							22888	54.69	7.11	52.90	38.71	89.89
	BMI (kg/m ²)							22888	25.91	4.96	24.89	14.23	59.58
	Weight (kg)							22888	70.03	14.15	68.04	38.56	175.09
	Height (m)							22888	1.64	0.06	1.65	1.30	2.01
YFS	Age (yrs)	1123	37.55	5.06	39.00	30.00	45.00	1320	37.57	5.01	39.00	30.00	45.00
	BMI (kg/m ²)	908	26.76	4.29	26.11	17.54	49.35	1081	25.32	5.03	24.34	16.56	58.82
	Weight (kg)	908	86.56	15.65	85.00	54.00	166.00	1083	69.82	14.55	67.00	42.00	166.00
	Height (m)	911	1.80	0.07	1.80	1.57	2.03	1084	1.66	0.06	1.66	1.45	1.89
<i>De novo</i> follow-up studies													
B58C-WTCCC & B58C replication set (R58)	Age (yrs)	683	45.30	0.36	45.42	44.25	46.00	657	45.33	0.34	45.42	44.25	46.00
	BMI (kg/m ²)	683	27.26	4.11	26.69	18.06	43.05	657	27.64	5.47	26.50	14.34	41.42
	Weight (kg)	683	85.54	14.14	84.00	55.60	145.00	657	71.97	15.10	69.30	40.00	127.00
	Height (m)	683	1.76	0.07	1.76	1.58	1.98	657	1.63	0.06	1.63	1.42	1.83
BCG	Age (yrs)	361	25.02	0.76	25.00	23.17	27.25	318	25.01	0.76	25.00	23.08	26.75
	BMI (kg/m ²)	359	25.08	3.91	24.76	15.84	41.78	314	25.23	5.35	23.83	16.97	47.85
	Weight (kg)	360	78.97	13.12	78.00	45.00	127.00	316	67.66	14.52	64.25	42.00	141.00
	Height (m)	359	1.77	0.06	1.77	1.53	1.96	315	1.64	0.06	1.64	1.46	1.80
BPPP	Age (yrs)	1633	49.26	15.95	51.07	19.06	76.39	1813	49.14	16.03	51.25	18.76	76.80
	BMI (kg/m ²)	1646	23.70	3.43	23.37	13.97	43.95	1825	21.29	3.84	20.79	13.39	49.85

Supplementary Note - Table 6

Study	Trait	Men						Women					
		n	mean	SD	median	min	max	n	mean	SD	median	min	max
BWHHS	Weight (kg)	1646	83.98	13.09	82.95	47.50	168.10	1825	69.80	13.00	68.20	43.00	165.50
	Height (m)	1646	1.77	0.07	1.77	1.58	2.00	1825	1.64	0.06	1.64	1.42	1.87
	Age (yrs)							4269	68.89	5.51	69.00	59.00	80.00
	BMI (kg/m ²)							3941	27.61	5.01	26.86	15.25	58.83
	Weight (kg)							3947	69.57	13.05	67.90	37.40	145.10
DESIR	Height (m)							3942	1.59	0.06	1.59	1.35	2.00
	Age (yrs)	2253	47.28	10.05	46.57	30.07	65.82	2283	47.33	10.03	46.91	30.13	65.50
	BMI (kg/m ²)	2253	25.46	3.36	25.10	16.70	45.40	2281	23.97	4.11	23.20	15.40	53.60
	Weight (kg)	2253	75.91	10.93	75.00	46.00	140.00	2281	61.03	10.52	59.00	37.00	127.00
	Height (m)	2253	1.73	0.07	1.72	1.50	1.96	2281	1.60	0.06	1.60	1.41	1.83
DIAGEN	Age (yrs)	830	60.96	13.47	60.00	22.00	96.00	991	62.05	15.00	61.00	18.00	97.00
	BMI (kg/m ²)	830	27.66	4.00	27.05	18.94	44.64	991	27.99	6.00	26.98	15.63	61.71
	Weight (kg)	830	84.73	13.96	83.00	50.00	137.00	991	74.00	16.98	71.00	40.00	182.00
	Height (m)	830	1.75	0.07	1.75	1.50	1.96	991	1.62	0.07	1.62	1.41	1.92
EGCUT	Age (yrs)	1891	42.97	18.43	41.00	18.00	101.00	1888	44.77	17.45	43.50	18.00	101.00
	BMI (kg/m ²)	1891	26.29	4.61	25.68	15.90	55.50	1888	26.18	5.60	25.12	11.51	53.15
	Weight (kg)	1891	83.75	15.37	82.00	46.00	168.00	1888	71.33	15.24	69.00	28.00	150.00
	Height (m)	1891	1.79	0.07	1.78	1.52	2.03	1888	1.65	0.06	1.65	1.40	1.90
EPIC-Norfolk	Age (yrs)	9084	59.08	9.30	59.00	39.00	79.00	9400	58.57	9.34	58.00	39.00	77.00
	BMI (kg/m ²)	9084	26.49	3.28	26.21	16.13	49.06	9400	26.15	4.23	25.49	15.23	58.69
	Weight (kg)	9084	80.31	11.40	79.40	44.60	160.00	9400	67.75	11.64	66.00	36.00	139.20
	Height (m)	9084	1.74	0.07	1.74	1.39	2.00	9400	1.61	0.06	1.61	1.29	2.00
EPIC-Potsdam	Age (yrs)	1981	51.80	8.10	52.00	24.00	69.00	3015	48.60	9.20	48.00	20.00	66.00
	BMI (kg/m ²)	1981	26.90	3.60	26.60	16.40	55.40	3015	25.80	4.80	24.90	17.20	58.70
	Weight (kg)	1981	82.40	11.90	81.20	48.00	162.00	3015	68.60	13.00	66.40	44.20	149.70
	Height (m)	1981	1.75	0.07	1.75	1.51	2.00	3015	1.63	0.06	1.63	1.41	1.88
FINRISK	Age (yrs)	3902	49.92	13.57	50.00	25.00	74.00	3916	47.40	12.85	47.00	25.00	74.00
	BMI (kg/m ²)	3902	27.04	3.97	26.58	14.70	48.39	3916	26.31	5.01	25.45	14.69	53.28
	Weight (kg)	3902	83.10	13.12	81.60	40.00	150.00	3916	69.27	13.17	67.15	37.10	137.80
	Height (m)	3902	1.75	0.07	1.76	1.34	2.04	3916	1.62	0.06	1.62	1.39	1.92
FUSION stage 2 - cases	Age (yrs)	728	58.28	9.04	59.00	28.00	82.00	498	61.19	7.98	61.00	34.36	89.00
	BMI (kg/m ²)	728	30.20	5.03	29.70	19.40	52.97	498	31.60	5.79	30.86	17.85	53.45
	Weight (kg)	728	92.48	17.20	90.00	42.20	167.00	498	81.23	16.10	78.90	48.00	143.50
	Height (m)	728	1.75	0.07	1.75	1.39	1.96	498	1.60	0.06	1.60	1.41	1.80
FUSION stage 2 - controls	Age (yrs)	1863	56.45	8.14	56.10	41.30	75.10	1833	57.23	8.31	56.80	40.99	75.20
	BMI (kg/m ²)	1863	26.81	3.94	26.40	15.78	93.00	1833	27.02	4.80	26.20	17.21	57.20
	Weight (kg)	1863	82.27	12.92	81.00	46.50	146.60	1833	70.70	12.86	68.80	42.80	153.80
	Height (m)	1863	1.75	0.06	1.75	1.53	1.95	1833	1.62	0.06	1.62	1.32	1.81
HUNT	Age (yrs)	2387	63.64	14.44	67.00	20.00	97.00	2477	66.52	14.21	71.00	20.00	92.00
	BMI (kg/m ²)	2387	27.02	3.64	26.60	16.30	45.60	2477	28.15	5.09	27.60	14.70	48.50
	Weight (kg)	2387	82.51	12.94	81.50	43.40	139.00	2477	72.77	13.88	71.50	31.00	135.00
	Height (m)	2387	1.75	0.07	1.75	1.49	1.98	2477	1.61	0.06	1.61	1.37	1.84
Inter99	Age (yrs)	3168	46.56	7.85	45.17	29.92	61.12	3342	45.82	8.00	45.06	29.69	61.35
	BMI (kg/m ²)	3168	26.82	4.02	26.33	16.73	56.90	3342	25.82	5.10	24.68	14.93	55.75
	Weight (kg)	3168	85.73	14.11	84.00	53.00	183.00	3342	71.10	14.70	68.50	36.80	159.30
	Height (cm)	3168	1.79	0.07	1.78	1.54	2.07	3342	1.65	0.06	1.66	1.29	1.88
KORA 2	Age (yrs)	1867	49.56	14.17	50.00	25.00	74.00	1824	48.70	13.88	49.00	25.00	74.00
	BMI (kg/m ²)	1852	27.06	3.60	26.84	17.54	46.38	1796	26.18	4.77	25.39	16.81	50.50
	Weight (kg)	1852	81.71	12.00	80.20	45.20	150.00	1796	67.87	12.09	66.20	37.50	122.90
	Height (m)	1857	1.74	0.07	1.74	1.33	1.98	1817	1.61	0.07	1.61	1.34	1.88
KORA 3	Age (yrs)	631	41.39	13.72	37.00	25.00	74.00	693	41.15	12.88	38.00	25.00	74.00
	BMI (kg/m ²)	626	26.59	3.43	26.21	17.59	40.99	678	25.26	4.74	24.17	16.89	49.31
	Weight (kg)	626	82.42	11.52	81.20	52.30	132.00	678	66.83	12.26	64.20	43.60	125.20
	Height (m)	630	1.76	0.07	1.76	1.55	2.00	690	1.63	0.06	1.63	1.42	1.80
KORA 4	Age (yrs)	714	43.51	15.00	38.00	25.00	74.00	788	42.19	13.96	37.00	25.00	74.00
	BMI (kg/m ²)	711	26.80	3.61	26.44	16.32	42.37	775	26.01	5.05	25.33	15.84	49.15
	Weight (kg)	711	83.12	11.64	82.40	52.30	136.50	775	68.97	13.32	67.10	41.10	137.00
	Height (m)	714	1.76	0.07	1.76	1.55	1.96	783	1.63	0.07	1.63	1.39	1.89
METSIM	Age (yrs)	7127	57.60	6.90	57.00	45.00	73.00						
	BMI (kg/m ²)	7127	27.25	4.16	26.67	16.18	55.54						
	Weight (kg)	7127	84.33	13.98	82.50	43.00	174.00						
	Height (m)	7127	1.76	0.06	1.76	1.47	2.03						
OBB	Age (yrs)	745	41.10	5.50	42.00	29.00	53.00	724	41.30	6.30	41.00	30.00	53.00
	BMI (kg/m ²)	745	26.70	3.90	26.20	15.30	46.20	724	26.70	3.90	26.20	15.30	46.20
	Weight (kg)	745	84.80	13.80	83.10	44.70	172.10	724	69.20	13.10	66.50	43.50	127.90
	Height (m)	745	1.78	0.07	1.78	1.50	2.03	724	1.65	0.07	1.65	0.83	1.82

*Includes individuals <18 years of age (BHS: n=3 (17.3, 17.6, and 17.6 years); HYPERGENES - cases: n=2 (17.6 and 17.8 years))

Supplementary Note

Supplementary Note - Table 7 Study design, number of individuals and sample quality control for studies used in extreme obesity case-control analyses, childhood BMI analyses, family-based analyses and polygenes analyses.

Supplementary Note - Table 7 -- Study design, number of individuals and sample quality control for studies used in extreme obesity case-control analyses, childhood BMI analyses, family-based analyses and polygenes analyses.

Study		Study design	Total sample size (N)	Call rate*	Sample QC	Samples in analyses (N)**	Anthropometric assessment method	References	
Short name	Full name				Study specific QC criteria				
<i>In silico</i> follow-up studies									
Essen Obesity Study (Essen Case-Control GWAS)	--	Case-Control	children and adolescents	888	≥ 95%	1) related individuals and duplicates; 2) Missing BMI and age.	888	measured	[PMID: 18159244] Hinney, A. et al. Genome wide association (GWA) study for early onset extreme obesity supports the role of fat mass and obesity associated gene (FTO) variants. PLoS One 26,2(12):e1361 (2007).
Essen Obesity Study (Essen Obesity Trio GWAS)	--	Family-based (TDT)	trios with obese children and adolescents	1,334	≥ 95%	1) Mendelian inconsistencies per family >5%; 2) duplicates; 3) Missing BMI and age in offspring.	1,272 (848 parents, 424 children and adolescents)	measured	[PMID: 12970296] Hinney A, Hohmann S, Geller F, Vogel C, Hess C, Wermter AK, Brokamp B, Goldschmidt H, Siegfried W, Remschmidt H, Schäfer H, Gudermann T, Hebebrand J. Melanocortin-4 receptor gene: case-control study and transmission disequilibrium test confirm that functionally relevant mutations are compatible with a major gene effect for extreme obesity. J Clin Endocrinol Metab.; 88(9):4258-67 (2003).
French obese CC- adults	French obese CC- adults	Case control study	adults	1,473	≥ 95%	1. ethnic outliers 2. related individuals	1,380	measured	[PMID: 19151714] Meyre D et al. Genome-wide association study for early-onset and morbid adult obesity identifies three new risk loci in European populations. Nat Genet.;41(2):139-40 (2009).
French obese CC - youth	French obese CC - young	Case control study	children and adolescents	1,396	≥ 95%	1. ethnic outliers 2. related individuals	1301	measured	[PMID: 19151714] Meyre D et al. Genome-wide association study for early-onset and morbid adult obesity identifies three new risk loci in European populations. Nat Genet.;41(2):139-40 (2009).
GINI/LISA	German Infant Nutritional Intervention / Influences of Lifestyle related Factors on the Immune System and the Development of Allergies in Childhood	Population-based	children	396	≥ 95%	1) Missing body weight and height 2) check for related individuals and duplicates	355	medical records	[PMID: 16452359] Zutavern A, Brockow I, Schaaf B, Bolte G, von Berg A, Diez U, Borte M, Herbarth O, Wichmann HE, Heinrich J. Timing of solid food introduction in relation to atopic dermatitis and atopic sensitization: results from a prospective birth cohort study. Pediatrics 2006; 117:401-411. [PMID: 19521784] Rzehak P, Sausenthaler S, Koletzko S, Bauer CP, Schaaf B, von Berg A et al. Period-specific growth, overweight and modification by breastfeeding in the GINI and LISA birth cohorts up to age 6 years. Eur J Epidemiol; 24:449-67 (2009).
SCOOP-UK	Severe Childhood Onset Obesity Project UK	Case series	children and adolescents	1,012	≥ 95%	1) heterozygosity <26% or >28%; 2) discrepancy with external identifying information; 3) ethnic outliers; 4) related individuals and duplicates	943	measured	[PMID: 19966786] Bochukova EG et al. Large, rare chromosomal deletions associated with severe early-onset obesity. Nature 463, 666-70 (2010)
QIMR	Twin study at Queensland Institute of Medical Research	Population-based	Adults (Polygenes)	3,038	≥ 95%	1) close relatives based on pedigree information; 2) ethnic outliers; 3) Missing BMI.	1,787	measured or self-report	[PMID: 19896111] Medland et al. Common Variants in the Trichohyalin Gene Are Associated with Straight Hair in Europeans. Am. J. Human Genet. 85(5):750-5 (2009).
<i>De novo</i> follow-up studies									
Berlin school girls (BESCS)/Berlin obese (BEPOC)	Berlin School Children's Study/Berlin Paediatric Obese Cohort	Population-based (enriched for overweight)	children and adolescents	2,785	N/A	1. Missing phenotypes 2. Missing DNA	2,723	measured	[PMID: 18974233.] Bau, A.M. et al. Is there a further acceleration in the age at onset of menarche? A cross-sectional study in 1840 school children focusing on age and bodyweight at the onset of menarche. Eur J Endocrinol. 160(1):107-113. (2009)

Supplementary Note - Table 7

Study		Study design		Total sample size (N)	Call rate*	Sample QC Study specific QC criteria	Samples in analyses (N)**	Anthropometric assessment method	References
Short name	Full name								
Datteln (DAPOC)	Datteln Paediatric Obese Cohort	Overweight and obese	children and adolescents	736	N/A	1. Missing phenotypes 2. Missing DNA	692	measured	[PMID: 17517246] Reinehr T, Hinney A, de Sousa G, Austrup F, Hebebrand J, Andler W. Definable somatic disorders in overweight children and adolescents. <i>J Pediatr.</i> ; 150(6):618-22, 622.e1-5.T (2007).
Essen/Marburg - obese adults	--	Obese	adults	1,411	N/A	1. Missing phenotypes 2. Missing DNA	1,024	measured	[PMID: 16492696] Hinney, A. et al. Prevalence, spectrum, and functional characterization of melanocortin-4 receptor gene mutations in a representative population-based sample and obese adults from Germany. <i>J Clin Endocrinol Metab</i> 91, 1761-1769 (2006).
Essen Obesity Trios (independent of GWAS)	--	Family-based (TDT)	trios with obese children and adolescents	1,101 (748 parents and 353 children and adolescents)	N/A	1. Missing phenotypes 2. Missing DNA, 3. Incomplete trio	942 (628 parents, 314 children and adolescents)	measured	[PMID: 18398438] Wermter, A.K. et al. Preferential reciprocal transfer of paternal/maternal DLK1 alleles to obese children: first evidence of polar overdominance in humans. <i>Eur J Hum Genet</i> 16(9), 1126-1134 (2008).
GINI/LISA	German Infant Nutritional Intervention / Influences of Lifestyle related Factors on the Immune System and the Development of Allergies in Childhood	Population-based	children and adolescents	2,151	N/A	1. Missing phenotypes 2. Missing DNA	1,969	measured	[PMID: 17889067] Filipiak, B. et al. Solid food introduction in relation to eczema: results from a four-year prospective birth cohort study. <i>J Pediatr</i> 151, 352-358 (2007) [PMID 17399781] Chen, C.M. et al. Longitudinal study on cat allergen exposure and the development of allergy in young children. <i>J Allergy Clin Immunol</i> 119, 1148-1155 (2007).
NFBC-1986	Northern Finland Birth Cohorts born in 1985-1986	Population-based	16 year olds	9,479	N/A	1. Missing DNA 2. Missing phenotypes 3. Non Caucasians 4. Related	5,370	measured	[PMID: 7485065] Olsen P, Laara E, Rantakallio P, Jarvelin MR, Sarpola A, Hartikainen AL. Epidemiology of preterm delivery in two birth cohorts with an interval of 20 years. <i>Am J Epidemiol</i> ;142(11):1184-93, (1995).
USCS	Ulm School Children Study	Population-based	children and adolescents	1,478	N/A	1. Missing phenotypes 2. Missing DNA	924	measured	[PMID: 19562371] Nagel, G. et al. Determinants of obesity in the Ulm Research on Metabolism, Exercise and Lifestyle in Children (URMEL-ICE). <i>Eur J Pediatr</i> 168(10):1259-1267 (2009).

*Sample genotyping success rate; i.e. percentage of successfully genotyped SNPs per sample

**Ns reported are numbers of individuals association data was provided for; Ns in paper may vary due to effective sample size used in meta-analyses

Supplementary Note

Supplementary Note - Table 8 Supplementary Note - Table 8 -- Information on genotyping methods, quality control of SNPs, imputation, and statistical analysis for studies used in extreme obesity case-control analyses, childhood BMI analyses, family-based analyses and polygenes analyses.

Supplementary Note - Table 8 -- Information on genotyping methods, quality control of SNPs, imputation, and statistical analysis for studies used in extreme obesity case-control analyses, childhood BMI analyses, family-based analyses and polygenes analyses.

Study	Platform	Genotyping					SNPs that met QC criteria	Imputation software	Imputation		Association analyses	
		Genotype calling algorithm	MAF	Inclusion criteria	Call rate*	p for HWE			MAF	Inclusion criteria	Imputation quality**	SNPs in meta-analysis
<i>In silico</i> follow-up studies												
Essen Obesity Study (Essen Case-Control GWAS)	Genome-Wide Human SNP Array 6.0 from Affymetrix	Birdseed	≥ 1%	≥ 95%	> 0.001	730,577	Beagle	≥1%	none	675	SNPtest	
Essen Obesity Study (Essen Obesity Trio GWAS)	Genome-Wide Human SNP Array 6.0 from Affymetrix	Birdseed	≥ 1%	≥ 95%	> 0.001	749,179	Beagle	≥1%	none	675	PLINK	
French obese CC- adults	Illumina Chip 370 duo	Illumina clustering	none	≥ 95%	> 10 ⁻⁴	112	IMPUTE	≥ 1%	proper-info ≥ 0.40	663	SNPtest	
French obese CC - young	Illumina Chip 370 duo	Illumina clustering	none	≥ 95%	> 10 ⁻⁴	114	IMPUTE	≥ 1%	proper-info ≥ 0.40	662	SNPtest	
GINA/LISA	Affymetrix Human SNP Array 5.0	BRLMM-P	0.01	0.95	0.01	371,004	IMPUTE	none	none	668	SNPtest	
SCOOP-UK (cases)	Affymetrix 6.0	Birdsuite versior	≥ 1%	≥ 95% if MAF ≥ 5%; ≥ 97% if 2% ≤ MAF < 5%; ≥ 99% if 1% ≤ MAF < 2%	> 10 ⁻⁴	722,714	IMPUTE	≥1%	proper-info ≥ 0.40	24	SNPtest	
QIMR	Illumina HumanHap 610 Quad	BeadStudio	≥ 1%	≥ 95%	> 10 ⁻⁶	515,961	MACH	≥ 1%	r2_hat ≥ 0.30			
<i>De novo</i> follow-up studies												
Study	Platform	Call rate*	Concordance rate for duplicates	p for HWE	Analysis software							
Berlin school girls (BESCS)/Berlin obese (BEPOC)	Sequenom MALDI-TOF MS; iPLEX™	≥ 90%	≥ 97%	> 10 ⁻⁶	SAS and R							
Datteln (DAPOC)	Sequenom MALDI-TOF MS; iPLEX™	≥ 95%	≥ 97%	> 10 ⁻⁶	SAS and R							
Essen/Marburg - obese adults	Sequenom MALDI-TOF MS; iPLEX™	≥ 95%	≥ 97%	> 10 ⁻⁶	SAS and R							
Essen Obesity Trios (independent of GWAS)	Sequenom MALDI-TOF MS; iPLEX™	≥ 95%	≥ 97%	> 10 ⁻⁶	SAS and R							
GINI/LISA	Sequenom MALDI-TOF MS; iPLEX™	≥ 95%	≥ 97%	> 10 ⁻⁶	SAS and R							
NFBC-1986	SNPlex	≥ 95%	none	≥ 10 ⁻⁶	R							
USCS	Sequenom MALDI-TOF MS; iPLEX™	≥ 95%	≥ 97%	> 10 ⁻⁶	SAS and R							

*Sample genotyping success rate; i.e. percentage of successfully genotyped SNPs per sample

**SNPtest calculates the 'proper_info' statistic as a measure of the relative statistical information about the additive genetic effect being estimated. The proper_info statistic has a direct relationship to the power of the test and ranges from 0 to 1, with 1 indicating perfect information. MACH calculates the 'rsq_hat', which is the r2 between each imputed genotype and its true underlying genotype. Rsq_hat ranges from 0 to 1, with 1 indicating perfect imputation.

Supplementary Note

Supplementary Note - Table 9 Study-specific descriptive statistics for studies used in extreme obesity case-control analyses, childhood BMI analyses, family-based analyses and polygenes analyses.

Supplementary Note - Table 9 -- Study-specific descriptive statistics for studies used in extreme obesity case-control analyses, childhood BMI analyses, family-based analyses and polygenes analyses.

Study	Trait	Men						Women					
		n	mean	SD	median	min	max	n	mean	SD	median	min	max
In silico follow-up studies													
Essen Obesity Study (Essen Case-Control GWAS) Cases	Age (yrs)	192	14.18	3.85	13.96	3.37	39.18	261	0.40	3.67	14.31	5.62	24.42
	BMI (kg/m ²)	192	33.11	6.48	32.34	21.88	56.86	261	1.00	6.84	32.33	20.35	61.88
	Weight (kg)	190	92.81	29.19	89.45	24.90	186.20	260	0.89	24.58	87.80	25.80	176.00
	Height (m)	190	1.66	0.15	1.68	1.00	1.94	260	0.47	0.13	1.63	1.12	1.82
Essen Obesity Study (Essen Case-Control GWAS) Controls	Age (yrs)	171	25.38	4.57	24.71	16.88	46.32	264	0.20	6.37	24.51	17.26	58.78
	BMI (kg/m ²)	171	18.86	0.94	18.95	16.29	21.80	264	1.00	0.95	17.69	13.22	19.78
	Weight (kg)	171	63.64	6.12	63.40	46.50	83.10	264	50.53	4.61	50.50	38.40	64.50
	Height (m)	171	1.84	0.07	1.83	1.64	2.04	264	1.69	0.07	1.74	1.50	1.89
Essen Obesity Study (Essen Obesity Trio GWAS) Parents	Age (yrs)	421	44.02	6.23	43.06	29.85	63.21	422	40.62	5.40	40.19	24.71	62.03
	BMI (kg/m ²)	424	30.22	5.36	29.61	18.04	54.33	424	30.58	7.36	29.33	17.15	64.34
	Weight (kg)	424	94.72	18.01	92.00	58.00	160.00	424	83.03	20.29	79.00	48.00	170.10
	Height (m)	424	1.77	0.07	1.77	1.54	1.99	424	1.64	0.07	1.64	1.00	1.80
Essen Obesity Study (Essen Obesity Trio GWAS) Offspring	Age (yrs)	200	13.00	2.72	13.46	5.14	19.47	224	13.38	2.86	13.52	4.96	20.54
	BMI (kg/m ²)	200	31.52	4.87	30.89	21.92	64.70	224	32.30	5.80	31.20	19.73	56.17
	Weight (kg)	198	85.05	24.24	84.85	31.00	214.30	223	83.91	21.88	83.60	26.00	174.00
	Height (m)	198	1.63	0.15	1.65	1.13	1.93	223	1.60	0.12	1.63	1.14	1.84
French obese (adults) Cases	Age (yrs)	143	44.90	11.64	46.00	16.00	75.00	521	43.87	12.07	44.00	15.00	77.00
	BMI (kg/m ²)	143	48.28	8.65	45.75	36.36	83.86	521	47.17	7.19	45.44	31.61	87.24
	Weight (kg)	143	149.45	29.20	142.50	99.00	270.00	521	123.82	21.63	120.00	74.00	231.80
	Height (m)	143	1.76	0.07	1.76	1.55	1.95	521	1.62	0.07	1.62	1.43	1.80
French obese (adults) Controls	Age (yrs)	178	53.10	5.59	52.65	45.04	64.96	538	49.52	8.53	49.80	30.25	65.02
	BMI (kg/m ²)	178	23.15	1.16	23.30	20.40	24.90	538	21.36	1.85	21.20	16.60	24.90
	Weight (kg)	178	69.47	6.49	70.00	51.00	89.00	538	54.84	5.86	55.00	40.00	71.00
	Height (m)	178	1.73	0.06	1.73	1.56	1.95	538	1.60	0.06	1.60	1.45	1.77
French obese (young) Cases	Age (yrs)	300	11.10	2.99	11.00	3.08	17.00	370	10.75	3.47	11.00	2.00	17.00
	BMI (kg/m ²)	300	29.70	6.24	28.27	19.20	69.36	370	29.40	6.65	28.15	18.67	54.36
	Weight (kg)	299	73.26	28.05	65.99	21.00	209.99	366	68.21	26.95	64.98	18.49	150.00
	Height (m)	299	1.54	0.16	1.53	1.00	1.97	366	1.49	0.17	1.53	0.92	1.83
French obese (young) Controls	Age (yrs)	309	11.89	2.37	11.83	6.08	17.08	322	11.93	2.21	11.92	5.92	17.17
	BMI (kg/m ²)	309	17.49	2.21	17.30	12.82	24.46	322	17.67	2.50	17.51	12.00	25.99
	Weight (kg)	309	40.44	12.34	37.60	19.60	79.20	322	40.07	10.59	39.40	19.20	69.60
	Height (m)	309	1.50	0.15	1.48	1.16	1.84	322	1.49	0.13	1.50	1.15	1.78
GINA/LISA	Age (yrs)	179	5.18	0.34	5.22	4.20	5.89	176	5.14	0.35	5.20	4.12	5.90
	BMI (kg/m ²)	179	15.16	1.34	15.15	11.78	20.83	176	15.30	1.37	15.14	11.96	22.84
	Weight (kg)	179	19.65	2.57	20.00	14.00	29.00	176	19.95	2.64	20.00	15.00	34.00
	Height (m)	179	1.14	0.05	1.14	0.99	1.25	176	1.14	0.04	1.14	1.02	1.24
SCOOP-UK cases	Age (yrs)	383	11.06	5.85	11.70	0.60	52.19	559	11.50	7.45	11.04	0.55	56.35
	BMI (kg/m ²)	380	34.23	8.91	32.73	21.06	89.62	551	34.17	9.59	32.40	17.07	96.38
	Weight (kg)	380	81.10	41.50	78.45	11.70	259.00	553	76.56	38.75	73.00	10.59	213.00
	Height (m)	380	147.97	25.75	153.40	70.40	199.60	552	143.69	23.80	150.40	67.50	192.90
QIMR	Age (yrs)	294	39.54	13.25	38.00	18.00	79.00	706	38.11	13.03	34.00	18.00	84.00
	Height (m)	294	1.79	0.07	1.79	1.60	2.01	706	1.64	0.07	1.64	1.44	1.93
	Weight (kg)	294	82.60	14.24	80.87	47.00	150.00	706	65.38	13.26	63.00	34.02	140.00
	BMI (kg/m ²)	294	25.78	4.26	25.25	14.83	55.03	706	24.36	4.61	23.42	14.17	45.11
De novo follow-up studies													
Berlin school girls (BESCS)/Berlin obese (BEPOC)	Age (yrs)	507	11.32	3.35	11.80	1.57	17.87	2216	12.19	2.35	12.71	0.81	17.61
	BMI (kg/m ²)	507	29.77	5.69	29.02	18.54	58.43	2216	21.95	5.84	20.34	12.50	55.37
	Weight (kg)	507	73.51	28.27	71.00	16.00	187.00	2216	52.87	18.19	49.00	16.00	164.00
	Height (m)	507	1.54	0.20	1.56	0.83	2.02	2216	1.55	0.12	1.57	1.02	1.90
Datteln (DAPOC)	Age (yrs)	305	10.65	2.60	10.90	0.90	17.70	387	10.75	2.84	10.90	2.90	17.10
	BMI (kg/m ²)	305	27.58	4.62	26.87	18.53	50.77	387	27.33	5.22	26.56	18.47	52.72
	Weight (kg)	305	64.00	20.58	61.70	13.60	144.20	387	62.90	21.48	60.50	22.80	138.70
	Height (m)	305	1.50	0.15	1.50	0.75	1.83	387	1.49	0.16	1.51	1.01	1.82
Essen/Marburg - obese adults	Age (yrs)	375	46.66	14.28	46.00	18.00	83.00	649	46.03	15.05	45.00	18.00	83.00
	BMI (kg/m ²)	375	34.94	4.75	33.52	29.04	63.27	649	36.28	5.71	34.72	27.99	65.58
	Weight (kg)	375	110.73	17.25	108.00	77.00	192.00	649	98.62	17.12	95.00	58.00	179.00
	Height (m)	375	1.78	0.07	1.77	1.60	2.01	649	1.65	0.07	1.65	1.35	1.84
Essen Obesity Trios (offspring)	Age (yrs)	148	13.72	3.21	13.40	4.19	24.90	205	13.67	3.29	14.00	3.76	22.18
	BMI (kg/m ²)	148	32.19	6.46	31.15	18.74	58.86	205	32.68	6.41	31.47	22.31	56.75
	Weight (kg)	147	89.80	27.67	86.50	21.30	170.00	203	86.23	25.18	85.00	26.10	173.80
	Height (m)	148	1.65	0.15	1.67	1.06	1.95	205	1.60	0.14	1.63	1.06	1.83
GINI/LISA	Age (yrs)	1010	5.22	0.17	5.20	4.84	5.83	959	5.22	0.17	5.20	4.87	5.78
	BMI (kg/m ²)	1010	15.40	1.34	15.27	11.98	24.72	959	15.37	1.36	15.26	11.89	21.95
	Weight (kg)	1010	19.96	2.57	19.85	13.80	35.00	959	19.59	2.56	19.40	13.00	34.30
	Height (m)	1010	1.13	0.05	1.17	0.99	1.28	959	1.13	0.05	1.13	0.99	1.26
NFBC-1986	Age (yrs)	2642	16.00	NA	16.00	16.00	16.00	2728	16.00	NA	16.00	16.00	16.00
	BMI (kg/m ²)	2634	21.54	3.98	20.77	11.07	44.66	2718	21.00	3.57	20.46	10.36	46.94
	Weight (kg)	2637	64.92	12.70	62.60	35.10	140.60	2721	57.23	9.90	55.40	34.70	123.20
	Height (m)	2634	1.74	0.08	1.74	1.46	1.98	2719	1.65	0.07	1.65	1.32	1.98
USCS	Age (yrs)	489	8.70	3.18	7.69	6.54	31.00	435	9.00	3.28	7.67	6.25	27.00
	BMI (kg/m ²)	489	19.77	9.55	16.14	12.75	87.10	435	20.72	10.59	16.12	11.90	73.20
	Weight (kg)	489	40.76	37.97	27.00	15.40	261.00	435	42.25	35.96	26.30	16.40	187.40
	Height (m)	489	1.34	0.18	1.29	1.09	1.99	435	1.34	0.17	1.28	1.09	1.84

Supplementary Note

Supplementary Note - Table 10 Nominally significant interactions ($P_{\text{interaction}} < 0.05$) for all pairwise tests between the lead SNPs at the 32 confirmed BMI loci. The results are not significant after correcting for multiple testing.

Supplementary Note - Table 10

Supplementary Note - Table 10 -- Nominally significant interactions ($P_{interaction} < 0.05$) for all pairwise tests between the lead SNPs at the 32 confirmed BMI loci. The results are not significant after correcting for multiple testing.

Markers		Pairwise interaction	Interaction P
SNP 1	SNP2	beta (SE)	
rs2287019	rs2890652	0.038 (0.012)	1.79E-03
rs13107325	rs29941	-0.048 (0.016)	2.64E-03
rs2287019	rs2815752	-0.025 (0.009)	7.56E-03
rs1555543	rs2241423	-0.020 (0.008)	9.45E-03
rs12622013	rs2815752	-0.026 (0.010)	1.12E-02
rs2241423	rs543874	-0.024 (0.010)	1.36E-02
rs12444979	rs2287019	-0.034 (0.014)	1.45E-02
rs1555543	rs2287019	-0.023 (0.009)	1.47E-02
rs12444979	rs713586	0.023 (0.010)	1.93E-02
rs2112347	rs4836133	0.016 (0.007)	2.31E-02
rs11847697	rs571312	-0.044 (0.020)	2.38E-02
rs10938397	rs571312	0.018 (0.008)	2.40E-02
rs1558902	rs987237	-0.019 (0.009)	2.52E-02
rs3810291	rs987237	-0.022 (0.010)	2.54E-02
rs2815752	rs7359397	-0.015 (0.007)	2.77E-02
rs11847697	rs13078807	0.045 (0.021)	2.79E-02
rs1514175	rs987237	0.018 (0.008)	2.98E-02
rs7138803	rs987237	-0.018 (0.009)	3.03E-02
rs1555543	rs7359397	0.014 (0.007)	3.11E-02
rs12444979	rs3817334	0.021 (0.010)	3.11E-02
rs10938397	rs206936	0.018 (0.008)	3.18E-02
rs2241423	rs571312	0.019 (0.009)	3.24E-02
rs29941	rs7138803	-0.015 (0.007)	3.26E-02
rs713586	rs987237	0.017 (0.008)	3.72E-02
rs1558902	rs4771122	-0.016 (0.008)	3.78E-02
rs10938397	rs4771122	-0.017 (0.008)	3.93E-02
rs7138803	rs7359397	-0.014 (0.007)	4.05E-02
rs13078807	rs29941	-0.017 (0.009)	4.17E-02
rs2241423	rs4771122	0.019 (0.009)	4.17E-02
rs3817334	rs7138803	0.013 (0.007)	4.34E-02
rs11847697	rs3810291	-0.038 (0.019)	4.38E-02
rs7359397	rs987237	-0.017 (0.008)	4.55E-02
rs12444979	rs29941	-0.020 (0.010)	4.96E-02

Supplementary Note

Supplementary Note - Table 11 Dominant, recessive and dominance deviation results for nominally significant (dominance deviation $P < 0.05$) lead SNPs at the 32 confirmed BMI loci. The effect allele is the BMI-increasing allele. Only SNPs with a dominance deviation $P < 0.05$ are presented. The results are not significant at $P < 0.05$ after correcting for the number of tests performed.

Supplementary Note - Table 11

Supplementary Note - Table 11 -- Dominant, recessive and dominance deviation results for nominally significant (dominance deviation $P < 0.05$) lead SNPs at the 32 confirmed BMI loci. The effect allele is the BMI-increasing allele. Only SNPs with a dominance deviation $P < 0.05$ are presented. The results are not significant at $P < 0.05$ after correcting for the number of tests performed.

SNP	Nearest gene	Effect Allele	Other Allele	Additive beta (SE)	Additive P-value	Dominant beta (SE)	Dominant P-value	Recessive beta (SE)	Recessive P-value	Dom Dev beta (SE)	Dom Dev P-value
rs1558902	<i>FTO</i>	a	t	0.084 (0.005)	1.20E-74	0.093 (0.007)	5.70E-45	0.132 (0.008)	3.50E-56	-0.019 (0.006)	3.47E-03
rs12444979	<i>GPRC5B</i>	c	t	0.054 (0.007)	5.80E-16	0.035 (0.025)	1.50E-01	0.063 (0.007)	1.70E-17	-0.035 (0.014)	1.15E-02

Supplementary Note

Supplementary Note - Table 12 PubMed and OMIM hits for genes within 500kb of 32 BMI-associated SNPs, using terms "obesity" and "BMI".

Supplementary Note - Table 12 -- PubMed and OMIM hits for genes within 500kb of 32 BMI-associated SNPs, using terms "obesity" and "BMI"

Gene	index SNP	Chr	Position	OMIM terms	Distance (nt)*	Pubmed hit with search terms "obesity" or "BMI"
TMEM18	rs2867125	2	612827	bmi	45,146	Almén MS et al. The obesity gene, TMEM18, is of ancient origin, found in majority of neuronal cells in all major brain regions and associated with obesity in severely obese children. <i>BMC Med Genet.</i> 2010 Apr 9;11:58. PMID: 20380707
ACP1	rs2867125	2	612827	no match	344,547	Apelt N et al. ACP1 genotype, glutathione reductase activity, and riboflavin uptake affect cardiovascular risk in the obese. <i>Metabolism.</i> 2009 Oct;58(10):1415-23. PMID: 19570551
ACP1	rs2867125	2	612827	no match	344,547	Bottini E et al. Enzyme polymorphism and clinical variability of diseases: study of acid phosphatase locus 1 (ACP1) in obese subjects. <i>Hum Biol.</i> 1990 Jun;62(3):403-11. PMID: 2373509
ACP1	rs2867125	2	612827	no match	344,547	Bottini N et al. Association of the acid phosphatase (ACP1) gene with triglyceride levels in obese women. <i>Mol Genet Metab.</i> 2002 Nov;77(3):226-9. PMID: 12409270
ACP1	rs2867125	2	612827	no match	344,547	Bottini N et al. Low-molecular-weight protein tyrosine phosphatase and human disease: in search of biochemical mechanisms. <i>Arch Immunol Ther Exp (Warsz).</i> 2002;50(2):95-104. PMID: 12022706
ACP1	rs2867125	2	612827	no match	344,547	Bottini N et al. Type 2 diabetes and the genetics of signal transduction: a study of interaction between adenosine deaminase and acid phosphatase locus 1 polymorphisms. <i>Metabolism.</i> 2004 Aug;53(8):995-1001. PMID: 15281007
ACP1	rs2867125	2	612827	no match	344,547	De Lorenzo A et al. A study of acid phosphatase locus 1 in women with high fat content and normal body mass index. <i>Metabolism.</i> 2009 Mar;58(3):351-4. PMID: 19217450
ACP1	rs2867125	2	612827	no match	344,547	Gloria-Bottini F et al. Body mass index and acid phosphatase locus 1 in diabetic disorders. <i>Acta Diabetol.</i> 2009 Oct 24; PMID: 19855922
ACP1	rs2867125	2	612827	no match	344,547	Gloria-Bottini F et al. Effect of ACP1*C on early life viability. <i>Hum Biol.</i> 2006 Jun;78(3):365-9. PMID: 17216808
ACP1	rs2867125	2	612827	no match	344,547	Gloria-Bottini F et al. Interaction at clinical level between erythrocyte acid phosphatase and adenosine deaminase genetic polymorphisms. <i>Hum Genet.</i> 1989 Jun;82(3):213-5. PMID: 2731933
ACP1	rs2867125	2	612827	no match	344,547	Gloria-Bottini F et al. The effect of genetic and seasonal factors on birth weight. <i>Early Hum Dev.</i> 2009 Jul;85(7):439-41. PMID: 19329262
ACP1	rs2867125	2	612827	no match	344,547	Gloria-Bottini F et al. The link between obesity and allergy: a role of ACP1 genetic polymorphism?. <i>Int J Obes (Lond).</i> 2007 Feb;31(2):392-3. PMID: 16770334
ACP1	rs2867125	2	612827	no match	344,547	Greene LS et al. Acid phosphatase locus 1 (ACP1): Possible relationship of allelic variation to body size and human population adaptation to thermal stress-A theoretical perspective. <i>Am J Hum Biol.</i> 2000 Sep;12(5):688-701. PMID: 11534062
ACP1	rs2867125	2	612827	no match	344,547	Lucarini N et al. A possible genetic component of obesity in childhood. Observations on acid phosphatase polymorphism. <i>Experientia.</i> 1990 Jan 15;46(1):90-1. PMID: 2298287
ACP1	rs2867125	2	612827	no match	344,547	Lucarini N et al. Low-molecular-weight acid phosphatase (ACP1), obesity, and blood lipid levels in subjects with non-insulin-dependent diabetes mellitus. <i>Hum Biol.</i> 1997 Aug;69(4):509-15. PMID: 9198310
ACP1	rs2867125	2	612827	no match	344,547	Lucarini N et al. Phosphotyrosine-protein-phosphatase and diabetic disorders. Further studies on the relationship between low molecular weight acid phosphatase genotype and degree of glycemic control. <i>Dis Markers.</i> 1998 Oct;14(2):121-5. PMID: 9868599
ACP1	rs2867125	2	612827	no match	344,547	Maccari R et al. Structure-based optimization of benzoic acids as inhibitors of protein tyrosine phosphatase 1B and low molecular weight protein tyrosine phosphatase. <i>ChemMedChem.</i> 2009 Jun;4(6):957-62. PMID: 19288492
ACP1	rs2867125	2	612827	no match	344,547	Paggi A et al. Further studies on acid phosphatase in obese subjects. <i>Dis Markers.</i> 1991 Jan-Feb;9(1):1-7. PMID: 1742941
ACP1	rs2867125	2	612827	no match	344,547	Pandey SK et al. Reduction of low molecular weight protein-tyrosine phosphatase expression improves hyperglycemia and insulin sensitivity in obese mice. <i>J Biol Chem.</i> 2007 May 11;282(19):14291-9. PMID: 17353188
CENPO	rs713586	2	25011512	no match	115,224	Obuse C et al. Proteomics analysis of the centromere complex from HeLa interphase cells: UV-damaged DNA binding protein 1 (DDB-1) is a component of the CEN-complex, while BMI-1 is transiently co-localized with the centromeric region in interphase. <i>Genes Cells.</i> 2004 Feb;9(2):105-20. PMID: 15009096
NCOA1	rs713586	2	25011512	obesity	164,438	Fan D et al. Mechanistic roles of leptin in osteogenic stimulation in thoracic ligament flavum cells. <i>J Biol Chem.</i> 2007 Oct 12;282(41):29958-66. PMID: 17702747
NCOA1	rs713586	2	25011512	obesity	164,438	Foryst-Ludwig A et al. Metabolic actions of estrogen receptor beta (ERbeta) are mediated by a negative cross-talk with PPARgamma. <i>PLoS Genet.</i> 2008 Jun 27;4(6):e1000108. PMID: 18584035
NCOA1	rs713586	2	25011512	obesity	164,438	Jeong JW et al. The genomic analysis of the impact of steroid receptor coactivators ablation on hepatic metabolism. <i>Mol Endocrinol.</i> 2006 May;20(5):1138-52. PMID: 16423883
NCOA1	rs713586	2	25011512	obesity	164,438	Lee HS et al. Epitope analysis of PPARgamma monoclonal antibody Pgamma48.34A and its application for screening PPARgamma ligands. <i>J Immunol Methods.</i> 2005 Jan;296(1-2):125-34. PMID: 15680157
NCOA1	rs713586	2	25011512	obesity	164,438	Miard S et al. Aging alters PPARgamma in rodent and human adipose tissue by modulating the balance in steroid receptor coactivator-1. <i>Aging Cell.</i> 2009 Aug;8(4):449-59. PMID: 19485965
NCOA1	rs713586	2	25011512	obesity	164,438	Picard F et al. SRC-1 and TIF2 control energy balance between white and brown adipose tissues. <i>Cell.</i> 2002 Dec 27;111(7):931-41. PMID: 12507421
NCOA1	rs713586	2	25011512	obesity	164,438	Tateishi K et al. Role of Jhd2a in regulating metabolic gene expression and obesity resistance. <i>Nature.</i> 2009 Apr 9;458(7239):757-61. PMID: 19194461
NCOA1	rs713586	2	25011512	obesity	164,438	Wang Z et al. Critical roles of the p160 transcriptional coactivators p/CIP and SRC-1 in energy balance. <i>Cell Metab.</i> 2006 Feb;3(2):111-22. PMID: 16459312
NCOA1	rs713586	2	25011512	obesity	164,438	Yin N et al. Molecular mechanisms involved in the growth stimulation of breast cancer cells by leptin. <i>Cancer Res.</i> 2004 Aug 15;64(16):5870-5. PMID: 15313931
POMC	rs713586	2	25011512	obesity, bmi	225,714	Alfieri A et al. Functional analysis of melanocortin-4-receptor mutants identified in severely obese subjects living in Southern Italy. <i>Gene.</i> 2010 Jun 1;457(1-2):35-41. PMID: 20214954
POMC	rs713586	2	25011512	obesity, bmi	225,714	Al-Qassab H et al. Dominant role of the p110beta isoform of PI3K over p110alpha in energy homeostasis regulation by POMC and AgRP neurons. <i>Cell Metab.</i> 2009 Nov;10(5):343-54. PMID: 19883613

Gene	index SNP	Chr	Position	OMIM terms	Distance (nt)*	Pubmed hit with search terms "obesity" or "BMI"
POMC	rs713586	2	25011512	obesity, bmi	225,714	Baker M et al. Association between common polymorphisms of the proopiomelanocortin gene and body fat distribution: a family study. <i>Diabetes</i> . 2005 Aug;54(8):2492-6. PMID: 16046320
POMC	rs713586	2	25011512	obesity, bmi	225,714	Battin J et al. [Genetic obesity in childhood]. <i>Bull Acad Natl Med</i> . 2009 Jun;193(6):1281-8. PMID: 20120159
POMC	rs713586	2	25011512	obesity, bmi	225,714	Biebermann H et al. A role for beta-melanocyte-stimulating hormone in human body-weight regulation. <i>Cell Metab</i> . 2006 Feb;3(2):141-6. PMID: 16459315
POMC	rs713586	2	25011512	obesity, bmi	225,714	Bienertova-Vasku J et al. No association of defined variability in leptin, leptin receptor, adiponectin, proopiomelanocortin and ghrelin gene with food preferences in the Czech population. <i>Nutr Neurosci</i> . 2008 Feb;11(1):2-8. PMID: 18510797
POMC	rs713586	2	25011512	obesity, bmi	225,714	Bjørbaek C et al. Central leptin receptor action and resistance in obesity. <i>J Investig Med</i> . 2009 Oct;57(7):789-94. PMID: 20029269
POMC	rs713586	2	25011512	obesity, bmi	225,714	Bruno OD et al. In what clinical settings should Cushing's syndrome be suspected?. <i>Medicina (B Aires)</i> . 2009;69(6):674-80. PMID: 20053613
POMC	rs713586	2	25011512	obesity, bmi	225,714	Buono P et al. Six novel mutations in the proopiomelanocortin and melanocortin receptor 4 genes in severely obese adults living in southern Italy. <i>Clin Chem</i> . 2005 Aug;51(8):1358-64. PMID: 15951321
POMC	rs713586	2	25011512	obesity, bmi	225,714	Cakir I et al. Hypothalamic Sirt1 regulates food intake in a rodent model system. <i>PLoS One</i> . 2009 Dec 15;4(12):e8322. PMID: 20020036
POMC	rs713586	2	25011512	obesity, bmi	225,714	Challis BG et al. A missense mutation disrupting a dibasic prohormone processing site in pro-opiomelanocortin (POMC) increases susceptibility to early-onset obesity through a novel molecular mechanism. <i>Hum Mol Genet</i> . 2002 Aug 15;11(17):1997-2004. PMID: 12165561
POMC	rs713586	2	25011512	obesity, bmi	225,714	Chen HH et al. Severe obesity is associated with novel single nucleotide polymorphisms of the ESR1 and PPARgamma locus in Han Chinese. <i>Am J Clin Nutr</i> . 2009 Aug;90(2):255-62. PMID: 19491387
POMC	rs713586	2	25011512	obesity, bmi	225,714	Chuang JC et al. A beta(3)-Adrenergic-Leptin-Melanocortin Circuit Regulates Behavioral and Metabolic Changes Induced by Chronic Stress. <i>Biol Psychiatry</i> . 2010 Jan 8; PMID: 20060958
POMC	rs713586	2	25011512	obesity, bmi	225,714	Corander MP et al. Science of self-preservation: how melanocortin action in the brain modulates body weight, blood pressure, and ischemic damage. <i>Circulation</i> . 2009 Dec 1;120(22):2260-8. PMID: 19948994
POMC	rs713586	2	25011512	obesity, bmi	225,714	Creemers JW et al. Mutations in the amino-terminal region of proopiomelanocortin (POMC) in patients with early-onset obesity impair POMC sorting to the regulated secretory pathway. <i>J Clin Endocrinol Metab</i> . 2008 Nov;93(11):4494-9. PMID: 18697863
POMC	rs713586	2	25011512	obesity, bmi	225,714	Darcan S et al. Transient salt wasting in POMC-deficiency due to infection induced stress. <i>Exp Clin Endocrinol Diabetes</i> . 2010 Apr;118(4):281-3. PMID: 19998238
POMC	rs713586	2	25011512	obesity, bmi	225,714	Delplanque J et al. Linkage and association studies between the proopiomelanocortin (POMC) gene and obesity in caucasian families. <i>Diabetologia</i> . 2000 Dec;43(12):1554-7. PMID: 11151766
POMC	rs713586	2	25011512	obesity, bmi	225,714	Doecke JD et al. Single nucleotide polymorphisms in obesity-related genes and the risk of esophageal cancers. <i>Cancer Epidemiol Biomarkers Prev</i> . 2008 Apr;17(4):1007-12. PMID: 18398047
POMC	rs713586	2	25011512	obesity, bmi	225,714	Dubern B et al. Mutational analysis of the pro-opiomelanocortin gene in French obese children led to the identification of a novel deleterious heterozygous mutation located in the alpha-melanocyte stimulating hormone domain. <i>Pediatr Res</i> . 2008 Feb;63(2):211-6. PMID: 18091355
POMC	rs713586	2	25011512	obesity, bmi	225,714	Echwald SM et al. Mutational analysis of the proopiomelanocortin gene in Caucasians with early onset obesity. <i>Int J Obes Relat Metab Disord</i> . 1999 Mar;23(3):293-8. PMID: 10193875
POMC	rs713586	2	25011512	obesity, bmi	225,714	Gout J et al. Metabolic and melanocortin gene expression alterations in male offspring of obese mice. <i>Mol Cell Endocrinol</i> . 2010 May 5;319(1-2):99-108. PMID: 20097259
POMC	rs713586	2	25011512	obesity, bmi	225,714	Grayson BE et al. Changes in melanocortin expression and inflammatory pathways in fetal offspring of nonhuman primates fed a high-fat diet. <i>Endocrinology</i> . 2010 Apr;151(4):1622-32. PMID: 20176722
POMC	rs713586	2	25011512	obesity, bmi	225,714	Hall JE et al. Obesity-induced Hypertension: Role of Sympathetic Nervous System, Leptin and Melanocortins. <i>J Biol Chem</i> . 2010 Mar 26; PMID: 20348094
POMC	rs713586	2	25011512	obesity, bmi	225,714	Hawke Z et al. PACAP neurons in the hypothalamic ventromedial nucleus are targets of central leptin signaling. <i>J Neurosci</i> . 2009 Nov 25;29(47):14828-35. PMID: 19940178
POMC	rs713586	2	25011512	obesity, bmi	225,714	Hetherington MM et al. Gene-environment interactions in obesity. <i>Forum Nutr</i> . 2010;63:195-203. PMID: 19955787
POMC	rs713586	2	25011512	obesity, bmi	225,714	Hinney A et al. Systematic mutation screening of the pro-opiomelanocortin gene: identification of several genetic variants including three different insertions, one nonsense and two missense point mutations in probands of different weight extremes. <i>J Clin Endocrinol Metab</i> . 1998 Oct;83(10):3737-41. PMID: 9768693
POMC	rs713586	2	25011512	obesity, bmi	225,714	Hochberg I et al. Hypothalamic Obesity. <i>Endocr Dev</i> . 2010;17:185-196. PMID: 19955767
POMC	rs713586	2	25011512	obesity, bmi	225,714	Iskandar K et al. PDK-1/FoxO1 pathway in POMC neurons regulates Pomc expression and food intake. <i>Am J Physiol Endocrinol Metab</i> . 2010 Apr;298(4):E787-98. PMID: 20103739
POMC	rs713586	2	25011512	obesity, bmi	225,714	Kim DH et al. Peptide designed to elicit apoptosis in adipose tissue endothelium reduces food intake and body weight. <i>Diabetes</i> . 2010 Apr;59(4):907-15. PMID: 20103704
POMC	rs713586	2	25011512	obesity, bmi	225,714	Krude H et al. Severe early-onset obesity, adrenal insufficiency and red hair pigmentation caused by POMC mutations in humans. <i>Nat Genet</i> . 1998 Jun;19(2):155-7. PMID: 9620771
POMC	rs713586	2	25011512	obesity, bmi	225,714	la Fleur SE et al. A free-choice high-fat high-sugar diet induces changes in arcuate neuropeptide expression that support hyperphagia. <i>Int J Obes (Lond)</i> . 2010 Mar;34(3):537-46. PMID: 20029382
POMC	rs713586	2	25011512	obesity, bmi	225,714	Lee YS et al. A POMC variant implicates beta-melanocyte-stimulating hormone in the control of human energy balance. <i>Cell Metab</i> . 2006 Feb;3(2):135-40. PMID: 16459314
POMC	rs713586	2	25011512	obesity, bmi	225,714	Li JY et al. Expression of ankyrin repeat and suppressor of cytokine signaling box protein 4 (Asb-4) in proopiomelanocortin neurons of the arcuate nucleus of mice produces a hyperphagic, lean phenotype. <i>Endocrinology</i> . 2010 Jan;151(1):134-42. PMID: 19934378

Gene	index SNP	Chr	Position	OMIM terms	Distance (nt)*	Pubmed hit with search terms "obesity" or "BMI"
POMC	rs713586	2	25011512	obesity, bmi	225,714	Li P et al. Expression of adiponectin receptors in mouse adrenal glands and the adrenocortical Y-1 cell line: Adiponectin regulates steroidogenesis. <i>Biochem Biophys Res Commun.</i> 2009 Dec 25;390(4):1208-13. PMID: 19878661
POMC	rs713586	2	25011512	obesity, bmi	225,714	Lin S et al. Critical role of arcuate Y4 receptors and the melanocortin system in pancreatic polypeptide-induced reduction in food intake in mice. <i>PLoS One.</i> 2009 Dec 30;4(12):e8488. PMID: 20041129
POMC	rs713586	2	25011512	obesity, bmi	225,714	Macia L et al. Interleukin-7, a new cytokine targeting the mouse hypothalamic arcuate nucleus: role in body weight and food intake regulation. <i>PLoS One.</i> 2010 Apr 1;5(4):e9953. PMID: 20376352
POMC	rs713586	2	25011512	obesity, bmi	225,714	Menzies JR et al. Direct and Indirect Effects of Cannabinoids on in vitro GABA Release in the Rat Arcuate Nucleus. <i>J Neuroendocrinol.</i> 2010 Mar 2;. PMID: 20236227
POMC	rs713586	2	25011512	obesity, bmi	225,714	Miraglia del Giudice E et al. Molecular screening of the proopiomelanocortin (POMC) gene in Italian obese children: report of three new mutations. <i>Int J Obes Relat Metab Disord.</i> 2001 Jan;25(1):61-7. PMID: 11244459
POMC	rs713586	2	25011512	obesity, bmi	225,714	Ohshiro Y et al. Sequence analysis of the pro-opiomelanocortin (POMC) gene in obese/diabetic Japanese. <i>Int J Obes Relat Metab Disord.</i> 2002 May;26(5):730-1. PMID: 12032760
POMC	rs713586	2	25011512	obesity, bmi	225,714	Oliszewski PK et al. Alpha-melanocyte stimulating hormone and ghrelin: central interaction in feeding control. <i>Peptides.</i> 2007 Oct;28(10):2084-9. PMID: 17719137
POMC	rs713586	2	25011512	obesity, bmi	225,714	Pankov luA et al. [Screening of mutations in genes of pro-opiomelanocortin in patients with constitutional exogenous obesity]. <i>Vopr Med Khim.</i> 2002 Jan- Feb;48(1):121-30. PMID: 12068494
POMC	rs713586	2	25011512	obesity, bmi	225,714	Pétevári E et al. Suppression of food intake by intracerebroventricular injection of alpha-MSH varies with age in rats. <i>Acta Physiol Hung.</i> 2009 Dec;96(4):483-7. PMID: 19942555
POMC	rs713586	2	25011512	obesity, bmi	225,714	Potoczna N et al. Gene variants and binge eating as predictors of comorbidity and outcome of treatment in severe obesity. <i>J Gastrointest Surg.</i> 2004 Dec;8(8):971-81; discussion 981-2. PMID: 15585384
POMC	rs713586	2	25011512	obesity, bmi	225,714	Reed AS et al. Functional role of suppressor of cytokine signaling 3 upregulation in hypothalamic leptin resistance and long-term energy homeostasis. <i>Diabetes.</i> 2010 Apr;59(4):894-906. PMID: 20068134
POMC	rs713586	2	25011512	obesity, bmi	225,714	Rosmond R et al. Polymorphisms in exon 3 of the proopiomelanocortin gene in relation to serum leptin, salivary cortisol, and obesity in Swedish men. <i>Metabolism.</i> 2002 May;51(5):642-4. PMID: 11979399
POMC	rs713586	2	25011512	obesity, bmi	225,714	Ruaño G et al. Physiogenomic comparison of edema and BMI in patients receiving rosiglitazone or pioglitazone. <i>Clin Chim Acta.</i> 2009 Feb;400(1-2):48-55. PMID: 18996102
POMC	rs713586	2	25011512	obesity, bmi	225,714	Santoro N et al. An insertional polymorphism of the proopiomelanocortin gene is associated with fasting insulin levels in childhood obesity. <i>J Clin Endocrinol Metab.</i> 2004 Oct;89(10):4846-9. PMID: 15472174
POMC	rs713586	2	25011512	obesity, bmi	225,714	Stefater MA et al. Sleeve Gastrectomy Induces Loss of Weight and Fat Mass in Obese Rats, but Does Not Affect Leptin Sensitivity. <i>Gastroenterology.</i> 2010 Mar 10;. PMID: 20226189
POMC	rs713586	2	25011512	obesity, bmi	225,714	Su H et al. Gamma-protocadherins regulate the functional integrity of hypothalamic feeding circuitry in mice. <i>Dev Biol.</i> 2010 Mar 1;339(1):38-50. PMID: 20025866
POMC	rs713586	2	25011512	obesity, bmi	225,714	Sutton GM et al. Protein malnutrition during pregnancy in C57BL/6J mice results in offspring with altered circadian physiology before obesity. <i>Endocrinology.</i> 2010 Apr;151(4):1570-80. PMID: 20160133
POMC	rs713586	2	25011512	obesity, bmi	225,714	Tung YC et al. Hypothalamic-specific manipulation of Fto, the ortholog of the human obesity gene FTO, affects food intake in rats. <i>PLoS One.</i> 2010 Jan 19;5(1):e8771. PMID: 20025866
POMC	rs713586	2	25011512	obesity, bmi	225,714	Valli-Jaakola K et al. Further evidence for the role of ENPP1 in obesity: association with morbid obesity in Finns. <i>Obesity (Silver Spring).</i> 2008 Sep;16(9):2113-9. PMID: 18551113
POMC	rs713586	2	25011512	obesity, bmi	225,714	Xiao E et al. Effects of estradiol on cerebrospinal fluid levels of agouti-related protein in ovariectomized rhesus monkeys. <i>Endocrinology.</i> 2010 Mar;151(3):1002-9. PMID: 20056830
POMC	rs713586	2	25011512	obesity, bmi	225,714	Yang G et al. FoxO1 inhibits leptin regulation of pro-opiomelanocortin promoter activity by blocking STAT3 interaction with specificity protein 1. <i>J Biol Chem.</i> 2009 Feb 6;284(6):3719-27. PMID: 19049975
DNMT3A	rs713586	2	25011512	no match	297,837	Kamei Y et al. Increased expression of DNA methyltransferase 3a in obese adipose tissue: studies with transgenic mice. <i>Obesity (Silver Spring).</i> 2010 Feb;18(2):314-21. PMID: 19680236
LRP1B	rs2890652	2	142676401	no match	70,661	Masson O et al. LRP1 receptor controls adipogenesis and is up-regulated in human and mouse obese adipose tissue. <i>PLoS One.</i> 2009 Oct 12;4(10):e7422. PMID: 19823686
IGF2BP2	rs9816226	3	187317193	obesity	291,672	Pecioska S et al. Association between type 2 diabetes loci and measures of fatness. <i>PLoS One.</i> 2010 Jan 1;5(1):e8541. PMID: 20049090
IGF2BP2	rs9816226	3	187317193	obesity	291,672	Ruchat SM et al. Combining genetic markers and clinical risk factors improves the risk assessment of impaired glucose metabolism. <i>Ann Med.</i> 2010 Apr;42(3):196-206. PMID: 20384434
IGF2BP2	rs9816226	3	187317193	obesity	291,672	Sjögren M et al. The search for putative genetic factors for components of the metabolic syndrome. <i>Diabetologia.</i> 2008 Dec;51(12):2242-51. PMID: 18853134
AHSG	rs9816226	3	187317193	no match	496,351	Andersen G et al. AHSG tag single nucleotide polymorphisms associate with type 2 diabetes and dyslipidemia: studies of metabolic traits in 7,683 white Danish subjects. <i>Diabetes.</i> 2008 May;57(5):1427-32. PMID: 18316360
AHSG	rs9816226	3	187317193	no match	496,351	Axelsson J et al. Is fetuin-A/alpha2-Heremans-Schmid glycoprotein associated with the metabolic syndrome in patients with chronic kidney disease?. <i>Am J Nephrol.</i> 2008;28(4):669-76. PMID: 18337634
AHSG	rs9816226	3	187317193	no match	496,351	Dahlman I et al. alpha2-Heremans-Schmid glycoprotein gene polymorphisms are associated with adipocyte insulin action. <i>Diabetologia.</i> 2004 Nov;47(11):1974-9. PMID: 15599699
AHSG	rs9816226	3	187317193	no match	496,351	Ix JH et al. Mechanisms linking obesity, chronic kidney disease, and fatty liver disease: the roles of fetuin-A, adiponectin, and AMPK. <i>J Am Soc Nephrol.</i> 2010 Mar;21(3):406-12. PMID: 20150538

Gene	index SNP	Chr	Position	OMIM terms	Distance (nt)*	Pubmed hit with search terms "obesity" or "BMI"
AHSG	rs9816226	3	187317193	no match	496,351	Lavebratt C et al. Polymorphism of the AHSG gene is associated with increased adipocyte beta2-adrenoceptor function. J Lipid Res. 2005 Oct;46(10):2278-81. PMID: 16024912
AHSG	rs9816226	3	187317193	no match	496,351	Mathews ST et al. Fetuin-null mice are protected against obesity and insulin resistance associated with aging. Biochem Biophys Res Commun. 2006 Nov 17;350(2):437-43. PMID: 17011519
AHSG	rs9816226	3	187317193	no match	496,351	Mathews ST et al. Improved insulin sensitivity and resistance to weight gain in mice null for the Ahsg gene. Diabetes. 2002 Aug;51(8):2450-8. PMID: 12145157
AHSG	rs9816226	3	187317193	no match	496,351	Müssig K et al. AHSG gene variation is not associated with regional body fat distribution—a magnetic resonance study." Exp Clin Endocrinol Diabetes. 2009 Sep;117(8):432-7. PMID: 19358088
AHSG	rs9816226	3	187317193	no match	496,351	Oikawa O et al. Evaluation of serum fetuin-A relationships with biochemical parameters in patients on hemodialysis. Clin Exp Nephrol. 2007 Dec;11(4):304-8. PMID: 18085392
AHSG	rs9816226	3	187317193	no match	496,351	Reinehr T et al. Fetuin-A and its relation to metabolic syndrome and fatty liver disease in obese children before and after weight loss. J Clin Endocrinol Metab. 2008 Nov;93(11):4479-85. PMID: 18728159
AHSG	rs9816226	3	187317193	no match	496,351	Siddiq A et al. A synonymous coding polymorphism in the alpha2-Heremans-schmid glycoprotein gene is associated with type 2 diabetes in French Caucasians. Diabetes. 2005 Aug;54(8):2477-81. PMID: 16046317
AHSG	rs9816226	3	187317193	no match	496,351	Stefan N et al. Alpha2-Heremans-Schmid glycoprotein/fetuin-A is associated with insulin resistance and fat accumulation in the liver in humans. Diabetes Care. 2006 Apr;29(4):853-7. PMID: 16567827
AHSG	rs9816226	3	187317193	no match	496,351	Stefan N et al. Plasma fetuin-A levels and the risk of type 2 diabetes. Diabetes. 2008 Oct;57(10):2762-7. PMID: 18633113
AHSG	rs9816226	3	187317193	no match	496,351	Yilmaz MI et al. Endothelial dysfunction in type-2 diabetics with early diabetic nephropathy is associated with low circulating adiponectin. Nephrol Dial Transplant. 2008 May;23(5):1621-7. PMID: 18175782
NFKB1	rs13107325	4	103407732	obesity	233,786	Haugen F et al. Activation of nuclear factor-kappaB by high molecular weight and globular adiponectin. Endocrinology. 2007 Nov;148(11):5478-86. PMID: 17702846
NFKB1	rs13107325	4	103407732	obesity	233,786	Hou B et al. Tumor necrosis factor alpha activates the human plasminogen activator inhibitor-1 gene through a distal nuclear factor kappaB site. J Biol Chem. 2004 Apr 30;279(18):18127-36. PMID: 14963043
NFKB1	rs13107325	4	103407732	obesity	233,786	Nareika A et al. Sodium lactate increases LPS-stimulated MMP and cytokine expression in U937 histiocytes by enhancing AP-1 and NF-kappaB transcriptional activities. Am J Physiol Endocrinol Metab. 2005 Oct;289(4):E534-42. PMID: 15941782
NFKB1	rs13107325	4	103407732	obesity	233,786	Prada PO et al. EGFR tyrosine kinase inhibitor (PD153035) improves glucose tolerance and insulin action in high-fat diet-fed mice. Diabetes. 2009 Dec;58(12):2910-9. PMID: 19696185
NFKB1	rs13107325	4	103407732	obesity	233,786	Sheu WH et al. Effect of weight loss on proinflammatory state of mononuclear cells in obese women. Obesity (Silver Spring). 2008 May;16(5):1033-8. PMID: 18356846
NFKB1	rs13107325	4	103407732	obesity	233,786	Silswal N et al. Human resistin stimulates the pro-inflammatory cytokines TNF-alpha and IL-12 in macrophages by NF-kappaB-dependent pathway. Biochem Biophys Res Commun. 2005 Sep 9;334(4):1092-101. PMID: 16039994
NFKB1	rs13107325	4	103407732	obesity	233,786	Vitseva OI et al. Inducible Toll-like receptor and NF-kappaB regulatory pathway expression in human adipose tissue. Obesity (Silver Spring). 2008 May;16(5):932-7. PMID: 18292749
HMGCR	rs2112347	5	75050998	no match	357,318	Chan MY et al. Sequential responses to high-fat and high-calorie feeding in an obese mouse model. Obesity (Silver Spring). 2008 May;16(5):972-8. PMID: 18292748
HMGCR	rs2112347	5	75050998	no match	357,318	Ruaño G et al. Physiogenomic comparison of edema and BMI in patients receiving rosiglitazone or pioglitazone. Clin Chim Acta. 2009 Feb;400(1-2):48-55. PMID: 18996102
HMGA1	rs206936	6	34410847	no match	88,861	Anand A et al. In vivo modulation of Hmgic reduces obesity. Nat Genet. 2000 Apr;24(4):377-80. PMID: 10742101
HMGA1	rs206936	6	34410847	no match	88,861	Auwerx J et al. Transcription, adipocyte differentiation, and obesity. J Mol Med. 1996 Jul;74(7):347-52. PMID: 8841947
HMGA1	rs206936	6	34410847	no match	88,861	Esposito F et al. Interaction between HMGA1 and retinoblastoma protein is required for adipocyte differentiation. J Biol Chem. 2009 Sep 18;284(38):25993-6004. PMID: 19633359
TFAP2B	rs987237	6	50911009	no match	0	Ugi S et al. Relation of the Expression of Transcriptional Factor TFAP2B to That of Adipokines in Subcutaneous and Omental Adipose Tissues. Obesity (Silver Spring). 2009 Dec 17;. PMID: 20019683
RIC3	rs4929949	11	8561169	no match	414,025	Sniieder H et al. TUB is a candidate gene for late-onset obesity in women. Diabetologia. 2008 Jan;51(1):54-61. PMID: 17955208
TUB	rs4929949	11	8561169	obesity	476,941	Bäckberg M et al. Abnormal cholinergic and GABAergic vascular innervation in the hypothalamic arcuate nucleus of obese tub/tub mice. Synapse. 2004 Jun 15;52(4):245-57. PMID: 15103691
TUB	rs4929949	11	8561169	obesity	476,941	Bäckberg M et al. Down-regulated expression of agouti-related protein (AGRP) mRNA in the hypothalamic arcuate nucleus of hyperphagic and obese tub/tub mice. Brain Res Mol Brain Res. 2004 Jun 18;125(1-2):129-39. PMID: 15193430
TUB	rs4929949	11	8561169	obesity	476,941	Boggon TJ et al. Implication of tubby proteins as transcription factors by structure-based functional analysis. Science. 1999 Dec 10;286(5447):2119-25. PMID: 10591637
TUB	rs4929949	11	8561169	obesity	476,941	Bouchard G et al. Cholesterol gallstone formation in overweight mice establishes that obesity per se is not linked directly to cholelithiasis risk. J Lipid Res. 2002 Jul;43(7):1105-13. PMID: 12091495
TUB	rs4929949	11	8561169	obesity	476,941	Caberoy NB et al. Efficient identification of tubby-binding proteins by an improved system of T7 phage display. J Mol Recognit. 2010 Jan- Feb;23(1):74-83. PMID: 19718693
TUB	rs4929949	11	8561169	obesity	476,941	Cantley LC et al. Transcription. Translocating tubby. Science. 2001 Jun 15;292(5524):2019-21. PMID: 11408644
TUB	rs4929949	11	8561169	obesity	476,941	Carroll K et al. Tubby proteins: the plot thickens. Nat Rev Mol Cell Biol. 2004 Jan;5(1):55-63. PMID: 14708010
TUB	rs4929949	11	8561169	obesity	476,941	Chung WK et al. Analysis of 30 genes (355 SNPs) related to energy homeostasis for association with adiposity in European-American and Yup'ik Eskimo populations. Hum Hered. 2009;67(3):193-205. PMID: 19077438

Gene	index SNP	Chr	Position	OMIM terms	Distance (nt)*	Pubmed hit with search terms "obesity" or "BMI"
TUB	rs4929949	11	8561169	obesity	476,941	Coyle CA et al. Reduced activity without hyperphagia contributes to obesity in Tubby mutant mice. <i>Physiol Behav.</i> 2008 Sep 3;95(1-2):168-75. PMID: 18619628
TUB	rs4929949	11	8561169	obesity	476,941	Fukagawa K et al. [Genetic abnormality and the mechanism of ingestive disorder in obese animal model]. <i>Nippon Rinsho.</i> 2001 Mar;59(3):466-71. PMID: 11268594
TUB	rs4929949	11	8561169	obesity	476,941	Giannaccini G et al. Tubby protein in human lymphocytes from normal weight and obese subjects. <i>Clin Biochem.</i> 2007 Jul;40(11):806-9. PMID: 17498679
TUB	rs4929949	11	8561169	obesity	476,941	He W et al. GFP-tagged expression and immunohistochemical studies to determine the subcellular localization of the tubby gene family members. <i>Brain Res Mol Brain Res.</i> 2000 Sep 30;81(1-2):109-17. PMID: 11000483
TUB	rs4929949	11	8561169	obesity	476,941	Ikeda A et al. Genetic modification of retinal degeneration in tubby mice. <i>Exp Eye Res.</i> 2002 Apr;74(4):455-61. PMID: 12076089
TUB	rs4929949	11	8561169	obesity	476,941	Ikeda A et al. Microtubule-associated protein 1A is a modifier of tubby hearing (moth1). <i>Nat Genet.</i> 2002 Apr;30(4):401-5. PMID: 11925566
TUB	rs4929949	11	8561169	obesity	476,941	Ikeda A et al. The tubby-like proteins, a family with roles in neuronal development and function. <i>J Cell Sci.</i> 2002 Jan 1;115(Pt 1):9-14. PMID: 11801719
TUB	rs4929949	11	8561169	obesity	476,941	Jones JM et al. Localization of insulin-2 (Ins-2) and the obesity mutant tubby (tub) to distinct regions of mouse chromosome 7. <i>Genomics.</i> 1992 Sep;14(1):197-9. PMID: 1358794
TUB	rs4929949	11	8561169	obesity	476,941	Kapeller R et al. Tyrosine phosphorylation of tub and its association with Src homology 2 domain-containing proteins implicate tub in intracellular signaling by insulin. <i>J Biol Chem.</i> 1999 Aug 27;274(35):24980-6. PMID: 10455176
TUB	rs4929949	11	8561169	obesity	476,941	Kleyn PW et al. Identification and characterization of the mouse obesity gene tubby: a member of a novel gene family. <i>Cell.</i> 1996 Apr 19;85(2):281-90. PMID: 8612280
TUB	rs4929949	11	8561169	obesity	476,941	Koritschoner NP et al. Thyroid hormone regulates the obesity gene tub. <i>EMBO Rep.</i> 2001 Jun;2(6):499-504. PMID: 11415982
TUB	rs4929949	11	8561169	obesity	476,941	Lee JH et al. Microarray analysis of differentially expressed genes in the brains of tubby mice. <i>Korean J Physiol Pharmacol.</i> 2009 Apr;13(2):91-7. PMID: 19885003
TUB	rs4929949	11	8561169	obesity	476,941	Mak HY et al. Polygenic control of <i>Caenorhabditis elegans</i> fat storage. <i>Nat Genet.</i> 2006 Mar;38(3):363-8. PMID: 16462744
TUB	rs4929949	11	8561169	obesity	476,941	Mukhopadhyay A et al. An endocytic pathway as a target of tubby for regulation of fat storage. <i>EMBO Rep.</i> 2007 Oct;8(10):931-8. PMID: 17762880
TUB	rs4929949	11	8561169	obesity	476,941	Nagai T et al. Cause of sudden, unexpected death of Prader-Willi syndrome patients with or without growth hormone treatment. <i>Am J Med Genet A.</i> 2005 Jul 1;136(1):45-8. PMID: 15937939
TUB	rs4929949	11	8561169	obesity	476,941	Quinn KV et al. Monitoring changes in membrane phosphatidylinositol 4,5-bisphosphate in living cells using a domain from the transcription factor tubby. <i>J Physiol.</i> 2008 Jun 15;586(Pt 12):2855-71. PMID: 18420701
TUB	rs4929949	11	8561169	obesity	476,941	Ronshaugen M et al. Structure and expression patterns of <i>Drosophila</i> TULP and TUSP, members of the tubby-like gene family. <i>Mech Dev.</i> 2002 Sep;117(1-2):209-15. PMID: 12204260
TUB	rs4929949	11	8561169	obesity	476,941	Santagata S et al. G-protein signaling through tubby proteins. <i>Science.</i> 2001 Jun 15;292(5524):2041-50. PMID: 11375483
TUB	rs4929949	11	8561169	obesity	476,941	Shiri-Sverdlov R et al. Identification of TUB as a novel candidate gene influencing body weight in humans. <i>Diabetes.</i> 2006 Feb;55(2):385-9. PMID: 16443771
TUB	rs4929949	11	8561169	obesity	476,941	Snieder H et al. TUB is a candidate gene for late-onset obesity in women. <i>Diabetologia.</i> 2008 Jan;51(1):54-61. PMID: 17955208
TUB	rs4929949	11	8561169	obesity	476,941	Stevenson DA et al. Unexpected death and critical illness in Prader-Willi syndrome: report of ten individuals. <i>Am J Med Genet A.</i> 2004 Jan 15;124A(2):158-64. PMID: 14699614
TUB	rs4929949	11	8561169	obesity	476,941	Stratigopoulos G et al. Regulation of Fto/Ftm gene expression in mice and humans. <i>Am J Physiol Regul Integr Comp Physiol.</i> 2008 Apr;294(4):R1185-96. PMID: 18256137
TUB	rs4929949	11	8561169	obesity	476,941	Stretton C et al. Expression and modulation of TUB by insulin and thyroid hormone in primary rat and murine 3T3-L1 adipocytes. <i>Biochem Biophys Res Commun.</i> 2009 Dec 25;390(4):1328-33. PMID: 19887065
TUB	rs4929949	11	8561169	obesity	476,941	Wang Y et al. Defective carbohydrate metabolism in mice homozygous for the tubby mutation. <i>Physiol Genomics.</i> 2006 Oct 11;27(2):131-40. PMID: 16849632
TUB	rs4929949	11	8561169	obesity	476,941	Way JM et al. Adipose tissue resistin expression is severely suppressed in obesity and stimulated by peroxisome proliferator-activated receptor gamma agonists. <i>J Biol Chem.</i> 2001 Jul 13;276(28):25651-3. PMID: 11373275
TUB	rs4929949	11	8561169	obesity	476,941	Xi Q et al. Mutation screen of the TUB gene in patients with retinitis pigmentosa and Leber congenital amaurosis. <i>Exp Eye Res.</i> 2006 Sep;83(3):569-73. PMID: 16643894
TUB	rs4929949	11	8561169	obesity	476,941	Xu XM et al. [Physiological and molecular control of lipid accumulation in <i>Caenorhabditis elegans</i>]. <i>Sheng Li Ke Xue Jin Zhan.</i> 2009 Apr;40(2):129-34. PMID: 19558141
BDNF	rs10767664	11	27682562	obesity, bmi	0	Araya AV et al. Evaluation of the effect of caloric restriction on serum BDNF in overweight and obese subjects: preliminary evidences. <i>Endocrine.</i> 2008 Jun;33(3):300-4. PMID: 19012000
BDNF	rs10767664	11	27682562	obesity, bmi	0	Arumugam TV et al. Age and energy intake interact to modify cell stress pathways and stroke outcome. <i>Ann Neurol.</i> 2010 Jan;67(1):41-52. PMID: 20186857
BDNF	rs10767664	11	27682562	obesity, bmi	0	Beckers S et al. Association of the BDNF Val66Met variation with obesity in women. <i>Mol Genet Metab.</i> 2008 Sep-Oct;95(1-2):110-2. PMID: 18667348
BDNF	rs10767664	11	27682562	obesity, bmi	0	Beckers S et al. The role of the leptin-melanocortin signalling pathway in the control of food intake. <i>Crit Rev Eukaryot Gene Expr.</i> 2009;19(4):267-87. PMID: 19817705
BDNF	rs10767664	11	27682562	obesity, bmi	0	Beyer CE et al. Depression-like phenotype following chronic CB(1) receptor antagonism. <i>Neurobiol Dis.</i> 2010 Apr 8; PMID: 20381618
BDNF	rs10767664	11	27682562	obesity, bmi	0	Byerly MS et al. Effects of BDNF, T3, and corticosterone on expression of the hypothalamic obesity gene network in vivo and in vitro. <i>Am J Physiol Regul Integr Comp Physiol.</i> 2009 Apr;296(4):R1180-9. PMID: 19158410
BDNF	rs10767664	11	27682562	obesity, bmi	0	Cao L et al. Molecular therapy of obesity and diabetes by a physiological autoregulatory approach. <i>Nat Med.</i> 2009 Apr;15(4):447-54. PMID: 19270710
BDNF	rs10767664	11	27682562	obesity, bmi	0	Chaldakov GN et al. NGF and BDNF: from nerves to adipose tissue, from neurokines to metabokines. <i>Riv Psichiatr.</i> 2009 Mar-Apr;44(2):79-87. PMID: 20066808
BDNF	rs10767664	11	27682562	obesity, bmi	0	Cordeira JW et al. Brain-derived neurotrophic factor regulates hedonic feeding by acting on the mesolimbic dopamine system. <i>J Neurosci.</i> 2010 Feb 17;30(7):2533-41. PMID: 20164338
BDNF	rs10767664	11	27682562	obesity, bmi	0	Correia CT et al. Pharmacogenetics of risperidone therapy in autism: association analysis of eight candidate genes with drug efficacy and adverse drug reactions. <i>Pharmacogenomics J.</i> 2009 Dec 8; PMID: 19997080

Gene	index SNP	Chr	Position	OMIM terms	Distance (nt)*	Pubmed hit with search terms "obesity" or "BMI"
BDNF	rs10767664	11	27682562	obesity, bmi	0	Ehrlich S et al. Serum brain-derived neurotrophic factor and peripheral indicators of the serotonin system in underweight and weight-recovered adolescent girls and women with anorexia nervosa. <i>J Psychiatry Neurosci</i> . 2009 Jul;34(4):323-9. PMID: 19568484
BDNF	rs10767664	11	27682562	obesity, bmi	0	Friedel S et al. Mutation screen of the brain derived neurotrophic factor gene (BDNF): identification of several genetic variants and association studies in patients with obesity, eating disorders, and attention- deficit/hyperactivity disorder. <i>Am J Med Genet B Neuropsychiatr Genet</i> . 2005 Jan 5;132B(1):96-9. PMID: 15457498
BDNF	rs10767664	11	27682562	obesity, bmi	0	Friedman JM et al. Obesity: Causes and control of excess body fat. <i>Nature</i> . 2009 May 21;459(7245):340-2. PMID: 19458707
BDNF	rs10767664	11	27682562	obesity, bmi	0	Golden E et al. Circulating brain-derived neurotrophic factor and indices of metabolic and cardiovascular health: data from the Baltimore Longitudinal Study of Aging. <i>PLoS One</i> . 2010 Apr 9;5(4):e10099. PMID: 20404913
BDNF	rs10767664	11	27682562	obesity, bmi	0	Gray J et al. Hyperphagia, severe obesity, impaired cognitive function, and hyperactivity associated with functional loss of one copy of the brain- derived neurotrophic factor (BDNF) gene. <i>Diabetes</i> . 2006 Dec;55(12):3366-71. PMID: 17130481
BDNF	rs10767664	11	27682562	obesity, bmi	0	Gunstad J et al. BDNF Val66Met polymorphism is associated with body mass index in healthy adults. <i>Neuropsychobiology</i> . 2006;53(3):153-6. PMID: 16707914
BDNF	rs10767664	11	27682562	obesity, bmi	0	Han JC et al. Brain-derived neurotrophic factor and obesity in the WAGR syndrome. <i>N Engl J Med</i> . 2008 Aug 28;359(9):918-27. PMID: 18753648
BDNF	rs10767664	11	27682562	obesity, bmi	0	Han JC et al. Lower Brain-Derived Neurotrophic Factor in Patients with Prader-Willi Syndrome Compared to Obese and Lean Control Subjects. <i>J Clin Endocrinol Metab</i> . 2010 Apr 28;. PMID: 20427492
BDNF	rs10767664	11	27682562	obesity, bmi	0	Kaplan AS et al. A DRD4/BDNF gene-gene interaction associated with maximum BMI in women with bulimia nervosa. <i>Int J Eat Disord</i> . 2008 Jan;41(1):22-8. PMID: 17922530
BDNF	rs10767664	11	27682562	obesity, bmi	0	Kohjima M et al. Increased food intake leads to obesity and insulin resistance in the tg2576 Alzheimer's disease mouse model. <i>Endocrinology</i> . 2010 Apr;151(4):1532-40. PMID: 20176720
BDNF	rs10767664	11	27682562	obesity, bmi	0	Krabbe KS et al. Brain-derived neurotrophic factor (BDNF) and type 2 diabetes. <i>Diabetologia</i> . 2007 Feb;50(2):431-8. PMID: 17151862
BDNF	rs10767664	11	27682562	obesity, bmi	0	Lee YS et al. The role of leptin-melanocortin system and human weight regulation: lessons from experiments of nature. <i>Ann Acad Med Singapore</i> . 2009 Jan;38(1):34-11. PMID: 19221669
BDNF	rs10767664	11	27682562	obesity, bmi	0	Luberg K et al. Human TrkB gene: novel alternative transcripts, protein isoforms and expression pattern in the prefrontal cerebral cortex during postnatal development. <i>J Neurochem</i> . 2010 May;113(4):952-64. PMID: 20193039
BDNF	rs10767664	11	27682562	obesity, bmi	0	Merhi ZO et al. Plasma brain-derived neurotrophic factor in women after bariatric surgery: a pilot study. <i>Fertil Steril</i> . 2009 Apr;91(4 Suppl):1544-8. PMID: 18950757
BDNF	rs10767664	11	27682562	obesity, bmi	0	Pedersen BK et al. Role of exercise-induced brain-derived neurotrophic factor production in the regulation of energy homeostasis in mammals. <i>Exp Physiol</i> . 2009 Dec;94(12):1153-60. PMID: 19748969
BDNF	rs10767664	11	27682562	obesity, bmi	0	Rask-Andersen M et al. Molecular mechanisms underlying anorexia nervosa: focus on human gene association studies and systems controlling food intake. <i>Brain Res Rev</i> . 2010 Mar;62(2):147-64. PMID: 19931559
BDNF	rs10767664	11	27682562	obesity, bmi	0	Shugart YY et al. Two British women studies replicated the association between the Val66Met polymorphism in the brain-derived neurotrophic factor (BDNF) and BMI. <i>Eur J Hum Genet</i> . 2009 Aug;17(8):1050-5. PMID: 19209189
BDNF	rs10767664	11	27682562	obesity, bmi	0	Uçeyler N et al. Lack of the serotonin transporter in mice reduces locomotor activity and leads to gender-dependent late onset obesity. <i>Int J Obes (Lond)</i> . 2010 Apr;34(4):701-11. PMID: 20084070
BDNF	rs10767664	11	27682562	obesity, bmi	0	Wang C et al. Brain-derived neurotrophic factor (BDNF) in the hypothalamic ventromedial nucleus increases energy expenditure. <i>Brain Res</i> . 2010 Apr 14;. PMID: 20398635
BDNF	rs10767664	11	27682562	obesity, bmi	0	Wang C et al. Chronic administration of brain-derived neurotrophic factor in the hypothalamic paraventricular nucleus reverses obesity induced by high fat diet. <i>Am J Physiol Regul Integr Comp Physiol</i> . 2010 Feb 17;. PMID: 20164202
BDNF	rs10767664	11	27682562	obesity, bmi	0	Xu L et al. TrkB Agonist Antibody Dose-Dependently Raises Blood Pressure in Mice With Diet-Induced Obesity. <i>Am J Hypertens</i> . 2010 Mar 18;. PMID: 20300069
BDNF	rs10767664	11	27682562	obesity, bmi	0	Yu Y et al. Energy-restricted pair-feeding normalizes low levels of brain- derived neurotrophic factor/tyrosine kinase B mRNA expression in the hippocampus, but not ventromedial hypothalamic nucleus, in diet-induced obese mice. <i>Neuroscience</i> . 2009 May 5;160(2):295-306. PMID: 19217934
BDNF	rs10767664	11	27682562	obesity, bmi	0	Zhang XY et al. BDNF levels and genotype are associated with antipsychotic- induced weight gain in patients with chronic schizophrenia. <i>Neuropsychopharmacology</i> . 2008 Aug;33(9):2200-5. PMID: 17987059
NR1H3	rs3817334	11	47607569	no match	360,592	Auboeuf D et al. Tissue distribution and quantification of the expression of mRNAs of peroxisome proliferator-activated receptors and liver X receptor- alpha in humans: no alteration in adipose tissue of obese and NIDDM patients. <i>Diabetes</i> . 1997 Aug;46(8):1319-27. PMID: 9231657
NR1H3	rs3817334	11	47607569	no match	360,592	Dahlman I et al. Liver X receptor gene polymorphisms and adipose tissue expression levels in obesity. <i>Pharmacogenet Genomics</i> . 2006 Dec;16(12):881-9. PMID: 17108812
NR1H3	rs3817334	11	47607569	no match	360,592	Jiang ZY et al. Increased expression of LXR alpha, ABCG5, ABCG8, and SR-BI in the liver from normolipidemic, nonobese Chinese gallstone patients. <i>J Lipid Res</i> . 2008 Feb;49(2):464-72. PMID: 18007013
NR1H3	rs3817334	11	47607569	no match	360,592	Phelan CA et al. Selective partial agonism of liver X receptor alpha is related to differential corepressor recruitment. <i>Mol Endocrinol</i> . 2008 Oct;22(10):2241-9. PMID: 18669643
RACGAP1	rs7138803	12	48533735	no match	135,477	Niyya F et al. Inhibition of cyclin-dependent kinase 1 induces cytokinesis without chromosome segregation in an ECT2 and MgcRacGAP-dependent manner. <i>J Biol Chem</i> . 2005 Oct 28;280(43):36502-9. PMID: 16118207
RACGAP1	rs7138803	12	48533735	no match	135,477	Obuse C et al. Proteomics analysis of the centromere complex from HeLa interphase cells: UV-damaged DNA binding protein 1 (DDB-1) is a component of the CEN-complex, while BMI-1 is transiently co-localized with the centromeric region in interphase. <i>Genes Cells</i> . 2004 Feb;9(2):105-20. PMID: 15009096

Gene	index SNP	Chr	Position	OMIM terms	Distance (nt)*	Pubmed hit with search terms "obesity" or "BMI"
GPD1	rs7138803	12	48533735	no match	250,333	Guindalini C et al. The influence of obstructive sleep apnea on the expression of glycerol-3-phosphate dehydrogenase 1 gene. <i>Exp Biol Med</i> (Maywood). 2010 Jan;235(1):52-6. PMID: 20404019
GPD1	rs7138803	12	48533735	no match	250,333	Park JJ et al. GRB14, GPD1, and GDF8 as potential network collaborators in weight loss-induced improvements in insulin action in human skeletal muscle. <i>Physiol Genomics</i> . 2006 Oct 11;27(2):114-21. PMID: 16849634
GPD1	rs7138803	12	48533735	no match	250,333	Poulain-Godefroy O et al. Inflammation is associated with a decrease of lipogenic factors in omental fat in women. <i>Am J Physiol Regul Integr Comp Physiol</i> . 2008 Jul;295(1):R1-7. PMID: 18448614
GPD1	rs7138803	12	48533735	no match	250,333	Swierczynski J et al. Enhanced glycerol 3-phosphate dehydrogenase activity in adipose tissue of obese humans. <i>Mol Cell Biochem</i> . 2003 Dec;254(1-2):55-9. PMID: 14674682
GP2	rs12444979	16	19841101	no match	388,211	Berkane AA et al. Proteomic of lipid rafts in the exocrine pancreas from diet-induced obese rats. <i>Biochem Biophys Res Commun</i> . 2007 Apr 13;355(3):813-9. PMID: 17320817
SH2B1	rs7359397	16	28793160	obesity	133	Bochukova EG et al. Large, rare chromosomal deletions associated with severe early-onset obesity. <i>Nature</i> . 2010 Feb 4;463(7281):666-70. PMID: 19966786
SH2B1	rs7359397	16	28793160	obesity	133	Burwell RG et al. Pathogenesis of adolescent idiopathic scoliosis in girls - a double neuro-osseous theory involving disharmony between two nervous systems, somatic and autonomic expressed in the spine and trunk: possible dependency on sympathetic nervous system and hormones with implications for medical therapy. <i>Scoliosis</i> . 2009 Oct 31;4:24. PMID: 19878575
SH2B1	rs7359397	16	28793160	obesity	133	Duan C et al. [Molecular mechanism of SH2B1 in regulating JAK2/IRS2 during obesity development.]. <i>Zhong Nan Da Xue Xue Bao Yi Xue Ban</i> . 2010 Mar;35(3):209-14. PMID: 20360640
SH2B1	rs7359397	16	28793160	obesity	133	Ghanayem BI et al. Diet-induced obesity in male mice is associated with reduced fertility and potentiation of acrylamide-induced reproductive toxicity. <i>Biol Reprod</i> . 2010 Jan;82(1):96-104. PMID: 19696015
SH2B1	rs7359397	16	28793160	obesity	133	Maures TJ et al. SH2B1 (SH2-B) and JAK2: a multifunctional adaptor protein and kinase made for each other. <i>Trends Endocrinol Metab</i> . 2007 Jan- Feb;18(1):38-45. PMID: 17140804
SH2B1	rs7359397	16	28793160	obesity	133	Morris DL et al. Recent advances in understanding leptin signaling and leptin resistance. <i>Am J Physiol Endocrinol Metab</i> . 2009 Dec;297(6):E1247-59. PMID: 19724019
SH2B1	rs7359397	16	28793160	obesity	133	Nelms K et al. Alternative splicing, gene localization, and binding of SH2-B to the insulin receptor kinase domain. <i>Mamm Genome</i> . 1999 Dec;10(12):1160-7. PMID: 10594240
SH2B1	rs7359397	16	28793160	obesity	133	Ren D et al. Neuronal SH2B1 is essential for controlling energy and glucose homeostasis. <i>J Clin Invest</i> . 2007 Feb;117(2):397-406. PMID: 17235396
SH2B1	rs7359397	16	28793160	obesity	133	Rider L et al. Adapter protein SH2B1beta cross-links actin filaments and regulates actin cytoskeleton. <i>Mol Endocrinol</i> . 2009 Jul;23(7):1065-76. PMID: 19342444
SH2B1	rs7359397	16	28793160	obesity	133	Senior K et al. Another piece in the molecular puzzle of obesity. <i>Drug Discov Today</i> . 2005 Nov 15;10(22):1488. PMID: 16257366
SH2B1	rs7359397	16	28793160	obesity	133	Song W et al. SH2B regulation of growth, metabolism, and longevity in both insects and mammals. <i>Cell Metab</i> . 2010 May 5;11(5):427-37. PMID: 20417156
CD19	rs7359397	16	28793160	no match	57,601	Asprey JM et al. Effect on immune indices of preoperative intravenous glutamine dipeptide supplementation in malnourished abdominal surgery patients in the preoperative and postoperative periods. <i>Nutrition</i> . 2009 Sep;25(9):920-5. PMID: 19647623
CD19	rs7359397	16	28793160	no match	57,601	Backé E et al. Immunological biomarkers in salt miners exposed to salt dust, diesel exhaust and nitrogen oxides. <i>Int Arch Occup Environ Health</i> . 2004 Jun;77(5):319-27. PMID: 15197551
CD19	rs7359397	16	28793160	no match	57,601	Başoğlu OK et al. [T-lymphocyte subgroups and tuberculin skin test reactivity in patients with chronic renal failure]. <i>Tuberk Toraks</i> . 2006;54(1):5-10. PMID: 16615011
CD19	rs7359397	16	28793160	no match	57,601	Grzegorzewska AE et al. Total lymphocyte count and subpopulation lymphocyte counts in relation to dietary intake and nutritional status of peritoneal dialysis patients. <i>Adv Perit Dial</i> . 2005;21:35-40. PMID: 16686282
CD19	rs7359397	16	28793160	no match	57,601	Haskell BD et al. The diabetes-prone NZO/HLLT strain. I. Immunophenotypic comparison to the related NZB/BINJ and NZW/LacJ strains. <i>Lab Invest</i> . 2002 Jul;82(7):833-42. PMID: 12118085
CD19	rs7359397	16	28793160	no match	57,601	Hudson CA et al. The dietary supplement ephedrine induces beta-adrenergic mediated exacerbation of systemic lupus erythematosus in NZM391 mice. <i>Lupus</i> . 2005;14(4):293-307. PMID: 15864916
CD19	rs7359397	16	28793160	no match	57,601	Marcos A et al. Evaluation of nutritional status by immunologic assessment in bulimia nervosa: influence of body mass index and vomiting episodes. <i>Am J Clin Nutr</i> . 1997 Aug;66(2):491S-497S. PMID: 9250137
CD19	rs7359397	16	28793160	no match	57,601	Martinka E et al. [Glutamic acid decarboxylase autoantibodies (antiGAD-Ab) in patients with non-insulin dependent diabetes mellitus (NIDDM)]. <i>Vnitr Lek</i> . 1998 Jan;44(1):17-22. PMID: 9750478
CD19	rs7359397	16	28793160	no match	57,601	Merhi ZO et al. Effect of bariatric surgery on peripheral blood lymphocyte subsets in women. <i>Surg Obes Relat Dis</i> . 2009 Mar-Apr;5(2):165-71. PMID: 18996763
CD19	rs7359397	16	28793160	no match	57,601	Santagostino A et al. An Italian national multicenter study for the definition of reference ranges for normal values of peripheral blood lymphocyte subsets in healthy adults. <i>Haematologica</i> . 1999 Jun;84(6):499-504. PMID: 10366792
CD19	rs7359397	16	28793160	no match	57,601	Sayarlioglu H et al. Nutritional status and immune functions in maintenance hemodialysis patients. <i>Mediators Inflamm</i> . 2006;2006(1):20264. PMID: 16864899
SULT1A1	rs7359397	16	28793160	no match	250,793	Glatt H et al. Pharmacogenetics of soluble sulfotransferases (SULTs). <i>Naunyn Schmiedebergs Arch Pharmacol</i> . 2004 Jan;369(1):55-68. PMID: 14600802
SULT1A1	rs7359397	16	28793160	no match	250,793	Hu MB et al. [Study on the relationship between polymorphisms of genes (CYP17, CYP19 and SULT1A1) and susceptibility to breast cancer in Chinese women]. <i>Zhonghua Liu Xing Bing Xue Za Zhi</i> . 2006 Apr;27(4):351-5. PMID: 16875543
SULT1A1	rs7359397	16	28793160	no match	250,793	Maglich JM et al. The nuclear receptor CAR is a regulator of thyroid hormone metabolism during caloric restriction. <i>J Biol Chem</i> . 2004 May 7;279(19):19832-8. PMID: 15004031

Gene	index SNP	Chr	Position	OMIM terms	Distance (nt)*	Pubmed hit with search terms "obesity" or "BMI"
SULT1A1	rs7359397	16	28793160	no match	250,793	Yang G et al. Modifying effects of sulfotransferase 1A1 gene polymorphism on the association of breast cancer risk with body mass index or endogenous steroid hormones. <i>Breast Cancer Res Treat.</i> 2005 Nov;94(1):63-70. PMID: 16175316
APOB48R	rs7359397	16	28793160	no match	375,377	Lumeng CN et al. Increased inflammatory properties of adipose tissue macrophages recruited during diet-induced obesity. <i>Diabetes.</i> 2007 Jan;56(1):16-23. PMID: 17192460
FTO	rs1558902	16	52361075	obesity	0	Adeyemo A et al. FTO Genetic Variation and Association with Obesity in West Africans and African-Americans. <i>Diabetes.</i> 2010 Mar 18;. PMID: 20299471
FTO	rs1558902	16	52361075	obesity	0	Attaoua R et al. Association of the FTO gene with obesity and the metabolic syndrome is independent of the IRS-2 gene in the female population of Southern France. <i>Diabetes Metab.</i> 2009 Dec;35(6):476-83. PMID: 19818665
FTO	rs1558902	16	52361075	obesity	0	Bassols J et al. Placental FTO expression relates to fetal growth. <i>Int J Obes (Lond).</i> 2010 Mar 30;. PMID: 20351740
FTO	rs1558902	16	52361075	obesity	0	Chen HH et al. Severe obesity is associated with novel single nucleotide polymorphisms of the ESR1 and PPARgamma locus in Han Chinese. <i>Am J Clin Nutr.</i> 2009 Aug;90(2):255-62. PMID: 19491387
FTO	rs1558902	16	52361075	obesity	0	Grau K et al. Macronutrient-specific effect of FTO rs9939609 in response to a 10-week randomized hypo-energetic diet among obese Europeans. <i>Int J Obes (Lond).</i> 2009 Nov;33(11):1227-34. PMID: 19687793
FTO	rs1558902	16	52361075	obesity	0	Han Z et al. Crystal structure of the FTO protein reveals basis for its substrate specificity. <i>Nature.</i> 2010 Apr 22;464(7292):1205-9. PMID: 20376003
FTO	rs1558902	16	52361075	obesity	0	Hardy R et al. Life course variations in the associations between FTO and MC4R gene variants and body size. <i>Hum Mol Genet.</i> 2010 Feb 1;19(3):545-52. PMID: 19880856
FTO	rs1558902	16	52361075	obesity	0	Hinney A et al. From monogenic to polygenic obesity: recent advances. <i>Eur Child Adolesc Psychiatry.</i> 2010 Mar;19(3):297-310. PMID: 20127379
FTO	rs1558902	16	52361075	obesity	0	Ho AJ et al. A commonly carried allele of the obesity-related FTO gene is associated with reduced brain volume in the healthy elderly. <i>Proc Natl Acad Sci U S A.</i> 2010 May 4;107(18):8404-9. PMID: 20404173
FTO	rs1558902	16	52361075	obesity	0	Hubacek JA et al. A FTO variant and risk of acute coronary syndrome. <i>Clin Chim Acta.</i> 2010 Apr 1;. PMID: 20362563
FTO	rs1558902	16	52361075	obesity	0	Karasawa S et al. Association of the common fat mass and obesity associated (FTO) gene polymorphism with obesity in a Japanese population. <i>Endocr J.</i> 2010;57(4):293-301. PMID: 20051647
FTO	rs1558902	16	52361075	obesity	0	Kowalska I et al. The FTO gene modifies weight, fat mass and insulin sensitivity in women with polycystic ovary syndrome, where its role may be larger than in other phenotypes. <i>Diabetes Metab.</i> 2009 Sep;35(4):328-31. PMID: 19625203
FTO	rs1558902	16	52361075	obesity	0	Liem ET et al. Influence of common variants near INSIG2, in FTO, and near MC4R genes on overweight and the metabolic profile in adolescence: the TRAILS (TRacking Adolescents' Individual Lives Survey) Study. <i>Am J Clin Nutr.</i> 2010 Feb;91(2):321-8. PMID: 20007308
FTO	rs1558902	16	52361075	obesity	0	Meyre D et al. Prevalence of loss-of-function FTO mutations in lean and obese individuals. <i>Diabetes.</i> 2010 Jan;59(1):311-8. PMID: 19833892
FTO	rs1558902	16	52361075	obesity	0	Pettersen E et al. Genetic heterogeneity in latent autoimmune diabetes is linked to various degrees of autoimmune activity: results from the Nord-Trøndelag Health Study. <i>Diabetes.</i> 2010 Jan;59(1):302-10. PMID: 19833889
FTO	rs1558902	16	52361075	obesity	0	Reinehr T et al. Aggravating effect of INSIG2 and FTO on overweight reduction in a one-year lifestyle intervention. <i>Arch Dis Child.</i> 2009 Dec;94(12):965-7. PMID: 19224890
FTO	rs1558902	16	52361075	obesity	0	Rendo T et al. Effects of the FTO Gene on Lifestyle Intervention Studies in Children. <i>Obes Facts.</i> 2009;2(6):393-399. PMID: 20090391
FTO	rs1558902	16	52361075	obesity	0	Rodríguez-López R et al. [Association of FTO gene polymorphisms and morbid obesity in the population of Extremadura (Spain)]. <i>Endocrinol Nutr.</i> 2010 Apr 23;. PMID: 20418190
FTO	rs1558902	16	52361075	obesity	0	Rutters F et al. Genetic associations with acute stress-related changes in eating in the absence of hunger. <i>Patient Educ Couns.</i> 2010 Apr 19;. PMID: 20409671
FTO	rs1558902	16	52361075	obesity	0	Speakman JR et al. FTO effect on energy demand versus food intake. <i>Nature.</i> 2010 Apr 1;464(7289):E1; discussion E2. PMID: 20360686
FTO	rs1558902	16	52361075	obesity	0	Switonski M et al. Genetics of fat tissue accumulation in pigs: a comparative approach. <i>J Appl Genet.</i> 2010;51(2):153-168. PMID: 20453303
FTO	rs1558902	16	52361075	obesity	0	Tung YC et al. Hypothalamic-specific manipulation of Fto, the ortholog of the human obesity gene FTO, affects food intake in rats. <i>PLoS One.</i> 2010 Jan 19;5(1):e8771. PMID: 20098739
FTO	rs1558902	16	52361075	obesity	0	van den Berg L et al. Investigation of a patient with a partial trisomy 16q including the fat mass and obesity associated gene (FTO): fine mapping and FTO gene expression study. <i>Am J Med Genet A.</i> 2010 Mar;152A(3):630-7. PMID: 20186806
FTO	rs1558902	16	52361075	obesity	0	Zhang G et al. Common SNPs in FTO gene are associated with obesity related anthropometric traits in an island population from the eastern Adriatic coast of Croatia. <i>PLoS One.</i> 2010 Apr 28;5(4):e10375. PMID: 20442772
RPGRIP1L	rs1558902	16	52361075	no match	65,803	Klötting N et al. Inverse relationship between obesity and FTO gene expression in visceral adipose tissue in humans. <i>Diabetologia.</i> 2008 Apr;51(4):641-7. PMID: 18251005
RPGRIP1L	rs1558902	16	52361075	no match	65,803	Sen Gupta P et al. Can faulty antennae increase adiposity? The link between cilia proteins and obesity. <i>J Endocrinol.</i> 2009 Dec;203(3):327-36. PMID: 19460851
RPGRIP1L	rs1558902	16	52361075	no match	65,803	Stratigopoulos G et al. Regulation of Fto/Ftm gene expression in mice and humans. <i>Am J Physiol Regul Integr Comp Physiol.</i> 2008 Apr;294(4):R1185-96. PMID: 18256137
RBL2	rs1558902	16	52361075	no match	278,014	Jowett JB et al. Genetic variation at the FTO locus influences RBL2 gene expression. <i>Diabetes.</i> 2010 Mar;59(3):726-32. PMID: 20009087
MC4R	rs571312	18	55990749	obesity, bmi	198,795	Alfieri A et al. Functional analysis of melanocortin-4-receptor mutants identified in severely obese subjects living in Southern Italy. <i>Gene.</i> 2010 Jun 1;457(1-2):35-41. PMID: 20214954
MC4R	rs571312	18	55990749	obesity, bmi	198,795	Atalayer D et al. Food demand and meal size in mice with single or combined disruption of melanocortin type 3 and 4 receptors. <i>Am J Physiol Regul Integr Comp Physiol.</i> 2010 Apr 7;. PMID: 20375267
MC4R	rs571312	18	55990749	obesity, bmi	198,795	Calton MA et al. Association of functionally significant Melanocortin-4 but not Melanocortin-3 receptor mutations with severe adult obesity in a large North American case-control study. <i>Hum Mol Genet.</i> 2009 Mar 15;18(6):1140-7. PMID: 19091795

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MC4R	rs571312	18	55990749	obesity, bmi	198,795	Chen HH et al. Severe obesity is associated with novel single nucleotide polymorphisms of the ESR1 and PPARgamma locus in Han Chinese. <i>Am J Clin Nutr.</i> 2009 Aug;90(2):255-62. PMID: 19491387
MC4R	rs571312	18	55990749	obesity, bmi	198,795	Cole SA et al. Evidence that multiple genetic variants of MC4R play a functional role in the regulation of energy expenditure and appetite in Hispanic children. <i>Am J Clin Nutr.</i> 2010 Jan;91(1):191-9. PMID: 19889825
MC4R	rs571312	18	55990749	obesity, bmi	198,795	Figueroa KP et al. Genetic variance in the spinocerebellar ataxia type 2 (ATXN2) gene in children with severe early onset obesity. <i>PLoS One.</i> 2009 Dec 14;4(12):e8280. PMID: 20016785
MC4R	rs571312	18	55990749	obesity, bmi	198,795	Grant SF et al. Investigation of the locus near MC4R with childhood obesity in Americans of European and African ancestry. <i>Obesity (Silver Spring).</i> 2009 Jul;17(7):1461-5. PMID: 19265794
MC4R	rs571312	18	55990749	obesity, bmi	198,795	Guan XM et al. Regulation of energy homeostasis by bombesin receptor subtype-3: selective receptor agonists for the treatment of obesity. <i>Cell Metab.</i> 2010 Feb 3;11(2):101-12. PMID: 20096642
MC4R	rs571312	18	55990749	obesity, bmi	198,795	Hardy R et al. Life course variations in the associations between FTO and MC4R gene variants and body size. <i>Hum Mol Genet.</i> 2010 Feb 1;19(3):545-52. PMID: 19880856
MC4R	rs571312	18	55990749	obesity, bmi	198,795	Hetherington MM et al. Gene-environment interactions in obesity. <i>Forum Nutr.</i> 2010;63:195-203. PMID: 19955787
MC4R	rs571312	18	55990749	obesity, bmi	198,795	Hinney A et al. Three at one swoop!. <i>Obes Facts.</i> 2009;2(1):3-8. PMID: 20054198
MC4R	rs571312	18	55990749	obesity, bmi	198,795	Hochberg I et al. Hypothalamic Obesity. <i>Endocr Dev.</i> 2010;17:185-196. PMID: 19955767
MC4R	rs571312	18	55990749	obesity, bmi	198,795	Kang SJ et al. Genome-wide association of anthropometric traits in African- and African-derived populations. <i>Hum Mol Genet.</i> 2010 May 6; PMID: 20400458
MC4R	rs571312	18	55990749	obesity, bmi	198,795	Kring SI et al. Common variants near MC4R in relation to body fat, body fat distribution, metabolic traits and energy expenditure. <i>Int J Obes (Lond).</i> 2010 Jan;34(1):182-9. PMID: 19844209
MC4R	rs571312	18	55990749	obesity, bmi	198,795	Krishna R et al. Potent and selective agonism of the melanocortin receptor 4 with MK-0493 does not induce weight loss in obese human subjects: energy intake predicts lack of weight loss efficacy. <i>Clin Pharmacol Ther.</i> 2009 Dec;86(6):659-66. PMID: 19741604
MC4R	rs571312	18	55990749	obesity, bmi	198,795	Park TJ et al. High-level expression and purification of the second transmembrane domain of wild-type and mutant human melanocortin-4 receptor for solid-state NMR structural studies. <i>Protein Expr Purif.</i> 2008 Dec;62(2):139-45. PMID: 18809499
MC4R	rs571312	18	55990749	obesity, bmi	198,795	Pichler M et al. Association of the melanocortin-4 receptor V103I polymorphism with dietary intake in severely obese persons. <i>Am J Clin Nutr.</i> 2008 Sep;88(3):797-800. PMID: 18779298
MC4R	rs571312	18	55990749	obesity, bmi	198,795	Rask-Andersen M et al. Molecular mechanisms underlying anorexia nervosa: focus on human gene association studies and systems controlling food intake. <i>Brain Res Rev.</i> 2010 Mar;62(2):147-64. PMID: 19931559
MC4R	rs571312	18	55990749	obesity, bmi	198,795	Rediger A et al. Heterodimerization of Hypothalamic G-Protein-Coupled Receptors Involved in Weight Regulation. <i>Obes Facts.</i> 2009;2(2):80-86. PMID: 20054210
MC4R	rs571312	18	55990749	obesity, bmi	198,795	Roth CL et al. A novel melanocortin-4 receptor gene mutation in a female patient with severe childhood obesity. <i>Endocrine.</i> 2009 Aug;36(1):52-9. PMID: 19214805
MC4R	rs571312	18	55990749	obesity, bmi	198,795	Sayk F et al. Sympathetic function in human carriers of melanocortin-4 receptor gene mutations. <i>J Clin Endocrinol Metab.</i> 2010 Apr;95(4):1998-2002. PMID: 20147580
MC4R	rs571312	18	55990749	obesity, bmi	198,795	Stokić E et al. Polymorphism Val103Ile of the melanocortin-4 receptor gene in the Serbian population. <i>Mol Biol Rep.</i> 2010 Jan;37(1):33-7. PMID: 19283510
MC4R	rs571312	18	55990749	obesity, bmi	198,795	Switonski M et al. Genetics of fat tissue accumulation in pigs: a comparative approach. <i>J Appl Genet.</i> 2010;51(2):153-168. PMID: 20453303
MC4R	rs571312	18	55990749	obesity, bmi	198,795	Tan K et al. Functional characterization and structural modeling of obesity associated mutations in the melanocortin 4 receptor. <i>Endocrinology.</i> 2009 Jan;150(1):114-25. PMID: 18801902
MC4R	rs571312	18	55990749	obesity, bmi	198,795	Tao YX et al. The Melanocortin-4 Receptor: Physiology, Pharmacology, and Pathophysiology. <i>Endocr Rev.</i> 2010 Feb 26; PMID: 20190196
MC4R	rs571312	18	55990749	obesity, bmi	198,795	Tolson KP et al. Postnatal Sim1 deficiency causes hyperphagic obesity and reduced Mc4r and oxytocin expression. <i>J Neurosci.</i> 2010 Mar 10;30(10):3803-12. PMID: 20220015
MC4R	rs571312	18	55990749	obesity, bmi	198,795	Yurtcu E et al. Melanocortin-4 receptor gene polymorphisms in obese patients. <i>Biochem Genet.</i> 2009 Apr;47(3-4):295-300. PMID: 19184404
CEBPG	rs29941	19	39001372	no match	435,940	Szczerbal I et al. Chromosomal localization of nine porcine genes encoding transcription factors involved in adipogenesis. <i>Cytogenet Genome Res.</i> 2008;121(1):50-4. PMID: 18544926
GIPR	rs2287019	19	50894012	obesity	16,455	Fulurija A et al. Vaccination against GIP for the treatment of obesity. <i>PLoS One.</i> 2008 Sep 9;3(9):e3163. PMID: 18779862
GIPR	rs2287019	19	50894012	obesity	16,455	Hansotia T et al. Extraprostatic incretin receptors modulate glucose homeostasis, body weight, and energy expenditure. <i>J Clin Invest.</i> 2007 Jan;117(1):143-52. PMID: 17187081
GIPR	rs2287019	19	50894012	obesity	16,455	Harada N et al. A novel GIP receptor splice variant influences GIP sensitivity of pancreatic beta-cells in obese mice. <i>Am J Physiol Endocrinol Metab.</i> 2008 Jan;294(1):E61-8. PMID: 17971513
GIPR	rs2287019	19	50894012	obesity	16,455	Isken F et al. Deficiency of glucose-dependent insulinotropic polypeptide receptor prevents ovariectomy-induced obesity in mice. <i>Am J Physiol Endocrinol Metab.</i> 2008 Aug;295(2):E350-5. PMID: 18505834
GIPR	rs2287019	19	50894012	obesity	16,455	Miyawaki K et al. Inhibition of gastric inhibitory polypeptide signaling prevents obesity. <i>Nat Med.</i> 2002 Jul;8(7):738-42. PMID: 12068290
GIPR	rs2287019	19	50894012	obesity	16,455	Naitoh R et al. Inhibition of GIP signaling modulates adiponectin levels under high-fat diet in mice. <i>Biochem Biophys Res Commun.</i> 2008 Nov 7;376(1):21-5. PMID: 18723001
GIPR	rs2287019	19	50894012	obesity	16,455	Prabakaran D et al. Glucose-dependent insulinotropic polypeptide stimulates the proliferation of colorectal cancer cells. <i>Regul Pept.</i> 2010 Apr 28; PMID: 20433877
GIPR	rs2287019	19	50894012	obesity	16,455	Rudovich N et al. GIP receptor mRNA expression in different fat tissue depots in postmenopausal non-diabetic women. <i>Regul Pept.</i> 2007 Aug 16;142(3):138-45. PMID: 17395281
GIPR	rs2287019	19	50894012	obesity	16,455	Song DH et al. Glucose-dependent insulinotropic polypeptide enhances adipocyte development and glucose uptake in part through Akt activation. <i>Gastroenterology.</i> 2007 Dec;133(6):1796-805. PMID: 18054552

Gene	index SNP	Chr	Position	OMIM terms	Distance (nt)*	Pubmed hit with search terms "obesity" or "BMI"
GIPR	rs2287019	19	50894012	obesity	16,455	Yamada Y et al. Physiology of GIP--a lesson from GIP receptor knockout mice. <i>Horm Metab Res.</i> 2004 Nov-Dec;36(11-12):771-4. PMID: 15655707
GIPR	rs2287019	19	50894012	obesity	16,455	Zhou H et al. Gastric inhibitory polypeptide modulates adiposity and fat oxidation under diminished insulin action. <i>Biochem Biophys Res Commun.</i> 2005 Sep 30;335(3):937-42. PMID: 16105663
DMPK	rs2287019	19	50894012	no match	70,804	Block MH et al. Discovery and optimization of a series of carbazole ureas as NPY5 antagonists for the treatment of obesity. <i>J Med Chem.</i> 2002 Aug 1;45(16):3509-23. PMID: 12139462
DMPK	rs2287019	19	50894012	no match	70,804	Sun L et al. An in vivo microdialysis coupled with liquid chromatography/tandem mass spectrometry study of cortisol metabolism in monkey adipose tissue. <i>Anal Biochem.</i> 2008 Oct 15;381(2):214-23. PMID: 18638442
FOXA3	rs2287019	19	50894012	no match	165,346	Brannian JD et al. Pioglitazone administration alters ovarian gene expression in aging obese lethal yellow mice. <i>Reprod Biol Endocrinol.</i> 2008 Mar 18;6:10. PMID: 18348723
VASP	rs2287019	19	50894012	no match	171,932	Cuisset T et al. Relationship between aspirin and clopidogrel responses in acute coronary syndrome and clinical predictors of non response. <i>Thromb Res.</i> 2009 Feb;123(4):597-603. PMID: 18499233
VASP	rs2287019	19	50894012	no match	171,932	Rider L et al. Adapter protein SH2B1beta cross-links actin filaments and regulates actin cytoskeleton. <i>Mol Endocrinol.</i> 2009 Jul;23(7):1065-76. PMID: 19342444
VASP	rs2287019	19	50894012	no match	171,932	Russo I et al. Platelet resistance to the antiaggregatory cyclic nucleotides in central obesity involves reduced phosphorylation of vasodilator-stimulated phosphoprotein. <i>Clin Chem.</i> 2007 Jun;53(6):1053-60. PMID: 17463178
NANOS2	rs2287019	19	50894012	no match	214,301	Ding F et al. Neonatal maternal deprivation response and developmental changes in gene expression revealed by hypothalamic gene expression profiling in mice. <i>PLoS One.</i> 2010 Feb 24;5(2):e9402. PMID: 20195375
FOSB	rs2287019	19	50894012	no match	223,735	Rowe GC et al. Increased energy expenditure and insulin sensitivity in the high bone mass DeltaFosB transgenic mice. <i>Endocrinology.</i> 2009 Jan;150(1):135-43. PMID: 18772235
FOSB	rs2287019	19	50894012	no match	223,735	Teegarden SL et al. Delta FosB-mediated alterations in dopamine signaling are normalized by a palatable high-fat diet. <i>Biol Psychiatry.</i> 2008 Dec 1;64(11):941-50. PMID: 18657800
ERCC1	rs2287019	19	50894012	no match	274,995	Borrmann L et al. High mobility group A2 protein and its derivatives bind a specific region of the promoter of DNA repair gene ERCC1 and modulate its activity. <i>Nucleic Acids Res.</i> 2003 Dec 1;31(23):6841-51. PMID: 14627817
ERCC1	rs2287019	19	50894012	no match	274,995	Hooker S et al. NAT2 and NER genetic variants and sporadic prostate cancer susceptibility in African Americans. <i>Prostate Cancer Prostatic Dis.</i> 2008;11(4):349-56. PMID: 18026184
ERCC1	rs2287019	19	50894012	no match	274,995	Weiss JM et al. Nucleotide excision repair genotype and the incidence of endometrial cancer: effect of other risk factors on the association. <i>Gynecol Oncol.</i> 2006 Dec;103(3):891-6. PMID: 16806437
ERCC1	rs2287019	19	50894012	no match	274,995	Zhou PK et al. Interaction between viral proteins and hosts and its disturbance in the cellular responses to ionising radiation. <i>Int J Radiat Biol.</i> 2009 Jul;85(7):587-97. PMID: 19479603
ERCC2	rs2287019	19	50894012	no match	328,327	Lee SA et al. Obesity and genetic polymorphism of ERCC2 and ERCC4 as modifiers of risk of breast cancer. <i>Exp Mol Med.</i> 2005 Apr 30;37(2):86-90. PMID: 15886521
ERCC2	rs2287019	19	50894012	no match	328,327	Lin J et al. Case-control analysis of nucleotide excision repair pathway and the risk of renal cell carcinoma. <i>Carcinogenesis.</i> 2008 Nov;29(11):2112-9. PMID: 18711149
ERCC2	rs2287019	19	50894012	no match	328,327	Weiss JM et al. Nucleotide excision repair genotype and the incidence of endometrial cancer: effect of other risk factors on the association. <i>Gynecol Oncol.</i> 2006 Dec;103(3):891-6. PMID: 16806437
GRLF1	rs3810291	19	52260843	no match	60,670	Rutters F et al. Leptin-adiposity relationship changes, plus behavioral and parental factors, are involved in the development of body weight in a Dutch children cohort. <i>Physiol Behav.</i> 2008 Mar 18;93(4-5):967-74. PMID: 18282590
GRLF1	rs3810291	19	52260843	no match	60,670	Vogels N et al. Relation of weight maintenance and dietary restraint to peroxisome proliferator-activated receptor gamma2, glucocorticoid receptor, and ciliary neurotrophic factor polymorphisms. <i>Am J Clin Nutr.</i> 2005 Oct;82(4):740-6. PMID: 16210701
GPR77	rs3810291	19	52260843	no match	271,368	MacLaren RE et al. Association of adipocyte genes with ASP expression: a microarray analysis of subcutaneous and omental adipose tissue in morbidly obese subjects. <i>BMC Med Genomics.</i> 2010 Jan 27;3:3. PMID: 20105310
GPR77	rs3810291	19	52260843	no match	271,368	Paglialunga S et al. Reduced adipose tissue triglyceride synthesis and increased muscle fatty acid oxidation in C5L2 knockout mice. <i>J Endocrinol.</i> 2007 Aug;194(2):293-304. PMID: 17641279
GPR77	rs3810291	19	52260843	no match	271,368	Wen Y et al. Palmitate and oleate induction of acylation stimulating protein resistance in 3T3-L1 adipocytes and preadipocytes. <i>J Cell Biochem.</i> 2008 May 15;104(2):391-401. PMID: 18004729
GPR77	rs3810291	19	52260843	no match	271,368	Wen Y et al. Sex steroid hormones induce acylation stimulating protein resistance in 3T3-L1 adipocytes. <i>J Cell Biochem.</i> 2008 Oct 1;105(2):404-13. PMID: 18615583
NAPA	rs3810291	19	52260843	no match	421,860	Balducci S et al. Exercise training can modify the natural history of diabetic peripheral neuropathy. <i>J Diabetes Complications.</i> 2006 Jul-Aug;20(4):216-23. PMID: 16798472
NAPA	rs3810291	19	52260843	no match	421,860	Brillon DJ et al. Mechanism of defective insulin-receptor kinase activity in NIDDM. Evidence for two receptor populations. <i>Diabetes.</i> 1989 Mar;38(3):397-403. PMID: 2465197
NAPA	rs3810291	19	52260843	no match	421,860	Christoff PB et al. Procainamide disposition in obesity. <i>Drug Intell Clin Pharm.</i> 1983 Jul-Aug;17(7-8):516-22. PMID: 6191939
NAPA	rs3810291	19	52260843	no match	421,860	Zhou D et al. Genetics of metabolic variations between <i>Yersinia pestis</i> biovars and the proposal of a new biovar, <i>microtus</i> . <i>J Bacteriol.</i> 2004 Aug;186(15):5147-52. PMID: 15262951

* Distance between SNP and gene

Supplementary Note

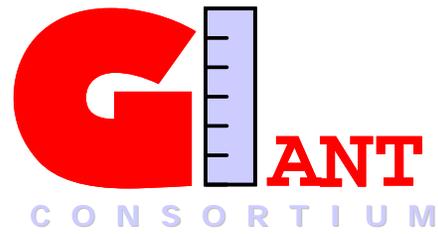
Supplementary Note - Table 13 Assessment of population-stratification by comparison of effect sizes and directions of association between family-based data from Framingham and overall meta-analysis using the QFAM -- within procedure from PLINK.

Supplementary Note - Table 13 -- Assessment of population-stratification by comparison of effect sizes and directions of association between family-based data from Framingham and overall meta-analysis using the QFAM --within procedure from PLINK.

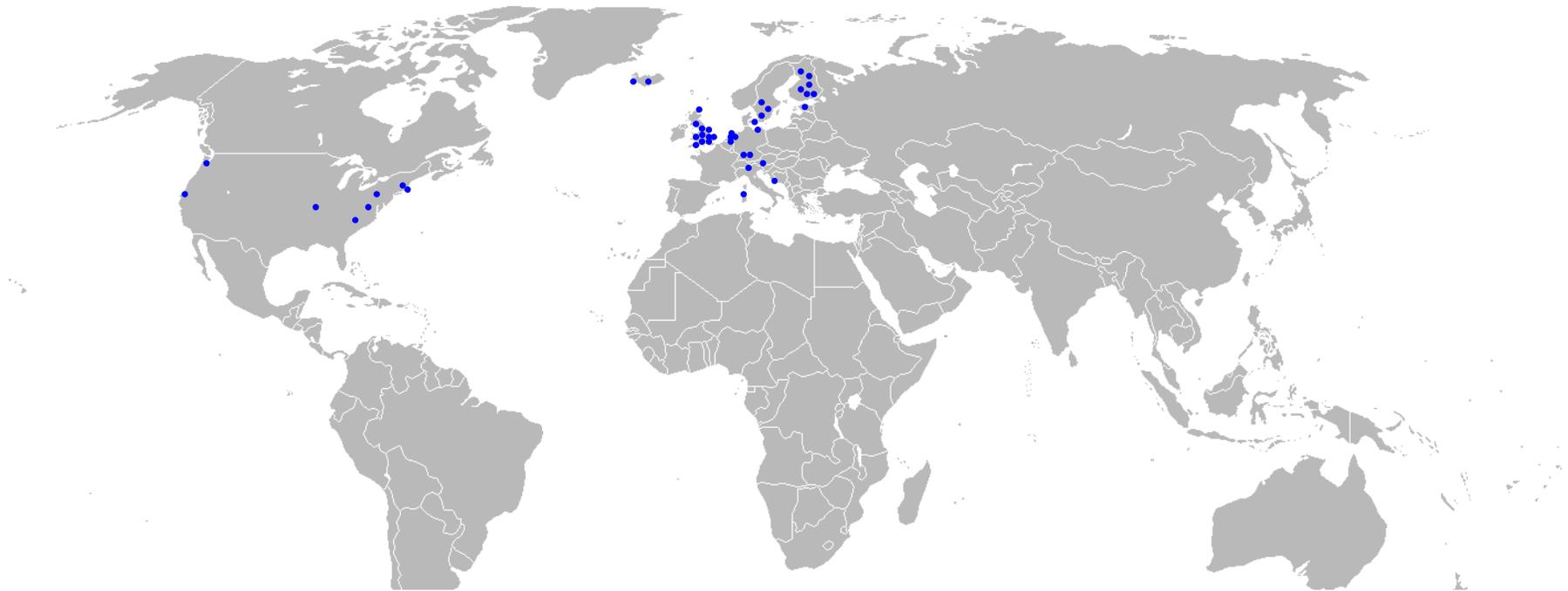
SNP	Nearest gene	Chr	Position** (bp)	Effect Allele	Framingham			Overall meta-analysis		Consistency of direction	Effect size
					N	beta	P-value	beta	P-value		
Previous BMI loci											
rs1558902	<i>FTO</i>	16	52,361,075	A	5,507	0.1028	6.1E-04	0.0819	2.3E-111	Same	Fram > Overall
rs2867125	<i>TMEM18</i>	2	612,827	T	5,507	-0.0016	9.7E-01	0.0658	3.7E-46	Opposite	Overall > Fram
rs571312	<i>MC4R</i>	18	55,990,749	A	5,504	0.0392	2.9E-01	0.0546	2.2E-40	Same	Overall > Fram
rs10938397	<i>GNPDA2</i>	4	44,877,284	A	5,423	0.0939	1.1E-03	0.0411	1.6E-29	Same	Fram > Overall
rs10767664	<i>BDNF</i>	11	27,682,562	A	5,507	-0.0128	7.3E-01	0.0438	2.7E-25	Opposite	Overall > Fram
rs2815752	<i>NEGR1</i>	1	72,585,028	A	5,503	0.0807	8.2E-03	0.0332	3.0E-20	Same	Fram > Overall
rs7359397	<i>SH2B1</i>	16	28,793,160	T	5,491	0.0383	2.1E-01	0.0319	9.5E-20	Same	Fram > Overall
rs9816226	<i>ETV5</i>	3	187,317,193	A	5,503	0.0691	6.9E-02	0.0405	1.7E-18	Same	Fram > Overall
rs3817334	<i>MTCH2</i>	11	47,607,569	T	5,507	0.0211	4.7E-01	0.0248	1.2E-11	Same	Overall > Fram
rs29941	<i>KCTD15</i>	19	39,001,372	A	5,506	-0.0136	6.8E-01	0.0209	3.3E-08	Opposite	Overall > Fram
Previous waist & weight loci											
rs543874	<i>SEC16B</i>	1	176,156,103	A	5,507	0.0861	2.5E-02	0.0455	7.7E-22	Same	Fram > Overall
rs987237	<i>TFAP2B</i>	6	50,911,009	A	5,487	0.0310	4.3E-01	0.0411	1.2E-19	Same	Overall > Fram
rs7138803	<i>FAIM2</i>	12	48,533,735	A	5,480	0.0416	1.9E-01	0.0304	1.6E-17	Same	Fram > Overall
rs10150332	<i>NRXN3</i>	14	79,006,717	T	5,507	0.0582	1.0E-01	0.0272	9.1E-10	Same	Fram > Overall
Newly identified BMI loci											
rs713586	<i>RBJ</i>	2	25,011,512	T	5,483	0.0547	7.1E-02	0.0298	5.0E-20	Same	Fram > Overall
rs12444979	<i>GPRC5B</i>	16	19,841,101	T	5,502	0.0112	7.9E-01	0.0435	3.3E-21	Same	Overall > Fram
rs2241423	<i>MAP2K5</i>	15	65,873,892	A	5,507	-0.0464	1.9E-01	0.0343	2.8E-18	Opposite	Fram > Overall
rs2287019	<i>QPCTL</i>	19	50,894,012	T	5,419	0.0746	5.1E-02	0.0350	7.6E-15	Same	Fram > Overall
rs1514175	<i>TNNI3K</i>	1	74,764,232	A	5,502	0.0280	3.5E-01	0.0235	2.5E-13	Same	Fram > Overall
rs13107325	<i>SLC39A8</i>	4	103,407,732	T	5,505	0.0964	5.5E-02	0.0497	7.1E-13	Same	Fram > Overall
rs2112347	<i>FLJ35779</i>	5	75,050,998	T	5,501	0.0173	6.0E-01	0.0238	5.0E-13	Same	Overall > Fram
rs10968576	<i>LRRN6C</i>	9	28,404,339	A	5,507	0.0570	8.2E-02	0.0257	2.7E-13	Same	Fram > Overall
rs3810291	<i>TMEM160</i>	19	52,260,843	A	5,460	-0.0217	5.1E-01	0.0239	1.2E-11	Opposite	Overall > Fram
rs887912	<i>FANCL</i>	2	59,156,381	T	5,505	0.0690	3.7E-02	0.0252	2.1E-13	Same	Fram > Overall
rs13078807	<i>CADM2</i>	3	85,966,840	A	5,501	0.0321	3.9E-01	0.0262	2.5E-11	Same	Fram > Overall
rs11847697	<i>PRKD1</i>	14	29,584,863	T	5,500	0.0193	7.9E-01	0.0536	2.2E-10	Same	Overall > Fram
rs2890652	<i>LRP1B</i>	2	142,676,401	T	5,498	0.0416	3.1E-01	0.0284	9.3E-11	Same	Fram > Overall
rs1555543	<i>PTBP2</i>	1	96,717,385	A	5,507	0.0664	3.1E-02	0.0188	3.1E-09	Same	Fram > Overall
rs4771122	<i>MTIF3</i>	13	26,918,180	A	5,504	0.0367	3.3E-01	0.0258	9.5E-10	Same	Fram > Overall
rs4836133	<i>ZNF608</i>	5	124,360,002	A	5,445	0.0149	6.1E-01	0.0193	1.6E-09	Same	Overall > Fram
rs4929949	<i>RPL27A</i>	11	8,561,169	T	5,507	0.0020	9.4E-01	0.0182	3.5E-09	Same	Overall > Fram
rs206936	<i>NUDT3</i>	6	34,410,847	A	5,507	-0.0261	4.6E-01	0.0208	3.3E-08	Opposite	Fram > Overall

2.3. SUPPLEMENTARY NOTE - FIGURES

Supplementary Note - Figure 1 The GIANT (Genetic Investigation of ANthropometric Traits) Consortium. Geographical location of the 46 genome-wide association cohorts that participated in the stage 1 meta-analyses.



Genome-wide association data of **123,865** individuals from 46 studies



ADVANCE, AGES-Reykjavik, Amish, ARIC, B58C, BRIGHT, CASP1&2, CHS, CoLaus, deCODE, DGI, EGCUT, EPIC-Norfolk, ERF, Fenland, FHS, FramHS, FTC, FUSION, GenMets, GerMiFS1&2, KORA3&4, MICROS, MIGEN, NFBC, NHS, NSPHS, NTR-NESDA, ORCADES, PLCO, Procardis, Rotterdam, RUNMC, SardinIA, SASBAC, Search/UKOPS, SHIP, TwinsUK, WTCCC-CHD, WTCCC-UKBS, WTCCC-T2D

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2nd Signals analyses

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Timothy M Frayling, Cecilia M Lindgren, John RB Perry, Michael N Weedon (chair), Cristen J Willer, Andrew R Wood (lead)

Copy Number Variant analyses

Supplementary Note

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Phenotyping of contributing cohorts

Stage 1 – Genome-wide association cohorts

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Stage 2 – de novo replication cohorts

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Genotyping of contributing cohorts

Stage 1 – Genome-wide association cohorts

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Stage 2 – de novo replication cohorts

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Supplementary Note

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Analyses of contributing cohorts

Stage 1 – Genome-wide association cohorts

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Supplementary Note

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Supplementary Note

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Supplementary Note

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Supplementary Note

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Supplementary Note

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Supplementary Note

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