

**Genome-wide meta-analysis identifies 11 new loci for anthropometric traits and provides
insights into genetic architecture**
SUPPLEMENTARY INFORMATION

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1. SUPPLEMENTARY TABLES

Supplementary Table 1. Total number of subjects for stage 1 and stage 2 for each anthropometric trait outcome

Trait	Stage 1 ^a	Stage 2 ^b	Stage 1 + Stage 2 ^c
	No. cases / No. controls	No. cases / No. controls	No. cases / No. controls
Overweight	93,015 / 65,840	65,332 / 39,294	158,306 / 105,101
Obesity Class 1	32,858 / 65,839	22,373 / 39,060	55,229 / 104,894
Obesity Class 2	9,889 / 62,657	5,476 / 35,430	15,334 / 97,858
Obesity Class 3	2,896 / 47,468	1,162 / 22,307	3,986 / 67,010
BMI tails	7,962 / 8,106	4,900 / 4,891	12,735 / 12,864
Height tails	8,097 / 8,099	4,872 / 4,831	12,926 / 12,888
WHR tails	4,774 / 5,481	3,351 / 3,352	7,969 / 8,683

^aThe study bases for the stage 1 cases and controls were: 158,864 (BMI), 168,267 (Height) and 100,605 (WHR)

^bThe study bases for the stage 2 cases and controls were: 109,703 (BMI), 107,740 (Height) and 75,220 (WHR)

^cThe study bases for the stage 1+2 cases and controls were: 268,567 (BMI), 276,007 (Height) and 175,825 (WHR)

Supplementary Table 2. Study design, number of individuals, and sample quality control for studies in stage 1, stage 2, or ancillary analyses

Study		Study design	Ethnicity	Total sample size (N)	Sample QC		Samples in analyses (N)	Anthropometric assessment method	References
Short name	Full name				Call rate*	Other exclusions			
Stage 1:									
ADVANCE controls	Atherosclerotic Disease, Vascular Function, and Genetic Epidemiology	Population-based case-control study	White European	311	≥ 98.5%	1) Gender Descrepancies, 2) related individuals and duplicates, 3) ethnic outliers	311	measured	[PMID: 18443000] Assimes, T. L., J. W. Knowles, et al. (2008). "Susceptibility locus for clinical and subclinical coronary artery disease at chromosome 9p21 in the multi-ethnic ADVANCE study." <i>Hum Mol Genet</i> 17(15): 2320-2328.
ADVANCE cases	Atherosclerotic Disease, Vascular Function, and Genetic Epidemiology	Population-based case-control study	White European	275	≥ 98.5%	1) Gender Descrepancies, 2) related individuals and duplicates, 3) ethnic outliers	275	measured	[PMID: 18443000] Assimes, T. L., J. W. Knowles, et al. (2008). "Susceptibility locus for clinical and subclinical coronary artery disease at chromosome 9p21 in the multi-ethnic ADVANCE study." <i>Hum Mol Genet</i> 17(15): 2320-2328.
AGES	Age, Gene/Environment Susceptibility-Reykjavik Study	Population-based	White European	3219	≥ 97%	1) mismatch with previous genotypes; 2) remove A/T & G/C SNPs; 3) remove SNPs not in HapMap	3219	measured	[PMID: 17351290] Harris et al. Age, Gene/Environment Susceptibility-Reykjavik Study: multidisciplinary applied phenomics. <i>American Journal of Epidemiology</i> (2007) vol. 165 (9) pp. 1076-87
ARIC	Atherosclerosis Risk in Communities Study	Population-based	White European	8861	≥ 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 height and/or weight or other covariate	8108	measured	[PMID: 2646917] Atherosclerosis Risk in Communities (ARIC) Study: design and objectives. 1989. ARIC Investigators. <i>Am. J. Epidemiol.</i> 129: 687-702.
B58C-T1DGC	British 1958 birth cohort (Type 1 Diabetes Genetic Consortium controls)	Population-based	White European	2592	≥ 98%	1) contamination; 2) non-European identity; 3) Missing or invalid height, weight, waist or hip.	2591	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. <i>Int J Epidemiol</i> 2007;36:522-531. [PMID: 19430480] Barrett JC, Clayton DG, Concannon P, Akolkar B, Cooper JD, Erlich HA, Julier C, Morahan G, Nerup J, Nierras C, Plagnol V, Pociot F, Schuilenburg H, Smyth DJ, Stevens H, Todd JA, Walker NM, Rich SS; The Type 1 Diabetes Genetics Consortium. Genome-wide

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Short name	Full name				Call rate*	Other exclusions			
									association study and meta-analysis find that over 40 loci affect risk of type 1 diabetes. Nat Genet 2009 May 10.
B58C-WTCCC	British 1958 birth cohort (Wellcome Trust Case Control Consortium controls)	Population-based	White European	1502	≥ 97%	1) contamination; 2) non-European identity; 3) Missing or invalid height, weight, waist or hip.	1479	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: 16155052] Power, C. & Elliott, J. Cohort profile: 1958 British birth cohort (National Child Development Study). Int. J. Epidemiol. 35, 34-41 (2006).
BRIGHT	British Genetics of Hypertension (BRIGHT) Study	Hypertensive cases	White European	2000	≥ 97%	1) heterozygosity <23% or >30% 2) external discordance 3) non-European ancestry 4) duplicate/first/second degree relatives.	1895	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).
CAD WTCCC	WTCCC Coronary Heart Disease cases	Case series	White European	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	White European	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	White European	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).

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Short name	Full name				Call rate*	Other exclusions			
CAPS2 Cases	Cancer Prostate in Sweden 2	Case-control study of prostate cancer	White European	1483	> 95%	1) related individuals and duplicates; 2) ethnic outliers; 3) missing body weight and height.	1483	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	White European	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	Caucasian	3329	≥ 95%	1) sex mismatch or discordance with prior genotyping; 2) none whites; 3) 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, et al. The Cardiovascular Health Study: design and rationale. Ann Epidemiol. 1991;1(3):263-276
CoLaus	Cohorte Lausannoise	Population-based	White European	6188	> 90%	1) ethnic outliers; 2) related individuals and duplicates; 3) Missing height	5435	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
COROGENE controls	COROGENE-STUDY Genetic Predisposition of Coronary Heart Disease in Patients Verified with Coronary Angiogram (controls for this study are a part of the National FINRISK Studies)	Case-control	White European: Finnish-only	2066	≥ 95%	1) SNP clustering probability for each genotype > 95% 2) Call rate > 95% both individuals and markers 3) MAF > 1% 4) HWE p > 1*10 ⁻⁶ 5)heterozygosity, gender check and relatedness checks have been performed and any discrebansies have been removed. 6) Missing phenotype.	1890	measured	[PMID: 21642350] Vaara, S. et al. Cohort profile:The Corogene Study. Int J Epidemiol. 2011 Jun 3. Epub ahead of print.
COROGENE cases	COROGENE-STUDY Genetic Predisposition of Coronary Heart Disease in Patients Verified with Coronary Angiogram	Case-control	White European: Finnish-only	2597	≥ 95%	1) SNP clustering probability for each genotype > 95% 2) Call rate > 95% both individuals and markers 3) MAF > 1% 4) HWE p >	2240	measured	[PMID: 21642350] Vaara, S. et al. Cohort profile:The Corogene Study. Int J Epidemiol. 2011 Jun 3. Epub ahead of print.

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Study		Study design	Ethnicity	Total sample size (N)	Sample QC		Samples in analyses (N)	Anthropometric assessment method	References
Short name	Full name				Call rate*	Other exclusions			
						1*10 ⁻⁶ 5)heterozygosity, gender check and relatedness checks have been performed and any discrebansies have been removed. 6) Missing phenotype.			
deCODE	deCODE genetics sample set	Population-based	White European	38446	≥ 96%	1) Missing body weight and height.	26799	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 controls	Diabetes Genetics Initiative	Case-control	White European	1464	≥ 95%	1) Related individuals and duplicates 2) Sex mismatch 3) Phenotype missing	1317	measured	[PMID: 17463246] Saxena, R. et al. Genome-wide association analysis identifies loci for type 2 diabetes and triglyceride levels. Science. Science 2007;316(5829):1331-6.
DGI Cases	Diabetes Genetics Initiative	Case-control	White European	1467	≥ 95%	1) Related individuals and duplicates 2) Sex mismatch 3) Phenotype missing	1090	measured	[PMID: 17463246] Saxena, R. et al. Genome-wide association analysis identifies loci for type 2 diabetes and triglyceride levels. Science. Science 2007;316(5829):1331-6.
EGCUT	Estonian Genme Center of University of Tartu	Population-based	Estonians	2734	≥ 95%	1) ethnic outliers; 2) related individuals and duplicates; 3) Missing body weight and height.	2685	measured	[PMID: 19424496] Nelis M. et al. Genetic structure of Europeans: a view from the North-East. PLoS One. 2009;4(5):e5472.
EPIC-Obesity Study	European Prospective Investigation into Cancer and Nutrition - Obesity Study	Population-based	White European	2566	≥ 94%	1) heterozygosity <23% or >30%; 2) >5.0% discordance in SNP pairs with r2= 1 in HapMap; 3) ethnic outliers; 4) related individuals and duplicates; 5) Missing body weight and height.	2415	measured	[PMID: 10466767] Day,N.E. et al. EPIC-Norfolk: study design and characteristics of the cohort. European Prospective Investigation of Cancer. British Journal of Cancer 80, 95-103 (1999). [PMID: 18454148] Loos,R.J. et al. Common variants near MC4R are ass
ERF	Erasmus Rucphen Family	Family-based	White European	2386	≥ 95%	1) heterozygosity; 2)high IBS > 95%; 3) ethnic outliers; 4) Gender mismatch; 5) Missing phenotypes.	2095	measured	[PMID: 15054401] Aulchenko YS, Heutink P, Mackay I, Bertoli-Avella AM, Pullen J, Vaessen N et al. Linkage disequilibrium in young genetically isolated Dutch population. Eur J Hum Genet 2004; 12(7): 527-534.

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Study		Study design	Ethnicity	Total sample size (N)	Sample QC		Samples in analyses (N)	Anthropometric assessment method	References
Short name	Full name				Call rate*	Other exclusions			
FamHS (Stage 1)	Family Heart Study	Case (Coronary Artery Calcification)-control	European American	3659	N.A.	1) Non- European American descent; 2) Missing body weight and height	856	measured	[PMID: 8651220] Higgins M, Province M, Heiss G, Eckfeldt J, Ellison RC, Folsom AR, Rao DC, Sprafka JM, Williams R. NHLBI Family Heart Study: objectives and design. <i>Am J Epidemiol.</i> 1996;143:1219-28.
FENLAND	Fenland Study	Population-based	White European	1500	≥ 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. Six new loci associated with body mass index highlight a neuronal influence on body weight regulation. <i>Nat Genet</i> 2009, 41:25-34.
FRAM	Framingham Heart Study	Population-based, multi-generational	White European	9274	≥ 97%	1) pHWE<1e-6call 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.
FUSION controls	Finland□United States Investigation of NIDDM Genetics	Case□control study	White European	1173	> 97.5%	1) related individuals; 2) missing BMI	1171	measured	[PMID: 17463248] Scott, L.J., Mohlke, K.L., Bonnycastle, L.L., Willer, C.J., Li, Y., Duren, W.L., Erdos, M.R., Stringham, H.M., Chines, P.S., Jackson, A.U., Prokunina-Olsson L., Ding, C.J., Swift, A.J., Narisu, N., Hu, T., Pruim, R., Xiao, R., Li, X.Y., Conneely, K.N., Riebow, N.L., Sprau, A.G., Tong, M., White, P.P., Hetrick, K.N., Barhnart, M.W., Bark, C.W., Goldstein, J.L., Watkins, L., Xiang, F., Saramies, J., Buchanan, T.A., Watanabe, R.M., Valle, T.T., Kinnunen, L., Abecasis, G.R., Pugh, E.W., Doheny, K.F., Bergman, R.N., Tuomilehto, J., Collins, F.S., Boehnke, M. A Genome-Wide Association Study of Type 2 Diabetes in Finns Detects Multiple Susceptibility Variants. <i>Science</i> 2007 Jun 1;316(5829):1341-5. Epub 2007 Apr 26.

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Study		Study design	Ethnicity	Total sample size (N)	Sample QC		Samples in analyses (N)	Anthropometric assessment method	References
Short name	Full name				Call rate*	Other exclusions			
FUSION Cases	Finland-United States Investigation of NIDDM Genetics	Case-control study	White European	1160	> 97.5%	1) related individuals; 2) missing BMI	1092	measured	[PMID: 17463248] Scott, L.J., Mohlke, K.L., Bonnycastle, L.L., Willer, C.J., Li, Y., Duren, W.L., Erdos, M.R., Stringham, H.M., Chines, P.S., Jackson, A.U., Prokunina-Olsson L., Ding, C.J., Swift, A.J., Narisu, N., Hu, T., Pruim, R., Xiao, R., Li, X.Y., Conneely, K.N., Riebow, N.L., Sprau, A.G., Tong, M., White, P.P., Hetrick, K.N., Barhnart, M.W., Bark, C.W., Goldstein, J.L., Watkins, L., Xiang, F., Saramies, J., Buchanan, T.A., Watanabe, R.M., Valle, T.T., Kinnunen, L., Abecasis, G.R., Pugh, E.W., Doheny, K.F., Bergman, R.N., Tuomilehto, J., Collins, F.S., Boehnke, M. A Genome-Wide Association Study of Type 2 Diabetes in Finns Detects Multiple Susceptibility Variants. Science 2007 Jun 1;316(5829):1341-5. Epub 2007 Apr 26. Health 2000. March 09th, 2009 [cited 2010 26.11.2010]; Available from: http://www.terveys2000.fi/indexe.html
GENMETS	Health 2000 study subset of Metabolic syndrome cases and their matched controls	Case - control	White European	2124	> 95%	1) heterozygosity 2) gender check 3) ethnic outliers 4) cryptic relatedness 5) related individuals and duplicates	1705	measured	HEALTH AND FUNCTIONAL CAPACITY IN FINLAND, Baseline Results of the Health 2000 Health Examination Survey. In: Aromaa A, Koskinen S, editors. Helsinki: National Public Health Institute; 2004.
GerMIFS1	German Myocard Infarct Family Study	Case - control study (only cases)	European (german people)	875	≥ 97%	1) heterozygosity + 3 * sd 2) related individuals and duplicates 3) Missing phenotypes	875	measured	[PMID: 17634449] Samani NJ et al. Genomewide association analysis of coronary artery disease.
GerMIFS2	German Myocard Infarct Family Study	Case - control study (only cases)	European (german people)	1222	≥ 97%	1) heterozygosity + 3 * sd 2) related individuals and duplicates 3) Missing phenotypes	1222	measured	[PMID: 19198612] Erdmann J. Genomewide association analysis of coronary artery disease.
GOOD	Gothenburg Osteoporosis and Obesity Determinants Study	Population-based	White European	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. J Bone Miner Res 20, 1334-1341 (2005).
HBCS	Helsinki Birth Cohort Study	Birth-cohort	White European: Finnish	1872	≥ 95%	1) SNP clustering probability for each genotype > 95% 2) Call rate > 95% both individuals and markers 3) MAF > 1% 4) HWE p > 1*10 ⁻⁶	1739	measured	[PMID: 16881894] Eriksson, J.G. Early growth and adult health outcomes -- lessons learned from the Helsinki Birth Cohort Study. Matern Child Nutr. 2005 Jul;149-54. Review.

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Short name	Full name				Call rate*	Other exclusions			
KORA S3	Cooperative Health Research in the Region of Augsburg	Population-based	White European	1644	≥ 93%	5) heterozygosity, gender check and relatedness checks have been performed and any discrepancies have been removed. 6) Missing phenotype. 1) German passport; 2) missing phenotypes.	1638-1643	measured	[PMID: 16032514] Wichmann HE et al. KORA-gen--resource for population genetics, controls and a broad spectrum of disease phenotypes. Gesundheitswesen 67 Suppl 1, S26-30 (2005).
KORA S4	Cooperative Health Research in the Region of Augsburg	Population-based	White European	1814	≥ 93%	1) German passport; 2) missing phenotypes.	1811-1812	measured	[PMID: 16032514] Wichmann HE et al. KORA-gen--resource for population genetics, controls and a broad spectrum of disease phenotypes. Gesundheitswesen 67 Suppl 1, S26-30 (2005).
MGS	Molecular Genetics of Schizophrenia/NIMH Repository Control Sample	Population-based (survey research method)	European ancestry (USA)	2681	0.997	1) call rate < 97% for samples, 95% for SNP; 2) heterozygosity < 26% or > 28.5%; 3) excess duplicate discordancies or mendelian errors (SNPs); 4) ethnic outliers (principal component scores); 5) related individuals and duplicates; 6) Missing body weight or height.	2597	self-report	[PMID: 19571809] Shi, J, et al. Common variants on chromosome 6p22.1 are associated with schizophrenia. Nature. 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. Am J Psychiatry. 2008 Apr;165(4):497-506.
MICROS	Microisolates in South Tyrol	Population-based	White European	1269	0.98	1) Gender mismatch 2) Genetic outliers	1097	measured	[PMID: 17550581] Pattaro C, Marroni F, Riegler A, Mascalzoni D, Pichler I, et al. (2007) The genetic study of three population microisolates in South Tyrol (MICROS): study design and epidemiological perspectives. BMC Med Genet 8: 29.
Migen (cases)	Myocardial Infarction Genetics Consortium	Case control forearly onset MI	European	1420	≥ 95%	1) Cohort deficient or excess heterozygosity 2) Population Stratification outliers 3) Close Relatives and Sample Duplicates 4) Sample Contamination	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).

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Short name	Full name				Call rate*	Other exclusions			
						(excess close low level genomes sharing)5) missing body weight or height			
Migen (controls)	Myocardial Infarction Genetics Consortium	Case control for early onset MI	European	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	White European	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)
NHS	The Nurses' Health Study	Nested Case-control	White European	2,287	>98%	1) Sex discrepancy with genetic data from X-linked markers; 2) Duplicates and first/second degree relatives; 3) Ancestry outliers; 4) Heterozygosity; 5) Autosomal chromosome aberrations; 6) Missing phenotype information	2,120	self-report	[PMID: 15864280] Colditz GA, Hankinson SE (2005) The Nurses' Health Study: lifestyle and health among women. [PMID: 20418489] Qi L, Cornelis MC, Kraft P, Stanya KJ, Linda Kao WH, et al. (2010) Genetic variants at 2q24 are associated with susceptibility to type 2 diabetes. Hum Mol Genet 19: 2706-2715.
NFBC	Northern Finland Birth Cohort 1966	Population-based	White European	5654	≥ 95%	1) gender discrepancy with genetic data from X-linked markers; 2)	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).

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Short name	Full name				Call rate*	Other exclusions			
NSPHS	Northern Sweden population health study	Population	White European	700	0.98	withdrawn consent; 3) duplicates and first and second degree relatives; 4) contaminated samples 1) Gender mismatch 2) Genetic outliers	656	measured	[PMID: 20568910] Igl W, Johansson A, Gyllensten U. The Northern Swedish Population Health Study (NSPHS)--a paradigmatic study in a rural population combining community health and basic research. <i>Rural Remote Health</i> . 2010 Apr-Jun;10(2):1363. Epub 2010 Jun 18. [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> . 2009 Jan 15;18(2):373-80. Epub 2008 Oct 24.
NTR and NESDA MDD controls	The Netherlands Twin Register and Nederlandse Studie naar Depressie en Angst	Case-control	White European	1802	>90%	1) Contamination; 2) cryptic relatedness and ethnic outliers ;3) gender mismatch; 4) Missing body weight and height	1800	questionnaire and measured	[PMID: 19065144] Sullivan PF et al. Genome-wide association for major depressive disorder: a possible role for the presynaptic protein piccolo. <i>Mol Psychiatry</i> . 14:650-2 (2009)
NTR and NESDA MDD cases	The Netherlands Twin Register and Nederlandse Studie naar Depressie en Angst	Case-control	White European	1738	>90%	1) Contamination; 2) cryptic relatedness and ethnic outliers ; 3) gender mismatch; 4) Missing body weight and height	1731	questionnaire and measured	[PMID: 2047772] Willemsen et al. The Netherlands Twin Register biobank: a resource for genetic epidemiological studies. <i>Twin Res Hum Genet</i> . 13:231-45 (2010)
ORCADES	Orkney Complex Disease Study	Population-based	White European	761	0.98	1) Gender mismatch 2) Genetic outliers	716	measured	[PMID: 18760389] McQuillan.R., Leutenegger.A.L., Abdel-Rahman.R., Franklin.C.S., Pericic.M., Barac-Lauc.L., Smolej-Narancic.N., Janicijevic.B., Polasek.O., Tenesa.A., et al. (2008) Runs of homozygosity in European populations. <i>American Journal of Human Genetics</i> , 83, 359-372.
PLCO	Prostate, Lung, Colorectal, and Ovarian Cancer Screening Trial	Population-based	Non-Hispanic Caucasian	2298	≥ 94%	1) Gender discordance 2) Non-European ancestry 3) Related individuals and duplicates; 4) Missing phenotype data	2238	self-report	[PMID: 17401363] Yeager M, et al. Genome-wide association study of prostate cancer identifies a second risk locus at 8q24. <i>Nat Genet</i> 39, 645-9 (2007).
PROCARDIS	Precocious Coronary Artery Disease (ProCARDIS)	Case-control	White European	2573	> 95%	none	2573	measured	[PMID: 18048406] 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. <i>Hum Mol Genet</i> 17, 806-14 (2008).

Supplementary Table 2. Study design, number of individuals, and sample quality control for studies in stage 1, stage 2, or ancillary analyses

Study		Study design	Ethnicity	Total sample size (N)	Sample QC		Samples in analyses (N)	Anthropometric assessment method	References
Short name	Full name				Call rate*	Other exclusions			
RUNMC	Radboud University Nijmegen Medical Centre, composed of Nijmegen Bladder Cancer Study (NBCS) and Nijmegen Biomedical Study (NBS)	Population-based	White European	3081	≥ 96%	1) Missing body weight and height.	2873	measured	[PMID: 17568781] Wetzels, J.F. et al. Age- and gender-specific reference values of estimated GFR in Caucasians: the Nijmegen Biomedical Study. <i>Kidney Int</i> 72, 632-637 (2007). [PMID: 18794855] Kiemeny, L.A. et al. Sequence variant on 8q24 confers susceptibility to urinary bladder cancer. <i>Nat Genet</i> 40, 1307-1312 (2008).
RS1	Rotterdam Study I	Population Based	White European	7983	≥ 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 from PLINK; 6) Missing body weight and height.	5743	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:21877163] {Hofman, 2011 The Rotterdam Study: 2012 objectives and design update.}; [PMID:1833235] {Hofman, 1991 Determinants of disease and disability in the elderly: the Rotterdam Elderly Study};
SASBAC controls	Swedish And Singapore Breast Association Consortium	Case-control study of breast cancer	White European	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. <i>Int J Cancer</i> 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. <i>Breast Cancer Res</i> 8, R67 (2006).
SASBAC cases	Swedish And Singapore Breast Association Consortium	Case-control study of breast cancer	White European	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. <i>Int J Cancer</i> 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. <i>Breast Cancer Res</i> 8, R67 (2006).

Supplementary Table 2. Study design, number of individuals, and sample quality control for studies in stage 1, stage 2, or ancillary analyses

Study		Study design	Ethnicity	Total sample size (N)	Sample QC		Samples in analyses (N)	Anthropometric assessment method	References
Short name	Full name				Call rate*	Other exclusions			
SARDINIA	SARDINIA study on aging	Family-based	White European	4305	>90%	1) Missing body weight and height 2) related individuals	889	measured	[PMID: 16934002] Pilia, G. et al. Heritability of cardiovascular and personality traits in 6,148 Sardinians. <i>PLoS Genet</i> 2, e132 (2006). [PMID 18193045] Sanna S et al. Common variants in the GDF5-UQCC region are associated with variation in human height. <i>Nat Genet.</i> 2008 Feb;40(2):198-203. Epub 2008 Jan 13
SEARCH/UKOPS	Studies of Epidemiology and Risk factors in Cancer Heredity / UK Ovarian Cancer Population Study	Population-based case series	White European	1710	≥95%	1) ethnic outliers 2) duplicates 3) Missing body weight and height	1556	self-assessed	[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. <i>Nature Genetics</i> , 41, p996-1000, (2009).
SHIP	Study of Health in Pomerania	Population-based	White European	4308	>92%	1) missing genotype or phenotype data 2) reported vs. genotyped gender mismatch 3) duplicates	4070	measured	[PMID: 20167617] Völzke H. et al. Cohort profile: the study of health in Pomerania. <i>Int J Epidemiol.</i> 2011 Apr;40(2):294-307.
Sorbs	population-based study of the Sorbs, self-contained population of Slavonic origin from Eastern Germany	Population-based	White European	1097	≥ 94%	1) ethnic outliers; 2) duplicates 3) gender mismatch	941	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. PMID: 21559053
TWINSUK	TwinsUK	Twins pop	White European	2226	≥ 95%	1) heterozygosity <33% or >37%; 2) ethnic outliers; 3) related individuals and duplicates; 4) Missing body weight and height.	1479	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).
T2D WTCCC	WTCCC Type 2 Diabetes cases	Case series	White European	1999	≥ 97%	1) heterozygosity <23% or >30%; 2) discrepancy with external identifying information; 3) ethnic outliers; 4) related individuals and duplicates;	1903	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. <i>Nature</i> 447, 661-678 (2007)
VIS	VIS and KORCULA (EUROSPAN)	Population-based	White European	983	0.98	1) Gender mismatch 2) Genetic outliers	795	measured	[PMID: 18327257] Vitart, V., Rudan, I., Hayward, C., Gray, N.K., Floyd, J., Palmer, C.N., Knott, S.A., Kolcic, I., Polasek, O., Graessler, J., et al. (2008) SLC2A9 is a newly identified urate transporter influencing serum urate concentration, urate excretion and gout. <i>Nature Genetics</i> , 40, 437-442.

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Study		Study design	Ethnicity	Total sample size (N)	Sample QC		Samples in analyses (N)	Anthropometric assessment method	References
Short name	Full name				Call rate*	Other exclusions			
WGHS	Women's Genome Health Study	Cohort	European	23294	≥ 98%	1) self-reported European ancestry confirmed by identity-by-state analysis using ancestry informative SNPs in PLINK.	23294	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
YFS	The Cardiovascular Risk in Young Finns Study	Population-based	White European: Finnish	2556	≥ 95%	1) SNP clustering probability for each genotype > 95% 2) Call rate > 95% both individuals and markers 3) MAF > 1% 4) HWE p > 1*10 ⁻⁶ 5) heterozygosity, gender check and relatedness checks have been performed and any discrepancies have been removed. 6) Missing phenotype.	2443	measured	[PMID: 18263651] Raitakari OT, Juonala M, Rönnemaa T, Keltikangas-Järvinen L, Räsänen L, Pietikäinen M, Hutri-Kähönen N, Taittonen L, Jokinen E, Marniemi J, Jula A, Telama R, Kähönen M, Lehtimäki T, Akerblom HK, Viikari JS. Cohort profile: the cardiovascular risk in Young Finns Study. Int J Epidemiol. 2008 Dec;37(6):1220-6.
Stage 2 - In silico:									
B58C-REPL	British 1958 birth cohort (replication subset)	Population-based	White European	2422	≥ 98%	1) contamination; 2) non-European identity; 3) Missing or invalid height, weight, waist or hip.	2422	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 19430480] Barrett JC, Clayton DG, Concannon P, Akolkar B, Cooper JD, Erlich HA, Julier C, Morahan G, Nerup J, Nierras C, Plagnol V, Pociot F, Schuilenburg H, Smyth DJ, Stevens H, Todd JA, Walker NM, Rich SS; The Type 1 Diabetes Genetics Consortium. Genome-wide association study and meta-analysis find that over 40 loci affect risk of type 1 diabetes. Nat Genet 2009 May 10. [PMID: 15486340] James AL et al. Am J Respir Crit Care Med Vol 171 pp 109-114 (2005)
BHS	Busselton Health Study	Population-based	White European	1281	≥ 97%	1) ethnic outliers; 2) related individuals and duplicates.	1206	measured	

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Study		Study design	Ethnicity	Total sample size (N)	Sample QC		Samples in analyses (N)	Anthropometric assessment method	References
Short name	Full name				Call rate*	Other exclusions			
HYPERGENES controls	Hypergenes	Case-control	White European	1934	>90%	1) ethnic outliers 2) Missing body weight and height.	1934	measured	[PMID: 22184326] Salvi E, Kotalik Z, Glorioso N, Benaglio P, Frau F, Kuznetsova T, Arima H, Hoggart C, Tichet J, Nikitin YP, Conti C, Seidlerova J, Tikhonoff V, Stolarz-Skrzypek K, Johnson T, Devos N, Zagato L, Guarrera S, Zaninello R, Calabria A, Stancanelli B, Troffa C, Thijs L, Rizzi F, Simonova G, Lupoli S, Argiolas G, Braga D, D'Alessio MC, Ortu MF, Ricceri F, Mercurio M, Descombes P, Marconi M, Chalmers J, Harrap S, Filipovsky J, Bochud M, Iacoviello L, Ellis J, Stanton AV, Laan M, Padmanabhan S, Dominiczak AF, Samani NJ, Melander O, Jeunemaitre X, Manunta P, Shabo A, Vineis P, Cappuccio FP, Caulfield MJ, Matullo G, Rivolta C, Munroe PB, Barlassina C, Staessen JA, Beckmann JS, Cusi D. Genome-wide association study using a high-density single nucleotide polymorphism array and case-control design identifies a novel essential hypertension susceptibility locus in the promoter region of endothelial NO synthase. <i>Hypertension</i> . 2012 Feb;59(2):248-55.
HYPERGENES Cases	Hypergenes	Case-control	White European	2124	>90%	1) ethnic outliers 2) Missing body weight and height.	2124	measured	[PMID: 22184326] Salvi E, Kotalik Z, Glorioso N, Benaglio P, Frau F, Kuznetsova T, Arima H, Hoggart C, Tichet J, Nikitin YP, Conti C, Seidlerova J, Tikhonoff V, Stolarz-Skrzypek K, Johnson T, Devos N, Zagato L, Guarrera S, Zaninello R, Calabria A, Stancanelli B, Troffa C, Thijs L, Rizzi F, Simonova G, Lupoli S, Argiolas G, Braga D, D'Alessio MC, Ortu MF, Ricceri F, Mercurio M, Descombes P, Marconi M, Chalmers J, Harrap S, Filipovsky J, Bochud M, Iacoviello L, Ellis J, Stanton AV, Laan M, Padmanabhan S, Dominiczak AF, Samani NJ, Melander O, Jeunemaitre X, Manunta P, Shabo A, Vineis P, Cappuccio FP, Caulfield MJ, Matullo G, Rivolta C, Munroe PB, Barlassina C, Staessen JA, Beckmann JS, Cusi D. Genome-wide association study using a high-density single nucleotide polymorphism array and case-control design identifies a novel essential hypertension susceptibility locus in the promoter region of endothelial NO synthase. <i>Hypertension</i> . 2012 Feb;59(2):248-55.
LifeLines	LifeLines	Population-based	White European	9480	≥ 95%	1) non-Caucasian (Principle Components Analysis); 2) related individuals (pi-hat)	8116	measured	[PMID:18075776] Stolk, R.P. Universal risk factors for multifactorial diseases: LifeLines: a three-generation population-based study. <i>Eur J Epidemiol</i> . 23(1):67-74 (2008)

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Study		Study design	Ethnicity	Total sample size (N)	Sample QC		Samples in analyses (N)	Anthropometric assessment method	References
Short name	Full name				Call rate*	Other exclusions			
PLCO2 controls	Prostate, Lung, Colorectal, and Ovarian Cancer Screening Trial	Population-based case-control	Non-Hispanic Caucasian	1216	>98%	> 0.4); 3) unexpected duplicates; 4) gender mismatch 5) missing phenotype 1) sex discordance with genetic data; 2) heterozygosity; 3) ancestry outliers; 4) related individuals and duplicates; 5) missing phenotype data	1193	self-report	[PMID: 21490707] Cornelis MC, et al. Genome-wide meta-analysis identifies regions on 7p21 (AHR) and 15q24 (CYP1A2) as determinants of habitual caffeine consumption. <i>PLoS Genet.</i> 2011 Apr;7(4):e1002033.
PLCO2 cases	Prostate, Lung, Colorectal, and Ovarian Cancer Screening Trial	Population-based case-control	Non-Hispanic Caucasian	3003	>98%	1) sex discordance with genetic data; 2) heterozygosity; 3) ancestry outliers; 4) related individuals and duplicates; 5) missing phenotype data	2976	self-report	[PMID: 21490707] Cornelis MC, et al. Genome-wide meta-analysis identifies regions on 7p21 (AHR) and 15q24 (CYP1A2) as determinants of habitual caffeine consumption. <i>PLoS Genet.</i> 2011 Apr;7(4):e1002033.
PREVEND	Prevention of Renal and Vascular Endstage Disease	Population-based	White European	3920	≥95%	1) ethnic outliers; 2) related individuals and duplicates; 3) missing phenotype;	3622	measured	[PMID: 11004219] Pinto-Sietsema SJ, et al. Urinary albumin excretion is associated with renal function abnormalities in a nondiabetic population. <i>J Am Soc Nephrol</i> 2000;11:1882-8.
QIMR	Twin study at Queensland Institute of Medical Research	Population-based	White European	11930	≥ 95%	1) age < 18; 2) ethnic outliers; 3) related individuals and duplicates; 4) Missing body weight and height.	3953	measured or self-reported	[PMID: 19896111] Medland, 2009 Common Variants in the Trichohyalin Gene Are Associated with Straight Hair in Europeans.
RS2	Rotterdam Study - II	Population-based	White European	3011	≥ 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 >	1911	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:21877163] {Hofman, 2011 The Rotterdam Study: 2012 objectives and design update}; [PMID:1833235] {Hofman, 1991 Determinants of disease and disability in the elderly: the Rotterdam Elderly Study};

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Study		Study design	Ethnicity	Total sample size (N)	Sample QC		Samples in analyses (N)	Anthropometric assessment method	References
Short name	Full name				Call rate*	Other exclusions			
						4SD mean HaMAP CEU cluster from PLINK; 5) Missing body weight and height.			
RS3	Rotterdam Study - III	Population-based	White European	3932	≥ 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) Missing body weight and height.	2006	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};
TRAILS	TRacking Adolescents' Individual Lives Survey	Population-based	Caucasian (Dutch)	1455	>95%	1) heterozygosity >3SD from mean; 2) duplicate and MZ samples; 3) sex mismatches; 4) non-caucasians; 5) missing phenotype data	1141	measured	[PMID: 18263649] Huisman, M. et al. Cohort profile: the Dutch 'TRacking Adolescents' Individual Lives' Survey'; TRAILS. Int J Epidemiol. 2008 Dec;37(6):1227-35.
TWINGENE	TWINGENE	Population-based	White European	9836	≥ 97%	1) Heterozygosity, (F-mean(F))/sd(Z) ≥ 5 2) Sex-check based on the heterozygosity rates on the X-chromosome 3) Missing all phenotypes	9740	measured	[PMID: 8981957] Hong Y, Pedersen NL, Brismar K, de Faire U. Genetic and environmental architecture of the features of the insulin-resistance syndrome. The American Journal of Human Genetics 1997 Jan;60(1):143-52.

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Study		Study design	Ethnicity	Total sample size (N)	Sample QC		Samples in analyses (N)	Anthropometric assessment method	References
Short name	Full name				Call rate*	Other exclusions			
UKBS2	Wellcome Trust Common Controls Consortium - Panel 2	Population-based	White European	1338	≥ 95%	1) missing body weight & height	1332	questionnaire	[PMID: 17554300] Committee., W.T.C.C.C., Burton, P.R., Clayton, D.G., Cardon, L.R., Craddock, N., Deloukas, P., Duncanson, A., Kwiatkowski, D.P., McCarthy, M.I., Ouwehand, W.H., Samani, N.J., Todd, J. & Donnelly, P.C. Genome-wide association study of 14,000 cases of seven common diseases and 3,000 shared controls. <i>Nature</i> , 447 , 661-678 (2007).
Stage 2 - Metabochip:									
AMC-PAS	Academic Medical Centre Amsterdam Premature Atherosclerosis Study	CAD cases	White European	490	0.97	1) heterozygosity; 2) ethnic outliers; 3) duplicate; 4) missing phenotypes; 5) call rate < 0.95	490	measured	[PMID: 19164808] Coronary Artery Disease Consortium, Samani NJ. et al. Large scale association analysis of novel genetic loci for coronary artery disease. <i>Arterioscler Thromb Vasc Biol.</i> 2009 May;29(5):774-80.
CARDIOGENICS controls	CARDIOGENICS	CAD controls	European	375	0.98	1) heterozygosity; 2) ethnic outliers; 3) duplicate; 4) missing phenotypes; 5) call rate < 0.95	375	measured	[PMID: 17634449] Samani NJ. et al. Genomewide association analysis of coronary artery disease. <i>N Engl J Med.</i> 2007 Aug 2;357(5):443-53.
CARDIOGENICS cases	CARDIOGENICS	CAD cases	European	379	0.98	1) heterozygosity; 2) ethnic outliers; 3) duplicate; 4) missing phenotypes; 5) call rate < 0.95	379	measured	[PMID: 17634449] Samani NJ. et al. Genomewide association analysis of coronary artery disease. <i>N Engl J Med.</i> 2007 Aug 2;357(5):443-53.
D2D2007.DPS.DRSEXTRA.FUSION2.METSIM controls	FIN-D2D 2007. Diabetes Prevention Study, The Dose Responses to Exercise Training Study, FUSION stage2, and METabolic Syndrome In Men	FIN-D2D 2007 and DR's EXTRA are population-based. DPS is a clinical prevention trial. For FUSION stage 2 and METSIM, samples genotyped followed a case/control design. Cases were analyzed separately from controls.	White European	6643	0.95	1) Gender discrepant; 2) unexpected duplicate; 3) missing all phenotype; 4) first-degree related	6108	measured	(FIN-D2D 2007) [PMID: 20459722] Kotronen A, Yki-Järvinen H, Männistö S, Saarikoski L, Korpi-Hyövälti E, Oksa H, Saltevo J, Saaristo T, Sundvall J, Tuomilehto J, Peltonen M. Non-alcoholic and alcoholic Fatty Liver Disease – two Diseases of Affluence associated with the Metabolic Syndrome and Type 2 Diabetes: the FIN-D2D Survey. <i>BMC Public Health</i> 2010; 10: 237. (DPS) [PMID: 11333990] Tuomilehto J, Lindström J, Eriksson JG, Valle TT, Hämäläinen H, Ilanne-Parikka P, Keinänen-Kiukaanniemi S, Laakso M, Louheranta A, Rastas M, Salminen V, Uusitupa M, for the Finnish Diabetes Prevention Study Group. Prevention of Type 2 Diabetes Mellitus by Changes in Lifestyle among Subjects with Impaired Glucose Tolerance. <i>N Engl J Med</i> 2001; 344: 1343-1350. (DR'S EXTRA) [PMID: 21186108] Diet, fitness and metabolic syndrome - The DR's EXTRA Study. Kouki R, Schwab U, Lakka TA, Hassinen M, Savonen K, Komulainen P, Krachler B, Rauramaa R. <i>Nutr Metab Cardiovasc Dis.</i> 2010 Dec 24. (FUSION stage 2) [PMID:

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Study		Study design	Ethnicity	Total sample size (N)	Sample QC		Samples in analyses (N)	Anthropometric assessment method	References
Short name	Full name				Call rate*	Other exclusions			
D2D2007.DPS.DRSEXTRA.FUSION2.METSIM cases	FIN-D2D 2007. Diabetes Prevention Study, The Dose Responses to Exercise Training Study, FUSIONstage2, and METabolic Syndrome In Men	FIN-D2D 2007 and DR's EXTRA are population-based. DPS is a clinical prevention trial. For FUSION stage 2 and METSIM, samples genotyped followed a case/control design. Cases were analyzed separately from controls.	White European	2923	0.95	1) Gender discrepant; 2) unexpected duplicate; 3) missing all phenotype; 4) first-degree related	2754	measured	17463248] Scott, L.J., Mohlke, K.L., Bonnycastle, L.L., Willer, C.J., Li, Y., Duren, W.L., Erdos, M.R., Stringham, H.M., Chines, P.S., Jackson, A.U., Prokunina-Olsson L., Ding, C.J., Swift, A.J., Narisu, N., Hu, T., Pruim, R., Xiao, R., Li, X.Y., Conneely, K.N., Riebow, N.L., Sprau, A.G., Tong, M., White, P.P., Hetrick, K.N., Barhnart, M.W., Bark, C.W., Goldstein, J.L., Watkins, L., Xiang, F., Saramies, J., Buchanan, T.A., Watanabe, R.M., Valle, T.T., Kinnunen, L., Abecasis, G.R., Pugh, E.W., Doheny, K.F., Bergman, R.N., Tuomilehto, J., Collins, F.S., Boehnke, M. (2007) A Genome-Wide Association Study of Type 2 Diabetes in Finns Detects Multiple Susceptibility Variants. Science 316:1341-1345. (METSIM) [PMID: 19223598] Stancakova A, Javorsky M, Kuulasmaa T, Haffner SM, Kuusisto J, Laakso M: Changes in insulin sensitivity and insulin release in relation to glycemia and glucose tolerance in 6416 Finnish men. Diabetes 58:1212-1221, 2009. (FIN-D2D 2007) [PMID: 20459722] Kotronen A, Yki-Järvinen H, Männistö S, Saarikoski L, Korpi-Hyövälti E, Oksa H, Saltevo J, Saaristo T, Sundvall J, Tuomilehto J, Peltonen M. Non-alcoholic and alcoholic Fatty Liver Disease – two Diseases of Affluence associated with the Metabolic Syndrome and Type 2 Diabetes: the FIN-D2D Survey. BMC Public Health 2010; 10: 237. (DPS) [PMID: 11333990] Tuomilehto J, Lindström J, Eriksson JG, Valle TT, Hämäläinen H, Ilanne-Parikka P, Keinänen-Kiukkaanniemi S, Laakso M, Louheranta A, Rastas M, Salminen V, Uusitupa M, for the Finnish Diabetes Prevention Study Group. Prevention of Type 2 Diabetes Mellitus by Changes in Lifestyle among Subjects with Impaired Glucose Tolerance. N Engl J Med 2001; 344: 1343-1350. (DR'S EXTRA) [PMID: 21186108] Diet, fitness and metabolic syndrome - The DR's EXTRA Study. Kouki R, Schwab U, Lakka TA, Hassinen M, Savonen K, Komulainen P, Krachler B, Rauramaa R. Nutr Metab Cardiovasc Dis. 2010 Dec 24. (FUSION stage 2) [PMID: 17463248] Scott, L.J., Mohlke, K.L., Bonnycastle, L.L., Willer, C.J., Li, Y., Duren, W.L., Erdos, M.R., Stringham, H.M., Chines, P.S., Jackson, A.U., Prokunina-Olsson L., Ding, C.J., Swift, A.J., Narisu, N., Hu, T., Pruim, R., Xiao, R., Li, X.Y., Conneely, K.N., Riebow,

Supplementary Table 2. Study design, number of individuals, and sample quality control for studies in stage 1, stage 2, or ancillary analyses

Study		Study design	Ethnicity	Total sample size (N)	Sample QC		Samples in analyses (N)	Anthropometric assessment method	References
Short name	Full name				Call rate*	Other exclusions			
									N.L., Sprau, A.G., Tong, M., White, P.P., Hetrick, K.N., Barhnart, M.W., Bark, C.W., Goldstein, J.L., Watkins, L., Xiang, F., Saramies, J., Buchanan, T.A., Watanabe, R.M., Valle, T.T., Kinnunen, L., Abecasis, G.R., Pugh, E.W., Doheny, K.F., Bergman, R.N., Tuomilehto, J., Collins, F.S., Boehnke, M. (2007) A Genome-Wide Association Study of Type 2 Diabetes in Finns Detects Multiple Susceptibility Variants. <i>Science</i> 316:1341-1345. (METSIM) [PMID: 19223598]
DILGOM	Dietary, life style, and genetic determinants of obesity and metabolic syndrome	Population-based	White European	3997	≥ 95%	1) heterozygosity <23.9% or >27.6%; 2) ethnic outliers; 3) related individuals and duplicates.	3938	measured	[PMID: 20138944] Kontinen H, Männistö S, Sarlio-Lähteenkorva S, Silventoinen K, Haukkala A. Emotional eating, depressive symptoms and self-reported food consumption. A population-based study. <i>Appetite</i> . 2010 Jun;54(3):473-9. [PMID: 20844574] Inouye M, Silander K, Hamalainen E, Salomaa V, Harald K, Jousilahti P, Männistö S, Eriksson JG, Saarela J, Ripatti S, Perola M, van Ommen GJ, Taskinen MR, Palotie A, Dermitzakis ET, Peltonen L. An immune response network associated with blood lipid levels. <i>PLoS Genet</i> . 2010 Sep 9;6(9). Peltonen, M., Harald, K., Männistö, S., Saarikoski, L., Peltomäki, P., Lund, L., et al. (2008). The National FINRISK 2007 Study. Helsinki: Publications of the National Public Health Institute, B 34/2008.

Supplementary Table 2. Study design, number of individuals, and sample quality control for studies in stage 1, stage 2, or ancillary analyses

Study		Study design	Ethnicity	Total sample size (N)	Sample QC		Samples in analyses (N)	Anthropometric assessment method	References
Short name	Full name				Call rate*	Other exclusions			
GoDARTS controls	GoDARTS controls	Population-based	White European	3962	For SNPs with MAF $\geq 5\%$: $\geq 95\%$ For SNPs with $\geq 1\%$ MAF $< 5\%$: $\geq 99\%$	MAF $\geq 1\%$; genotype clustering, call rate, HWE, MI of alleles, concordance rate, correct SNP mapping	3709	measured	[PMID: 17429603] Kimber CH, Doney AS, Pearson ER, McCarthy MI, Hattersley AT, Leese GP, Morris AD, Palmer CN: TCF7L2 in the Go-DARTS study: evidence for a gene dose effect on both diabetes susceptibility and control of glucose levels. <i>Diabetologia</i> 2007, 50:1186-1191. [PMID:21186350] Zhou K, Bellenguez C, Spencer CC, Bennett AJ, Coleman RL, Tavendale R, Hawley SA, Donnelly LA, Schofield C, Groves CJ, Burch L, Carr F, Strange A, Freeman C, Blackwell JM, Bramon E, Brown MA, Casas JP, Corvin A, Craddock N, Deloukas P, Dronov S, Duncanson A, Edkins S, Gray E, Hunt S, Jankowski J, Langford C, Markus HS, Mathew CG, Plomin R, Rautanen A, Sawcer SJ, Samani NJ, Trembath R, Viswanathan AC, Wood NW, Harries LW, Hattersley AT, Doney AS, Colhoun H, Morris AD, Sutherland C, Hardie DG, Peltonen L, McCarthy MI, Holman RR, Palmer CN, Donnelly P, Pearson ER: Common variants near ATM are associated with glycemic response to metformin in type 2 diabetes. <i>Nature genetics</i> 2011, 43:117-120.
GoDARTS cases	GoDARTS cases	Population-based	White European	3582	For SNPs with MAF $\geq 5\%$: $\geq 95\%$ For SNPs with $\geq 1\%$ MAF $< 5\%$: $\geq 99\%$	MAF $\geq 1\%$; genotype clustering, call rate, HWE, MI of alleles, concordance rate, correct SNP mapping	3313	measured	[PMID: 17429603] Kimber CH, Doney AS, Pearson ER, McCarthy MI, Hattersley AT, Leese GP, Morris AD, Palmer CN: TCF7L2 in the Go-DARTS study: evidence for a gene dose effect on both diabetes susceptibility and control of glucose levels. <i>Diabetologia</i> 2007, 50:1186-1191. [PMID:21186350] Zhou K, Bellenguez C, Spencer CC, Bennett AJ, Coleman RL, Tavendale R, Hawley SA, Donnelly LA, Schofield C, Groves CJ, Burch L, Carr F, Strange A, Freeman C, Blackwell JM, Bramon E, Brown MA, Casas JP, Corvin A, Craddock N, Deloukas P, Dronov S, Duncanson A, Edkins S, Gray E, Hunt S, Jankowski J, Langford C, Markus HS, Mathew CG, Plomin R, Rautanen A, Sawcer SJ, Samani NJ, Trembath R, Viswanathan AC, Wood NW, Harries LW, Hattersley AT, Doney AS, Colhoun H, Morris AD, Sutherland C, Hardie DG, Peltonen L, McCarthy MI, Holman RR, Palmer CN, Donnelly P, Pearson ER: Common variants near ATM are associated with glycemic response to metformin in type 2 diabetes. <i>Nature genetics</i> 2011, 43:117-120.

Supplementary Table 2. Study design, number of individuals, and sample quality control for studies in stage 1, stage 2, or ancillary analyses

Study		Study design	Ethnicity	Total sample size (N)	Sample QC		Samples in analyses (N)	Anthropometric assessment method	References
Short name	Full name				Call rate*	Other exclusions			
EGCUT controls	Estonian Genome Center of University of Tartu	Case-control	White European	940	≥ 95%	1) Ambiguous sex; 2) cryptic relatedness; 3) missing phenotype; 4) non-European descent	940	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. PLoS ONE 4(5): e5472. Metspalu A. The Estonian Genome Project. Drug Development Research 62, 97-101 (2004).
EGCUT CAD	Estonian Genome Center of University of Tartu	Case-control	White European	700	≥ 95%	1) Ambiguous sex; 2) cryptic relatedness; 3) missing phenotype; 4) non-European descent	700	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. PLoS ONE 4(5): e5472. Metspalu A. The Estonian Genome Project. Drug Development Research 62, 97-101 (2004).
EGCUT T2D	Estonian Genome Center of University of Tartu	Case-control	White European	967	≥ 95%	1) Ambiguous sex; 2) cryptic relatedness; 3) missing phenotype; 4) non-European descent	967	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. PLoS ONE 4(5): e5472. Metspalu A. The Estonian Genome Project. Drug Development Research 62, 97-101 (2004).
ELY	Ely Study	Population-based	White European	1625	≥ 95%	1) Missing body weight and height. 2) Heterozygosity 3) gender check	1600	measured	[PMID: 17257284] Forouhi NG, et al. Incidence of Type 2 diabetes in England and its association with baseline impaired fasting glucose: the Ely study 1990-2000. Diabet Med. 2007 Feb;24(2):200-7.
EPIC Controls	EPIC (European Prospective Investigation into Cancer) Norfolk Cohort, non-DM controls	Population-based	White European	978	≥ 95%	1) Missing body weight and height, or case-control status 2) Heterozygosity 3) gender check	963	measured	[PMID: 10466767] Day N, Oakes S, Luben R, et al. EPIC-Norfolk: study design and characteristics of the cohort. European Prospective Investigation of Cancer. Br J Cancer 1999, 80:95-103 [PMID: 18454148] Loos RJ et al. Common variants near MC4R are associated with fat mass, weight and risk of obesity. Nat Genet 2008, 40:768-75
EPIC Cases	EPIC (European Prospective Investigation into Cancer) Norfolk Cohort, DM cases	T2D case series	White European	735	≥ 95%	1) Missing body weight and height, or case-control status 2) Heterozygosity 3) gender check	727	measured	[PMID: 10466767] Day N, Oakes S, Luben R, et al. EPIC-Norfolk: study design and characteristics of the cohort. European Prospective Investigation of Cancer. Br J Cancer 1999, 80:95-103 [PMID: 18454148] Loos RJ et al. Common variants near MC4R are associated with fat mass, weight and risk of obesity. Nat Genet 2008, 40:768-75
FENLAND	Fenland Study	Population-based	White European	3251	≥ 95%	1) Missing body weight and height. 2) Heterozygosity 3) gender check	3186	measured	[PMID: 19079261] Willer CJ, Speliotes EK, Loos RJ, et al. Six new loci associated with body mass index highlight a neuronal influence on body weight regulation. Nat Genet 2009, 41:25-34.

Supplementary Table 2. Study design, number of individuals, and sample quality control for studies in stage 1, stage 2, or ancillary analyses

Study		Study design	Ethnicity	Total sample size (N)	Sample QC		Samples in analyses (N)	Anthropometric assessment method	References
Short name	Full name				Call rate*	Other exclusions			
GLACIER	Gene x Lifestyle interactions And Complex traits Involved in Elevated disease Risk	Prospective cohort study	White European	6311	≥ 95%	1. Missing phenotype 2. Duplicates 3. Call rate 4. Heterozygosity 5. Gender fails	6047	measured	[PMID: 19164386] Renström F, Shungin D, Johansson I, the MAGIC investigators, Florez JC, Hallmans G, Hu FB, Franks PW. Genetic predisposition to long-term non-diabetic deteriorations in glucose homeostasis: ten-year follow-up of the GLACIER Study. <i>Diabetes</i> . 60(1): 345-54 2011 [PMID: 14660243] Renström F, Payne F, Nordström A, Brito EC, Rolandsson O, Hallmans G, Barosso I, Nordström P, Franks PW: the GIANT consortium. Replication and extension of genome-wide association study results for obesity in 4,923 adults from Northern Sweden. <i>Hum Mol Genet</i> . 18(8):1489-96 2009
HNR	Heinz Nixdorf Recall Study	Population-based	White European	4570	≥ 97%	1) heterozygosity; 2) ethnic outliers; 3) related individuals and duplicates; 4) missing phenotypes	4518	measured	[PMID: 16121757] Stang A. et al. Baseline recruitment and analyses of nonresponse of the Heinz Nixdorf Recall Study: identifiability of phone numbers as the major determinant of response. <i>Eur J Epidemiol</i> . 2005;20(6):489-96. [PMID: 20616309] Pechlivanis S. et al. Coronary artery calcification and its relationship to validated genetic variants for diabetes mellitus assessed in the Heinz Nixdorf recall cohort. <i>Arterioscler Thromb Vasc Biol</i> . 2010 Sep;30(9):1867-72.
HUNT.TROMSO controls	HUNT2 and Tromsø 4	HUNT2 and Tromsø 4 are population-based studies, but samples genotyped followed a case/control design. Cases were analyzed separately from controls.	White European	1685	0.95	1) Gender discrepant; 2) unexpected duplicate; 3) missing all phenotypes; 4) first-degree related	1290	measured	(HUNT2) Holmen J, Midtjell K, Krüger Ø, Langhammer A, Holmen TL, Bratberg G. The Nord-Trøndelag Health Study 1995-97 (HUNT 2): Objectives, contents, methods and participation. <i>Norsk Epidemiologi</i> 2003; 13:19-32. (Tromsø 4) [PMID: 21422063] Jacobsen BK, Eggen AE, Mathiesen EB, Wilsgaard T, Njolstad I. Cohort profile: The Tromsø Study. <i>Int J Epidemiol</i> ; April 21, 2011.
HUNT.TROMSO cases	HUNT2 and Tromsø 4	HUNT2 and Tromsø 4 are population-based studies, but samples genotyped followed a case/control design. Cases were analyzed separately from controls.	White European	1303	0.95	1) Gender discrepant; 2) unexpected duplicate; 3) missing all phenotypes; 4) first-degree related	1078	measured	(HUNT2) Holmen J, Midtjell K, Krüger Ø, Langhammer A, Holmen TL, Bratberg G. The Nord-Trøndelag Health Study 1995-97 (HUNT 2): Objectives, contents, methods and participation. <i>Norsk Epidemiologi</i> 2003; 13:19-32. (Tromsø 4) [PMID: 21422063] Jacobsen BK, Eggen AE, Mathiesen EB, Wilsgaard T, Njolstad I. Cohort profile: The Tromsø Study. <i>Int J Epidemiol</i> ; April 21, 2011.
IMPROVE	Carotid Intima-Media Thickness and IMT-Progression as predictors of Vascular Events in a high risk European population	Individuals with high CVD risk	White European	3449	≥ 95%	1) ambiguous sex; 2) cryptic relatedness; 3) non-european descent	3449	measured	[PMID: 19952003] Baldassarre D. et al. Cross-sectional analysis of baseline data to identify the major determinants of carotid intima-media thickness in a European population: the IMPROVE study. <i>Eur Heart J</i> . 2010 Mar;31(5):614-22.
KORA S3 METABO	Cooperative Health Research in the Region of Augsburg	Population-based	White European	1278	0.93	none	1278	measured	[PMID: 16032514] Wichmann HE et al. KORA-gen--resource for population genetics, controls and a broad spectrum of disease phenotypes. <i>Gesundheitswesen</i> 67

Supplementary Table 2. Study design, number of individuals, and sample quality control for studies in stage 1, stage 2, or ancillary analyses

Study		Study design	Ethnicity	Total sample size (N)	Sample QC		Samples in analyses (N)	Anthropometric assessment method	References
Short name	Full name				Call rate*	Other exclusions			
									Suppl 1, S26-30 (2005).
KORA S4 METABO	Cooperative Health Research in the Region of Augsburg	Population-based	White European	1222	0.93	none	1222	measured	[PMID: 16032514] Wichmann HE et al. KORA-gen--resource for population genetics, controls and a broad spectrum of disease phenotypes. <i>Gesundheitswesen</i> 67 Suppl 1, S26-30 (2005).
LURIC	Ludwigshafen Risk and Cardiovascular Health Study	Case-control	White European	2929	≥ 95%	1) Gender ambiguity; 2) Missing hip and waist measurement; 3) age < 18	2895	measured	[PMID: 11258203] Winkelmann, BR, Marz, W, Boehm, BO, Zotz, R, Hager, J, Hellstern, P and Senges, J, Rationale and design of the LURIC study--a resource for functional genomics, pharmacogenomics and long-term prognosis of cardiovascular disease, <i>Pharmacogenomics</i> , 2001, 2: S1-73.
MORGAM Controls	MONica Risk, Genetics, Archiving and Monograph	Population-based follow-up cohorts.	White European	~2650	≥ 95%	1) heterozygosity <18.1% or >20.7%; 2) >10.0% discordance with Sequenom genotypes 3) ethnic outliers; 4) related individuals and duplicates.	2473	measured	[PMID: 15561751] Evans A, Salomaa V, Kulathinal S, Asplund K, Cambien F, Ferrario M, Perola M, Peltonen L, Shields D, Tunstall-Pedoe H, Kuulasmaa K, for the MORGAM Project. MORGAM (an international pooling of cardiovascular cohorts). <i>Int J Epidemiol</i> 2005;34:21-27. 2. Tunstall-Pedoe H, editor. Prepared by Tunstall-Pedoe H, Kuulasmaa K, Tolonen H, Davidson M, Mendis S with 64 other contributors for The WHO MONICA Project. MONICA Monograph and Multimedia Sourcebook. Geneva: World Health Organization; 2003. ISBN 92 4 156223 4. Also available from http://www.ktl.fi/monica/public/monograph.html .
MORGAM Cases	MONica Risk, Genetics, Archiving and Monograph	CVD cases from population-based follow-up cohorts.	White European	~2150	≥ 95%	1) heterozygosity <18.1% or >20.7%; 2) >10.0% discordance with Sequenom genotypes 3) ethnic outliers; 4) related individuals and duplicates.	2052	measured	[PMID: 15561751] Evans A, Salomaa V, Kulathinal S, Asplund K, Cambien F, Ferrario M, Perola M, Peltonen L, Shields D, Tunstall-Pedoe H, Kuulasmaa K, for the MORGAM Project. MORGAM (an international pooling of cardiovascular cohorts). <i>Int J Epidemiol</i> 2005;34:21-27. 2. Tunstall-Pedoe H, editor. Prepared by Tunstall-Pedoe H, Kuulasmaa K, Tolonen H, Davidson M, Mendis S with 64 other contributors for The WHO MONICA Project. MONICA Monograph and Multimedia Sourcebook. Geneva: World Health Organization; 2003. ISBN 92 4 156223 4. Also available from http://www.ktl.fi/monica/public/monograph.html .
NSHD	MRC National Survey of Health & Development	Birth cohort	White European	5362	≥ 95%	1) Missing body weight and height. 2) Heterozygosity 3) gender check	988	measured	[PMID: 16204333] Wadsworth M, Kuh D, Richards M, Hardy R. Cohort Profile: The 1946 National Birth Cohort (MRC National Survey of Health and Development). <i>Int J Epidemiol</i> . 35(1):49-54. (2006) [PMID: 19880856] Hardy R, Wills AK, Wong A, Elks CE, Wareham NJ, Loos RJ, Kuh D, Ong KK. Life course

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Study		Study design	Ethnicity	Total sample size (N)	Sample QC		Samples in analyses (N)	Anthropometric assessment method	References
Short name	Full name				Call rate*	Other exclusions			
									variations in the associations between FTO and MC4R gene variants and body size. Hum Mol Genet. 19(3):545-52 (2010).
PIVUS	Prospective Investigation of the Vasculature in Uppsala Seniors	Population-based	White European	999	≥90%	1) Related individuals and duplicates 2) Heterozygosity, (F-mean(F))/sd(Z) ≥ 5 3) Missing all phenotypes	978	measured	[PMID: 16141402] Lind, L. et al. A comparison of three different methods to evaluate endothelium-dependent vasodilation in the elderly: the Prospective Investigation of the Vasculature in Uppsala Seniors (PIVUS) study. Arterioscler Thromb Vasc Biol 25, 2368-75 (2005).
ULSAM	Uppsala Longitudinal Study of Adult Men	Population-based	White European	1221	≥90%	1) Related individuals and duplicates 2) Heterozygosity, (F-mean(F))/sd(Z) ≥ 5 3) Missing all phenotypes	1116	measured	[PMID: 16030278] Ingelsson, E. et al. Insulin resistance and risk of congestive heart failure. JAMA 20, 334-41 (2005).
STR	Swedish Twin Registry	Population-based	White European	2702	≥90%	1) Heterozygosity, (F-mean(F))/sd(Z) ≥ 5 2) Missing all phenotypes	2430	measured	[PMID: 17254424] Lichtenstein, P. et al. The Swedish Twin Registry in the third millennium: an update. Twin Res Hum Genet 9, 875-82 (2006).
THISEAS controls	The Hellenic study of Interactions between Snps and Eating in Atherosclerosis Susceptibility	CAD controls	European	981	0.97	1) heterozygosity; 2) ethnic outliers; 3) duplicates; 4) missing phenotypes; 5) call rate < 0.95	981	measured	[PMID: 20167083] Theodoraki EV. et al. Fibrinogen beta variants confer protection against coronary artery disease in a Greek case-control study. BMC Med Genet. 2010 Feb 18;11:28.
THISEAS cases	The Hellenic study of Interactions between Snps and Eating in Atherosclerosis Susceptibility	CAD cases	European	514	0.97	1) heterozygosity; 2) ethnic outliers; 3) duplicates; 4) missing phenotypes; 5) call rate < 0.95	514	measured	[PMID: 20167083] Theodoraki EV. et al. Fibrinogen beta variants confer protection against coronary artery disease in a Greek case-control study. BMC Med Genet. 2010 Feb 18;11:28.
WHITEHALL	The Whitehall II study	Cohort of London-based civil servants	European descent	3413	≥ 95%	1) Missing body weight and height. 2) Heterozygosity 3) gender check	3377	measured	[PMID: 15576467] Marmot M, Brunner E. Cohort Profile: the Whitehall II study. Int J Epidemiol. 2005; 34:251-6. [PMID: 21441441] Jensen AC et al. Associations of common genetic variants with age-related changes in fasting and postload glucose: evidence from 18 years of follow-up of the Whitehall II cohort. Diabetes. 2011; 60:1617-23.

Supplementary Table 2. Study design, number of individuals, and sample quality control for studies in stage 1, stage 2, or ancillary analyses

Study		Study design	Ethnicity	Total sample size (N)	Sample QC		Samples in analyses (N)	Anthropometric assessment method	References
Short name	Full name				Call rate*	Other exclusions			
WTCCC T2D nonGWAS	Wellcome Trust Case Control Consortium T2D	Population-based	White European	1335	For SNPs with MAF $\geq 5\%$: $\geq 95\%$ For SNPs with $\geq 1\%$ MAF $< 5\%$: $\geq 99\%$	MAF $\geq 1\%$; genotype clustering, call rate, HWE, MI of alleles, concordance rate, correct SNP mapping	1077	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)
Ancillary Studies									
Essen Obesity Study (Essen Case-Control GWAS)	Essen Obesity Study (Essen Case-Control GWAS)	Case-Control	White European	888	$\geq 95\%$	1) ethnic outliers; 2) related individuals and duplicates; 3) Missing BMI and age; 4) per sample call rate $\geq 95\%$.	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)	Essen Obesity Study (Essen Obesity Trio GWAS)	Family-based (trios)	White European	2,183	$\geq 95\%$	1) Mendelian inconsistencies per family $> 5\%$; 2) ethnic outliers; 3) duplicates; 4) Missing BMI and age in offspring; 5) per sample call rate $\geq 95\%$.	2,115	measured	[PMID: 21131291] Jarick, I. et al. Novel common copy number variation for early onset extreme obesity on chromosome 11q11 identified by a genome-wide analysis. Hum Mol Genet. 15, 20(4):840-52 (2011).
French Extreme Obesity Study	French Extreme Obesity Study	Case-Control	White European	2,796	$\geq 95\%$	1) Ethnic outliers 2) Missing BMI 3) Per sample call rate > 0.9	2,652	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. Nature Genetics 2009 Feb;41(2):157-9. Epub 2009 Jan 18. Unpublished data
GEO-IT	Genetic of Extreme Obesity-Italy	Case-Control	White European	3948	$\geq 98\%$	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 phenotype data	3509	measured	Unpublished data
GOYA	Genetics of Overweight Young Adults	Women - birth cohort, men - population based	White European	5536	$\geq 95\%$	2) individuals who did not cluster with CEU individuals 3) heterozygosity $< 30.2\%$ or $> 35\%$ 4) related individuals and duplicates 5) sex discrepancies	5373	women - self-reported men - taken from draft board records	[PMID: 21935397] Paternoster, L. et al. Genome-wide population-based association study of extremely overweight young adults - The GOYA study

Supplementary Table 2. Study design, number of individuals, and sample quality control for studies in stage 1, stage 2, or ancillary analyses

Study		Study design	Ethnicity	Total sample size (N)	Sample QC		Samples in analyses (N)	Anthropometric assessment method	References
Short name	Full name				Call rate*	Other exclusions			
						6) genotypes discordant with previous project			

* Call rate to exclude individuals for whom genotyping success rate is less than a certain percentage (to exclude 'bad' samples/DNA)

Supplementary Table 3. Information on genotyping methods, quality control, imputation, and statistical analysis for studies included in stage 1 and stage 2 and ancillary studies

Study	Genotyping						Imputation			Association analyses	
	Platform	Genotype calling algorithm	Inclusion criteria			SNPs that met QC criteria	Imputation software	Inclusion criteria		SNPs in meta-analysis	Analyses software
			MAF	Call rate*	P for HWE			MAF	Imputation quality*		
Stage 1:											
ADVANCE controls	Illumina HumanHap550 v.3	BeadStudio (GenCall)	NA	≥ 98.5%	> 0.001	557,883	IMPUTE	NA	proper-info ≥ 0.40	2307251	SNPTEST
ADVANCE cases	Illumina HumanHap550 v.3	BeadStudio (GenCall)	NA	≥ 95%	> 0.001	557,883	IMPUTE	NA	proper-info ≥ 0.40	2295533	SNPTEST
AGES	Illumina Human370CNV	BeadStudio	≥ 1%	≥ 95%	> 10 ⁻⁶	308,340	MACH	none	none	2465835	ProbABEL
ARIC	Affymetrix Genome-Wide Human SNP Array 6.0	Birdseed	≥ 1%	≥ 90%	> 10 ⁻⁶	685,812	MACH v1.0.16	>1%		2557252	ProbABEL
B58C-T1DGC	Illumina HumanHap 550 V.1	ILLUMINUS	> 0%	-	-	539,458	MACH	> 0%	r2-hat ≥ 0.30	2507988	ProbABEL
B58C-WTCCC	Affymetrix GeneChip Human Mapping 500K	CHIAMO	> 0%	-	-	490,032	IMPUTE	≥ 0%	proper-info ≥ 0.40	2448428	SNPTEST
BRIGHT	Affymetrix GeneChip Human Mapping 500K	CHIAMO	≥ 5%	≥ 95%	> 10 ⁻⁶	387,666	IMPUTE	none	proper-info ≥ 0.40	2429493	SNPTEST
CAD WTCCC	Affymetrix GeneChip Human Mapping 500K	CHIAMO	> 5%	≥ 95%	> 10 ⁻⁶	387,667	IMPUTE	none	proper-info ≥ 0.40	2430162	SNPTEST
CAPS1 cases	Affymetrix GeneChip Human Mapping 500K	BRLMM	≥ 1%	≥ 95%	> 10 ⁻⁷	330,124	IMPUTE	none	proper-info ≥ 0.40	2388288	SNPTEST
CAPS1 controls	Affymetrix GeneChip Human Mapping 500K	BRLMM	≥ 1%	≥ 95%	> 10 ⁻⁷	330,124	IMPUTE	none	proper-info ≥ 0.40	2391197	SNPTEST
CAPS2 cases	Affymetrix GeneChip Human Mapping 5.0K	BLRMM-P	≥ 1%	≥ 95%	> 10 ⁻⁷	348,163	IMPUTE	none	proper-info ≥ 0.40	2416628	SNPTEST
CAPS2 controls	Affymetrix GeneChip Human Mapping 5.0K	BLRMM-P	≥ 1%	≥ 95%	> 10 ⁻⁷	348,163	IMPUTE	none	proper-info ≥ 0.40	2392236	SNPTEST
CHS	Illumina 370CNV	BeadStudio	NA	≥ 97%	≥ 10 ⁻⁵	306,655	BimBam v0.99	NA	NA	2333818	R 2.10
CoLaus	Affymetrix GeneChip Human Mapping 500K	BRLMM	≥ 1%	≥ 70%	> 10 ⁻⁷	390,631	IMPUTE	≥ 0%	r2-hat ≥ 0.30	2483593	Matlab
COROGENE controls	Illumina Human 660 000 BeadChip	ILLUMINUS	≥ 5%	≥ 95%	> 10 ⁻⁶	554,987	MACH	-	-	2543887	PLINK, ProbABEL
COROGENE cases	Illumina Human 660 000 BeadChip	ILLUMINUS	≥ 5%	≥ 95%	> 10 ⁻⁶	554,987	MACH	-	-	2543887	PLINK, ProbABEL
deCODE	Illumina HumanHap300 or HumanHapCNV370	BeadStudio	≥ 1%	≥ 96%	> 10 ⁻⁶	290,447	IMPUTE	> 0%	proper-info ≥ 0.40	2456118	SNPTEST
DGI controls	Affymetrix 500K	BRLMM	≥ 1%	≥ 95%	> 10 ⁻⁶	386,731	MACH	> 0%	r2-hat ≥ 0.30	2375087	MACH2QTL
DGI Cases	Affymetrix 500K	BRLMM	≥ 1%	≥ 95%	> 10 ⁻⁶	386,731	MACH	> 0%	r2-hat ≥ 0.30	2375087	MACH2QTL
EGCUT	Illumina Human370CNV-duo /-quad	GenomeStudio	≥ 1%	≥ 95%	> 10 ⁻⁶	316,924	IMPUTE	NA	NA	2552661	SNPTEST
EPIC-Obesity Study	Affymetrix GeneChip Human Mapping 500K	BRLMM	≥ 1%	≥ 90%	> 10 ⁻⁶	382,037	IMPUTE	≥ 1%	proper-info ≥ 0.40	2565027	SNPTEST
ERF	Illumina 318k, 370k and affymetrix 250k	Beadstudio, BRLMM	≥ 1%	≥ 95%	> 10 ⁻⁶	up to 427922	MACH	≥ 1%	NA	2543887	ProbABEL
FamHS	ILLUMINA 550K	BEADSTUDIO-GENCALL v3.0	≥ 1%	N.A.	≥ 10 ⁻⁶	456,293	MACH	exclusions	no exclusions	2543887	SAS
Fenland	Affymetrix SNPs5.0	BRLMM	≥ 1%	≥ 90%	> 10 ⁻⁶	362,055	IMPUTE	> 0%	proper-info ≥ 0.40	2555899	SNPTEST
FRAM	Affymetrix 500K supplemental	BRLMM	≥ 1%	≥ 97%	> 10 ⁻⁶	378,163	MACH v1.0.15	> 1%	RATIO ≥ 0.3	2543800	R
FUSION controls	Illumina Infinium™ II HumanHap300 BeadChip	BeadStudio	≥ 1%	≥ 90%	≥ 10 ⁻⁶	315,635	MACH	> 5%	r2-hat ≥ 0.30	2466546	ProbABEL
FUSION Cases	Illumina Infinium™ II HumanHap300 BeadChip	BeadStudio	≥ 1%	≥ 90%	≥ 10 ⁻⁶	315,635	MACH	> 5%	r2-hat ≥ 0.30	2466546	ProbABEL
GENMETS	Illumina Human610K	ILLUMINUS	> 1%	> 95%	0.000001	555,388	MACH 1.16	> 1%	Rsq > 0.3	2543887	ProbABEL

Supplementary Table 3. Information on genotyping methods, quality control, imputation, and statistical analysis for studies included in stage 1 and stage 2 and ancillary studies

Study	Genotyping						Imputation			Association analyses	
	Platform	Genotype calling algorithm	Inclusion criteria			SNPs that met QC criteria	Imputation software	Inclusion criteria		SNPs in meta-analysis	Analyses software
			MAF	Call rate*	P for HWE			MAF	Imputation quality*		
GerMIFS1	Affymetrix Mapping 500k Array Set	BRLMM	≥ 1%	≥ 98%	> 10 ⁻⁴	266,622	IMPUTE	≥ 1%	Proper-info ≥ 0	2543887	SNPTEST
GerMIFS2	Affymetrix Genome-Wide Human SNP Array 6.0	Birdseed	≥ 1%	≥ 98%	> 10 ⁻⁴	560,682	IMPUTE	≥ 1%	Proper-info ≥ 0	2543887	SNPTEST
GOOD	Illumina Infinium HumanHap 610K	BeadStudio	≥ 1%	≥ 98%	> 10 ⁻⁶	521,160	MACH	-	-	2543887	MACH2QTL
HBCS	Illumina custom made BeadChip Human 670-Quad	Illuminus	≥ 5%	≥ 95%	> 10 ⁻⁶	533,491	MACH	-	-	2543887	PLINK, ProbABEL
KORA S3	Affymetrix 500K	BRLMM	none	≥ 90%	none	490,032	MACH v1.0.9	none	none	2557252	MACH2DAT
KORA S4	Affymetrix 6.0	Birdseed	none	≥ 93%	none	651,596	MACH v1.0.15	none	none	2543887	MACH2DAT
MGS	Affymetrix Genome-Wide Human SNP Array 6.0	Birdsuite 2.0	≥ 1%	≥ 95%	> 10 ⁻⁶	696,492	MACH 1.0	≥ 1%	R2 > 0.3	696492	PLINK and local software
MICROS	Illumina Infinium II HumanHap 300v2	BeadStudio	≥ 1%	≥ 98%	> 10 ⁻⁶	292,917	MACH	≥ 0%	r2_hat ≥ 0.3	2437189	ProbABEL
Migen (cases)	Affymetrix 6.0 Array	Birdseed	> 0.01	> 0.95	1 x 10 ⁻⁶	618,475	MACH(1.0.15)	> 0.01	r2_hat ≥ 0.30	2271906	MACH2QTL
Migen (controls)	Affymetrix 6.0 Array	Birdseed	> 0.02	> 0.96	2 x 10 ⁻⁶	618,475	MACH(1.0.15)	> 0.02	r2_hat ≥ 0.31	2271906	MACH2QTL
NBS WTCCC	Affymetrix GeneChip Human Mapping 500K	CHIAMO	> 5%	≥ 95%	> 10 ²⁶	387,667	IMPUTE	none	proper_info ≥ 0.40	2415681	SNPTEST
NFBC	Illumina HumanCNV 370DUO Analysis BeadChip	Standard Illumina BeadStudio	> 5%	≥ 95%	> 10 ²⁴	328,007	IMPUTE	none	proper_info ≥ 0.40	2458713	SNPTEST
NHS	Affymetrix Genome-Wide Human 6.0 array	Birdseed calling algorithm v2	≥ 2%	≥ 98%	> 10 ⁻⁴	704,409	MACH	≥ 2%	MACH r2_hat ≥ 0.8	2509969	ProbABEL
NSPHS	Illumina Infinium II HumanHap 300v2	BeadStudio	≥ 0%	≥ 98%	> 10 ⁻⁹	318,236	MACH	≥ 1%	r2_hat ≥ 0.3	2382230	ProbABEL
NTR and NESDA MDD controls	Perlegen / Affymetrix 600K gene chip	Perlegen proprietary	≥ 1%	≥ 95%	> 10 ⁻⁵	435,291	IMPUTE	≥ 1%	proper-info ≥ 0.40	2543887	SNPTEST
NTR and NESDA MDD cases	Perlegen / Affymetrix 600K gene chip	Perlegen proprietary	≥ 1%	≥ 95%	> 10 ⁻⁵	435,291	IMPUTE	≥ 1%	proper-info ≥ 0.40	2543887	SNPTEST
ORCADES	Illumina Infinium II HumanHap 300v2	BeadStudio	≥ 1%	≥ 98%	> 10 ⁻⁶	306,207	MACH	≥ 0%	r2_hat ≥ 0.3	2442003	ProbABEL
PLCO	Illumina HumanHap300 and Illumina HumanHap240	Bead Studio	none	≥ 90%	none	523,231	MACH	≥ 1%	MACH r2_hat ≥ 0.30	2527018	MACH2DAT
PROCARDIS	Illumina HumanHap300 BeadChips	Illumina Beadstudio 2.0 software	> 5%	≥ 95%	> 5x10 ²⁷	~820 000	IMPUTE	none	proper_info ≥ 0.40	2581539	SNPTEST
RUNMC	Illumina HumanHapCNV370	BeadStudio	≥ 1%	≥ 96%	> 10 ⁻⁶	312,199	IMPUTE	> 0%	proper-info ≥ 0.40	2465662	SNPTEST
RS1	Illumina / HumanHap 550 V.3	Beadstudio Genecall	≥ 1%	≥ 97.5%	> 10 ⁻⁶	512,349	MACH	≥ 1%	(O/E)σ2 ratio ≥ 0.1	2543887	MACH
SASBAC cases	Illumina HumanHap300+240S	BeadStudio (GenCall)	≥ 3%	≥ 90%	> 10 ⁻⁷	510,578	IMPUTE	none	proper-info ≥ 0.40	2491837	SNPTEST
SASBAC controls	Illumina HumanHap550 Affymetrix GeneChip	BeadStudio (GenCall)	≥ 3%	≥ 90%	> 10 ⁻⁷	512,223	IMPUTE	none	proper-info ≥ 0.40	2474796	SNPTEST
SardiNIA	Human Mapping 500K and Affymetrix 10K	BRLMM	≥ 5%	≥ 90%	> 10 ⁻⁶	356,359	MACH	> 0%	r2_hat ≥ 0.30	2251689	MACH2DAT
SEARCH	Illumina 610	Bead Studio	≥ 1%	≥ 95%	> 10 ⁻⁴	495,229	In house similar to Impute	≥ 1%	none	2552155	Routines written in C++
SHIP	Affymetrix Human SNP Array 6.0	Birdseed V2	≥ 0%	≥ 0%	none	869,224	IMPUTE v0.5.0	≥ 0%	NA	2748910	QUICKTEST 0.94 InforSense
Sorbs	500K Affymetrix GeneChip (250K Sty and 250K Nsp arrays, Affymetrix, Inc) and	BRLMM (Affy 500K); Birdseed (Array 6.0)	0.01	0.95	10 ⁻⁴	378,513	IMPUTE	MAF>1%, HWE<10 ⁻⁴	Proper-info > 0.4	2352557	SNPTEST

Supplementary Table 3. Information on genotyping methods, quality control, imputation, and statistical analysis for studies included in stage 1 and stage 2 and ancillary studies

Study	Genotyping						Imputation			Association analyses	
	Platform	Genotype calling algorithm	Inclusion criteria			SNPs that met QC criteria	Imputation software	Inclusion criteria		SNPs in meta-analysis	Analyses software
			MAF	Call rate*	P for HWE			MAF	Imputation quality*		
TWINSUK	Affymetrix Genome-Wide Human SNP Array 6.0 Illumina / HumanHap 300 & 550	Illuminus	≥ 1%	≥ 95%	> 10 ⁻⁶	295,702	IMPUTE	≥ 1%	proper-info ≥ 0.40	2544232	SNPTEST
T2D WTCCC	Affymetrix GeneChip Human Mapping 500K	CHIAMO	> 5%	≥ 95%	> 10 ⁻⁶	387,667	IMPUTE	none	proper-info ≥ 0.40	2425154	SNPTEST
VIS	Illumina Infinium II HumanCNV370v1	BeadStudio	≥ 1%	≥ 98%	> 10 ⁻⁶	305,068	MACH	≥ 0%	r ² _{hat} ≥ 0.3	2428707	ProbABEL
WGHS	Illumina humanHap 300 DuoPlus	Illumina BeadStudio 3.3	≥ 1%	≥ 98%	> 10 ⁻⁶	339,596	MACH v1.0.15	≥ 1%	MACH R2 ≥ 0.3	2608509	ProbABEL
YFS	Illumina custom made BeadChip Human 670-Quad	Illuminus	≥ 5%	≥ 95%	> 10 ⁻⁶	546,677	MACH	-	-	2543887	PLINK, ProbABEL

Supplementary Table 3. Information on genotyping methods, quality control, imputation, and statistical analysis for studies included in stage 1 and stage 2 and ancillary studies

Study	Genotyping					Imputation			Association analyses		
	Platform	Genotype calling algorithm	Inclusion criteria			SNPs that met QC criteria	Imputation software	Inclusion criteria		SNPs in meta-analysis	Analyses software
			MAF	Call rate*	P for HWE			MAF	Imputation quality*		
Stage 2 - In silico:											
B58C-REPL	Illumina HumanHap 550 / 610	Gencall	> 1%	≥ 95%	> 10 ⁻⁴	519,080	MACH	> 0%	r2-hat ≥ 0.30	replication set	ProbABEL
BHS	Illumina 610-Quad	Beadstudio	≥ 1%	≥ 95%	> 10 ⁻⁷	529,596	MACH	≥ 1%	r2 ≥ 0.30	2510147	MACH2DAT, MACH2QTL
HYPERGENES controls	Illumina Human1M-Duov3_B	Genome Studio	≥ 1%	≥ 90%	> 10 ⁻⁷	872,576	MACH	-	-	2543887	Matlab
HYPERGENES Cases	Illumina Human1M-Duov3_B	Genome Studio	≥ 1%	≥ 90%	> 10 ⁻⁷	872,576	MACH	-	-	2543887	Matlab
LifeLines	Illumina Cyto SNP12 v2	GenomeStudio	≥ 1%	≥ 95%	> 10 ⁻⁴	257581	BEAGLE v3.1.0	> 0%	no filtering on imputation quality	2472812	PLINK
PLCO2 controls	Illumina HumanHap 550K; Illumina HumanHap 610K	Bead Studio	> 0%	≥ 90%	-	525,262	IMPUTE	≥ 1%	proper-info ≥ 0.40	2538067	PLINK
PLCO2 cases	Illumina HumanHap 550K; Illumina HumanHap 610K	Bead Studio	> 0%	≥ 90%	-	525,262	IMPUTE	≥ 1%	proper-info ≥ 0.40	2564030	PLINK
PREVEND	Illumina HumanCytoSNP-12	GenomeStudio	≥ 1%	> 95%	0.0001	232,571	BEAGLE	≥ 1%	at meta-level	2289210	PLINK
QIMR	Illumina HumanHap 370 or 610	BeadStudio	≥ 1%	≥ 95%	> 10 ⁻⁶	271,069	MACH	NA	NA	2543887	PLINK
RS2	Illumina / HumanHap 550 V.3 DUO; Illumina / HumanHap 610 QUAD	Genomestudio Genecall	≥ 1%	≥ 97.5%	> 10 ⁻⁶	466,389	MACH	≥ 1%	(O/E)σ2 ratio ≥ 0.1	2543887	MACH
RS3	Illumina / HumanHap 610 QUAD	Genomestudio Genecall	≥ 1%	≥ 97.5%	> 10 ⁻⁶	466,389	MACH	≥ 1%	(O/E)σ2 ratio ≥ 0.1	2543887	MACH
TRAILS	Illumina Cyto SNP12 v2	GenomeStudio	≥ 1%	≥ 95%	> 10 ⁻⁴	260,127	IMPUTE2	≥ 0%	proper-info ≥ 0.0	2633433	SNPTEST
TWINGENE	Illumina HumanOmniExpress	GenomeStudio 2010.3	≥ 1%	≥ 97%	> 10 ⁻⁷	644,556	IMPUTE2	n.a.	n.a.	2585373	PLINK
UKBS2	Affymetrix Genome-Wide Human SNP Array 6.0	CHIAMO	≥ 1%	≥ 95%	> 10 ⁻⁶	754,588	IMPUTE	≥ 1%	info ≥ 0.40	2536100	SNPTEST

Supplementary Table 3. Information on genotyping methods, quality control, imputation, and statistical analysis for studies included in stage 1 and stage 2 and ancillary studies

Study	Genotyping								Analyses software
	Genotyping center	Genotype calling algorithm	Inclusion criteria					SNPs that met QC criteria	
			MAF	Call rate*	P for HWE	Discordance rate	Other		
Stage 2 - Metabochip:									
AMC-PAS	Wellcome Trust Sanger Institute	GenoSNP	none	≥ 98%	> 10 ⁻⁴	< 10%	na	169807	PLINK
CARDIOGENIC S controls	Wellcome Trust Sanger Institute	GenoSNP	none	≥ 98%	> 10 ⁻⁴	< 10%	na	177614	PLINK
CARDIOGENIC S cases	Wellcome Trust Sanger Institute	GenoSNP	none	≥ 98%	> 10 ⁻⁴	< 10%	na	177614	PLINK
D2D2007.DPS.D RSEXTRA.FUSION2.METSIM controls	Johns Hopkins University Genetic Resources Core Facility SNP Center at the Center for Inherited Disease Research, Baltimore MD	BeadStudio version 3.3.7, Gentrain version 1.0	> 0%	≥ 95%	none	none	Cluster Separation score less than 0.2 or which had more than 1 Replicate error as defined with the HapMap control samples.	145498	PLINK
D2D2007.DPS.D RSEXTRA.FUSION2.METSIM cases	Johns Hopkins University Genetic Resources Core Facility SNP Center at the Center for Inherited Disease Research, Baltimore MD	BeadStudio version 3.3.7, Gentrain version 1.0	> 0%	≥ 95%	none	none	Cluster Separation score less than 0.2 or which had more than 1 Replicate error as defined with the HapMap control samples.	138627	PLINK
DILGOM	The FIMM Technology Centre, Helsinki, Finland	GenCall	none	≥ 95%	none	none	none	183872	PLINK
GoDARTS controls	Cambridge Genomic Services, Department of Pathology, University of Cambridge, UK	GeneCall	≥ 1%	≥ 95%	> 10 ⁻⁶	1 duplicate pair with genotype concordance rate=1 and IBD=1	NA	180399	SNPTEST
GoDARTS cases	Cambridge Genomic Services, Department of Pathology, University of Cambridge, UK	GeneCall	≥ 1%	≥ 95%	> 10 ⁻⁶	1 duplicate pair with genotype concordance rate=1 and	NA	180399	SNPTEST

Supplementary Table 3. Information on genotyping methods, quality control, imputation, and statistical analysis for studies included in stage 1 and stage 2 and ancillary studies

Study	Genotyping center	Genotype calling algorithm	Genotyping					SNPs that met QC criteria	Analyses software
			Inclusion criteria						
			MAF	Call rate*	P for HWE	Discordance rate	Other		
						IBD=1			
EGCUT controls	Estonian Genome Center of University of Tartu	GeneCall	> 0%	≥ 95%	> 10 ⁻⁶	NA	NA	120720	SNPTEST
EGCUT CAD	Estonian Genome Center of University of Tartu	GeneCall	> 0%	≥ 95%	> 10 ⁻⁶	NA	NA	120720	SNPTEST
EGCUT T2D	Estonian Genome Center of University of Tartu	GeneCall	> 0%	≥ 95%	> 10 ⁻⁶	NA	NA	120720	SNPTEST
ELY	Cambridge Genomic Services, Department of Pathology, University of Cambridge, UK	GeneCall	> 0%	≥ 90%	> 10 ⁻⁶	< 3%	NA	149302	PLINK
EPIC Controls & cases	Cambridge Genomic Services, Department of Pathology, University of Cambridge, UK	GeneCall	> 0%	≥ 90%	> 10 ⁻⁶	< 3%	NA	143294	PLINK
FENLAND	Cambridge Genomic Services, Department of Pathology, University of Cambridge, UK	GeneCall	> 0%	≥ 90%	> 10 ⁻⁶	< 3%	NA	167085	PLINK
GLACIER	Wellcome Trust Sanger Institute, Wellcome Trust Genome Campus, CB10 1SA, Hinxton, UK	Illuminus	> 0%	≥ 95%	> 0.00001	< 3%	none	149782	PLINK
HNR	Institute of Human Genetics, University of Bonn, Bonn, Germany. Department of Genomics, Life & Brain Center, University of Bonn, Bonn, Germany	Genetrain 2	none	≥ 95%	> 10 ⁻⁶	< 3%	NA	192261	PLINK
HUNT.TROMSO controls	Johns Hopkins University Genetic Resources Core Facility SNP Center at the Center for Inherited Disease Research, Baltimore MD	BeadStudio version 3.3.7, Genetrain version 1.0	> 0%	≥ 95%	none	none	Cluster Separation score less than 0.2 or which had more than 1 Replicate error as defined with the HapMap control samples. Additional hand editing was done for X, Y and	143846	PLINK

Supplementary Table 3. Information on genotyping methods, quality control, imputation, and statistical analysis for studies included in stage 1 and stage 2 and ancillary studies

Study	Genotyping								Analyses software
	Genotyping center	Genotype calling algorithm	Inclusion criteria					SNPs that met QC criteria	
			MAF	Call rate*	P for HWE	Discordance rate	Other		
HUNT:TROMSO cases	Johns Hopkins University Genetic Resources Core Facility SNP Center at the Center for Inherited Disease Research, Baltimore MD	BeadStudio version 3.3.7, GenTrain version 1.0	> 0%	≥ 95%	none	none	Mitochondrial loci.	138863	PLINK
IMPROVE	Wallenberg Genotyping Platform, Molecular Medicine, Department of Medical Sciences, Uppsala University, Uppsala, Sweden	GeneCall	> 0%	≥ 95%	$> 10^{-6}$	-	-	185704	PLINK
KORA S3 METABO	Helmholtz Center Munich	Genome Studio	none	none	none	none	none	185781	ProbABEL/PLINK
KORA S4 METABO	Helmholtz Center Munich	Genome Studio	none	none	none	none	none	185781	ProbABEL/PLINK
LURIC	Institute of Human Genetics, Department of Genomics, Life & Brain Center, University of Bonn, Germany	GeneTrain 2	-	≥ 90%	$> 10^{-6}$	-	-	191341	PLINK
MORGAM Controls	The Wellcome Trust Sanger Institute, UK	GenCall	none	none	none	none	none	196725	PLINK
MoRGAM Cases	The Wellcome Trust Sanger Institute, UK	GenCall	none	none	none	none	none	196725	PLINK
NSHD	Cambridge Genomic Services, Department of Pathology, University of Cambridge, UK	GeneCall	> 0%	≥ 90%	$> 10^{-6}$	< 3%	NA	146474	PLINK
PIVUS	Uppsala SNP&SEQ Technology Platform	GenCall	none	≥ 90%	$> 10^{-6}$	NA	NA	185450	PLINK

Supplementary Table 3. Information on genotyping methods, quality control, imputation, and statistical analysis for studies included in stage 1 and stage 2 and ancillary studies

Study	Genotyping								Analyses software
	Genotyping center	Genotype calling algorithm	Inclusion criteria					SNPs that met QC criteria	
			MAF	Call rate*	P for HWE	Discordance rate	Other		
ULSAM	Uppsala SNP&SEQ Technology Platform	GenCall	none	≥ 90%	> 10 ⁻⁶	NA	NA	183357	PLINK
STR	Uppsala SNP&SEQ Technology Platform	GenCall	none	≥ 90%	> 10 ⁻⁶	NA	NA	183961	R GWAF
THISEAS controls	Wellcome Trust Sanger Institute	GenoSNP	none	≥ 98%	> 10 ⁻⁴	< 10%	na	169779	PLINK
THISEAS cases	Wellcome Trust Sanger Institute	GenoSNP	none	≥ 98%	> 10 ⁻⁴	< 10%	na	169779	PLINK
WHITEHALL	Cambridge Genomic Services, Department of Pathology, University of Cambridge, UK; and UCL Genomics, Molecular Haematology and Cancer Biology Unit, London, UK	GeneCall	> 0%	≥ 90%	> 10 ⁻⁶	NA	NA	171257	PLINK
WTCCC T2D nonGWAS	Cambridge Genomic Services, Department of Pathology, University of Cambridge, UK	GeneCall	≥ 1%	≥ 95%	> 10 ⁻⁶	1 duplicate pair with genotype concordance rate=1 and IBD=1	NA	178205	SNPTEST

Supplementary Table 3. Information on genotyping methods, quality control, imputation, and statistical analysis for studies included in stage 1 and stage 2 and ancillary studies

Study	Platform	Genotyping					SNPs that met QC criteria	Imputation software	Imputation		Association analyses	
		Genotype calling algorithm	Inclusion criteria			MAF			MAF	Inclusion criteria		SNPs in meta-analysis
MAF	Call rate		P for HWE	MAF	Imputation quality							
Ancillary												
Essen Obesity Study (Essen Case-Control GWAS)	Affymetrix Genome-Wide Human SNP Array 6.0	Birdseed	≥ 1%	≥ 95%	> 0.001 in controls	727,239	MACH	≥ 1%	$r2_hat \geq 0.30$	only in replication	SNPT	
Essen Obesity Study (Essen Obesity Trio GWAS)	Affymetrix Genome-Wide Human SNP Array 6.0	Birdseed	≥ 1%	≥ 95%	> 0.001 in parents	752,216	MACH	≥ 1%	$r2_hat \geq 0.30$	only in replication	PLIN	
French Extreme Obesity Study	Illumina Human CNV370-Duo array & Illumina HAP300 array	GenCall software	≥ 1%	≥ 95%	> 0.01	308,846	MACH 1.0	≥ 1%	proper-info > 0.30	~ 2,3 millions	PLIN	
GEO-IT	Illumina Infinium 660W-Quad	GenomeStudio	≥ 1%	≥ 98%	$> 10^{-6}$	519,612	MACH	≥ 1%	$Rsq \geq 0.30$	249,678	PLIN	
GOYA	Illumina 610k Quad	GenomeStudio	≥ 1%	≥ 95%	$> 10^{-7}$	545,349	MACH 1.0	≥ 1%	$r2 \geq 0.3$	2,449,993	MACH DA1	

Supplementary Table 4. Study-specific descriptive statistics for studies included in stage 2 and ancillary analyses

Study	Trait	Men								Women							
		N	Mean	SD	Median	Min	Max	Correlation with BMI	Correlation with height	N	Mean	SD	Median	Min	Max	Correlation with BMI	Correlation with height
Stage 2 - In silico:																	
B58C-REPL	Age (yrs)	1230	45.19	0.36	45.17	44.25	46.00	-0.01	-0.01	1192	45.20	0.38	45.17	44.25	46.00	0.03	0.00
	BMI (kg/m ²)	1230	27.76	4.40	27.26	16.53	56.65	1.00	-0.02	1192	26.85	5.52	25.54	16.29	54.25	1.00	-0.12
	Weight (kg)	1230	86.29	14.98	84.60	47.00	172.50	0.89	0.41	1192	70.85	14.82	67.55	41.60	143.50	0.94	0.24
	Height (m)	1230	1.76	0.06	1.76	1.56	2.01	-0.02	1.00	1192	1.63	0.06	1.63	1.22	1.88	-0.12	1.00
	WC (cm)	1170	98.40	11.41	97.20	70.00	160.20	0.87	0.19	1158	85.04	12.79	82.80	57.20	133.80	0.89	0.06
	Hip (cm)	1170	105.77	7.73	105.10	86.00	168.20	0.82	0.10	1158	104.94	11.28	102.80	81.20	157.60	0.92	0.09
	WHR (cm/cm)	1170	0.93	0.06	0.93	0.76	1.21	0.58	0.00	1158	0.81	0.06	0.81	0.62	1.04	0.43	0.00
BHS	Age (yrs)	512	54.30	16.80	54.30	17.60	91.40	0.13	-0.37	694	54.50	17.00	54.10	17.30	90.50	0.10	-0.44
	BMI (kg/m ²)	512	26.58	3.56	26.20	15.77	40.02	1.00	-0.07	693	25.52	4.47	24.65	16.82	40.77	1.00	-0.15
	Weight (kg)	512	81.65	12.40	80.20	46.40	127.00	0.84	0.48	693	67.00	12.09	64.60	34.80	109.00	0.90	0.28
	Height (m)	512	1.75	0.07	1.75	1.53	1.99	-0.07	1.00	694	1.62	0.06	1.62	1.35	1.81	-0.16	1.00
	WC (cm)	494	93.78	10.38	93.00	62.80	130.00	0.86	0.07	672	80.99	11.71	78.70	58.65	132.00	0.89	-0.06
	Hip (cm)	494	100.10	7.51	99.35	74.80	132.80	0.91	0.26	672	101.10	9.26	99.95	77.00	138.80	0.89	0.10
	WHR (cm/cm)	494	0.94	0.06	0.93	0.75	1.17	0.51	-0.17	672	0.80	0.07	0.79	0.64	1.05	0.51	-0.21
HYPERGENES Cases	Age (yrs)	1080	49.81	10.32	50.00	17.78	84.00	0.04	-0.32	542	48.22	9.37	48.44	18.38	93.00	0.11	-0.20
	BMI (kg/m ²)	1072	27.45	3.50	27.16	16.00	47.43	1.00	-0.07	538	26.89	4.99	26.21	17.45	52.35	1.00	-0.09
	Weight (kg)	1073	81.38	12.05	80.00	49.00	139.50	0.82	0.51	540	68.61	13.75	67.00	44.00	164.00	0.89	0.36
	Height (m)	1072	1.72	0.07	1.72	1.49	1.96	-0.07	1.00	538	1.60	0.07	1.60	1.40	1.80	-0.09	1.00
	WC (cm)	170	98.37	12.04	98.00	71.00	139.00	0.89	0.15	152	89.30	13.71	88.00	61.00	133.00	0.89	0.05
	Hip (cm)	170	104.51	8.82	104.00	86.00	150.00	0.84	0.21	152	107.26	11.51	104.40	89.00	158.00	0.91	0.12
	WHR (cm/cm)	170	0.94	0.07	0.95	0.79	1.15	0.53	0.01	152	0.83	0.07	0.83	0.64	1.01	0.47	-0.05
HYPERGENES Controls	Age (yrs)	1031	62.21	10.69	59.67	28.00	98.00	-0.10	-0.12	709	63.65	10.82	60.58	44.93	113.00	-0.13	-0.12
	BMI (kg/m ²)	998	25.93	3.26	25.56	10.15	40.77	1.00	-0.13	684	24.98	3.75	24.52	16.53	41.35	1.00	-0.17
	Weight (kg)	1025	76.34	11.62	75.00	29.00	186.00	0.81	0.47	701	64.32	10.16	63.00	42.00	110.00	0.87	0.34
	Height (m)	998	1.71	0.07	1.70	1.50	1.92	-0.13	1.00	684	1.61	0.06	1.60	1.40	1.81	-0.17	1.00
	WC (cm)	130	95.21	9.85	93.88	72.50	120.30	0.85	0.27	159	84.96	10.69	83.50	62.80	119.80	0.81	0.21
	Hip (cm)	130	102.56	6.30	102.75	87.00	118.50	0.82	0.32	159	103.19	8.30	102.00	84.00	134.50	0.84	0.23

Supplementary Table 4. Study-specific descriptive statistics for studies included in stage 2 and ancillary analyses

Study	Trait	Men								Women							
		N	Mean	SD	Median	Min	Max	Correlation with BMI	Correlation with height	N	Mean	SD	Median	Min	Max	Correlation with BMI	Correlation with height
	WHR (cm/cm)	130	0.93	0.06	0.93	0.80	1.07	0.60	0.13	159	0.82	0.07	0.82	0.68	1.19	0.40	0.10
LifeLines	Age (yrs)	3477	48.1	11.3	47.8	19.6	87.6	0.16	-0.25	4639	44.0	11.1	47.2	18.1	89.0	0.21	-0.28
	BMI (kg/m ²)	3477	26.6	3.6	26.2	14.4	48.9	1.00	-0.09	4639	25.7	4.7	25.1	13.9	51.8	1.00	-0.17
	Weight (kg)	3477	88.3	13.2	87.0	49.5	165.0	0.83	0.43	4639	73.9	13.6	72.0	42.5	152.0	0.88	0.27
	Height (m)	3477	1.82	0.07	1.82	1.58	2.08	-0.09	1.00	4639	1.70	0.06	1.69	1.45	1.93	-0.17	1.00
	WC (cm)	3477	96.4	10.7	96.0	65.0	158.0	0.84	0.08	4638	86.8	12.2	86.5	58.0	146.0	0.83	-0.02
	Hip (cm)	3477	100.2	7.4	100.0	73.0	163.0	0.70	0.30	4637	100.7	10.3	100.0	69.0	181.0	0.84	0.08
	WHR (cm/cm)	3477	1.0	0.1	1.0	0.6	1.3	0.58	-0.15	4637	0.9	0.1	0.9	0.5	1.3	0.39	-0.11
PLCO2 controls	Age (yrs)	649	63.60	5.20	64.00	55.00	74.00	-0.16	-0.08	544	63.60	5.20	64.00	55.00	74.00	-0.06	-0.07
	BMI (kg/m ²)	645	27.00	3.90	26.50	17.20	40.50	1.00	0.00	544	26.40	5.00	25.70	16.70	50.30	1.00	-0.08
	Weight (kg)	645	86.60	14.00	83.90	52.20	145.10	0.89	0.45	544	70.30	14.20	68.00	41.30	149.70	0.92	0.31
	Height (m)	649	1.78	0.07	1.78	1.63	1.98	0.00	1.00	544	1.63	0.06	1.63	1.45	1.85	-0.08	1.00
	WC (cm)	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
	Hip (cm)	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
	WHR (cm/cm)	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
PLCO2 cases	Age (yrs)	2042	64.20	5.10	64.00	55.00	74.00	-0.08	-0.08	934	64.00	5.30	64.00	55.00	74.00	-0.02	-0.11
	BMI (kg/m ²)	2040	27.50	4.20	26.90	15.90	50.20	1.00	-0.02	933	26.70	5.10	25.90	16.50	52.20	1.00	-0.07
	Weight (kg)	2040	87.60	14.80	85.30	52.20	176.90	0.90	0.42	933	71.00	14.50	68.00	40.80	137.90	0.91	0.33
	Height (m)	2042	1.79	0.07	1.78	1.55	2.11	-0.02	1.00	934	1.63	0.06	1.63	1.24	1.93	-0.07	1.00
	WC (cm)	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
	Hip (cm)	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
	WHR (cm/cm)	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
PREVEND	Age (yrs)	1870	50.93	12.77	50.00	28.00	75.00	0.20	-0.34	1752	48.19	12.03	47.00	28.00	75.00	0.29	-0.37
	BMI (kg/m ²)	1870	26.41	3.68	26.02	17.21	44.66	1.00	-0.18	1752	25.82	4.77	24.99	17.06	53.73	1.00	-0.22
	Weight (kg)	1870	85.11	12.54	84.00	49.00	146.00	0.85	0.36	1752	72.31	13.18	70.00	44.90	144.50	0.91	0.20
	Height (m)	1870	1.80	0.07	1.79	1.58	2.07	-0.18	1.00	1752	1.68	0.07	1.68	1.45	1.93	-0.22	1.00
	WC (cm)	1868	94.50	11.17	94.00	31.00	141.50	0.84	-0.02	1752	83.33	12.99	81.50	57.00	150.00	0.86	-0.08
	Hip (cm)	1868	100.11	6.70	100.00	70.00	128.00	0.78	0.23	1752	100.95	10.35	100.00	68.00	170.00	0.86	0.02

Supplementary Table 4. Study-specific descriptive statistics for studies included in stage 2 and ancillary analyses

Study	Trait	Men								Women							
		N	Mean	SD	Median	Min	Max	Correlation with BMI	Correlation with height	N	Mean	SD	Median	Min	Max	Correlation with BMI	Correlation with height
	WHR (cm/cm)	1868	0.94	0.07	0.94	0.27	1.36	0.59	-0.23	1752	0.82	0.08	0.82	0.45	1.39	0.45	-0.15
QIMR	Age (yrs)	1646	43.23	15.78	42.00	18.00	91.00	0.28	-0.17	2307	43.03	15.14	41.00	18.00	90.00	0.29	-0.18
	BMI (kg/m ²)	1646	25.75	3.81	25.38	16.60	60.93	1.00	-0.15	2307	24.86	5.04	23.73	14.17	64.52	1.00	-0.16
	Weight (kg)	1646	81.40	12.71	80.00	50.00	161.00	0.86	0.37	2307	66.28	13.63	63.50	34.02	160.00	0.91	0.25
	Height (m)	1646	1.78	0.07	1.78	1.45	2.08	-0.15	1.00	2307	1.63	0.07	1.63	1.32	1.96	-0.16	1.00
	WC (cm)	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
	Hip (cm)	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
	WHR (cm/cm)	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
RS2	Age (yrs)	876	64.38	7.59	61.81	55.14	93.95	-0.13	-0.21	1035	64.71	8.23	61.73	55.12	95.33	-0.02	-0.30
	BMI (kg/m ²)	876	26.83	3.29	26.70	16.78	40.52	1.00	-0.10	1035	27.48	4.47	26.78	16.66	50.12	1.00	-0.06
	Weight (kg)	876	85.05	11.36	82.00	54.00	126.80	0.84	0.45	1035	72.77	12.80	71.00	44.10	150.00	0.90	0.38
	Height (m)	876	1.76	0.06	1.76	1.57	2.03	-0.10	1.00	1035	1.63	0.06	1.63	1.42	1.90	-0.06	1.00
	WC (cm)	876	99.00	10.18	98.60	70.00	198.60	0.82	0.09	1035	89.84	11.56	88.90	62.00	145.00	0.86	0.10
	Hip (cm)	876	101.80	5.87	101.30	86.50	163.20	0.75	0.25	1035	104.08	8.61	103.00	82.00	154.00	0.86	0.20
	WHR (cm/cm)	876	0.97	0.07	0.97	0.58	1.95	0.54	-0.08	1035	0.86	0.08	0.86	0.60	1.18	0.45	-0.05
RS3	Age (yrs)	877	55.93	5.43	56.10	45.46	84.15	0.08	-0.26	1129	56.20	6.03	56.42	45.75	97.22	0.07	-0.23
	BMI (kg/m ²)	877	28.03	4.07	27.31	18.42	46.68	1.00	-0.07	1129	27.48	5.06	26.55	14.02	56.87	1.00	-0.10
	Weight (kg)	877	89.76	14.33	87.70	58.30	153.50	0.88	0.41	1129	74.90	14.28	72.80	35.00	158.60	0.92	0.29
	Height (m)	877	1.79	0.07	1.79	1.61	1.98	-0.07	1.00	1129	1.65	0.06	1.65	1.47	1.85	-0.10	1.00
	WC (cm)	842	99.98	11.71	98.50	65.20	174.00	0.85	0.14	1085	89.59	12.87	88.10	62.10	160.00	0.88	0.05
	Hip (cm)	842	107.50	7.33	107.00	86.00	144.50	0.76	0.31	1085	107.80	10.24	106.30	79.80	161.10	0.89	0.13
	WHR (cm/cm)	842	0.93	0.08	0.92	0.66	2.02	0.54	-0.05	1085	0.83	0.07	0.83	0.66	1.54	0.45	-0.06
TRAILS	Age (yrs)	539	19.2	0.5	19.2	18.2	20.7	0.04	-0.09	602	19.1	0.6	19.1	18.2	20.8	0.09	-0.07
	BMI (kg/m ²)	539	22.6	3.8	21.9	15.6	43.2	1.00	-0.04	602	23.0	3.7	22.4	15.9	45.6	1.00	-0.15
	Weight (kg)	539	76.3	14.0	73.5	48.4	138.0	0.84	0.46	602	66.5	10.9	65.5	42.5	125.0	0.84	0.35
	Height (m)	539	1.84	0.07	1.84	1.65	2.06	-0.04	1.00	602	1.70	0.07	1.71	1.42	1.92	-0.15	1.00
	WC (cm)	537	81.8	9.8	79.5	64.5	130.8	0.77	0.23	602	75.3	8.8	73.6	46.0	130.0	0.78	0.09
	Hip (cm)	537	99.4	8.4	98.0	75.5	138.5	0.74	0.33	602	99.9	8.5	99.3	51.0	133.8	0.75	0.18

Supplementary Table 4. Study-specific descriptive statistics for studies included in stage 2 and ancillary analyses

Study	Trait	Men								Women							
		N	Mean	SD	Median	Min	Max	Correlation with BMI	Correlation with height	N	Mean	SD	Median	Min	Max	Correlation with BMI	Correlation with height
	WHR (cm/cm)	537	0.8	0.1	0.8	0.7	1.1	0.35	-0.05	602	0.8	0.1	0.7	0.6	1.0	0.29	-0.10
TWINGENE	Age (yrs)	4594	65.51	8.06	65.20	47.57	93.30	-0.05	-0.29	5146	64.64	8.21	64.11	47.38	93.87	0.04	-0.32
	BMI (kg/m ²)	4410	26.33	3.53	25.93	15.43	57.82	1.00	-0.07	4886	25.80	4.32	25.13	15.02	66.99	1.00	-0.12
	Weight (kg)	4410	81.90	12.20	80.50	50.00	179.10	0.86	0.45	4886	68.78	12.02	67.10	37.60	171.50	0.90	0.32
	Height (m)	4494	1.76	0.07	1.76	1.43	2.05	-0.07	1.00	5007	1.63	0.06	1.63	1.37	1.93	-0.12	1.00
	WC (cm)	4536	97.24	10.21	97.00	52.50	193.00	0.82	0.14	5068	86.92	11.59	86.00	40.00	199.00	0.83	0.06
	Hip (cm)	4523	103.22	7.97	103.00	41.00	197.00	0.64	0.25	5063	103.42	9.40	102.30	38.00	172.00	0.83	0.13
	WHR (cm/cm)	4517	0.94	0.09	0.94	0.49	2.61	0.36	-0.05	5056	0.84	0.08	0.84	0.45	1.88	0.37	-0.04
UKBS2	Age (yrs)	684	45.09	11.92	46	17	69	0.05	-0.14	648	42.5	12.3	43.0	17.0	69.0	0.09	-0.17
	BMI (kg/m ²)	684	26.45	3.63	26.2	18.01	44.44	1	-0.07	648	26.0	4.5	25.1	16.6	42.9	1	-0.17
	Weight (kg)	684	83.99	12.81	82.73	48.18	152.7	0.86	0.43	648	70.4	12.6	68.2	50.0	117.7	0.9	0.27
	Height (m)	684	1.78	0.07	1.78	1.52	2.08	-0.08	1	648	1.6	0.064	1.7	1.4	1.8	-0.17	1
	WC (cm)	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
	Hip (cm)	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
	WHR (cm/cm)	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-

Supplementary Table 4. Study-specific descriptive statistics for studies included in stage 2 and ancillary analyses

Study	Trait	Men								Women							
		N	Mean	SD	Median	Min	Max	Correlation with BMI	Correlation with height	N	Mean	SD	Median	Min	Max	Correlation with BMI	Correlation with height
Stage 2 - Metabochip:																	
AMC-PAS	Age (yrs)	368	43.50	5.14	44.50	24.00	50.00	-0.05	-0.06	122	42.78	5.62	44.00	25.00	50.00	0.09	-0.09
	BMI (kg/m ²)	368	27.13	3.81	26.70	18.50	40.60	1.00	0.08	122	26.19	5.09	25.30	17.40	41.20	1.00	-0.09
	Weight (kg)	368	88.09	14.84	86.00	50.00	150.00	0.88	0.54	122	73.08	14.85	70.00	49.00	122.00	0.93	0.29
	Height (m)	368	1.80	0.07	1.80	1.59	2.04	0.08	1.00	122	1.67	0.06	1.67	1.50	1.83	-0.09	1.00
	WC (cm)	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
	Hip (cm)	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
	WHR (cm/cm)	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
CARDIOGENICS controls	Age (yrs)	148	53.90	7.10	55.00	42.00	65.00	-0.01	-0.12	227	53.20	7.00	54.00	41.00	65.00	0.11	-0.07
	BMI (kg/m ²)	148	25.90	3.40	25.30	19.00	37.20	1.00	-0.24	227	25.60	3.90	25.10	18.10	39.20	1.00	-0.07
	Weight (kg)	148	82.20	10.60	81.00	57.00	114.00	0.87	0.27	227	69.30	11.50	68.00	53.00	127.00	0.89	0.37
	Height (m)	148	1.78	0.06	1.78	1.63	1.98	-0.24	1.00	227	1.64	0.06	1.65	1.50	1.80	-0.07	1.00
	WC (cm)	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
	Hip (cm)	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
	WHR (cm/cm)	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
CARDIOGENICS cases	Age (yrs)	328	56.50	8.70	57.50	26.00	87.00	-0.10	-0.19	51	58.70	8.70	59.00	39.00	79.00	-0.23	-0.16
	BMI (kg/m ²)	328	28.50	4.20	27.80	17.10	42.20	1.00	0.02	51	27.60	5.90	26.70	17.70	43.80	1.00	0.20
	Weight (kg)	328	88.27	14.63	86.40	36.00	133.00	0.87	0.50	51	73.59	17.89	70.00	43.00	122.00	0.94	0.51
	Height (m)	328	1.80	0.10	1.80	1.50	2.00	0.02	1.00	51	1.60	0.10	1.60	1.50	1.80	0.20	1.00
	WC (cm)	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
	Hip (cm)	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
	WHR (cm/cm)	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
D2D2007.DPS.DRSEXTRA.FUSION2.MET SIM controls	Age (yrs)	3394	58.00	8.08	58.00	41.30	78.46	0.05	-0.22	2714	59.59	8.44	60.02	40.09	78.70	0.07	-0.28
	BMI (kg/m ²)	3394	26.77	3.60	26.43	16.48	48.95	1.00	-0.01	2714	27.37	5.01	26.67	15.96	61.34	1.00	-0.13
	Weight (kg)	3394	82.62	12.75	81.40	45.00	160.00	0.85	0.48	2714	71.55	13.60	69.70	41.90	158.60	0.90	0.27

Supplementary Table 4. Study-specific descriptive statistics for studies included in stage 2 and ancillary analyses

Study	Trait	Men								Women							
		N	Mean	SD	Median	Min	Max	Correlation with BMI	Correlation with height	N	Mean	SD	Median	Min	Max	Correlation with BMI	Correlation with height
	Height (m)	339 4	1.76	0.06	1.75	1.45	1.98	-0.01	1.00	271 4	1.62	0.06	1.62	1.40	1.83	-0.13	1.00
	WC (cm)	339 1	96.75	10.3 7	96.00	63.0 0	155.0 0	0.86	0.19	270 6	88.54	12.4 9	87.00	58.0 0	150.0 0	0.88	0.05
	Hip (cm)	339 1	100.0 9	6.80	99.75	72.0 0	160.0 0	0.77	0.34	270 8	102.8 8	9.85	101.50	72.0 0	161.0 0	0.89	0.11
	WHR (cm/cm)	339 1	0.97	0.06	0.96	0.78	1.89	0.61	-0.02	270 5	0.86	0.07	0.86	0.67	1.29	0.53	-0.03
D2D2007.DPS.DRSEXTRA.FUSION2.MET SIM cases	Age (yrs)	203 3	60.50	7.74	61.00	28.0 0	82.00	-0.13	-0.19	721	61.72	7.95	62.00	34.3 6	89.00	-0.12	-0.19
	BMI (kg/m ²)	203 3	30.13	5.05	29.41	18.2 9	55.54	1.00	0.01	721	31.83	5.81	31.20	17.8 5	53.45	1.00	0.00
	Weight (kg)	203 3	92.25	16.8 4	90.00	42.2 0	174.0 0	0.91	0.40	721	82.36	16.0 6	80.00	48.0 0	137.4 0	0.93	0.34
	Height (m)	203 3	1.75	0.06	1.75	1.39	1.96	0.01	1.00	721	1.61	0.06	1.61	1.45	1.77	0.00	1.00
	WC (cm)	203 2	106.7 4	13.1 1	105.00	73.0 0	167.0 0	0.90	0.17	719	100.5 3	14.0 4	99.50	62.0 0	145.0 0	0.87	0.15
	Hip (cm)	203 3	105.2 4	9.13	104.00	74.0 0	164.0 0	0.84	0.25	719	110.8 1	11.6 0	109.50	84.0 0	152.0 0	0.90	0.15
	WHR (cm/cm)	203 2	1.01	0.07	1.01	0.72	1.44	0.59	0.02	719	0.91	0.07	0.90	0.61	1.18	0.37	0.04
DILGOM	Age (yrs)	179 7	53.30	13.5 0	55.00	25.0 0	74.00	0.15	-0.32	213 9	51.70	13.6 0	53.00	25.0 0	74.00	0.24	-0.38
	BMI (kg/m ²)	179 1	27.20	4.20	26.70	15.8 0	63.10	1.00	-0.06	213 9	26.90	5.40	25.80	16.0 0	52.50	1.00	-0.18
	Weight (kg)	179 1	84.00	14.1 0	82.20	50.1 0	192.6 0	0.89	0.41	213 9	70.80	14.2 0	68.50	38.5 0	144.5 0	0.92	0.20
	Height (m)	179 7	1.76	0.07	1.75	1.52	2.01	-0.06	1.00	213 8	1.63	0.06	1.63	1.38	1.84	-0.18	1.00
	WC (cm)	178 7	96.80	12.0 0	95.50	64.0 0	172.0 0	0.91	0.07	212 2	87.00	13.5 0	85.00	58.0 0	143.0 0	0.92	-0.03
	Hip (cm)	179 0	100.5 0	7.60	100.00	80.5 0	161.0 0	0.85	0.25	212 2	102.0 0	10.8 0	100.50	75.5 0	162.0 0	0.92	0.02
	WHR (cm/cm)	178 4	1.00	0.10	1.00	0.80	1.20	0.65	-0.15	211 6	0.90	0.10	0.80	0.70	1.10	0.56	-0.09
GoDARTS controls	Age (yrs)	191 8	60.20	11.0 9	61.00	36.0 0	79.00	0.01	-0.26	178 9	57.99	11.4 5	57.00	36.0 0	79.00	0.01	-0.24
	BMI (kg/m ²)	191 8	27.26	3.97	26.90	16.2 0	54.90	1.00	-0.08	178 8	26.81	5.02	26.00	14.3 0	51.60	1.00	-0.15
	Weight (kg)	191 8	84.63	13.3 4	83.80	46.9 0	168.0 0	0.88	0.40	178 8	70.46	13.5 0	68.55	33.0 0	125.6 0	0.91	0.27
	Height (m)	191 8	1.76	0.07	1.76	1.51	1.98	-0.08	1.00	178 9	1.62	0.07	1.62	1.43	1.85	-0.15	1.00
	WC (cm)	191 7	98.49	10.7 3	98.00	69.0 0	172.0 0	0.86	0.11	178 7	86.69	12.3 0	86.00	61.0 0	153.0 0	0.84	0.02

Supplementary Table 4. Study-specific descriptive statistics for studies included in stage 2 and ancillary analyses

Study	Trait	Men								Women							
		N	Mean	SD	Median	Min	Max	Correlation with BMI	Correlation with height	N	Mean	SD	Median	Min	Max	Correlation with BMI	Correlation with height
GoDARTS cases	Hip (cm)	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
	WHR (cm/cm)	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
	Age (yrs)	1929	63.19	9.49	64.00	36.00	88.00	-0.26	-0.18	1351	64.02	9.76	66.00	36.00	89.00	-0.33	-0.18
	BMI (kg/m ²)	1924	31.12	5.64	30.30	16.20	61.50	1.00	-0.06	1346	32.93	7.05	32.00	16.40	62.30	1.00	0.00
	Weight (kg)	1925	94.31	18.16	91.70	46.90	177.80	0.91	0.35	1347	84.58	19.33	82.58	42.20	164.00	0.94	0.35
	Height (m)	1928	1.74	0.07	1.74	1.50	2.00	-0.06	1.00	1348	1.60	0.06	1.60	1.40	1.86	0.00	1.00
	WC (cm)	1750	108.51	13.36	107.00	74.00	164.00	0.89	0.13	1215	103.67	15.16	103.00	62.00	177.00	0.84	0.16
Hip (cm)	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	
WHR (cm/cm)	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	
EGCUT controls	Age (yrs)	341	54.4	11.0	55.0	35.0	86.0	0.21	-0.30	601	50.0	10.1	49.0	34.0	93.0	0.23	-0.33
	BMI (kg/m ²)	340	22.8	2.5	22.7	16.7	29.4	1.00	-0.12	600	22.1	2.5	22.1	14.6	35.5	1.00	-0.21
	Weight (kg)	340	70.9	8.5	70.0	48.0	95.0	0.81	0.48	600	59.5	7.6	60.0	41.0	99.0	0.76	0.46
	Height (m)	340	1.76	0.07	1.76	1.49	1.97	-0.12	1.00	600	1.64	0.06	1.64	1.41	1.82	-0.21	1.00
	WC (cm)	340	84.1	6.8	85.0	54.0	110.0	0.66	0.11	600	72.8	5.8	73.0	52.0	105.0	0.45	0.21
	Hip (cm)	340	94.9	7.7	96.0	57.0	115.0	0.55	0.26	600	95.1	7.2	95.0	64.0	122.0	0.37	0.19
	WHR (cm/cm)	340	0.9	0.1	0.9	0.7	1.2	0.15	-0.14	600	0.8	0.1	0.8	0.6	1.2	0.08	0.00
EGCUT CAD	Age (yrs)	346	62.5	11.6	62.0	22.0	87.0	0.04	-0.24	354	67.1	9.2	69.0	30.0	86.0	-0.36	-0.26
	BMI (kg/m ²)	346	28.2	4.4	28.0	17.2	43.9	1.00	-0.07	354	29.3	5.5	28.6	17.5	51.0	1.00	0.06
	Weight (kg)	346	86.8	15.6	85.0	51.0	137.0	0.92	0.32	354	76.0	15.0	74.0	42.0	129.0	0.91	0.47
	Height (m)	346	1.75	0.07	1.76	1.51	1.98	-0.07	1.00	354	1.61	0.06	1.61	1.35	1.78	0.06	1.00
	WC (cm)	346	100.5	11.6	100.0	70.0	135.0	0.73	0.10	354	93.1	13.2	92.0	62.0	125.0	0.79	0.18
	Hip (cm)	346	103.1	8.7	102.0	80.0	132.0	0.75	0.17	354	107.0	12.0	105.0	70.0	160.0	0.69	0.27
	WHR (cm/cm)	346	1.0	0.1	1.0	0.7	1.4	0.24	-0.06	354	0.9	0.1	0.9	0.7	1.1	0.44	-0.04
EGCUT T2D	Age (yrs)	355	63.5	10.2	64.0	36.0	94.0	-0.29	-0.25	613	64.5	10.5	64.0	32.0	93.0	-0.24	-0.27
	BMI (kg/m ²)	354	32.6	4.7	31.9	20.5	51.4	1.00	-0.01	613	33.7	5.7	32.8	21.2	56.9	1.00	-0.04
	Weight (kg)	354	100.8	16.1	99.0	62.0	160.0	0.90	0.41	613	88.3	16.3	86.0	47.0	160.0	0.88	0.43

Supplementary Table 4. Study-specific descriptive statistics for studies included in stage 2 and ancillary analyses

Study	Trait	Men								Women							
		N	Mean	SD	Median	Min	Max	Correlation with BMI	Correlation with height	N	Mean	SD	Median	Min	Max	Correlation with BMI	Correlation with height
ELY	Height (m)	355	1.76	0.07	1.75	1.58	2.00	-0.01	1.00	613	1.62	0.06	1.62	1.43	1.84	-0.04	1.00
	WC (cm)	272	111.7	11.2	110.5	82.0	145.0	0.78	0.25	417	104.6	14.2	103.0	62.0	150.0	0.79	0.06
	Hip (cm)	272	110.0	11.2	110.0	70.0	155.0	0.76	0.27	417	116.0	14.0	115.0	63.0	165.0	0.64	0.13
	WHR (cm/cm)	272	1.0	0.1	1.0	0.7	1.6	0.19	0.02	417	0.9	0.1	0.9	0.7	1.1	0.16	-0.08
	Age (yrs)	744	61.49	9.13	61.51	35.67	77.44	-0.04	-0.26	856	60.82	9.25	60.17	36.34	78.88	0.08	-0.30
	BMI (kg/m ²)	744	27.36	3.99	26.81	15.98	45.82	1.00	-0.05	855	27.29	5.37	26.33	16.89	59.28	1.00	-0.13
	Weight (kg)	744	83.07	13.31	81.00	48.50	134.50	0.85	0.43	856	71.10	14.33	68.50	38.00	145.00	0.89	0.29
	Height (m)	744	1.74	0.07	1.74	1.55	2.00	-0.05	1.00	855	1.61	0.06	1.61	1.46	1.80	-0.13	1.00
WC (cm)	736	100.00	10.60	99.40	71.25	144.00	0.84	0.14	853	87.55	12.59	85.85	57.25	143.25	0.85	0.03	
Hip (cm)	736	105.33	7.56	104.53	85.15	139.25	0.78	0.26	852	107.13	11.77	105.25	80.10	171.00	0.87	0.11	
WHR (cm/cm)	735	0.95	0.06	0.95	0.75	1.14	0.54	-0.06	852	0.82	0.06	0.81	0.58	1.04	0.34	-0.08	
EPIC Controls	Age (yrs)	411	59.35	9.15	60.00	40.00	79.00	0.07	-0.22	552	58.36	9.61	58.00	39.00	77.00	0.15	-0.26
	BMI (kg/m ²)	410	26.36	3.25	26.07	16.89	40.31	1.00	-0.08	551	25.91	3.97	25.31	16.29	46.04	1.00	-0.08
	Weight (kg)	411	79.77	11.35	78.40	51.00	135.00	0.81	0.48	551	67.20	11.16	65.40	41.20	126.60	0.86	0.40
	Height (m)	410	1.74	0.07	1.74	1.50	1.96	-0.08	1.00	552	1.61	0.06	1.61	1.42	1.79	-0.08	1.00
	WC (cm)	411	94.92	9.74	94.40	71.80	134.00	0.82	0.14	550	81.46	10.34	79.95	56.50	122.10	0.82	0.05
	Hip (cm)	410	102.51	6.30	102.30	83.20	125.30	0.77	0.30	549	102.67	8.60	101.50	81.60	153.80	0.84	0.23
	WHR (cm/cm)	410	0.92	0.06	0.92	0.76	1.16	0.58	-0.05	549	0.79	0.06	0.79	0.65	1.00	0.45	-0.14
EPIC Cases	Age (yrs)	432	61.54	8.12	62.00	41.00	77.00	-0.01	-0.21	295	62.07	8.32	63.00	41.00	77.00	-0.17	-0.22
	BMI (kg/m ²)	432	29.10	3.79	28.56	19.58	49.06	1.00	0.00	291	30.12	5.11	29.79	19.49	45.51	1.00	0.06
	Weight (kg)	432	87.56	12.90	86.20	56.00	160.00	0.83	0.51	292	77.09	14.61	75.00	45.80	124.40	0.90	0.45
	Height (m)	432	1.73	0.06	1.73	1.54	1.93	0.00	1.00	294	1.60	0.06	1.60	1.44	1.78	0.06	1.00
	WC (cm)	432	103.51	10.03	103.00	79.00	145.60	0.81	0.19	295	92.82	12.46	92.00	64.40	141.30	0.85	0.18
	Hip (cm)	432	106.57	7.42	105.75	88.80	141.90	0.77	0.27	295	110.04	11.45	108.60	85.00	151.60	0.90	0.25

Supplementary Table 4. Study-specific descriptive statistics for studies included in stage 2 and ancillary analyses

Study	Trait	Men								Women							
		N	Mean	SD	Median	Min	Max	Correlation with BMI	Correlation with height	N	Mean	SD	Median	Min	Max	Correlation with BMI	Correlation with height
Fenland	WHR (cm/cm)	432	0.97	0.06	0.97	0.81	1.15	0.43	0.02	295	0.84	0.06	0.84	0.69	1.20	0.27	0.01
	Age (yrs)	1488	46.87	7.19	46.74	30.90	59.93	0.12	-0.10	1698	46.90	7.04	47.13	30.48	59.95	0.16	-0.10
	BMI (kg/m ²)	1486	27.04	4.09	26.52	16.38	50.60	1.00	-0.09	1698	26.48	5.48	25.37	16.86	59.85	1.00	-0.09
	Weight (kg)	1486	85.41	14.13	83.40	52.00	177.60	0.86	0.39	1698	71.18	15.47	67.90	41.50	181.00	0.90	0.32
	Height (m)	1486	1.78	0.07	1.78	1.57	1.98	-0.09	1.00	1698	1.64	0.06	1.64	1.43	1.89	-0.09	1.00
	WC (cm)	1485	96.44	11.23	95.25	65.60	148.95	0.89	0.07	1698	85.19	12.78	82.75	59.00	144.30	0.89	0.07
	Hip (cm)	1485	103.15	7.18	102.45	83.35	155.00	0.81	0.28	1698	103.59	10.98	101.73	78.65	180.40	0.88	0.19
WHR (cm/cm)	1484	0.93	0.07	0.93	0.76	1.17	0.68	-0.14	1698	0.82	0.07	0.81	0.50	1.17	0.53	-0.08	
GLACIER	Age (yrs)	2381	50.06	8.34	50.00	30.00	62.00	0.14	-0.18	3666	49.26	8.82	50.00	29.00	64.00	0.20	-0.18
	BMI (kg/m ²)	2381	26.08	3.43	25.75	17.79	59.03	1.00	-0.15	3666	25.54	4.39	24.80	16.02	57.85	1.00	-0.20
	Weight (kg)	2381	81.65	11.37	80.90	51.00	135.00	0.84	0.40	3666	68.55	11.69	67.00	40.00	146.00	0.88	0.27
	Height (m)	2381	1.77	0.06	1.77	1.50	2.07	-0.15	1.00	3666	1.64	0.06	1.64	1.23	1.91	-0.20	1.00
	WC (cm)	874	96.97	10.06	96.00	71.00	156.00	0.81	0.11	1445	86.70	11.68	85.00	59.00	131.00	0.85	0.06
	Hip (cm)	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
	WHR (cm/cm)	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
HNR	Age (yrs)	2261	59.65	7.81	60.00	45.00	76.00	0.07	-0.27	2256	59.62	7.81	60.00	45.00	76.00	0.07	-0.23
	BMI (kg/m ²)	2250	27.98	4.86	27.60	18.10	49.40	1.00	-0.09	2242	27.42	5.92	26.90	16.50	59.20	1.00	-0.16
	Weight (kg)	2252	85.75	14.82	84.50	53.50	150.00	0.86	0.42	2242	72.05	15.18	70.80	43.30	150.00	0.92	0.23
	Height (m)	2256	1.71	0.62	1.75	1.51	2.00	-0.09	1.00	2244	1.56	0.78	1.62	1.43	1.83	-0.16	1.00
	WC (cm)	2256	99.96	12.52	99.30	48.60	172.00	0.87	0.09	2246	88.03	14.41	87.20	46.00	144.70	0.88	0.00
	Hip (cm)	2256	101.97	10.50	101.30	77.70	160.40	0.83	0.20	2245	103.58	13.93	103.00	57.00	160.60	0.88	0.06
	WHR (cm/cm)	2255	0.94	0.61	0.98	0.54	1.24	0.43	-0.11	2245	0.80	0.70	0.85	0.53	1.28	0.37	-0.09

Supplementary Table 4. Study-specific descriptive statistics for studies included in stage 2 and ancillary analyses

Study	Trait	Men								Women							
		N	Mean	SD	Median	Min	Max	Correlation with BMI	Correlation with height	N	Mean	SD	Median	Min	Max	Correlation with BMI	Correlation with height
HUNT.TROMSO controls	Age (yrs)	664	61.40	13.40	63.00	20.00	93.00	0.02	-0.30	626	65.50	13.40	68.00	20.00	92.00	0.16	-0.45
	BMI (kg/m ²)	660	26.20	3.30	26.00	17.80	39.90	1.00	-0.02	620	26.80	4.40	26.20	16.60	44.00	1.00	-0.13
	Weight (kg)	660	80.00	11.80	79.30	51.10	132.20	0.83	0.49	621	69.40	11.90	68.50	39.90	112.00	0.89	0.31
	Height (m)	661	1.70	0.10	1.80	1.50	2.00	-0.02	1.00	621	1.60	0.10	1.60	1.40	1.80	-0.13	1.00
	WC (cm)	510	93.80	9.00	94.00	70.00	121.00	0.78	0.18	484	86.10	10.70	85.00	62.00	117.00	0.83	0.03
	Hip (cm)	510	102.30	5.90	102.00	85.00	123.00	0.72	0.26	483	104.30	9.30	104.00	82.00	134.00	0.87	0.15
	WHR (cm/cm)	510	0.92	0.06	0.91	0.75	1.13	0.52	0.02	483	0.82	0.06	0.82	0.67	1.07	0.41	-0.14
HUNT.TROMSO cases	Age (yrs)	547	62.00	12.20	63.00	27.00	90.00	-0.26	-0.20	531	66.00	13.10	68.00	28.00	91.00	-0.15	-0.30
	BMI (kg/m ²)	546	28.40	4.00	28.20	18.30	46.10	1.00	0.03	524	30.00	5.40	29.70	17.30	48.00	1.00	-0.04
	Weight (kg)	546	87.00	14.10	86.00	52.90	144.40	0.86	0.49	524	77.10	14.80	76.10	38.40	129.10	0.91	0.34
	Height (m)	546	1.70	0.10	1.80	1.50	2.00	0.03	1.00	525	1.60	0.10	1.60	1.40	1.80	-0.04	1.00
	WC (cm)	422	100.10	10.00	100.00	73.00	139.50	0.81	0.16	392	95.50	12.60	95.00	50.00	133.00	0.86	0.02
	Hip (cm)	422	104.90	6.80	105.00	87.00	131.00	0.73	0.31	392	110.30	11.30	108.80	83.00	150.00	0.87	0.06
	WHR (cm/cm)	422	0.95	0.06	0.95	0.74	1.47	0.50	-0.02	392	0.87	0.07	0.86	0.44	1.06	0.34	-0.04
IMPROVE	Age (yrs)	1666	64.03	5.35	64.45	54.49	79.47	-0.09	-0.03	1783	64.38	5.45	64.62	54.47	79.78	-0.02	-0.02
	BMI (kg/m ²)	1665	27.40	3.64	27.08	18.93	47.16	1.00	0.06	1783	27.11	4.77	26.48	15.93	47.46	1.00	0.05
	Weight (kg)	1665	83.48	13.17	82.00	51.40	160.00	0.84	0.55	1783	70.30	14.07	68.40	39.00	131.00	0.90	0.45
	Height (m)	1665	1.74	0.07	1.74	1.52	1.97	0.06	1.00	1783	1.61	0.06	1.60	1.42	1.83	0.05	1.00
	WC (cm)	1664	98.70	10.82	98.00	64.00	150.00	0.82	0.30	1775	89.62	12.47	89.00	54.00	145.00	0.82	0.23
	Hip (cm)	1664	101.70	8.13	101.00	73.00	155.00	0.51	0.00	1775	102.84	10.79	102.00	67.00	160.00	0.34	0.01
	WHR (cm/cm)	1664	0.97	0.07	0.96	0.67	1.45	0.68	0.43	1775	0.87	0.07	0.87	0.58	1.31	0.84	0.32
KORA S3 METABO	Age (yrs)	604	40.90	13.27	37.00	25.00	74.00	0.22	-0.38	674	40.91	12.68	38.00	25.00	74.00	0.31	-0.39
	BMI (kg/m ²)	599	26.54	3.43	26.16	18.03	40.99	1.00	-0.14	659	25.18	4.72	24.09	16.89	40.99	1.00	-0.25

Supplementary Table 4. Study-specific descriptive statistics for studies included in stage 2 and ancillary analyses

Study	Trait	Men								Women							
		N	Mean	SD	Median	Min	Max	Correlation with BMI	Correlation with height	N	Mean	SD	Median	Min	Max	Correlation with BMI	Correlation with height
KORA S4 METABO	Weight (kg)	599	82.39	11.58	81.00	52.30	132.00	0.84	0.42	659	66.73	12.19	64.20	43.60	132.00	0.91	0.18
	Height (m)	603	1.76	0.07	1.77	1.59	2.00	-0.14	1.00	671	1.63	0.06	1.63	1.42	2.00	-0.25	1.00
	WC (cm)	603	92.84	9.32	92.00	70.50	127.00	0.85	-0.01	660	79.20	11.41	76.50	59.00	127.00	0.89	-0.14
	Hip (cm)	603	102.94	6.23	102.50	86.00	125.00	0.78	0.25	660	101.33	9.57	100.00	79.00	125.00	0.90	0.02
	WHR (cm/cm)	603	0.90	0.06	0.90	0.76	1.10	0.58	-0.24	660	0.78	0.06	0.77	0.63	1.10	0.51	-0.29
	Age (yrs)	585	42.70	15.85	36.00	24.00	75.00	0.34	-0.48	637	41.19	14.83	36.00	24.00	75.00	0.31	-0.47
	BMI (kg/m ²)	582	26.64	3.59	26.38	16.32	41.79	1.00	-0.23	624	25.94	5.09	25.34	15.84	41.79	1.00	-0.24
	Weight (kg)	582	82.66	11.65	81.45	52.30	136.50	0.82	0.36	624	68.88	13.38	67.15	41.10	136.50	0.90	0.19
	Height (m)	585	1.76	0.07	1.76	1.56	1.96	-0.23	1.00	632	1.63	0.07	1.63	1.39	1.96	-0.24	1.00
	WC (cm)	585	94.38	9.99	93.50	65.40	134.20	0.89	-0.12	624	82.16	12.00	80.60	61.00	134.20	0.90	-0.13
Hip (cm)	585	104.15	6.43	103.75	87.20	136.00	0.81	0.21	624	103.94	9.99	102.77	81.87	136.00	0.92	0.03	
WHR (cm/cm)	585	0.91	0.06	0.90	0.72	1.07	0.63	-0.36	624	0.79	0.06	0.78	0.62	1.07	0.53	-0.27	
LURIC	Age (yrs)	2007	61.70	10.70	62.60	18.60	88.50	-0.03	-0.27	888	64.80	10.30	65.30	27.00	92.10	-0.01	-0.18
	BMI (kg/m ²)	2007	27.50	3.80	27.00	16.30	57.10	1.00	-0.08	888	27.20	4.70	26.80	16.40	46.10	1.00	-0.07
	Weight (kg)	2007	83.10	12.70	82.00	51.00	185.00	0.86	0.44	888	71.00	13.10	70.00	41.00	125.00	0.91	0.35
	Height (m)	2007	1.74	0.07	1.73	1.52	2.02	-0.08	1.00	888	1.61	0.06	1.61	1.41	1.89	-0.07	1.00
	WC (cm)	1978	100.70	10.70	100.00	61.00	165.00	0.77	0.91	876	94.10	13.00	94.00	58.00	135.00	0.79	0.10
	Hip (cm)	1978	102.50	8.80	102.00	66.00	165.00	0.68	0.17	876	103.80	11.60	103.00	70.00	150.00	0.79	0.16
	WHR (cm/cm)	1978	0.98	0.06	0.98	0.63	1.61	0.34	-0.07	876	0.91	0.08	0.90	0.65	1.22	0.25	-0.04
	Age (yrs)	2138	59.40	8.00	59.80	25.10	76.40	0.05	-0.14	335	57.30	9.00	58.80	27.30	73.70	0.22	-0.25
BMI (kg/m ²)	2095	26.90	4.00	26.50	16.70	51.90	1.00	-0.02	319	27.60	5.00	26.80	15.40	51.10	1.00	-0.08	
Weight (kg)	2095	80.60	13.30	79.60	42.80	154.00	0.89	0.43	320	69.90	13.50	68.20	40.80	134.00	0.90	0.36	
Height (m)	2094	1.73	0.07	1.73	1.51	1.94	-0.02	1.00	319	1.59	0.07	1.59	1.39	1.78	-0.08	1.00	

Supplementary Table 4. Study-specific descriptive statistics for studies included in stage 2 and ancillary analyses

Study	Trait	Men								Women							
		N	Mean	SD	Median	Min	Max	Correlation with BMI	Correlation with height	N	Mean	SD	Median	Min	Max	Correlation with BMI	Correlation with height
MoRGAM Cases	WC (cm)	1084	95.10	10.90	94.50	64.00	144.00	0.88	0.12	287	85.30	12.30	83.50	60.50	121.00	0.87	0.04
	Hip (cm)	1084	100.60	7.70	100.00	74.50	144.00	0.78	0.24	287	103.60	9.40	102.00	82.50	144.00	0.87	0.16
	WHR (cm/cm)	1084	0.90	0.10	1.00	0.70	1.20	0.59	-0.07	287	0.80	0.10	0.80	0.70	1.10	0.56	-0.11
	Age (yrs)	1772	58.80	7.80	58.60	25.50	75.80	-0.01	-0.06	280	57.90	8.50	59.50	30.60	73.90	0.14	-0.29
	BMI (kg/m ²)	1707	27.50	3.90	27.20	17.00	48.00	1.00	-0.04	246	29.10	5.70	28.50	17.90	50.70	1.00	-0.26
	Weight (kg)	1708	81.70	12.80	80.60	50.20	129.00	0.88	0.43	246	72.80	14.10	71.40	46.90	132.00	0.92	0.14
	Height (m)	1707	1.72	0.06	1.72	1.50	1.97	-0.04	1.00	246	1.58	0.06	1.58	1.38	1.80	-0.26	1.00
	WHR (cm/cm)	983	97.50	10.70	96.50	72.00	138.50	0.86	0.12	222	88.90	13.40	87.50	61.50	131.00	0.87	-0.10
NSHD	Hip (cm)	984	101.00	7.80	100.00	75.00	137.00	0.76	0.20	222	105.80	10.30	104.80	82.50	147.50	0.90	-0.06
	WHR (cm/cm)	983	1.00	0.10	1.00	0.70	1.20	0.53	-0.03	222	0.80	0.10	0.80	0.60	1.40	0.43	-0.10
	Age (yrs)	466	53.00		53.00	53.00	53.00			522	53.00		53.00	53.00	53.00		
	BMI (kg/m ²)	464	27.61	4.16	26.97	18.19	43.05	1.00	-0.09	515	27.72	5.54	26.63	18.62	57.12	1.00	-0.10
	Weight (kg)	464	83.51	13.37	81.30	54.40	136.50	0.87	0.36	516	72.04	14.62	69.80	42.20	154.00	0.91	0.28
	Height (m)	464	1.74	0.06	1.74	1.53	1.95	-0.09	1.00	520	1.61	0.06	1.61	1.39	1.79	-0.10	1.00
	WC (cm)	463	98.00	10.76	97.00	74.00	135.00	0.84	0.14	519	86.77	13.30	85.00	63.00	159.00	0.85	0.05
	Hip (cm)	463	103.85	7.10	103.00	89.00	143.00	0.77	0.28	519	106.23	10.89	105.00	75.00	148.00	0.88	0.11
WHR (cm/cm)	463	0.94	0.06	0.94	0.75	1.14	0.62	-0.03	518	0.81	0.07	0.81	0.65	1.10	0.45	-0.03	
PIVUS	Age (yrs)	490	70.13	0.17	70.13	69.84	72.28	0.07	-0.06	488	70.26	0.15	70.28	69.94	70.76	-0.07	-0.02
	BMI (kg/m ²)	490	27.05	3.72	26.90	17.70	43.40	1.00	-0.05	488	27.12	4.92	26.50	16.60	49.80	1.00	-0.13
	Weight (kg)	490	83.70	13.00	82.00	53.00	138.00	0.87	0.45	488	71.33	13.20	71.00	42.00	126.00	0.93	0.25
	Height (m)	490	1.76	0.07	1.75	1.55	1.98	-0.05	1.00	488	1.62	0.06	1.62	1.48	1.84	-0.13	1.00
	WC (cm)	485	94.86	10.45	95.00	64.00	134.00	0.89	0.14	483	87.67	11.61	87.00	60.00	134.00	0.89	0.03
	Hip (cm)	485	100.26	6.75	100.00	86.00	130.00	0.80	0.33	483	101.41	9.26	101.00	71.00	143.00	0.89	0.13

Supplementary Table 4. Study-specific descriptive statistics for studies included in stage 2 and ancillary analyses

Study	Trait	Men								Women							
		N	Mean	SD	Median	Min	Max	Correlation with BMI	Correlation with height	N	Mean	SD	Median	Min	Max	Correlation with BMI	Correlation with height
	WHR (cm/cm)	485	0.94	0.06	0.94	0.64	1.19	0.65	-0.10	483	0.86	0.06	0.86	0.60	1.07	0.51	-0.10
STR	Age (yrs)	1046	74.33	9.01	75.24	45.76	93.03	-0.12	-0.23	1384	76.10	9.73	78.25	39.76	103.78	-0.11	-0.21
	BMI (kg/m ²)	838	25.13	3.36	24.82	14.53	38.57	1.00	-0.09	1378	25.03	4.24	24.55	14.06	46.07	1.00	-0.04
	Weight (kg)	1043	76.67	11.69	76.00	45.00	132.00	0.84	0.46	1377	63.84	11.68	63.00	35.00	126.00	0.90	0.38
	Height (m)	808	1.74	0.07	1.73	1.52	1.95	-0.09	1.00	1377	1.59	0.06	1.60	1.36	1.78	-0.04	1.00
	WC (cm)	563	96.63	9.36	96.00	71.00	130.00	0.80	0.11	780	85.54	10.02	84.00	61.00	131.00	0.78	0.05
	Hip (cm)	563	100.78	7.98	101.00	80.00	131.00	0.65	0.07	778	99.33	9.95	99.00	49.00	133.00	0.75	0.09
	WHR (cm/cm)	563	0.96	0.05	0.96	0.79	1.19	0.40	0.09	778	0.86	0.07	0.86	0.63	1.61	0.20	-0.04
THISEAS - controls	Age (yrs)	432	54.30	13.31	52.00	24.00	89.00	0.09	-0.41	549	60.63	14.07	63.00	25.00	91.86	0.36	-0.57
	BMI (kg/m ²)	416	28.86	4.26	28.43	19.15	44.58	1.00	-0.18	530	28.60	5.26	28.14	17.67	49.56	1.00	-0.29
	Weight (kg)	432	87.43	14.32	86.00	52.00	140.00	0.80	0.44	548	71.82	13.44	71.00	5.50	161.00	0.86	0.22
	Height (m)	430	1.74	0.08	1.75	1.08	1.94	-0.18	1.00	548	1.59	0.08	1.59	1.36	1.83	-0.29	1.00
	WC (cm)	357	101.97	12.39	100.50	53.00	193.00	0.73	0.03	363	90.33	13.92	89.00	38.00	131.00	0.77	-0.15
	Hip (cm)	357	105.47	8.28	105.00	79.00	138.00	0.71	0.08	363	105.17	13.56	105.00	11.00	148.00	0.67	-0.06
	WHR (cm/cm)	357	0.97	0.09	0.96	0.51	1.87	0.35	-0.02	363	0.89	0.48	0.85	0.36	8.27	0.06	-0.03
THISEAS cases	Age (yrs)	435	62.15	10.69	62.00	33.00	88.00	-0.20	-0.20	79	66.19	11.06	67.00	37.00	89.00	-0.23	-0.16
	BMI (kg/m ²)	423	27.91	4.00	27.45	18.37	44.00	1.00	-0.06	78	28.42	4.51	27.68	19.92	40.70	1.00	-0.06
	Weight (kg)	401	82.86	13.16	80.00	48.00	140.00	0.86	0.45	75	72.95	13.06	72.40	49.70	106.80	0.84	0.48
	Height (m)	404	1.72	0.07	1.72	1.55	1.98	-0.06	1.00	75	1.60	0.08	1.60	1.34	1.75	-0.06	1.00
	WC (cm)	341	100.25	12.21	100.00	17.00	140.00	0.68	0.06	62	95.92	17.09	95.50	10.00	134.00	0.31	0.18
	Hip (cm)	199	101.28	9.70	101.00	44.00	123.00	0.57	0.22	54	107.52	12.03	106.00	85.00	144.00	0.84	0.25
	WHR (cm/cm)	199	0.99	0.13	0.99	0.15	2.36	0.14	-0.14	54	0.89	0.14	0.90	0.08	1.15	-0.20	0.07
ULSAM	Age (yrs)	111	71.00	0.63	71.00	69.4	74.10	-0.06	0.02	-	-	-	-	-	-	-	-

Supplementary Table 4. Study-specific descriptive statistics for studies included in stage 2 and ancillary analyses

Study	Trait	Men								Women								
		N	Mean	SD	Median	Min	Max	Correlation with BMI	Correlation with height	N	Mean	SD	Median	Min	Max	Correlation with BMI	Correlation with height	
Whitehall	BMI (kg/m ²)	1112	26.26	3.40	25.93	16.69	46.34	1.00	-0.07	-	-	-	-	-	-	-	-	
	Weight (kg)	1116	80.41	11.42	79.50	46.00	138.70	0.87	0.42	-	-	-	-	-	-	-	-	
	Height (m)	1112	1.75	0.06	1.75	1.56	2.00	-0.07	1.00	-	-	-	-	-	-	-	-	
	WC (cm)	1095	94.77	9.55	94.00	51.00	137.00	0.87	0.13	-	-	-	-	-	-	-	-	
	Hip (cm)	1095	100.17	7.08	100.00	51.00	141.00	0.78	0.28	-	-	-	-	-	-	-	-	
	WHR (cm/cm)	1095	0.95	0.05	0.95	0.78	1.14	0.58	-0.11	-	-	-	-	-	-	-	-	-
	Age (yrs)	2555	60.75	5.91	59.62	50.69	73.71	-0.03	-0.15	822	61.06	6.01	60.55	50.47	73.31	0.10	-0.19	
	BMI (kg/m ²)	2548	26.60	3.66	26.30	17.90	48.70	1.00	-0.08	820	27.29	5.53	26.20	15.80	49.00	1.00	-0.13	
	Weight (kg)	2549	81.71	12.35	80.40	43.00	144.20	0.84	0.42	821	71.05	14.79	68.70	38.90	138.30	0.90	0.28	
	Height (m)	2550	1.75	0.07	1.75	1.44	1.98	-0.08	1.00	821	1.61	0.07	1.61	1.39	1.86	-0.13	1.00	
WC (cm)	2554	95.14	10.35	94.45	66.55	138.00	0.89	0.11	821	86.37	13.51	85.15	55.15	132.50	0.90	0.01		
Hip (cm)	2554	99.91	6.61	99.40	76.00	156.00	0.79	0.33	820	102.52	10.78	101.10	73.20	142.20	0.88	0.15		
WHR (cm/cm)	2554	0.95	0.06	0.95	0.72	1.20	0.66	-0.13	820	0.84	0.07	0.84	0.66	1.11	0.58	-0.14		
WTCCC T2D nonGWAS	Age (yrs)	626	56.27	10.24	57.00	20.83	87.00	-0.19	-0.11	448	56.37	11.07	57.87	19.44	80.09	-0.15	-0.11	
	BMI (kg/m ²)	624	31.19	5.60	30.40	16.80	60.30	1.00	0.04	445	33.83	7.61	33.00	18.70	59.60	1.00	-0.05	
	Weight (kg)	624	96.63	19.19	95.00	57.00	190.51	0.92	0.42	445	88.19	20.89	84.70	43.00	165.50	0.94	0.30	
	Height (m)	625	1.76	0.07	1.76	1.56	2.20	0.04	1.00	445	1.61	0.07	1.61	1.39	1.79	-0.05	1.00	
	WC (cm)	606	108.7	14.9	107.0	75.0	200.7	0.87	0.19	431	104.3	15.4	104.0	60.0	149.0	0.86	0.09	
	Hip (cm)	606	110.2	12.0	109.0	86.4	215.9	0.76	0.22	431	116.2	15.5	114.0	79.5	172.0	0.89	0.09	
	WHR (cm/cm)	604	0.99	0.07	1.00	0.80	1.28	0.48	0.05	431	0.90	0.07	0.90	0.70	1.16	0.14	0.01	

Supplementary Table 4. Study-specific descriptive statistics for studies included in stage 2 and ancillary analyses

Study	Trait	Men								Women							
		N	Mean	SD	Median	Min	Max	Correlation with BMI	Correlation with height	N	Mean	SD	Median	Min	Max	Correlation with BMI	Correlation with height
Ancillary																	
Essen Obesity Study (Essen Case-Control GWAS)	Age (yrs)	171	25.38	4.57	24.71	16.88	46.32	0.20	0.03	264	26.54	6.37	24.51	17.26	58.78	0.44	-0.15
Controls	BMI (kg/m ²)	171	18.86	0.94	18.95	16.29	21.80	1.00	0.07	264	17.58	0.95	17.69	13.22	19.78	1.00	-0.09
	Weight (kg)	171	63.64	6.12	63.40	46.50	83.10	0.58	0.85	264	50.53	4.61	50.50	38.40	64.50	0.52	0.80
	Height (m)	171	1.84	0.07	1.83	1.64	2.04	0.07	1.00	264	1.69	0.07	1.70	1.50	1.90	-0.09	1.00
	WC (cm)	167	73.21	4.16	73.00	62.00	85.00	0.48	0.50	257	64.71	4.31	64.00	55.50	85.00	0.37	0.27
	Hip (cm)	167	91.34	4.79	91.00	67.00	102.00	0.41	0.55	256	87.13	5.22	87.00	62.00	101.00	0.31	0.42
	WHR (cm/cm)	167	0.80	0.04	0.80	0.68	1.04	0.09	0.00	256	0.74	0.05	0.74	0.62	0.97	0.10	-0.11
Essen Obesity Study (Essen Case-Control GWAS)	Age (yrs)	192	14.18	3.85	13.96	3.37	39.18	0.40	0.70	261	14.50	3.67	14.31	5.62	24.42	0.59	0.61
Cases	BMI (kg/m ²)	192	33.11	6.48	32.34	21.88	56.86	1.00	0.47	261	33.18	6.84	32.33	20.35	61.88	1.00	0.40
	Weight (kg)	190	92.81	29.19	89.45	24.90	186.20	0.89	0.80	260	87.59	24.58	87.80	25.80	176.00	0.90	0.74
	Height (m)	190	1.66	0.15	1.68	1.01	1.94	0.47	1.00	260	1.61	0.13	1.64	1.13	1.82	0.40	1.00
	WC (cm)	130	102.87	17.03	102.00	50.00	152.00	0.86	0.62	180	96.12	16.08	94.00	45.00	150.00	0.85	0.50
	Hip (cm)	129	108.75	15.67	108.00	40.00	150.00	0.83	0.66	178	112.16	16.74	112.00	42.00	160.00	0.84	0.62
	WHR (cm/cm)	129	0.95	0.08	0.93	0.75	1.25	0.23	0.07	178	0.86	0.12	0.85	0.55	2.07	0.08	-0.10
Essen Obesity Study (Essen Obesity Trio GWAS) Parents	Age (yrs)	687	44.21	6.13	43.63	29.85	71.65	0.06	-0.20	696	40.89	5.44	40.49	24.71	62.03	0.08	-0.13
Parents	BMI (kg/m ²)	696	30.34	5.43	29.62	18.04	54.33	1.00	-0.02	698	30.23	7.13	28.88	17.15	64.34	1.00	-0.10
	Weight (kg)	696	95.83	18.72	93.00	56.50	180.00	0.91	0.40	698	82.35	20.03	78.00	46.00	170.10	0.92	0.28
	Height (m)	696	1.78	0.07	1.77	1.53	1.99	-0.02	1.00	698	1.65	0.07	1.65	1.01	1.83	-0.10	1.00
	WC (cm)	524	105.26	14.75	104.00	55.00	166.00	0.89	0.13	565	93.94	17.04	91.50	56.00	167.00	0.86	0.06
	Hip (cm)	524	107.91	10.98	106.50	51.00	158.00	0.81	0.20	565	111.24	15.74	109.00	55.50	223.00	0.83	0.05
	WHR (cm/cm)	524	0.97	0.08	0.97	0.56	1.35	0.53	-0.05	565	0.84	0.09	0.83	0.47	1.43	0.37	0.02
Essen Obesity Study (Essen Obesity Trio GWAS) Offspring	Age (yrs)	318	13.31	2.98	13.43	4.19	24.90	0.42	0.82	387	13.54	3.04	13.71	3.76	22.18	0.55	0.74
Offspring	BMI (kg/m ²)	318	31.60	5.52	30.86	18.74	64.70	1.00	0.45	387	32.36	6.04	31.21	19.73	56.75	1.00	0.43
	Weight (kg)	316	86.50	25.51	85.30	21.30	214.30	0.87	0.82	386	84.76	23.58	84.00	26.00	174.00	0.90	0.77
	Height (m)	316	1.64	0.15	1.66	1.07	1.95	0.45	1.00	386	1.60	0.13	1.63	1.06	1.84	0.43	1.00

Supplementary Table 4. Study-specific descriptive statistics for studies included in stage 2 and ancillary analyses

Study	Trait	Men								Women							
		N	Mean	SD	Median	Min	Max	Correlation with BMI	Correlation with height	N	Mean	SD	Median	Min	Max	Correlation with BMI	Correlation with height
	WC (cm)	250	100.31	14.41	99.50	57.00	142.00	0.81	0.68	300	95.43	13.23	95.00	55.00	150.00	0.83	0.47
	Hip (cm)	250	107.54	13.68	108.50	60.00	178.00	0.76	0.71	298	110.80	15.74	110.00	71.00	213.00	0.83	0.63
	WHR (cm/cm)	250	0.93	0.08	0.93	0.64	1.34	0.21	0.06	298	0.86	0.08	0.86	0.46	1.18	-0.01	-0.25
French Extreme Obesity Study	Age (yrs)	591	11.61	2.66	11.83	4.00	17.00	0.22	0.85	662	11.39	2.95	11.42	2.00	17.17	0.29	0.85
Children	BMI (kg/m ²)	591	23.76	7.97	21.69	12.82	70.76	1.00	0.43	662	24.29	8.04	22.54	12.95	59.52	1.00	0.40
	Weight (kg)	591	57.70	28.24	51.00	19.60	237.00	0.91	0.74	662	56.24	25.96	49.00	18.49	166.00	0.92	0.70
	Height (m)	591	1.53	0.16	1.52	1.15	1.97	0.43	1.00	662	1.49	0.15	1.52	0.95	1.83	0.40	1.00
	WC (cm)	533	79.78	21.79	73.00	46.00	170.00	0.94	0.54	589	76.15	20.46	69.00	47.00	155.00	0.92	0.43
	WHR (cm/cm)	530	0.89	0.10	0.87	0.71	1.58	0.61	0.05	588	0.84	0.12	0.82	0.63	2.35	0.41	-0.19
	Hip (cm)	530	88.67	18.53	87.00	56.96	200.00	0.91	0.67	588	90.16	18.70	87.00	57.00	155.00	0.90	0.67
French Extreme Obesity Study	Age (yrs)	318	49.48	9.07	50.00	20.00	69.00	-0.39	-0.29	1055	46.58	10.63	47.00	19.00	77.00	-0.21	-0.27
Adult	BMI (kg/m ²)	318	34.18	13.70	24.60	20.40	83.86	1.00	0.16	1055	34.00	13.91	24.80	16.60	87.24	1.00	0.11
	Weight (kg)	318	104.51	44.06	78.00	51.00	270.00	0.98	0.35	1055	88.59	37.86	67.00	40.00	231.80	0.98	0.30
	Height (m)	318	1.74	0.07	1.74	1.55	1.95	0.16	1.00	1055	1.61	0.06	1.61	1.43	1.80	0.11	1.00
	WC (cm)	310	109.84	31.35	92.00	73.00	200.00	0.97	0.25	984	96.38	30.18	79.00	57.00	195.00	0.96	0.18
	WHR (cm/cm)	310	0.94	0.10	0.92	0.75	1.46	0.56	0.12	982	0.83	0.10	0.80	0.59	1.41	0.64	0.07
	Hip (cm)	310	115.31	27.42	99.05	84.01	245.00	0.94	0.25	981	114.83	26.58	101.00	70.00	204.00	0.96	0.20
GEO-IT	Age (yrs)	680	40.17	11.43	39	19	73	0.06	-	1112	39.1	9.8	38.0	19.0	66.0	0.2	-
Controls	BMI (kg/m ²)	680	21.7	1.55	22	16	24.5	1	-	1112	20.9	1.7	21.0	14.0	24.0	1	-
	Weight (kg)	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
	Height (m)	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
	WC (cm)	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
	Hip (cm)	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
	WHR (cm/cm)	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
GEO-IT	Age (yrs)	580	47.89	14	48	18	82	-0.21	-0.26	1137	52.9	14.6	55.0	18.0	85.0	-0.1	-0.37
Cases	BMI (kg/m ²)	580	47.49	6.86	45.91	40	85.6	1	0.05	1137	47.3	6.4	46.0	40.5	82.7	1	-0.06
	Weight (kg)	580	142.96	24.78	138	96.4	265	0.86	0.54	1137	117.8	18.7	115.0	70.6	207.4	0.81	0.51
	Height (m)	580	1.73	0.08	1.73	1.43	1.98	0.05	1	1137	1.58	0.07	1.58	1.30	1.82	-0.06	1

Supplementary Table 4. Study-specific descriptive statistics for studies included in stage 2 and ancillary analyses

Study	Trait	Men								Women							
		N	Mean	SD	Median	Min	Max	Correlation with BMI	Correlation with height	N	Mean	SD	Median	Min	Max	Correlation with BMI	Correlation with height
	WC (cm)	580	141.82	13.4	140	110	197	0.82	0.28	1137	126.4	13.2	125.0	90.0	260.0	0.69	0.13
	Hip (cm)	580	139.06	15.76	137	90	202	0.81	0.22	1137	140.7	13.4	139.0	108.0	290.0	0.79	0.11
	WHR (cm/cm)	580	1.02	0.07	1.02	0.77	1.33	-0.17	0.01	1137	0.89	0.07	0.9	0.61	1.22	-0.03	-0.01
GOYA	Age (yrs)	808	20	1.9	19	18	31	0.1	0.06	86219	29.6	4.3	29.4	15.6	47.0	-0.02	0.01
	BMI (kg/m ²)	808	21.7	2.5	21.3	15.7	36.2	1	-0.08	79322	23.6	4.2	22.6	13.9	64.4	1.00	-0.05
	Weight (kg)	808	67.8	8.9	67	47	116	0.83	0.5	79393	67.1	12.8	65.0	38.0	177.0	0.92	0.33
	Height (m)	808	1.77	0.07	1.77	1.57	1.96	-0.08	1	80622	1.69	0.06	1.69	1.42	1.98	-0.05	1.00
	WC (cm)	na	na	na	na	na	na	na	na	na	na	na	na	na	na	na	na
	Hip (cm)	na	na	na	na	na	na	na	na	na	na	na	na	na	na	na	na
	WHR (cm/cm)	na	na	na	na	na	na	na	na	na	na	na	na	na	na	na	na

Supplementary Table 5. Number of individuals in each study for the tails and clinical class of obesity categories

Study name		Normal-weight	Overweight	Obese class I	Obese class II	Obese class III	Height tails		BMI tails		WHR tails	
		BMI < 25	BMI ≥ 25	BMI ≥ 30	BMI ≥ 35	BMI ≥ 40	95 th -100 th pct	0 th -5 th pct	95 th -100 th pct	0 th -5 th pct	95 th -100 th pct	0 th -5 th pct
		N	N	N	N	N	N	N	N	N	N	N
Stage 1:												
ADVANCE Controls	Men	43	85	24	7	2	7	7	7	7	-	-
	Women	103	82	38	15	11	10	10	10	10	-	-
	Pooled	146	167	62	22	13	17	17	17	17	-	-
ADVANCE Cases	Men	14	100	68	27	9	6	6	6	6	-	-
	Women	44	117	84	53	20	9	9	9	9	-	-
	Pooled	58	217	152	80	29	15	15	15	15	-	-
AGES	Men	590	761	93	-	-	68	68	68	68	-	-
	Women	1087	1087	169	-	-	94	96	93	93	-	-
	Pooled	1677	1848	262	47	8	162	164	161	161	-	-
ARIC	Men	1017	2815	855	168	36	192	192	191	191	191	191
	Women	1987	2301	1000	365	112	214	214	215	215	215	215
	Pooled	3004	5116	1855	533	148	406	406	406	406	406	406
B58C-T1DGC	Men	281	978	341	76	14	64	64	63	63	60	60
	Women	579	749	304	120	53	67	67	67	67	65	65
	Pooled	860	1727	645	196	67	131	131	130	130	125	125
B58C-WTCCC	Men	194	547	191	48	7	38	38	38	38	35	35
	Women	324	414	181	57	20	37	37	37	37	37	37
	Pooled	518	961	372	105	27	75	75	75	75	72	72
BRIGHT	Men	149	570	176	-	-	36	36	36	36	34	34
	Women	319	768	282	-	-	55	55	55	55	50	50
	Pooled	468	-	-	49	-	-	-	-	-	-	-
CAD WTCCC	Men	419	1109	345	62	10	59	58	57	57	-	-
	Women	123	276	103	35	10	19	19	19	19	-	-
	Pooled	542	1385	448	97	20	78	77	76	76	-	-
CASP1 controls	Men	172	311	306	67	7	25	25	24	24	-	-
	Women	-	-	-	-	-	-	-	-	-	-	-
	Pooled	-	-	-	-	-	-	-	-	-	-	-
CASP1 cases	Men	178	306	69	9	-	24	24	24	24	-	-
	Women	-	-	-	-	-	-	-	-	-	-	-

Supplementary Table 5. Number of individuals in each study for the tails and clinical class of obesity categories

Study name		Normal-weight	Overweight	Obese class I	Obese class II	Obese class III	Height tails		BMI tails		WHR tails	
		BMI < 25	BMI ≥ 25	BMI ≥ 30	BMI ≥ 35	BMI ≥ 40	95 th -100 th pct	0 th -5 th pct	95 th -100 th pct	0 th -5 th pct	95 th -100 th pct	0 th -5 th pct
		N	N	N	N	N	N	N	N	N	N	N
	Pooled	-	-	-	-	-	-	-	-	-	-	-
CASP2 controls	Men	202	298	53	5	3	26	26	25	25	-	-
	Women	-	-	-	-	-	-	-	-	-	-	-
	Pooled	-	-	-	-	-	-	-	-	-	-	-
CASP2 cases	Men	538	885	190	17	2	73	73	71	71	-	-
	Women	-	-	-	-	-	-	-	-	-	-	-
	Pooled	-	-	-	-	-	-	-	-	-	-	-
CHS	Men	459	817	189	19	5	65	68	64	63	63	64
	Women	845	1107	374	104	31	113	99	98	97	98	98
	Pooled	1304	1924	563	123	36	178	167	162	160	161	162
CoLaus	Men	941	1619	444	93	19	155	141	130	127	127	127
	Women	1626	1248	425	131	37	157	198	144	146	144	143
	Pooled	2567	2867	869	224	56	306	343	277	281	271	270
COROGENE controls	Men	302	703	214	44	9	51	51	51	51	51	51
	Women	370	510	184	62	17	44	44	44	44	44	44
	Pooled	672	1213	398	106	26	95	95	95	95	95	95
COROGENE cases	Men	360	814	265	75	17	57	57	57	57	0	0
	Women	233	347	147	43	12	28	28	28	28	0	0
	Pooled	593	1161	412	118	29	85	85	85	85	0	0
deCODE	Men	2597	6616	2447	646	187	450	453	465	465	145	132
	Women	7403	10183	4128	1361	436	870	867	875	875	149	145
	Pooled	10000	16799	6575	2007	623	1320	1320	1340	1340	294	277
DGI Controls	Men	182	387	81	-	-	28	28	29	29	12	12
	Women	201	345	107	-	-	29	29	28	28	11	11
	Pooled	383	732	188	27	6	57	57	57	57	23	23
DGI Cases	Men	145	551	81	-	-	32	32	35	35	26	25
	Women	144	490	214	-	-	35	35	32	32	23	23
	Pooled	289	1041	295	90	16	67	67	67	67	49	48
EGCUT	Men	531	587	170	38	5	56	56	56	57	31	32
	Women	641	521	215	82	14	59	59	63	57	36	36

Supplementary Table 5. Number of individuals in each study for the tails and clinical class of obesity categories

Study name		Normal-weight	Overweight	Obese class I	Obese class II	Obese class III	Height tails		BMI tails		WHR tails	
		BMI < 25	BMI ≥ 25	BMI ≥ 30	BMI ≥ 35	BMI ≥ 40	95 th -100 th pct	0 th -5 th pct	95 th -100 th pct	0 th -5 th pct	95 th -100 th pct	0 th -5 th pct
		N	N	N	N	N	N	N	N	N	N	N
	Pooled	1172	1108	385	120	29	115	115	119	114	67	68
EPIC - Obesity	Men	348	782	156	21	2	57	56	57	56	57	56
	Women	572	712	223	56	14	68	63	65	64	65	64
	Pooled	920	1494	379	77	16	125	119	122	120	122	120
ERF	Men	265	644	189	38	5	45	45	45	45	45	45
	Women	527	668	241	73	4	59	59	59	59	59	59
	Pooled	792	1312	430	111	9	-	-	-	-	-	-
FamHS	Men	90	326	123	-	-	-	-	-	-	-	-
	Women	180	260	126	-	-	-	-	-	-	-	-
	Pooled	270	586	249	63	25	43	42	43	43	43	43
FENLAND	Men	156	459	131	24	10	31	31	31	31	31	31
	Women	358	429	177	61	21	40	40	40	40	40	40
	Pooled	514	888	308	85	31	71	71	71	71	71	71
FRAM	Men	1284	2566	732	171	39	232	202	127	171	39	55
	Women	2908	1620	616	243	86	172	202	310	255	124	107
	Pooled	4192	4186	1348	414	125	404	404	437	426	163	162
FUSION Controls	Men	180	392	104	-	-	28	28	28	28	28	29
	Women	192	407	138	-	-	30	29	30	30	29	29
	Pooled	372	799	242	35	5	58	57	58	58	57	58
FUSION Cases	Men	79	424	265	-	-	31	31	31	31	31	31
	Women	45	544	263	-	-	22	24	23	23	23	24
	Pooled	124	968	528	155	36	53	55	54	54	54	55
GENMETS Controls	Men	231	218	34	5	0	21	20	21	20	21	20
	Women	276	214	59	11	3	22	21	23	23	24	22
	Pooled	507	432	93	16	3	43	41	44	43	45	42
GENMETS Cases	Men	30	438	171	39	8	21	20	22	21	22	23
	Women	85	393	207	68	20	22	21	23	23	23	22
	Pooled	115	831	378	107	28	43	41	45	44	45	45
GerMIFS1	Men	154	487	138	20	-	32	32	32	32	-	-

Supplementary Table 5. Number of individuals in each study for the tails and clinical class of obesity categories

Study name	Normal-weight	Overweight	Obese class I	Obese class II	Obese class III	Height tails		BMI tails		WHR tails		
	BMI < 25	BMI ≥ 25	BMI ≥ 30	BMI ≥ 35	BMI ≥ 40	95 th -100 th pct	0 th -5 th pct	95 th -100 th pct	0 th -5 th pct	95 th -100 th pct	0 th -5 th pct	
	N	N	N	N	N	N	N	N	N	N	N	
	Women	96	213	81	16	-	16	15	16	16	-	-
	Pooled	250	700	219	36	-	-	-	-	-	-	-
GerMIFS2	Men	280	672	212	27	-	43	43	43	43	-	-
	Women	86	153	69	21	-	11	11	11	11	-	-
	Pooled	366	825	281	48	-	-	-	-	-	-	-
GOOD	Men	795	143	30	5	1	47	48	47	47	48	50
	Women	-	-	-	-	-	-	-	-	-	-	-
	Pooled	-	-	-	-	-	-	-	-	-	-	-
HBCS	Men	194	378	123	30	9	37	37	37	37	37	37
	Women	316	399	179	66	24	50	50	50	50	50	50
	Pooled	510	777	302	96	33	87	87	87	87	87	87
KORA S3	Men	160	653	168	31	3	44	42	41	41	41	41
	Women	307	522	188	51	10	46	43	42	42	42	42
	Pooled	467	1175	356	82	13	90	85	83	83	83	83
KORA S4	Men	167	716	223	41	12	46	45	45	45	45	45
	Women	335	593	255	76	17	49	47	47	47	47	47
	Pooled	502	1309	478	117	29	95	92	92	92	92	92
MGS	Men	171	1076	597	247	90	53	98	63	61	-	-
	Women	312	1038	705	398	213	99	81	67	69	-	-
	Pooled	483	2114	1302	645	303	152	179	130	130	-	-
MICROS	Men	198	269	69	14	3	23	23	23	23	8	8
	Women	345	267	105	27	8	30	30	31	30	8	8
	Pooled	543	536	174	41	11	50	50	54	54	14	14
Migen Cases	Men	617	158	158	-	-	42	52	42	43	-	-
	Women	320	150	167	-	-	36	56	27	27	-	-
	Pooled	937	308	325	149	-	78	108	69	70	-	-
Migen Controls	Men	596	248	167	-	-	40	42	38	38	-	-
	Women	305	253	143	-	-	34	23	23	23	-	-
	Pooled	901	501	310	91	-	74	65	61	61	-	-
NBS WTCCC	Men	241	458	119	22	7	36	35	34	33	-	-
	Women	390	356	114	28	7	36	35	36	35	-	-

Supplementary Table 5. Number of individuals in each study for the tails and clinical class of obesity categories

Study name		Normal-weight	Overweight	Obese class I	Obese class II	Obese class III	Height tails		BMI tails		WHR tails	
		BMI < 25	BMI ≥ 25	BMI ≥ 30	BMI ≥ 35	BMI ≥ 40	95 th -100 th pct	0 th -5 th pct	95 th -100 th pct	0 th -5 th pct	95 th -100 th pct	0 th -5 th pct
		N	N	N	N	N	N	N	N	N	N	N
	Pooled	631	814	233	50	14	72	70	70	68	-	-
NFBC66	Men	1183	1071	190	34	8	135	134	128	128	137	137
	Women	1534	717	215	77	25	113	112	115	114	112	112
	Pooled	2717	1788	405	111	33	248	246	243	242	249	249
NHS	Men	-	-	-	-	-	-	-	-	-	-	-
	Women	1419	868	278	76	20	113	124	273	106	772	79
	Pooled	-	-	-	-	-	-	-	-	-	-	-
NSPHS	Men	112	195	59	14	7	15	15	15	15	-	-
	Women	170	170	61	20	5	17	17	17	16	-	-
	Pooled	282	365	120	34	12	32	32	32	32	-	-
NTR and NESDA MDD Controls	Men	314	287	75	9	0	32	32	34	34	33	33
	Women	670	324	88	26	7	54	30	54	55	55	55
	Pooled	984	611	163	35	7	73	64	89	89	89	89
NTR and NESDA MDD Cases	Men	216	215	79	12	6	26	24	26	25	26	26
	Women	679	317	128	60	19	59	55	60	60	59	59
	Pooled	895	532	207	72	25	75	75	86	84	86	86
ORCADES	Men	71	254	94	19	7	16	16	16	16	18	18
	Women	132	240	95	35	13	18	18	19	19	18	18
	Pooled	203	494	189	54	20	35	35	35	35	34	34
PLCO	Men	579	1659	504	85	14	113	113	112	112	-	-
	Women	-	-	-	-	-	-	-	-	-	-	-
	Pooled	-	-	-	-	-	-	-	-	-	-	-
PROCARDIS	Men	457	1407	421	75	11	94	94	94	94	31	30
	Women	283	373	150	48	11	33	33	33	33	17	18
	Pooled	740	1780	571	123	22	127	127	127	127	48	48
RUNMC	Men	767	1010	171	34	8	91	86	88	91	-	-
	Women	506	506	134	33	11	53	58	56	53	-	-
	Pooled	1273	1516	305	67	19	144	144	144	144	-	-

Supplementary Table 5. Number of individuals in each study for the tails and clinical class of obesity categories

Study name		Normal-weight	Overweight	Obese class I	Obese class II	Obese class III	Height tails		BMI tails		WHR tails	
		BMI < 25	BMI ≥ 25	BMI ≥ 30	BMI ≥ 35	BMI ≥ 40	95 th -100 th pct	0 th -5 th pct	95 th -100 th pct	0 th -5 th pct	95 th -100 th pct	0 th -5 th pct
		N	N	N	N	N	N	N	N	N	N	N
RS1	Men	993	1399	177	8	0	118	118	118	118	112	112
	Women	1219	2152	649	113	18	169	165	169	168	157	156
	Pooled	2212	3551	826	121	18	-	-	-	-	-	-
SASBAC controls	Men	-	-	-	-	-	-	-	-	-	-	-
	Women	356	399	89	19	4	38	37	38	38	-	-
	Pooled	-	-	-	-	-	-	-	-	-	-	-
SASBAC cases	Men	-	-	-	-	-	-	-	-	-	-	-
	Women	377	416	102	22	6	40	39	40	40	-	-
	Pooled	-	-	-	-	-	-	-	-	-	-	-
SardiNIA	Men	87	336	124	20	1	22	22	22	22	22	22
	Women	158	307	132	34	13	24	24	24	24	24	24
	Pooled	245	643	256	54	14	45	50	45	50	45	50
SEARCH	Men	-	-	-	-	-	-	-	-	-	-	-
	Women	637	919	368	113	41	79	79	78	78	-	-
	Pooled	-	-	-	-	-	-	-	-	-	-	-
SHIP	Men	514	1491	507	86	13	100	100	100	100	100	100
	Women	866	1199	535	181	37	102	102	103	103	103	103
	Pooled	1380	2690	1042	267	50	202	202	203	203	203	203
Sorbs	Men	117	271	76	14	5	19	19	19	19	19	19
	Women	242	311	139	43	17	27	27	27	27	27	27
	Pooled	359	582	215	57	22	46	46	46	46	46	46
TWINSUK	Men	-	-	-	-	-	-	-	-	-	-	-
	Women	872	605	185	67	24	76	79	74	74	55	56
	Pooled	-	-	-	-	-	-	-	-	-	-	-
T2D WTCCC	Men	171	947	526	179	64	46	46	51	50	48	47
	Women	104	702	476	261	114	40	40	40	40	39	39
	Pooled	275	1649	1002	440	178	86	86	91	90	87	86
VIS	Men	76	249	81	8	2	16	16	16	16	16	16
	Women	154	291	122	24	4	22	22	23	23	22	22
	Pooled	230	540	203	32	5	39	39	39	39	39	39

Supplementary Table 5. Number of individuals in each study for the tails and clinical class of obesity categories

Study name		Normal-weight	Overweight	Obese class I	Obese class II	Obese class III	Height tails		BMI tails		WHR tails	
		BMI < 25	BMI ≥ 25	BMI ≥ 30	BMI ≥ 35	BMI ≥ 40	95 th -100 th pct	0 th -5 th pct	95 th -100 th pct	0 th -5 th pct	95 th -100 th pct	0 th -5 th pct
		N	N	N	N	N	N	N	N	N	N	N
WGHS	Men	-	-	-	-	-	-	-	-	-	-	-
	Women	11826	11062	4001	1273	389	1155	1155	1145	1145	1027	1027
	Pooled	-	-	-	-	-	-	-	-	-	-	-
YFS	Men	332	576	169	42	9	46	46	46	46	46	46
	Women	605	476	171	50	15	55	55	55	55	54	54
	Pooled	937	1052	340	92	26	101	101	101	101	100	100

Supplementary Table 5. Number of individuals in each study for the tails and clinical class of obesity categories

Study name		Normal-weight	Overweight	Obese class I	Obese class II	Obese class III	Extreme height		Extreme BMI		Extreme WHR adjusted for BMI	
		BMI < 25	BMI ≥ 25	BMI ≥ 30	BMI ≥ 35	BMI ≥ 40	95 th -100 th pct	0 th -5 th pct	95 th -100 th pct	0 th -5 th pct	95 th -100 th pct	0 th -5 th pct
		N	N	N	N	N	N	N	N	N	N	N
Stage 2 - In silico												
B58C-REPL	Men	324	906	303	66	19	61	61	62	62	58	59
	Women	539	653	266	111	41	60	59	60	59	58	58
	Pooled	863	1559	569	177	60	121	120	122	121	116	117
BHS	Men	184	328	87	14	1	26	26	26	26	25	25
	Women	373	320	117	29	3	35	35	35	35	34	34
	Pooled	-	-	-	-	-	-	-	-	-	-	-
HYPERGENES Controls	Men	405	593	108	10	1	50	71	50	50	7	7
	Women	387	297	64	14	2	34	49	34	34	8	8
	Pooled	792	890	172	24	3	88	94	84	84	14	14
HYPERGENES Cases	Men	250	822	203	34	8	54	62	54	54	8	8
	Women	218	320	112	37	10	27	32	27	27	8	8
	Pooled	468	1142	315	71	18	81	88	81	81	16	16
LifeLines	Men	1202	2275	539	89	17	175	173	175	175	175	175
	Women	2285	2354	754	231	63	233	233	233	233	233	233
	Pooled	3487	4629	1293	320	80	408	406	408	408	408	408
PLCO2 controls	Men	200	445	127	26	2	32	32	32	32	-	-
	Women	254	290	100	38	10	28	28	28	28	-	-
	Pooled	454	735	227	64	12	60	60	60	60	-	-
PLCO2 cases	Men	572	1468	457	102	28	102	102	102	102	-	-
	Women	410	523	210	57	24	47	47	47	47	-	-
	Pooled	982	1991	667	159	52	149	149	149	149	-	-
PREVEND	Men	707	1163	285	42	6	93	95	93	87	93	87
	Women	876	876	293	82	26	87	88	94	88	94	88
	Pooled	1583	2039	578	124	32	180	183	187	175	187	175
QIMR	Men	748	898	202	32	4	83	83	83	83	-	-
	Women	1421	886	314	107	37	117	117	116	116	-	-
	Pooled	2169	1784	516	139	41	200	200	199	199	-	-
RS2	Men	252	624	125	13	1	44	44	44	44	44	44

Supplementary Table 5. Number of individuals in each study for the tails and clinical class of obesity categories

Study name	Normal-weight	Overweight	Obese class I	Obese class II	Obese class III	Extreme height		Extreme BMI		Extreme WHR adjusted for BMI		
	BMI < 25	BMI ≥ 25	BMI ≥ 30	BMI ≥ 35	BMI ≥ 40	95 th -100 th pct	0 th -5 th pct	95 th -100 th pct	0 th -5 th pct	95 th -100 th pct	0 th -5 th pct	
	N	N	N	N	N	N	N	N	N	N	N	
	Women	327	708	249	75	11	52	52	52	52	52	52
	Pooled	579	1332	374	88	12	-	-	-	-	-	-
RS3	Men	201	676	219	57	11	44	44	44	44	43	43
	Women	384	745	279	93	25	57	57	57	57	55	55
	Pooled	585	1421	498	150	36	-	-	-	-	-	-
TRAILS	Men	440	99	32	9	2	27	27	27	27	27	27
	Women	469	133	31	7	5	31	31	31	31	31	31
	Pooled	909	232	63	16	7	58	58	58	58	58	58
TWINGENE	Men	1660	2750	564	79	18	225	225	220	221	219	218
	Women	2380	2506	732	175	34	251	251	245	245	244	244
	Pooled	4040	5256	1296	254	52	475	476	464	464	463	462

Supplementary Table 5. Number of individuals in each study for the tails and clinical class of obesity categories

Study -name		Normal-weight	Overweight	Obese class I	Obese class II	Obese class III	Extreme height		Extreme BMI		Extreme WHR adjusted for BMI	
		BMI < 25	BMI ≥ 25	BMI ≥ 30	BMI ≥ 35	BMI ≥ 40	95 th -100 th pct	0 th -5 th pct	95 th -100 th pct	0 th -5 th pct	95 th -100 th pct	0 th -5 th pct
		N	N	N	N	N	N	N	N	N	N	N
Stage 2 - Metabochip												
AMC-PAS	Men	112	256	78	12	1	19	18	18	18	-	-
	Women	54	66	25	8	2	6	6	6	6	-	-
	Pooled	166	322	103	20	3	25	24	24	24	-	-
Cardiogenics Controls	Men	69	79	20	2	-	7	7	7	7	-	-
	Women	110	117	29	6	-	11	11	10	11	-	-
	Pooled	179	196	49	8	-	18	18	17	18	-	-
Cardiogenics Cases	Men	65	262	112	26	3	16	16	16	16	-	-
	Women	19	31	17	6	1	2	2	2	2	-	-
	Pooled	84	293	129	32	4	18	18	18	18	-	-
D2D2007.DPS.DRSE XTRA.FUSION2.ME TSIM controls	Men	1091	2300	543	87	15	169	169	169	169	170	170
	Women	950	1755	698	201	53	136	136	135	135	136	136
	Pooled	2041	4055	1241	288	68	305	305	304	304	306	306
D2D2007.DPS.DRSE XTRA.FUSION2.ME TSIM cases	Men	270	1763	930	281	93	102	102	101	102	102	102
	Women	71	648	423	187	63	36	36	36	36	35	35
	Pooled	341	2411	1353	468	156	137	137	137	138	137	138
DILGOM	Men	547	1244	357	83	20	91	91	90	90	90	89
	Women	908	1231	499	168	55	108	108	108	106	106	107
	Pooled	1455	2475	856	251	75	199	199	198	196	196	196
GoDARTS controls	Men	535	1383	389	75	17	96	96	96	96	-	-
	Women	706	1082	394	121	36	90	90	90	90	-	-
	Pooled	1241	2465	783	196	53	186	186	186	186	-	-
GoDARTS cases	Men	193	1731	1011	400	140	97	97	97	97	-	-
	Women	144	1202	846	460	205	68	68	68	68	-	-
	Pooled	337	2933	1857	860	345	165	165	165	165	-	-
EGCUT controls	Men	278	62	0	0	0	19	17	17	18	17	17
	Women	535	65	2	1	0	31	36	30	30	30	30

Supplementary Table 5. Number of individuals in each study for the tails and clinical class of obesity categories

Study -name		Normal-weight	Overweight	Obese class I	Obese class II	Obese class III	Extreme height		Extreme BMI		Extreme WHR adjusted for BMI	
		BMI < 25	BMI ≥ 25	BMI ≥ 30	BMI ≥ 35	BMI ≥ 40	95 th -100 th pct	0 th -5 th pct	95 th -100 th pct	0 th -5 th pct	95 th -100 th pct	0 th -5 th pct
		N	N	N	N	N	N	N	N	N	N	N
	Pooled	813	127	2	1	0	50	53	47	48	47	47
EGCUT CAD	Men	93	253	114	25	5	19	18	18	18	18	18
	Women	77	277	145	54	16	22	20	18	18	18	18
	Pooled	170	530	259	79	21	41	38	36	36	36	36
EGCUT T2D	Men	11	343	253	96	24	20	19	18	18	14	14
	Women	22	591	450	217	79	39	38	31	31	21	21
	Pooled	33	934	703	313	103	59	57	49	49	35	35
Ely	Men	210	534	162	31	7	38	38	38	38	37	37
	Women	330	525	202	71	24	43	43	43	43	43	43
	Pooled	540	1059	364	102	31	81	81	81	81	80	80
EPIC_CONTROLS	Men	148	262	56	5	1	21	21	21	21	21	21
	Women	254	297	82	14	3	28	28	28	28	28	28
	Pooled	402	559	138	19	4	49	49	49	49	49	49
EPIC_CASES	Men	40	392	142	26	7	22	22	22	22	22	22
	Women	45	246	136	45	14	15	15	15	15	15	15
	Pooled	85	638	278	71	21	37	37	37	37	37	37
Fenland	Men	494	992	297	66	12	75	75	75	75	75	75
	Women	806	892	350	137	39	85	85	85	85	85	85
	Pooled	1300	1884	647	203	51	160	160	160	160	160	160
GLACIER	Men	943	1434	269	31	9	119	119	120	120	-	-
	Women	1898	1754	521	121	27	183	183	184	183	-	-
	Pooled	2841	3188	790	152	36	302	302	304	303	-	-
HNR	Men	438	1823	596	125	127	107	114	113	114	115	119
	Women	772	1473	637	192	56	109	146	113	114	117	110
	Pooled	1210	3296	1233	317	183	216	260	226	228	232	229
HUNT.TROMSO controls	Men	237	422	72	9	-	34	33	33	33	25	25
	Women	226	389	151	23	3	31	32	31	31	24	23
	Pooled	463	811	223	32	3	65	64	64	64	50	50
HUNT.TROMSO cases	Men	103	443	180	29	4	27	28	27	28	21	21

Supplementary Table 5. Number of individuals in each study for the tails and clinical class of obesity categories

Study -name		Normal-weight	Overweight	Obese class I	Obese class II	Obese class III	Extreme height		Extreme BMI		Extreme WHR adjusted for BMI	
		BMI < 25	BMI ≥ 25	BMI ≥ 30	BMI ≥ 35	BMI ≥ 40	95 th -100 th pct	0 th -5 th pct	95 th -100 th pct	0 th -5 th pct	95 th -100 th pct	0 th -5 th pct
		N	N	N	N	N	N	N	N	N	N	N
	Women	95	427	258	85	29	27	27	26	27	20	19
	Pooled	198	870	438	114	33	54	53	54	54	41	40
IMPROVE	Men	425	1240	333	60	11	84	84	84	84	74	86
	Women	647	1129	422	120	35	90	88	89	89	89	102
	Pooled	1072	2369	755	180	46	174	172	173	173	163	188
KORA S3 METABO	Men	207	392	87	9	2	31	31	30	30	30	30
	Women	387	272	99	30	9	35	34	33	33	33	33
	Pooled	594	664	186	39	11	66	65	63	63	63	63
KORA S4 METABO	Men	197	385	90	13	2	30	30	30	30	30	30
	Women	294	330	107	41	7	32	32	32	32	32	32
	Pooled	491	715	197	54	9	62	62	62	62	62	62
LURIC	Men	544	1464	426	64	13	101	101	101	98	98	98
	Women	292	596	224	49	14	44	44	44	44	44	44
	Pooled	836	2060	650	113	27	145	145	145	142	142	142
MORGAM controls	Men	719	1376	361	70	14	106	106	106	102	53	55
	Women	108	211	95	25	7	17	17	17	17	14	15
	Pooled	827	1587	456	95	21	123	123	123	119	67	70
MORGAM cases	Men	430	1277	393	65	12	86	84	87	86	49	49
	Women	64	182	99	41	8	13	13	13	13	12	12
	Pooled	494	1459	492	106	20	99	97	100	99	61	61
NSHD	Men	131	333	117	23	5	24	24	24	24	24	24
	Women	184	331	144	50	16	26	26	26	26	26	26
	Pooled	315	664	261	73	21	50	50	50	50	50	50
PIVUS	Men	145	344	93	14	1	25	25	25	25	25	24
	Women	169	315	127	30	8	24	24	25	25	24	25
	Pooled	314	659	220	44	9	49	49	50	50	49	49
STR	Men	394	354	62	3	-	36	36	38	38	15	15
	Women	643	570	143	35	4	63	63	63	63	34	35
	Pooled	1037	924	205	38	4	99	99	101	101	49	50
THISEAS Controls	Men	77	337	149	37	5	21	20	19	20	17	16

Supplementary Table 5. Number of individuals in each study for the tails and clinical class of obesity categories

Study -name		Normal-weight	Overweight	Obese class I	Obese class II	Obese class III	Extreme height		Extreme BMI		Extreme WHR adjusted for BMI	
		BMI < 25	BMI ≥ 25	BMI ≥ 30	BMI ≥ 35	BMI ≥ 40	95 th -100 th pct	0 th -5 th pct	95 th -100 th pct	0 th -5 th pct	95 th -100 th pct	0 th -5 th pct
		N	N	N	N	N	N	N	N	N	N	N
	Women	142	386	195	58	17	26	27	27	27	18	17
	Pooled	219	723	344	95	22	47	47	46	47	35	33
THISEAS Cases	Men	88	304	97	22	3	19	20	18	18	9	9
	Women	19	55	19	6	-	3	3	3	3	2	1
	Pooled	107	359	116	28	3	22	23	21	21	11	10
ULSAM	Men	409	701	139	17	4	56	55	56	56	55	54
	Women	-	-	-	-	-	-	-	-	-	-	-
	Pooled	409	701	139	17	4	56	55	56	56	55	54
Whitehall	Men	573	1126	276	42	5	85	85	85	85	85	85
	Women	198	337	127	46	18	27	27	27	27	27	27
	Pooled	771	1463	403	88	23	112	112	112	112	112	112
WTCCC T2D nonGWAS	Men	62	562	336	136	42	26	24	32	32	28	22
	Women	41	402	286	163	93	20	21	21	21	19	25
	Pooled	103	964	622	299	135	46	45	53	53	47	47

Supplementary Table 5. Number of individuals in each study for the tails and clinical class of obesity categories

Study name and case/control description		Controls N	Cases	Case/control definition
N Ancillary				
Essen Obesity Study (Essen Case-Control GWAS)	Men	171	192	Cases: early onset obese children and adolescent recruited in a specialized hospital; controls: healthy lean individuals
	Women	264	261	
	Pooled	435	453	
Essen Obesity Study (Essen Obesity Trio GWAS), Parents	Men	96	326	Biological parents of the offspring. Family-based association analysis has been used for this study
	Women	182	310	
	Pooled	278	636	
Essen Obesity Study (Essen Obesity Trio GWAS), Offspring	Men	0	318	Obese children and adolescent (≥ 90 th percentile, most ≥ 97 th percentile) recruited in a specialized hospital. Family-based association analysis has been used for this study
	Women	0	387	
	Pooled	0	705	
French Extreme Obesity	Men	485	435	Cases: early onset (< 6 yo) obese children and class III obese adults recruited in a specialized hospital; controls: lean adults and children
	Women	854	878	
	Pooled	1339	1313	
GEO-IT	Men	680	581	Cases: Class III obese adults recruited in a specialized hospital. Controls: lean adults
	Women	1112	1136	
	Pooled	1792	1717	
GOYA	Men	792	673	Cases: women with the 3.6% largest residuals after regressing BMI on age and parity. Men with BMI ≥ 31 kg/m ² , corresponding to >99th percentile. Controls: randomly selected subjects from the same populations
	Women	1948	1960	
	Pooled	2740	2633	

Supplementary Table 6. Results for all SNPs taken forward for replication that reached genome-wide significance ($P < 5 \times 10^{-8}$)

SNP	Chr	Position	Gene	Effect allele	Other allele	Effect allele freq	Stage 1				Stage 2				Stage 1 + Stage 2				Trait
							No. cases	No. controls	OR	P	No. cases	No. controls	OR	P	No. cases	No. controls	OR	P	
Novel loci^a																			
rs584438	17	35852698	<i>IGFBP4</i>	c	a	0.62	7830	7850	1.18	1.11E-09	1814	1814	1.19	1.20E-03	9644	9664	1.18	5.22E-12	Height tails
rs6662509	1	9240191	<i>H6PD</i>	t	c	0.15	5462	5461	1.23	2.21E-06	3615	3566	1.23	3.37E-05	9077	9027	1.23	3.19E-10	Height tails
rs2362965	3	159592073	<i>RSRC1/SHOX2</i>	t	a	0.50	7989	7993	1.14	1.45E-07	4819	4775	1.10	2.46E-03	12808	12768	1.12	2.14E-09	Height tails
rs1594829	8	26261994	<i>PPP2R2A</i>	c	t	0.77	6693	6697	1.18	5.51E-07	4166	4115	1.11	1.08E-02	10859	10812	1.15	3.88E-08	Height tails
rs7989336	13	95815549	<i>HS6ST3</i>	a	g	0.47	9825	62114	1.12	5.88E-09	1664	17113	1.04	2.49E-01	11489	79226	1.10	1.06E-08	Obesity class 2
rs17381664	1	77820919	<i>ZZZ3</i>	c	t	0.39	9833	62114	1.11	7.61E-08	5351	33841	1.05	3.60E-02	15184	95955	1.09	2.85E-08	Obesity class 2
rs17024258	1	109948844	<i>GNAT2</i>	t	c	0.04	18662	38427	1.23	1.41E-06	8956	15471	1.28	1.12E-06	27618	53898	1.25	8.66E-12	Obesity class 1
rs4735692	8	76778218	<i>HNFB4G</i>	a	g	0.58	32675	65697	1.07	5.03E-08	22086	38352	1.04	4.51E-03	54761	104049	1.06	2.48E-09	Obesity class 1
rs13041126	20	50526403	<i>MRPS33P4</i>	t	c	0.72	32020	64015	1.07	3.05E-07	22088	37595	1.04	7.03E-03	54108	101610	1.06	2.16E-08	Obesity class 1
rs2531995	16	3953468	<i>ADCY9</i>	t	c	0.61	32433	65542	1.06	3.17E-06	6680	16602	1.07	3.94E-03	39113	82144	1.07	4.04E-08	Obesity class 1
rs4735692	8	76778218	<i>HNFB4G</i>	a	g	0.58	92703	65698	1.05	6.13E-09	65323	39290	1.03	3.01E-03	158026	104988	1.04	3.51E-10	Overweight
rs7503807	17	76205706	<i>RPTOR</i>	a	c	0.57	92855	65723	1.04	4.20E-06	64535	38813	1.03	9.20E-04	157390	104536	1.04	1.98E-08	Overweight
Known loci																			
rs11075990	16	52377394	<i>FTO</i>	g	a	0.40	7744	7893	1.36	9.28E-33	4209	4204	1.35	2.39E-20	11953	12097	1.35	1.63E-51	BMI tails
rs8089364	18	56009809	<i>MC4R</i>	c	t	0.27	6802	6933	1.22	9.07E-11	4518	4503	1.27	5.59E-12	11320	11436	1.24	4.42E-21	BMI tails
rs2903492	2	614678	<i>TMEM18</i>	a	g	0.83	5893	6024	1.29	3.89E-11	3756	3745	1.21	1.67E-05	9649	9769	1.26	5.77E-15	BMI tails
rs2568958	1	72537704	<i>NEGR1</i>	a	g	0.60	7766	7915	1.18	6.84E-10	4900	4891	1.15	5.56E-06	12666	12806	1.16	2.26E-14	BMI tails
rs10938397	4	44877284	<i>GNPDA2</i>	g	a	0.43	7873	8017	1.15	1.54E-07	4862	4847	1.17	2.22E-07	12735	12865	1.16	1.76E-13	BMI tails
rs633715	1	176119203	<i>SEC16B</i>	c	t	0.19	6167	6296	1.24	3.37E-09	3604	3590	1.17	2.15E-04	9771	9886	1.21	4.90E-12	BMI tails
rs987237	6	50911009	<i>TFAP2B</i>	g	a	0.18	6160	6300	1.20	4.34E-07	3877	3874	1.21	1.25E-05	10037	10174	1.20	2.49E-11	BMI tails
rs2030323	11	27685115	<i>BDNF</i>	c	a	0.79	6248	6384	1.21	5.20E-08	4318	4304	1.13	1.23E-03	10566	10688	1.18	5.58E-10	BMI tails
rs7138803	12	48533735	<i>LOC144233</i>	a	g	0.38	7668	7810	1.14	5.83E-07	4718	4714	1.11	8.49E-04	12386	12524	1.13	2.37E-09	BMI tails
rs1516725	3	187306698	<i>ETV5</i>	c	t	0.86	5175	5327	1.30	2.06E-08	3265	3253	1.11	5.85E-02	8440	8580	1.21	3.65E-08	BMI tails
rs1991431	3	142616140	<i>ZBTB38</i>	a	g	0.44	8013	8031	1.33	4.33E-29	4869	4828	1.32	1.03E-19	12882	12859	1.33	3.93E-47	Height tails
rs224333	20	33487376	<i>GDF5</i>	a	g	0.36	7682	7697	1.33	7.50E-25	4664	4612	1.28	8.49E-14	12346	12309	1.31	8.17E-37	Height tails
rs1351394	12	64638093	<i>HMGA2</i>	t	c	0.49	7950	7960	1.28	5.95E-22	4872	4831	1.23	9.76E-12	12822	12791	1.26	6.59E-32	Height tails
rs42235	7	92086012	<i>CDK6</i>	t	c	0.30	7528	7541	1.31	9.37E-21	4672	4621	1.24	8.71E-10	12200	12162	1.28	1.36E-28	Height tails
rs806794	6	26308656	<i>HIST1H2BF</i>	a	g	0.70	7328	7347	1.25	6.10E-14	4701	4662	1.28	2.63E-13	12029	12009	1.26	1.31E-25	Height tails
rs3118906	13	50004789	<i>DLEU7</i>	g	a	0.72	7538	7560	1.29	6.26E-18	4539	4487	1.22	2.01E-08	12077	12047	1.26	1.51E-24	Height tails
rs11205303	1	148173037	<i>MTMR11</i>	c	t	0.38	7597	7612	1.26	3.81E-15	4656	4601	1.23	1.32E-09	12253	12213	1.25	3.72E-23	Height tails

Supplementary Table 6. Results for all SNPs taken forward for replication that reached genome-wide significance ($P < 5 \times 10^{-8}$)

SNP	Chr	Position	Gene	Effect allele	Other allele	Effect allele freq	Stage 1				Stage 2				Stage 1 + Stage 2				Trait
							No. cases	No. controls	OR	P	No. cases	No. controls	OR	P	No. cases	No. controls	OR	P	
rs798554	7	2726321	AMZ1	c	t	0.68	7531	7552	1.27	3.58E-17	4810	4769	1.20	6.35E-08	12341	12321	1.24	4.36E-23	Height tails
rs6845078	4	17561306	LCORL	c	t	0.85	5627	5628	1.34	4.90E-12	3483	3435	1.45	5.40E-12	9110	9063	1.38	3.16E-22	Height tails
rs7689420	4	145787802	HHIP	c	t	0.84	5842	5836	1.26	7.17E-09	3866	3815	1.44	1.16E-14	9708	9651	1.33	4.02E-21	Height tails
rs7741741	6	142697494	GPR126	t	a	0.72	7346	7370	1.22	1.28E-10	4640	4587	1.28	7.28E-12	11986	11957	1.24	1.04E-20	Height tails
rs17720281	4	145763226	HHIP	t	c	0.41	7875	7897	1.22	5.03E-13	4872	4831	1.19	1.39E-08	12747	12728	1.21	4.91E-20	Height tails
rs11259933	15	82371160	ADAMTSL3	a	g	0.51	8011	8017	1.17	9.48E-11	4821	4779	1.22	1.56E-10	12832	12796	1.19	1.39E-19	Height tails
rs2079795	17	56851431	C17orf82	t	c	0.32	7561	7585	1.18	1.44E-09	4838	4796	1.24	6.06E-11	12399	12381	1.20	1.08E-18	Height tails
rs1878528	4	82403258	PRKG2	g	a	0.33	7848	7861	1.19	7.75E-11	4808	4768	1.21	8.83E-09	12656	12629	1.20	4.21E-18	Height tails
rs2145270	20	6569685	BMP2	c	t	0.37	7516	7533	1.21	1.53E-11	4669	4618	1.20	5.86E-08	12185	12151	1.20	5.18E-18	Height tails
rs1885486	6	7640972	BMP6	a	g	0.48	8001	8007	1.18	1.08E-10	4904	4863	1.18	8.82E-08	12905	12870	1.18	5.01E-17	Height tails
rs1490384	6	126892853	C6orf173	t	c	0.49	7961	7971	1.19	8.16E-12	4872	4829	1.16	2.36E-06	12833	12800	1.18	1.20E-16	Height tails
rs9292468	5	32854830	C5orf23	t	c	0.40	7863	7881	1.18	1.80E-10	4903	4862	1.17	2.34E-07	12766	12743	1.18	2.11E-16	Height tails
rs7534091	1	118666139	SPAG17	a	g	0.74	6951	6935	1.20	7.06E-09	4409	4356	1.24	4.78E-09	11360	11291	1.22	2.47E-16	Height tails
rs11144688	9	77732106	PCSK5	g	a	0.89	4876	4884	1.46	7.03E-08	2360	2307	1.49	1.14E-09	7236	7191	1.48	4.27E-16	Height tails
rs4535251	3	173409067	FND3B	t	c	0.50	8011	8019	1.16	1.10E-08	4904	4863	1.20	5.34E-09	12915	12882	1.18	4.39E-16	Height tails
rs3791679	2	55950396	EFEMP1	a	g	0.77	6896	6882	1.22	8.19E-10	4525	4485	1.21	1.92E-07	11421	11367	1.22	7.81E-16	Height tails
rs8114671	20	33252803	PROCR	c	a	0.56	7671	7693	1.17	7.88E-10	4836	4794	1.17	2.75E-07	12507	12487	1.17	1.09E-15	Height tails
rs2280470	15	87196630	ACAN	a	g	0.33	7753	7773	1.18	3.01E-09	4865	4829	1.19	8.64E-08	12618	12602	1.18	1.45E-15	Height tails
rs11082304	18	18974971	CABLES1	t	g	0.50	8022	8025	1.15	1.31E-07	4327	4276	1.21	3.33E-09	12349	12301	1.18	4.75E-15	Height tails
rs1759645	6	34302844	HMGA1	c	t	0.16	5734	5719	1.27	5.25E-09	3610	3560	1.31	1.77E-07	9344	9279	1.29	5.23E-15	Height tails
rs2284746	1	17179262	MFAP2	g	c	0.52	7972	7980	1.16	1.03E-08	4732	4677	1.18	9.15E-08	12704	12657	1.17	5.37E-15	Height tails
rs10748128	12	68113925	FRS2	t	g	0.35	7564	7592	1.18	1.17E-08	4703	4650	1.20	1.06E-07	12267	12242	1.19	6.61E-15	Height tails
rs1074683	20	31768314	PXMP4	c	g	0.76	6819	6808	1.23	2.94E-10	4414	4371	1.18	8.21E-06	11233	11179	1.21	1.46E-14	Height tails
rs2941551	17	59332280	CSH1	g	a	0.27	7427	7425	1.21	1.85E-10	4634	4582	1.16	2.57E-05	12061	12008	1.19	3.16E-14	Height tails
rs1029534	7	28155608	JAZF1	t	g	0.31	7575	7595	1.17	2.54E-08	4417	4381	1.19	5.95E-07	11992	11976	1.18	8.19E-14	Height tails
rs3764419	17	26188149	ATAD5	c	a	0.61	7865	7881	1.16	1.51E-08	4840	4798	1.16	1.18E-06	12705	12679	1.16	8.95E-14	Height tails
rs11895026	2	24890362	CENPO, ADCY3, POMC	a	c	0.24	6933	6918	1.19	4.82E-08	4542	4500	1.20	5.68E-07	11475	11418	1.19	1.41E-13	Height tails
rs3814333	1	182273742	GLT25D2	t	c	0.33	7643	7639	1.17	5.88E-09	4607	4555	1.16	8.66E-06	12250	12194	1.17	2.34E-13	Height tails
rs9832740	3	72475831	RYBP	g	c	0.49	7989	7992	1.15	2.50E-08	4734	4681	1.16	4.46E-06	12723	12673	1.15	5.16E-13	Height tails
rs1415701	6	130387528	L3MBTL3	g	a	0.73	6830	6818	1.18	1.17E-08	3628	3579	1.20	9.25E-06	10458	10397	1.19	5.39E-13	Height tails
rs2093210	14	60027032	C14orf39	c	t	0.42	7769	7766	1.18	1.61E-09	4698	4644	1.14	7.10E-05	12467	12410	1.16	7.66E-13	Height tails

Supplementary Table 6. Results for all SNPs taken forward for replication that reached genome-wide significance ($P < 5 \times 10^{-8}$)

SNP	Chr	Position	Gene	Effect allele	Other allele	Effect allele freq	Stage 1				Stage 2				Stage 1 + Stage 2				Trait
							No. cases	No. controls	OR	P	No. cases	No. controls	OR	P	No. cases	No. controls	OR	P	
rs2744971	6	34689614	<i>C6orf106</i>	g	t	0.15	5909	5894	1.29	6.10E-10	3544	3492	1.21	1.91E-04	9453	9386	1.26	8.21E-13	Height tails
rs572169	3	173648421	<i>GHSR</i>	t	c	0.31	7617	7617	1.19	2.99E-10	4638	4586	1.13	3.91E-04	12255	12203	1.17	1.11E-12	Height tails
rs11880992	19	2127403	<i>DOT1L</i>	a	g	0.39	7711	7730	1.14	5.53E-07	4857	4816	1.17	5.12E-07	12568	12546	1.15	1.54E-12	Height tails
rs2564921	3	53100625	<i>RFT1</i>	t	c	0.43	7905	7921	1.16	1.07E-08	4725	4690	1.14	3.92E-05	12630	12611	1.15	2.06E-12	Height tails
rs2025151	9	98201333	<i>ZNF367</i>	g	c	0.18	6048	6037	1.23	6.82E-08	4150	4104	1.21	7.12E-06	10199	10141	1.22	2.28E-12	Height tails
rs2247056	6	31373469	<i>HLA-C</i>	t	c	0.27	7182	7171	1.21	1.50E-10	4280	4228	1.13	1.69E-03	11462	11399	1.18	3.61E-12	Height tails
rs12455370	18	19069221	<i>CABLES1</i>	t	c	0.74	6875	6860	1.18	3.32E-07	4493	4441	1.19	2.41E-06	11368	11301	1.19	3.65E-12	Height tails
rs10058074	5	131714045	<i>FLJ44796</i>	a	g	0.45	7845	7848	1.17	1.43E-09	4731	4693	1.12	4.36E-04	12576	12541	1.15	4.46E-12	Height tails
rs9391253	6	105474309	<i>LIN28B</i>	t	a	0.32	7565	7587	1.19	1.42E-10	4610	4557	1.11	2.03E-03	12175	12144	1.16	4.79E-12	Height tails
rs6141600	20	34175724	<i>EPB41L1</i>	c	t	0.28	7351	7376	1.20	2.87E-09	1715	1713	1.23	4.36E-04	9066	9089	1.21	5.63E-12	Height tails
rs10843164	12	28460981	<i>CCDC91</i>	t	c	0.68	7293	7314	1.16	2.64E-07	4740	4701	1.16	4.93E-06	12033	12015	1.16	5.84E-12	Height tails
rs10185077	2	56051616	<i>EFEMP1</i>	t	g	0.60	7765	7795	1.17	2.38E-09	4077	4023	1.13	3.97E-04	11842	11818	1.16	5.92E-12	Height tails
rs13428823	2	25226802	<i>POMC</i>	g	a	0.37	7925	7941	1.17	2.80E-09	1983	1995	1.18	5.07E-04	9908	9936	1.17	5.98E-12	Height tails
rs12075079	1	170486618	<i>DNM3</i>	g	a	0.21	6394	6385	1.20	1.34E-07	4183	4131	1.20	1.18E-05	10577	10516	1.20	7.09E-12	Height tails
rs10838798	11	48047879	<i>PTPRJ</i>	t	g	0.31	7595	7608	1.17	4.02E-08	4770	4731	1.15	3.78E-05	12365	12339	1.16	7.10E-12	Height tails
rs13210323	6	35113062	<i>ANKS1A</i>	a	c	0.73	7262	7288	1.19	1.12E-08	4475	4422	1.15	1.35E-04	11737	11710	1.17	7.59E-12	Height tails
rs1432559	2	56043122	<i>EFEMP1</i>	g	t	0.20	6185	6186	1.24	2.95E-09	4061	4013	1.17	4.07E-04	10246	10199	1.21	8.93E-12	Height tails
rs1950500	14	23900690	<i>NFATC4</i>	t	c	0.30	7360	7369	1.16	1.22E-07	4659	4620	1.16	2.25E-05	12019	11989	1.16	1.17E-11	Height tails
rs2856321	12	11747040	<i>ETV6</i>	g	a	0.37	7833	7850	1.17	1.17E-09	4810	4770	1.10	1.82E-03	12643	12620	1.14	2.66E-11	Height tails
rs17361789	1	170389224	<i>DNM3</i>	g	t	0.30	7292	7318	1.21	7.57E-10	1820	1829	1.15	1.19E-02	9112	9147	1.20	3.82E-11	Height tails
rs10948222	6	45352393	<i>SUPT3H</i>	c	t	0.60	7767	7795	1.16	1.67E-07	4225	4173	1.14	6.33E-05	11992	11968	1.15	4.54E-11	Height tails
rs7741091	6	31460610	<i>HLA-B</i>	a	g	0.68	7640	7664	1.16	6.75E-08	4720	4679	1.13	1.65E-04	12360	12343	1.15	5.49E-11	Height tails
rs10492321	12	92504219	<i>SOCS2</i>	a	t	0.21	6390	6377	1.27	9.44E-10	1070	1069	1.24	2.39E-02	7460	7446	1.26	7.07E-11	Height tails
rs310405	6	81857081	<i>FAM46A</i>	a	g	0.52	7964	7967	1.17	1.05E-09	4478	4429	1.09	8.16E-03	12443	12396	1.14	1.18E-10	Height tails
rs9388768	6	130415795	<i>L3MBTL3</i>	c	a	0.33	7664	7686	1.13	1.28E-06	4777	4738	1.15	2.15E-05	12441	12424	1.14	1.32E-10	Height tails
rs6450922	5	32725475	<i>NPR3</i>	c	g	0.75	6804	6801	1.24	4.27E-08	3743	3688	1.16	4.15E-04	10547	10488	1.20	1.46E-10	Height tails
rs7466269	9	132453905	<i>FUBP3</i>	a	g	0.64	7872	7889	1.13	2.33E-06	4785	4745	1.14	2.09E-05	12657	12634	1.14	2.10E-10	Height tails
rs1545552	2	33213842	<i>LTBP1</i>	g	a	0.72	7420	7440	1.17	2.62E-07	4738	4699	1.14	1.73E-04	12158	12139	1.15	2.33E-10	Height tails
rs718444	13	32045052	<i>APRN</i>	t	c	0.37	7878	7894	1.17	2.21E-09	4837	4795	1.09	7.22E-03	12715	12689	1.14	2.37E-10	Height tails
rs2844479	6	31680935	<i>AIF1</i>	a	c	0.66	7628	7642	1.16	2.01E-07	4734	4682	1.13	2.82E-04	12362	12324	1.14	2.68E-10	Height tails
rs153750	5	171113842	<i>FBXW11</i>	g	t	0.62	7827	7845	1.18	1.67E-09	1814	1814	1.11	4.12E-02	9641	9659	1.16	2.93E-10	Height tails

Supplementary Table 6. Results for all SNPs taken forward for replication that reached genome-wide significance ($P < 5 \times 10^{-8}$)

SNP	Chr	Position	Gene	Effect allele	Other allele	Effect allele freq	Stage 1				Stage 2				Stage 1 + Stage 2				Trait
							No. cases	No. controls	OR	P	No. cases	No. controls	OR	P	No. cases	No. controls	OR	P	
rs2982712	6	152399872	<i>ESR1</i>	c	t	0.47	7860	7874	1.17	1.33E-08	1814	1814	1.15	7.66E-03	9674	9688	1.17	3.66E-10	Height tails
rs6957923	7	23449210	<i>IGF2BP3</i>	g	a	0.63	7628	7645	1.16	5.63E-08	4513	4466	1.11	1.70E-03	12141	12111	1.14	5.39E-10	Height tails
rs4965593	15	98575908	<i>ADAMTS17</i>	c	g	0.32	7545	7558	1.16	1.00E-07	4477	4427	1.12	1.40E-03	12022	11985	1.15	7.24E-10	Height tails
rs526896	5	134384604	<i>PITX1</i>	t	g	0.72	7376	7374	1.16	6.00E-07	4775	4734	1.13	3.07E-04	12151	12108	1.15	8.62E-10	Height tails
rs9596219	13	49461448	<i>TRIM13</i>	t	c	0.25	6874	6865	1.19	4.93E-08	4500	4449	1.11	3.64E-03	11374	11314	1.16	1.44E-09	Height tails
rs10838708	11	47398089	<i>PSMC3</i>	g	a	0.53	7983	7987	1.17	9.93E-10	1899	1908	1.07	1.71E-01	9882	9895	1.15	1.52E-09	Height tails
rs780151	10	80601487	<i>ZMIZ1</i>	g	a	0.57	7957	7962	1.14	2.53E-07	4561	4520	1.11	1.43E-03	12518	12482	1.13	1.74E-09	Height tails
rs4075154	8	57371800	<i>RDHE2</i>	a	g	0.87	5206	5206	1.33	4.00E-09	1267	1266	1.16	1.39E-01	6473	6472	1.29	2.70E-09	Height tails
rs2322633	6	80902708	<i>BCKDHB</i>	t	c	0.50	7982	7987	1.14	1.94E-07	4900	4860	1.10	2.31E-03	12882	12847	1.12	2.72E-09	Height tails
rs10475992	5	171267898	<i>FBXW11</i>	t	c	0.52	7502	7506	1.20	8.93E-09	4673	4633	1.09	8.35E-03	12175	12139	1.14	2.97E-09	Height tails
rs11209718	1	41461525	<i>SCMH1</i>	t	c	0.44	7920	7924	1.14	2.22E-07	4817	4778	1.10	2.39E-03	12737	12702	1.12	3.09E-09	Height tails
rs10231759	7	150143105	<i>TMEM176A</i>	c	t	0.28	7307	7343	1.17	1.64E-08	1951	1963	1.10	5.66E-02	9258	9306	1.15	3.91E-09	Height tails
rs4953076	2	44334454	<i>PPM1B</i>	g	c	0.25	7116	7107	1.15	3.20E-06	4670	4631	1.14	3.35E-04	11786	11738	1.15	4.35E-09	Height tails
rs9967417	18	45213498	<i>DYM</i>	g	c	0.42	7667	7676	1.14	1.60E-07	4704	4651	1.09	5.22E-03	12371	12327	1.13	5.11E-09	Height tails
rs4337252	15	72013818	<i>LOXL1</i>	c	g	0.50	8049	8050	1.14	2.10E-07	1983	1995	1.13	9.32E-03	10032	10045	1.14	6.58E-09	Height tails
rs1724888	7	2707545	<i>AMZ1</i>	c	g	0.73	5348	5357	1.21	4.29E-06	4391	4337	1.15	4.06E-04	9739	9693	1.18	1.15E-08	Height tails
rs9285425	6	117978529	<i>DCBLD1</i>	g	a	0.50	7951	7952	1.13	1.02E-06	1983	1995	1.14	4.74E-03	9934	9947	1.14	1.60E-08	Height tails
rs6750795	2	232086475	<i>C2orf52</i>	t	c	0.44	7950	7969	1.13	1.06E-06	4903	4862	1.09	3.13E-03	12853	12831	1.12	1.73E-08	Height tails
rs6855629	4	106353765	<i>KIAA1546</i>	g	a	0.63	7791	7807	1.14	1.07E-06	1814	1814	1.15	5.36E-03	9605	9621	1.14	2.01E-08	Height tails
rs17511102	2	37814117	<i>CDC42EP3</i>	t	a	0.09	4104	4100	1.41	9.89E-08	708	709	1.33	8.78E-02	4812	4809	1.40	2.36E-08	Height tails
rs7552186	1	217061896	<i>LYPLAL1</i>	c	t	0.62	7801	7816	1.13	2.90E-06	4874	4833	1.10	2.30E-03	12675	12649	1.12	3.03E-08	Height tails
rs3103267	2	232696826	<i>DIS3L2</i>	c	a	0.72	7181	7201	1.17	1.49E-07	1814	1814	1.11	5.74E-02	8995	9015	1.16	3.19E-08	Height tails
rs8043757	16	52370951	<i>FTO</i>	t	a	0.40	32577	65746	1.24	2.44E-66	21792	37471	1.22	2.42E-45	54369	103217	1.23	4.98E-110	Obesity class 1
rs6711012	2	614034	<i>TMEM18</i>	c	g	0.82	32108	64305	1.19	3.18E-25	20696	35123	1.17	8.89E-17	52804	99428	1.18	2.96E-40	Obesity class 1
rs538656	18	56001402	<i>MC4R</i>	t	g	0.24	31746	63734	1.14	2.13E-19	21733	36568	1.15	1.02E-18	53480	100302	1.15	2.03E-36	Obesity class 1
rs10938397	4	44877284	<i>GNPDA2</i>	g	a	0.43	32789	65639	1.11	5.04E-16	22334	39032	1.13	3.91E-20	55123	104671	1.12	2.85E-34	Obesity class 1
rs633715	1	176119203	<i>SEC16B</i>	c	t	0.19	32294	64436	1.12	7.21E-13	18412	34561	1.13	1.78E-11	50706	98997	1.12	8.57E-23	Obesity class 1
rs2030323	11	27685115	<i>BDNF</i>	c	a	0.79	32222	64668	1.11	2.52E-12	21746	38028	1.12	1.61E-11	53968	102696	1.12	2.92E-22	Obesity class 1
rs2206277	6	50906485	<i>TFAP2B</i>	t	c	0.18	30821	63877	1.14	1.35E-16	20843	34865	1.10	2.29E-07	51664	98742	1.12	4.90E-22	Obesity class 1
rs7138803	12	48533735	<i>LOC144233</i>	a	g	0.38	32843	65810	1.09	2.61E-11	21795	37472	1.10	6.32E-11	54638	103282	1.09	9.67E-21	Obesity class 1
rs10182181	2	25003800	<i>ADCY3,POMC</i>	g	a	0.46	32834	65801	1.08	3.30E-09	22269	38897	1.09	4.99E-10	55103	104698	1.08	1.11E-17	Obesity class 1

Supplementary Table 6. Results for all SNPs taken forward for replication that reached genome-wide significance ($P < 5 \times 10^{-8}$)

SNP	Chr	Position	Gene	Effect allele	Other allele	Effect allele freq	Stage 1				Stage 2				Stage 1 + Stage 2				Trait
							No. cases	No. controls	OR	P	No. cases	No. controls	OR	P	No. cases	No. controls	OR	P	
rs7141420	14	78969207	<i>NRXN3</i>	t	c	0.52	32834	65789	1.08	2.34E-10	22371	39054	1.08	1.04E-08	55205	104843	1.08	1.27E-17	Obesity class 1
rs7531118	1	72609827	<i>NEGR1</i>	c	t	0.56	32812	65711	1.08	4.62E-10	21511	36320	1.09	6.90E-09	54323	102031	1.08	1.94E-17	Obesity class 1
rs10835211	11	27657941	<i>BDNF</i>	a	g	0.25	32641	64798	1.09	4.36E-09	21670	36999	1.08	4.45E-07	54311	101797	1.08	9.43E-15	Obesity class 1
rs1800437	19	50873232	<i>GIPR</i>	g	c	0.78	27534	58036	1.09	1.03E-06	21166	36441	1.11	3.94E-09	48700	94478	1.10	2.86E-14	Obesity class 1
rs9816226	3	187317193	<i>ETV5</i>	t	a	0.82	32057	64298	1.12	1.02E-11	20860	35337	1.07	6.36E-04	52917	99634	1.10	1.57E-13	Obesity class 1
rs7498665	16	28790742	<i>SH2B1</i>	g	a	0.40	32790	65703	1.07	6.05E-08	21547	37516	1.07	9.57E-07	54337	103219	1.07	2.50E-13	Obesity class 1
rs13333228	16	52351299	<i>FTO</i>	c	t	0.67	32813	65015	1.08	4.67E-08	22159	38017	1.07	2.75E-06	54971	103032	1.07	5.77E-13	Obesity class 1
rs8028313	15	65830111	<i>MAP2K5</i>	c	g	0.78	32321	64522	1.11	1.68E-11	21670	37028	1.06	1.13E-03	53991	101550	1.08	6.35E-13	Obesity class 1
rs2307111	5	75039434	<i>C5orf37</i>	t	c	0.60	32799	65689	1.07	3.35E-08	22367	39048	1.06	2.05E-05	55166	104737	1.07	3.39E-12	Obesity class 1
rs11042023	11	8619092	<i>RPL27A</i>	c	t	0.65	32855	65834	1.08	1.07E-08	22373	39060	1.05	1.59E-04	55228	104894	1.07	1.38E-11	Obesity class 1
rs12996547	2	592036	<i>TMEM18</i>	t	c	0.36	32852	65829	1.08	5.37E-09	21793	37473	1.05	8.33E-04	54645	103302	1.07	5.10E-11	Obesity class 1
rs887912	2	59156381	<i>FANCL</i>	t	c	0.28	32641	64784	1.09	2.32E-09	21766	37007	1.05	2.79E-03	54407	101791	1.07	1.22E-10	Obesity class 1
rs12446632	16	19842890	<i>GPRC5B</i>	g	a	0.86	31389	62749	1.10	7.70E-08	19994	34518	1.08	3.43E-04	51383	97267	1.09	1.50E-10	Obesity class 1
rs2817419	6	50920865	<i>TFAP2B</i>	a	g	0.74	31081	63970	1.08	5.25E-07	22106	37114	1.06	2.36E-04	53187	101084	1.07	6.89E-10	Obesity class 1
rs9302652	16	52423476	<i>FTO</i>	c	t	0.28	32267	63887	1.09	9.59E-10	21442	36520	1.04	2.16E-02	53709	100407	1.07	1.16E-09	Obesity class 1
rs11075986	16	52362845	<i>FTO</i>	c	g	0.92	27644	55775	1.15	4.28E-07	17700	30380	1.10	1.19E-03	45344	86155	1.12	3.54E-09	Obesity class 1
rs1514177	1	74763990	<i>TNNI3K</i>	c	g	0.43	32839	65817	1.06	9.01E-07	21512	36324	1.05	1.05E-03	54351	102141	1.06	4.65E-09	Obesity class 1
rs1879523	2	641599	<i>TMEM18</i>	t	a	0.33	32503	64582	1.06	3.93E-06	21831	37737	1.05	4.68E-04	54334	102319	1.06	7.77E-09	Obesity class 1
rs4438957	6	51253131	<i>TFAP2B</i>	c	g	0.25	30807	63684	1.08	1.24E-06	6353	15531	1.08	5.63E-03	37160	79215	1.08	2.24E-08	Obesity class 1
rs10968576	9	28404339	<i>LINGO2</i>	g	a	0.32	31375	64518	1.07	3.89E-07	6353	15531	1.06	2.75E-02	37728	80049	1.07	3.34E-08	Obesity class 1
rs7185735	16	52380152	<i>FTO</i>	g	a	0.40	9840	62087	1.33	1.34E-50	5312	32805	1.33	1.40E-30	15152	94892	1.33	1.39E-79	Obesity class 2
rs10189761	2	636364	<i>TMEM18</i>	a	t	0.82	9315	56184	1.26	5.04E-19	4447	25793	1.20	1.09E-06	13762	81977	1.24	6.12E-24	Obesity class 2
rs11152213	18	56003928	<i>MC4R</i>	c	a	0.24	9515	59270	1.19	6.68E-14	4855	27911	1.20	5.38E-10	14370	87181	1.19	2.65E-22	Obesity class 2
rs2207139	6	50953449	<i>TFAP2B</i>	g	a	0.18	8603	54397	1.19	8.18E-12	4755	27337	1.22	5.35E-09	13358	81734	1.20	2.72E-19	Obesity class 2
rs633715	1	176119203	<i>SEC16B</i>	c	t	0.19	9384	56080	1.18	2.65E-11	4153	27392	1.21	1.82E-09	13538	83472	1.19	4.00E-19	Obesity class 2
rs13130484	4	44870448	<i>GNPDA2</i>	t	c	0.43	9830	61932	1.13	1.58E-09	5475	35421	1.16	1.78E-10	15305	97353	1.14	2.84E-18	Obesity class 2
rs7138803	12	48533735	<i>LOC144233</i>	a	g	0.38	9818	61723	1.14	2.03E-11	5352	33839	1.13	1.00E-06	15170	95562	1.14	1.23E-16	Obesity class 2
rs3101336	1	72523773	<i>NEGR1</i>	c	t	0.61	9808	61868	1.12	5.42E-09	5474	35427	1.12	4.20E-06	15282	97295	1.12	1.17E-13	Obesity class 2
rs10423928	19	50874144	<i>GIPR</i>	t	a	0.77	8422	53974	1.14	1.12E-06	4859	28377	1.20	3.07E-08	13281	82351	1.16	3.58E-13	Obesity class 2
rs9956279	18	56093779	<i>MC4R</i>	t	c	0.31	9618	60277	1.13	6.33E-09	4520	24736	1.13	2.93E-05	14138	85013	1.13	7.99E-13	Obesity class 2
rs2030323	11	27685115	<i>BDNF</i>	c	a	0.79	9419	57138	1.14	5.21E-08	5174	31604	1.12	2.33E-04	14593	88742	1.13	5.89E-11	Obesity class 2

Supplementary Table 6. Results for all SNPs taken forward for replication that reached genome-wide significance ($P < 5 \times 10^{-8}$)

SNP	Chr	Position	Gene	Effect allele	Other allele	Effect allele freq	Stage 1				Stage 2				Stage 1 + Stage 2				Trait
							No. cases	No. controls	OR	P	No. cases	No. controls	OR	P	No. cases	No. controls	OR	P	
rs1412239	9	28415515	LINGO2	g	c	0.32	9054	60650	1.10	1.63E-06	5269	32150	1.11	2.76E-05	14323	92800	1.11	1.92E-10	Obesity class 2
rs2817419	6	50920865	TFAP2B	a	g	0.74	8894	58222	1.13	8.29E-08	5343	33634	1.10	8.03E-04	14237	91856	1.12	3.78E-10	Obesity class 2
rs9568867	13	53005353	OLFM4	a	g	0.13	8975	52803	1.14	7.52E-06	4433	26219	1.17	5.25E-05	13408	79021	1.15	1.71E-09	Obesity class 2
rs1516725	3	187306698	ETV5	c	t	0.86	8886	52129	1.21	1.17E-10	4504	26646	1.05	2.06E-01	13390	78775	1.15	2.68E-09	Obesity class 2
rs9302652	16	52423476	FTO	c	t	0.28	9708	60599	1.12	1.15E-07	5219	32045	1.08	5.36E-03	14927	92644	1.10	3.89E-09	Obesity class 2
rs2112347	5	75050998	C5orf37	t	g	0.63	9839	62112	1.11	3.68E-07	5475	35430	1.08	2.09E-03	15314	97542	1.09	3.95E-09	Obesity class 2
rs11639988	16	19851864	GPRC5B	a	g	0.86	9043	53231	1.19	8.29E-09	4459	25840	1.09	4.00E-02	13502	79071	1.15	4.02E-09	Obesity class 2
rs887912	2	59156381	FANCL	t	c	0.28	9750	61125	1.11	9.93E-07	5219	32050	1.09	1.66E-03	14969	93175	1.10	6.46E-09	Obesity class 2
rs7184597	16	28829310	RABEP2,SH2B1	t	c	0.33	9702	60757	1.12	5.11E-07	5351	33836	1.08	2.42E-03	15053	94593	1.10	6.77E-09	Obesity class 2
rs1421085	16	52358455	FTO	c	t	0.41	2825	44703	1.45	3.93E-26	1162	22307	1.47	2.11E-14	3986	67010	1.45	6.25E-39	Obesity class 3
rs1558902	16	52361075	FTO	a	t	0.41	92910	65769	1.15	1.53E-49	63198	37852	1.13	3.71E-34	156108	103621	1.14	1.84E-81	Overweight
rs6711012	2	614034	TMEM18	c	g	0.82	91720	65561	1.12	3.54E-21	61797	37446	1.11	1.43E-15	153517	103007	1.11	5.91E-35	Overweight
rs13130484	4	44870448	GNPDA2	t	c	0.43	92824	65636	1.07	3.95E-14	65326	39285	1.08	1.30E-15	158150	104921	1.08	4.10E-28	Overweight
rs10871777	18	56002743	MC4R	g	a	0.24	90380	64984	1.10	2.35E-20	22395	16437	1.11	1.46E-08	112775	81421	1.10	1.67E-27	Overweight
rs633715	1	176119203	SEC16B	c	t	0.20	92101	65685	1.08	4.05E-12	56249	35297	1.08	2.86E-09	148350	100982	1.08	6.78E-20	Overweight
rs2030323	11	27685115	BDNF	c	a	0.79	91950	65670	1.08	1.15E-12	64289	39168	1.06	2.94E-06	156238	104838	1.07	5.40E-17	Overweight
rs2568958	1	72537704	NEGR1	a	g	0.61	92598	65803	1.06	1.05E-11	65307	39283	1.05	2.96E-06	157905	105086	1.06	3.57E-16	Overweight
rs2206277	6	50906485	TFAP2B	t	c	0.18	89774	65154	1.08	5.58E-12	61253	36486	1.06	1.32E-05	151027	101640	1.07	6.61E-16	Overweight
rs9816226	3	187317193	ETV5	t	a	0.82	91789	65612	1.07	2.02E-09	61133	36478	1.07	2.02E-06	152921	102089	1.07	1.81E-14	Overweight
rs10182181	2	25003800	ADCY3,POMC	g	a	0.46	92951	65801	1.06	2.09E-10	64962	39128	1.04	1.38E-05	157913	104929	1.05	3.04E-14	Overweight
rs12446554	16	19842574	GPRC5B	g	t	0.86	91280	65564	1.08	1.95E-09	60393	36469	1.06	2.36E-04	151673	102033	1.07	3.69E-12	Overweight
rs7498665	16	28790742	SH2B1	g	a	0.40	92810	65704	1.05	5.39E-08	62819	37694	1.04	1.57E-05	155629	103398	1.05	4.74E-12	Overweight
rs8028313	15	65830111	MAP2K5	c	g	0.79	92150	65726	1.07	2.02E-09	63530	37893	1.04	5.11E-04	155680	103619	1.06	1.42E-11	Overweight
rs10835211	11	27657941	BDNF	a	g	0.25	92601	65800	1.06	1.41E-07	63831	37271	1.05	2.52E-05	156432	103071	1.05	1.74E-11	Overweight
rs13078807	3	85966840	CADM2	g	a	0.20	92163	65726	1.06	1.11E-07	61529	37385	1.05	6.44E-05	153692	103111	1.06	3.47E-11	Overweight
rs10875976	12	48512734	LOC144233	a	g	0.49	92859	65716	1.05	7.09E-07	63290	37708	1.04	6.50E-05	156149	103424	1.04	2.30E-10	Overweight
rs4615388	6	51254979	TFAP2B	a	t	0.25	89366	64757	1.06	4.27E-08	22395	16440	1.06	3.87E-03	111762	81197	1.06	6.47E-10	Overweight
rs12996547	2	592036	TMEM18	t	c	0.36	92997	65830	1.05	6.16E-08	63285	37704	1.03	1.91E-03	156282	103534	1.04	1.04E-09	Overweight
rs6731302	2	58686997	FANCL	a	g	0.44	92994	65828	1.04	1.12E-06	63776	38748	1.04	3.99E-04	156769	104576	1.04	2.48E-09	Overweight
rs2370983	14	78973129	NRXN3	a	g	0.62	92992	65815	1.04	2.26E-06	63269	37700	1.04	3.31E-04	156260	103515	1.04	3.31E-09	Overweight
rs9947301	18	56020570	MC4R	c	t	0.90	87722	64194	1.08	7.16E-07	54330	32704	1.06	1.72E-03	142052	96898	1.08	5.82E-09	Overweight

Supplementary Table 6. Results for all SNPs taken forward for replication that reached genome-wide significance ($P < 5 \times 10^{-8}$)

SNP	Chr	Position	Gene	Effect allele	Other allele	Effect allele freq	Stage 1				Stage 2				Stage 1 + Stage 2				Trait
							No. cases	No. controls	OR	P	No. cases	No. controls	OR	P	No. cases	No. controls	OR	P	
rs1514174	1	74765651	<i>TNNI3K</i>	c	t	0.44	92962	65797	1.05	1.13E-07	63258	37699	1.03	7.28E-03	156219	103496	1.04	1.07E-08	Overweight
rs2112347	5	75050998	<i>C5orf37</i>	t	g	0.63	92974	65807	1.05	2.87E-07	65333	39294	1.03	4.46E-03	158306	105101	1.04	1.25E-08	Overweight
rs1294421	6	6688148	<i>LY86</i>	g	t	0.62	4556	5247	1.21	1.62E-09	3351	3352	1.10	6.85E-03	7907	8599	1.16	2.19E-10	WHR tails
rs7745274	6	127550850	<i>RSPO3</i>	a	g	0.55	4683	5394	1.15	3.91E-06	3122	3128	1.16	5.33E-05	7805	8522	1.15	8.80E-10	WHR tails
rs2820464	1	217759843	<i>LYPLAL1</i>	g	a	0.66	4384	5074	1.23	4.54E-10	3089	3088	1.06	1.16E-01	7473	8162	1.16	7.35E-09	WHR tails
rs13389219	2	165237122	<i>COBLL1</i>	c	t	0.57	4584	5296	1.14	2.68E-06	3385	3387	1.11	3.09E-03	7969	8683	1.13	3.24E-08	WHR tails

^aLoci were considered novel if > 1 Mb from a SNP previously reported to be associated with the trait.

Supplementary Table 7. Variance explained^a by newly discovered and previously discovered SNPs for BMI and height.

	Twingene	LifeLines	Meta-analysis
	BMI		
32 previously identified SNPs	1.03%	1.11%	1.07%
32 previously identified SNPs + 7 new SNPs	1.06%	1.24%	1.16%
	Height		
180 previously identified SNPs	7.77%	6.98%	7.33%
180 previously identified SNPs + 4 new SNPs	7.77%	7.00%	7.35%

^a Variance explained is calculated as the improvement in R² over a base model.

Supplementary Table 8. Association results for novel genome-wide significant SNPs ($P < 5 \times 10^{-8}$) and cis gene expression (cis-eqtl)

SNP	Ch r	Position	Trait increasing allele	Other allele	Tissue	Gene	Effect direction	P for Giant SNP	P _{adj} for Giant SNP	Peak SNP	r ²	P for peak SNP	P _{adj} for peak SNP	Trait
<i>Novel loci</i>														
rs584438	17	35852698	c	a	Blood ¹	<i>TNS4</i>	+	2.50E-06	1	rs584438	1	2.50E-06	1	Height tails
rs2362965	3	159592073	t	a	Omental fat	<i>Contig42711_RC</i>	+	3.87E-12	0.002	rs7624161	0.07	1.27E-63	2.44E-50	Height tails
rs2362965	3	159592073	t	a	Lymphocyte	<i>GFM1</i>	-	4.21E-05	0.18	rs17642107	0.01	7.64E-26	1.68E-21	Height tails
rs2362965	3	159592073	t	a	Subcutaneous fat	<i>LXN</i>	-	2.13E-06	0.45	rs4680449	0.29	1.83E-27	2.19E-16	Height tails
rs2362965	3	159592073	t	a	Omental fat	<i>RARRES1</i>	-	1.39E-06	0.04	rs7646881	0.12	8.95E-15	3.07E-09	Height tails
rs2362965	3	159592073	t	a	Lymphocyte	<i>RSRC1</i>	+	1.72E-08	4.40E-05	rs827183	0.07	9.91E-22	7.90E-19	Height tails
rs17024258	1	109948844	t	c	Blood ¹	<i>GSTM1L</i>	+	7.30E-07	0.07	rs366631	0.06	4.30E-22	6.10E-17	Obesity class 1
rs17024258	1	109948844	t	c	Adipose	<i>GSTM3</i>	+	3.80E-11	0.01	rs11101992	0.08	6.10E-94	3.90E-80	Obesity class 1
rs17024258	1	109948844	t	c	Blood ¹	<i>GSTM3</i>	+	3.60E-08	0.002	rs4970777	0.03	2.60E-75	2.40E-106	Obesity class 1
rs2531995	16	3953468	t	c	Omental fat	<i>ADCY9</i>	+	5.83E-40	1	rs2531995	1	5.83E-40	1	Obesity class 1
rs2531995	16	3953468	t	c	Subcutaneous fat	<i>ADCY9</i>	+	4.66E-33	1	rs2531995	1	4.66E-33	1	Obesity class 1
rs2531995	16	3953468	t	c	Adipose	<i>ADCY9</i>	+	7.10E-28	2.00E-20	rs2601788	0.10	9.20E-33	4.60E-25	Obesity class 1
rs2531995	16	3953468	t	c	Liver	<i>ADCY9</i>	+	7.67E-24	1	rs2531995	1	7.67E-24	1	Obesity class 1
rs2531995	16	3953468	t	c	Blood ¹	<i>ADCY9</i>	+	3.70E-20	3.20E-12	rs2601788	0.10	8.80E-26	1.40E-17	Obesity class 1
rs17381664	1	77820919	c	t	Liver	<i>GIPC2</i>	-	4.88E-09	0.33	rs526723	0.13	1.66E-65	7.77E-49	Obesity class 2
rs17381664	1	77820919	c	t	Omental fat	<i>GIPC2</i>	+	3.67E-08	0.76	rs501774	0.15	2.46E-52	2.45E-37	Obesity class 2
rs17381664	1	77820919	c	t	Subcutaneous fat	<i>GIPC2</i>	+	2.54E-07	0.30	rs513977	0.12	1.79E-41	1.27E-30	Obesity class 2
rs17381664	1	77820919	c	t	Lymphocyte	<i>NEXN</i>	-	5.04E-07	0.54	rs6699769	0.19	1.73E-26	9.67E-20	Obesity class 2
rs7503807	17	76205706	a	c	Blood ¹	<i>NPTX1</i>	+	8.60E-06	1	rs9907193	0.18	8.20E-14	3.50E-09	Overweight

¹Results from DeCode

²Results from MolOBB

Supplementary Table 9. Association results for BMI tails and clinical classes of obesity for previously identified loci for adult body mass index

SNP	Gene	Published associated trait	Reference	Effect Other		BMI tails		Obesity class 3		Obesity class 2		Obesity class 1		Overweight	
				allele	allele	OR	P	OR	P	OR	P	OR	P	OR	P
rs1558902	<i>FTO</i>	Adult BMI	Speliotes et al. Nat Genet 2010	a	t	1.35	1.95E-51	1.45	5.86E-39	1.34	3.99E-83	1.24	1.85E-114	1.14	1.84E-81
rs2815752	<i>NEGR1</i>	Adult BMI	Speliotes et al. Nat Genet 2010	a	g	1.17	1.19E-14	1.19	9.56E-09	1.13	5.93E-14	1.08	1.77E-15	1.06	3.55E-16
rs2867125	<i>TMEM18</i>	Adult BMI	Speliotes et al. Nat Genet 2010	c	t	1.27	1.34E-14	1.25	1.46E-06	1.25	5.01E-24	1.18	8.85E-38	1.11	1.06E-31
rs571312	<i>MC4R</i>	Adult BMI	Speliotes et al. Nat Genet 2010	a	c	1.22	1.62E-14	1.21	4.72E-07	1.18	5.47E-20	1.14	1.21E-30	1.10	9.83E-32
rs10938397	<i>GNPDA2</i>	Adult BMI	Speliotes et al. Nat Genet 2010	g	a	1.16	1.76E-13	1.20	5.97E-10	1.14	3.64E-18	1.12	2.85E-34	1.08	3.19E-28
rs543874	<i>SEC16B</i>	Adult BMI	Speliotes et al. Nat Genet 2010	g	a	1.22	5.39E-12	1.23	8.52E-07	1.17	1.41E-15	1.12	1.06E-20	1.08	8.40E-17
rs987237	<i>TFAP2B</i>	Adult BMI	Speliotes et al. Nat Genet 2010	g	a	1.20	2.49E-11	1.29	1.22E-08	1.19	1.41E-18	1.12	6.07E-22	1.07	2.19E-16
rs3810291	<i>TMEM160</i>	Adult BMI	Speliotes et al. Nat Genet 2010	a	g	1.17	3.68E-10	1.12	0.001	1.10	1.01E-07	1.07	1.74E-09	1.04	8.93E-08
rs10767664	<i>BDNF</i>	Adult BMI	Speliotes et al. Nat Genet 2010	a	t	1.19	1.85E-09	1.10	0.02	1.15	4.08E-11	1.12	2.89E-18	1.08	4.69E-16
rs7138803	<i>FAIM</i>	Adult BMI	Speliotes et al. Nat Genet 2010	a	g	1.13	2.37E-09	1.17	4.03E-08	1.14	1.23E-16	1.09	9.67E-21	1.05	1.53E-12
rs9816226	<i>ETV5</i>	Adult BMI	Speliotes et al. Nat Genet 2010	t	a	1.18	1.25E-08	1.15	0.001	1.13	9.45E-09	1.10	1.57E-13	1.07	1.81E-14
rs2287019	<i>QPCTL</i>	Adult BMI	Speliotes et al. Nat Genet 2010	c	t	1.19	1.72E-08	1.22	1.65E-05	1.16	4.26E-11	1.10	1.58E-12	1.05	3.00E-08
rs3817334	<i>MTCH2</i>	Adult BMI	Speliotes et al. Nat Genet 2010	t	c	1.12	4.27E-08	1.09	0.003	1.07	8.97E-06	1.06	1.14E-10	1.05	3.73E-10
rs713586	<i>RBJ</i>	Adult BMI	Speliotes et al. Nat Genet 2010	c	t	1.13	3.62E-07	1.11	0.001	1.09	1.84E-06	1.07	1.68E-09	1.05	4.65E-10
rs7359397	<i>SH2B1</i>	Adult BMI	Speliotes et al. Nat Genet 2010	t	c	1.11	4.17E-07	1.08	0.006	1.08	3.88E-07	1.06	5.57E-11	1.05	1.01E-10
rs12444979	<i>GPRC5B</i>	Adult BMI	Speliotes et al. Nat Genet 2010	c	t	1.19	1.95E-06	1.19	0.0015	1.16	1.66E-09	1.09	2.89E-09	1.07	4.50E-11
rs2241423	<i>MAP2K5</i>	Adult BMI	Speliotes et al. Nat Genet 2010	g	a	1.13	1.07E-05	1.15	0.0004	1.13	6.71E-10	1.09	5.08E-15	1.06	1.42E-13
rs10968576	<i>LRRN6C</i>	Adult BMI	Speliotes et al. Nat Genet 2010	g	a	1.11	1.22E-05	1.12	0.001	1.10	2.89E-07	1.07	3.34E-08	1.04	1.39E-06
rs1514175	<i>TNNI3K</i>	Adult BMI	Speliotes et al. Nat Genet 2010	a	g	1.11	1.49E-05	1.06	0.07	1.05	0.01	1.05	5.86E-06	1.04	1.34E-06
rs4929949	<i>RPL27A</i>	Adult BMI	Speliotes et al. Nat Genet 2010	c	t	1.09	1.52E-05	1.09	0.005	1.07	1.80E-05	1.05	1.33E-07	1.03	4.16E-05
rs1555543	<i>PTBP2</i>	Adult BMI	Speliotes et al. Nat Genet 2010	c	a	1.11	2.83E-05	1.08	0.02	1.04	0.02	1.05	1.85E-05	1.03	0.0001
rs2890652	<i>LRP1B</i>	Adult BMI	Speliotes et al. Nat Genet 2010	c	t	1.15	0.0001	1.17	0.002	1.07	0.004	1.07	7.07E-06	1.04	0.0003
rs29941	<i>KCTD15</i>	Adult BMI	Speliotes et al. Nat Genet 2010	g	a	1.09	0.0001	1.10	0.002	1.09	6.47E-07	1.05	1.75E-07	1.03	2.97E-05
rs2112347	<i>FLJ35779</i>	Adult BMI	Speliotes et al. Nat Genet 2010	t	g	1.08	0.0002	1.11	0.0004	1.09	3.95E-09	1.07	2.88E-13	1.04	1.25E-08
rs887912	<i>FANCL</i>	Adult BMI	Speliotes et al. Nat Genet 2010	t	c	1.09	0.0002	1.13	0.0003	1.10	6.46E-09	1.07	1.22E-10	1.04	2.39E-08
rs13078807	<i>CADM2</i>	Adult BMI	Speliotes et al. Nat Genet 2010	g	a	1.10	0.0005	1.08	0.07	1.10	5.24E-07	1.06	1.10E-07	1.06	3.47E-11
rs206936	<i>NUDT3</i>	Adult BMI	Speliotes et al. Nat Genet 2010	g	a	1.10	0.0006	1.09	0.03	1.04	0.03	1.04	0.0005	1.03	0.0004
rs7227255	<i>MC4R</i>	Adult BMI	Speliotes et al. Nat Genet 2010	g	a	1.48	0.007	1.10	0.62	1.64	2.87E-06	1.30	8.37E-07	1.16	3.80E-07
rs4836133	<i>ZNF608</i>	Adult BMI	Speliotes et al. Nat Genet 2010	a	c	1.07	0.004	1.06	0.14	1.05	0.02	1.05	4.59E-05	1.04	6.26E-05
rs10150332	<i>NRXN3</i>	Adult BMI	Speliotes et al. Nat Genet 2010	c	t	1.09	0.004	1.12	0.007	1.10	1.70E-05	1.08	3.15E-08	1.04	2.10E-05
rs4771122	<i>MTIF3</i>	Adult BMI	Speliotes et al. Nat Genet 2010	g	a	1.07	0.03	1.10	0.02	1.09	3.43E-05	1.06	5.83E-06	1.04	5.48E-05
rs13107325	<i>SLC39A8</i>	Adult BMI	Speliotes et al. Nat Genet 2010	t	c	1.17	0.05	1.29	0.005	1.22	2.42E-05	1.12	1.65E-06	1.09	6.28E-07
rs11847697	<i>PRKD1</i>	Adult BMI	Speliotes et al. Nat Genet 2010	t	c	1.15	0.16	1.01	0.94	1.22	0.002	1.15	8.76E-06	1.09	1.46E-05

Supplementary Table 10. Association results for all novel genome-wide significant ($P < 5 \times 10^{-8}$) obesity-related SNPs in studies of extremely obese with different ascertainment strategies

SNP	Chr	Position	Gene	Trait increasing allele	Other allele	Extremely obese studies (case-control)				Extremely obese studies (case-control and family-based)				Trait
						No. cases	No. controls	OR	P	No. cases	No. controls	Effect direction	P	
rs7989336	13	95815549	<i>HS6ST3</i>	a	g	6088	6279	1.10	3.79E-04	6787	6979	+	0.005	Obesity class 2
rs17381664	1	77820919	<i>ZZZ3</i>	c	t	6084	6257	1.01	0.64	6682	6852	+	0.23	Obesity class 2
rs17024258	1	109948844	<i>GNAT2</i>	t	c	6103	6299	1.06	0.44	6795	6997	+	0.25	Obesity class 1
rs4735692	8	76778218	<i>HNF4G</i>	a	g	6101	6291	1.07	0.01	6745	6929	+	0.006	Obesity class 1
rs13041126	20	50526403	<i>MRPS33P4</i>	t	c	6124	6314	1.06	0.05	6829	7019	+	0.02	Obesity class 1
rs2531995	16	3953468	<i>ADCY9</i>	t	c	6130	6310	1.10	8.06E-04	6825	7006	+	6.28E-04	Obesity class 1
rs4735692	8	76778218	<i>HNF4G</i>	a	g	6101	6291	1.07	0.01	6745	6929	+	0.006	Overweight
rs7503807	17	76205706	<i>RPTOR</i>	a	c	6137	6318	1.09	0.002	6842	7022	+	0.001	Overweight

Supplementary Table 11. Association results for BMI tails and clinical classes of obesity for published loci for extreme childhood and adult obesity

SNP	Gene	Published associated trait	Reference	Effect	Other	BMI tails		Obesity Class 3		Obesity Class 2		Obesity Class 1		Overweight	
				allele	allele	OR	P	OR	P	OR	P	OR	P	OR	P
rs1421085	<i>FTO</i>	extreme childhood obesity, extreme adult obesity	Dina et al. Nat Genet 2007	c	t	1.35	6.48E-52	1.45	6.25E-39	1.34	2.06E-83	1.24	6.65E-113	1.14	5.50E-82
rs17782313	<i>MC4R</i>	extreme childhood obesity, extreme adult obesity	Loos et al., Nat Genet 2008	c	t	1.20	4.67E-13	1.20	2.18E-06	1.19	1.56E-20	1.15	3.24E-32	1.11	9.12E-34
rs6235	<i>PCSK1</i>	extreme childhood obesity, extreme adult obesity	Benzinou et al., Nat Genet 2008	c	g	1.05	0.06	1.12	0.002	1.08	1.84E-04	1.05	2.52E-04	1.03	3.76E-04
rs6232	<i>PCSK1</i>	extreme childhood obesity, extreme adult obesity	Benzinou et al., Nat Genet 2008	c	t	0.998	0.98	1.13	0.18	1.10	0.06	1.06	0.05	1.04	0.07
rs1424233	<i>MAF</i>	extreme childhood obesity, extreme adult obesity	Meyre et al., Nat Genet 2009	t	c	0.996	0.86	1.01	0.85	0.996	0.81	1.01	0.25	1.02	0.06
rs1805081	<i>NPC1</i>	extreme childhood obesity, extreme adult obesity	Meyre et al., Nat Genet 2009	t	c	1.07	6.48E-04	1.07	0.02	1.05	0.004	1.04	8.05E-05	1.02	0.002
rs10508503	<i>PTER</i>	extreme childhood obesity, extreme adult obesity	Meyre et al., Nat Genet 2009	c	t	1.04	0.52	1.02	0.74	1.03	0.36	1.04	0.04	1.02	0.13
rs12145833	<i>SDCCAG8</i>	childhood extreme obesity	Scherag et al. PLOS Genet 2010	t	g	1.03	0.48	1.04	0.42	1.06	0.02	1.03	0.09	1.03	0.01
rs17150703	<i>TNKS</i>	childhood extreme obesity	Scherag et al. PLOS Genet 2010	a	g	1.11	0.04	1.09	0.18	1.07	0.04	1.03	0.11	1.02	0.16
rs6548238	<i>TMEM18</i>	childhood extreme obesity	Scherag et al. PLOS Genet 2010	c	t	1.27	1.10E-13	1.26	2.09E-06	1.26	7.39E-25	1.18	4.29E-36	1.12	1.38E-32
rs7132908	<i>FAIM2</i>	Severe young adulthood obesity	Paternoster et al., PLOS One 2011	a	g	1.15	9.23E-07	1.19	5.54E-06	1.13	1.49E-08	1.11	5.62E-15	1.06	4.04E-09
rs2116830	<i>KCNMA1</i>	extreme adult obesity	Jiao et al. BMC Medical Genomics 2011	g	t	1.03	0.40	1.03	0.63	1.02	0.53	1.01	0.43	1.01	0.38
rs9299	<i>HOXB5</i>	childhood obesity	Bradfield et al. Nat Genet 2012	t	c	1.05	0.049	1.01	0.85	1.02	0.32	1.04	0.004	1.02	0.02
rs9568856	<i>OLFM4</i>	childhood obesity	Bradfield et al. Nat Genet 2012	a	g	1.14	0.0035	1.08	0.19	1.12	1.53E-04	1.06	0.002	1.04	8.93E-04

Supplementary Table 12. Differences between the effect sizes observed in the tails for BMI and those expected based on the overall distribution

SNP	Gene	Effective N	Beta difference ^a	Standard error difference	P-value for difference
Previously published SNPs associated with BMI in GIANT					
rs1558902	<i>FTO</i>	5446	0.059	0.024	0.015
rs2815752	<i>NEGR1</i>	5421	-0.059	0.025	0.016
rs10938397	<i>GNPDA2</i>	5538	-0.058	0.025	0.018
rs10767664	<i>BDNF</i>	4344	-0.078	0.033	0.019
rs2112347	<i>FLJ35779</i>	5446	0.054	0.025	0.029
rs4771122	<i>MTIF3</i>	4444	-0.064	0.031	0.041
rs13078807	<i>CADM2</i>	4184	-0.062	0.033	0.062
rs887912	<i>FANCL</i>	4974	0.048	0.027	0.080
rs3817334	<i>MTCH2</i>	5445	-0.036	0.024	0.135
rs12444979	<i>GPRC5B</i>	3547	0.063	0.044	0.151
rs571312	<i>MC4R</i>	4205	0.044	0.031	0.162
rs4929949	<i>RPL27A</i>	5612	0.027	0.024	0.253
rs2241423	<i>MAP2K5</i>	4483	-0.029	0.032	0.355
rs2287019	<i>QPCTL</i>	3600	-0.035	0.039	0.368
rs29941	<i>KCTD15</i>	5134	-0.023	0.026	0.382
rs4836133	<i>ZNF608</i>	5610	-0.021	0.024	0.387
rs13107325	<i>SLC39A8</i>	2320	-0.074	0.093	0.428
rs11847697	<i>PRKD1</i>	1771	0.075	0.103	0.467
rs9816226	<i>ETV5</i>	4198	-0.024	0.035	0.494
rs10150332	<i>NRXN3</i>	4341	-0.022	0.032	0.504
rs1514175	<i>TNNI3K</i>	5520	0.013	0.024	0.573
rs543874	<i>SEC16B</i>	4190	0.019	0.034	0.580
rs3810291	<i>TMEM160</i>	4930	-0.012	0.028	0.669
rs1555543	<i>PTBP2</i>	5440	0.010	0.024	0.675
rs7359397	<i>SH2B1</i>	5498	-0.010	0.024	0.692
rs206936	<i>NUDT3</i>	4217	0.008	0.034	0.803
rs7138803	<i>FAIM</i>	5392	0.005	0.025	0.826
rs2867125	<i>TMEM18</i>	4078	-0.006	0.037	0.862
rs10968576	<i>LRRN6C</i>	5217	-0.003	0.026	0.901
rs987237	<i>TFAP2B</i>	4141	-0.003	0.035	0.935
rs713586	<i>RBJ</i>	5517	0.002	0.024	0.937
rs2890652	<i>LRP1B</i>	3872	-0.003	0.037	0.942
Previously published SNPs for association with extreme obesity					
rs1421085	<i>FTO</i>	5445	-0.057	0.024	0.018
rs17782313	<i>MC4R</i>	4294	-0.041	0.031	0.190
rs6235	<i>PCSK1</i>	4859	-0.032	0.028	0.257
rs1424233	<i>MAF</i>	5521	0.024	0.023	0.296
rs1805081	<i>NPC1</i>	5539	0.022	0.024	0.356
rs12145833	<i>SDCCAG8</i>	3939	0.033	0.037	0.373
rs9568856	<i>OLFM4</i>	3644	0.034	0.044	0.443
rs2116830	<i>KCNMA1</i>	4296	-0.026	0.036	0.464
rs6548238	<i>TMEM18</i>	4006	-0.007	0.038	0.859
rs9299	<i>HOXB5</i>	5178	-0.004	0.026	0.880
rs7132908	<i>FAIM2</i>	3097	-0.001	0.033	0.983

rs17150703	<i>TNKS</i>	3098	0.000	0.051	0.997
Novel genome-wide significant SNPs for the tails of BMI and clinical classes of obesity					
rs7503807	<i>RPTOR</i>	5400	-0.049	0.024	0.040
rs2531995	<i>ADCY9</i>	5446	0.030	0.026	0.251
rs7989336	<i>HS6ST3</i>	5453	-0.024	0.024	0.309
rs17381664	<i>ZZZ3</i>	5378	0.018	0.025	0.477
rs4735692	<i>HNF4G</i>	5475	0.015	0.024	0.541
rs13041126	<i>MRPS33P4</i>	4768	-0.005	0.028	0.872

^aThe beta represents the difference in standardized effects.

Supplementary Table 13. Summary of the studies in the polygene/variance explained analyses

Study	No. of cases/ No. of controls (extreme BMI)	No. subjects for BMI (continuous)*	Covariates	Outcome definition
TwinGene	328/328	6559	age,sex,3 PCA	Cases: upper 5% of BMI distribution; Controls: lower 5% of BMI distribution
LifeLines	405/408	8116	age,sex,10 PCA	Cases: upper 5% of BMI distribution; Controls: lower 5% of BMI distribution
French Obesity Study	1313/1339	N.A.	age,sex	Cases: obese children and class III obese adults; Controls: lean adults and children
Essen Obesity Study	453/435	N.A.	sex	Cases: extremely obese children and adolescents; Controls: healthy lean individuals
GOYA	2633/2740	N.A.	age,sex	Case: extremely obese men (>99th percentile) and women (>96% percentile); Controls: randomly selected from the same population
GEO	1717/1792	N.A.	age,sex, 6 PCA	Cases: BMI>40; Controls: BMI<25

*Some studies did not contribute to the overall analysis as indicated by N.A.

Supplementary Table 14. Variance explained and AUC improvement in six studies of extreme BMI traits

	Twingene	LifeLines	GEO	GOYA	Essen Obesity Study	French Obesity Study	Meta-analysis
Variance explained^a							
GIANT-extreme	7.99%	1.3%	2.2%	4.0%	7.2%	7.1%	4.13%
GIANT-bmi	10.52%	4.6%	3.2%	4.7%	12.9%	8.2%	5.50%
GIANT-extreme polygene^b	8.21%	10.3%	2.8%	4.1%	8.0%	7.6%	6.40%
GIANT-bmi polygene^b	21.36%	16.6%	8.1%	9.0%	17.9%	17.4%	15.30%
AUC improvement^c							
GIANT-extreme polygene^b	0.082	0.065	0.013	0.071	0.127	0.079	0.071
GIANT-bmi polygene^b	0.162	0.096	0.034	0.118	0.218	0.148	0.129
N. SNPs (GIANT-extreme)	10/10	10/10	9/10	10/10	8/10	10/10	10
N. SNPs (GIANT-bmi)	32/32	30/32	29/32	32/32	26/32	30/32	30
N. SNPs (GIANT-extremes polygene)^b	16939	17595	14634	16750	8120	16002	15475
N. SNPs (GIANT-bmi polygene)^b	24970	22527	22294	24792	9144	23882	22800
R² for base model	0.030	0.110	0.472	0.004	0.001	0.025	0.171
AUC for base model	0.581	0.667	0.849	0.531	0.515	0.581	0.654

^a Variance explained is calculated as the improvement in Nagelkerke R² over a base model.

^b Including all the SNPs with P-value lower than 0.05 in GIANT-extremes or GIANT-bmi

^c AUC improvement is calculated over a base model.

Supplementary Table 15. Secondary signals that reached genome-wide significance ($P < 5 \times 10^{-8}$)

SNP	Chr	Position	Gene	Effect allele	Other allele	Effect allele freq	No. cases	No. controls	OR ^a	P ^a	Peak SNP ^b	r ^{2c}	Conditional OR ^d	Conditional P ^d
BMI tails														
rs2058908	16	52363646	<i>FTO</i>	c	t	0.7234	10361.7	10494.4	1.37	2.253E-31	rs8043757	0.184	1.19	2.750E-09
Height tails														
rs10990303	9	97450226	<i>PTCH1</i>	t	c	0.2297	9993.87	9932.94	1.20	1.421E-11	rs1885427	0.001	1.20	6.608E-12
rs572169	3	173648421	<i>GHSR</i>	t	c	0.3155	12254.8	12202.9	1.17	1.109E-12	rs12493901	0.006	1.15	3.541E-11
rs2295887	20	33177848	<i>EDEM2</i>	g	a	0.3874	11407	11372	1.16	8.602E-13	rs224333	0.025	1.15	4.511E-11
rs2814993	6	34726871	<i>C6orf106</i>	a	g	0.1525	8343	8289	1.29	4.942E-14	rs1759645	0.016	1.25	8.623E-11
rs12817549	12	92645445	<i>CRADD</i>	t	c	0.5628	11776	11745	1.11	4.228E-09	rs11107116	0	1.12	1.531E-10
rs1432559	2	56043122	<i>EFEMP1</i>	g	t	0.1969	10246	10199.1	1.21	8.932E-12	rs3791679	0.008	1.19	2.824E-10
rs17720281	4	145763226	<i>HHIP</i>	t	c	0.4207	12747	12728	1.21	4.912E-20	rs1812175	0.114	1.15	4.468E-10
rs1368380	5	171218237	<i>FBXW11</i>	t	c	0.4349	7894	7910	1.15	1.209E-08	rs153750	0.017	1.16	3.285E-09
rs7731703	5	32730699	<i>NPR3</i>	c	t	0.6712	11184.2	11157.1	1.16	2.473E-10	rs13154066	0	1.15	3.783E-09
rs6750795	2	232086475	<i>C2orf52</i>	t	c	0.4427	12852.7	12830.8	1.12	1.728E-08	rs6437061	0.002	1.12	1.081E-08
rs648831	6	81012927	<i>BCKDHB</i>	t	c	0.5067	11861	11813	1.13	1.363E-09	rs310405	0.005	1.12	1.330E-08
rs10512248	9	97299524	<i>PTCH1</i>	g	t	0.3462	11589.6	11562.8	1.13	6.171E-09	rs1885427	0.002	1.13	1.861E-08
rs1866146	2	25234077	<i>EFR3B</i>	g	a	0.3504	9377	9394	1.19	4.32E-13	rs11895026	0.187	1.15	2.011E-08
WHR tails														
rs2745359	6	127423649	<i>RSPO3</i>	c	t	0.0704	2544.9	2542.9	1.82	1.321E-09	rs7745274	0.023	1.74	3.128E-08
Obesity class 1														
rs929641	2	58645881	<i>FANCL</i>	a	g	0.5876	51698	94588.9	1.06	6.581E-09	rs887912	0	1.06	5.780E-09
Overweight														
rs9675886	18	56120302	<i>MC4R</i>	g	a	0.2832	143868	94079	1.07	1.088E-19	rs538656	0.099	1.05	1.169E-08
rs887912	2	59156381	<i>FANCL</i>	t	c	0.2813	154367	102137	1.04	2.39E-08	rs6731302	0.007	1.04	3.18E-08

^aOR and P for the secondary signal from the unconditional meta-analysis

^bMost significant SNP in the region conditioned on.

^cR² between the secondary signal and peak SNP in the region

^dOR and P for the secondary signal from the approximate conditional and joint analysis

Supplementary Table 16. Study-specific haplotype results for the regions that exceeded a prior odds of 1 in 30,000

Study	Sex	No. of SNPs	No. of subjects upper 5% tails	No. of subjects lower 5% tails	Bayes' factor	Log10 Bayes' factor
Results for <i>ID4</i>. Chr6:19,909,472-19,989,472						
ARIC	Male	41	192	192	1.47	0.17
ARIC	Female	41	214	214	7.85	0.89
COLAUS	Male	18	143	143	8.66	0.94
COLAUS	Female	18	127	127	1.81	0.26
LIFELINES	Male	17	175	175	3.73	0.57
LIFELINES	Female	17	233	233	1.69	0.23
NFBC	Male	21	56	56	0.88	-0.05
NFBC	Female	21	58	58	0.55	-0.26
PLCO	Male	28	113	113	1.06	0.03
QIMR	Male	20	83	83	1.79	0.25
QIMR	Female	20	117	117	0.90	-0.05
RS1	Male	31	118	119	0.83	-0.08
RS1	Female	31	162	159	0.71	-0.15
SHIP	Male	44	100	100	44.53	1.65
SHIP	Female	44	102	102	1.75	0.24
TWINGENE	Male	38	172	173	0.23	-0.64
TWINGENE	Female	38	159	160	3.69	0.57
WGHS	Female	10	1155	1155	3.25	0.51
Total			3479	3479	118839	5.07
Results for <i>LIN28B</i>. Chr6:105,445,647-105,525,647						
ARIC	Male	11	192	192	1.01	0.01
ARIC	Female	11	214	214	10.83	1.03
COLAUS	Male	3	143	143	1.01	0.00
COLAUS	Female	3	127	127	0.55	-0.26
LIFELINES	Male	3	175	175	0.65	-0.18
LIFELINES	Female	3	233	233	1.14	0.06
NFBC	Male	8	56	56	0.97	-0.01
NFBC	Female	8	58	58	58.30	1.77
PLCO	Male	12	113	113	1.40	0.15
QIMR	Male	6	83	83	108.85	2.04
QIMR	Female	6	117	117	1.37	0.14
RS1	Male	13	118	119	1.33	0.13
RS1	Female	13	162	159	0.19	-0.73
SHIP	Male	12	100	100	0.56	-0.26
SHIP	Female	12	102	102	0.58	-0.24
TWINGENE	Male	11	172	173	19.14	1.28
TWINGENE	Female	11	159	160	0.50	-0.30
WGHS	Female	3	1155	1155	2.54	0.41
Total			3479	3479	105478	5.02
Results for <i>DLEU7</i>. Chr13:9,963,335-50,043,335						
ARIC	Male	25	192	192	0.99	0.00
ARIC	Female	25	214	214	0.59	-0.23
COLAUS	Male	15	143	143	0.61	-0.21
COLAUS	Female	15	127	127	0.73	-0.14
LIFELINES	Male	12	175	175	0.51	-0.29
LIFELINES	Female	12	233	233	1.87	0.27
NFBC	Male	16	56	56	0.91	-0.04
NFBC	Female	16	58	58	14.47	1.16
PLCO	Male	32	113	113	1.74	0.24
QIMR	Male	15	83	83	1.56	0.19
QIMR	Female	15	117	117	12.95	1.11
RS1	Male	34	118	119	1.36	0.13

Supplementary Table 16. Study-specific haplotype results for the regions that exceeded a prior odds of 1 in 30,000

Study	Sex	No. of SNPs	No. of subjects upper 5% tails	No. of subjects lower 5% tails	Bayes' factor	Log10 Bayes' factor
RS1	Female	34	162	159	2.90	0.46
SHIP	Male	28	100	100	9.13	0.96
SHIP	Female	28	102	102	1.10	0.04
TWINGENE	Male	23	172	173	1.29	0.11
TWINGENE	Female	23	159	160	1.06	0.03
WGHS	Female	10	1155	1155	10.73	1.03
Total			3479	3479	66599	4.82

Supplementary Table 17. SNPs identified in other GWAS near novel loci

Lead SNP	GWAS SNP within 1Mb from lead SNP	Distance from lead SNP (bases)	R ² with lead SNP	Information from GWAS catalog				Same Gene reported for the lead SNP?	PMID
				First author	Study	Disease Trait	Reported Gene		
rs9568867	rs10507577	139261	0.01	Benjamin EJ	Genome-wide association with select biomarker traits in the Framingham Heart Study.	Select biomarker traits	Intergenic		17903293
rs9568867	rs9536591	473735	0.00	Matarin M	A genome-wide genotyping study in patients with ischaemic stroke: initial analysis and data release.	Stroke	Intergenic		17434096
rs7989336	rs2038823	66115	0.24	Huang YC	Genome-wide association study of diabetic retinopathy in a Taiwanese population.	Diabetic retinopathy	<i>HS6ST3</i>	Yes	21310492
rs7989336	rs9556711	998868	0.00	Heath AC	A quantitative-trait genome-wide association study of alcoholism risk in the community: findings and implications.	Alcoholism (alcohol use disorder factor score)	<i>MBNL2</i>		21529783
rs7503807	rs10445407	670698	0.02	Yashin AI	Joint influence of small-effect genetic variants on human longevity.	Longevity	<i>SLC38A10</i>		20834067
rs7503807	rs6565681	242617	0.01	Kamada F	A genome-wide association study identifies RNF213 as the first Moyamoya disease gene.	Moyamoya disease	<i>RNF213</i>		21048783
rs6662509	rs12025126	558050	0.03	Ramdas WD	A genome-wide association study of optic disc parameters.	Vertical cup-disc ratio	<i>RERE</i>		20548946
rs6662509	rs2252865	894928	0.00	The Schizophrenia consortium	Genome-wide association study identifies five new schizophrenia loci	Schizophrenia	<i>NR</i>		21926974
rs6662509	rs4908760	791462	0.00	Jin Y	Variant of TYR and autoimmunity susceptibility loci in generalized vitiligo.	Vitiligo	<i>RERE</i>		20410501
rs584438	rs11078927	534767	0.01	Torgerson DG	Meta-analysis of genome-wide association studies of asthma in ethnically diverse North American populations.	Asthma	<i>GSDMB</i>		21804549
rs584438	rs17609240	488483	0.01	Soranzo N	A genome-wide meta-analysis identifies 22 loci associated with eight hematological parameters in the HaemGen consortium.	Hematological parameters	<i>GSDMA, ORMDL3</i>		19820697
rs584438	rs2290400	532932	0.01	Barrett JC	Genome-wide association study and meta-analysis find that over 40 loci affect risk of type 1 diabetes.	Type 1 diabetes	<i>ORMDL3</i>		19430480
rs584438	rs2305480	536976	0.02	Moffatt MF	A large-scale, consortium-based genomewide association study of asthma.	Asthma	<i>GSDMB</i>		20860503
rs584438	rs2305480	536976	0.02	McGovern DP	Genome-wide association identifies multiple ulcerative colitis susceptibility loci.	Ulcerative colitis	<i>ORMDL3, ZPBP2M, GSDML</i>		20228799
rs584438	rs2315504	447709	0.07	Kim JJ	Identification of 15 loci influencing height in a Korean population.	Height	<i>KRT23, KRT20</i>		19893584
rs584438	rs2872507	558409	0.01	Plagnol V	Genome-wide association analysis of autoantibody positivity in type 1 diabetes cases.	Type 1 diabetes autoantibodies	<i>ORMDL3</i>		21829393
rs584438	rs2872507	558409	0.01	Anderson CA	Meta-analysis identifies 29 additional ulcerative colitis risk loci, increasing the number of confirmed associations to 47.	Ulcerative colitis	<i>IKZF3, ORMDL3, IKZF3, PNMT, ZPBP2, GSDML</i>		21297633
rs584438	rs2872507	558409	0.01	Stahl EA	Genome-wide association study meta-analysis identifies seven new rheumatoid arthritis risk loci.	Rheumatoid arthritis	<i>IKZF3</i>		20453842
rs584438	rs2872507	558409	0.01	Barrett JC	Genome-wide association defines more than 30 distinct susceptibility loci for Crohn's disease.	Crohn's disease	<i>ORMDL3</i>		18587394
rs584438	rs3859192	470524	0.03	Crosslin DR	Genetic variants associated with the white blood cell count in 13,923 subjects in the eMERGE Network.	White blood cell count	<i>GSDMA</i>		22037903
rs584438	rs3894194	477179	0.00	Moffatt MF	A large-scale, consortium-based genomewide	Asthma	<i>GSDMA</i>		20860503

Supplementary Table 17. SNPs identified in other GWAS near novel loci

Information from GWAS catalog									
Lead SNP	GWAS SNP within 1Mb from lead SNP	Distance from lead SNP (bases)	R ² with lead SNP	First author	Study	Disease Trait	Reported Gene	Same Gene reported for the lead SNP?	PMID
					association study of asthma.				
rs584438	rs4794822	442460	0.02	Okada Y	Identification of nine novel loci associated with white blood cell subtypes in a Japanese population.	White blood cell types	<i>PSMD3, CSF3</i>		21738478
rs584438	rs4794822	442460	0.02	Okada Y	Common variations in <i>PSMD3-CSF3</i> and <i>PLCB4</i> are associated with neutrophil count.	Neutrophil count	<i>PSMD3, CSF3</i>		20172861
rs584438	rs6503525	503998	0.01	Ferreira MA	Association between <i>ORMDL3</i> , <i>IL1RL1</i> and a deletion on chromosome 17q21 with asthma risk in Australia.	Asthma	<i>ORMDL3</i>		21150878
rs584438	rs7216389	529223	0.01	Moffatt MF	Genetic variants regulating <i>ORMDL3</i> expression contribute to the risk of childhood asthma.	Asthma	<i>ORMDL3</i>		17611496
rs584438	rs7221109	171114	0.00	Barrett JC	Genome-wide association study and meta-analysis find that over 40 loci affect risk of type 1 diabetes.	Type 1 diabetes	Intergenic		19430480
rs584438	rs8067378	547824	0.01	McGovern DP	Genome-wide association identifies multiple ulcerative colitis susceptibility loci.	Ulcerative colitis	<i>GSDMB</i>		20228799
rs584438	rs907092	676913	0.01	Hirschfield GM	Primary biliary cirrhosis associated with <i>HLA</i> , <i>IL12A</i> , and <i>IL12RB2</i> variants.	Primary biliary cirrhosis	<i>IKZF3</i>		19458352
rs584438	rs9303277	622703	0.01	Liu X	Genome-wide meta-analyses identify three loci associated with primary biliary cirrhosis.	Primary biliary cirrhosis	<i>IKZF3,ZPBP2,GSDMB,ORMDL3</i>		20639880
rs4735692	rs16939046	467709	0.00	Luciano M	Whole genome association scan for genetic polymorphisms influencing information processing speed.	Information processing speed	<i>AC022274.1</i>		21130836
rs4735692	rs2922763	41952	0.53	Speliotes EK	Association analyses of 249,796 individuals reveal 18 new loci associated with body mass index.	Body mass index	<i>HNF4G</i>	Yes	20935630
rs17381664	rs17391694	575295	0.26	Lango Allen H	Hundreds of variants clustered in genomic loci and biological pathways affect human height.	Height	<i>GIPC2</i>		20881960
rs17024258	rs10494112	205156	0.03	Albagha OM	Genome-wide association identifies three new susceptibility loci for Paget's disease of bone.	Paget's disease	<i>CSF1, EPS8LS</i>		21623375
rs17024258	rs12049330	116133	0.02	Shi J	Genome-wide association study of recurrent early-onset major depressive disorder.	Major depressive disorder	<i>ATXN7L2, SYPL2, CYB561D1</i>		20125088
rs17024258	rs12740374	329731	0.00	Lette G	Genome-wide association study of coronary heart disease and its risk factors in 8,090 African Americans: the NHLBI CARE Project.	Coronary heart disease	<i>CELSR2</i>		21347282
rs17024258	rs12740374	329731	0.00	Kathiresan S	Common variants at 30 loci contribute to polygenic dyslipidemia.	LDL cholesterol	<i>CELSR2, PSRC1, SORT1</i>		19060906
rs17024258	rs1933182	147483	0.12	Kottgen A	New loci associated with kidney function and chronic kidney disease.	Chronic kidney disease	<i>SYPL2,ATXN7L2,CYB561D1,PSMA5,AMIGO1,SORT1</i>		20383146
rs17024258	rs484959	218762	0.01	Albagha OM	Genome-wide association study identifies variants at <i>CSF1</i> , <i>OPTN</i> and <i>TNFRSF11A</i> as genetic risk factors for Paget's disease of bone.	Paget's disease	<i>CSF1</i>		20436471
rs17024258	rs599839	325155	0.02	Kim YJ	Large-scale genome-wide association studies in East Asians identify new genetic loci influencing metabolic traits.	Metabolic traits	<i>CELSR2, PSRC1, SORT1</i>		21909109
rs17024258	rs599839	325155	0.02	Schunkert H	Large-scale association analysis identifies 13 new susceptibility loci for coronary artery disease.	Coronary heart disease	<i>SORT1</i>		21378990

Supplementary Table 17. SNPs identified in other GWAS near novel loci

Lead SNP	GWAS SNP within 1Mb from lead SNP	Distance from lead SNP (bases)	R ² with lead SNP	Information from GWAS catalog				Same Gene reported for the lead SNP?	PMID
				First author	Study	Disease Trait	Reported Gene		
rs17024258	rs599839	325155	0.02	Suchindran S	Genome-wide association study of Lp-PLA(2) activity and mass in the Framingham Heart Study.	Lipoprotein-associated phospholipase A2 activity and mass	<i>PSRC1</i>		20442857
rs17024258	rs599839	325155	0.02	Sandhu MS	LDL-cholesterol concentrations: a genome-wide association study.	LDL cholesterol	<i>CELSR2,PSRC1</i>		18262040
rs17024258	rs599839	325155	0.02	Willer CJ	Newly identified loci that influence lipid concentrations and risk of coronary artery disease.	LDL cholesterol	<i>CELSR2,PSRC1,SORT1</i>		18193043
rs17024258	rs599839	325155	0.02	Wallace C	Genome-wide association study identifies genes for biomarkers of cardiovascular disease: serum urate and dyslipidemia.	LDL cholesterol	<i>CELSR2,PSRC1</i>		18179892
rs17024258	rs599839	325155	0.02	Samani NJ	Genomewide association analysis of coronary artery disease.	Coronary heart disease	<i>PSRC1</i>		17634449
rs17024258	rs629301	329015	0.00	Teslovich TM	Biological, clinical and population relevance of 95 loci for blood lipids.	Lipid traits	<i>SORT1</i>		20686565
rs17024258	rs646776	328791	0.00	The Coronary Artery Consortium	A genome-wide association study in Europeans and South Asians identifies five new loci for coronary artery disease.	Coronary heart disease	<i>CELSR2, PSRC1, SORT1</i>		21378988
rs17024258	rs646776	328791	0.00	Carrasquillo MM	Genome-wide screen identifies rs646776 near sortilin as a regulator of progranulin levels in human plasma.	Progranulin levels	<i>SORT1,CELSR2,PSRC1</i>		21087763
rs17024258	rs646776	328791	0.00	Barber MJ	Genome-wide association of lipid-lowering response to statins in combined study populations.	Response to statin therapy	<i>CELSR2,PSRC1, SORT1</i>		20339536
rs17024258	rs646776	328791	0.00	Kathiresan S	Genome-wide association of early-onset myocardial infarction with single nucleotide polymorphisms and copy number variants.	Myocardial infarction (early onset)	<i>CELSR2, PSRC1, SORT1</i>		19198609
rs17024258	rs646776	328791	0.00	Aulchenko YS	Loci influencing lipid levels and coronary heart disease risk in 16 European population cohorts.	Cholesterol, total	<i>CELSR2</i>		19060911
rs17024258	rs646776	328791	0.00	Aulchenko YS	Loci influencing lipid levels and coronary heart disease risk in 16 European population cohorts.	LDL cholesterol	<i>CELSR2</i>		19060911
rs17024258	rs646776	328791	0.00	Sabatti C	Genome-wide association analysis of metabolic traits in a birth cohort from a founder population.	LDL cholesterol	<i>CELSR2, PSRC1, SORT1</i>		19060910
rs17024258	rs646776	328791	0.00	Kathiresan S	Six new loci associated with blood low-density lipoprotein cholesterol, high-density lipoprotein cholesterol or triglycerides in humans.	LDL cholesterol	<i>CELSR2,PSRC1,SORT1</i>		18193044
rs17024258	rs6537837	27589	0.02	Shi J	Genome-wide association study of recurrent early-onset major depressive disorder.	Major depressive disorder	<i>GNAT2, GNAI3, AMPD2</i>	Yes	20125088

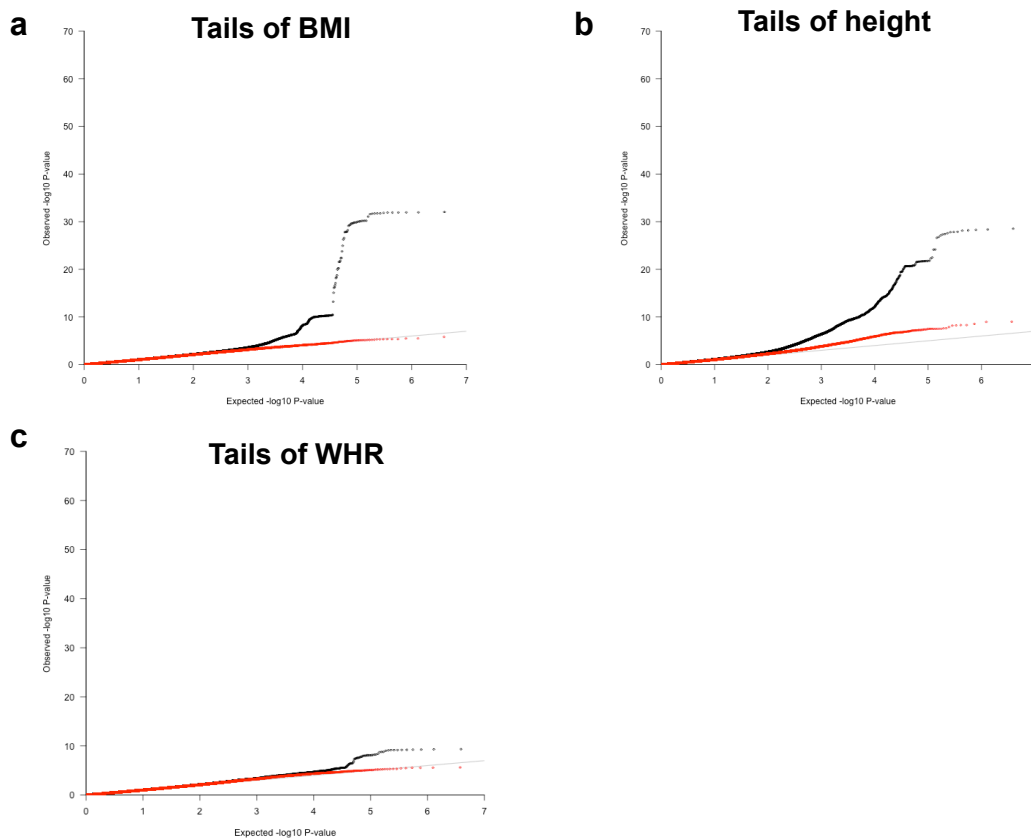
Supplementary Table 17. SNPs identified in other GWAS near novel loci

Information from GWAS catalog									
Lead SNP	GWAS SNP within 1Mb from lead SNP	Distance from lead SNP (bases)	R ² with lead SNP	First author	Study	Disease Trait	Reported Gene	Same Gene reported for the lead SNP?	PMID
rs17024258	rs660240	329483	0.00	Middelberg RP	Genetic variants in LPL, OASL and TOMM40/APOE-C1-C2-C4 genes are associated with multiple cardiovascular-related traits.	Cardiovascular disease risk factors	<i>CELSR2</i>		21943158
rs17024258	rs660240	329483	0.00	Waterworth DM	Genetic variants influencing circulating lipid levels and risk of coronary artery disease.	LDL cholesterol	<i>CELSR2</i>		20864672
rs17024258	rs7528419	330129	0.00	Grallert H	Eight genetic loci associated with variation in lipoprotein-associated phospholipase A2 mass and activity and coronary heart disease: meta-analysis of genome-wide association studies from five community-based studies.	Lipoprotein-associated phospholipase A2 activity and mass	<i>CELSR2</i>		22003152
rs17024258	rs958798	622902	0.01	Mosing MA	A genome-wide association study of self-rated health.	Self-rated health	<i>KCNC4</i>		20707712

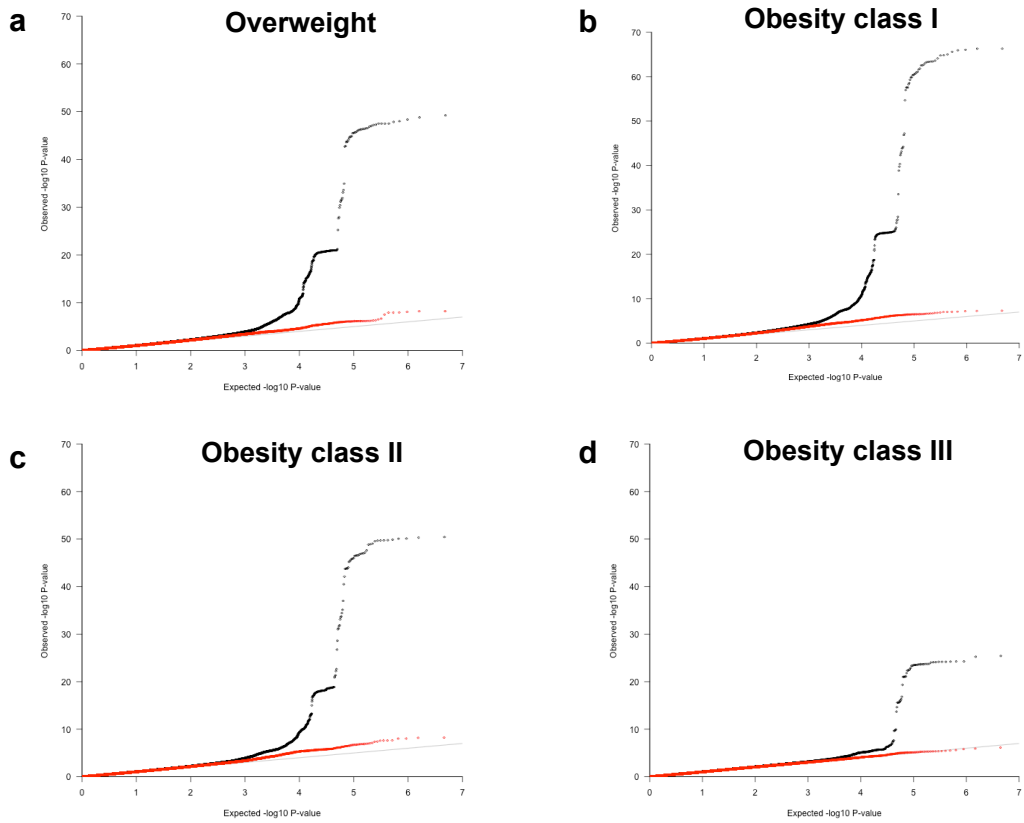
2. SUPPLEMENTARY FIGURES

Supplementary Figures 1-2. Quantile-quantile (Q-Q) plots of SNPs in stage 1 meta-analyses (black) and after removing any SNPs within 1 Mb of the previously reported genome-wide significant hits for BMI, height or WHR and our novel loci (red). The outcomes represented are tails of BMI (panel a; Supplementary Figure [SF] 1), height (panel b; SF1), WHR (panel c; SF 1), overweight (panel a; SF 2), obesity class I (panel b; SF 2), obesity class II (panel c; SF 2), and obesity class III (panel d; SF2).

Supplementary Figure 1. Quantile-quantile (Q-Q) plots of the stage 1 meta-analyses

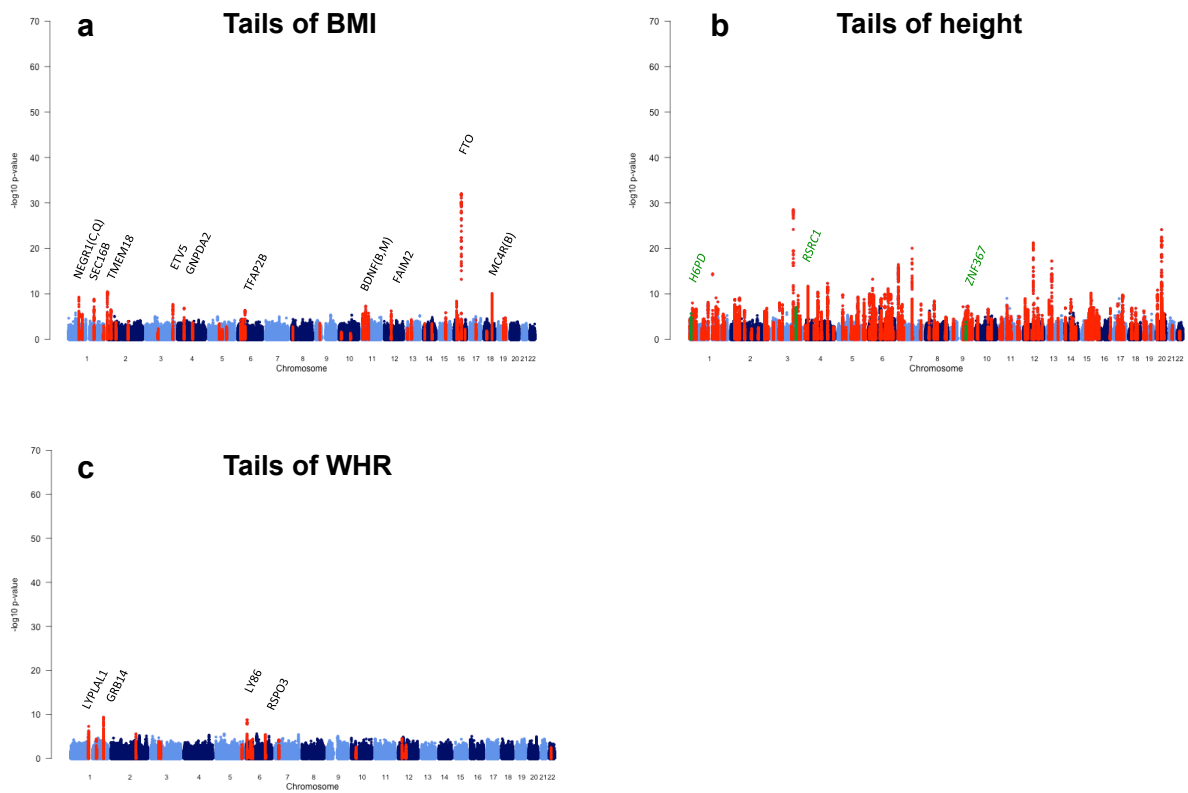


Supplementary Figure 2. Quantile-quantile (Q-Q) plots of the stage 1 meta-analyses

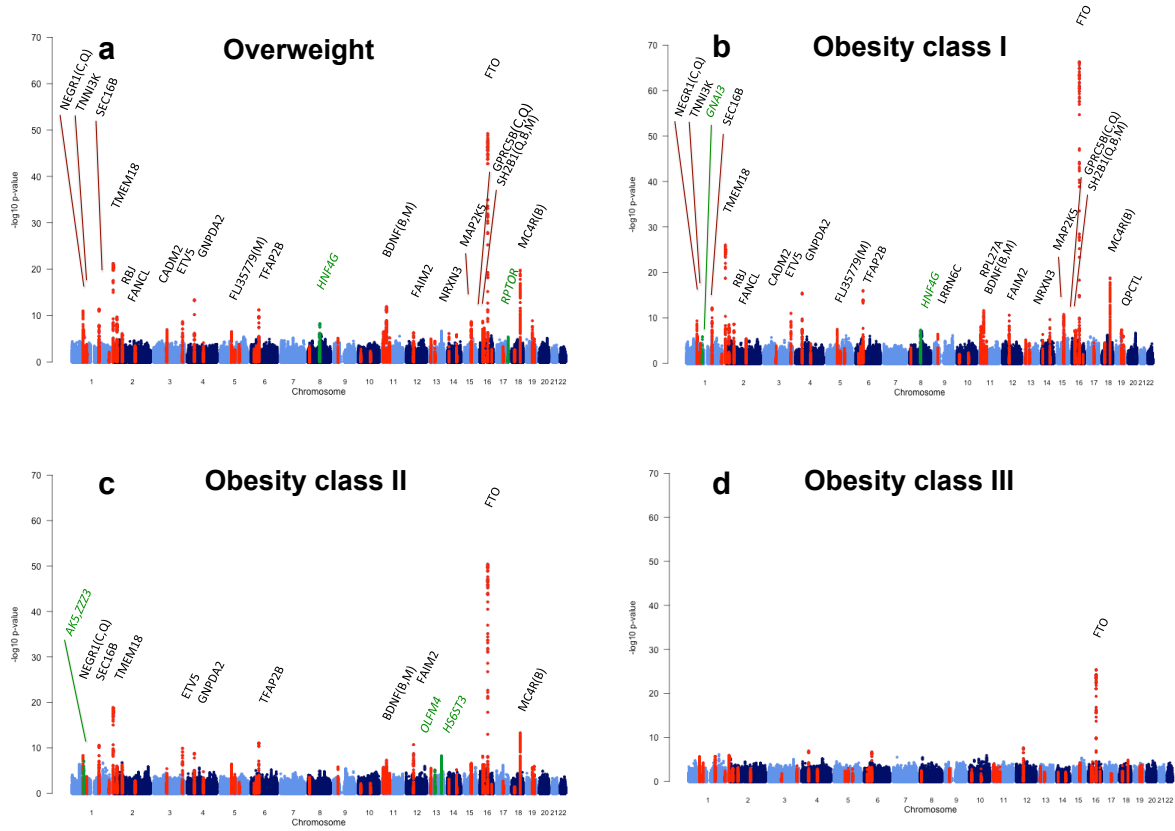


Supplementary Figures 3-4. Manhattan plots showing the significance of association for all SNPs in the stage 1 meta-analyses. SNPs are plotted on the x-axis according to their position on each chromosome against association with respective outcome on the y-axis (shown as $-\log_{10}$ P-value). SNPs previously reported to show genome-wide significant association with BMI, WHR or height are shown in red, and those reaching genome-wide significance for the first time in our analyses (stage 1+2) are highlighted in green. The outcomes represented are tails of BMI (panel a; Supplementary Figure [SF] 3), height (panel b; SF3), WHR (panel c; SF3), overweight (panel a; SF4), obesity class I (panel b; SF4), obesity class II (panel c; SF4) and obesity class III (panel d; SF4).

Supplementary Figure 3. Manhattan plots showing the significance of association for all SNPs in the stage 1 meta-analyses for the tails of BMI, height, and WHR

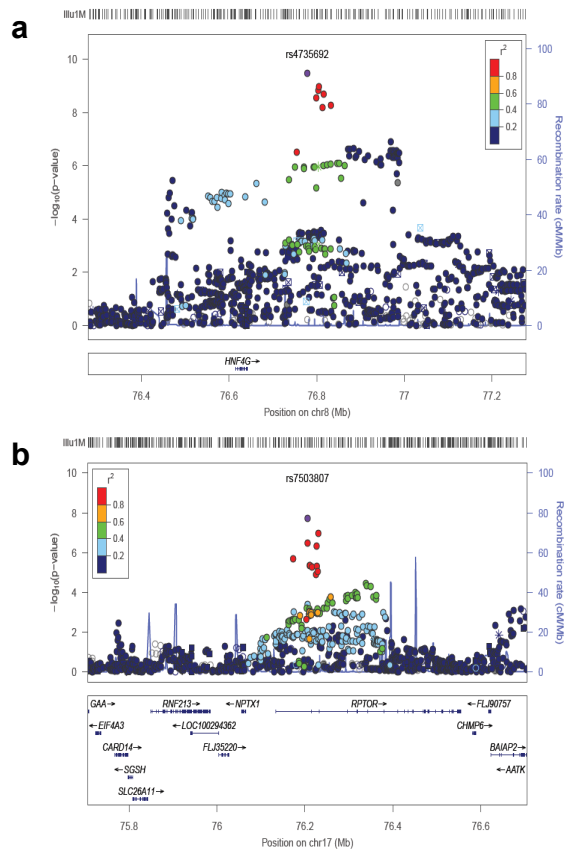


Supplementary Figure 4. Manhattan plots showing the significance of association for all SNPs in the stage 1 meta-analyses for the clinical classes of obesity

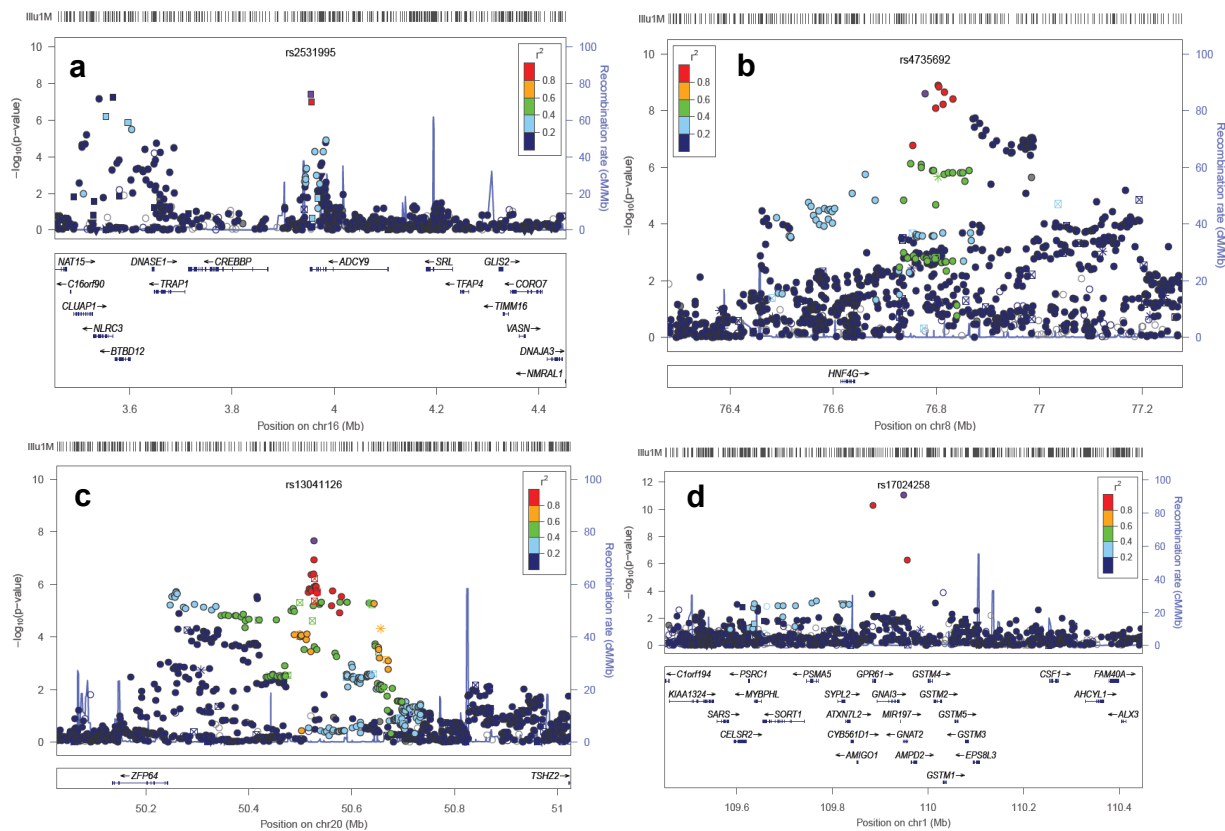


Supplementary Figures 5-8. Regional plots of the 11 novel loci associated with overweight (Supplementary Figure [SF] 5), obesity class I (SF6), obesity class II (SF7), and tails of height (SF8). SNPs are plotted by position on chromosome against association with respective outcome ($-\log_{10}$ P-value). The SNP name shown on the plot was the most significant SNP in the combined stage 1+2 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>) using a distance of 500kb from the SNP noted and HapMap CEU as the reference panel with the following variant annotations: Upright triangles=framestop/splice; upside-down triangle=nonsynonymous variant; filled square=synonymous/UTR.

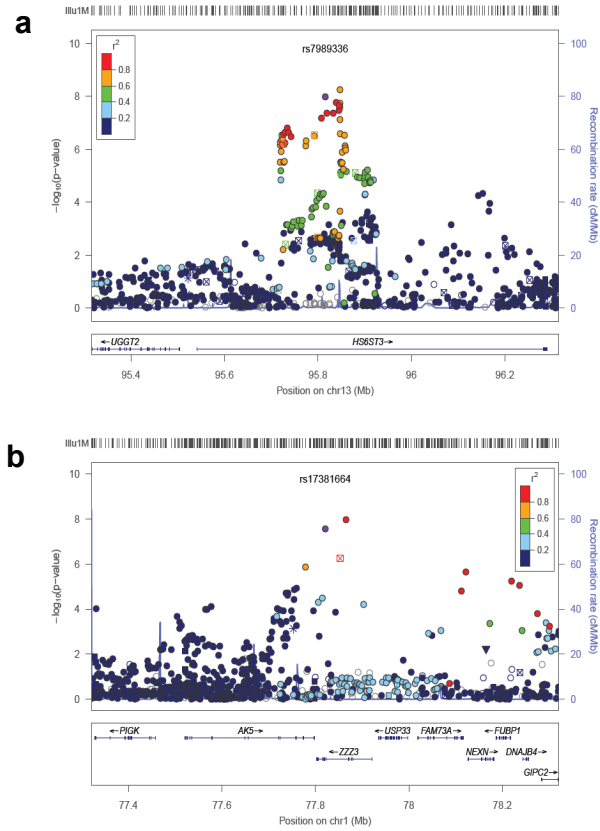
Supplementary Figure 5. Regional plots of the novel loci associated with overweight



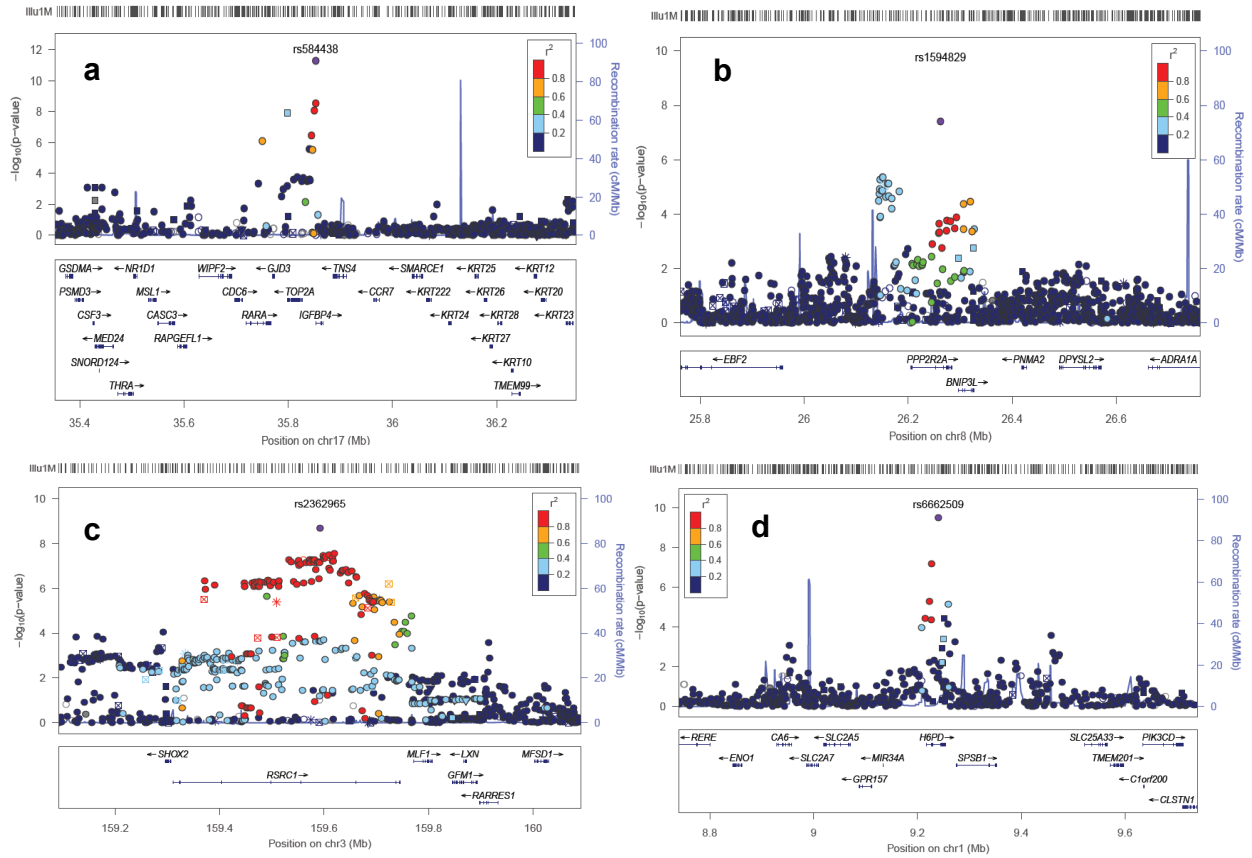
Supplementary Figure 6. Regional plots of the novel loci associated with obesity class I



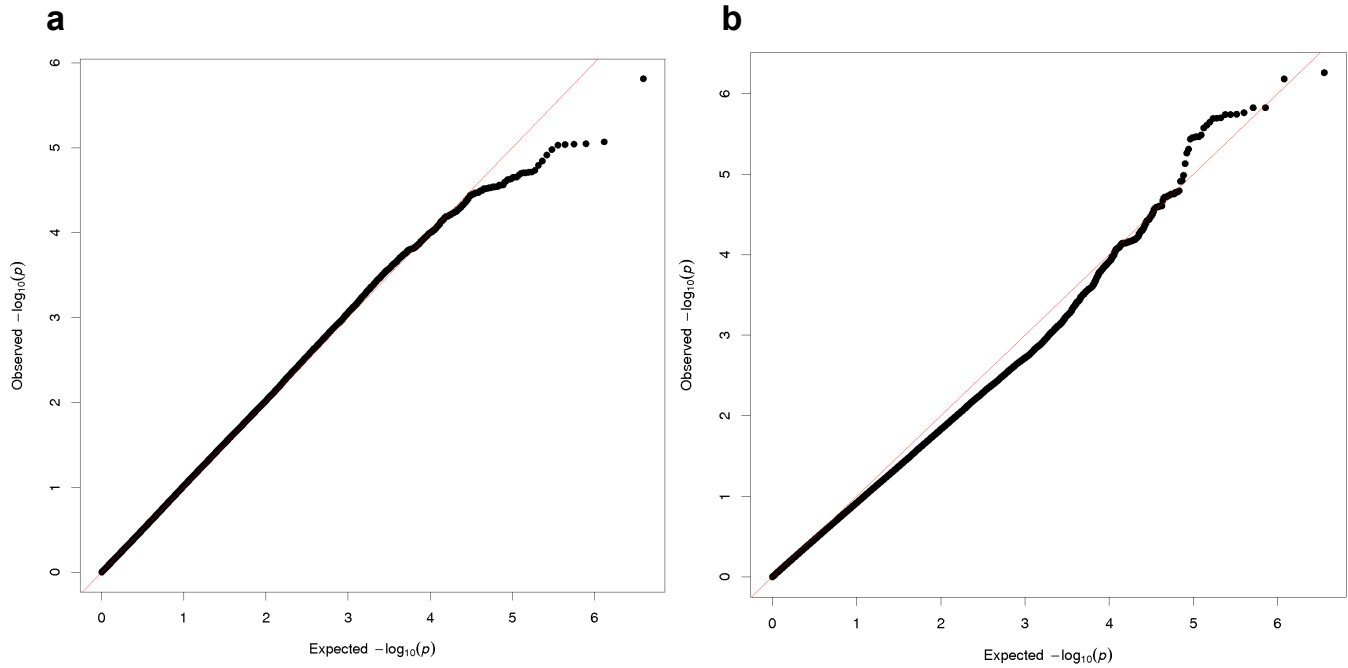
Supplementary Figure 7. Regional plots of the novel loci associated with obesity class II



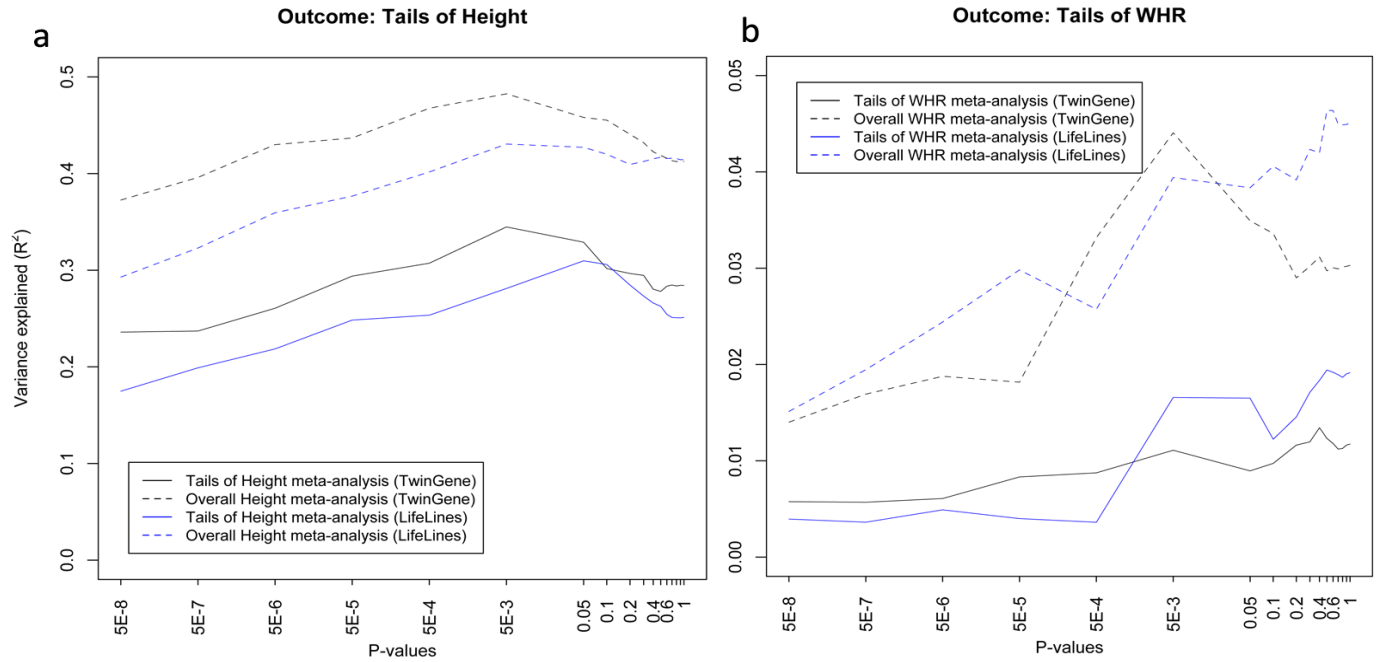
Supplementary Figure 8. Regional plot of the novel loci associated with tails of height



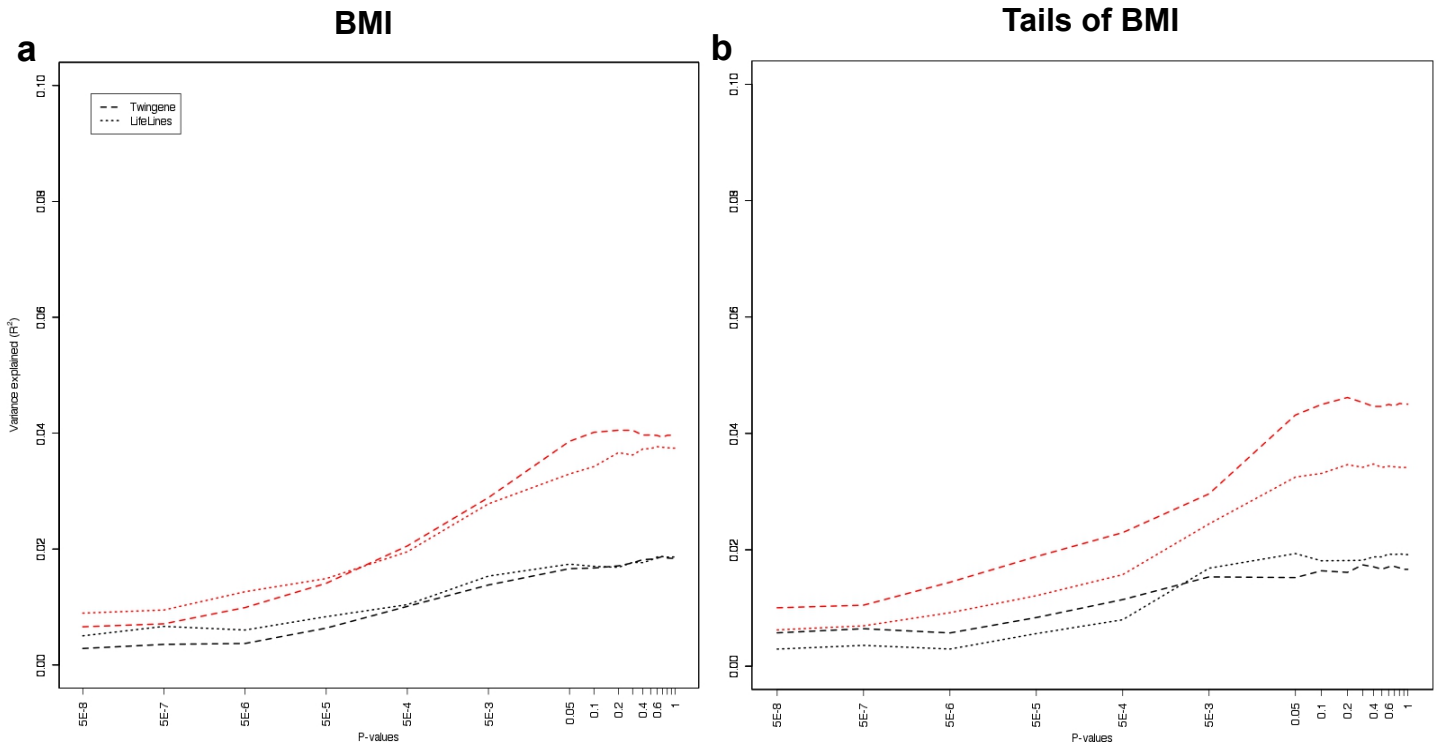
Supplementary Figure 9. Q-Q plots of the $-\log_{10}$ p-values for the difference between the observed associations for the tails and expected associations based on the overall distributions for height (panel a) and WHR (panel b).



Supplementary Figure 10. Variance in tails of height and WHR explained by common variants. The y-axis represents the proportion of variance explained (Nagelkerke R^2) in two studies not included in the discovery meta-analysis. The prediction model was based on the results from the stage 1 meta-analysis of the tails (solid line) or full distribution (dotted line) of height (panel A) and WHR (panel B).

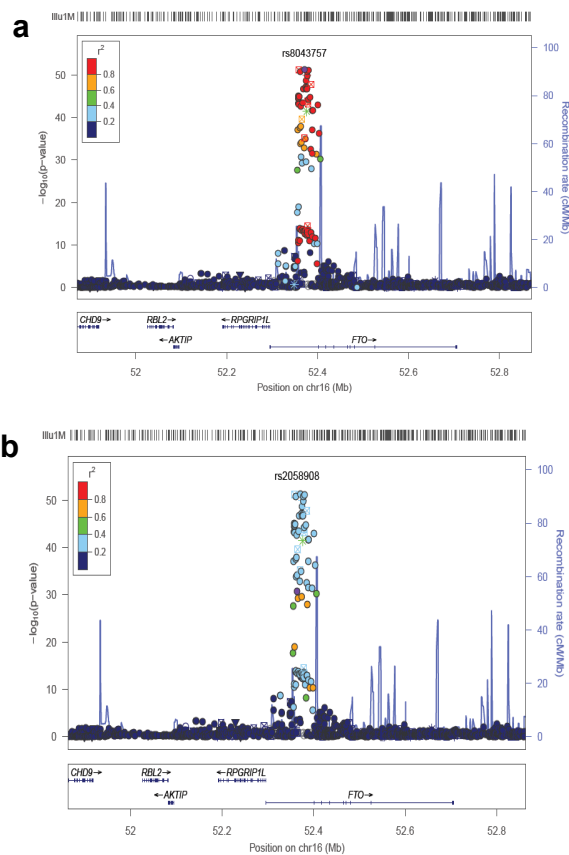


Supplementary Figure 11. Comparison of the phenotypic variance in overall BMI (Panel A) and tails of BMI (Panel B) explained by common genetic variants. By using methods for reweighting to a liability scale (described by Lee *et al*¹), we compared the variance explained for overall BMI and tails of BMI using two different polygene predictors (overall BMI and tails of BMI). In red, the prediction model was based on BMI from the full distribution. In black, the prediction model was based on the results from the stage I meta-analysis of tails of BMI. Only the two population-based studies where the sampling fraction for the tails of BMI was known were included (TWINGENE and LifeLines).

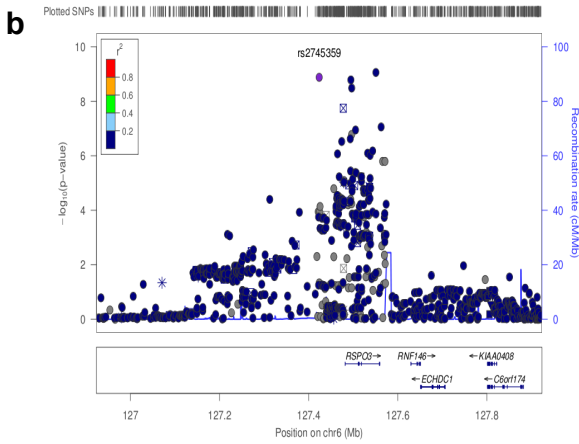
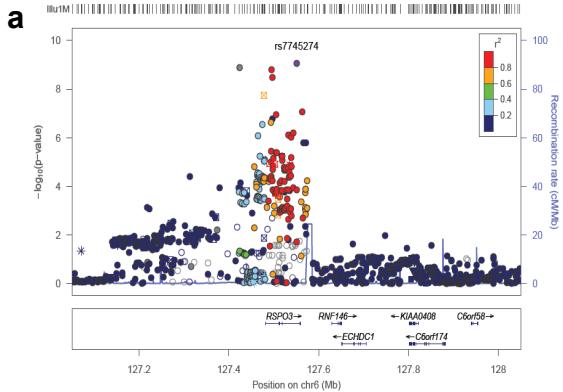


Supplementary Figures 12-14. Secondary signals at the *FTO* (Supplementary Figure [SF] 12), *RSPO3* (SF13), and *FANCL* (SF14) loci contributing to tails of BMI, tails of WHR, and overweight (upper panel) and obesity class I (lower panel), respectively. SNPs are plotted by position on chromosome against association with respective outcome ($-\log_{10}$ P-value). Panel A highlights the most significant SNP in stage 1+2 meta-analysis; panel B the secondary signal that also retained genome-wide significance after conditioning on all other SNPs in the region. 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, exons, and direction of transcription from UCSC genome browser (genome.ucsc.edu) are noted. Hash marks at the top of the figure represent positions of SNPs in the meta-analysis. Regional plots were generated using LocusZoom (<http://csg.sph.umich.edu/locuszoom>) using a distance of 500kb from the SNP noted and HapMap CEU as the reference panel with the following variant annotations: Upright triangles=framestop/splice; upside-down triangle=nonsynonymous variant; filled square=synonymous/UTR.

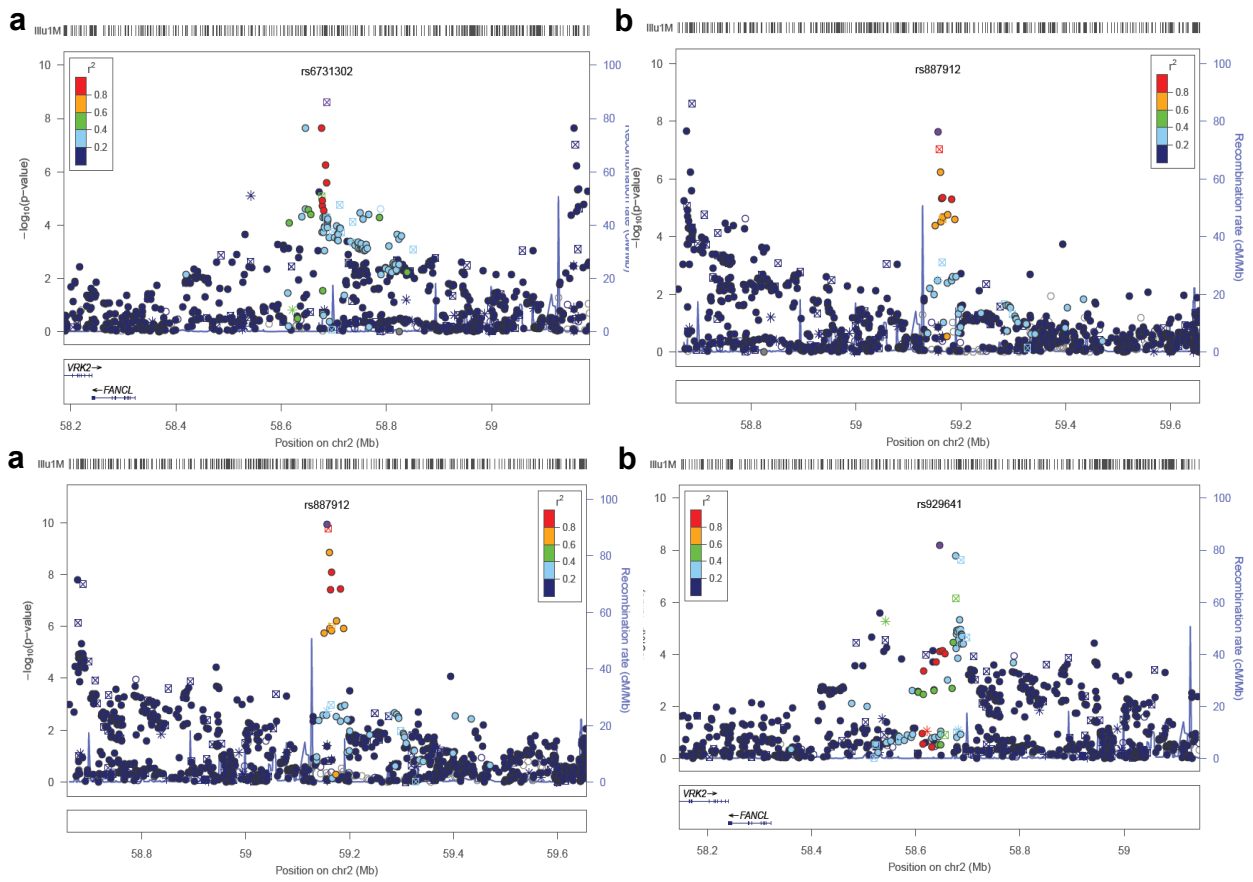
Supplementary Figure 12. Secondary signals at the *FTO* locus



Supplementary Figure 13. Secondary signals at the *RSPO3* locus



Supplementary Figure 14. Secondary signals at the *FANCL* locus



3. SUPPLEMENTARY NOTE

A. Summary Of Literature Search On Genes Nearest To The 11 Novel Loci

We utilized SNIPPER (<http://csg.sph.umich.edu/boehnke/snipper/>) and SNAP² to derive potential biological links of genes in the proximity of the novel association signals (± 500 kb of the index SNP), and present a summary in this section.

Tails of Height

rs584438 (*IGFBP4*): A total of 30 genes are found within 500 kb of the lead marker, rs584438. Two of the more biologically relevant genes that lie within the association signal region are *IGFBP4* and *CDC6*. *IGFBP4* (insulin-like growth factor binding protein) is located 504bp from rs584438. This protein is abundant in skeletal tissue and binds class I and II IGF hormones.³ *IGFBP4* activity seems to play an important role in the level of bone mineral density.⁴ In mice, increased expression of *IGFBP4* in bone leads to decreased cancellous bone growth and remodelling during postnatal growth.³ *CDC6* (cell division cycle 6 homolog) is located 140 kb downstream from the lead marker and plays an important role in regulating the early stages of DNA replication. The *CDC6* gene has been associated with Meier-Gorlin Syndrome, a form of primordial dwarfism, which presents with a mosaic of features including short stature.⁵ While outside of the main signal region, *KRT20* (keratin 20) is 423 kb upstream of the lead marker. *KRT20* encodes a protein that is a member of the keratin family and is found in a large cluster of type I cytokeratin genes in the region of 17q12-q21. The keratins are intermediate filament proteins responsible for the structural integrity of epithelial cells. A previous GWA study identified a SNP near *KRT20* to be associated with height in a Korean population.⁶ Several additional GWA studies have reported associations within the 1 Mb region of rs584438 for asthma,⁷⁻¹⁰ ulcerative colitis,^{11,12} type I diabetes,^{13,14} rheumatoid arthritis,¹⁵ Crohn's disease,¹⁶ haematological variables,¹⁷⁻¹⁹ and primary biliary cirrhosis.^{20,21} All of these signals appear to be independent of our lead SNP signal ($r^2 < 0.2$; **Supplementary Table 17**).

rs6662509 (*H6PD*): A total of 14 genes are found within 500 kb of the lead marker, rs6662509. The SNP is located within an intron in the hexose-6-phosphate dehydrogenase glucose 1-dehydrogenase gene (*H6PD*). *H6PD* is a luminal enzyme of the endoplasmic reticulum involved in several pathways, including insulin resistance and the metabolic syndrome.^{22,23} It interacts

with hydroxysteroid (11-beta) dehydrogenase 1 (HSD11B1), and mutations in both *H6PD* and *HSD11B1* lead to cortisone reductase deficiency, a disease characterized by excessive androgen production and accelerated bone aging.²⁴ *H6PD* gene expression has been associated with type 2 diabetes mellitus²⁵ and *H6PD* null mice develop a severe skeletal myopathy.²⁶ GWA studies have reported associations within the 1 Mb region of rs6662509 for vertical cup-disc ratio among optic disk parameters,²⁷ schizophrenia,²⁸ and vitiligo.²⁹ However, all of these associated markers seem to be independent of the lead SNP, rs6662509 ($r^2 < 0.2$; **Supplementary Table 17**).

rs2362965 (*RSRC1*/*SHOX2*): A total of seven genes are found near our lead marker, rs2362965. The lead marker lies within the *RSRC1* gene (arginine/serine-rich coiled-coil 1, 3q25.32), which encodes a protein involved in mRNA splicing and has been associated with schizophrenia.³⁰ The *SHOX2* gene lies 285 kb upstream of our lead marker. This gene is a member of the SHOX (short stature homeobox) gene family, which plays a major role in skeletal limb development. The *SHOX2* gene is found in all vertebrates and acts as a transcription activator or repressor in different cell types. While it has not been definitively implicated in any human phenotype, restricting the SHOX2 protein in mice results in a shortening of the proximal hind and fore limbs.³¹

rs1594829 (*PPP2R2A*): There are six genes within 500 kb of rs1594829. Many of these genes are associated with cancer-related phenotypes, but do not show an obvious association with height. The lead marker is an intronic SNP within the *PPP2R2A* (protein phosphatase 2, regulatory subunit B, alpha) gene, which codes for the β subunit of the regulatory protein phosphatase 2 (PP2A). PP2A plays a role in the downregulation of cellular growth.³² Although *EBF2* (early B-cell factor 2) lies outside of the main signal region (303kb upstream), its function may relate to height. This protein belongs to the COE (Collier/Olf/EBF) family of helix-loop-helix transcription factors, which plays an important role in variety of developmental processes including neuronal cell differentiation and adipogenesis.³³ Studies with mice suggest that this gene may be involved in the differentiation of osteoblasts.³⁴

Overweight

rs7503807 (*RPTOR*): A total of 13 genes are found within 500 kb of our lead marker, rs7503807. Two of these genes (*RPTOR* and *BAIAP2*) have functions related to obesity. The SNP lies within the *RPTOR* gene (regulatory associated protein of MTOR, complex 1), which encodes an element of the raptor-mTOR complex that regulates cell growth by directing the accumulation of mass based on nutrient availability in the cellular environment. This complex operates in response to nutrient and insulin levels.³⁵ While outside of the main signal region (~418 kb upstream from the lead marker), *BAIAP2* (BAI1-associated protein 2) encodes a brain-specific angiogenesis inhibitor (BAI1)-binding protein. This protein plays a role in insulin uptake as part of the insulin receptor substrate, and likely plays a role in insulin uptake in the central nervous system. This gene has also shown an association with several neurological and behavioral phenotypes, such as brain lateralization, ADHD, autism and neurodegenerative disorders.^{36,37} Two GWA studies have found associations within 1 Mb of the lead marker, rs7503807, including associations with longevity³⁸ and with Moyamoya disease.³⁹ Both of these SNPs appear to be independent of the lead marker (**Supplementary Table 17**).

Overweight and obesity class I

rs4735692 (*HNF4G*): The peak signal is intergenic and only one gene, *HNF4G*, is found in the 1Mb surrounding region. rs4735692 is 137 kb downstream of *HNF4G* (Hepatocyte nuclear factor 4-gamma), which is a transcription factor which is expressed in the pancreas, kidney, small intestine, and testes. In a previous GIANT study of BMI, we detected suggestive evidence for association between another SNP in this region, rs2922763 ($D'=0.91$ and $r^2=0.52$ with rs4735692) and BMI ($p=6.46 \times 10^{-8}$).⁴⁰ In addition, two previous studies have reported associations between SNPs near *HNF4G* and inflammatory bowel disease (rs830772, $D'=0.74$ and $r^2=0.20$)⁴¹ and information processing speed (rs16939046, $r^2 = 0.00$),⁴² although it is unclear if these signals are localizing the same genetic signals (**Supplementary Table 17**).

Obesity class I

rs17024258 (*GNAT2*): There are 27 genes within 500 kb of the peak signal, rs17024258. The lead SNP is intronic in *GNAT2* (guanine nucleotide binding protein (G protein), alpha transducing activity polypeptide 2, 1p13.1), which is part of a 3 subunit G protein, transducin.⁴³

Perhaps more interesting, the SNP is located 207kb from well-known lipid gene *SORT1* (sortilin 1, 1p13.3), which is expressed in multiple cell types and has been reported to be involved in insulin responsiveness in adipose cells.⁴⁴ Sortilin is an essential player in adipocyte and muscle glucose metabolism through the control of *GLUT4* localization. *SORT1* is downregulated in obesity and studies have shown decreased levels of sortilin in the adipose tissues of morbidly obese humans and mice, and in skeletal muscle of obese mice.⁴⁵ TNF-alpha is thought to be involved in this downregulation, suggesting that chronic low-grade inflammation in obesity could thus contribute to insulin resistance by modulating proteins that control *GLUT4*. In addition, *SORT1* is an intracellular sorting receptor for apolipoprotein (apo) B100 that facilitates the formation and hepatic export of apoB100-containing lipoproteins, and regulates plasma low-density lipoprotein cholesterol (LDL-C).⁴⁶ rs17024258 is also 6 kb downstream of *MIR197* gene (microRNA 197) and 9 kb downstream of *GNAI3* (guanine nucleotide binding protein (G protein), alpha inhibiting activity polypeptide 3), which encodes a GNP-binding protein. The heterotrimeric GNP-binding proteins are signal transducers that communicate signals from many hormones, neurotransmitters, chemokines, and autocrine and paracrine factors.⁴⁷ Eighty-four kb upstream of our signal is the *GSTM1* (glutathione S-transferase mu 1) gene, which encodes a glutathione S-transferase belonging to the μ class of enzymes. *GSTM1* functions in the detoxification of electrophilic compounds, including carcinogens, therapeutic drugs, and environmental toxins, by conjugation with glutathione and has been implicated in numerous diseases, such as essential hypertension,⁴⁸ type 2 diabetes,^{49,50} and smoking-related coronary artery risk and disease.^{51,52}

Several GWA studies have reported associations within the 1 MB region of rs17024258, notably with LDL and total cholesterol,⁵³⁻⁵⁹ coronary heart disease,^{54,60-65} progranulin,⁶⁶ metabolic traits,⁶⁷ lipoprotein-associated phospholipase A2 activity and mass,^{68,69} response to statins,⁷⁰ and multiple cardiovascular disease risk factors;⁷¹ however, all signals were independent of our reported SNP. Additional studies have identified associations with SNPs within 500 kb of rs17024258 for Paget's disease,^{72,73} major depressive disorder,⁷⁴ kidney function,⁷⁵ and self-rated health.⁷⁶ Finally, a suggestive GWA was reported between SNPs in *GNAT2* with recurrent early-onset major depressive disorder, although our signal is most likely distinct from this signal (rs6537837-intronic *GNAI3*; $r^2 = 0.012$)⁷⁴ (**Supplementary Table 17**).

rs2531995 (*ADCY9*): There are seventeen genes located within 1 Mb of rs2531995. The SNP is located within the 3' untranslated region of *ADCY9* (adenylate cyclase 9), a membrane-bound enzyme that catalyses the formation of cyclic AMP from ATP. *ADCY9* is regulated by a family of G protein-coupled receptors, protein kinases, and calcium. A previous study by GIANT reported a suggestive association between another SNP in the *ADCY9* region (rs2444217) and body mass index ($r^2 = 0.23$; BMI: p-meta= 9.49×10^{-8}).⁴⁰ Our lead SNP, rs2531995, is also 83 kb from *CREBBP* (CREB binding protein), which is ubiquitously expressed and known to play critical roles in embryonic development, growth control, and homeostasis by coupling chromatin remodeling to transcription factor recognition. Mutations in *CREBBP* cause Rubinstein-Taybi syndrome (RTS), which is characterized by distinctive anthropometric characteristics. rs2531995 is also 226 kb from *SRL* (sarcalumenin), a gene potentially involved in the regulation of calcium transport and 246 kb from *TRAPI* (TNF receptor-associated protein 1), which encodes a highly conserved molecular chaperone that has key roles in signal transduction, protein folding, protein degradation, and morphologic evolution. Several GWA studies have reported associations within the 1 MB region of rs2531995, notably with partial epilepsies,⁷⁷ metabolic syndrome,⁷⁸ although, it is unlikely that these signals are localizing the same genetic signals (**Supplementary Table 17**).

rs13041126 (*MRPS33P4*): There are only two genes within 500 kb of the lead marker, rs13041126. One of these genes, zinc finger protein 64 homolog, mouse (*ZFP64*), lies within the peak signal region. *ZFP64* is a human zinc finger protein, a family of proteins responsible for transcriptional regulation. Associations have been found between *ZFP64* and non-progressive, autosomal recessive, congenital cerebellar ataxia⁷⁹ and with mantle cell lymphoma (MCL) indicating *ZFP64*'s role as a tumor suppressor.⁸⁰ Several GWA studies have reported associations within the 1 Mb region of rs13041126, notably with amyotrophic lateral sclerosis,⁸¹ haemoglobin concentrations,⁸² and male infertility⁸³ (**Supplementary Table 17**).

Obesity class II

rs7989336 (*HS6ST3*): There are only two genes within 500 kb of the lead marker, both lying within the main signal region. The lead marker, rs7989336, is located in an intron of heparan

sulfate 6-O-sulfotransferase 3 (*HS6ST3*), which functions to generate structures required for interactions between heparan sulfate and a variety of proteins. *HS6ST3* is ubiquitously expressed⁸⁴ and is regulated in tissue-specific manners. rs7989336 is 312 kb upstream of the *UGGT2* gene (UDP-glucose glycoprotein glucosyltransferase 2), which encodes a soluble protein that provides quality control for protein transport out of the endoplasmic reticulum. GWA studies have indicated an association within the 1 MB region around rs7989336, with diabetic retinopathy (rs2038823, $r^2 = 0.24$)⁸⁵ and alcoholism;⁸⁶ although these signals may be independent from our lead SNP (**Supplementary Table 17**).

rs17381664 (ZZZ3): There are a total of nine genes that lie within 500 kb of the peak signal. The lead marker, rs17381664, is located within an intron of the *ZZZ3* (ZZ-type zinc finger-containing protein 3) gene encoding a component of the Ada-Two-A-containing (ATAC) complex. ATAC complexes have an essential role in mammalian development, histone acetyltransferase activity on histones H3 and H4, cell cycle progression, and prevention of apoptosis during embryogenesis.^{87,88} Twenty-three kb from our lead marker is the *AK5* (adenylate kinase 5) gene, which encodes a member of the adenylate kinase family, involved in regulating the adenine nucleotide composition within a cell. *AK5* is specifically expressed in brain, and is primarily located in the neuronal cytosolic fraction.⁸⁹ The SNP is 306 kb upstream from the *NEXN* (nexilin F actin binding protein). This gene may function in regulating cell migration through its association with the actin cytoskeleton and is abundant in the heart and skeletal muscle. Mutations in this gene have been associated with dilated cardiomyopathy⁹⁰ and hypertrophic cardiomyopathy.⁹¹ The lead marker is 463 kb upstream of *GIPC2* (GIPC PDZ domain containing family, member 2). GIPC genes may play an important role in the early stages of growth and development and in tumor cell proliferation through the interaction of several growth factor proteins, including IGF1.^{92,93}

B. Detailed Methods Description

Study design

We conducted a two-stage study for the tails of three anthropometric traits (BMI, WHR, and height) and four clinical classes of obesity (overweight and obesity classes I,II, and II). Stage 1 consisted of a meta-analysis of GWAS utilizing data on up to 168,267 individuals of European ancestry from 51 studies participating in the Genetic Investigation of ANthropometric Traits (GIANT) consortium. In stage 2, 273 SNPs with P -values $< 5 \times 10^{-6}$ were followed up in up to additional 109,703 individuals of European descent, which included 67,243 individuals from 24 studies with data from the MetaboChip, and 42,460 additional individuals from 12 studies with *in silico* replication GWAS data. This gave us a study base (or sampling frame) of up to 276,007 individuals of European descent for the joint analysis of stage 1 and stage 2.

Stage 1 – GWA meta-analysis

Samples and genotyping

A total of 51 studies from the GIANT consortium contributed to stage 1 with genotype data from study bases of up to 158,864 (BMI), 168,267 (height), and 100,605 (WHR) adult individuals of European ancestry (**Supplementary Tables 1-5**). The total number of cases and controls for each phenotype is given in **Supplementary Table 1**. Samples from these studies were genotyped using Affymetrix or Illumina whole genome genotyping arrays (**Supplementary Table 3**). After applying appropriate quality control metrics, each study then imputed the ~2.8 million polymorphic autosomal SNPs in the HapMap European CEU population using MACH,⁹⁴ IMPUTE,⁹⁵ or BimBam.⁹⁶

Phenotype definition

The tails of the three anthropometric traits (i.e., BMI, height, and WHR) were defined as the upper 5th percentile (cases) and lower 5th percentile (controls) of the distribution stratified by sex and disease status after controlling for the following covariates: age, age² and principal components for BMI; age and principal components for height, and age, age², BMI and principal components for WHR. For the clinical obesity classes, cases were defined as BMI ≥ 25 kg/m² for overweight, BMI ≥ 30 kg/m² for obesity class I, BMI ≥ 35 kg/m² for obesity class II, and BMI ≥ 40 kg/m² for obesity class 3. Controls were subjects with BMI < 25 kg/m². A minimum of 30 cases and 30 controls for each study-specific stratum was required.

Association analysis of tails of BMI, height and WHR and clinical classes of obesity

Each study conducted a single marker association analysis assuming an additive genetic model taking the genotype imputation uncertainty into account utilizing either MACH2DAT (Y. Li, C.J. Willer, J. Ding, and G.R. Abecasis, unpublished data), Merlin,⁹⁷ SNPTEST,⁹⁵ PLINK,⁹⁸ ProbABEL,⁹⁹ GenABEL,¹⁰⁰ or LME in R. Analyses were stratified by sex (except for studies with related individuals) and disease status for studies that ascertained participants based on a relevant disease (e.g., diabetes). To allow for relatedness, in deCODE, Erasmus Rucphen Family, Family Heart Study, Framingham Heart Study, Essen Obesity study, Swedish Twin Registry, TWINGENE, and TwinsUK, regression coefficients were estimated in the context of a variance component model that modeled the relatedness in men and women combined with sex as a covariate. Before meta-analyzing the data, results from each study were extensively reviewed using standardized quality control procedures to identify potential problems, such as strand issues, discrepancies between the reported standard errors and p-values, and allele frequency differences. SNPs with poor imputation quality scores (e.g., $r^2 < 0.3$ for MACH, observed/expected ratio < 0.3 for BimBam, proper info < 0.4 for IMPUTE, and information < 0.8 for PLINK) or estimated minor allele count ≤ 20 (i.e. $2 \times N \times$ minor allele frequency) in each stratum (men/women or pooled for family-based studies) of each study were removed from analysis. Each stratum- and study-specific GWAS was corrected for genomic control; the average study-specific genomic control values were 1.033 for tails of BMI, 1.052 for tails of height, 1.018 for tails of WHR, 1.019 for overweight, 1.029 for obesity class I, 1.031 for obesity class II, and 1.018 for obesity class III.

Meta-analysis of stage 1 association results

For stage 1, a meta-analysis was performed for each phenotype using METAL¹⁰¹ and the fixed effects inverse variance method based on the β estimates and standard errors from each GWAS. Results were similar using the weighted z-score method. The results of the inverse variance meta-analysis were followed by an additional genomic control correction. Before correction, the median genomic control values were 1.151 for tails of BMI, 1.169 for tails of height, 0.986 for tails of WHR, 1.253 for overweight, 1.275 for obesity class I, 1.142 for obesity class II, and 1.043 for obesity class III.

Selection of SNPs for follow-up

For each phenotype, the most strongly associated SNPs ($P < 5 \times 10^{-6}$) from each locus after filtering using an $r^2 < 0.1$ were taken forward for replication. A total of 237 SNPs, with 70 SNPs selected for more than one trait, were selected for replication. This included 17 SNPs for tails of BMI, 134 for tails of height, 10 for tails of WHR, 37 for overweight, 59 for obesity class I, 37 for obesity class II and 13 for obesity class III.

Stage 2 – Follow-up

Samples and genotyping

A total of 12 studies with *in silico* GWAS data from study bases of up to 42,460 (BMI), 41,565 (Height), and 28,897 (WHR) subjects of European ancestry and 24 studies with MetaboChip data from study bases of up to 67,246 (BMI), 66,177 (Height), and 46,323 (WHR) subjects of European ancestry were included in stage 2 (**Supplemental Tables 1-5**). The total number of cases and controls for each phenotype is given in **Supplementary Table 1**. For the 12 studies providing *in silico* GWAS data, samples were genotyped using Affymetrix or Illumina whole genome platforms, and the ~2.8 million polymorphic autosomal SNPs in the HapMap European CEU population were imputed using MACH,⁹⁴ IMPUTE,⁹⁵ or BimBam.⁹⁶ Twenty-four studies provided genotype data from a custom iSelect MetaboChip array, which was designed for a large-scale follow-up of putative associations for metabolic and cardiovascular traits and contains approximately 195K SNPs. Of the 273 SNPs selected for replication, 102 SNPs were directly genotyped on the chip and 113 SNPs had proxies ($r^2 > 0.8$) that were genotyped on the MetaboChip.

Association analyses and meta-analysis

As described for stage 1, each study tested the association between the replication SNPs and the relevant phenotypes stratified by sex and disease status. The inverse variance method was used to meta-analyze the results separately for the *in silico* GWAS studies and the MetaboChip studies. For the studies with MetaboChip data, a genomic control correction was estimated and applied to each study using SNPs unrelated to anthropometric traits. The results of the MetaboChip meta-analysis were followed by an additional genomic control correction using the same unrelated SNPs. Before correction, the median genomic control values were 0.994 for tails of BMI, 1.087 for tails of height, 0.980 for tails of WHR, 1.021 for overweight, 1.025 for obesity

class I, 1.022 for obesity class II, 0.979 for obesity class III. The meta-analysis results from the *in silico* GWAS studies and MetaboChip studies were then meta-analyzed together (stage 2) and jointly with the stage 1 meta-analysis (stage3). In the final stage, only SNPs separated by at least 1 Mb were considered independent, and loci were only considered novel if located at least 1 Mb from any previously established locus for that trait.

Conditional analyses of secondary signals within loci

To identify potential secondary signals, we utilized the approximate conditional and joint analysis proposed by Yang et al,¹⁰² which uses summary-level statistics and the LD structure from a reference sample to approximate conditional p-values. The meta-analysis results for each trait were analyzed separately with LD correction between SNPs estimated from 6,654 unrelated individuals from the ARIC cohort.

Effects in samples of clinically extreme obese individuals

We tested the association of 91 SNPs (10 SNPs for tails of BMI, 25 for overweight, 33 for obesity class I, 22 for obesity class II and 1 for obesity class III) reaching $P < 5 \times 10^{-8}$ in the joint analysis of stage 1 and stage 2 results, in five studies of clinically extreme obese individuals (**Supplementary Tables 2-5**). In four case-control studies (French Extreme Obesity Study, Essen Obesity Study [Essen Case-Control GWAS], GEO-IT and GOYA), the fixed effects inverse variance method was used to meta-analyze the results, similarly to what previously done for stage 1 and 2 meta-analyses. To further increase our power, we included a fifth study (Essen Obesity Study [Essen Obesity Trio GWAS]) that has a nuclear family structure and includes obese offspring and biological parents. To analyze the four case-control studies and the nuclear family study together, we used the weighted z-score method taking into account the direction but not the magnitude of the effect.

Association analyses with metabolic traits

The effects of the trait-raising alleles on anthropometric traits from the overall distribution, lipids, blood pressure, and glucose- and insulin-related traits were investigated using data from the overall distributions of BMI, height, waist and hip circumference from the GIANT consortium (available at <http://www.broadinstitute.org/collaboration/giant/index.php/>)

GIANT_consortium),^{40,103,104} Global Lipids Genetics Consortium (GLGC; available at <http://www.sph.umich.edu/csg/abecasis/public/lipids2010/>),⁵³ International Consortium for Blood Pressure (ICBP; available at <http://www.igm.jhmi.edu/~gehret/icbp32413ahsfd134/>),¹⁰⁵ and Meta-Analyses of Glucose and Insulin-related traits Consortium (MAGIC; available at <ftp.sanger.ac.uk/pub/magic/>).^{106,107}

Functional variants

We extracted SNPs from either HapMap (version 2, CEU) or 1000G pilot 1 that were in LD with the index SNP ($R^2 > 0.7$) and within 500kb of our best associating variant using SNAP.² We then used Annovar¹⁰⁸ to annotate these variants for functional effects based on RefSeq annotations. When an identical change was found both in HapMap and 1000G, we noted only the 1000G output. Otherwise, results from both datasets are noted. Only effects on the longest transcript were noted.

eQTL analyses

To evaluate functional implications, we examined the *cis* associations between each of the 165 SNPs that reached genome-wide significance ($P < 5 \times 10^{-8}$) and expression of nearby genes in adipose tissue, whole blood, lymphocytes, liver, and brain (cortical tissue). SNPs were tested for *cis* associations with transcripts within a 1 Mb region, assuming an additive effect of the BMI allele or using an ANOVA test. For reporting, p-value thresholds correspond to a false discovery rate of 1% for the lymphocytes and 5% for the remaining tissues were utilized. Conditional analyses were performed by conditioning the trait-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 low statistical power.

Subcutaneous adipose tissue and whole blood from deCode: As described previously,¹⁰⁹ 603 individuals from Iceland with adipose tissue and 747 individuals with whole blood samples were genotyped with the Illumina 317K or 370K platform with subsequent imputation based on the HapMap CEU population. Using RNA from the adipose and blood samples, gene expression profiles were conducted by hybridizing the RNA to a custom-made human array containing 23,720 unique oligonucleotide probes. *Cis* associations were tested by regressing the mean

logarithm (log₁₀) expression ratio (MLR) on the number of effect alleles adjusting for age, sex and age x sex, and the differential cell count for the blood analyses and accounting for familial relatedness. Only *cis* associations with a p-value < 1x10⁻⁵ corresponding to a false discovery rate <5% are reported.

Lymphoblastoid cell lines from a childhood asthma study: In brief, peripheral blood lymphocytes were transformed into lymphoblastoid cell lines for 830 parents and offspring from 206 families of European descent.¹¹⁰ Using extracted RNA from these cell lines, gene expression was assessed with the Affymetrix HG-U133 Plus 2.0 chip, which contains 54,675 transcripts, representing 20,599 genes. Genotyping was conducted using the Illumina Human1M Beadchip and Illumina HumanHap300K Beadchip, and imputation performed using the HapMap CEU population. *Cis* associations (defined as genes within 1 Mb of the SNP) were tested, assuming an additive genetic model and adjusting for non-genetic effects in the gene expression value. Only *cis* associations that reached a $P < 6.8 \times 10^{-5}$ corresponding to a false discovery rate of 1% are reported.

Liver and subcutaneous and omental fat tissue from bariatric surgery: As described previously,¹¹¹ liver (n=707), subcutaneous (n=870) and omental (n=916) fat tissue were obtained from patients who underwent bariatric surgery. RNA from these tissues 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, and *cis* associations with each SNPs were tested. A Bonferroni-correct p-value cutoff of 1×10^{-5} (0.05/5313 transcripts) was denoted as statistically significant.

Subcutaneous adipose tissue, gluteal adipose tissue, and whole blood from MolOBB: As described previously,¹¹² 73 individuals donated subcutaneous adipose tissue from the abdominal wall and gluteus. Total RNA from these tissues was hybridized onto the Affymetrix Human Genome U133 Plus 2.0 gene-expression microarray (hgu133plus2 array), containing 17,726 non-overlapping probes. Subjects were genotyped with the Illumina 317K Beadchip chip array and imputation conducted using IMPUTE. After quality control filters were applied to the expression and genotype data, 52 individuals with abdominal adipose tissue, 62 subjects with gluteal fat, and 65 subjects with whole blood remained for expression analysis. *Cis* associations

within 500kb on either side of the gene were evaluated by regressing expression level on genotype and adjusting for sex and plate effects. A false discovery rate filter was applied,¹¹³ and only those associations with a false discovery rate <5% are presented.

Brain tissue: 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.¹¹⁴ 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 evaluated using the Illumina Human Refseq-8 Expression BeadChip system. *Cis* association analyses assumed an additive model and were adjusted for sex and age at death. A Bonferroni-corrected p-value threshold of 0.0003 was assumed.

Systematic comparisons of the tails and overall distributions of anthropometric traits from the general population

The goal of these analyses was to determine if the observed genetic effects in the tails of distributions of anthropometric traits are different from what we could infer from the overall distributions. To examine the same underlying population, we included all GWAS studies that provided genome-wide results both for the overall trait (e.g. BMI) and for the tails (e.g. tails of BMI) of the distribution. The analyses were done separately for BMI, height and WHRadjBMI.

Calculation of expected association with the tails of the distributions

First, assuming Hardy-Weinberg Equilibrium, we used the minor allele frequencies and the number of analyzed individuals to calculate the number of subjects with each genotype (AA, Aa, aa). Second, we assumed the overall trait to be normally distributed and calculated the expected number of subjects in the upper and lower 5% tails. Third, we used the beta for association between the overall trait and the genotype frequency to calculate, for each genotype, the expected number of individuals in the upper and lower 5% tails. Therefore, for each SNP, we obtained six values, one for each genotype in the lower and upper tail. We used these values to perform a logistic regression, comparing the upper and lower tail, and obtained the ‘expected beta’ and ‘expected standard error’.

Testing of the differences between ‘expected betas’ and ‘observed betas’

We compared the ‘expected betas’ described above with the ‘observed betas’ obtained from the actual analyses of the tails of the distributions. To test the differences between these two quantities, we needed both the variability (‘expected standard error’ and ‘observed standard error’) and the correlation. The latter quantity was not available from the summary results and can only be estimated through bootstrapping of individual level data. We therefore used 10,000 random SNPs from TwinGene, one of the largest studies with individual level data available to us, to estimate the median correlation between ‘expected betas’ and ‘observed betas’. This was estimated to be equal to 0.65. We performed sensitivity analyses varying this correlation coefficient, and found the value estimated from TwinGene to be appropriate in explaining the correlation in the studies used in the actual analyses. Finally, for each SNP, we tested: (‘expected beta’ - ‘observed beta’), where the standard error of the differences was estimated as: $\sqrt{\text{expected standard error}^2 + \text{observed standard error}^2 - 2 \cdot 0.65 \cdot (\text{expected standard error} \cdot \text{observed standard error})}$.

Meta-analysis of the differences between ‘expected betas’ and ‘observed betas’

Differences between ‘expected betas’ and ‘observed betas’ were meta-analyzed using the inverse variance method in METAL. We applied a minor allele count cut-off of 10 in each individual study, and excluded all SNPs that were analyzed in less than half of the maximum sample size from the meta-analysis results. We plotted the q-q plots from the meta-analysis results for BMI, height and WHRadjBMI in **Figure 1** and **Supplementary Figure 10**, respectively.

Comparisons of genetic determinants of overall BMI and tails of BMI from the general population with clinically extremely obese using a polygene approach

For these analyses, we included all GWAS studies that provided genome-wide results both for overall BMI and BMI tails to examine the same underlying population. In these studies, we ran two meta-analyses, one with tails of BMI as outcome (defined as 0-5th vs. 95-100th percentile), and one with overall BMI as outcome. From the results of these meta-analyses, we excluded SNPs with low N (<50% than the max N observed). We pruned the remaining SNPs to be independent by using HapMap release 23 and 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. After the filtering and pruning, we included 124,196 (tails of BMI) and 207,361 (overall BMI) SNPs in

the creation of polygene scores as proposed by the International Schizophrenia Consortium.¹¹⁵ The polygene scores were calculated in four samples of extremely obese (GOYA, GEO-IT, Essen Obesity Study and French Obesity Study; total N=14,153), and in two independent cohort studies (TwinGene and LifeLines) using the same definition of tails of BMI (0-5th vs. 95-100th percentile) as in the discovery analyses (**Supplementary Table 12**). In a polygene approach, instead of limiting the genetic score to genome-wide significant SNPs, the statistically significant threshold is gradually lowered to include more and more SNPs, acknowledging that even if most SNPs are false positives, their impact on the variance explained would be low or negligible, whereas the effect of true positives may be picked up as signals. To estimate the phenotypic variance explained, we fit logistic or linear regression models including age, sex, study-specific covariates and the polygene score as predictors, and tails of BMI or overall BMI as outcomes, in separate models. The phenotypic variance explained by the polygene score is defined as the difference in R^2 (linear regression) or Nagelkerke R^2 (logistic regression) between these models and a basic model including only age, sex and study-specific covariates as predictors. The Nagelkerke R^2 obtained from the logistic regression with tails of BMI as outcome cannot be directly compared with the R^2 from the linear regression with BMI as outcome. In the two population-based studies where the sampling fraction was known (TWINGENE and LifeLines), we made the R^2 and Nagelkerke R^2 comparable using the method suggested by Lee et al¹ that uses the sampling fraction (0.05) to recalibrate the Nagelkerke R^2 to a liability scale (**Supplementary Table 13**).

Haplotype-based analyses

We tested for association of the tails of of height, BMI and WHR with haplotypes across each established and novel locus for the traits in each study using GENE-BPM.¹¹⁵ Analyses were performed separately for males and females. In these analyses, haplotypes were first re-constructed from unphased GWAS SNP data by means of an expectation-maximisation algorithm. These haplotypes are then clustered according to their similarity in terms of their allelic make-up. Within a logistic regression-modelling framework, haplotypes within the same cluster are assigned the same allelic effect, reducing the required number of parameters. Markov-chain Monte Carlo techniques are employed to sample over the space of haplotype clusters and regression model parameters. Evidence in favour of association of haplotypes with each trait

within each study is assessed by means of a Bayes factor. Within each study, we also obtain estimates of population haplotype frequencies, together with posterior mean and standard deviation of haplotype log-odds ratios. In addition, we obtain an approximation to the posterior probability that each pair of haplotypes appear in the same cluster. Association evidence across studies was combined by summing \log_{10} Bayes factors.

C. Full List of Acknowledgements

Stage 1 – Genome-wide association studies

ADVANCE - The ADVANCE study was supported by a grant from the Reynold's Foundation and NHLBI grant HL087647.

AGES - The Age, Gene/Environment Susceptibility Reykjavik Study has been funded by NIH contract N01-AG-12100, the NIA Intramural Research Program, Hjartavernd (the Icelandic Heart Association), and the Althingi (the Icelandic Parliament). The study is approved by the Icelandic National Bioethics Committee, (VSN: 00-063) and the Data Protection Authority. The researchers are indebted to the participants for their willingness to participate in the study.

ARIC - The Atherosclerosis Risk in Communities Study is carried out as a collaborative study supported by National Heart, Lung, and Blood Institute contracts (HHSN268201100005C, HHSN268201100006C, HHSN268201100007C, HHSN268201100008C, HHSN268201100009C, HHSN268201100010C, HHSN268201100011C, and HHSN268201100012C), R01HL087641, R01HL59367 and R01HL086694; National Human Genome Research Institute contract U01HG004402; and National Institutes of Health contract HHSN268200625226C. The authors thank the staff and participants of the ARIC study for their important contributions. Infrastructure was partly supported by Grant Number UL1RR025005, a component of the National Institutes of Health and NIH Roadmap for Medical Research.

BHS - The Busselton Health Study acknowledges the generous support for the 1994/5 follow-up study from Healthway, Western Australia and the numerous Busselton community volunteers who assisted with data collection and the study participants from the Shire of Busselton. The Busselton Health Study is supported by The Great Wine Estates of the Margaret River region of Western Australia. The BHS gratefully acknowledges the assistance of the Western Australian DNA Bank (NHMRC Enabling Facility) with DNA samples and the support provided by the Ark (NHMRC Enabling Facility) for this study.

BRIGHT - The BRIGHT study is supported by the Medical Research Council of Great Britain (G9521010D) and the British Heart Foundation (PG/02/128). This work was facilitated by the forms part of the research themes contributing to the translational research portfolio National Institute for Health Biomedical Research Unit at Barts. The BRIGHT study is extremely grateful to all the patients who participated in the study and the BRIGHT nursing team.

B58C (all subsets) WTCCC/T1DGC/REPL - We acknowledge use of phenotype and genotype data from the British 1958 Birth Cohort DNA collection, funded by the Medical Research Council grant G0000934 and the Wellcome Trust grant 068545/Z/02. (<http://www.b58cgene.sgul.ac.uk/>). Genotyping for the B58C-WTCCC subset was funded by the Wellcome Trust grant 076113/B/04/Z. The B58C-T1DGC genotyping utilized resources provided by the Type 1 Diabetes Genetics Consortium, a collaborative clinical study sponsored by the National Institute of Diabetes and Digestive and Kidney Diseases (NIDDK), National Institute of Allergy and Infectious Diseases (NIAID), National Human Genome Research Institute (NHGRI), National Institute of Child Health and Human Development (NICHD), and Juvenile Diabetes Research Foundation International (JDRF) and supported by U01 DK062418. B58C-T1DGC GWAS data were deposited by the Diabetes and Inflammation Laboratory, Cambridge Institute for Medical Research (CIMR), University of Cambridge, which is funded by Juvenile Diabetes Research Foundation International, the Wellcome Trust and the National Institute for Health Research Cambridge Biomedical Research Centre; the CIMR is in receipt of a Wellcome Trust Strategic Award (079895). Some of the B58C-REPL genotyping was supported by a contract from the European Commission Framework Programme 6 (018996) and grants from the French Ministry of Research.

CAD WTCCC (WTCCC Coronary Artery Disease cases) - Collection of the CAD-WTCCC cases (BHF Family Heart Study) was funded by the British Heart Foundation and the Medical Research Council and genotyping by the Wellcome Trust as part of the WTCCC. We thank the members of the BHF Family Heart Study Research Group for recruitment. NJS hold a Chair supported by the British Heart Foundation and is an NIHR Senior Investigator.

CAPS 1 & 2 - The CAPS study was supported by grants from the Swedish Research Council, the Swedish Cancer Society, and the National Cancer Institute. E.I. was supported by grants from the Swedish Research Council, the Swedish Heart-Lung Foundation, the Swedish Society of Medicine, the Swedish Foundation for Strategic Research, and the Royal Swedish Academy of Science while working with this article.

CHS - Cardiovascular Health Study: This CHS research was supported by NHLBI contracts N01-HC-85239, N01-HC-85079 through N01-HC-85086; N01-HC-35129, N01 HC-15103, N01 HC-55222, N01-HC-75150, N01-HC-45133, HHSN268201200036C and NHLBI grants HL080295, HL087652, HL105756 with additional contribution from NINDS. Additional support

was provided through AG-023629, AG-15928, AG-20098, AG-031890, and AG-027058 from the NIA. See also <http://www.chs-nhlbi.org/pi.htm>. DNA handling and genotyping at Cedars-Sinai Medical Center was supported in part by the National Center for Research Resources grant UL1RR033176, and is now at the National Center for Advancing Translational Sciences, CTSI grant UL1TR000124; in addition to the National Institute of Diabetes and Digestive and Kidney Disease grant DK063491 to the Southern California Diabetes Endocrinology Research Center.

COLAUS - The CoLaus study received financial contributions from GlaxoSmithKline, the Faculty of Biology and Medicine of Lausanne, and the Swiss National Science Foundation (33CSCO-122661). The authors thank Dawn Waterworth, Co-PI of the CoLaus study. Special thanks to Sven Bergmann, Jacques Beckmann, Murielle Bochud, Yolande Barreau, Mathieu Firmann, Vladimir Mayor, Anne-Lise Bastian, Binasa Ramic, Martine Moranville, Martine Baumer, Marcy Sagette, Jeanne Ecoffey and Sylvie Mermoud for data collection. VM is a part-time employee of the GlaxoSmithKline.

COROGENE - The study was supported in part by the Aarno Koskelo Foundation, the Finnish Foundation for Cardiovascular Research, and the EVO funds of Helsinki University Central Hospital. The Corogene study is not supported by industry and all presentations have been carried out in academic environment.

CROATIA-VIS - The CROATIA-Vis study was funded by grants from the Medical Research Council (UK), European Commission Framework 6 project EUROSPAN (Contract No. LSHG-CT-2006-018947) and Republic of Croatia Ministry of Science, Education and Sports research grants. (108-1080315-0302). The funders had no role in study design, data collection and analysis, decision to publish, or preparation of the manuscript. We would like to acknowledge the staff of several institutions in Croatia that supported the field work, including but not limited to The University of Split and Zagreb Medical Schools, Institute for Anthropological Research in Zagreb and Croatian Institute for Public Health. The SNP genotyping for the CROATIA-Vis cohort was performed in the core genotyping laboratory of the Wellcome Trust Clinical Research Facility at the Western General Hospital, Edinburgh, Scotland, UK.

deCODE - We would like to thank participants in deCODE cardiovascular- and obesity studies and collaborators for their cooperation. The research performed at deCODE Genetics was part funded through the European Community's Seventh Framework Programme (FP7/2007-2013), ENGAGE project, grant agreement HEALTH-F4-2007- 201413.

DGI - The Botnia (DGI) study have been supported by grants from Folkhälsan Research Foundation, Sigrid Juselius Foundation, Ministry of Education, Nordic Center of Excellence in Disease Genetics, Gyllenberg Foundation, Swedish Cultural Foundation in Finland, Finnish Diabetes Research Foundation, Foundation for Life and Health in Finland, Finnish Medical Society, Paavo Nurmi Foundation, Perklén Foundation, Ollqvist Foundation, Närpes Health Care Foundation, the Municipal Health Care Center and Hospital in Jakobstad, Health Care Centers in Vasa, Närpes and Korsholm. This work was also partially supported by NIH grant R01-DK075787 to JNH and March of Dimes grant 6-FY-09-507 to JNH.

Estonian Genome Center of University of Tartu (EGCUT) - EGCUT received financing by FP7 grants (201413, 245536), grant from Estonian Government SF0180142s08, from the EU through the European Regional Development Fund, OPENGENE, in the frame of Centre of Excellence in Genomics and Univ. of Tartu grant SP1GVARENG.

EPIC - The EPIC Norfolk Study is funded by Cancer Research United Kingdom and the Medical Research Council.

ERF - The genotyping for the ERF study was supported by EUROSPAN (European Special Populations Research Network) and the European Commission FP6 STRP grant (018947; LSHG-CT-2006-01947). The ERF study was further supported by grants from the Netherlands Organisation for Scientific Research, Erasmus MC, the Centre for Medical Systems Biology (CMSB) and the Netherlands Brain Foundation (HersenStichting Nederland). We are grateful to all patients and their relatives, general practitioners and neurologists for their contributions and to P. Veraart for her help in genealogy, Jeannette Vergeer for the supervision of the laboratory work and P. Snijders for his help in data collection. We would also like to acknowledge Internationale Stichting Alzheimer Onderzoek (ISAO) and Hersenstichting Netherlands.

FamHS - The Family Heart Study (FamHS) was supported by NIH grants R01-HL-087700 and R01-HL-088215 (Michael A. Province, PI) from NHLBI; and R01-DK-8925601 and R01-DK-075681 (Ingrid B. Borecki, PI) from NIDDK.

FENLAND - The Fenland Study is funded by the Wellcome Trust and the Medical Research Council, as well as by the Support for Science Funding programme and CamStrad. We are grateful to all the volunteers for their time and help, and to the General Practitioners and practice staff for help with recruitment. We thank the Fenland Study co-ordination team and the Field Epidemiology team of the MRC Epidemiology Unit for recruitment and clinical testing.

FRAM - This research was conducted in part using data and resources from the Framingham Heart Study of the National Heart Lung and Blood Institute of the National Institutes of Health and Boston University School of Medicine. The analyses reflect intellectual input and resource development from the Framingham Heart Study investigators participating in the SNP Health Association Resource (SHARe) project. This work was partially supported by the National Heart, Lung and Blood Institute's Framingham Heart Study (Contract No. N01-HC-25195) and its contract with Affymetrix, Inc for genotyping services (Contract No. N02-HL-6-4278). A portion of this research utilized the Linux Cluster for Genetic Analysis (LinGA-II) funded by the Robert Dawson Evans Endowment of the Department of Medicine at Boston University School of Medicine and Boston Medical Center.

FUSION - Support for FUSION was provided by NIH grants R01-DK062370 (to M.B.), R01-DK072193 (to K.L.M.), and intramural project number 1Z01-HG000024 (to F.S.C.). Genome-wide genotyping was conducted by the Johns Hopkins University Genetic Resources Core Facility SNP Center at the Center for Inherited Disease Research (CIDR), with support from CIDR NIH contract no. N01-HG-65403.

Genmets - Genmets was supported through funds from The European Community's Seventh Framework Programme (FP7/2007-2013), BioSHaRE Consortium, grant agreement 261433. V.S. was supported by the Sigrid Juselius Foundation, Finnish Foundation for Cardiovascular research, and the Finnish Academy (grant number 129494). S.R. was supported by the Academy of Finland Center of Excellence in Complex Disease Genetics (213506 and 129680), Academy of Finland (251217), the Finnish foundation for Cardiovascular Research and the Sigrid Juselius Foundation.

GerMIFS1+2 - GerMIFS1, GerMIFS2: Supported by the Deutsche Forschungsgemeinschaft and the German Federal Ministry of Education and Research (BMBF) in the context of the German National Genome Research Network (NGFN-2 and NGFN-plus), the FP6 and FP7 EU funded integrated projects Cardiogenics (LSHM-CT-2006-037593), ENGAGE (201413), the bi-national BMBF/ANR funded project CARDomics (01KU0908A), and the Nordic Center of Cardiovascular Research (NCCR).

GOOD - Financial support was received from the Swedish Research Council (K2010-54X-09894-19-3, 2006-3832 and K2010-52X-20229-05-3), the Swedish Foundation for Strategic Research, the ALF/LUA research grant in Gothenburg, the Lundberg Foundation, the Torsten

and Ragnar Söderberg's Foundation, Petrus and Augusta Hedlunds Foundation, the Västra Götaland Foundation, the Göteborg Medical Society, the Novo Nordisk foundation, the Sahlgrenska Center for Cardiovascular and Metabolic Research (CMR, no. A305:188), which is supported by the Swedish Foundation for Strategic Research, and the European Commission grant HEALTH-F2-2008-201865-GEFOS. We would like to acknowledge Maria Nethander at the Genomics core facility at the University of Gothenburg for statistical analyses. We would like to thank Dr. Tobias A. Knoch, Luc V. de Zeeuw, Anis Abuseiris, and Rob de Graaf as well as their institutions the Erasmus Computing Grid, Rotterdam, The Netherlands, and especially the national German MediGRID and Services@MediGRID part of the German D-Grid, both funded by the German Bundesministerium fuer Forschung und Technology under grants #01 AK 803 A-H and # 01 IG 07015 G for access to their grid resources. We would also like to thank Karol Estrada, Department of Internal Medicine, Erasmus MC, Rotterdam, Netherlands for advice regarding the grid resources.

HBCS - Helsinki Birth Cohort Study has been supported by grants from the Academy of Finland (Grant No. 120386 and 125876 to JGE), the Finnish Diabetes Research Society, Folkhälsan Research Foundation, Novo Nordisk Foundation, Finska Läkaresällskapet, the European Science Foundation (EuroSTRESS), the Wellcome Trust (Grant No. 89061/Z/09/Z and 089062/Z/09/Z), Samfundet Folkhälsan, Finska Läkaresällskapet and the Signe and Ane Gyllenberg foundation. We thank all study participants as well as everybody involved in the Helsinki Birth Cohort Study.

KORA S3 and S4 - The KORA Augsburg studies were financed by the Helmholtz Zentrum München, German Research Center for Environmental Health, Neuherberg, Germany and supported by grants from the German Federal Ministry of Education and Research (BMBF). Part of this work was financed by the German National Genome Research Network (grant number NGFNPLUS 01GS023 and 01GS0834). Our research was supported within the Munich Center of Health Sciences (MC Health) as part of LMUinnovativ.

MGS - The Molecular Genetics of Schizophrenia project was carried out by 10 research sites and PIs: Pablo V. Gejman, Study Coordinator (Department of Psychiatry and Behavioral Sciences, NorthShore University HealthSystem, Evanston, IL, and Department of Psychiatry and Behavioral Sciences, University of Chicago, Chicago, IL), Douglas F. Levinson (Stanford University), Bryan J. Mowry (University of Queensland), Donald Black (University of Iowa),

Robert Freedman (University of Colorado), C. Robert Cloninger (Washington University), Jeremy Silverman (Mt. Sinai Medical School), Nancy Buccola (Louisiana State University - New Orleans), William Byerley (University of California at San Francisco), and Farooq Amin (Emory University). This study was supported by NIH R01 grants (MH67257 to N.G.B., MH59588 to B.J.M., MH59571 to P.V.G., MH59565 to R.F., MH59587 to F.A., MH60870 to W.F.B., MH59566 to D.W.B., MH59586 to J.M.S., MH61675 to D.F.L., MH60879 to C.R.C., and MH81800 to P.V.G.), MH79469 to P.V.G., and MH79470 to D.F.L.), NARSAD (National Alliance for Research on Schizophrenia and Depression) Young Investigator Awards (to J.D. and A.R.S.), the Genetic Association Information Network (GAIN), the Walter E. Nichols, M.D., and Eleanor Nichols endowments, at Stanford University, and by The Paul Michael Donovan Charitable Foundation. Genotyping was carried out by the Genotyping and Analysis at the Broad Institute of Harvard and MIT (S. Gabriel and D.B.M.), which is supported by grant U54 RR020278 from the National Center for Research Resources. Genotyping of half of the control sample presented here was carried out with support from GAIN. The GAIN quality control team (G.R. Abecasis and J. Paschall) made important contributions to the project.

MICROS - For the MICROS study, we thank the primary care practitioners in the villages of the Val Venosta and the personnel of the Hospital of Silandro (Department of Laboratory Medicine) for their participation and collaboration in the research project. In South Tyrol, the study was supported by the Ministry of Health and Department of Educational Assistance, University and Research of the Autonomous Province of Bolzano, and the South Tyrolean Sparkasse Foundation.

MIGEN - The MIGen study was supported by the National Heart, Lung, and Blood Institute's STAMPEED genomics research program (R01 HL087676) and the National Center for Research Resources (U54 RR020278). This work was also partially supported by NIH grant K23-DK080145 to EKS, NIH grant R01-DK075787 to JNH and March of Dimes grant 6-FY-09-507 to JNH.

NHS - This study was supported by grants HL71981, CA65725, CA87969, CA49449, CA67262, CA50385 and 5UO1CA098233 from the National Institutes of Health. Dr. Lu Qi is a recipient of the American Heart Association Scientist Development Award (0730094N)

NSPHS - The Northern Swedish Population Health Study (NSPHS) was funded by the Swedish Medical Research Council (project number K2007-66X-20270-01-3), and the Foundation for

Strategic Research (SSF). The NSPHS as part of EUROSPAN (European Special Populations Research Network) was also supported by European Commission FP6 STRP grant number 01947 (LSHG-CT-2006-01947). This work was also supported by the Swedish Society for Medical Research (ÅJ). The authors are grateful for the contribution of district nurse Svea Hennix for data collection and Inger Jonasson for logistics and coordination of the health survey. Finally, the authors thank all the community participants for their interest and willingness to contribute to the study.

NTR and NESDA - Funding was obtained from the Netherlands Organization for Scientific Research (NWO: MagW/ZonMW): Genetic basis of anxiety and depression (904-61-090); Genetics of individual differences in smoking initiation and persistence (NWO 985-10-002); Resolving cause and effect in the association between exercise and well-being (904-61-193); Twin family database for behavior genomics studies (480-04-004); Twin research focusing on behavior (400-05-717); Genetic determinants of risk behavior in relation to alcohol use and alcohol use disorder (Addiction-31160008); Genotype/phenotype database for behavior genetic and genetic epidemiological studies (40-0056-98-9032); Spinozapremie (SPI 56-464-14192); CMSB: Center for Medical Systems Biology (NWO Genomics); NBIC/BioAssist/RK/2008.024); BBMRI –NL: Biobanking and Biomolecular Resources Research Infrastructure (184.021.007); the VU University: Institute for Health and Care Research (EMGO+) and Neuroscience Campus Amsterdam (NCA); the European Science Foundation (ESF): Genomewide analyses of European twin and population cohorts (EU/QLRT-2001-01254); European Community's Seventh Framework Program (FP7/2007-2013): ENGAGE (HEALTH-F4-2007-201413); the European Research Council (ERC) Genetics of Mental Illness (230374); Rutgers University Cell and DNA Repository cooperative agreement (NIMH U24 MH068457-06); Collaborative study of the genetics of DZ twinning (NIH R01D0042157-01A); National Institute of Mental Health (NIMH; 1RC2 MH089951-01 and 1RC2MH089995-01); the Genetic Association Information Network, a public–private partnership between the NIH and Pfizer Inc., Affymetrix Inc. and Abbott Laboratories.

PLCO - The Prostate, Lung, Colorectal, and Ovarian (PLCO) Cancer Screening Trial was funded by the Intramural Research Program of the Division of Cancer Epidemiology and Genetics, National Cancer Institute, NIH

PROCARDIS - European Commission (LSHM-CT-2007-037273), the Swedish Heart-Lung Foundation, the Swedish Research Council (8691), the Knut and Alice Wallenberg Foundation, the Foundation for Strategic Research, the Torsten and Ragnar Söderberg Foundation, the Strategic Cardiovascular Programme of Karolinska Institutet and the Stockholm County Council, the Stockholm County Council (560183) and the Wellcome trust core award [090532/Z/09/Z]. Martin Farrall and Hugh Watkins acknowledge support from the British Heart Foundation Centre of Research Excellence, Oxford.

RSI - The generation and management of GWAS genotype data for the Rotterdam Study is supported by the Netherlands Organisation of Scientific Research NWO Investments (nr. 175.010.2005.011, 911-03-012). This study is funded by the Research Institute for Diseases in the Elderly (014-93-015; RIDE2), the Netherlands Genomics Initiative (NGI)/Netherlands Organisation for Scientific Research (NWO) project nr. 050-060-810. We thank Pascal Arp, Mila Jhamai, Marijn Verkerk, Lizbeth Herrera and Marjolein Peters for their help in creating the GWAS database, and Karol Estrada and Maksim V. Struchalin for their support in creation and analysis of imputed data. The Rotterdam Study is funded by Erasmus Medical Center and Erasmus University, Rotterdam, Netherlands Organization for the Health Research and Development (ZonMw), the Research Institute for Diseases in the Elderly (RIDE), the Ministry of Education, Culture and Science, the Ministry for Health, Welfare and Sports, the European Commission (DG XII), and the Municipality of Rotterdam. The authors are grateful to the study participants, the staff from the Rotterdam Study and the participating general practitioners and pharmacists.

ORCADES - ORCADES was supported by the Chief Scientist Office of the Scottish Government, the Royal Society, and the European Union framework program 6 European Special Populations Research Network project [contract LSHG-CT-2006-018947]. ORCADES would like to acknowledge the invaluable contributions of Lorraine Anderson, the research nurses in Orkney, and the administrative team in Edinburgh. DNA extraction for ORCADES was carried out by the Genetics Core Laboratory at the Wellcome Trust Clinical Research Facility, WGH, Edinburgh, Scotland and SNP genotyping was performed by Helmholtz Zentrum München, GmbH, Neuherberg, Germany.

SardiNIA - We thank all the volunteers who generously participated in this study, Monsignore Piseddu, Bishop of Ogliastra and the mayors and citizens of the Sardinian towns (Lanusei,

Ilbono, Arzana, and Elini). This work was supported by the Intramural Research Program of the National Institute on Aging (NIA), National Institutes of Health (NIH). The SardiNIA (“Progenia”) team was supported by Contract NO1-AG-1–2109 from the NIA; the efforts of GRA were supported in part by contract 263-MA-410953 from the NIA to the University of Michigan and by research grant HG002651 and HL084729 from the NIH (to GRA).

SASBAC - The SASBAC study was supported by funding from the Agency for Science, Technology and Research of Singapore (A*STAR), the United States National Institute of Health (NIH) and the Susan G. Komen Breast Cancer Foundation.

SEARCH - Paul Pharoah, Douglas Easton, Simon Gayther and the SEARCH and UKOPS teams. SEARCH was funded by Cancer Research UK (C490/A10124 and C490/A8339).

SHIP - SHIP is part of the Community Medicine Research net of the University of Greifswald, Germany, which is funded by the Federal Ministry of Education and Research (grants no. 01ZZ9603, 01ZZ0103, and 01ZZ0403), the Ministry of Cultural Affairs as well as the Social Ministry of the Federal State of Mecklenburg-West Pomerania, and the network ‘Greifswald Approach to Individualized Medicine (GANI_MED)’ funded by the Federal Ministry of Education and Research (grant 03IS2061A). Genome-wide data have been supported by the Federal Ministry of Education and Research (grant no. 03ZIK012) and a joint grant from Siemens Healthcare, Erlangen, Germany and the Federal State of Mecklenburg- West Pomerania. The University of Greifswald is a member of the ‘Center of Knowledge Interchange’ program of the Siemens AG and the Caché Campus program of the InterSystems GmbH.

Sorbs - This work was supported by grants from the Interdisciplinary Centre for Clinical Research at the University of Leipzig (B27 to AT, MS) from the German Diabetes Association (to AT), a Travel Grant from BIF (to AT) and by the DHFD, Diabetes Hilfs- und Forschungsfonds Deutschland (MS). We thank all those who participated in the study. Sincere thanks are given to Anke Tönjes who was significantly involved in the design and performing of the Sorbs study. We also thank Knut Krohn (Microarray Core Facility of the Interdisciplinary Centre for Clinical Research, University of Leipzig) for the genotyping support. Reedik Mägi acknowledges financial support from the European Commission under a Marie Curie Intra-European Fellowship. The research of Inga Prokopenko is funded in part through the European Community's Seventh Framework Programme (FP7/2007-2013), ENGAGE project, grant agreement HEALTH-F4-2007- 201413.

TwinsUK - The study was funded by the Wellcome Trust; European Community's Seventh Framework Programme (FP7/2007-2013)/grant agreement HEALTH-F2-2008-ENGAGE and the European Union FP-5 GenomEUtwin Project (QLG2-CT-2002-01254) and Framework 6 Project EUroClot. The study also receives support from the National Institute for Health Research (NIHR) comprehensive Biomedical Research Centre award to Guy's & St Thomas' NHS Foundation Trust in partnership with King's College London. We thank the staff from the TwinsUK, the DNA Collections and Genotyping Facilities at the Wellcome Trust Sanger Institute for sample preparation; Quality Control of the Twins UK cohort for genotyping (in particular Amy Chaney, Radhi Ravindrarajah, Douglas Simpkin, Cliff Hinds, and Thomas Dibling); Paul Martin and Simon Potter of the DNA and Genotyping Informatics teams for data handling; Le Centre National de Génotypage, France, led by Mark Lathrop, for genotyping; Duke University, North Carolina, USA, led by David Goldstein, for genotyping; and the Finnish Institute of Molecular Medicine, Finnish Genome Center, University of Helsinki, led by Aarno Palotie. Nicole Soranzo acknowledges financial support from the Wellcome Trust (Grant 091746/Z/10/Z).

UKBS2 - We acknowledge use of DNA from The UK Blood Services collection of Common Controls (UKBS-CC collection), funded by the Wellcome Trust grant 084183/Z/07/Z and by NIHR programme grant to NHSBT (RP-PG-0310-1002). The collection was established as part of the Wellcome Trust Case Control Consortium (WTCCC). We acknowledge funding from the British Heart Foundation to Augusto Rendon grant RG/09/012 and NIHR funding through the Cambridge Biomedical Research Centre.

WGHS - The WGHS is supported by HL 043851 and HL69757 from the National Heart, Lung, and Blood Institute and CA 047988 from the National Cancer Institute, the Donald W. Reynolds Foundation and the Fondation Leducq, with collaborative scientific support and funding for genotyping provided by Amgen.

WTCCC-T2D - Research funding for sample collection, genotyping and data analysis for the T2D-WTCCC and other cohorts for which the Oxford group had responsibility came from the British Diabetes Association, BDA Research, Diabetes UK, Oxford NIHR Biomedical Research Centre, European Commission (ENGAGE: HEALTH-F4-2007-201413; EURO DIA: LSHG-CT-2004-518153, Wellcome Trust (072960, 076113/B/04/Z, 076113/K/04/Z, 083270, 085301, 079557, 081682, 075491) UK Medical Research Council (G0000649, G0601261) and NIDDK

(R01-DK-073490). In addition, Cecilia Lindgren is funded by WT086596/Z/08/Z (Wellcome Trust Research Career Development Fellowship); Reedik Mägi is funded by European Commission under the Marie Curie Intra-European Fellowship; and Mark McCarthy receives personal funding from the Oxford NIHR Biomedical Research Centre.

YFS - The Young Finns Study has been financially supported by the Academy of Finland: grants 126925, 121584, 124282, 129378 (Salve), 117787 (Gendi), and 41071 (Skidi), the Social Insurance Institution of Finland, Kuopio, Tampere and Turku University Hospital Medical Funds (grant 9M048 and 9N035 for TeLeht), Juho Vainio Foundation, Paavo Nurmi Foundation, Finnish Foundation of Cardiovascular Research and Finnish Cultural Foundation, Tampere Tuberculosis Foundation and Emil Aaltonen Foundation (T.L). The expert technical assistance in the statistical analyses by Irina Lisinen is gratefully acknowledged.

Stage 2 - Insilico replication

BHS - The Busselton Health Study acknowledges the generous support for the 1994/5 follow-up study from Healthway, Western Australia and the numerous Busselton community volunteers who assisted with data collection and the study participants from the Shire of Busselton. The Busselton Health Study is supported by The Great Wine Estates of the Margaret River region of Western Australia. The BHS gratefully acknowledges the assistance of the Western Australian DNA Bank (NHMRC Enabling Facility) with DNA samples and the support provided by the Ark (NHMRC Enabling Facility) for this study.

B58C (all subsets) WTCCC/T1DGC/REPL - We acknowledge use of phenotype and genotype data from the British 1958 Birth Cohort DNA collection, funded by the Medical Research Council grant G0000934 and the Wellcome Trust grant 068545/Z/02.

(<http://www.b58cgene.sgul.ac.uk/>). Genotyping for the B58C-WTCCC subset was funded by the Wellcome Trust grant 076113/B/04/Z. The B58C-T1DGC genotyping utilized resources provided by the Type 1 Diabetes Genetics Consortium, a collaborative clinical study sponsored by the National Institute of Diabetes and Digestive and Kidney Diseases (NIDDK), National Institute of Allergy and Infectious Diseases (NIAID), National Human Genome Research Institute (NHGRI), National Institute of Child Health and Human Development (NICHD), and Juvenile Diabetes Research Foundation International (JDRF) and supported by U01 DK062418. B58C-T1DGC GWAS data were deposited by the Diabetes and Inflammation Laboratory,

Cambridge Institute for Medical Research (CIMR), University of Cambridge, which is funded by Juvenile Diabetes Research Foundation International, the Wellcome Trust and the National Institute for Health Research Cambridge Biomedical Research Centre; the CIMR is in receipt of a Wellcome Trust Strategic Award (079895). Some of the B58C-REPL genotyping was supported by a contract from the European Commission Framework Programme 6 (018996) and grants from the French Ministry of Research.

Estonian Genome Center of University of Tartu (EGCUT) - EGCUT received financing by FP7 grants (201413, 245536), grant from Estonian Government SF0180142s08, from the EU through the European Regional Development Fund, OPENGENE, in the frame of Centre of Excellence in Genomics and Univ. of Tartu grant SP1GVARENG.

HYPERGENES - funding: HYPERGENES (FP7 - HEALTH-F4-2007-201550); INTEROMICS (MIUR - CNR Italian Flagship Project). To HYPERGENES consortium took part: 1) University of Milano and Fondazione Filarete with Daniele Cusi, Project Coordinator, Cristina Barlassina, Erika Salvi, Sara Lupoli, Maurizio Marconi, Gianna Petrini, Vincenzo Toschi, Francesca Frau. 2) Katholieke Universiteit Leuven, with Jan Staessen, Jan Staessen, Tatiana Kuznetsova, Lutgarde Thijs. 3) Jagiellonian University Medical College, Krakow, with Kalina Kawecka-Jaszcz, Katarzyna Stolarz, Agnieszka Olszanecka, Wiktoria Wojciechowska. 4) IBM Israel – Science and Technology LTD, with Amnon Shabo, Ariel Frakash, Simona Cohen, Boaz Carmeli, Dan Pelleg, Michal Rosen-Zvi, Hani Neuvrith-Telem. 5) I.M.S. – Istituto di Management Sanitario S.r.l., Milan, with Pietro Conti, Costanza Conti, Mariella D’Alessio. 6) Institute of Internal Medicine, Siberian Branch of Russian Academy of Medical Sciences, Novosibirsk, with Yuri Nikitin, Sofia Malyutina, M. Voevoda, Andrew Ryabikov, E. Pello, Maxim Ryabikov. 7) Imperial College of Science, Technology and Medicine, with Paolo Vineis and Clive J Hoggart. 8) INSERM – Institut National de la Santé et de la Recherche Médicale U772, with Xavier Jeunemaitre, Pierre-François Plouin, Anne-Paule Gimenez-Roqueplo, Rosa Vargas-Poussou, Geneviève Beaurain. 9) University of Warwick. Cardiovascular Medicine & Epidemiology Group, Clinical Sciences Research Institute, with Francesco P Cappuccio, Michelle A Miller, Chen Ji. 10) Università degli Studi di Sassari. Hypertension and cardiovascular prevention centre, with Nicola Glorioso, Chiara Maria Troffa, Giuseppe Argiolas, Silvia Pitzoi. 11) STMICROELECTRONICS SRL, with Enrico Rosario Alessi. 12) Universite de Lausanne. Department of Medical Genetics, with Carlo Rivolta, Jacques S. Beckmann, Zoltan Kutalik,

Paola Benaglio. 13) Pharnext S.A.S., Paris, with Daniel Cohen and Ilya Chumakov. 14) Softeco Sismat Spa, Genova, with Stefano Bianchi. 15) Shanghai Institute of Hypertension, with Jiguang Wang and Li Yan. 16) Charles University in Prague. Department of Internal Medicine II, Pilsen, with Jan Filipovsky, Otto Mayer, Milan Hromadka, Jitka Seidlerova, Milena Dolejsova, Lukas Handl. 17) Università degli Studi di Padova. Department of Clinical and Experimental Medicine, with Edoardo Casiglia, Valerie Tikhonoff, Laura Schiavon, Anna Bascelli, Elisa Pagnin. 18) Medical University of Gdansk. Hypertension Unit, Department of Hypertension and Diabetology, with Krzysztof Narkiewicz, Marzena Chrostowska, Radoslaw Szczech, Michal Hoffmann. 19) University Vita-Salute San Raffaele, with Paolo Manunta, Chiara Lanzani, Maria Teresa Sciarrone, Lorena Citterio, Laura Zagato.

LifeLines Cohort Study - The LifeLines Cohort Study, and generation and management of GWAS genotype data for the LifeLines Cohort Study is supported by the Netherlands Organization of Scientific Research NWO (grant 175.010.2007.006), the Economic Structure Enhancing Fund (FES) of the Dutch government, the Ministry of Economic Affairs, the Ministry of Education, Culture and Science, the Ministry for Health, Welfare and Sports, the Northern Netherlands Collaboration of Provinces (SNN), the Province of Groningen, University Medical Center Groningen, the University of Groningen, Dutch Kidney Foundation and Dutch Diabetes Research Foundation. We thank Behrooz Alizadeh, Annemieke Boesjes, Marcel Bruinenberg, Noortje Festen, Ilja Nolte, Lude Franke, Mitra Valimohammadi for their help in creating the GWAS database, and Rob Bieringa, Chris Döling, Martin Elderson, Joost Keers, Elise Klaver, René Oostergo, Salome Scholtens, Rosalie Visser, Judith Vonk for their work related to data-collection and validation. The authors are grateful to the study participants, the staff from the LifeLines Cohort Study and Medical Biobank Northern Netherlands, and the participating general practitioners and pharmacists. Statistical analyses were carried out on the Genetic Cluster Computer (<http://www.geneticcluster.org>) which is financially supported by the Netherlands Scientific Organization (NWO 480-05-003) along with a supplement from the Dutch Brain Foundation and the VU University Amsterdam.

PLCO2 - The Prostate, Lung, Colorectal, and Ovarian (PLCO) Cancer Screening Trial was funded by the Intramural Research Program of the Division of Cancer Epidemiology and Genetics, National Cancer Institute, NIH

PREVEND - PREVEND genetics is supported by the Dutch Kidney Foundation (Grant E033), the EU project grant GENECURE (FP-6 LSHM CT 2006 037697), the National Institutes of Health (grant LM010098), The Netherlands organisation for health research and development (NWO VENI grant 916.761.70), and the Dutch Inter University Cardiology Institute Netherlands (ICIN). Folkert W. Asselbergs is supported by a clinical fellowship from the Netherlands Organisation for Health Research and Development (ZonMw grant 90700342).

QIMR - We are grateful to the twins and their families for their generous participation in these studies. We would like to thank staff at the Queensland Institute of Medical Research: Dixie Statham, Ann Eldridge and Marlene Grace for sample collection, Anjali Henders, Megan Campbell, Lisa Bowdler, Steven Crooks and staff of the Molecular Epidemiology Laboratory for sample processing and preparation, Dale Nyholt, Scott Gordon, Brian McEvoy, Belinda Cornes and Beben Benyamin for data QC and preparation, and David Smyth and Harry Beeby for IT support. We acknowledge funding from the Australian National Health and Medical Research Council (NHMRC grants 241944, 389875, 389891, 389892, 389938, 442915, 442981, 496739, 496688, 552485, 613672, 613601 and 1011506), the U.S. National Institute of Health (grants AA07535, AA10248, AA014041, AA13320, AA13321, AA13326 and DA12854) and the Australian Research Council (ARC grants DP0770096 and DP1093900).

RS-II, RS-III - The generation and management of GWAS genotype data for the Rotterdam Study is supported by the Netherlands Organisation of Scientific Research NWO Investments (nr. 175.010.2005.011, 911-03-012). This study is funded by the Research Institute for Diseases in the Elderly (014-93-015; RIDE2), the Netherlands Genomics Initiative (NGI)/Netherlands Organisation for Scientific Research (NWO) project nr. 050-060-810. We thank Pascal Arp, Mila Jhamai, Marijn Verkerk, Lizbeth Herrera and Marjolein Peters for their help in creating the GWAS database, and Karol Estrada and Maksim V. Struchalin for their support in creation and analysis of imputed data. The Rotterdam Study is funded by Erasmus Medical Center and Erasmus University, Rotterdam, Netherlands Organization for the Health Research and Development (ZonMw), the Research Institute for Diseases in the Elderly (RIDE), the Ministry of Education, Culture and Science, the Ministry for Health, Welfare and Sports, the European Commission (DG XII), and the Municipality of Rotterdam. The authors are grateful to the study participants, the staff from the Rotterdam Study and the participating general practitioners and pharmacists.

TRAILS - TRAILS (TRacking Adolescents' Individual Lives Survey) is a collaborative project involving various departments of the University Medical Center and University of Groningen, the Erasmus University Medical Center Rotterdam, the University of Utrecht, the Radboud Medical Center Nijmegen, and the Parnassia Bavo group, all in the Netherlands. TRAILS has been financially supported by grants from the Netherlands Organization for Scientific Research NWO (Medical Research Council program grant GB-MW 940-38-011; ZonMW Brainpower grant 100-001-004; ZonMw Risk Behavior and Dependence grants 60-60600-98-018 and 60-60600-97-118; ZonMw Culture and Health grant 261-98-710; Social Sciences Council medium-sized investment grants GB-MaGW 480-01-006 and GB-MaGW 480-07-001; Social Sciences Council project grants GB-MaGW 457-03-018, GB-MaGW 452-04-314, and GB-MaGW 452-06-004; NWO large-sized investment grant 175.010.2003.005; NWO Longitudinal Survey and Panel Funding 481-08-013); the Sophia Foundation for Medical Research (projects 301 and 393), the Dutch Ministry of Justice (WODC), the European Science Foundation (EuroSTRESS project FP-006), and the participating universities. We are grateful to all adolescents, their parents and teachers who participated in this research and to everyone who worked on this project and made it possible.

TWINGENE - This work was supported by grants from the Ministry for Higher Education, the Swedish Research Council (M-2005-1112 and 2009-2298), GenomEUtwin (EU/QLRT-2001-01254; QLG2-CT-2002-01254), NIH grant DK U01-066134, The Swedish Foundation for Strategic Research (SSF; ICA08-0047).

Stage 2 - MetaboChip replication

AMC-PAS - AMC-PAS is grateful to M.D. Trip MD, PhD and S. Sivapalaratnam MD for their input in collecting the data.

1958BC - Collection of DNA in the 1958 Birth Cohort was funded by the Medical Research Council grant G0000934 and Wellcome Trust grant 068545/Z/02. Dr Sue Ring and Dr Wendy McArdle (University of Bristol), and Mr Jon Johnson (Centre for Longitudinal Studies, Institute of Education, London) are thanked for help with data linkage. Work was undertaken at Great Ormond Street Hospital /University College London, Institute of Child Health which received a proportion of funding from the Department of Health's National Institute of Health Research

('Biomedical Research Centres' funding). The Medical Research Council provides funds for the MRC Centre of Epidemiology for Child Health.

CARDIOGENICS / THISEAS / AMC-PAS - This work was funded by the Wellcome Trust. We like to thank the members of the WTSI Genotyping Facility in particular Sarah Edkins and Cordelia Langford

CARDIOGENICS - Sample collection in the Cardiogenics Consortium (<http://www.cardiogenics.eu/web/>) was funded by the 6th Framework Program of the European Union (LSHM-CT-2006-037593) and supported through the Cambridge Bioresource which is funded by the NIHR Cambridge Biomedical research Centre. We thank all the participants and clinicians involved in the recruitment process at Cambridge and Leicester (UK), Luebeck and Regensburg (Germany), and Paris (France).

DPS - The DPS has been financially supported by grants from the Academy of Finland (117844 and 40758, 211497, and 118590; The EVO funding of the Kuopio University Hospital from Ministry of Health and Social Affairs (5254), Finnish Funding Agency for Technology and Innovation (40058/07), Nordic Centre of Excellence on Systems biology in controlled dietary interventions and cohort studies, SYSDIET (070014), The Finnish Diabetes Research Foundation, Yrjö Jahnsson Foundation (56358), Sigrid Juselius Foundation, Juho Vainio Foundation and TEKES grants 70103/06 and 40058/07.

DILGOM - The DILGOM study has been funded by the Academy of Finland (grant numbers 139635, 129494, 118065, 129322, 250207), the Orion-Farmos Research Foundation, the Finnish Foundation for Cardiovascular Research, and the Sigrid Jusélius Foundation.

DR's EXTRA - The DR.s EXTRA Study was supported by grants to R. Rauramaa by the Ministry of Education and Culture of Finland (627;2004-2011), Academy of Finland (102318; 123885), Kuopio University Hospital, Finnish Diabetes Association, Finnish Heart Association, Päivikki and Sakari Sohlberg Foundation and by grants from European Commission FP6 Integrated Project (EXGENESIS); LSHM-CT-2004-005272, City of Kuopio and Social Insurance Institution of Finland (4/26/2010).

Ely - The MRC Ely Study was funded by the Medical Research Council and the Wellcome Trust. We are most grateful to all study participants and to the staff of the St. Mary's Street Surgery, Ely. We thank all the staff who worked on the study.

EPIC - The EPIC Norfolk Study is funded by Cancer Research United Kingdom and the Medical Research Council.

FIN-D2D 2007 - The FIN-D2D study has been financially supported by the hospital districts of Pirkanmaa, South Ostrobothnia, and Central Finland, the Finnish National Public Health Institute (current National Institute for Health and Welfare), the Finnish Diabetes Association, the Ministry of Social Affairs and Health in Finland, the Academy of Finland (grant number 129293), Commission of the European Communities, Directorate C-Public Health (grant agreement no. 2004310) and Finland's Slottery Machine Association.

FENLAND - The Fenland Study is funded by the Wellcome Trust and the Medical Research Council, as well as by the Support for Science Funding programme and CamStrad. We are grateful to all the volunteers for their time and help, and to the General Practitioners and practice staff for help with recruitment. We thank the Fenland Study co-ordination team and the Field Epidemiology team of the MRC Epidemiology Unit for recruitment and clinical testing.

GLACIER - The GLACIER Study was funded by grants from the Swedish Diabetes Association, Swedish Heart-Lung Foundation, Swedish Research Council, Medical Research Foundation of Umeå University, and Novo Nordisk (all to PWF). Genotyping for the GLACIER Study was funded by the Wellcome Trust. We thank the participants for their outstanding contributions to the GLACIER Study. We also thank the staff of the Umeå Medical Biobank, especially Göran Hallmans, Åsa Agren, John Huttilainen, and Ann-Marie Ahren for data retrieval and organisation and Kerstin Enquist and Tore Johansson for expert assistance with DNA extraction and plating. The GLACIER Study is nested within the Västerbottens Intervention Project (VIP); we thank the staff of the VIP Study for phenotype data collection, particularly Lars Wennhall who leads the VIP Study. Inês Barroso acknowledges funding from the Wellcome Trust grant 077016/Z/05/Z, United Kingdom NIHR Cambridge Biomedical Research Centre and the MRC Centre for Obesity and Related Metabolic Diseases. We would like to thank Emma Gray, Douglas Simpkin, Sarah Hunt and staff of the WTSI Sample Logistics, Genotyping and Variation Informatics Facilities.

Go-DARTS (DUNDEE) - We acknowledge the support of the Health Informatics Centre, University of Dundee for managing and supplying the anonymised data and NHS Tayside, the original data owner. We are grateful to all the participants who took part in the Go-DARTS study, to the general practitioners, to the Scottish School of Primary Care for their help in

recruiting the participants, and to the whole team, which includes interviewers, computer and laboratory technicians, clerical workers, research scientists, volunteers, managers, receptionists, and nurses. The Wellcome Trust provides support for Wellcome Trust United Kingdom Type 2 Diabetes Case Control Collection (Go-DARTS) and the Scottish Health Informatics Programme. Further informatics support is provided by the Chief Scientist Office of Scotland. This work was also supported by the UK Medical Research Council (G0601261)

HNR - We thank the Heinz Nixdorf Foundation, Germany, and Deutsche Forschungsgemeinschaft (project ER 155/6-2) for the generous support of this study. We acknowledge the support of the Sarstedt AG & Co. (Nümbrecht, Germany) concerning laboratory equipment. We are grateful to Prof. Raimund Erbel (Clinic of Cardiology, West German Heart Centre, University Hospital of Essen, University Duisburg-Essen, Germany), Prof. Dr. Susanne Moebus (Institute for Medical Informatics, Biometry and Epidemiology (IMIBE), University Hospital of Essen, University of Duisburg-Essen, Essen, Germany) and Prof. Dirk Schadendorf (Clinic Department of Dermatology, University Hospital Essen, University Duisburg-Essen, Essen, Germany) for funding this study.

HUNT2 - The Nord-Trøndelag Health Study (The HUNT Study) is a collaboration between HUNT Research Centre (Faculty of Medicine, Norwegian University of Science and Technology NTNU), Nord-Trøndelag County Council, Central Norway Health Authority, and the Norwegian Institute of Public Health.

IMPROVE - The IMPROVE study was funded by the European Commission (Contract number: QLG1-CT-2002-00896), the Swedish Heart-Lung Foundation, the Swedish Research Council (8691), the Knut and Alice Wallenberg Foundation, the Foundation for Strategic Research, the Torsten and Ragnar Söderberg Foundation, the Strategic Cardiovascular Programme of Karolinska Institutet and the Stockholm County Council, the Strategic support for epidemiological research at Karolinska Institutet and the Stockholm County Council and the Stockholm County Council (560183).

KORA S3 and S4 - The KORA Augsburg studies were financed by the Helmholtz Zentrum München, German Research Center for Environmental Health, Neuherberg, Germany and supported by grants from the German Federal Ministry of Education and Research (BMBF). Part of this work was financed by the German National Genome Research Network (grant number

NGFNPLUS 01GS0823). Our research was supported within the Munich Center of Health Sciences (MC Health) as part of LMUinnovativ.

LURIC - LURIC received funding through the 6th Framework Program (integrated project Bloodomics, grant LSHM-CT-2004-503485) and 7th of Framework Program (integrated project AtheroRemo, Grant Agreement number 201668) of the European Union. The authors extend appreciation to the participants of the LURIC study without their collaboration this article would not have been written. We thank the LURIC study team either temporarily or permanently involved in patient recruitment, sample and data handling, and the laboratory staff at the Ludwigshafen General Hospital and the Universities of Freiburg and Ulm, Germany.

METSIM - The METSIM study was funded by the Academy of Finland (grants no. 77299 and 124243).

MORGAM - The MORGAM study was part funded through the European Community's Sixth Framework Programme Cardiogenics project, grant agreement LSHM-CT-2006-037593 and Seventh Framework Programme ENGAGE project, grant agreement HEALTH-F4-2007-201413. Sites and key personnel of contributing MORGAM Centres: Finland) FINRISK, National Institute for Health and Welfare, Helsinki: V. Salomaa (principal investigator), A. Juolevi, E. Vartiainen, P. Jousilahti; ATBC, National Institute for Health and Welfare, Helsinki: J. Virtamo (principal investigator), H. Kumpulainen; MORGAM Data Centre, National Institute for Health and Welfare, Helsinki: K. Kuulasmaa (responsible person), Z. Cepaitis, A. Haukijärvi, B. Joseph, J. Karvanen, S. Kulathinal, M. Niemelä, O. Saarela; MORGAM Central Laboratory, National Institute for Health and Welfare, Helsinki: M. Perola (responsible person), P. Laiho, M. Sauramo. The ATBC Study was supported by US Public Health Service contracts N01-CN-45165, N01-RC-45035 and N01-RC-37004 from the National Cancer Institute. France) National Coordinating Centre, National Institute of Health and Medical Research (U258), Paris: P. Ducimetière (national coordinator), A. Bingham; PRIME/Strasbourg, Department of Epidemiology and Public Health, EA 3430, University of Strasbourg, Faculty of Medicine, Strasbourg: D. Arveiler (principal investigator), B. Haas, A. Wagner; PRIME/Toulouse, Department of Epidemiology, Toulouse University School of Medicine, Toulouse: J. Ferrières (Principal Investigator), J-B. Ruidavets, V. Bongard, D. Deckers, C. Saulet, S. Barrere; PRIME/Lille, Department of Epidemiology and Public Health, INSERM U744-Université Lille Nord de France – Institut Pasteur de Lille: P. Amouyel (principal investigator), M. Montaye, B.

Lemaire, S. Beauchant, D. Cotel, C. Graux, N. Marecaux, C. Steclebout, S. Szeremeta; MORGAM Laboratory, INSERM U937, Paris: F. Cambien (responsible person), L. Tiret, V. Nicaud. Italy) Centro Ricerche EPIMED - Epidemiologia e Medicina Preventiva, Dipartimento di Medicina Sperimentale. Università degli Studi dell'Insubria, Varese: M. Ferrario (principal investigator), G. Veronesi. Research Centre on Public Health, University of Milano-Bicocca, Monza, Italy: Giancarlo Cesana. This study was supported by the Health Administration of Regione Lombardia [grant numbers 9783/1986, 41795/1993, 31737/1997 and 17155/2004], for the baseline examinations and the follow-up. United Kingdom) PRIME/Belfast, Queen's University Belfast, Belfast, Northern Ireland: F. Kee (principal investigator) A. Evans (former principal investigator), J. Yarnell, E. Gardner; MORGAM Coordinating Centre, Queen's University Belfast, Belfast, Northern Ireland: A. Evans (MORGAM coordinator), S. Cashman, F. Kee. MORGAM Management Group: A. Evans (chair, Belfast, UK), S. Blankenberg (Hamburg, Germany), F. Cambien (Paris, France), M. Ferrario (Varese, Italy), K. Kuulasmaa (Helsinki, Finland), A. Palotie (Cambridge, UK), M. Perola (Helsinki, Finland), A. Peters (Neuherberg, Germany), V. Salomaa (Helsinki, Finland), H. Tunstall-Pedoe (Dundee, Scotland), P.G. Wiklund (Umeå, Sweden); Previous members: K. Asplund (Stockholm, Sweden), L. Peltonen (Helsinki, Finland), D. Shields (Dublin, Ireland), B. Stegmayr (Umeå, Sweden).

NSHD - This work was funded by the Medical Research Council [Unit Programme number U123092720]. We are very grateful to the members of this birth cohort for their continuing interest and participation in the study.

PIVUS - This project was supported by grants from the Swedish Research Council, the Swedish Heart-Lung Foundation, the Swedish Foundation for Strategic Research, the Royal Swedish Academy of Sciences, Swedish Diabetes Foundation, Swedish Society of Medicine, and Novo Nordisk Fonden. Genotyping was performed by the SNP&SEQ Technology Platform in Uppsala (www.genotyping.se). We thank Tomas Axelsson, Ann-Christine Wiman and Caisa Pöntinen for their excellent assistance with genotyping. The SNP Technology Platform is supported by Uppsala University, Uppsala University Hospital and the Swedish Research Council for Infrastructures.

STR - This work was supported by grants from the US National Institutes of Health (AG028555, AG08724, AG04563, AG10175, AG08861), the Swedish Research Council, the Swedish Heart-Lung Foundation, the Swedish Foundation for Strategic Research, the Royal Swedish Academy

of Science, and ENGAGE (within the European Union Seventh Framework Programme, HEALTH-F4-2007-201413). Genotyping was performed by the SNP&SEQ Technology Platform in Uppsala (www.genotyping.se). We thank Tomas Axelsson, Ann-Christine Wiman and Caisa Pöntinen for their excellent assistance with genotyping. The SNP Technology Platform is supported by Uppsala University, Uppsala University Hospital and the Swedish Research Council for Infrastructures.

T2D WTCCC - Research funding for sample collection, genotyping and data analysis for the T2D-WTCCC and other cohorts for which the Oxford group had responsibility came from the British Diabetes Association, BDA Research, Diabetes UK, Oxford NIHR Biomedical Research Centre, European Commission (ENGAGE: HEALTH-F4-2007-201413; EURODIA: LSHG-CT-2004-518153, Wellcome Trust (072960, 076113/B/04/Z, 076113/K/04/Z, 083270, 085301, 079557, 081682, 075491) UK Medical Research Council (G0000649,G0601261) and NIDDK (R01-DK-073490). In addition, Cecilia Lindgren is funded by WT086596/Z/08/Z (Wellcome Trust Research Career Development Fellowship); Reedik Mägi is funded by European Commission under the Marie Curie Intra-European Fellowship; and Mark McCarthy receives personal funding from the Oxford NIHR Biomedical Research Centre.

THISEAS - Recruitment for THISEAS was partially funded by a research grant (PENED 2003) from the Greek General Secretary of Research and Technology; we thank all the dieticians and clinicians for their contribution to the project.

Tromsø 4 - University of Tromsø, Norwegian Research Council (project number 185764)

ULSAM - This project was supported by grants from the Swedish Research Council, the Swedish Heart-Lung Foundation, the Swedish Foundation for Strategic Research, the Royal Swedish Academy of Sciences, Swedish Diabetes Foundation, Swedish Society of Medicine, and Novo Nordisk Fonden. Genotyping was performed by the SNP&SEQ Technology Platform in Uppsala (www.genotyping.se). We thank Tomas Axelsson, Ann-Christine Wiman and Caisa Pöntinen for their excellent assistance with genotyping. The SNP Technology Platform is supported by Uppsala University, Uppsala University Hospital and the Swedish Research Council for Infrastructures.

Whitehall II - The WHII study has been supported by grants from the Medical Research Council; Economic and Social Research Council; BHF; Health and Safety Executive; Department of Health; National Heart Lung and Blood Institute (HL36310), US, NIH: National

Institute on Aging (AG13196), US, NIH; Agency for Health Care Policy Research (HS06516); and the John D and Catherine T MacArthur Foundation Research Networks on Successful Midlife Development and Socioeconomic Status and Health. Genotyping in WHII was supported by BHF grant PG/07/133/24260

Other contributing studies: clinically extremes

Essen Obesity Study (Essen Case-Control GWAS & Essen Obesity Trio GWAS) - We thank all the participants of this study. This work was supported by grants from the Federal Ministry of Education and Research (BMBF: 01GI0823; NGFN-plus: 01GS0820, 01GS0830, 01KU0903) and the Deutsche Forschungsgemeinschaft (DFG; HE 1446/4-1).

French Extreme Obesity Study - The study was supported by le Conseil Régional Nord Pas de Calais/FEDER and the Agence Nationale de la Recherche. D.M. is funded by a Canada Research Chair. We are indebted to all subjects who participated in these studies.

GEO-IT - Financial support has been provided by the Italian Health Ministry. Istituto Auxologico Italiano: S. Mai, S. Maestrini, M. Mencarelli. Monica/Brianza Research Group : G. Cesana, P. Brambilla, M. Ferrario, R. Sega, C. Menni and Lombardy Health Directorate. Brianza cohort study was mainly funded by the Health Administration of Regione Lombardia, Italy.

GOYA - This study was conducted as part of the activities of the Danish Obesity Research Centre (DanORC, www.danorc.dk) and the MRC centre for Causal Analyses in Translational Epidemiology (MRC CAiTE). LP was supported and the genotyping for GOYA funded by the Wellcome Trust (WT 084762). LP and DME were supported by a Medical Council New Investigator Award (MRC G0800582 to DME). GOYA is a nested study within The Danish National Birth Cohort which was established with major funding from the Danish National Research Foundation. Additional support for this cohort has been obtained from the Pharmacy Foundation, the Egmont Foundation, The March of Dimes Birth Defects Foundation, the Augustinus Foundation, and the Health Foundation.

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