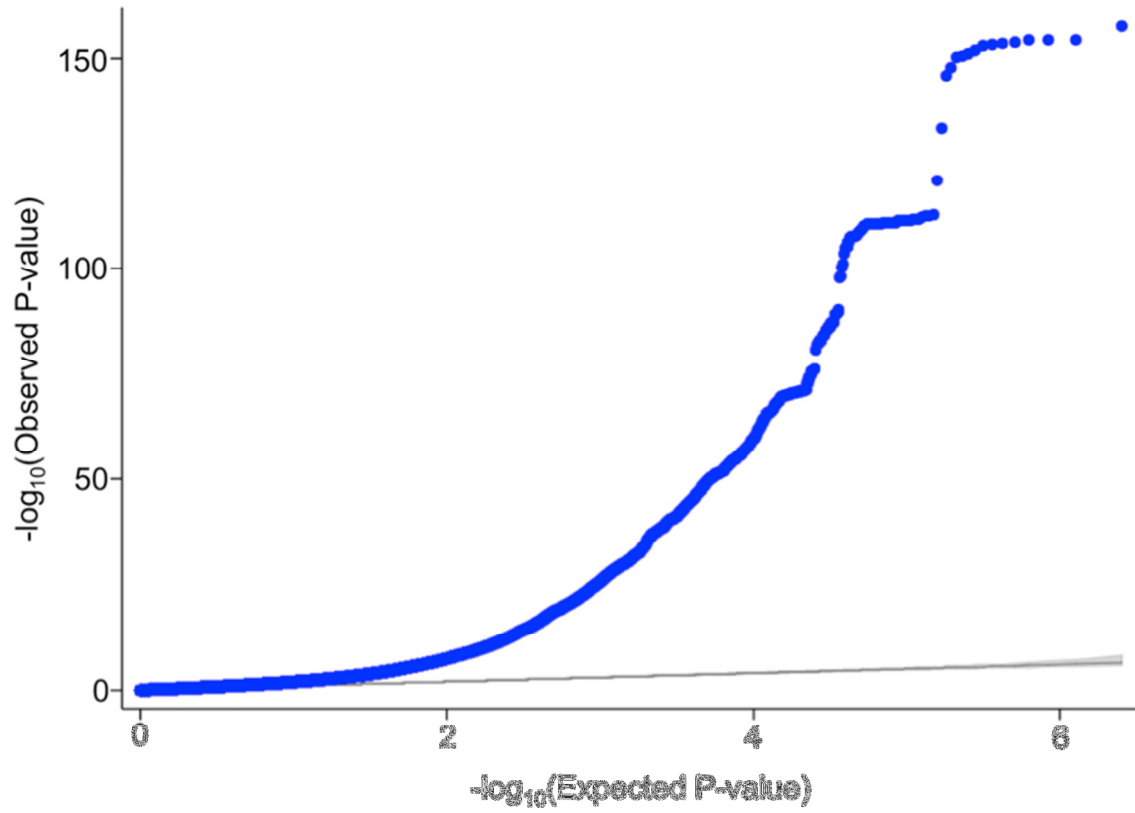


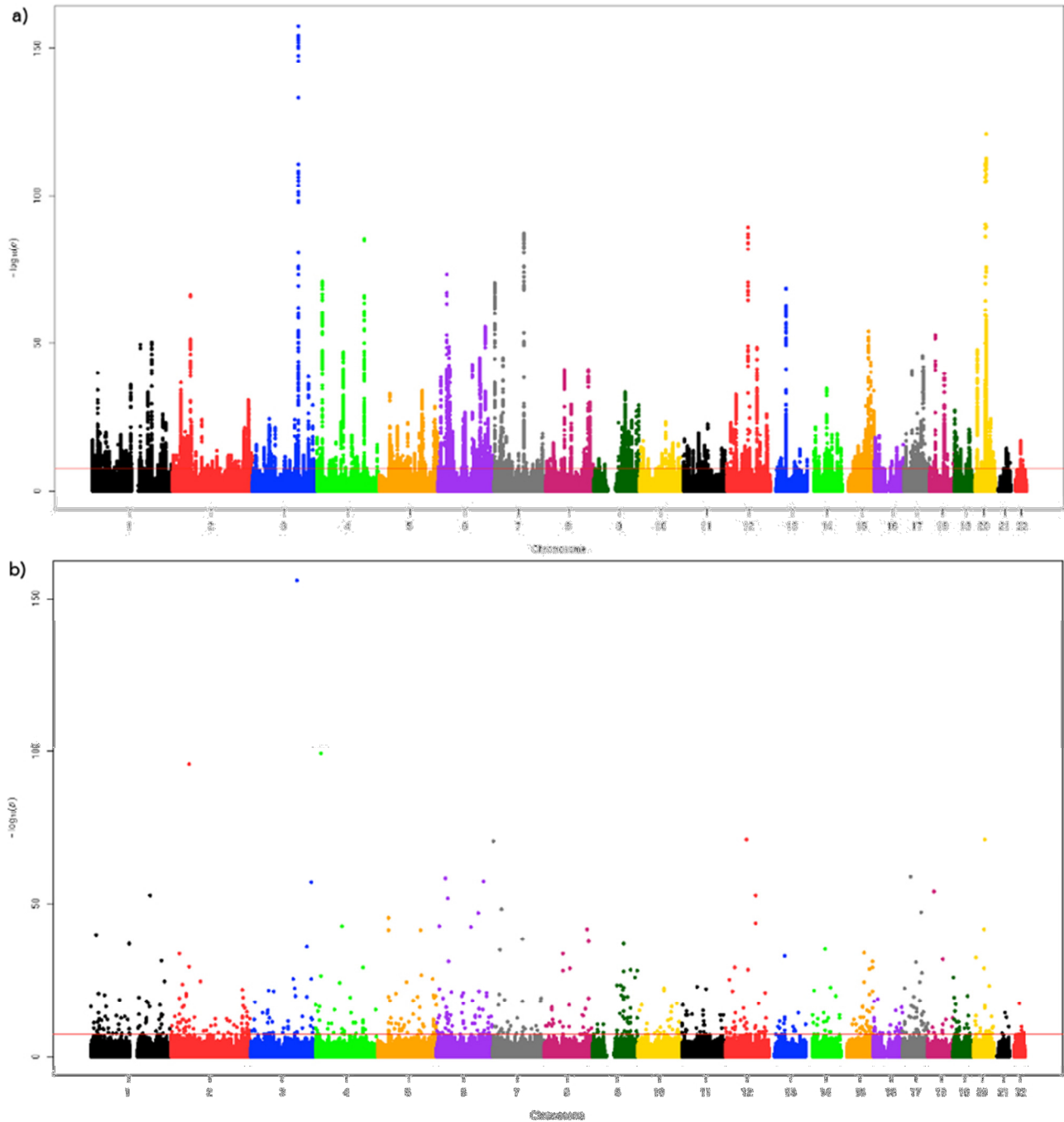
Supplementary Figure 1

Overview of the analysis strategy implemented.



Supplementary Figure 2

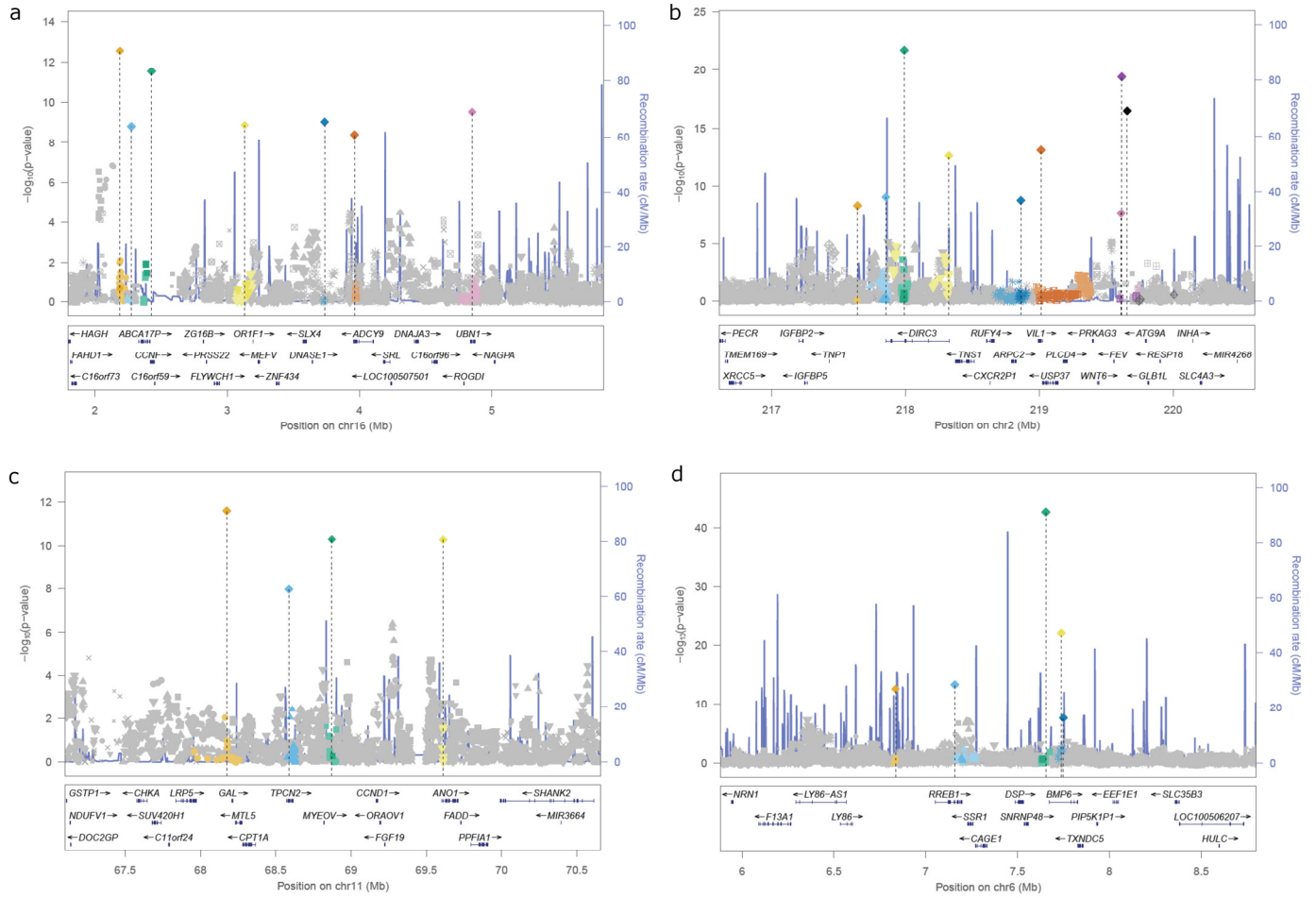
Quantile-quantile plot showing the P -value distribution of GWAS meta-analysis results after applying a single genomic control.



Supplementary Figure 3

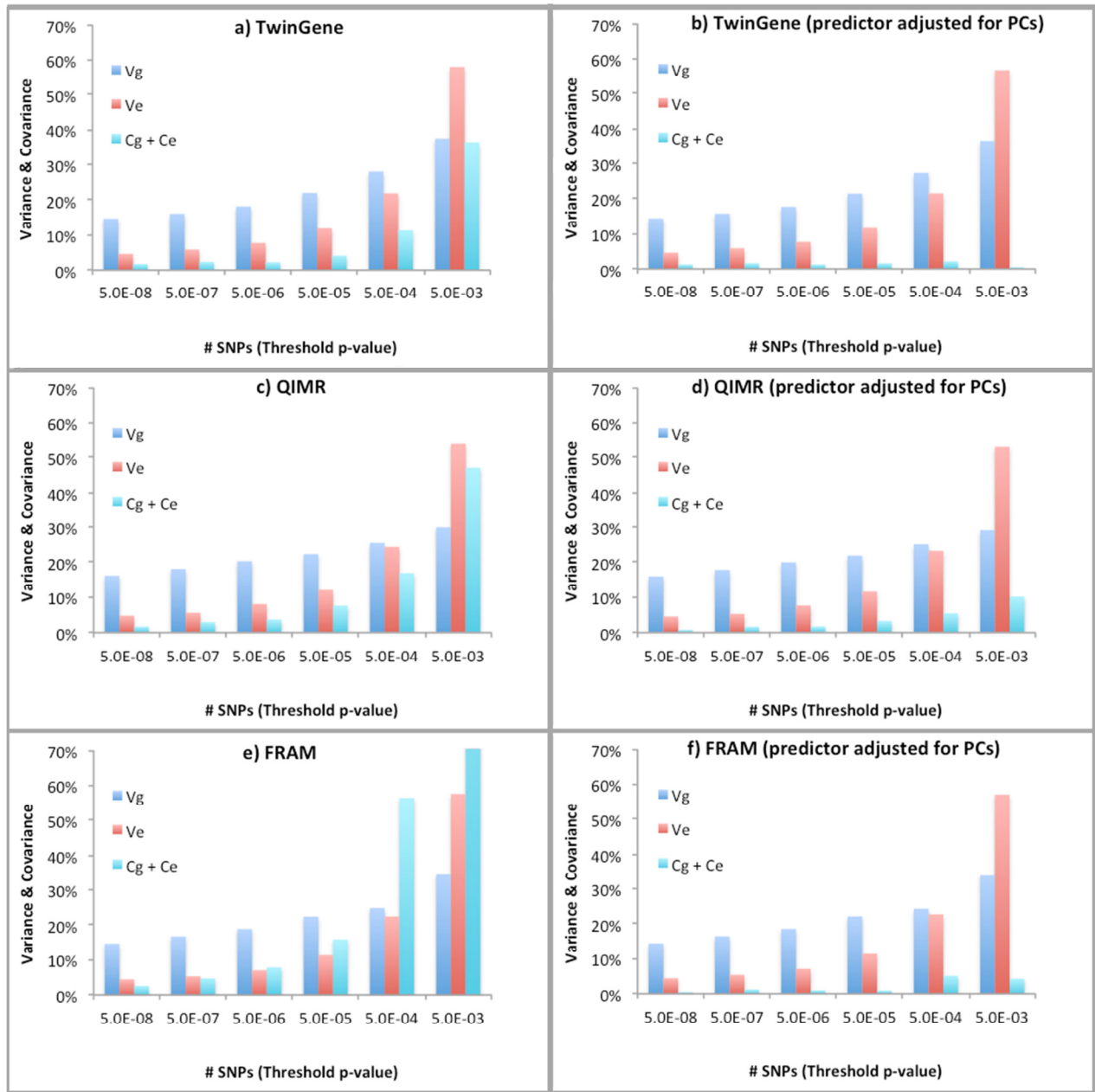
Manhattan plots.

Plots of (a) results obtained from meta-analysis after applying a single genomic control correction and (b) results obtained from performing an approximate conditional and joint multiple-SNP analysis on the meta-analysis results. The red horizontal line in both plots represents the genome-wide significance threshold of $P = 5 \times 10^{-8}$.



Supplementary Figure 4

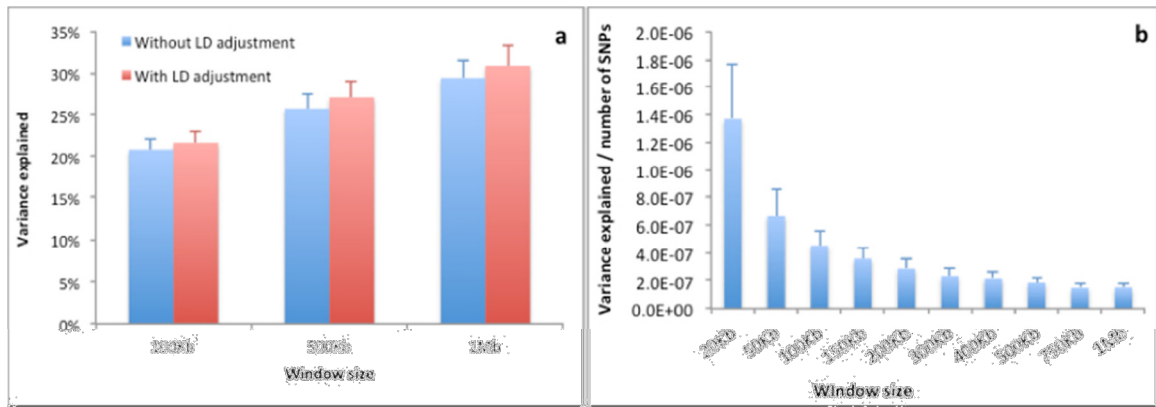
Examples of loci with multiple signals that cluster around the same gene or are in close proximity to others.



Supplementary Figure 5

Partitioning the variance in the SNP-derived genetic predictor using a within-family analysis.

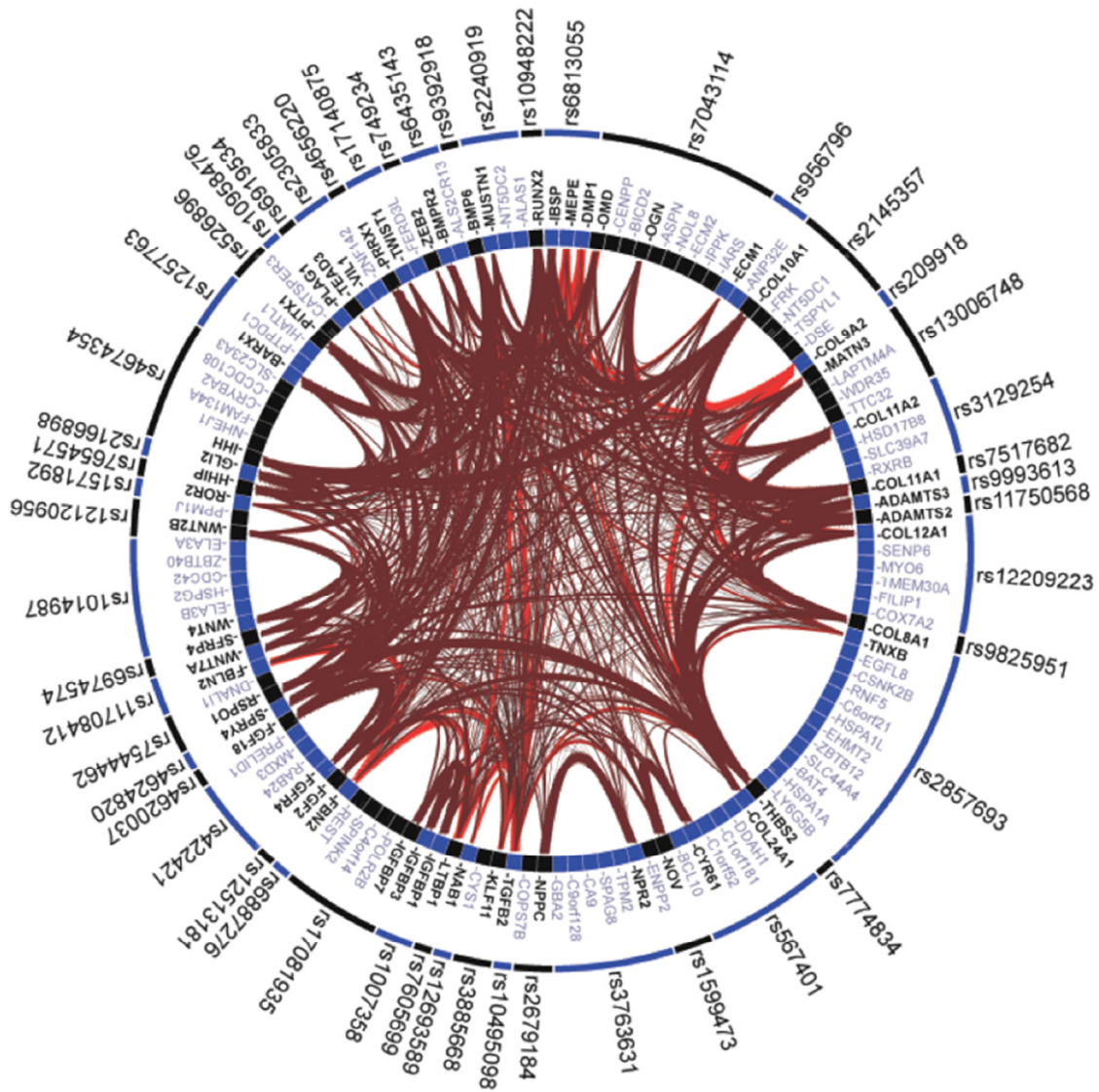
The variance of the SNP-based genetic predictor was partitioned into components due to real SNP effects (V_g), errors in estimating SNP effects (V_e) and population stratification ($C_g + C_e$) (Online Methods). The SNPs were selected from the approximate conditional and joint multiple-SNP association analysis with the target cohort being excluded from the meta-analysis. In **b**, **d** and **f**, the SNP-based predictor was adjusted by the first 20 principal components (PCs). The comparison between the results with and without PC adjustment demonstrates clearly the partitioning of the variance component due to population stratification ($C_g + C_e$).



Supplementary Figure 6

Variance explained by the SNPs at known loci.

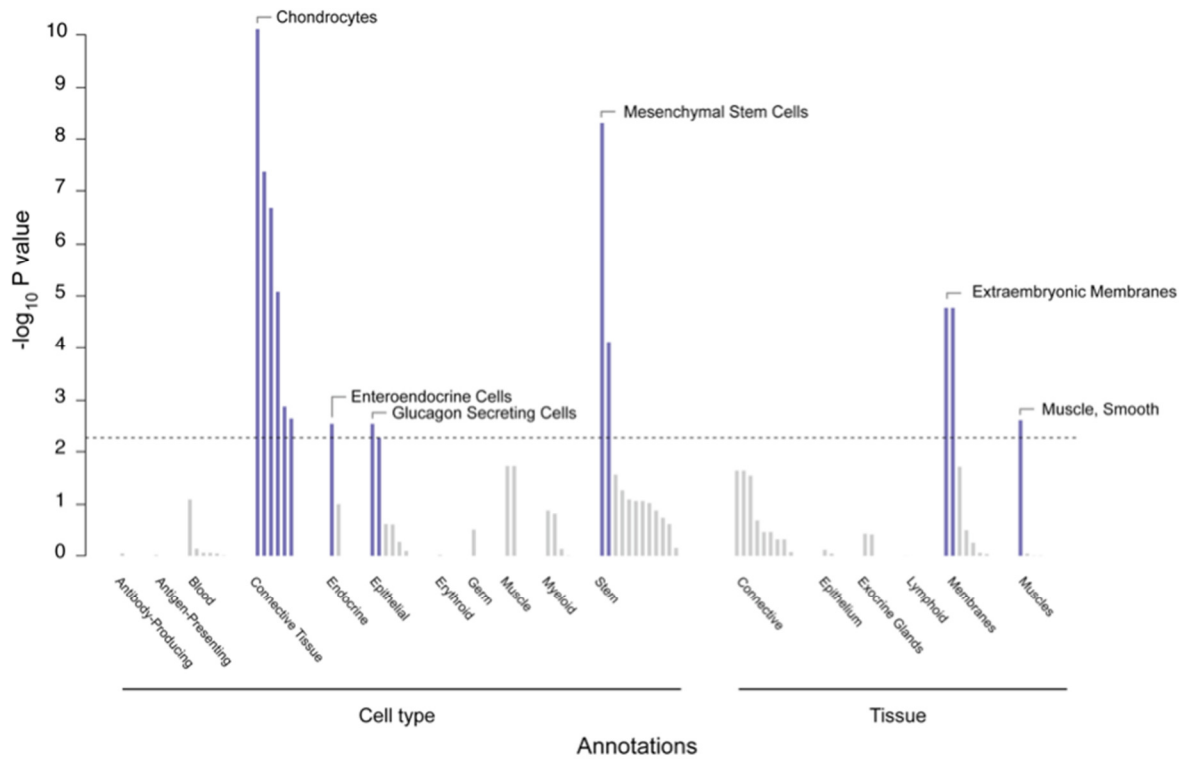
Details of data and analyses can be found in the Online Methods. **(a)** Variance explained by all the SNPs within a certain physical distance of the top associated SNPs (including the top SNPs). **(b)** Variance explained by the SNPs at the known loci excluding the top SNPs divided by the number of these SNPs. Estimates in **b** are without LD adjustment. Error bars are the standard errors of the estimates.



Supplementary Figure 7

Results from GRAIL analysis.

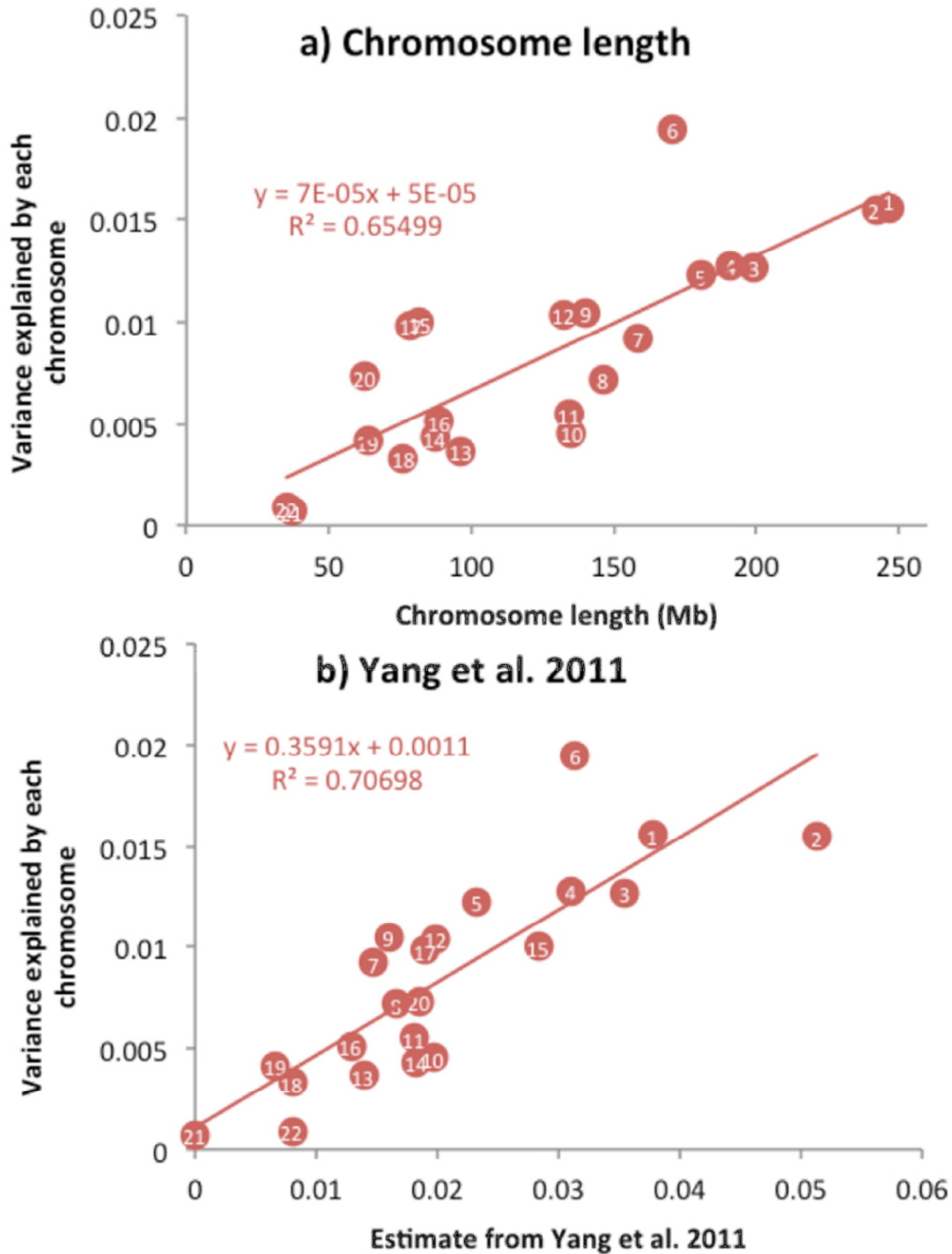
A subset of the 697 lead height SNPs are arranged along the outer circle alternating with colors. The inner circle represents the individual prioritized ($P < 1 \times 10^{-6}$) genes. Gene names shown in black have literature connections, and the ones in gray do not. The redness and thickness of the lines connecting pairs of genes represent the strength of the connections. Top-ranking keywords prioritized by GRAIL were 'transcription', 'growth', 'nuclear', 'factor', 'binding', 'collagen', 'differentiation', 'promoter', 'development', 'ribosomal', 'bone', 'mice', 'expression' and 'cartilage'.



Supplementary Figure 8

DEPICT cell type enrichment analysis.

Genes in associated height loci tended to be highly expressed in, among other cell types, chondrocytes and mesenchymal stem cells. The analysis was conducted based on the DEPICT method and 37,427 human microarray samples. Significantly enriched (FDR < 0.05) cell types are color coded in violet. The figure also includes tissue enrichment results, which were not shown in **Figure 3** of the main text.



Supplementary Figure 9

Partitioning the variance explained by chromosome length.

- (a) The proportion of variance explained by the genome-wide significant SNPs on a chromosome plotted against chromosome length.
 (b) The correlation of the variance explained by the top GWAS SNPs on each chromosome against the estimate from Yang *et al.* (2011) for that chromosome. Note: the numbers in red circles indicate chromosome number.

Defining the role of common variation in the genomic and biological architecture of adult human height

Andrew R Wood, Tonu Esko, Jian Yang, Sailaja Vedantam, Tune H Pers, *et al.* *

*A full list of author names appears in the main paper.

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Supplementary Table 2. Results from cluster analyses of multiple signals using different sized windows around index SNPs. These results are based on 10,000 SNP sets matched for allele frequency, locus gene density and distance to nearest gene but otherwise randomly selected from across the genome. The table provides a breakdown of the number of loci the 697 SNPs fit into based on different locus size definitions and the clustering of SNP pairs among the 697 within varying genomic distances.

Locus definition	# Loci 697 SNPs fit into	Average # loci that random 697 SNPs fall into	5% - 95% confidence intervals around average # loci for random SNPs	P-value
+/- 1.25 Mb	399	462	445-480	<0.0001
+/- 1 Mb	423	496	479-513	<0.0001
+/- 500 kb	502	574	559-589	<0.0001
+/- 250 kb	563	624	611-636	<0.0001
+/- 100 kb	623	660	650-669	<0.0001
Distance range	# SNP-pairs within distance	Average # random SNP pairs within distance	Range of random SNP pairs within distance	P-value
50kb to 100kb	55	37	24-53	<0.0001
100kb to 200kb	63	47	31-69	0.0014
200kb to 300kb	53	44	29-64	0.032
300kb to 400kb	49	42	27-61	0.081
400kb to 500kb	44	41	27-58	0.25
500kb to 600kb	39	39	26-57	0.61
600kb to 700kb	38	38	26-56	0.57
700kb to 800kb	39	37	24-54	0.38
800kb to 900kb	34	36	24-56	0.81
900kb to 1Mb	39	36	23-51	0.27
+/- 1Mb	274	200	162-243	<0.0001
100kb to 1Mb	222	186	144-277	0.0002
250kb to 1Mb	162	150	114-182	0.12
100kb to 500kb	143	108	78-138	<0.0001
250kb to 500kb	83	72	48-96	0.053
500kb to 1Mb	101	101	74-135	0.51

Supplementary Table 4. Genomic inflation factors derived from performing linear mixed model association analyses. Lambda inflation factors from subsets of data analysed using genomic relationship matrices and used to validate inflated statistics from the main meta-analysis. All studies performed leave one chromosome out (genomic relationship matrix GRM-LOO) analyses as well as using data from all chromosomes to calculate genomic relationship matrices (GRM-ALL) except WGHS, whose large size precluded the generation of 22 separate matrices, and who performed an odd-even analysis (GRM-O/E). Results are compared to those from standard single GC corrected GWAS (Std single GC GWAS). Meta-analysis lambdas in square brackets indicate the values from scaling the result in the 59,380 samples to a total of 250,000 individuals.

Study	N	EMMAX-ALL	EMMAX-LOO or O/E	Std Single GC GWAS	In Main GWAS Meta-analysis
COLAUS	5,410	0.989	1.031	1.029	Yes
DGI	2,407	1.002	1.035	1.037	Yes
EGCUT (OMNI)	1,293	0.997	1.014	1.085	Yes
EPIC	3,585	1.005	1.036	1.042	Yes
FENLAND	1,402	0.996	1.023	1.056	Yes
FUSION	2,249	0.994	1.034	1.184	Yes
PIVUS	948	0.987	1.002	1.011	No
InCHIANTI	1,138	0.99	1.004	1.051	Yes
MIGEN	2,659	1.002	1.018	1.009	Yes
QIMR	3,672	0.994	1.022	1.02	Yes
RISC	1,106	1.001	1.008	1.014	Yes
RSI	5,373	1.004	1.062	1.088	Yes
TWINGENE	5,668	1.001	1.055	1.052	Yes
ULSAM	1,116	0.995	1.01	1.031	No
WGHS*	21,354	0.974	1.07	1.13	No
Meta-analysis of 14 Studies (without WGHS)					
GC M/A	38,026	1.16	1.21	1.2	
Meta-analysis of 15 Studies (with WGHS)					
GC M/A	59,380	1.2 [1.83]	1.29 [2.21]	1.23 [1.94]	

Supplementary Table 5. Family based analysis and quantification of over-estimation (“Winner’s Curse”) effects in discovery GWAS. The discovery meta-analysis as repeated, excluding either a set of family-based studies or, in turn, three random sets of non-family-based studies (with total sample sizes matched to the effective sample size of the combined family-based studies). The multiple loci in each meta-analysis were selected after clumping of SNPs based on $r^2 < 0.01$ and 1 Mb distance windows, and effect sizes were estimated. The power to replicate in the excluded samples was calculated at different P -value thresholds using the effect size estimates and the effective sample size. The estimated deficit in replication due to the winner’s curse is the average deficit in observed, compared with expected replications for the three sets of random non-family-based studies, after adjustment to match the number of significant SNPs (416) seen in the meta-analysis that excluded the family-based samples. The upper limit of false positives due to stratification is calculated as the difference between the deficit observed in the non-family-based samples (attributable to winner's curse alone) and the deficit of replication seen in the family-based samples (attributable to winner’s curse plus stratification).

Threshold	Family Based N=14,963,SNPs=416		Random cohort Set1 N=14897,SNPs=416		Random cohort Set2 N=15,648,SNPs=423		Random cohort Set3 N=16,462,SNPs=435		*Deficit in replication from winner's curse	**Deficit in replication from stratification	***Upper limit of completely false positives from stratification
	Predicted	Observed	Predicted	Observed	Predicted	Observed	Predicted	Observed			
Same Direction	399.98	371	404.88	386	408.23	386	422.03	412	16.78	12.20	24.45
P < 0.05	209.63	142	233.15	169	208.57	162	229.05	186	50.37	17.26	17.7
P < 0.01	127.99	82	147.7	112	120.07	85	137.09	112	31.39	14.60	14.67

* At each P value threshold, winner’s curse calculated as the average deficit of predicted vs. observed replicated signals in 3 random cohorts after adjusting to match the same number of 416 SNPs as that of family based studies (see Supplementary Methods 1.5.2).

**Deficit due to stratification calculated as the difference in expected minus the observed replications in family based studies that cannot be explained by winner's curse.

***Upper limit of completely false positives from stratification calculated as the number of false positive loci if the deficit in replication attributable to stratification is completely explained by a set of false positive loci, with no effect size inflation from stratification in the remaining loci (see Supplementary Methods).

Supplementary Table 6. Within family association analysis of the variants identified in the discovery GWAS. The directional concordance of the SNPs that could be analyzed within each study and after meta-analysis is shown below.

Study	SNPs	# Same Direction	% Concordant	Effective N
SardiNIA	238	160	67	3743
QIMR	416	319	77	3426
TWINGENE	415	303	73	2766
Amish	403	257	64	399
Framingham	414	310	75	2920
ERF	385	227	59	634
FamHS	416	301	72	1949
Meta Analysis	416	371	89	14963

Supplementary Table 9. Overlap with genes involved in abnormal skeletal growth syndromes. List of 266 abnormal skeletal/growth genes identified in the OMIM database (<http://www.ncbi.nlm.nih.gov/omim>) using the following keywords: short stature, overgrowth, skeletal dysplasia, brachydactyly. Of these, 39 were prioritised by DEPICT and are highlighted.

*ACAN	CTDP1	GLI3	NBN	SHH
ACP5	CTSK	*GNAS	NEU1	SHOX
*ADAMTS10	CUL4B	GNPAT	NF1	SHROOM4
*ADAMTS2	CUL7	GNPTAB	NFIX	SIL1
ADAMTSL2	CYP11B1	GPC3	*NIPBL	SLC26A2
AGPS	*CYP19A1	GPC6	NKX3-2	SLC29A3
ALG12	CYP21A2	GUSB	NOG	SLC2A2
ALMS1	CYP27B1	HCCS	*NPPC	SLC34A3
ALPL	DDR2	HDAC4	*NPR2	SLC35C1
ANKH	DHCR7	*HES7	*NSD1	*SLC35D1
ARL6	*DYM	HESX1	OBSL1	SLC37A4
ARSB	DYNC2H1	*HMGA2	OCRL	SLC39A13
ARSE	EBP	HOXD13	OFD1	SLC4A4
ATP6V0A2	EFNB1	HPRT1	PAPSS2	SLC6A8
ATP7A	*EIF2AK3	HRAS	PAX3	SMARCAL1
ATP8B1	ERCC2	HSPG2	PAX8	SMC1A
ATR	ERCC3	HYAL1	PCNT	SMC3
ATRX	ESCO2	ICK	PEX7	SMPD1
B3GALTL	*ESR1	IDUA	PHEX	SMS
B4GALT7	EVC	IFT122	PHF6	SNAP47
BBS1	EVC2	IFT80	PITX2	SOS1
BBS10	EXT1	IGBP1	POU1F1	SOST
BBS12	EXT2	IGF1	PQBP1	SOX3
BBS2	FAM123B	*IGF1R	PROP1	*SOX9
BBS4	FAM20C	IGF2	*PTCH1	SPG20
BBS5	*FANCA	IHH	PTCH2	SRY
BBS7	FANCB	IKBKKG	PTEN	STAT5B
BBS9	FANCC	JAG1	PTH1R	TAZ
BMPR1B	FANCD2	KCNJ2	*PTHLH	TBCE
BRAF	FANCE	KDM5C	PTPN11	TBX1
BRCA2	FANCF	KIAA1279	RAB23	*TBX15
BTK	FANCG	KRAS	RAB3GAP1	TCF4
BUB1B	*FANCI	LBR	RAB3GAP2	TGFBR1
C7orf11	FANCL	LEMD3	*RAF1	TGFBR2
CA2	FANCM	LEPRE1	RAI1	THRB
CCDC28B	*FBN1	LFNG	RBM28	TNFRSF11B
CEP290	*FBN2	LHX4	RECQL4	TP63
CHD7	FGD1	LIFR	RMRP	TRAPPC2
CHRNA2	FGF23	LIG4	RNF135	TRIM32
CHST3	*FGFR2	LMBR1	ROR2	TRIM37
CLCN5	FGFR3	LMNA	RPL11	*TRIP11
COL10A1	FLNA	LRP5	RPL35A	*TRPS1
*COL11A1	*FLNB	MAP2K1	RPL5	TRPV4
*COL11A2	FOXC1	MAP2K2	RPS17	TWIST1
COL1A1	FUCA1	MATN3	RPS19	UBR1
COL1A2	G6PC	*MC4R	RPS24	*WDR35
COL2A1	GALNS	MECP2	RPS6KA3	*WNT3
COL5A1	*GDF5	MGP	RPS7	WNT7A
COL5A2	GH1	MKKS	RUNX2	WRN
COL9A1	*GHR	MKS1	SBDS	ZBTB16
*COL9A2	GHRHR	MMP13	SDHA	
COL9A3	*GHSR	MMP2	SECISBP2	
COMP	GJA1	MRPS16	SEMA3E	
CRTAP	GLB1	MYCN	*SERPINH1	

Supplementary Table 10. Overlapping associations with “unrelated” traits in NHGRI GWAS catalog. GIANT lead height variants or strong proxies ($r^2>0.8$) associated with other traits and diseases reported in the NHGRI catalog of published GWAS at genome-wide level of significance ($P<5\times 10^{-8}$).

GIANT height SNP	Proxy SNP from NHGRI catalog	Chr	Position (bp)	Height P-value for NHGRI SNP (SingleGC)	Disease/ Trait	NHGRI Catalog Trait P-value	Distance (bp) between SNPs	LD (r2)	Height increasing allele	Effect relative to height-increasing allele	Reference (PubMed ID)	
rs2284746	rs2284746	1	17306675	1.17E-40	Pulmonary function	8E-16	0	1	G	reduced pulmonary function	21946350	
rs926438	rs3091242	1	25674785	5.16E-07	Erythrocyte sedimentation rate	2E-13	78853	0.934	T	n/a	21700265	
	rs10903129	1	25768937	2.70E-06	Total cholesterol	5E-10	15299	0.967	G	higher total cholesterol	19060911	
	rs12027135	1	25775733	3.10E-06	Erythrocyte sedimentation rate	5E-13				n/a	21700265	
					Total cholesterol	4E-11	22095	0.967	T	higher total cholesterol	20686565	
					LDL cholesterol	1E-10				higher LDL cholesterol	20686565	
rs12120956	rs17030613	1	113190807	2.63E-12	Blood pressure	1E-08	11764	1	A	lowe blood pressure	21572416	
rs780094	rs1260326	2	27730940	1.35E-11	C-reactive protein	5E-40				lower protein levels	21300955	
					Cardiovascular disease	2E-08				lower CAD risk	21943158	
					Total cholesterol	7E-27				lower total cholesterol	20686565	
					Chronic kidney disease	3E-14				lower CKD risk	20383146	
					Hypertriglyceridemia	7E-09				lower hypertriglyceridemia risk	20657596	
					Liver enzyme levels (GGT)	4E-13				lower enzyme levels	22001757	
					Metabolic traits	4E-10				lower metabolite levels	19060910	
					Metabolite levels	1E-12	10297	0.932	C	n/a	22916037	
					Metabolite levels	3E-18				n/a	22286219	
					Non-albumin protein levels	3E-09				lower protein levels	22558069	
					Platelet counts	9E-10				lower platelet count	22139419	
					Serum albumin level	2E-08				lower protein levels	23022100	
					Triglycerides	2E-31				lower triglycerides	19060906	
					Triglycerides	6E-133				lower triglycerides	20686565	
					Two-hour glucose challenge	3E-10				reduced effect	20081857	
	rs780094	rs780094	2	27741237	5.71E-12	C-reactive protein	7E-15				lower protein levels	18439548
						Fasting glucose-related traits	4E-20				n/a	20081858
						Fasting glucose-related traits	4E-24				n/a	22581228
						Metabolic syndrome	6E-20				lower MS risk	22399527
						Metabolic traits	6E-53	0	1	C	lower metabolite levels	21886157
						Phospholipid levels	9E-09				lower particle levels	21829377
						Triglycerides	3E-14				lower triglycerides	18193044
						Triglycerides	3E-20				lower triglycerides	19060911
						Triglycerides	6E-32				lower triglycerides	18193043
						Uric acid levels	1E-09				lower UA levels	19503597
rs780093	2	27742603	9.56E-12	Crohn's disease	5E-11					lower CD risk	21102463	
				Sex hormone-binding globulin levels	9E-11	1366	1	C	higher protein levels	22829776		
				Triglycerides	3E-10				lower triglycerides	21386085		
				Urate levels	4E-17				lower urate levels	20884846		
rs11684404	rs1260333	2	27748624	2.17E-08	Triglycerides	2E-19	7387	0.87	G	lower triglycerides	20864672	
rs11684404	rs7571971	2	88895351	5.07E-23	Progressive supranuclear palsy	4E-13	29271	0.848	T	n/a	21685912	
rs724016	rs6763931	3	141102833	5.26E-154	Cancer (Prostate)	2E-08	2737	1	A	increased cancer risk	21743467	
rs2300921	rs2002675	3	185629568	1.89E-08	Menarche (age at onset)	1E-09	21433	1	G	later menarche	21102462	

rs9292468	rs1173766	5	32804528	1.96E-19	Blood pressure	2E-08	14545	0.84	T	lower blood pressure	21572416
	rs1173771	5	32815028	2.79E-31	Blood pressure	5E-09	4045	1	A	lower blood pressure	21909110
rs6894139	rs1366594	5	88376061	2.87E-23	Bone mineral density	8E-10	48279	0.967	A	lower BMD	21533022
					Bone mineral density (hip)	1E-13				higher BMD	19801982
rs12186664	rs6235	5	95728898	5.35E-11	Proinsulin levels	1E-26	98673	0.92	G	n/a	21873549
rs7701414	rs3091338	5	131402738	7.18E-24	Crohn's disease	4E-08	183220	0.872	T	increased CD risk	22412388
rs4624820	rs4624820	5	141681788	1.05E-09	Cancer (Testicular)	1E-14	0	1	A	increased cancer risk	20543847
					Cancer (Testicular)	3E-13				increased cancer risk	19483681
rs6457374	rs2524054	6	31252396	3.04E-30	CD4:CD8 lymphocyte ratio	6E-21	19865	0.933	A	reduced ratio	20045101
	rs2247056	6	31265490	6.90E-34	Triglycerides	2E-15	6771	1	T	lower triglycerides	20686565
rs314263	rs7759938	6	105378954	2.59E-43	Menarche (age at onset)	7E-09	13791	0.965	C	later menarche	19448620
					Menarche (age at onset)	5E-60				later menarche	21102462
	rs314276	6	105407999	6.78E-42	Menarche (age at onset)	4E-16	15254	1	A	later menarche	19448623
rs2145357	rs1999930	6	116387134	6.99E-08	Age-related macular degeneration	1E-08	64308	0.924	T	n/a	21665990
rs1155939	rs9388489	6	126698719	1.32E-38	Type 1 diabetes	4E-13	167414	0.842	G	increased T1D risk	19430480
	rs1361108	6	126767600	1.34E-38	Menarche (age at onset)	2E-08	98533	0.839	T	earlier menarche	21102462
rs798497	rs798502	7	2789880	1.80E-66	Ulcerative colitis	3E-15	6077	0.959	A	increased UC risk	21297633
rs11783655	rs6995402	8	145005561	2.07E-09	Platelet counts	5E-10	32012	0.902	T	lower platelet count	22139419
rs10766065	rs900145	11	13293905	1.07E-07	Menarche (age at onset)	2E-08	15944	0.832	C	later menarche	21102462
rs2306694	rs2066808	12	56737973	1.97E-14	Psoriasis	1E-09	57337	1	G	n/a	19169254
rs8756	rs10784502	12	66343810	1.12E-82	Brain structure	1E-12	15942	0.967	C	increased brain volume	22504417
	rs1042725	12	66358347	6.47E-88	Head circumference (infant)	3E-10	1405	0.875	C	increased head circumference	22504419
rs7980687	rs7980687	12	123822711	1.03E-26	Head circumference (infant)	8E-09	0	1	A	increased head circumference	22504419
rs11616380	rs1359790	13	80717156	1.63E-07	Type 2 diabetes	6E-09	11841	1	A	lower T2D risk	20862305
rs2093210	rs10483727	14	61072875	1.69E-34	Glaucoma (primary open-angle)	4E-11	115596	0.802	T	increased cancer risk	22570617
					Vertical cup-disc ratio	1E-11				increased ratio	20548946
rs1036477	rs1036476	15	48914775	7.71E-11	Thoracic aortic aneurysms and dissections	6E-13	151	0.892	T	n/a	21909107
rs7162825	rs2652822	15	63422772	1.03E-07	Metabolic traits	7E-27	16414	0.935	C	lower metabolite levels	21886157
rs5742915	rs5742915	15	74336633	1.52E-29	Paget's disease	2E-14	0	1	C	increased PD risk	21623375
rs2238300	rs2307449	15	89863928	1.01E-10	Menopause (age at onset)	4E-13	12348	0.866	T	later menopause	22267201
rs1659127	rs1659127	16	14388305	2.85E-19	Menarche (age at onset)	4E-09	0	1	A	later menarche	21102462
rs217181	rs217181	16	72114002	3.74E-10	Metabolite levels	1E-36	0	1	T	n/a	22916037
rs1625895	rs1625895	17	7578115	3.88E-10	Sex hormone-binding globulin levels	2E-21	0	1	C	lower protein levels	22829776
rs199515	rs199533	17	44828931	1.22E-05	Parkinson's disease	1E-14	27706	0.947	G	n/a	19915575
	rs199515	17	44856641	1.56E-05	Parkinson's disease	3E-17	0	1	C	n/a	22451204
rs318095	rs46522	17	46988597	1.03E-14	Coronary heart disease	2E-08	13863	1	C	lower CAD risk	21378990
rs11152213	rs12967135	18	57849023	2.93E-12	HDL cholesterol	7E-09	3925	1	A	lower HDL cholesterol	20686565
rs11880992	rs12982744	19	2177193	1.61E-27	Osteoarthritis	1E-11	790	1	G	lower OA risk	22566624
rs2057291	rs6026584	20	57469073	4.57E-10	Renal function	9E-09	2970	1	T	increased renal function	22797727

Supplementary Table 11. Enrichment of functional genomic elements catalogued by ENCODE and the Roadmap Epigenomics Projects. Annotation of the GIANT height SNPs with epigenomic features. For each “seed” height SNPs, we retrieved all markers in linkage disequilibrium (LD, $r^2 \geq 0.8$) using the European panel from the 1000 Genomes Project (CEU). We then annotated each “seed” and LD proxy using epigenomic tracks and counted the number of loci where a SNP overlaps an ENCODE or Roadmap Epigenomics Projects peak (for a given track, a locus can count only once even if several SNPs within the locus overlap ENCODE peaks). We repeated the annotation and counting procedure on a 1,000 sets of matched SNPs. Here we report the number of loci that overlap peaks in each track for ENCODE cell lines. For all marks tested the empirical enrichment *P*-value is <0.001. NA: track not available when we accessed the data.

	ENCODE					Roadmap Epigenomics	
	GM12878	H1hESC	HEPG2	HUVEC	K562	Chondrocytes	Osteoblasts
Open chromatin							
DNA polymerase II	150	170	218	198	197	NA	NA
DNAse I hypersensitive sites	238	259	235	254	228	NA	247
FAIRE	139	87	137	231	197	NA	NA
DNA methylation							
Illumina 450K array	156	156	155	156	155	NA	NA
Histone modification							
Ctcf	245	243	199	228	257	NA	98
Ezh2	436	345	393	371	426	NA	NA
H2az	317	402	399	383	335	NA	NA
H3k27ac	248	373	258	309	263	NA	371
H3k4me1	376	411	411	396	361	269	435
H3k4me2	323	320	335	358	278	NA	378
H3k4me3	330	248	279	269	266	271	344
H3k79me2	271	277	304	318	274	NA	NA
H3k9ac	230	354	270	284	269	239	NA
H4k20me1	410	388	359	365	424	NA	NA

Supplementary Table 12. Summary of results from gene-set enrichment analysis as implemented by MAGENTA for gene set enrichment analysis. Shown are gene sets that met significance after correcting for multiple hypotheses testing. Gene set shown in **bold** was significant enriched in the previous GIANT height GWAS study (Lango Allen *et al*; 2010).

Database	Biological pathways or Gene sets	Original # genes in gene set	# genes in gene set analyzed by GSEA	Nominal GSEA P-value	False discovery rate (FDR)	Expected # genes above 95th percentile cut-off	Observed # genes above 95th percentile cut-off
Gene Ontology	nucleosome	64	35	8.00E-06	0.022	2	10
Gene Ontology	heart morphogenesis	29	28	3.90E-05	0.026	1	8
Gene Ontology	nucleosome assembly	80	47	1.30E-05	0.036	2	11
Gene Ontology	chromosome	147	112	9.00E-06	0.037	6	18
KEGG	Hedgehog signaling pathway	56	53	2.00E-04	0.04	3	10
PANTHER	Hedgehog signaling pathway	14	14	6.00E-04	0.023	1	5
PANTHER (BioProc)	Lactation mammary development	13	11	6.00E-06	2.00E-04	1	6
PANTHER (BioProc)	Chromatin packaging and remodeling	237	140	1.00E-06	0.002	7	22
PANTHER (MolFunc)	Histone	86	31	9.90E-07	<<1E-5	2	11
REACTOME	PACKAGING_OF_TELOMERE_ENDS	49	24	9.90E-07	<<1E-5	1	10
REACTOME	RNA_POLYMERASE_I_PROMOTER_OPENING	59	23	9.90E-07	<<1E-5	1	10
REACTOME	RNA_POLYMERASE_I_PROMOTER_CLEARANCE	82	43	5.00E-06	9.00E-04	2	11
REACTOME	TELOMERE_MAINTENANCE	77	50	1.00E-04	0.005	3	11
REACTOME	APOPTOSIS_INDUCED_DNA_FRAGMENTATION	11	11	9.00E-04	0.019	1	4
REACTOME	RNA_POLYMERASE_I_III_AND_MITOCHONDRIAL_TRANSCRIPTION	120	81	4.00E-04	0.028	4	13
REACTOME	TRANSCRIPTION	197	154	3.00E-04	0.044	8	19

Supplementary Table 15. DEPICT analysis of tissue enrichment. The Data-driven Enrichment-Prioritized Integration for Complex Traits (DEPICT) method uses expression data from 37,427 human microarray samples to identify cell-types and tissues (defined by MeSH [*Medical Subject Headings*] terms) in which genes from associated loci are highly expressed. Locus gene sets were constructed by mapping genes to a given locus if they were within, or overlapped with, regions defined by linkage disequilibrium $r^2 > 0.5$ around the lead height SNP of that locus.

MeSH ID	Name	MeSH first level term	MeSH second level term	P-value	False-discovery rate
Lango-Allen et al. 199 Height SNPs					
A02.165	Cartilage	Musculoskeletal System	Cartilage	0.0001	0.0400
A11.329.171	Chondrocytes	Cells	Connective Tissue Cells	0.0011	0.0700
A10.615.284	Extraembryonic Membranes	Tissues	Membranes	0.0012	0.0467
A10.615.284.473	Chorion	Tissues	Membranes	0.0012	0.0350
A11.872.580	Mesenchymal Stem Cells	Cells	Stem Cells	0.0017	0.0440
697 Height SNPs					
A02.165	Cartilage	Musculoskeletal System	Cartilage	1.55E-12	< 0.0011
A11.329.171	Chondrocytes	Cells	Connective Tissue Cells	7.84E-11	< 0.0011
A11.872.580	Mesenchymal Stem Cells	Cells	Stem Cells	4.97E-09	< 0.0011
A02.835.583.443.800	Synovial Membrane	Musculoskeletal System	Skeleton	5.49E-09	< 0.0011
A02.835.583	Joints	Musculoskeletal System	Skeleton	5.49E-09	< 0.0011
A02.835.583.443	Joint Capsule	Musculoskeletal System	Skeleton	5.49E-09	< 0.0011
A11.329.830	Stromal Cells	Cells	Connective Tissue Cells	4.12E-08	< 0.0011
A11.329.228	Fibroblasts	Cells	Connective Tissue Cells	2.09E-07	< 0.0011
A11.329.629	Osteoblasts	Cells	Connective Tissue Cells	8.34E-06	< 0.0011
A10.615.284	Extraembryonic Membranes	Tissues	Membranes	1.68E-05	< 0.0011
A10.615.284.473	Chorion	Tissues	Membranes	1.68E-05	< 0.0011
A02.835.232.834.151	Cervical Vertebrae	Musculoskeletal System	Skeleton	2.64E-05	< 0.0011
A02.835.232.834	Spine	Musculoskeletal System	Skeleton	2.87E-05	< 0.0011
A11.872	Stem Cells	Cells	Stem Cells	7.59E-05	< 0.0011
A03.734.414	Islets of Langerhans	Digestive System	Pancreas	8.19E-05	< 0.0011
A04.411	Lung	Respiratory System	Lung	8.59E-05	< 0.0011
A07.231.114	Arteries	Cardiovascular System	Blood Vessels	0.0002	0.0012
A05.360.319.114.373	Fallopian Tubes	Urogenital System	Genitalia	0.0003	0.0011
A15.145.846	Serum	Hemic and Immune Systems	Blood	0.0005	0.0032
A05.360.319	Genitalia Female	Urogenital System	Genitalia	0.0005	0.0030
A05.360.319.679.690	Myometrium	Urogenital System	Genitalia	0.0005	0.0038
A05.360.319.679	Uterus	Urogenital System	Genitalia	0.0008	0.0064
A05.360.319.679.490	Endometrium	Urogenital System	Genitalia	0.0011	0.0078
A11.329	Connective Tissue Cells	Cells	Connective Tissue Cells	0.0013	0.0100
A07.541.510	Heart Valves	Cardiovascular System	Heart	0.0014	0.0104
A07.541.510.110	Aortic Valve	Cardiovascular System	Heart	0.0014	0.0100
A07.231	Blood Vessels	Cardiovascular System	Blood Vessels	0.0017	0.0111
A05.360.319.114.630	Ovary	Urogenital System	Genitalia	0.0019	0.0107
A05.360.319.114	Adnexa Uteri	Urogenital System	Genitalia	0.0019	0.0110
A11.329.114	Adipocytes	Cells	Connective Tissue Cells	0.0022	0.0107
A10.690.467	Muscle Smooth	Tissues	Muscles	0.0024	0.0116
A11.382.625	Enteroendocrine Cells	Cells	Endocrine Cells	0.0028	0.0169
A11.436.294.064	Glucagon Secreting Cells	Cells	Epithelial Cells	0.0028	0.0164
A03.556.875.875	Stomach	Digestive System	Gastrointestinal Tract	0.0037	0.0224
A06.407	Endocrine Glands	Endocrine System	Endocrine Glands	0.0039	0.0217
A06.407.312	Gonads	Endocrine System	Endocrine Glands	0.0041	0.0239

A11.436.275	Endothelial Cells	Cells	Epithelial Cells	0.0055	0.0314
A05.360	Genitalia	Urogenital System	Genitalia	0.0055	0.0311
A03.734	Pancreas	Digestive System	Pancreas	0.0064	0.0349
A03.556.875	Upper Gastrointestinal Tract	Digestive System	Gastrointestinal Tract	0.0086	0.0480
A07.231.908.670.874	Umbilical Veins	Cardiovascular System	Blood Vessels	0.0089	0.0473
A07.231.908.670	Portal System	Cardiovascular System	Blood Vessels	0.0089	0.0462
A07.231.908	Veins	Cardiovascular System	Blood Vessels	0.0099	0.0474

Supplementary Table 17. Study design, number of individuals and sample quality control for genome-wide association study cohorts

Study		Study design	Total sample size (N)	Sample QC		Samples in analyses(N)	Anthropometric assessment method	References
Short name	Full name			Call rate*	other exclusions			
ACTG	The AIDS Clinical Trials Group	Population-based	2648	>95%	1) Non-Europeans (based on PCA); 2) High individual missingness (>5%); 3) High heterozygosity (Inbreeding coefficient > 0.1 or < -0.1); 4) Related individuals 5) duplicates	1055	measured	International, H.I.V.C.S. et al. The major genetic determinants of HIV-1 control affect HLA class I peptide presentation. <i>Science</i> 330, 1551-7 (2010).
AE	Athero-Express Biobank Study	patient-cohort	2512	≥ 97%	1) Heterozygosity (\hat{H}) ± 3 standard deviations of the mean; 2) Ethnic outliers through Principal Component Analysis compared to HapMap 2 (r22); 3) Related individuals and duplicates, $\pi > 0.20$; 4) Missing body weight and height.	686	measured	1) Verhoeven, B.A. et al. Athero-express: differential atherosclerotic plaque expression of mRNA and protein in relation to cardiovascular events and patient characteristics. <i>Rationale and design. Eur J Epidemiol</i> 19, 1127-33 (2004). 2) Hurks, R. et al. Aneurysm-express: human abdominal aortic aneurysm wall expression in relation to heterogeneity and vascular events - rationale and design. <i>Eur Surg Res</i> 45, 34-40 (2010).
ASCOT	Anglo-Scandinavian Cardiac Outcome Trial	Randomised control clinical trial	3868	≥ 95%	1) Sample cryptic duplicates 2) Sample outliers in ancestry principle components analysis 3) First and second degree relatives; 4) Missing body weight and height.	3802	measured	1) Sever, P.S. et al. Anglo-Scandinavian Cardiac Outcomes Trial: a brief history, rationale and outline protocol. <i>J Hum Hypertens</i> 15 Suppl 1, S11-2 (2001). 2) Sever, P.S. et al. Rationale, design, methods and baseline demography of participants of the Anglo-Scandinavian Cardiac Outcomes Trial. <i>ASCOT investigators. J Hypertens</i> 19, 1139-47 (2001).
BLSA	Baltimore Longitudinal Study on Aging	Population-based	848	≥ 98.5%	1) sex mismatch 2) ethnic outliers	844	measured	Shock, N.W., et al., Normal Human Aging: The Baltimore Study of Aging: NIH Publication No. 84-2450 (1984).
B-PROOF	B-vitamins for the Prevention of Osteoporotic Fractures	RCT	2919	≥ 95%	1) gender mismatches 2) related individuals	2669	measured	van Wijngaarden, J.P. et al. Rationale and design of the B-PROOF study, a randomized controlled trial on the effect of supplemental intake of vitamin B12 and folic acid on fracture incidence. <i>BMC Geriatr</i> 11, 80 (2011).
DESIR	Data from an Epidemiological Study on the Insulin Resistance syndrome	Population-based	731	≥ 90%	1) Missing data; 2) ethnicity outliers (N=15)	716	measured	Balkau, B. [An epidemiologic survey from a network of French Health Examination Centres, (D.E.S.I.R.): epidemiologic data on the insulin resistance syndrome]. <i>Rev Epidemiol Sante Publique</i> 44, 373-5 (1996).
DNBC	Danish National Birth Cohort - Preterm Delivery Study	Case-control	1937	≥ 95%	1) persons born outside of Scandinavia or with parents born outside of Scandinavia; 2) Missing body weight and height.	1802	self-reported	1) Olsen, J. et al. The Danish National Birth Cohort--its background, structure and aim. <i>Scand J Public Health</i> 29, 300-7 (2001). 2) Ryckman, K.K. et al. Replication of a genome-wide association study of birth weight in preterm neonates. <i>J Pediatr</i> 160, 19-24 e4 (2012).

EGCUT-370	Estonian Genome Center, University of Tartu	Population-based	866	≥ 97%	1) ethnic outliers; 2) related individuals and duplicates; 3) Missing body weight and height.	866	measured	1) Nelis, M. et al. Genetic structure of Europeans: a view from the North-East. PLoS One 4, e5472 (2009). 2) Metspalu, A. The Estonian Genome Project. Drug Development Research 62, 97-101 (2004).
EGCUT-OMNI	Estonian Genome Center, University of Tartu	Population-based	1356	≥ 97%	1) ethnic outliers; 2) related individuals and duplicates; 3) Missing body weight and height.	1356	measured	1) Nelis, M. et al. Genetic structure of Europeans: a view from the North-East. PLoS One 4, e5472 (2009). 2) Metspalu, A. The Estonian Genome Project. Drug Development Research 62, 97-101 (2004).
ERF	Erasmus Rucphen Family Study	Family-based	2726	≥98%	Mendelian inconsistencies	2726	measured	Aulchenko, Y.S. et al. Linkage disequilibrium in young genetically isolated Dutch population. Eur J Hum Genet 12, 527-34 (2004).
FamHS	Family Heart Study	Population-based	1486	≥ 98%	1) ethnic outliers; 2) related individuals and duplicates; 3) Missing body weight and height. 4)sex inconsistencies.	1463	measured	1) Higgins, M. et al. NHLBI Family Heart Study: objectives and design. Am J Epidemiol 143, 1219-28 (1996). 2) O'Donnell, C.J. et al. Genome-wide association study for coronary artery calcification with follow-up in myocardial infarction. Circulation 124, 2855-64 (2011).
Health ABC	Health, Aging, and Body Composition Study	longitudinal cohort study	1655	≥ 97%	1) related individuals and duplicates; 2) Missing body weight and height.	1655	measured	Harris, T.B. et al. Waist circumference and sagittal diameter reflect total body fat better than visceral fat in older men and women. The Health, Aging and Body Composition Study. Ann N Y Acad Sci 904, 462-73 (2000).
HERITAGE Family Study	Health, Risk Factors, Training and Genetics (HERITAGE) Family Study	Family Study, baseline data from an exercise training intervention	500	>99%	NA	500	measured	1) Bouchard, C. et al. The HERITAGE family study. Aims, design, and measurement protocol. Med Sci Sports Exerc 27, 721-9 (1995). 2) Bouchard C, Sarzynski MA, Rice T, Kraus W, Church T, Sung YJ, Rao DC, Rankinen T. Genomic predictors of maximal oxygen uptake response to standardized exercise training programs. Journal of Applied Physiology 110:1160-1170 (2011).
HYPERGENES Controls	HYPERGENES	Case-control	1934	>90%	1) ethnic outliers 2) Missing body weight and height.	1900	measured	Salvi, E. et al. Genomewide 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. Hypertension 59, 248-55 (2012).
HYPERGENES Cases	HYPERGENES	Case-control	2124	>90%	1) ethnic outliers 2) Missing body weight and height.	1841	measured	Salvi, E. et al. Genomewide 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. Hypertension 59, 248-55 (2012).
InCHIANTI	Invecchiare in Chianti	Population-based	1210	≥ 98%	1) Sex mismatch 2) heterozygosity <30%; 3) Missing body weight or height.	1138	measured	Ferrucci, L. et al. Subsystems contributing to the decline in ability to walk: bridging the gap between epidemiology and geriatric practice in the InCHIANTI study. J Am Geriatr Soc 48, 1618-25 (2000).

IPM Mount Sinai BioMe	The Charles Bronfman Institute for Personalized Medicine BioMe Biobank Program	Hospital-based	3069	≥ 95%	1) heterozygosity (Z-value > 6) 2) Gender mismatch errors 3) ethnic outliers; 4) First degree relatives and duplicates; 5) Missing body weight, height, or BMI (then excluded only from that group).	2867	measured	Tayo, B.O. et al. Genetic background of patients from a university medical center in Manhattan: implications for personalized medicine. PLoS One 6, e19166 (2011).
LifeLines	LifeLines Cohort study	Population-based	9480	≥ 95%	1) heterozygosity >4SD from mean; 2) ethnic outliers; 3) related individuals and duplicates; 4) sex mismatch with database; 5) Missing age, sex, height	8118	measured	Stolk, R.P. et al. Universal risk factors for multifactorial diseases: LifeLines: a three-generation population-based study. Eur J Epidemiol 23, 67-74 (2008).
LLS	Leiden Longevity Study	Family based	2415	≥ 95%	1) ethnic outliers 2) Missing body weight and height.	1903	self-reported	1) Schoenmaker, M. et al. Evidence of genetic enrichment for exceptional survival using a family approach: the Leiden Longevity Study. Eur J Hum Genet 14, 79-84 (2006). 2) Westendorp, R.G. et al. Nonagenarian siblings and their offspring display lower risk of mortality and morbidity than sporadic nonagenarians: The Leiden Longevity Study. J Am Geriatr Soc 57, 1634-7 (2009).
LOLIPOP_EW610	London Life Sciences Prospective Population Study	Population-based	945	≥ 95%	1) Duplicates 2) gender discrepancy 3) contaminated samples 4) relatedness	927	measured	1) Wain, L.V. et al. Genome-wide association study identifies six new loci influencing pulse pressure and mean arterial pressure. Nat Genet 43, 1005-11 (2011). 2) International Consortium for Blood Pressure Genome-Wide Association, S. et al. Genetic variants in novel pathways influence blood pressure and cardiovascular disease risk. Nature 478, 103-9 (2011).
LOLIPOP_EWA	London Life Sciences Prospective Population Study	Population-based with some enrichment	878	≥ 95%	1) Duplicates 2) contaminated samples 3) relatedness 4) samples already in EW610	513	measured	Yuan, X. et al. Population-based genome-wide association studies reveal six loci influencing plasma levels of liver enzymes. Am J Hum Genet 83, 520-8 (2008).
LOLIPOP_EWP	London Life Sciences Prospective Population Study	Population-based with some enrichment	1006	≥ 95%	1) Duplicates 2) contaminated samples 3) samples already in EW610	651	measured	Kooner, J.S. et al. Genome-wide scan identifies variation in MLXIPL associated with plasma triglycerides. Nat Genet 40, 149-51 (2008).
NELSON	Dutch and Belgian Lung Cancer Screening Trial	Current and former heavy smokers	3082	≥ 98%	1) heterozygosity > 2d from mean; 2) ethnic outliers; 4) related individuals (r2>0.2) and duplicates; 5) Missing height.	2668	measured	1) van Iersel, C.A. et al. Risk-based selection from the general population in a screening trial: selection criteria, recruitment and power for the Dutch-Belgian randomised lung cancer multi-slice CT screening trial (NELSON). Int J Cancer 120, 868-74 (2007). 2) van Setten, J. et al. Genome-wide association study of coronary and aortic calcification implicates risk loci for coronary artery disease and myocardial infarction. Atherosclerosis 228, 400-5 (2013).

PLCO2 controls	Prostate, Lung, Colorectal, and Ovarian Cancer Screening Trial	Population-based case-control	1216	>98%	1) sex discordance with genetic data; 2) heterozygosity; 3) ancestry outliers; 4) related individuals and duplicates; 5) missing phenotype data	1193	self-report	Cornelis, M.C. et al. Genome-wide meta-analysis identifies regions on 7p21 (AHR) and 15q24 (CYP1A2) as determinants of habitual caffeine consumption. <i>PLoS Genet</i> 7, e1002033 (2011).
PLCO2 cases	Prostate, Lung, Colorectal, and Ovarian Cancer Screening Trial	Population-based case-control	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	Cornelis, M.C. et al. Genome-wide meta-analysis identifies regions on 7p21 (AHR) and 15q24 (CYP1A2) as determinants of habitual caffeine consumption. <i>PLoS Genet</i> 7, e1002033 (2011).
PREVEND	Prevention of REnal and Vascular ENdstage Disease (PREVEND) Study	Population-based	3920	≥ 95%	1) Ethnic outliers 2) related individuals and duplicates 3) missing phenotype	3624	measured	Hillege, H.L. et al. Urinary albumin excretion predicts cardiovascular and noncardiovascular mortality in general population. <i>Circulation</i> 106, 1777-82 (2002).
PROCARDIS	Precocious Coronary Artery Disease	Population-based	13000	≥ 95%	1) ethnic outliers; 2) Missing body weight and height.	~7000	measured	1) Coronary Artery Disease Genetics, C. A genome-wide association study in Europeans and South Asians identifies five new loci for coronary artery disease. <i>Nat Genet</i> 43, 339-44 (2011). 2) 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).
PROSPER/ PHASE	The PROspective study of Pravastatin in the Elderly at Risk for vascular disease	Randomized controlled trial	5.784	≥ 97.5%	1) heterozygosity ; 2) ethnic outliers; 3) related individuals and duplicates;	5244	measured	1) Shepherd, J. et al. The design of a prospective study of Pravastatin in the Elderly at Risk (PROSPER). PROSPER Study Group. PROspective Study of Pravastatin in the Elderly at Risk. <i>Am J Cardiol</i> 84, 1192-7 (1999). 2) Shepherd, J. et al. Pravastatin in elderly individuals at risk of vascular disease (PROSPER): a randomised controlled trial. <i>Lancet</i> 360, 1623-30 (2002). 3) Trompet, S. et al. Replication of LDL GWAs hits in PROSPER/PHASE as validation for future (pharmacogenetic) analyses. <i>BMC Med Genet</i> 12, 131 (2011).
QFS	Quebec Family Study		951	≥ 95%	N/A	860	measured	Bouchard, C. et al. Inheritance of the amount and distribution of human body fat. <i>Int J Obes</i> 12, 205-15 (1988).
QIMR	Twin study at Queensland Institute of Medical Research	Population-based	11930	≥ 95%	1) age < 18; 2) ethnic outliers; 3) related individuals and duplicates; 4) Missing body weight and height.	3627	measured / self-reported	Medland, S.E. et al. Common variants in the trichohyalin gene are associated with straight hair in Europeans. <i>Am J Hum Genet</i> 85, 750-5 (2009).

RISC	Relationship between Insulin Sensitivity and Cardiovascular disease Study	Population-based	1566	>95%	1) Non-European Descent 2) Sex mismatches	1031	measured	Hills, S.A. et al. The EGIR-RISC STUDY (The European group for the study of insulin resistance: relationship between insulin sensitivity and cardiovascular disease risk): I. Methodology and objectives. <i>Diabetologia</i> 47, 566-70 (2004).
SHIP-TREND	Study of Health in Pomerania - TREND	Population-based	997	≥ 94%	1) duplicate samples (by estimated IBD) 2) reported/genotyped gender mismatch	986	measured	Volzke, H. et al. Cohort profile: the study of health in Pomerania. <i>Int J Epidemiol</i> 40, 294-307 (2011).
TRAILS	Tracking Adolescents' Individual Lives Survey	Population-based (measured at 18yrs of age)	1491	≥ 95%	1) heterozygosity >4SD from mean; 2) ethnic outliers; 3) related individuals and duplicates; 4) sex mismatch with database; 5) Missing height, age, or sex.	1139	measured	Huisman, M. et al. Cohort profile: the Dutch 'TRacking Adolescents' Individual Lives' Survey'; TRAILS. <i>Int J Epidemiol</i> 37, 1227-35 (2008).
TWINGENE	TWINGENE	Population-based	9836	≥ 97%	1) Heterozygosity, $(F - \text{mean}(F))/\text{sd}(Z) \geq 5$ 2) Sex-check based on the heterozygosity rates on the X-chromosome 3) Missing phenotypes used in analysis	9380	measured	Hong, Y. et al. Genetic and environmental architecture of the features of the insulin-resistance syndrome. <i>Am J Hum Genet</i> 60, 143-52 (1997).

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

Supplementary Table 18. Information on genotyping methods, quality control of SNPs, imputation, and statistical analysis for genome-wide association study cohort

Study	GENOTYPING						IMPUTATION			ASSOCIATION ANALYSIS	
	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		
ACTG	Illumina HumanHap650Y and Illumina-1Mduo	Beadstudio	>1%	≥98%	$>5 \times 10^{-6}$	477,986 (650Y); 917,487 (1Mduo)	BEAGLE	0.0002	info-score≥0.1	2,491,886	PLINK
AE	Affymetrix 5.0	BRLMM-P	≥3%	≥97%	$>10^{-6}$	403,789	BEAGLE	ALL	ALL	2,409,615	PLINK & GoldenHelix SVS
ASCOT	Illumina HumanCNV 370	Beadstudio	>0%	≥97%	$>10^{-7}$	283,291	MACH	ALL	r2-hat≥0.30	2,477,983	MACH2QTL
BSA	Illumina 550K	Beadstudio	≥1%	≥99%	$>10^{-4}$	514,027	MACH	≥1%	r2-hat≥0.30	2,557,230	MERLIN-OFFLINE
B-PROOF	Illumina OMNIEXPRESS	GenomeStudio	ALL	≥97.5%	$>10^{-6}$	722,053	MACH	≥1%	r2-hat≥0.30	2,450,937	GRIMP
DESIR	Illumina Human CNV370-Duo and Illumina HAP300	GenomeStudio	≥1%	≥95%	$>10^{-4}$	291,609	IMPUTE	≥1%	proper-info≥0.40	2,544,187	SNPTEST
DNBC	Illumina 660w-quad	BeadStudio	≥0.5%	≥98%	$>10^{-3}$	518097	MACH	ALL	ALL	2549123	MACH2QTL
EGCUT-370	Illumina Human370CNV	GenomeStudio	≥1%	≥97%	$>10^{-6}$	320,955	IMPUTE	≥1%	proper-info≥0.40	2,341,594	SNPTEST
EGCUT-OMNI	Illumina OMNI-Express	GenomeStudio	≥1%	≥97%	$>10^{-6}$	647,357	IMPUTE	≥1%	proper-info≥0.40	2,375,437	SNPTEST
ERF	Illumina 318K, Illumina 370K and Affymetrix 250K	BRLMM, BeadStudio	≥1%	≥98%	$>10^{-6}$	450,877	MACH	≥1%	ALL	ALL	ProbABEL/GenABEL
FamHS	Illumina HumMap 550K, Human 610-Quadv1, and Illumina Human 1M-Duov3	GenCall, BeadStudio	≥1%	≥98%	$>10^{-6}$	849,551	MACH	≥1%	r2-hat≥0.30	2,543,887	Local Software
Health ABC	Illumina-1Mduo	Beadchip	≥1%	≥97%	$>10^{-6}$	914,263	MACH	NA	r2-hat≥0.30	2,543,887	R
HERITAGE Family Study	Illumina HumanCNV370-Quad v3.0	GenomeStudio	≥1%	≥99%	$>10^{-6}$	324,607	MACH	≥1%	r2-hat≥0.30	2,453,887	ProbABEL
Hypergenes Controls	Illumina-1Mduo	GenCall, BeadStudio	≥1%	≥90%	$>10^{-7}$	Center I: 861759 Center II: 872576	MACH	>0%	r2-hat≥0.30	2,543,887	Matlab
Hypergenes Cases	Illumina-1Mduo	GenCall, BeadStudio	≥1%	≥90%	$>10^{-7}$	Center I: 861759 Center II: 872576	MACH	>0%	r2-hat≥0.30	2,543,887	Matlab
InCHIANTI	Illumina 550K	Birdstudio	≥1%	≥99%	$>10^{-4}$	495,343	MACH	≥1%	r2-hat≥0.30	2,461,088	MERLIN
IPM Mount Sinai BioMe	Affymetrix SNP 6.0	Birdsuite	≥1%	≥95%	$>10^{-3}$	665,889	MACH	ALL	ALL	3,021,329	MACH2QTL
Lifelines	Illumina Cyto SNP12 v2	GenomeStudio	≥1%	≥95%	$>10^{-4}$	257,581	BEAGLE	>0%	ALL	2,472,812	PLINK
LLS	Illumina Infinium HD Human660W-Quad and Infinium HD Human OmniExpress	BeadStudio	≥1%	≥95%	$>10^{-4}$	516,712	IMPUTE	≥1%	proper-info≥0.40	3,801,289	QT-assoc

LOLIPOP_EW610	Illumina Human610	BeadStudio	≥1%	≥90%	>10 ⁻⁶	544,620	MACH	ALL	ALL	2,543,887	MACH2QTL
LOLIPOP_EWA	Affymetrix 500K	BRLMN	≥1%	≥90%	>10 ⁻⁶	374,773	MACH	ALL	ALL	2557252	MACH2QTL
LOLIPOP_EWP	Perlegen custom	Perlegen custom	≥1%	≥90%	>10 ⁻⁶	184,469	MACH	ALL	ALL	2,177,742	MACH2QTL
NELSON	Illumina Quad610 BeadChip	GenomeStudio	≥5%	≥98%	>10 ⁻³	581,712	BEAGLE	ALL	ALL	2,543,807	PLINK
PLCO2 controls	Illumina HumanHap 550K and Illumina HumanHap 610K	BeadStudio	ALL	≥90%	none	525,262	IMPUTE	≥1%	proper-info≥0.40	2,538,067	SNPTEST
PLCO2 cases	Illumina HumanHap 550K and Illumina HumanHap 610K	BeadStudio	ALL	≥90%	none	525,262	IMPUTE	≥1%	proper-info≥0.40	2,564,030	SNPTEST
PREVEND	Illumina cytoSNP12 v2	GenomeStudio	≥1%	>95%	>10 ⁻⁵	232,571	BEAGLE	≥1%	ALL	2289210	PLINK
PROCARDIS	Illumina 1M, Illumina 610K	Beadstudio	≥1%	≥95%	>10 ⁻⁶	460,920	MACH	ALL	ALL	2,543,887	STATA
PROSPER/ PHASE	Illumina 660K beadchip	Beadstudio	≥1%	≥97.5%	>10 ⁻⁶	557,192	MACH	ALL	ALL	2,543,887	ProbABEL
QFS	Illumina 610-Quad chip	BeadStudio	≥1%	≥95%	>10 ⁻⁴	543,713	MACH	≥1%	r2-hat≥0.30	2,562,194	GWAF
QIMR	Illumina HumanHap 370 / 610	BeadStudio	≥1%	≥95%	>10 ⁻⁶	271,069	MACH	≥1%	r2-hat≥0.30	2,543,887	PLINK
RISC	Affymetrix 6.0	Birdseed	>1%	≥98%	>10 ⁻⁴	747,423	MACH	>1%	r2-hat≥0.30	2,500,497	MACH2QTL
SHIP-TREND	Illumina Human Omni 2.5	GenCall	ALL	>90%	>10 ⁻⁴	1,782,967	IMPUTE	ALL	ALL	3,437,411	QUICKTEST
TRAILS	Illumina Cyto SNP12 v2	GenomeStudio	≥1%	≥95%	>10 ⁻⁴	260,077	IMPUTE2	ALL	ALL	2,631,501	SNPTEST
TWINGENE	Illumina HumanOmniExpress	GenomeStudio	≥1%	≥97%	>10 ⁻⁷	644556	IMPUTE2	ALL	ALL	2,585,373	PLINK
TwinsUK	Illumina HumanHap300 and Illumina HumanHap610Q	Illuminus	≥1%	>97% (MAF>5%); >99% (<5%)	>10 ⁻⁶	303,940 (Hap300); 553,487 (Hap610Q)	IMPUTE	≥1%	proper-info≥0.40	3,044,097	SNPTEST

* Call rate to exclude SNPs for which less than a certain percentage of individuals were successfully genotyped (i.e. to exclude 'bad' SNPs)

Supplementary Table 19: Study-specific descriptive statistics genome-wide association study cohorts

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
ACTG	Age (yrs)	1055	39.2	9.20	39.0	17.0	77.0	NA	-0.03	NA	NA	NA	NA	NA	NA	NA	NA
	BMI (kg/m ²)	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA
	Weight (kg)	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA
	Height (m)	1055	1.8	0.07	1.8	1.4	2.0	NA	1.00	NA	NA	NA	NA	NA	NA	NA	NA
	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
AE	Age (yrs)	469	68.1	8.88	69.0	39.0	88.0	-0.06	-0.20	217	68.2	9.60	69.0	37.0	92.0	-0.05	-0.05
	BMI (kg/m ²)	433	26.3	3.18	26.0	17.6	39.9	1.00	-0.01	189	26.4	4.71	26.0	15.2	50.7	1.00	-0.19
	Weight (kg)	435	81.6	11.60	80.0	49.0	130.0	0.81	0.53	191	71.4	12.95	70.0	43.0	133.0	0.90	0.21
	Height (m)	433	1.8	0.07	1.8	1.6	2.0	-0.01	1.00	191	1.6	0.06	1.7	1.5	1.8	-0.19	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
ASCOT	Age (yrs)	3122	63.5	8.2	64.0	40.0	80.0	-0.22	-0.18	673	64.7	7.6	65.0	41.0	80.0	-0.21	-0.13
	BMI (kg/m ²)	3122	28.9	4.6	28.4	15.0	88.8	1.00	-0.11	673	29.5	5.7	28.7	16.5	52.1	1.00	-0.11
	Weight (kg)	3122	87.2	14.8	85.4	44.0	188.0	0.84	0.43	673	75.3	15.1	72.8	43.6	150.6	0.91	0.30
	Height (m)	3122	1.7	0.1	1.7	1.0	2.0	-0.11	1.00	673	1.6	0.1	1.6	1.3	1.9	-0.11	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
BLSA	Age (yrs)	462	71.3	15.70	74.0	22.0	96.0	-0.20	-0.45	382	66.5	17.15	67.0	21.0	97.0	-0.01	-0.51
	BMI (kg/m ²)	462	27.6	4.52	26.7	19.1	46.7	1.00	0.01	382	25.9	4.83	24.9	18.2	46.9	1.00	-0.01
	Weight (kg)	462	84.2	16.01	82.0	53.1	170.3	0.86	0.48	382	68.3	13.95	66.0	41.8	122.4	0.88	0.42
	Height (m)	462	1.7	0.07	1.7	1.5	2.0	0.01	1.00	382	1.6	0.07	1.6	1.4	1.8	-0.01	1.00
	WC (cm)	462	98.6	12.02	98.0	68.0	146.5	0.83	0.14	382	84.4	11.63	84.0	57.0	123.0	0.81	0.02
	Hip (cm)	462	103.5	9.71	102.0	82.0	155.5	0.80	0.32	382	103.1	10.58	101.9	70.1	145.0	0.85	0.25
	WHR (cm/cm)	462	1.0	0.06	1.0	0.8	1.1	0.41	-0.16	382	0.8	0.07	0.8	0.6	1.1	0.29	-0.23
B-PROOF	Age (yrs)	1459	73.4	6.1	72.0	63.0	98.0	0.85	-0.23	1460	74.9	6.8	74.0	63.0	97.0	-0.21	-0.34
	BMI (kg/m ²)	1456	26.8	3.4	26.4	17.0	56.0	1.00	-0.04	1445	27.5	4.5	27.0	14.0	47.0	1.00	-0.11
	Weight (kg)	1456	83.1	11.9	82.0	48.0	180.0	0.85	0.48	1447	72.6	12.5	72.0	37.0	135.0	0.89	0.36
	Height (cm)	1456	175.9	6.6	176.0	151.4	197.5	-0.04	1.00	1446	162.6	6.5	163.0	138.7	183.4	-0.11	1.00
DESIR	Age (yrs)	178	52.7	5.59	52.0	45.0	64.0	0.02	-0.21	538	49.0	8.55	49.0	30.0	65.0	0.44	-0.20
	BMI (kg/m ²)	178	23.2	1.16	23.3	20.4	24.9	1.00	0.08	538	21.4	1.85	21.2	16.6	24.9	1.00	-0.11
	Weight (kg)	178	69.5	6.49	70.0	51.0	89.0	0.60	0.85	538	54.8	5.86	55.0	40.0	71.0	0.74	0.58
	Height (m)	178	173.1	6.46	173.0	156.0	195.0	0.08	1.00	538	160.1	5.80	160.0	145.0	177.0	-0.11	1.00
	WC (cm)	177	84.6	5.38	84.0	73.0	101.0	0.50	0.41	536	70.9	5.79	70.0	57.0	98.0	0.69	0.12
	Hip (cm)	177	95.1	4.05	95.0	84.0	107.0	0.45	0.62	536	93.0	5.42	93.0	77.0	108.0	0.65	0.28
	WHR (cm/cm)	177	0.9	0.04	0.9	0.8	1.0	0.23	0.01	537	0.8	0.05	0.8	0.6	1.1	0.28	-0.06

DNBC	Age (yrs)	NA	NA	NA	NA	NA	NA	NA	NA	1802	29.3	4.20	29.2	16.3	43.5	0.02	0.03
	BMI (kg/m ²)	NA	NA	NA	NA	NA	NA	NA	NA	1802	23.5	4.31	22.6	15.2	47.8	1.00	-0.06
	Weight (kg)	NA	NA	NA	NA	NA	NA	NA	NA	1802	67.1	12.93	65.0	36.0	135.0	0.89	0.36
	Height (m)	NA	NA	NA	NA	NA	NA	NA	NA	1802	1.7	0.06	1.7	1.5	1.9	-0.06	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
EGCUT_370	Age (yrs)	416	37.0	15.60	33.0	18.0	87.0	0.28	-0.47	450	37.3	15.61	34.0	18.0	83.0	0.42	-0.32
	BMI (kg/m ²)	416	25.6	4.13	25.2	17.3	48.3	1.00	-0.11	450	24.4	4.78	23.4	17.0	42.9	1.00	-0.18
	Weight (kg)	416	83.8	14.45	83.0	50.0	160.0	0.89	0.35	450	67.8	13.20	66.0	40.0	136.0	0.92	0.20
	Height (m)	416	180.9	7.09	181.0	158.0	204.0	-0.11	1.00	450	166.7	6.19	167.0	144.0	184.0	-0.18	1.00
	WC (cm)	416	90.5	11.94	89.0	64.0	148.0	0.86	-0.01	450	79.1	11.72	77.0	46.0	121.0	0.86	-0.10
	Hip (cm)	416	102.0	7.17	101.0	82.0	148.0	0.82	0.18	450	102.0	9.63	100.0	78.0	167.0	0.84	0.06
	WHR (cm/cm)	416	0.9	0.08	0.9	0.7	1.1	0.63	-0.16	450	0.8	0.07	0.8	0.5	1.0	0.52	-0.23
EGCUT_OMNI	Age (yrs)	517	46.4	15.77	46.0	20.0	74.0	0.22	-0.37	839	67.9	21.62	80.0	18.0	103.0	0.21	-0.49
	BMI (kg/m ²)	517	26.9	4.44	26.5	18.1	44.1	1.00	-0.10	839	27.1	5.40	26.3	11.5	74.5	1.00	-0.11
	Weight (kg)	517	86.6	15.21	85.0	57.0	153.0	0.89	0.35	839	70.3	14.53	69.0	28.0	172.0	0.90	0.32
	Height (m)	517	179.4	7.01	180.0	158.0	205.0	-0.10	1.00	839	161.1	7.28	161.0	140.0	193.0	-0.11	1.00
	WC (cm)	517	95.7	13.05	95.0	56.0	144.0	0.87	0.02	839	88.5	13.32	88.0	60.0	160.0	0.79	0.03
	Hip (cm)	517	103.3	8.88	103.0	58.0	136.0	0.71	0.19	839	104.7	11.74	103.0	63.0	172.0	0.77	0.15
	WHR (cm/cm)	517	0.9	0.09	0.9	0.7	1.2	0.59	-0.15	839	0.8	0.08	0.8	0.6	1.6	0.35	-0.13
ERF	Age (yrs)	1218	49.5	14.21	49.6	17.6	86.5	0.15	-0.46	1508	48.3	14.42	48.0	18.0	86.1	0.25	-0.42
	BMI (kg/m ²)	1218	27.2	4.14	26.7	15.9	50.8	1.00	-0.08	1507	26.7	5.16	25.7	15.5	61.8	1.00	-0.12
	Weight (kg)	1218	83.1	13.92	81.6	41.9	154.7	0.87	0.42	1507	69.7	14.07	67.2	42.1	161.0	0.91	0.29
	Height (m)	1218	174.7	7.21	174.7	152.2	196.5	-0.08	1.00	1508	161.8	6.56	161.6	139.3	182.8	-0.12	1.00
	WC (cm)	1218	94.2	11.70	93.0	62.5	146.6	0.87	-0.03	1503	82.3	12.70	80.1	54.3	149.0	0.89	-0.06
	Hip (cm)	1218	99.4	7.27	98.6	76.5	147.5	0.85	0.27	1504	79.0	173.00	100.3	101.6	9.7	0.86	0.17
	WHR (cm/cm)	1218	0.9	0.08	0.9	0.7	1.2	0.56	-0.28	1503	0.8	0.08	0.8	0.5	1.3	0.52	-0.25
FamHS	Age (yrs)	632	48.1	13.76	46.5	25.6	85.7	0.15	-0.24	831	47.5	12.94	45.2	25.7	81.4	0.19	-0.26
	BMI (kg/m ²)	632	27.8	4.32	27.2	18.4	46.2	1.00	-0.09	831	27.1	6.09	26.1	16.5	55.0	1.00	-0.12
	Weight (kg)	632	87.1	14.63	85.3	55.3	140.6	0.88	0.39	831	72.3	16.62	68.9	41.7	144.2	0.94	0.22
	Height (m)	632	177.0	6.92	177.0	157.0	203.0	-0.09	1.00	831	163.3	6.45	163.0	141.0	196.0	-0.12	1.00
	WC (cm)	631	99.3	12.54	99.0	44.0	147.0	0.86	0.06	831	92.6	16.93	90.0	27.0	170.0	0.89	-0.01
	Hip (cm)	631	104.2	8.32	104.0	53.0	140.0	0.82	0.27	831	106.8	12.38	104.0	59.0	162.0	0.91	0.10
	WHR (cm/cm)	631	1.0	0.08	1.0	0.4	1.8	0.47	-0.17	831	0.9	0.09	0.9	0.3	1.6	0.52	-0.15
Health ABC	Age (yrs)	873	73.9	2.87	74.0	69.0	80.0	-0.08	-0.13	782	73.6	2.80	73.0	69.0	80.0	-0.08	-0.15
	BMI (kg/m ²)	873	27.1	3.69	26.6	17.6	44.2	1.00	-0.05	782	26.1	4.53	25.6	15.6	44.7	1.00	-0.09
	Weight (kg)	873	81.6	12.39	80.0	52.2	134.5	0.87	0.44	782	66.5	12.09	65.1	40.8	123.0	0.92	0.31
	Height (m)	873	1.7	0.06	1.7	1.5	1.9	-0.05	1.00	782	1.6	0.06	1.6	1.4	1.8	-0.09	1.00
	WC (cm)	873	102.1	10.98	101.3	59.4	224.7	0.81	0.16	782	96.1	12.35	96.4	65.0	144.0	0.75	0.13
	Hip (cm)	n/a	n/a	n/a	n/a	n/a	n/a	n/a	n/a	n/a	n/a	n/a	n/a	n/a	n/a	n/a	n/a
	WHR (cm/cm)	n/a	n/a	n/a	n/a	n/a	n/a	n/a	n/a	n/a	n/a	n/a	n/a	n/a	n/a	n/a	n/a
HERITAGE	Age (yrs)	244	36.5	14.95	31.9	17.0	64.3	0.35	-0.21	256	35.1	14.14	31.2	17.2	65.2	0.41	-0.11
	BMI (kg/m ²)	244	26.7	4.86	25.8	17.3	44.2	1.00	-0.06	256	25.0	4.89	23.9	17.0	47.5	1.00	-0.08

	Weight (kg)	244	84.4	16.20	81.2	54.0	140.2	0.93	0.32	256	67.1	13.69	64.0	39.6	115.7	0.92	0.31
	Height (cm)	244	177.8	6.36	177.9	160.0	196.8	-0.06	1.00	256	163.6	6.41	163.7	147.4	184.7	-0.08	1.00
	WC (cm)	244	94.7	13.89	93.6	69.2	135.6	0.95	0.06	256	86.2	14.53	84.7	64.2	143.8	0.89	0.05
	Hip (cm)	244	103.1	8.80	102.1	86.5	131.1	0.91	0.24	256	102.5	10.67	100.3	81.3	146.8	0.91	0.20
	WHR (cm/cm)	244	0.9	0.07	0.9	0.8	1.1	0.75	-0.16	256	0.8	0.08	0.8	0.7	1.1	0.60	-0.14
HYPERGENES Controls	Age (yrs)	1105	62.2	10.6	59.8	28.0	98.0	-0.09	-0.12	791	64.2	11.2	61.0	44.9	113.0	-0.15	-0.14
	BMI (kg/m ²)	1092	26.0	3.3	25.6	10.2	40.8	1.00	-0.13	776	25.0	3.7	24.6	16.5	41.4	1.00	-0.15
	Weight (kg)	1119	76.4	11.5	75.0	29.0	186.0	0.82	0.46	793	64.3	10.2	63.0	41.0	110.0	0.87	0.35
	Height (m)	1092	1.7	0.1	1.7	1.5	2.0	-0.13	1.00	776	1.6	0.1	1.6	1.4	1.8	-0.15	1.00
	WC (cm)	178	95.6	10.7	93.7	58.0	140.0	0.75	0.27	192	84.7	10.5	82.4	62.8	119.8	0.81	0.18
	Hip (cm)	178	102.8	6.8	102.6	87.0	137.0	0.72	0.32	192	103.3	8.1	102.5	84.0	134.5	0.83	0.25
	WHR (cm/cm)	178	0.93	0.06	0.93	0.66	1.09	0.57	0.14	192	0.82	0.07	0.81	0.68	1.19	0.40	0.04
HYPERGENES Controls	Age (yrs)	1197	49.4	10.4	50.0	17.6	84.0	0.04	-0.33	602	48.5	9.6	49.0	18.4	93.0	0.10	-0.19
	BMI (kg/m ²)	1208	27.4	3.5	27.1	16.0	47.4	1.00	-0.08	621	26.9	5.0	26.3	17.5	52.3	1.00	-0.08
	Weight (kg)	1209	81.4	12.1	80.0	49.0	139.5	0.82	0.51	623	68.8	13.7	67.0	44.0	164.0	0.89	0.37
	Height (m)	1208	1.7	0.1	1.7	1.5	2.0	-0.08	1.00	621	1.6	0.1	1.6	1.4	2.0	-0.08	1.00
	WC (cm)	214	97.7	11.7	97.0	71.0	139.0	0.87	0.17	189	89.2	13.7	88.0	61.0	133.0	0.89	0.06
	Hip (cm)	214	104.8	9.8	104.0	86.0	180.0	0.71	0.28	189	107.5	11.5	105.0	88.5	158.0	0.91	0.11
	WHR (cm/cm)	214	0.93	0.07	0.94	0.52	1.15	0.47	-0.05	189	0.8	0.1	0.8	0.6	1.0	0.46	-0.03
inCHIANTI	Age (yrs)	513	66.6	15.31	70.0	23.0	94.0	0.03	-0.50	625	68.3	15.14	71.0	21.0	95.0	0.20	-0.50
	BMI (kg/m ²)	511	27.0	3.40	27.0	18.1	37.4	1.00	0.02	624	27.3	4.67	27.0	18.0	46.6	1.00	-0.21
	Weight (kg)	511	75.7	12.03	75.5	44.0	120.0	0.80	0.61	624	64.9	11.44	64.3	41.0	107.5	0.85	0.32
	Height (m)	513	167.1	7.98	167.0	142.0	193.0	0.02	1.00	625	154.3	7.36	155.0	135.0	176.0	-0.21	1.00
	WC (cm)	499	94.5	9.34	95.0	65.0	126.0	0.79	0.20	611	88.8	11.78	89.0	58.0	124.0	0.84	-0.08
	Hip (cm)	499	99.2	7.30	99.0	75.0	140.0	0.73	0.33	611	101.6	9.76	101.0	75.0	166.0	0.81	0.09
	WHR (cm/cm)	499	1.0	0.06	1.0	0.6	1.2	0.42	-0.09	611	0.9	0.07	0.9	0.5	1.1	0.40	-0.25
IPM EA controls	Age (yrs)	334	58.0	13.59	59.0	26.0	88.0	0.14	-0.26	170	54.0	16.25	55.0	24.0	87.0	0.28	-0.29
	BMI (kg/m ²)	334	27.4	5.08	26.5	18.4	45.6	1.00	-0.05	170	27.6	7.45	25.9	15.8	62.0	1.00	-0.20
	Weight (kg)	334	86.4	17.08	81.8	51.8	148.2	0.88	0.38	170	72.9	19.48	68.2	41.8	159.1	0.95	0.10
	Height (m)	334	1.8	0.07	1.8	1.5	2.0	-0.05	1.00	170	1.6	0.07	1.6	1.4	1.9	-0.20	1.00
IPM EA cases	Age (yrs)	138	67.3	11.16	67.5	34.0	93.0	-0.15	-0.03	62	69.7	11.55	69.0	34.0	90.0	-0.22	-0.24
	BMI (kg/m ²)	138	28.3	5.75	27.2	15.2	48.9	1.00	-0.02	62	28.6	6.74	27.2	16.6	48.7	1.00	-0.02
	Weight (kg)	138	87.8	19.30	83.9	45.5	145.5	0.91	0.36	62	73.9	17.81	70.0	43.6	125.0	0.94	0.26
	Height (m)	138	1.8	0.07	1.8	1.6	1.9	-0.02	1.00	62	1.6	0.06	1.6	1.5	1.8	-0.02	1.00
Lifelines	Age (yrs)	3480	48.1	11.30	47.8	19.6	87.6	0.16	-0.25	4641	44.0	11.06	47.2	18.1	89.0	0.21	-0.28
	BMI (kg/m ²)	3479	26.6	3.64	26.2	14.4	48.9	1.00	-0.09	4639	25.7	4.69	25.1	13.9	51.8	1.00	-0.17
	Weight (kg)	3479	88.3	13.25	87.0	49.5	165.0	0.83	0.43	4639	73.9	13.58	72.0	42.5	152.0	0.88	0.27
	Height (m)	3479	182.1	6.70	182.0	158.0	208.0	-0.09	1.00	4639	169.7	6.42	169.0	145.0	193.0	-0.17	1.00
	WC (cm)	3479	96.4	10.70	96.0	65.0	158.0	0.84	0.08	4638	86.8	12.25	86.5	58.0	146.0	0.83	-0.02
	Hip (cm)	3479	100.2	7.44	100.0	73.0	163.0	0.70	0.30	4637	100.7	10.32	100.0	69.0	181.0	0.84	0.08
	WHR (cm/cm)	3479	1.0	0.07	1.0	0.6	1.3	0.58	-0.15	4637	0.9	0.07	0.9	0.5	1.3	0.39	-0.11
LLS	Age (yrs)	872	60.0	6.60	60.0	33.6	80.3	0.03	-0.21	1049	58.6	6.60	58.5	30.2	76.1	0.07	-0.10
	BMI (kg/m ²)	869	25.7	2.91	25.4	18.1	35.9	1.00	-0.04	1034	24.9	3.50	24.5	16.3	35.8	1.00	-0.12
	Weight (kg)	864	82.1	10.60	82.0	49.0	113.0	0.82	0.51	1045	69.6	10.90	68.0	45.0	113.0	0.89	0.33

	Height (m)	864	178.8	6.60	178.0	155.0	201.0	-0.07	1.00	1045	166.7	6.00	167.0	143.0	186.0	-0.14	1.00
	WC (cm)	317	102.0	8.50	102.0	77.3	130.0	0.82	0.17	314	95.3	10.30	95.0	69.0	126.0	0.78	0.12
	Hip (cm)	318	105.4	6.10	105.0	87.5	128.0	0.69	0.30	308	104.0	7.00	104.0	82.5	126.0	0.81	0.14
	WHR (cm/cm)	315	1.0	0.06	1.0	0.8	1.1	0.48	-0.05	315	0.9	0.06	0.9	0.7	1.1	0.23	0.10
LOLIPOP_EW610	Age (yrs)	678	56.1	9.80	56.4	35.1	74.9	0.06	-0.23	249	55.7	9.70	56.6	35.3	75.0	0.07	-0.23
	BMI (kg/m ²)	678	27.8	4.40	27.3	18.0	46.4	1.00	-0.05	249	26.6	5.10	25.4	17.7	47.9	1.00	-0.14
	Weight (kg)	678	85.6	15.00	83.5	46.0	150.4	0.89	0.40	249	70.6	13.80	68.2	43.4	118.1	0.91	0.28
	Height (m)	678	175.4	6.90	175.5	150.0	196.0	-0.05	1.00	249	162.8	6.60	163.0	146.0	187.5	-0.14	1.00
	WC (cm)	677	98.4	11.60	97.3	72.0	146.0	0.90	0.12	248	88.3	13.60	87.0	62.0	134.0	0.85	-0.03
	Hip (cm)	677	103.8	8.10	103.0	85.0	142.0	0.84	0.24	248	102.1	9.90	101.0	83.0	134.0	0.86	0.15
	WHR (cm/cm)	677	0.9	0.06	0.9	0.8	1.2	0.62	-0.06	248	0.9	0.10	0.8	0.7	1.2	0.46	-0.18
LOLIPOP_EWA	Age (yrs)	513	54.4	10.10	55.5	22.7	75.0	0.05	-0.30	NA	NA	NA	NA	NA	NA	NA	NA
	BMI (kg/m ²)	513	28.3	4.60	27.9	16.9	51.5	1.00	-0.02	NA	NA	NA	NA	NA	NA	NA	NA
	Weight (kg)	513	86.2	15.50	84.5	44.8	172.0	0.89	0.43	NA	NA	NA	NA	NA	NA	NA	NA
	Height (m)	513	174.5	7.08	174.0	153.5	197.2	-0.02	1.00	NA	NA	NA	NA	NA	NA	NA	NA
	WC (cm)	511	99.6	12.12	99.0	40.0	156.0	0.88	0.11	NA	NA	NA	NA	NA	NA	NA	NA
	Hip (cm)	511	104.0	8.60	104.0	39.0	153.0	0.75	0.23	NA	NA	NA	NA	NA	NA	NA	NA
	WHR (cm/cm)	511	1.0	0.07	1.0	0.7	1.2	0.61	-0.08	NA	NA	NA	NA	NA	NA	NA	NA
LOLIPOP_EWP	Age (yrs)	651	55.7	9.10	58.4	32.4	67.3	-0.02	-0.18	NA	NA	NA	NA	NA	NA	NA	NA
	BMI (kg/m ²)	651	28.6	5.30	27.9	16.5	58.3	1.00	0.00	NA	NA	NA	NA	NA	NA	NA	NA
	Weight (kg)	651	87.4	17.30	85.3	45.8	152.4	0.92	0.39	NA	NA	NA	NA	NA	NA	NA	NA
	Height (m)	651	174.5	6.81	175.0	154.0	201.4	0.00	1.00	NA	NA	NA	NA	NA	NA	NA	NA
	WC (cm)	650	100.9	13.60	63.0	100.0	152.0	0.92	0.17	NA	NA	NA	NA	NA	NA	NA	NA
	Hip (cm)	650	104.9	9.00	81.0	104.0	152.0	0.80	0.27	NA	NA	NA	NA	NA	NA	NA	NA
	WHR (cm/cm)	650	1.0	0.07	0.6	1.0	1.2	0.60	0.01	NA	NA	NA	NA	NA	NA	NA	NA
NELSON	Age (yrs)	2668	59.9	5.51	59.0	50.0	79.0	0.01	-0.18	NA	NA	NA	NA	NA	NA	NA	NA
	BMI (kg/m ²)	1135	27.1	3.64	26.8	15.3	42.4	1.00	0.00	NA	NA	NA	NA	NA	NA	NA	NA
	Weight (kg)	1135	86.1	13.11	85.0	50.0	147.0	0.86	0.51	NA	NA	NA	NA	NA	NA	NA	NA
	Height (m)	2668	178.8	6.25	179.0	157.0	201.0	0.00	1.00	NA	NA	NA	NA	NA	NA	NA	NA
PLCO2 controls	Age (yrs)	649	63.6	5.20	64.0	55.0	74.0	-0.16	-0.08	544	63.6	5.20	64.0	55.0	74.0	-0.06	-0.07
	BMI (kg/m ²)	645	27.0	3.90	26.5	17.2	40.5	1.00	0.00	544	26.4	5.00	25.7	16.7	50.3	1.00	-0.08
	Weight (kg)	645	86.6	14.00	83.9	52.2	145.1	0.89	0.45	544	70.3	14.20	68.0	41.3	149.7	0.92	0.31
	Height (m)	649	1.8	0.07	1.8	1.6	2.0	0.00	1.00	544	1.6	0.06	1.6	1.5	1.9	-0.08	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
PLCO2 cases	Age (yrs)	2042	64.2	5.10	64.0	55.0	74.0	-0.08	-0.08	934	64.0	5.30	64.0	55.0	74.0	-0.02	-0.11
	BMI (kg/m ²)	2040	27.5	4.20	26.9	15.9	50.2	1.00	-0.02	933	26.7	5.10	25.9	16.5	52.2	1.00	-0.07
	Weight (kg)	2040	87.6	14.80	85.3	52.2	176.9	0.90	0.42	933	71.0	14.50	68.0	40.8	137.9	0.91	0.33
	Height (m)	2042	1.8	0.07	1.8	1.6	2.1	-0.02	1.00	934	1.6	0.06	1.6	1.2	1.9	-0.07	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
PREVEND	Age (yrs)	1870	50.9	12.8	50.0	28.0	75.0	0.20	-0.34	1752	48.2	12.0	47.0	28.0	75.0	0.29	-0.37

	BMI (kg/m ²)	1870	26.4	3.7	26.0	17.2	44.7	1.00	-0.18	1752	25.8	4.8	25.0	17.1	53.7	1.00	-0.22	
	Weight (kg)	1870	85.1	12.5	84.0	49.0	146.0	0.85	0.36	1752	72.3	13.2	70.0	44.9	144.5	0.91	0.20	
	Height (m)	1870	179.5	7.0	179.0	157.5	207.0	-0.18	1.00	1752	167.5	6.5	167.5	145.0	193.0	-0.22	1.00	
	WC (cm)	1868	94.5	11.2	94.0	31.0	141.5	0.84	-0.02	1752	83.3	13.0	81.5	57.0	150.0	0.86	-0.08	
	Hip (cm)	1868	100.1	6.7	100.0	70.0	128.0	0.78	0.23	1752	101.0	10.4	100.0	68.0	170.0	0.86	0.02	
	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	
PROSPER/ PHASE	Age (yrs)	2524	75.0	3.27	74.5	70.2	83.3	-0.05	-0.11	2720	75.7	3.40	75.4	69.4	83.4	-0.05	-0.08	
	BMI (kg/m ²)	2524	26.6	3.58	26.3	15.2	45.1	1.00	-0.06	2720	27.1	4.70	26.7	15.6	50.1	1.00	-0.08	
	Weight (kg)	2524	78.7	11.93	78.0	40.0	127.0	0.86	0.47	2719	68.3	12.70	67.0	35.5	138.0	-0.89	0.38	
	Height (m)	2524	172.1	6.67	172.0	143.0	198.0	-0.06	1.00	2718	158.8	6.60	159.0	135.0	180.0	-0.08	1.00	
PROCARDIS	Age (yrs)	5334	60.4	8.10	61.0	20.0	83.0	-0.01	-0.15	1948	62.0	7.60	63.0	30.0	86.0	0.01	-0.19	
	BMI (kg/m ²)	5332	28.0	4.10	27.5	17.0	55.4	1.00	-0.05	1947	28.2	5.50	27.2	15.4	62.0	1.00	-0.17	
	Weight (kg)	5327	84.5	13.40	83.0	46.0	174.0	0.85	0.39	1947	72.8	14.40	71.0	33.0	159.0	0.90	0.26	
	Height (cm)	5334	173.7	6.90	174.0	139.0	206.0	-0.09	1.00	1948	160.9	6.90	161.0	130.0	185.0	-0.17	1.00	
	WC (cm)	687	99.8	10.00	99.0	71.0	141.0	0.86	0.10	360	88.9	12.20	88.0	64.0	137.0	0.86	0.09	
	Hip (cm)	687	101.8	6.60	101.0	85.0	130.0	0.77	0.22	360	101.4	9.10	100.0	80.0	173.0	0.86	0.14	
	WHR (cm/cm)	687	1.0	0.06	1.0	0.8	1.2	0.55	-0.06	360	0.9	0.08	0.9	0.7	1.2	0.42	0.00	
QFS	Age (yrs)	370	42.5	16.32	44.5	18.3	88.7	0.19	-0.42	490	42.9	16.92	44.2	18.1	93.5	0.09	-0.44	
	BMI (kg/m ²)	370	27.5	6.45	26.0	17.4	57.3	1.00	-0.06	490	27.9	8.45	25.2	16.8	64.9	1.00	-0.12	
	Weight (kg)	370	82.0	19.80	77.5	45.5	167.5	0.95	0.26	490	71.3	21.76	64.5	35.0	181.1	0.96	0.16	
	Height (m)	370	1.7	0.07	1.7	1.5	1.9	-0.06	1.00	490	1.6	0.07	1.6	1.4	1.8	-0.12	1.00	
	WC (cm)	366	94.2	16.66	90.7	65.4	164.5	0.95	0.01	466	84.9	18.68	79.1	57.9	151.0	0.95	-0.06	
	Hip (cm)	363	100.9	11.41	98.1	78.6	169.5	0.94	0.15	465	105.7	16.83	101.0	79.4	200.0	0.95	0.02	
	WHR (cm/cm)	363	0.9	0.08	0.9	0.8	1.1	0.67	-0.19	465	0.8	0.08	0.8	0.5	1.1	0.55	-0.17	
QIMR	Age (yrs)	1470	44.7	16.34	44.0	18.0	91.0	0.27	-0.16	2157	44.0	15.31	42.0	18.0	90.0	0.30	-0.19	
	BMI (kg/m ²)	1470	26.0	3.93	25.6	14.8	60.9	1.00	-0.13	2157	25.0	5.08	23.9	14.2	64.5	1.00	-0.17	
	Weight (kg)	1470	82.2	13.17	81.0	47.0	180.0	0.87	0.36	2157	66.6	13.69	64.0	34.0	160.0	0.91	0.25	
	Height (m)	1470	1.8	0.07	1.8	1.5	2.0	-0.13	1.00	2157	1.6	0.07	1.6	1.3	2.0	-0.17	1.00	
	WC (cm)	NA	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	NA
	WHR (cm/cm)	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA
RISC	Age (yrs)	453	43.3	8.60	43.0	30.0	60.0	0.06	-0.12	578	44.5	8.20	44.0	30.0	61.0	0.18	-0.16	
	BMI (kg/m ²)	453	26.4	3.50	26.1	17.0	43.9	1.00	-0.03	578	24.8	4.20	23.9	16.9	41.7	1.00	-0.13	
	Weight (kg)	453	84.0	12.90	82.0	48.8	143.9	0.86	0.48	578	67.5	11.90	65.8	42.1	119.0	0.90	0.31	
	Height (m)	453	178.4	6.90	178.0	149.0	203.0	-0.03	1.00	578	165.0	6.30	165.0	147.0	187.0	-0.13	1.00	
	WC (cm)	447	93.8	10.20	93.0	65.0	147.0	0.80	0.19	570	80.7	11.50	79.0	49.0	120.0	0.82	0.08	
	Hip (cm)	447	101.7	8.40	101.0	62.0	140.0	0.65	0.32	570	99.6	9.70	99.0	57.0	135.0	0.80	0.13	
	WHR (cm/cm)	447	0.9	0.07	0.9	0.7	1.7	0.42	-0.10	570	0.8	0.08	0.8	0.6	1.3	0.35	-0.01	
SHIP-TREND	Age (yrs)	432	50.1	14.25	50.0	22.0	80.0	0.28	-0.36	554	50.1	13.26	51.0	20.0	81.0	0.25	-0.41	
	BMI (kg/m ²)	432	27.8	3.80	27.7	17.7	43.9	1.00	-0.07	554	27.0	5.08	26.3	18.5	53.6	1.00	-0.19	
	Weight (kg)	432	86.9	13.05	85.5	53.3	129.8	0.87	0.43	554	72.7	13.73	71.2	45.5	136.3	0.91	0.23	
	Height (m)	432	1.8	0.07	1.8	1.6	2.0	-0.07	1.00	554	1.6	0.06	1.7	1.4	1.8	-0.19	1.00	
	WC (cm)	432	94.4	11.20	94.0	66.4	138.8	0.88	0.06	554	83.1	11.98	81.5	54.5	122.8	0.90	-0.07	
	Hip (cm)	432	101.2	7.41	101.0	80.5	135.0	0.79	0.27	554	101.4	11.02	100.0	72.5	149.5	0.87	0.06	

	WHR (cm/cm)	432	0.9	0.07	0.9	0.7	1.1	0.64	-0.17	554	0.8	0.07	0.8	0.6	1.3	0.43	-0.20
TRAILS	Age (yrs)	539	19.2	0.54	19.2	18.2	20.7	0.06	-0.09	603	19.1	0.57	19.1	18.2	20.8	0.07	-0.08
	BMI (kg/m ²)	539	22.6	3.80	21.9	15.6	43.2	1.00	-0.03	602	23.0	3.71	22.4	15.9	45.6	1.00	-0.16
	Weight (kg)	539	76.3	13.99	73.5	48.4	138.0	0.90	0.40	602	66.5	10.94	65.5	42.5	125.0	0.89	0.31
	Height (m)	539	183.6	7.28	183.5	165.0	206.0	-0.03	1.00	603	170.1	6.56	170.5	141.5	191.5	-0.16	1.00
	WC (cm)	537	81.8	9.81	79.5	64.5	130.8	0.87	0.20	602	75.3	8.76	73.6	46.0	130.0	0.86	0.05
	Hip (cm)	537	99.4	8.39	98.0	75.5	138.5	0.82	0.31	602	99.9	8.49	99.3	51.0	133.8	0.79	0.16
	WHR (cm/cm)	537	0.8	0.05	0.8	0.7	1.1	0.49	-0.03	602	0.8	0.05	0.7	0.6	1.0	0.41	-0.10
TWINGENE	Age (yrs)	4432	65.6	8.04	65.2	47.6	93.3	-0.05	-0.29	4948	64.7	8.22	64.3	47.4	93.9	0.04	-0.32
	BMI (kg/m ²)	4349	26.3	3.54	25.9	15.4	57.8	1.00	-0.07	4827	25.8	4.31	25.1	15.0	67.0	1.00	-0.13
	Weight (kg)	4349	81.9	12.22	80.5	50.0	179.1	0.86	0.44	4827	68.8	11.96	67.1	37.6	171.5	0.90	0.31
	Height (m)	4432	1.8	0.07	1.8	1.4	2.1	-0.07	1.00	4948	1.6	0.06	1.6	1.4	1.9	-0.13	1.00
	WC (cm)	4414	97.2	10.25	97.0	52.5	193.0	0.82	0.14	4929	87.0	11.58	86.0	40.0	199.0	0.83	0.06
	Hip (cm)	4399	103.2	7.99	103.0	41.0	197.0	0.64	0.25	4922	103.4	9.39	102.0	38.0	172.0	0.83	0.12
	WHR (cm/cm)	4396	0.9	0.09	0.9	0.5	2.6	0.37	-0.05	4917	0.8	0.08	0.8	0.5	1.9	0.37	-0.04
TwinsUK	Age (yrs)	NA	NA	NA	NA	NA	NA	NA	NA	3003	47.2	12.81	48.9	16.1	79.0	0.18	-0.21
	BMI (kg/m ²)	NA	NA	NA	NA	NA	NA	NA	NA	3003	25.2	4.71	24.3	13.2	52.4	1.00	-0.14
	Weight (kg)	NA	NA	NA	NA	NA	NA	NA	NA	3003	66.3	12.63	64.1	35.1	140.9	0.91	0.26
	Height (m)	NA	NA	NA	NA	NA	NA	NA	NA	3003	1.6	0.06	1.6	1.4	1.8	-0.14	1.00
	WC (cm)	NA	NA	NA	NA	NA	NA	NA	NA	2201	79.1	10.54	77.0	53.0	134.0	0.85	0.04
	Hip (cm)	NA	NA	NA	NA	NA	NA	NA	NA	2200	101.5	9.86	100.0	69.0	168.0	0.87	0.12
	WHR (cm/cm)	NA	NA	NA	NA	NA	NA	NA	NA	2200	0.8	0.06	0.8	0.6	1.4	0.35	-0.07

Supplementary Table 20. Study design, number of individuals and sample quality control for MetaboChip study cohorts.

Study		Study design	Total sample Size (N)	Sample QC		Samples in analyses (N)	Anthropometric assessment method	References
Short name	Full name			Call rate*	other exclusions			
ADVANCE-CAD controls	Atherosclerotic Disease, Vascular Function, & Genetic Epidemiology study	Population-based case control study of CAD	705	≥95%	1) missing body weight and/or height data 2) sex discrepancies	663	measured	1) Go, A.S. et al. Statin and beta-blocker therapy and the initial presentation of coronary heart disease. <i>Ann Intern Med</i> 144, 229-38 (2006). 2) Taylor-Piliae, R.E. et al. Validation of a new brief physical activity survey among men and women aged 60-69 years. <i>Am J Epidemiol</i> 164, 598-606 (2006). 3) Iribarren, C. et al. Metabolic syndrome and early-onset coronary artery disease: is the whole greater than its parts? <i>J Am Coll Cardiol</i> 48, 1800-7 (2006). 4) Assimes, T.L. et al. Susceptibility locus for clinical and subclinical coronary artery disease at chromosome 9p21 in the multi-ethnic ADVANCE study. <i>Hum Mol Genet</i> 17, 2320-8 (2008).
ADVANCE-CAD cases	Atherosclerotic Disease, Vascular Function, & Genetic Epidemiology study	Population-based case control study of CAD	974	≥95%	1) missing body weight and/or height data 2) sex discrepancies	895	measured	1) Go, A.S. et al. Statin and beta-blocker therapy and the initial presentation of coronary heart disease. <i>Ann Intern Med</i> 144, 229-38 (2006). 2) Taylor-Piliae, R.E. et al. Validation of a new brief physical activity survey among men and women aged 60-69 years. <i>Am J Epidemiol</i> 164, 598-606 (2006). 3) Iribarren, C. et al. Metabolic syndrome and early-onset coronary artery disease: is the whole greater than its parts? <i>J Am Coll Cardiol</i> 48, 1800-7 (2006). 4) Assimes, T.L. et al. Susceptibility locus for clinical and subclinical coronary artery disease at chromosome 9p21 in the multi-ethnic ADVANCE study. <i>Hum Mol Genet</i> 17, 2320-8 (2008).
AMC-PAS	Academic Medical Centre Amsterdam Premature Atherosclerosis Study	CAD cases	706	0.97	1) heterozygosity 2) ethnic outliers 3) duplicates 4) missing phenotypes 5) call rate < 0.95	490	measured	Coronary Artery Disease, C. et al. Large scale association analysis of novel genetic loci for coronary artery disease. <i>Arterioscler Thromb Vasc Biol</i> 29, 774-80 (2009).
B1958C	1958 British Birth Cohort	Population-based	2359	MAF ≥5%: ≥95% 1% ≤MAF <5% ≥99%	1) MAF ≥1%; genotype clustering 2) call rate 3) HWE 4) MI of alleles 5) concordance rate 6) correct SNP mapping	2211	measured	
BHS	Busselton Health Study	Population-based	270	≥99%	1) Relatedness 2) Missing height measurement	263	measured	James, A.L. et al. Decline in lung function in the Busselton Health Study: the effects of asthma and cigarette smoking. <i>Am J Respir Crit Care Med</i> 171, 109-14 (2005).
CARDIOGENICS	CARDIOGENICS	CAD case-control	806	0.98	1) heterozygosity 2) ethnic outliers 3) duplicates 4) missing phenotypes 5) call rate < 0.95	754	measured	Samani, N.J. et al. Genomewide association analysis of coronary artery disease. <i>N Engl J Med</i> 357, 443-53 (2007).

D2D 2007	FIN-D2D study, 2007 cohort	Population-based	2720	≥98.15%	1) Missing BMI, age, age<18. 2) Gender discrepancy or anomaly 3) Unexplained duplicate sample	2688	measured	Kotronen, A. et al. 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 10, 237 (2010).
DESIR	Data from an Epidemiological Study on the Insulin Resistance syndrome	Population-based	4993	≥90%	1) Missing data	3925	measured	Balkau, B. [An epidemiologic survey from a network of French Health Examination Centres, (D.E.S.I.R.): epidemiologic data on the insulin resistance syndrome]. Rev Epidemiol Sante Publique 44, 373-5 (1996).
DIAGEN	The DIAbetes GENetics Study	Clinical prevention study	1535	≥98.15%	1) Missing BMI, age, age<18. 2) Gender discrepancy or anomaly 3) Unexplained duplicate sample	1462	measured	Schwarz, P.E., et al. Hypoadiponectinemia is associated with progression toward type 2 diabetes and genetic variation in the ADIPOQ gene promoter. Diabetes Care 29, 1645-50 (2006).
DILGOM	Dietary, life style, and genetic determinants of obesity and metabolic syndrome	Population-based	3997	≥95%	1) heterozygosity <23.9% or >27.6%; 2) ethnic outliers; 3) related individuals and duplicates.	3938	measured	1) Konttinen, H. et al. Emotional eating, depressive symptoms and self-reported food consumption. A population-based study. Appetite 54, 473-9 (2010). 2) Inouye, M. et al. An immune response network associated with blood lipid levels. PLoS Genet 6, e1001113 (2010). 3) Peltonen, M. et al. The National FINRISK 2007 Study. Helsinki: Publications of the National Public Health Institute, B 34/2008 (2008).
DPS	Diabetes Prevention Study	Population-based	522	≥98.15%	1) Missing BMI, age, age<18. 2) Gender discrepancy or anomaly 3) Unexplained duplicate sample	472	measured	Tuomilehto, J. et al. Prevention of type 2 diabetes mellitus by changes in lifestyle among subjects with impaired glucose tolerance. N Engl J Med 344, 1343-50 (2001).
DR'S EXTRA	DR'S EXTRA	Population-based	1408	≥98.15%	1) Missing BMI, age, age<18. 2) Gender discrepancy or anomaly 3) Unexplained duplicate sample	1295	measured	Kouki, R. et al. Diet, fitness and metabolic syndrome--the DR's EXTRA study. Nutr Metab Cardiovasc Dis 22, 553-60 (2012).
DUNDEE cases	DUNDEE cases	Population-based	3582	MAF ≥5%: ≥95% ≥1% MAF<5%: ≥99%	1) MAF≥1% 2) genotype clustering 3) call rate 4) HWE 5) MI of alleles 6) concordance rate 7) correct SNP mapping	3582	measured	

DUNDEE controls	DUNDEE controls	Population-based	3962	MAF $\geq 5\%$: $\geq 95\%$ $\geq 1\%$ MAF $< 5\%$: $\geq 99\%$	1) MAF $\geq 1\%$ 2) genotype clustering 3) call rate 4) HWE 5) MI of alleles 6) concordance rate 7) correct SNP mapping	3709	measured	
EAS	Edinburgh Artery Study	Population based	733	$\geq 90\%$	1) Missing body weight and height. 2) Heterozygosity	733	measured	Fowkes, F.G. et al. Edinburgh Artery Study: prevalence of asymptomatic and symptomatic peripheral arterial disease in the general population. <i>Int J Epidemiol</i> 20, 384-92 (1991).
EGCUT-CAD	Estonian Genome Center of University of Tartu - Cardiovascular Disease cases	Case-Controls	700	$\geq 84.6\%$	1) Missing body weight and height. 2) Cryptic relatedness	700	measured	Nelis, M. et al. Genetic structure of Europeans: a view from the North-East. <i>PLoS One</i> 4, e5472 (2009).
EGCUT-CONTROLS	Estonian Genome Center of University of Tartu - Controls	Case-Controls	942	$\geq 84.6\%$	1) Missing body weight and height. 2) Cryptic relatedness	942	measured	Nelis, M. et al. Genetic structure of Europeans: a view from the North-East. <i>PLoS One</i> 4, e5472 (2009).
EGCUT-T2D	Estonian Genome Center of University of Tartu - Type 2 Diabetes cases	Case-Controls	968	$\geq 84.6\%$	1) Missing body weight and height. 2) Cryptic relatedness	968	measured	Nelis, M. et al. Genetic structure of Europeans: a view from the North-East. <i>PLoS One</i> 4, e5472 (2009).
Ely Study	MRC Ely Study	Population-based	1625	$\geq 95\%$	1) missing phenotype data 2) gender check; 3) duplicates check;	1600	measured	Forouhi, N.G. et al. Incidence of Type 2 diabetes in England and its association with baseline impaired fasting glucose: the Ely study 1990-2000. <i>Diabet Med</i> 24, 200-7 (2007).
EMIL	Echinococcus Multilocularis and Internal Diseases in Leutkirch (EMIL) study	Population based	N/A	$\geq 95\%$	1) Missing phenotype 2) non-European ancestry	1722	measured	1) Boehm, B.O. et al. Prevalence of the metabolic syndrome in southwest Germany. <i>Scand J Clin Lab Invest Suppl</i> 240, 122-8 (2005). 2) Haenle, M.M. et al. Overweight, physical activity, tobacco and alcohol consumption in a cross-sectional random sample of German adults. <i>BMC Public Health</i> 6, 233 (2006).
EPIC-Norfolk T2D	EPIC (European Prospective Investigation into Cancer) Norfolk T2D case-Cohort Study	Population-based	978 - cohort	$\geq 95\%$	1) Missing body weight and height, or case-control status. 2) Heterozygosity 3) gender check	963 - cohort	measured	1) Day, N. et al. EPIC-Norfolk: study design and characteristics of the cohort. <i>European Prospective Investigation of Cancer. Br J Cancer</i> 80 Suppl 1, 95-103 (1999). 2) Loos, R.J. et al. Common variants near MC4R are associated with fat mass, weight and risk of obesity. <i>Nat Genet</i> 40, 768-75 (2008).
			735 - T2D			727 - T2D		
Fenland	The Fenland Study	Population-based	3251	$\geq 95\%$	1) missing phenotype data 2) gender check; 3) duplicates check;	3186	measured	Willer, C.J. et al. Six new loci associated with body mass index highlight a neuronal influence on body weight regulation. <i>Nat Genet</i> 41, 25-34 (2009).
FUSION stage 2	FUSION stage 2	case-control	2951	$\geq 98.15\%$	1) Missing BMI, age, age < 18 . 2) Gender discrepancy or anomaly 3) Unexplained duplicate sample	2929	measured	Scott, L.J. et al. A genome-wide association study of type 2 diabetes in Finns detects multiple susceptibility variants. <i>Science</i> 316, 1341-5 (2007).

GLACIER	Gene x Lifestyle interactions And Complex traits Involved in Elevated disease Risk	Prospective cohort study	6,311	≥95%	1) Missing phenotype 2) Duplicates 3) Call rate 4) Heterozygosity 5) Gender fails	6047	measured	1) Renstrom, F. et al. Genetic predisposition to long-term nondiabetic deteriorations in glucose homeostasis: Ten-year follow-up of the GLACIER study. <i>Diabetes</i> 60, 345-54 (2011). 2) Hallmans, G. et al. Cardiovascular disease and diabetes in the Northern Sweden Health and Disease Study Cohort - evaluation of risk factors and their interactions. <i>Scand J Public Health Suppl</i> 61, 18-24 (2003). 3) Renstrom, F. et al. Replication and extension of genome-wide association study results for obesity in 4923 adults from northern Sweden. <i>Hum Mol Genet</i> 18, 1489-96 (2009).
HNR	Heinz Nixdorf Recall	Population-based	4570	≥97%	1) Missing BMI 2) heterozygosity 3) ethnic outlier 4) related individuals 5) duplicates	4500	measured	1) 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> 20, 489-96 (2005). 2) 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> 30, 1867-72 (2010).
HUNT 2	The Nord-Trøndelag Health Study 2	Population-based, although T2D case-control samples selected for metabochip typing	1567	≥98.15%	1) Missing BMI, age, age<18, forms of diabetes other than T2D (cases) 2) Gender discrepancy or anomaly 3) Unexplained duplicate sample	1334	measured	Holmen J, Midthjell K, et al. The Nord-Trøndelag Health Study 1995-97 (HUNT 2): Objectives, contents, methods and participation. <i>Nor J Epidemiol</i> 13(1):19-32 (2003).
IMPROVE	Carotid Intima Media Thickness [IMT] and IMT-Progression as Predictors of Vascular Events in a High Risk European Population	Population-based	NA	≥95%	1) ambiguous sex 2) cryptic relatedness 3) non-european descent	3,425	measured	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> 31, 614-22 (2010).
KORA S3	Cooperative Health Research in the Region of Augsburg (third survey), KOoperative Gesundheitsforschung in der Region Augsburg (dritte Studie)	Population-based	3113	none	none	3,113	measured	Wichmann, H.E., Gieger, C., Illig, T. & Group, M.K.S. KORA-gen--resource for population genetics, controls and a broad spectrum of disease phenotypes. <i>Gesundheitswesen</i> 67 Suppl 1, S26-30 (2005).
KORA S4	Cooperative Health Research in the Region of Augsburg (forth survey), KOoperative Gesundheitsforschung in der Region Augsburg (vierte Studie)	Population-based	3028	none	none	3,028	measured	Wichmann, H.E., Gieger, C., Illig, T. & Group, M.K.S. KORA-gen--resource for population genetics, controls and a broad spectrum of disease phenotypes. <i>Gesundheitswesen</i> 67 Suppl 1, S26-30 (2005).

Leipzig Adults	Leipzig adults	Population-based	1005	≥95%	1) ethnic outliers 2) gender discordance	1,005	measured	Speliotes, E.K. et al. Association analyses of 249,796 individuals reveal 18 new loci associated with body mass index. <i>Nat Genet</i> 42, 937-48 (2010).
LURIC - Cases	Ludwigshafen Risk and Cardiovascular Health Study	Case - Control	2583	≥95%	1) missing metabochip data 2) ambiguous sex 3) duplicates, 4) relatedness	2251	measured	Winkelmann, B.R. et al. Rationale and design of the LURIC study--a resource for functional genomics, pharmacogenomics and long-term prognosis of cardiovascular disease. <i>Pharmacogenomics</i> 2, S1-73 (2001).
LURIC - Controls	Ludwigshafen Risk and Cardiovascular Health Study	Case - Control	733	≥95%	1) missing metabochip data 2) ambiguous sex 3) duplicates, 4) relatedness	645	measured	Winkelmann, B.R. et al. Rationale and design of the LURIC study--a resource for functional genomics, pharmacogenomics and long-term prognosis of cardiovascular disease. <i>Pharmacogenomics</i> 2, S1-73 (2001).
METSIM	Metabolic Syndrome In Men	Population-based, although T2D case-control samples selected for metabochip typing	2176	≥98.15%	1) Missing BMI and age 2) age<18 3) Gender discrepancy or anomaly 4) Unexplained duplicate sample	2014	measured	Stancakova, A. et al. Changes in insulin sensitivity and insulin release in relation to glycemia and glucose tolerance in 6,414 Finnish men. <i>Diabetes</i> 58, 1212-21 (2009).
MORGAM Cases	MOnica Risk, Genetics, Archiving and Monograph	CVD cases from population-based follow-up cohorts.	~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	1) Evans, A. et al. MORGAM (an international pooling of cardiovascular cohorts). <i>Int J Epidemiol</i> 34, 21-7 (2005). 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 Controls	MOnica Risk, Genetics, Archiving and Monograph	Population-based follow-up cohorts.	~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	1) Evans, A. et al. MORGAM (an international pooling of cardiovascular cohorts). <i>Int J Epidemiol</i> 34, 21-7 (2005). 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	5,362	≥95%	1) Missing body weight and height. 2) Heterozygosity 3) gender check	988	measured	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, 49-54 (2006).
PIVUS	Prospective Investigation of the Vasculature in Uppsala Seniors	Population-based	999	≥90%	1) Related individuals and duplicates 2) Heterozygosity, (F-mean(F))/sd(Z) ≥ 5 3) Missing phenotypes used in analysis	978	measured	Lind, L., Fors, N., Hall, J., Marttala, K. & Stenborg, A. 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. <i>Arterioscler Thromb Vasc Biol</i> 25, 2368-75 (2005).
STR	Swedish Twin Registry	Population-based	2702	≥90%	1) Heterozygosity, (F-mean(F))/sd(Z) ≥ 5 2) Missing phenotypes used in analysis	2185	measured	Lichtenstein, P. et al. The Swedish Twin Registry in the third millennium: an update. <i>Twin Res Hum Genet</i> 9, 875-82 (2006).

THISEAS	The Hellenic study of Interactions between Snps and Eating in Atherosclerosis Susceptibility	CAD case-control	1887	≥97%	1) heterozygosity 2) ethnic outliers 3) duplicates 4) missing phenotypes 5) call rate< 0.95	1413	measured	
Tromsø	Tromsø 4	Population-based, although T2D case-control samples selected for metabochip typing	1421	≥98.15%	1) Missing BMI, age, age<18. 2) Gender discrepancy or anomaly 3) Unexplained duplicate sample	1418	measured	Jacobsen, B.K., Eggen, A.E., Mathiesen, E.B., Wilsgaard, T. & Njolstad, I. Cohort profile: the Tromso Study. <i>Int J Epidemiol</i> 41, 961-7 (2012).
ULSAM	Uppsala Longitudinal Study of Adult Men	Population-based	1221	≥90%	1) Related individuals and duplicates 2) Heterozygosity, (F-mean(F))/sd(Z) ≥ 5 3) Missing phenotypes used in analysis	1112	measured	Ingelsson, E., Sundstrom, J., Arnlov, J., Zethelius, B. & Lind, L. Insulin resistance and risk of congestive heart failure. <i>JAMA</i> 294, 334-41 (2005).
WHITEHALL	The Whitehall II study	Cohort of London-based civil servants	3413	≥95%	1) missing phenotype data 2) gender check; 3) duplicates check;	2234	measured	1) Marmot, M. & Brunner, E. Cohort Profile: the Whitehall II study. <i>Int J Epidemiol</i> 34, 251-6 (2005). 2) Jensen, A.C. 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. <i>Diabetes</i> 60, 1617-23 (2011).
WTCCC-T2D	Wellcome Trust Case Control Consortium - T2D non GWAS	Population-based	1335	MAF ≥5% : ≥95% ≥1% MAF<5%: ≥99%	1) MAF ≥ 1% 2) call rate 3) HWE 4) MI of alleles 5) concordance rate 6) correct SNP mapping 7) genotype clustering	1077	measured	Wellcome Trust Case Control, C. Genome-wide association study of 14,000 cases of seven common diseases and 3,000 shared controls. <i>Nature</i> 447, 661-78 (2007).

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

Supplementary Table 21. Information on genotyping methods, quality control of SNPs, imputation, and statistical analysis for Metabochip study cohort

Study	GENOTYPING					ASSOCIATION ANALYSIS	
	Genotype calling algorithm	Inclusion criteria			SNPs that met QC criteria	SNPs in meta-analysis	Analyses software
		MAF	Call rate*	P for HWE			
ADVANCE-CAD controls (males)	GenCall	>0%	ALL	>0.001	195,405	195,405	PLINK
ADVANCE-CAD controls (females)	GenCall	>0%	ALL	>0.001	195,679	195,679	PLINK
ADVANCE-CAD cases (males)	GenCall	>0%	ALL	>0.001	195,050	195,050	PLINK
ADVANCE-CAD cases (females)	GenCall	>0%	ALL	>0.001	195,827	195,827	PLINK
AMC-PAS	GenoSNP	ALL	≥98%	>10 ⁻⁴	169,807	169,807	PLINK
B1958C	GeneCall	≥1%	≥95%	>10 ⁻⁶	180,655	180,655	SNPTEST
BHS	GenomeStudio	ALL	ALL	ALL	129,565	129,565	GenABEL
CARDIOGENICS	GenoSNP	ALL	≥98%	>10 ⁻⁵	177,614	177,614	PLINK
D2D 2007 cases	BeadStudio version 3.3.7, GentraIn version 1.0	>0%	≥98.15%	ALL	138,569	138,569	EMMAX
D2D 2007 controls	BeadStudio version 3.3.7, GentraIn version 1.0	>0%	≥98.15%	none	145,465	145,465	EMMAX
DIAGEN cases	BeadStudio version 3.3.7, GentraIn version 1.0	>0%	≥98.15%	none	137,937	137,937	EMMAX
DIAGEN controls	BeadStudio version 3.3.7, GentraIn version 1.0	>0%	≥98.15%	none	142,744	142,744	EMMAX
DESIR	GeneCall	ALL	ALL	none	194,763	194,763	PLINK
DILGOM	GenCall	ALL	≥95%	none	183,872	183,872	PLINK
DPS cases	BeadStudio version 3.3.7, GentraIn version 1.0	>0%	≥98.15%	none	138,569	138,569	EMMAX
DPS controls	BeadStudio version 3.3.7, GentraIn version 1.0	>0%	≥98.15%	none	145,465	145,465	EMMAX
DR'S EXTRA cases	BeadStudio version 3.3.7, GentraIn version 1.0	>0%	≥98.15%	none	138,569	138,569	EMMAX
DR'S EXTRA controls	BeadStudio version 3.3.7, GentraIn version 1.0	>0%	≥98.15%	none	145,465	145,465	EMMAX
DUNDEE cases	GeneCall	≥1%	≥95%	>10 ⁻⁶	180,399	180,399	SNPTEST
DUNDEE controls	GeneCall	≥1%	≥95%	>10 ⁻⁶	180,399	180,399	SNPTEST

EAS	GeneCall	>0%	≥90%	>10 ⁻⁶	184,558	134,470	PLINK
EGCUT-CAD	GeneCall	>0%	≥90%	>10 ⁻⁶	182,516	130,213	GenABEL
EGCUT-CONTROLS	GeneCall	>0%	≥90%	>10 ⁻⁶	182,516	132,363	GenABEL
EGCUT-T2D	GeneCall	>0%	≥90%	>10 ⁻⁶	182,516	131,317	GenABEL
EMIL	GenCall	NA	≥95%	>10 ⁻⁶	183,031	183,031	PLINK
Ely Study	GeneCall	>0%	≥90%	>10 ⁻⁶	149,302	149,302	PLINK
EPIC-Norfolk T2D	GeneCall	>0%	≥90%	>10 ⁻⁶	143,294	143,294	PLINK
Fenland	GeneCall	>0%	≥90%	>10 ⁻⁶	167,085	167,085	PLINK
FUSION stage 2 cases	BeadStudio version 3.3.7, Gentrain version 1.0	>0%	≥98.15%	none	138,569	138,569	EMMAX
FUSION stage 2 controls	BeadStudio version 3.3.7, Gentrain version 1.0	>0%	≥98.15%	none	145,465	145,465	EMMAX
GLACIER	Illuminus	>0%	≥95%	>10 ⁻⁵	149,782	149,782	PLINK
HNR	Genetrain2	ALL	≥95%	>10 ⁻⁶	192,261	192,168	PLINK
HUNT 2 cases	BeadStudio version 3.3.7, Gentrain version 1.0	>0%	≥98.15%	NA	143,806	143,806	EMMAX
HUNT 2 controls	BeadStudio version 3.3.7, Gentrain version 1.0	>0%	≥98.15%	NA	143,806	143,806	EMMAX
IMPROVE	GeneCall	>0%	≥95%	>10 ⁻⁶	185,704	183,645	PLINK
KORA S3	Genome Studio	ALL	ALL	none	185,781	185,781	PLINK
KORA S4	Genome Studio	ALL	ALL	none	185,781	185,781	PLINK
Leipzig Adults	GeneCall	>0%	≥95%	MAF >0.05: >5.7x10 ⁻⁷ MAF <0.05: >1x10 ⁻⁴	141,071	141,071	SNPTEST
LURIC	GeneTrain 2	ALL	≥90%	>10 ⁻⁶	191,341	191,341	PLINK
METSIN cases	BeadStudio version 3.3.7, Gentrain version 1.0	>0%	≥98.15%	none	138,569	138,569	EMMAX
METSIN controls	BeadStudio version 3.3.7, Gentrain version 1.0	>0%	≥98.15%	none	145,465	145,465	EMMAX
MORGAM Cases	GenCall	ALL	ALL	none	196,725	196,725	PLINK
MORGAM Controls	GenCall	ALL	ALL	none	196,725	196,725	PLINK
NSHD	GeneCall	>0%	≥90%	>10 ⁻⁶	146,474	146,474	PLINK
PIVUS	GenCall	ALL	≥90%	>10 ⁻⁶	185,450	185,450	PLINK

STR	GenCall	ALL	≥90%	>10 ⁻⁶	183,961	183,961	Merlin
THISEAS	GenoSNP	ALL	≥98%	>10 ⁻⁷	169,779	169,779	PLINK
Tromsø cases	BeadStudio version 3.3.7, Gentrain version 1.0	>0%	≥98.15%	none	139,415	139,415	EMMAX
Tromsø controls	BeadStudio version 3.3.7, Gentrain version 1.0	>0%	≥98.15%	none	143,806	143,806	EMMAX
ULSAM	GenCall	ALL	≥90%	>10 ⁻⁶	183,357	183,357	PLINK
WHITEHALL	GeneCall	>0%	≥90%	>10 ⁻⁶	171,257	171,257	PLINK
WTCCC-T2D	GeneCall	≥1%	≥95%	>10 ⁻⁶	180,655	180,655	SNPTEST

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

Supplementary Table 22: Study-specific descriptive statistics of MetaboChip cohorts

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
ADVANCE-CAD controls	Age (yrs)	420	65.80	2.90	65.30	61.00	72.30	-0.03	-0.02	243	65.70	2.90	65.50	60.60	71.20	-0.15	-0.14
	BMI (kg/m ²)	420	28.40	4.50	27.60	19.90	46.60	1.00	-0.09	243	28.00	6.20	27.00	17.40	50.40	1.00	-0.13
	Weight (kg)	420	87.90	14.90	85.20	59.60	136.40	0.88	0.39	243	73.70	16.50	70.70	44.20	128.20	0.94	0.21
	Height (m)	420	1.76	0.07	1.76	1.57	1.95	-0.09	1.00	243	1.62	0.06	1.63	1.44	1.78	-0.13	1.00
	WC (cm)	419	99.50	12.60	98.50	66.50	154.00	0.89	0.11	243	85.80	13.40	84.80	58.70	131.60	0.92	-0.04
	Hip (cm)	NA	NA	NA	NA	NA	NA	NA	NA	0	NA	NA	NA	NA	NA	NA	NA
	WHR (cm/cm)	NA	NA	NA	NA	NA	NA	NA	NA	0	NA	NA	NA	NA	NA	NA	NA
ADVANCE-CAD cases	Age (yrs)	675	61.50	8.20	61.20	34.00	87.50	-0.19	-0.09	220	66.20	6.30	66.40	47.90	80.10	-0.81	-0.22
	BMI (kg/m ²)	675	29.10	4.80	28.20	17.80	53.70	1.00	-0.03	220	29.00	6.40	28.00	16.90	66.10	1.00	-0.17
	Weight (kg)	675	90.29	16.04	87.91	48.40	164.65	0.90	0.40	220	75.57	16.52	73.44	44.91	149.10	0.92	0.22
	Height (m)	675	1.80	0.10	1.80	1.60	2.00	-0.03	1.00	220	1.60	0.10	1.60	1.40	1.80	-0.17	1.00
	WC (cm)	675	99.90	12.00	98.40	70.20	148.00	0.89	0.17	217	89.20	14.10	88.40	59.90	132.30	0.90	0.03
	Hip (cm)	0	NA	NA	NA	NA	NA	NA	NA	0	NA	NA	NA	NA	NA	NA	NA
	WHR (cm/cm)	0	NA	NA	NA	NA	NA	NA	NA	0	NA	NA	NA	NA	NA	NA	NA
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)	0	NA	NA	NA	NA	NA	NA	NA	0	NA	NA	NA	NA	NA	NA	NA
	Hip (cm)	0	NA	NA	NA	NA	NA	NA	NA	0	NA	NA	NA	NA	NA	NA	NA
	WHR (cm/cm)	0	NA	NA	NA	NA	NA	NA	NA	0	NA	NA	NA	NA	NA	NA	NA
B1958C	Age (yrs)	1234	45.00	0.00	45.00	45.00	45.00	NA	NA	902	45.00	0.00	45.00	45.00	45.00	NA	NA
	BMI (kg/m ²)	1232	27.80	4.20	27.30	16.50	53.60	1.00	-0.02	902	26.60	5.30	25.30	17.70	54.30	1.00	-0.16
	Weight (kg)	1234	85.90	14.80	84.60	47.00	158.90	0.89	0.41	902	70.10	14.30	67.00	42.00	143.50	0.92	0.23
	Height (m)	1234	1.76	0.07	1.76	1.56	1.98	-0.02	1.00	902	1.62	0.06	1.63	1.22	1.84	-0.16	1.00
	WC (cm)	1234	98.10	11.80	97.20	73.10	151.20	0.80	0.20	902	83.90	14.30	82.40	60.50	133.80	0.76	0.05
	Hip (cm)	1234	105.30	9.10	105.10	86.20	152.40	0.64	0.26	902	103.90	13.80	102.40	82.80	157.60	0.71	0.10
	WHR (cm/cm)	1234	0.90	0.50	0.90	0.74	1.20	0.08	0.01	902	0.80	0.70	0.80	0.63	1.10	0.02	0.06
BHS	Age (yrs)	170	69.80	8.73	71.30	35.90	91.10	-0.01	-0.20	93	70.50	9.73	71.70	41.60	91.60	-0.06	-0.33
	BMI (kg/m ²)	169	26.70	3.59	26.30	18.00	38.70	1.00	-0.03	93	26.30	4.65	25.40	18.70	44.50	1.00	0.07
	Weight (kg)	169	79.90	12.20	79.80	51.40	109.00	0.86	0.49	93	67.60	13.10	66.00	46.00	107.00	0.94	0.42
	Height (m)	170	1.73	0.07	1.72	1.56	1.89	-0.03	1.00	93	1.60	0.06	1.60	1.49	1.76	0.07	1.00
	WC (cm)	166	97.00	10.40	95.90	69.00	129.50	0.88	0.19	91	84.80	11.10	82.00	65.20	119.00	0.88	0.18
	Hip (cm)	164	100.70	7.70	100.90	82.00	137.00	0.83	0.25	91	102.70	10.90	100.20	86.00	140.20	0.93	0.19
	WHR (cm/cm)	164	0.96	0.06	0.97	0.78	1.12	0.49	0.01	91	0.82	0.06	0.82	0.70	0.96	0.26	0.05
CARDIOGENICS	Age (yrs)	476	55.72	8.32	56.00	26.00	87.00	-0.04	-0.19	278	54.25	7.64	55.00	39.00	79.00	0.06	-0.12
	BMI (kg/m ²)	476	27.73	4.11	27.10	17.10	42.20	1.00	-0.09	278	26.01	4.37	25.30	17.70	43.80	1.00	-0.02

	Weight (kg)	476	86.41	13.80	84.00	36.00	133.00	0.87	0.40	278	70.13	12.99	68.00	43.00	127.00	0.91	0.39
	Height (m)	476	1.76	0.07	1.78	1.45	1.98	-0.09	1.00	278	1.64	0.06	1.65	1.50	1.80	-0.02	1.00
	WC (cm)	0	NA	NA	NA	NA	NA	NA	NA	0	NA	NA	NA	NA	NA	NA	NA
	Hip (cm)	0	NA	NA	NA	NA	NA	NA	NA	0	NA	NA	NA	NA	NA	NA	NA
	WHR (cm/cm)	0	NA	NA	NA	NA	NA	NA	NA	0	NA	NA	NA	NA	NA	NA	NA
D2D 2007 cases	Age (yrs)	270	63.13	7.82	65.00	45.00	74.00	-0.06	-0.21	187	63.66	7.11	65.00	45.00	74.00	-0.08	-0.26
	BMI (kg/m ²)	270	29.95	5.01	29.14	20.20	48.27	1.00	0.03	187	31.38	6.12	30.66	19.27	52.68	1.00	0.06
	Weight (kg)	270	92.26	16.46	90.40	57.70	150.00	0.90	0.41	187	81.68	16.75	79.10	49.90	137.40	0.95	0.35
	Height (m)	270	1.75	0.06	1.75	1.56	1.95	0.03	1.00	187	1.61	0.05	1.61	1.48	1.74	0.06	1.00
	WC (cm)	269	107.74	12.19	106.00	79.00	150.00	0.91	0.21	187	101.14	14.92	101.50	67.00	145.00	0.91	0.12
	Hip (cm)	270	104.57	8.95	103.00	84.50	145.00	0.86	0.27	187	109.11	11.63	108.00	85.00	152.00	0.90	0.19
	WHR (cm/cm)	269	1.03	0.06	1.03	0.83	1.22	0.55	0.06	187	0.93	0.07	0.92	0.73	1.14	0.53	-0.02
D2D 2007 controls	Age (yrs)	1014	59.62	8.42	60.00	45.00	74.00	0.05	-0.30	1217	58.78	8.24	58.00	45.00	74.00	0.12	-0.28
	BMI (kg/m ²)	1014	26.82	3.75	26.45	16.49	48.95	1.00	-0.03	1217	26.97	4.93	26.30	17.21	61.34	1.00	-0.12
	Weight (kg)	1014	83.10	13.33	81.50	46.50	160.00	0.85	0.46	1217	71.45	13.50	69.80	41.90	158.60	0.91	0.27
	Height (m)	1014	1.76	0.07	1.76	1.45	1.98	-0.03	1.00	1217	1.63	0.06	1.63	1.41	1.83	-0.12	1.00
	WC (cm)	1014	98.05	10.86	97.00	63.00	149.00	0.89	0.15	1215	88.89	12.23	87.50	62.00	150.00	0.91	0.05
	Hip (cm)	1014	99.59	6.78	99.00	79.00	141.00	0.79	0.33	1215	102.23	9.35	101.00	79.50	150.00	0.90	0.13
	WHR (cm/cm)	1014	0.98	0.06	0.98	0.79	1.24	0.68	-0.07	1214	0.87	0.06	0.86	0.72	1.17	0.59	-0.04
DESIR	Age (yrs)	2108	46.14	10.16	45.00	30.00	65.00	0.30	-0.32	1817	46.18	10.35	45.00	30.00	65.00	0.32	-0.30
	BMI (kg/m ²)	2081	25.61	3.38	25.30	16.70	45.40	1.00	-0.12	1777	24.79	4.28	24.00	15.40	53.60	1.00	-0.20
	Weight (kg)	2081	76.32	10.98	75.00	46.00	140.00	0.85	0.41	1777	62.98	10.94	61.00	37.00	127.00	0.90	0.24
	Height (m)	2082	172.63	6.57	172.00	150.00	196.00	-0.12	1.00	1778	159.47	6.04	160.00	141.00	188.00	-0.20	1.00
	WC (cm)	2077	90.12	9.70	90.00	64.00	133.00	0.88	0.03	1773	78.78	10.67	77.00	57.00	125.00	0.87	-0.05
	Hip (cm)	2077	97.65	6.43	97.00	78.00	134.00	0.79	0.26	1773	98.58	8.97	98.00	69.00	160.00	0.84	0.08
	WHR (cm/cm)	2077	0.92	0.06	0.92	0.71	1.21	0.60	-0.20	1773	0.80	0.07	0.79	0.54	1.06	0.46	-0.17
DIAGEN cases	Age (yrs)	210	63.80	10.80	64.00	37.00	96.00	-0.23	-0.19	215	67.83	12.54	67.00	33.00	93.00	-0.33	-0.25
	BMI (kg/m ²)	210	29.25	4.63	28.66	18.94	44.64	1.00	0.02	215	30.97	7.12	30.04	15.63	61.71	1.00	0.07
	Weight (kg)	210	88.18	15.36	86.25	58.00	134.40	0.88	0.45	215	79.77	20.29	77.70	40.00	168.00	0.93	0.39
	Height (m)	210	1.73	0.07	1.74	1.56	1.92	0.02	1.00	215	1.60	0.06	1.60	1.41	1.79	0.07	1.00
	WC (cm)	210	106.59	11.65	105.00	74.00	138.00	0.84	0.22	215	102.40	15.97	102.00	67.00	150.00	0.81	0.23
	Hip (cm)	210	106.56	8.65	106.00	81.00	129.00	0.72	0.25	215	110.94	15.07	108.00	71.00	154.00	0.80	0.27
	WHR (cm/cm)	210	1.00	0.06	0.99	0.83	1.15	0.52	0.05	215	0.92	0.08	0.92	0.72	1.16	0.19	0.01
DIAGEN controls	Age (yrs)	450	62.80	14.28	63.00	22.00	96.00	-0.19	-0.38	587	61.72	15.44	62.00	18.00	94.00	0.04	-0.34
	BMI (kg/m ²)	450	26.85	3.42	26.30	18.94	39.88	1.00	0.01	587	27.00	5.04	26.22	17.55	58.94	1.00	-0.01
	Weight (kg)	450	82.38	12.67	80.90	50.00	137.00	0.80	0.56	587	71.74	14.63	70.00	40.00	149.00	0.89	0.40
	Height (m)	450	1.75	0.07	1.75	1.53	1.96	0.01	1.00	587	1.63	0.07	1.63	1.42	1.85	0.40	1.00
	WC (cm)	450	99.15	10.15	99.00	68.00	142.00	0.76	0.13	587	91.43	13.74	90.00	60.00	148.00	0.74	0.06
	Hip (cm)	450	103.47	7.05	103.00	84.00	131.00	0.61	0.31	587	105.49	12.00	105.00	72.00	165.00	0.83	0.14
	WHR (cm/cm)	450	0.96	0.07	0.95	0.65	1.20	0.50	-0.11	587	0.87	0.09	0.86	0.63	1.26	0.27	-0.08
DILGOM	Age (yrs)	1797	53.28	13.47	55.00	25.00	74.00	0.15	-0.32	2139	51.74	13.58	53.00	25.00	74.00	0.24	-0.38
	BMI (kg/m ²)	1791	27.24	4.17	26.65	15.82	63.11	1.00	-0.06	2139	26.85	5.36	25.77	16.04	52.53	1.00	-0.18
	Weight (kg)	1791	84.00	14.06	82.20	50.10	192.60	0.89	0.41	2139	70.82	14.22	68.50	38.50	144.50	0.92	0.20
	Height (m)	1797	175.50	6.79	175.40	151.70	201.00	-0.06	1.00	2138	162.50	6.25	162.50	138.40	184.10	-0.18	1.00

	WC (cm)	1787	96.78	11.97	95.50	64.00	172.00	0.91	0.07	2122	87.02	13.50	85.00	58.00	143.00	0.92	-0.03
	Hip (cm)	1790	100.50	7.63	100.00	80.50	161.00	0.85	0.25	2122	102.00	10.82	100.50	75.50	162.00	0.92	0.02
	WHR (cm/cm)	1784	0.96	0.07	0.96	0.75	1.22	0.65	-0.15	2116	0.85	0.06	0.85	0.68	1.10	0.56	-0.09
DPS cases	Age (yrs)	32	56.00	6.69	58.59	43.69	64.78	-0.12	-0.21	57	54.90	6.22	54.93	43.28	67.79	-0.13	-0.18
	BMI (kg/m ²)	32	30.63	3.13	31.54	24.37	37.81	1.00	-0.08	57	32.92	4.36	32.19	25.59	45.76	1.00	-0.26
	Weight (kg)	32	94.01	11.43	90.00	78.20	122.50	0.86	0.34	57	87.36	12.51	86.60	67.00	136.50	0.77	0.34
	Height (m)	32	1.75	0.06	1.76	1.65	1.90	-0.08	1.00	57	1.63	0.06	1.62	1.48	1.77	-0.26	1.00
	WC (cm)	32	107.22	9.23	105.50	88.30	127.00	0.75	0.22	57	103.00	10.31	102.50	82.30	126.00	0.73	0.14
	Hip (cm)	32	106.19	6.46	105.15	97.00	119.00	0.79	0.24	57	113.98	8.61	113.60	99.00	140.00	0.79	0.10
	WHR (cm/cm)	32	1.01	0.05	1.01	0.89	1.09	0.35	-0.01	57	0.90	0.06	0.90	0.79	1.03	0.26	0.04
DPS controls	Age (yrs)	116	56.40	7.08	59.00	41.00	65.90	0.07	-0.35	267	55.01	7.29	56.94	40.09	67.91	-0.18	-0.20
	BMI (kg/m ²)	116	29.55	3.45	29.13	23.50	44.80	1.00	0.02	267	31.72	4.85	30.64	23.99	50.50	1.00	0.05
	Weight (kg)	116	90.64	12.45	88.95	62.90	148.10	0.82	0.55	267	82.99	14.30	80.00	52.90	131.10	0.88	0.48
	Height (m)	116	1.75	0.06	1.75	1.56	1.89	0.02	1.00	267	1.62	0.06	1.62	1.44	1.78	0.05	1.00
	WC (cm)	116	103.09	9.32	102.25	83.50	139.00	0.85	0.22	265	99.28	11.30	98.00	78.00	139.00	0.84	0.23
	Hip (cm)	116	104.94	7.58	104.65	88.00	143.50	0.80	0.32	267	112.12	10.78	110.00	90.30	161.00	0.87	0.25
	WHR (cm/cm)	116	0.98	0.05	0.98	0.86	1.08	0.41	-0.04	265	0.89	0.06	0.89	0.72	1.06	0.19	0.09
DR'S EXTRA Cases	Age (yrs)	60	69.06	5.88	70.84	57.81	78.12	-0.24	-0.40	61	68.28	5.74	69.67	57.92	77.72	-0.05	-0.35
	BMI (kg/m ²)	60	29.41	4.76	28.68	20.96	41.23	1.00	0.19	61	32.39	6.14	31.71	19.96	48.40	1.00	-0.12
	Weight (kg)	60	86.75	16.43	83.65	47.60	120.50	0.91	0.53	61	81.80	16.19	80.60	54.10	129.90	0.90	0.25
	Height (m)	60	1.71	0.07	1.70	1.47	1.96	0.19	1.00	61	1.59	0.06	1.60	1.40	1.74	-0.12	1.00
	WC (cm)	60	104.35	12.79	104.25	80.00	133.75	0.93	0.32	61	103.27	14.37	101.75	72.00	136.50	0.91	0.04
	Hip (cm)	60	103.32	10.41	101.00	81.00	132.50	0.87	0.41	61	110.29	11.53	109.00	91.00	145.00	0.88	0.00
	WHR (cm/cm)	60	1.01	0.06	1.02	0.87	1.14	0.46	0.00	61	0.93	0.07	0.92	0.77	1.18	0.53	-0.03
DR'S EXTRA Controls	Age (yrs)	542	66.37	5.44	66.11	57.36	78.46	-0.13	-0.27	632	66.40	5.24	66.20	57.71	78.70	0.02	-0.30
	BMI (kg/m ²)	542	27.14	3.51	26.83	19.31	44.21	1.00	0.06	632	27.16	4.62	26.26	15.96	48.57	1.00	-0.12
	Weight (kg)	542	82.13	12.40	80.80	51.30	135.90	0.86	0.53	632	69.47	12.22	67.85	41.90	143.20	0.90	0.29
	Height (m)	542	1.74	0.06	1.74	1.57	1.93	0.06	1.00	632	1.60	0.06	1.60	1.42	1.78	-0.12	1.00
	WC (cm)	542	97.49	10.07	96.63	73.00	136.50	0.87	0.28	632	87.44	11.99	86.25	59.50	130.50	0.89	0.03
	Hip (cm)	542	98.93	7.20	99.00	78.00	134.00	0.81	0.37	632	101.16	9.36	99.75	78.50	139.50	0.89	0.09
	WHR (cm/cm)	542	0.98	0.05	0.98	0.84	1.12	0.59	0.05	632	0.86	0.06	0.86	0.70	1.05	0.56	-0.06
DUNDEE controls	Age (yrs)	1918	60.20	11.09	61.00	36.00	79.00	0.01	-0.26	1789	57.99	11.45	57.00	36.00	79.00	0.01	-0.24
	BMI (kg/m ²)	1918	27.26	3.97	26.90	16.20	54.90	1.00	-0.08	1788	26.81	5.02	26.00	14.30	51.60	1.00	-0.15
	Weight (kg)	1918	84.63	13.34	83.80	46.90	168.00	0.88	0.40	1788	70.46	13.50	68.55	33.00	125.60	0.91	0.27
	Height (m)	1918	1.76	0.07	1.76	1.51	1.98	-0.08	1.00	1789	1.62	0.07	1.62	1.43	1.85	-0.15	1.00
	WC (cm)	1917	98.49	10.73	98.00	69.00	172.00	0.86	0.11	1787	86.69	12.30	86.00	61.00	153.00	0.84	0.02
	Hip (cm)	0	NA	NA	NA	NA	NA	NA	NA	0	NA	NA	NA	NA	NA	NA	NA
	WHR (cm/cm)	0	NA	NA	NA	NA	NA	NA	NA	0	NA	NA	NA	NA	NA	NA	NA
DUNDEE cases	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)	0	NA	NA	NA	NA	NA	NA	NA	0	NA	NA	NA	NA	NA	NA	NA

	WHR (cm/cm)	0	NA	NA	NA	NA	NA	NA	NA	0	NA	NA	NA	NA	NA	NA	NA	
EAS	Age (yrs)	353	64.73	5.60	64.76	54.73	75.36	-0.01	-0.22	378	64.24	5.71	63.93	54.92	75.20	0.03	-0.24	
	BMI (kg/m ²)	353	25.28	3.08	25.15	16.69	39.55	1.00	-0.06	378	25.27	4.19	24.61	17.38	51.46	1.00	-0.11	
	Weight (kg)	353	75.02	10.61	74.30	48.80	123.91	0.82	0.52	378	64.45	11.37	63.09	38.90	118.89	0.88	0.37	
	Height (m)	353	172.17	6.98	172.00	154.00	191.00	-0.06	1.00	378	1.60	0.06	1.60	1.41	1.79	-0.11	1.00	
	WC (cm)	353	NA	NA	NA	NA	NA	NA	NA	378	NA	NA	NA	NA	NA	NA	NA	NA
	Hip (cm)	353	NA	NA	NA	NA	NA	NA	NA	378	NA	NA	NA	NA	NA	NA	NA	NA
	WHR (cm/cm)	353	NA	NA	NA	NA	NA	NA	NA	378	NA	NA	NA	NA	NA	NA	NA	NA
EGCUT-CAD	Age (yrs)	355	63.50	10.20	64.00	36.00	94.00	-0.24	-0.27	613	64.50	10.50	64.00	32.00	93.00	-0.29	-0.25	
	BMI (kg/m ²)	354	32.60	4.70	31.90	20.50	51.40	1.00	-0.04	613	33.70	5.70	32.90	21.20	56.90	1.00	-0.01	
	Weight (kg)	354	100.80	16.10	99.00	62.00	160.00	0.88	0.43	613	88.30	16.30	86.00	47.00	160.00	0.90	0.41	
	Height (m)	355	1.76	0.07	1.75	1.58	2.00	-0.04	1.00	613	1.62	0.06	1.62	1.43	1.84	-0.01	1.00	
	WC (cm)	272	111.70	11.20	110.50	82.00	145.00	0.79	0.06	417	104.60	14.20	103.00	62.00	150.00	0.78	0.25	
	Hip (cm)	272	110.10	11.20	110.00	70.00	155.00	0.64	0.13	417	116.00	14.00	115.00	63.00	165.00	0.76	0.27	
	WHR (cm/cm)	272	1.00	0.10	1.00	0.70	1.60	0.16	-0.08	417	0.90	0.10	0.90	0.70	1.10	0.19	0.02	
EGCUT-T2D	Age (yrs)	355	63.50	10.20	64.00	36.00	94.00	-0.24	-0.27	613	64.50	10.50	64.00	32.00	93.00	-0.29	-0.25	
	BMI (kg/m ²)	354	32.60	4.70	31.90	20.50	51.40	1.00	-0.04	613	33.70	5.70	32.90	21.20	56.90	1.00	-0.01	
	Weight (kg)	354	100.84	16.09	99.00	62.00	160.00	0.88	0.43	613	88.28	16.33	86.00	47.00	160.00	0.90	0.41	
	Height (m)	355	1.80	0.10	1.80	1.60	2.00	-0.04	1.00	613	1.60	0.10	1.60	1.40	1.80	-0.01	1.00	
	WC (cm)	272	111.70	11.20	110.50	82.00	145.00	0.79	0.06	417	104.60	14.20	103.00	62.00	150.00	0.78	0.25	
	Hip (cm)	272	110.10	11.20	110.00	70.00	155.00	0.64	0.13	417	116.00	14.00	115.00	63.00	165.00	0.76	0.27	
	WHR (cm/cm)	272	1.02	0.09	1.01	0.70	1.57	0.16	-0.08	417	0.90	0.07	0.90	0.71	1.12	0.19	0.02	
EGCUT-CONTROLS	Age (yrs)	341	54.40	11.00	55.00	35.00	86.00	0.23	-0.33	601	50.00	10.10	49.00	34.00	93.00	0.21	-0.30	
	BMI (kg/m ²)	340	22.80	2.50	22.70	16.70	29.40	1.00	-0.21	600	22.10	2.50	22.10	14.60	35.50	1.00	-0.12	
	Weight (kg)	340	70.86	8.50	70.00	48.00	95.00	0.76	0.46	600	59.46	7.64	60.00	41.00	99.00	0.81	0.48	
	Height (m)	340	1.80	0.10	1.80	1.50	2.00	-0.21	1.00	600	1.60	0.10	1.60	1.40	1.80	-0.12	1.00	
	WC (cm)	340	84.10	6.80	85.00	54.00	110.00	0.45	0.21	600	72.80	5.80	73.00	52.00	105.00	0.66	0.11	
	Hip (cm)	340	94.90	7.70	96.00	57.00	115.00	0.37	0.19	600	95.10	7.20	95.00	64.00	122.00	0.55	0.26	
	WHR (cm/cm)	340	0.89	0.08	0.88	0.67	1.22	0.08	0.00	600	0.77	0.06	0.76	0.58	1.15	0.15	-0.14	
Ely Study	Age (yrs)	744	61.49	9.13	61.51	35.67	77.44	-0.04	-0.26	855	60.82	9.26	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	855	71.11	14.34	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	852	87.56	12.60	85.88	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	851	107.13	11.78	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	851	0.82	0.06	0.81	0.58	1.04	0.34	-0.08	
EMIL	Age (yrs)	807	45.70	11.40	45.00	25.00	75.00	0.20	-0.29	915	46.10	11.20	45.00	25.00	73.00	0.34	-0.29	
	BMI (kg/m ²)	807	27.70	4.70	26.90	17.20	57.50	1.00	-0.09	908	26.70	6.30	25.00	14.80	60.20	1.00	-0.11	
	Weight (kg)	807	86.20	15.60	84.00	49.00	180.00	0.90	0.35	908	71.70	17.30	67.50	37.20	174.00	0.94	0.23	
	Height (m)	807	1.76	0.67	1.76	1.49	1.96	-0.09	1.00	915	1.64	0.66	1.64	1.44	1.91	-0.11	1.00	
	WC (cm)	767	95.30	12.20	94.00	60.00	153.00	0.88	0.03	855	82.80	13.10	80.00	53.00	137.00	0.89	-0.03	
	Hip (cm)	767	104.60	8.00	104.00	72.00	158.00	0.82	0.23	855	103.20	11.10	101.00	56.00	157.00	0.90	0.07	
	WHR (cm/cm)	767	0.91	0.07	0.91	0.64	1.150	0.63	-0.18	855	0.80	0.07	0.80	0.56	1.27	0.49	-0.14	
EPIC-cohort	Age (yrs)	410	59.82	9.15	60.30	40.00	79.10	0.07	-0.22	551	58.83	9.60	58.40	39.90	77.40	0.15	-0.25	

	Hip (cm)	373	120.39	24.78	116.00	71.00	204.00	0.92	-0.01	526.00	125.69	26.09	126.00	63.00	192.00	0.91	0.08
	WHR (cm/cm)	376	1.00	0.11	1.00	0.76	1.30	0.25	0.07	544.00	0.91	0.12	0.90	0.60	1.40	0.38	-0.07
LURIC Cases	Age (yrs)	1678	63.06	9.82	63.78	26.87	88.46	-0.08	-0.22	573	66.24	9.62	67.19	34.00	92.00	-0.04	-0.13
	BMI (kg/m ²)	1678	27.50	3.78	27.05	17.51	57.10	1.00	-0.06	573	27.21	4.62	27.02	16.35	46.09	1.00	-0.05
	Weight (kg)	1678	82.67	12.68	81.80	51.00	185.00	0.87	0.44	573	70.48	12.85	70.00	43.00	122.30	0.91	0.37
	Height (cm)	1678	173.32	6.59	173.00	152.00	202.00	-0.06	1.00	573	160.87	6.10	160.00	146.00	178.00	-0.05	1.00
	WC (cm)	1652	101.00	10.70	100.00	61.00	165.00	0.76	0.12	566	94.86	13.32	94.00	63.00	135.00	0.80	0.12
	Hip (cm)	1652	102.53	8.89	102.00	66.00	165.00	0.68	0.18	566	103.78	11.48	103.00	70.00	150.00	0.79	0.18
	WHR (cm/cm)	1652	0.99	0.06	0.98	0.63	1.61	0.33	-0.05	566	0.91	0.08	0.91	0.68	1.22	0.29	-0.04
LURIC Controls	Age (yrs)	330	54.89	12.45	55.62	17.25	87.85	1.00	-0.31	315	62.14	11.02	62.69	26.97	90.71	0.03	-0.22
	BMI (kg/m ²)	330	27.24	3.72	26.65	16.34	40.33	1.00	-0.14	315	27.25	4.86	26.51	16.77	44.82	1.00	-0.10
	Weight (kg)	330	84.94	12.63	83.00	54.80	130.00	0.84	0.41	315	71.92	13.39	70.00	41.00	125.00	0.91	0.32
	Height (cm)	330	176.59	7.09	176.00	160.00	198.00	-0.14	1.00	315	162.40	6.53	163.00	141.00	189.00	-0.10	1.00
	WC (cm)	327	99.37	10.69	98.00	68.00	139.00	0.78	0.02	310	92.86	12.40	93.00	58.00	134.00	0.78	0.11
	Hip (cm)	327	102.14	8.31	101.00	75.00	138.00	0.69	0.11	310	103.98	11.69	103.00	71.00	147.00	0.79	0.13
	WHR (cm/cm)	327	0.97	0.07	0.97	0.70	1.20	0.40	-0.10	310	0.89	0.07	0.89	0.65	1.18	0.19	0.01
METSIM cases	Age (yrs)	1109	60.26	6.56	60.00	45.00	74.00	-0.16	-0.21	0	NA	NA	NA	NA	NA	NA	NA
	BMI (kg/m ²)	1109	30.22	5.22	29.41	18.29	55.54	1.00	0.00	0	NA	NA	NA	NA	NA	NA	NA
	Weight (kg)	1109	92.46	17.06	90.00	51.00	174.00	0.91	0.37	0	NA	NA	NA	NA	NA	NA	NA
	Height (m)	1109	1.75	0.06	1.75	1.57	1.93	0.00	1.00	0	NA	NA	NA	NA	NA	NA	NA
	WC (cm)	1108	107.19	13.32	105.00	71.50	167.00	0.91	0.15	0	NA	NA	NA	NA	NA	NA	NA
	Hip (cm)	1108	105.05	8.91	104.00	86.00	147.00	0.83	0.24	0	NA	NA	NA	NA	NA	NA	NA
	WHR (cm/cm)	1108	1.02	0.07	1.01	0.80	1.44	0.63	0.01	0	NA	NA	NA	NA	NA	NA	NA
METSIM controls	Age (yrs)	905	53.94	5.00	54.00	45.00	72.00	-0.01	-0.20	0	NA	NA	NA	NA	NA	NA	NA
	BMI (kg/m ²)	905	26.40	3.37	26.04	18.51	48.06	1.00	-0.06	0	NA	NA	NA	NA	NA	NA	NA
	Weight (kg)	905	82.55	11.90	81.50	45.00	154.00	0.84	0.45	0	NA	NA	NA	NA	NA	NA	NA
	Height (m)	905	1.77	0.06	1.77	1.55	1.96	-0.06	1.00	0	NA	NA	NA	NA	NA	NA	NA
	WC (cm)	905	95.85	9.63	95.00	74.00	152.00	0.86	0.17	0	NA	NA	NA	NA	NA	NA	NA
	Hip (cm)	905	100.31	6.26	100.00	72.00	160.00	0.72	0.34	0	NA	NA	NA	NA	NA	NA	NA
	WHR (cm/cm)	905	0.95	0.06	0.95	0.78	1.19	0.65	-0.05	0	NA	NA	NA	NA	NA	NA	NA
MoRGAM Cases	Age (yrs)	1772	58.76	7.78	58.58	25.49	75.84	-0.01	-0.06	280	57.85	8.53	59.49	30.60	73.89	0.14	-0.29
	BMI (kg/m ²)	1707	27.50	3.88	27.15	17.04	47.96	1.00	-0.04	246	29.09	5.71	28.46	17.87	50.66	1.00	-0.26
	Weight (kg)	1708	81.71	12.79	80.60	50.20	129.00	0.88	0.43	246	72.82	14.05	71.35	46.90	132.00	0.92	0.14
	Height (m)	1707	172.30	6.29	172.00	150.00	197.00	-0.04	1.00	246	158.40	6.29	158.00	138.00	180.00	-0.26	1.00
	WC (cm)	983	97.48	10.72	96.50	72.00	138.50	0.86	0.12	222	88.92	13.42	87.50	61.50	131.00	0.87	-0.10
	Hip (cm)	984	101.00	7.78	100.00	75.00	137.00	0.76	0.20	222	105.80	10.28	104.80	82.50	147.50	0.90	-0.06
	WHR (cm/cm)	983	0.96	0.06	0.96	0.73	1.20	0.53	-0.03	222	0.84	0.08	0.83	0.61	1.38	0.43	-0.10
MoRGAM Controls	Age (yrs)	2138	59.42	7.98	59.77	25.14	76.37	0.05	-0.14	335	57.33	9.00	58.78	27.28	73.66	0.22	-0.25
	BMI (kg/m ²)	2095	26.88	3.98	26.50	16.67	51.90	1.00	-0.02	319	27.61	4.97	26.81	15.43	51.06	1.00	-0.08
	Weight (kg)	2095	80.58	13.26	79.60	42.80	154.00	0.89	0.43	320	69.86	13.49	68.20	40.80	134.00	0.90	0.36
	Height (m)	2094	173.00	6.52	173.00	151.00	194.00	-0.02	1.00	319	159.00	6.72	159.00	139.00	178.00	-0.08	1.00
	WC (cm)	1084	95.14	10.94	94.50	64.00	144.00	0.88	0.12	287	85.26	12.32	83.50	60.50	121.00	0.87	0.04
	Hip (cm)	1084	100.60	7.65	100.00	74.50	144.00	0.78	0.24	287	103.60	9.43	102.00	82.50	144.00	0.87	0.16
	WHR (cm/cm)	1084	0.95	0.07	0.95	0.67	1.16	0.59	-0.07	287	0.82	0.07	0.82	0.68	1.08	0.56	-0.11

	Weight (kg)	1112	80.40	11.42	79.50	46.00	138.70	0.87	0.42	0	NA	NA	NA	NA	NA	NA	NA
	Height (m)	1112	1.75	0.06	1.75	1.56	2.00	-0.07	1.00	0	NA	NA	NA	NA	NA	NA	NA
	WC (cm)	1094	94.75	9.54	94.00	51.00	137.00	0.87	0.13	0	NA	NA	NA	NA	NA	NA	NA
	Hip (cm)	1094	100.16	7.07	100.00	51.00	141.00	0.78	0.28	0	NA	NA	NA	NA	NA	NA	NA
	WHR (cm/cm)	1094	0.95	0.05	0.95	0.78	1.14	0.58	-0.11	0	NA	NA	NA	NA	NA	NA	NA
WHITEHALL	Age (yrs)	1699	60.51	5.82	59.38	50.69	72.93	-0.06	-0.13	535	60.60	5.79	59.81	50.47	72.85	0.06	-0.16
	BMI (kg/m ²)	1699	26.65	3.63	26.30	18.10	44.80	1.00	-0.07	535	27.14	5.31	26.20	16.30	47.70	1.00	-0.10
	Weight (kg)	1699	81.85	12.38	80.40	43.00	133.00	0.85	0.42	535	70.82	14.44	68.50	41.30	130.10	0.89	0.32
	Height (m)	1699	1.75	0.07	1.75	1.44	1.98	-0.07	1.00	535	1.62	0.06	1.61	1.39	1.80	-0.10	1.00
	WC (cm)	1699	95.12	10.25	94.45	66.55	138.00	0.89	0.12	535	86.00	12.92	85.40	55.55	126.30	0.89	0.04
	Hip (cm)	1699	99.85	6.53	99.30	76.00	131.00	0.79	0.34	534	101.95	10.19	100.70	77.20	139.20	0.88	0.18
	WHR (cm/cm)	1699	0.95	0.06	0.95	0.77	1.20	0.66	-0.13	534	0.84	0.07	0.84	0.68	1.11	0.57	-0.13
WTCCC-T2D	Age (yrs)	626	56.30	10.20	57.00	20.80	87.00	-0.19	-0.11	448	56.40	11.10	57.90	19.40	80.10	-0.15	-0.11
	BMI (kg/m ²)	624	31.20	5.60	30.40	16.80	60.30	1.00	0.04	445	33.80	7.60	33.00	18.70	59.60	1.00	-0.05
	Weight (kg)	624	96.60	19.20	95.00	57.00	190.50	0.92	0.42	445	88.20	20.90	84.70	43.00	165.50	0.94	0.30
	Height (m)	625	1.80	0.10	1.80	1.60	2.20	0.04	1.00	445	1.60	0.10	1.60	1.40	1.80	-0.05	1.00
	WC (cm)	606	108.72	14.88	107.00	75.00	200.66	0.87	0.19	431	104.28	15.39	104.00	60.00	149.00	0.86	0.09
	Hip (cm)	606	110.22	11.95	109.00	86.40	215.90	0.76	0.22	431	116.24	15.54	114.00	79.50	172.00	0.89	0.09
	WHR (cm/cm)	604	1.00	0.10	1.00	0.80	1.30	0.48	0.05	431	0.90	0.10	0.90	0.70	1.20	0.14	0.01

Supplementary Note

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3. Acknowledgments

4. Affiliations of investigators from acknowledged consortia

1. Genome-wide association meta-analysis

We combined the height summary statistics from 79 genome-wide association (GWA) studies in a meta-analysis of 253,288 individuals.

1.1.1 Description of individual cohorts, genotyping methods, and quality control

Descriptive characteristics, study design, sample size, sample quality control (QC) and anthropometric measurement technique for studies included in the primary GWA meta-analysis but not previously reported in Lango Allen *et al.* (2010)¹ are provided in **Supplementary Tables 17-19**. All participants provided written informed consent and the studies were approved by the respective Local Research Ethics committees or Institutional Review Boards.

Details on the genotyping platform used and genotype quality control procedures employed for each study are presented in **Supplementary Table 18**, while the basic anthropometric measures are summarised in **Supplementary Table 19**.

1.1.2 Imputation

All cohorts were genotyped using commercially available Affymetrix (Affymetrix, Inc., Santa Clara, CA, USA), Illumina (Illumina, Inc. San Diego, CA, USA) genotyping arrays, or custom Perlegen (Perlegen Sciences, Inc. Mountain View, CA, USA) arrays. To facilitate meta-analysis, each group performed genotype imputation using BIMBAM², MACH^{3,4}, IMPUTE⁵ BEAGLE⁶ or PLINK⁷ and genotypes from the Phase II CEU HapMap.

1.1.3 GWA analyses in individual studies

Each GWA study tested association between each imputed or genotyped SNP and sex standardized height, assuming an additive inheritance model and adjusting for age and other appropriate covariates specific to the study (e.g. genotype-based principal components). Studies with unrelated individuals tested association under a linear regression framework. Studies with related samples used variance component or other linear mixed effects modelling to account for relatedness in the regression. The uncertainty of the imputed genotypes was taken into account in the association analysis using methods appropriate for the imputation software used.

The genomic control (GC) inflation factor was calculated for each of the GWA scans separately. The average GC inflation factor was 1.03. Genomic control correction was applied to results for each study prior to meta-analysis by multiplying SNP standard errors by the square root of the inflation factor.

1.1.4 Quality control checks of individual studies

Each file was processed through a cleaning script⁸ that performed several quality checks, including calculating the number of markers, ranges of test statistics, the genomic correction inflation factor, and NxMAF. From each study we excluded monomorphic SNPs and SNPs with poor imputation quality: $rsq_hat < 0.3$ (BEAGLE⁶, BIMBAM² and MACH^{3,4}), $proper\ info < 0.4$ (IMPUTE⁵), or $info < 0.8$ (PLINK⁷). Additional checks to identify outlying study files were also made in attempt to ensure that correct transformations to the phenotype had been applied, correct assignment of the effect allele and effect allele frequency had been made, and that genomic control inflation factors and the proportion of previously published height SNPs¹ with directional consistency were consistent relative to sample size amongst other study files.

1.1.5 Meta-analysis of GWA studies

A total of 2,550,858 autosomal SNPs were meta-analyzed across 174 input files (many of the 79 cohorts had separate male-female and/or case-control files). We did not apply a minor allele frequency cut-off, but we did apply an arbitrary cut-off of $NxMAF > 3$ (equivalent

to a minor allele count of 6) to guard against extremely rare variants present in only one or two samples (possible genotyping/imputation errors or private mutations), for which regression coefficients are not estimated well using the standard statistical methods employed in most GWA statistical programs. We used the inverse-variance fixed effects meta-analysis method to combine the results from the individual studies. For comparison purposes, we also performed a sample size weighted Z -score based fixed effects meta-analysis. The correlation coefficient between the \log_{10} of the P -values of the inverse variance and sample size weighted meta-analysis was >0.99 . Meta-analyses were performed using the software program METAL⁹ (www.sph.umich.edu/csg/abecasis/metal).

1.2 GCTA-COJO: conditional and joint multiple SNPs analysis

We used a conditional and joint (COJO) multiple SNPs analysis implemented in GCTA^{10,11} to select the top associated SNPs. This method uses the summary statistics from the meta-analysis and LD correlations between SNPs estimated from a reference sample to perform a conditional association analysis¹⁰. The method starts with an initial model of the SNP that shows the strongest evidence of association across the whole genome. It then implements the association analysis conditioning on the selected SNP(s) to select the top SNPs one-by-one iteratively via a stepwise model selection procedure until no SNP has a conditional P -value that passes the significance level. Finally, all the selected SNPs are fitted jointly in the model for effect size estimation. The advantage of this method is that i) it uses summary statistics of the meta-analysis without having to access the individual-level genotype data which is usually unavailable in a large-scale meta-analysis; ii) it performs the conditional analysis iteratively so that there is no need to use arbitrary distance and LD thresholds to define the top associated SNP at a locus; iii) it is able to detect multiple associated SNPs at a single genomic locus.

We used the ARIC cohort as the reference sample for LD estimation¹. We used 593,521 genotyped SNPs to create the genetic relatedness between 8,682 individuals of European ancestry. We removed one of each pair of individuals with an estimate of genetic relatedness > 0.025 and retained 6,654 unrelated individuals. There were $\sim 3.0\text{M}$ SNPs included in the original meta-analysis. We included in this analysis only the SNPs ($\sim 2.48\text{M}$) on HapMap2 and with sample size $> 50,000$. We used the conventional genome-wide significance level ($P < 5 \times 10^{-8}$) for SNP discovery and chose a range of less stringent significance levels (5×10^{-3} , 5×10^{-4} , ..., 5×10^{-7}) to select SNPs for prediction analysis. We ignored the LD correlations between SNPs in $> 20\text{Mb}$ distance or on different chromosomes. In addition, to avoid sample overlap in the subsequent prediction and estimation analyses, the target cohort in which the prediction or GCTA^{10,11} estimation analysis (see below) was performed was excluded from the original meta-analysis. Using this method we identified 697 signals (**Supplementary Table 1**).

1.2.1 Cluster analysis of signals within genomic loci

To test whether height-associated SNPs were non-randomly distributed over the genome, we constructed 10,000 random sets of 697 SNPs each that were matched based on allele frequency (± 5 percentage points), nearby number of genes (using LD cut-off $r^2 > 0.5$; $\pm 10\%$ of seed SNP gene neighbor count), and gene proximity ($\pm 20\text{kb}$). We used the number of secondary signals (distance less than 1Mb between neighboring SNPs) to test for non-random clustering. Among the 10,000 matched random sets, we observed an average of 200 (range 162 to 243) SNP-pairs by the matched SNPs compared to 273 with the set of 697 height SNPs. The observed distribution resulted in an empirical enrichment P -value < 0.0001 .

We repeated the clustering analysis by estimating the total number of distance-wise genomic loci (distance between physically neighbouring SNPs more than the given distance

threshold, like for example 1Mb) by using same set of 10,000 sets of random matched SNP-sets across a range of locus size definitions, from 100kb to 1.25Mb. We also repeated the analyses using 50kb and 100kb sections of DNA at various distances from index variants – e.g. 50kb-100kb, 100kb-200kb, 200kb-300kb. We observed evidence of clustering regardless of locus size, but showed that the majority of clustering was driven by variants within windows <250kb. The results for the whole range of locus sizes are shown in **Supplementary Table 2**.

1.3 Replication using independent studies typed with MetaboChip

We combined the height summary statistics from 37 MetaboChip-based association studies in a meta-analysis of 80,067 individuals.

1.3.1 Description of individual studies and genotyping methods

Descriptive characteristics, study design, sample size, sample quality control (QC) and anthropometric measurement technique for the studies included in the MetaboChip meta-analysis are provided in **Supplementary Tables 20-22**. All individuals were Caucasians of European ancestry. All participants provided written informed consent and the studies were approved by the respective Local Research Ethics committees or Institutional Review Boards.

Details on the genotype quality control procedures employed for each study are presented in **Supplementary Table 21**, while the basic anthropometric measures are summarised in **Supplementary Table 22**.

Studies were genotyped using Illumina's commercially available custom MetaboChip genotyping array¹². Briefly, the array holds ~200,000 SNPs of interest for metabolic and atherosclerotic / cardiovascular disease traits. In addition, SNPs known to have reached genome-wide significance for any trait at the time of design are also present on the array.

1.3.2 MetaboChip analysis in Individual studies

Each GWA study tested association between each genotyped SNP and the same height transformations, adjustment, and inheritance model as described above. All studies comprised of unrelated individuals. Studies that also contributed to the primary GWA analysis removed any subjects that contributed to the primary GWA prior to analysis. We tested for association between SNPs and height using a linear regression framework.

The genomic control (GC) inflation factor was calculated for each of the MetaboChip scans separately. The average GC inflation factor was 1.03. Genomic control correction was applied to results for each study prior to meta-analysis. However, as MetaboChip contains SNPs associated with height and other anthropometric traits, we used a set 4,427 SNPs associated with QT interval present on MetaboChip to calculate, and control for, study inflation factors. This was to avoid over correction by incorporating SNPs already known to influence height.

1.3.3 Quality control checks of individual studies

Each file was processed through a cleaning script that performed several quality checks, including calculating the number of markers, ranges of test statistics, the genomic correction inflation factor, and $N_x\text{MAF}$. From each study we excluded monomorphic SNPs and SNPs with a call rate <95% and/or Hardy-Weinberg $P < 1 \times 10^{-6}$. Additional checks to identify outlying study files were also made as described in section 1.1.4.

1.3.4 Meta-analysis of MetaboChip

A total of 110,743 autosomal SNPs were meta-analyzed across 82 input files. We did not apply a minor allele frequency cut-off, but did apply the same $N \times \text{MAF} > 3$ cut-off for SNP inclusion as used for the genome-wide analyses. We used exactly the same meta-analysis procedure as described for the GWAS. The correlation coefficient between the \log_{10} of the P -values of the inverse variance and sample size weighted meta-analysis was >0.99 .

1.3.5 Metachip replication results

We aimed to replicate 403/697 lead SNPs identified through conditional analysis, significant prior to conditional analysis (i.e. genome-wide significance not driven through haplotype effects), and were either found on Metachip or had a proxy with $r^2 \geq 0.8$ located $\pm 500\text{kb}$ from the lead SNP. The “best” proxy was selected based on having the highest r^2 and closest to the lead SNP, breaking any r^2 ties (**Supplementary Table 3**). Proxies were obtained using the SNP Annotation and Proxy Search (SNAP) program¹³ using data from HapMap release 22, HapMap 3, and 1000 Genomes Pilot 1.

We compared the number of signals that were significant based on the Metachip meta-analysis and identified 370/403 that reached $P < 0.05$ (92%) and 126/403 (31%) that reached $P < 5 \times 10^{-8}$ (**Supplementary Table 3**). We also compared effect sizes to analyse directional consistency between the two meta-analyses. As proxies were used and effect alleles not directly comparable, we limited the number of comparisons to 339/403 whereby either the same SNP was found on Metachip or the minor allele frequency (MAF) of both the original signal SNP and the proxy SNP was < 0.4 . Proxy SNPs were compared by comparing the effect sizes of the minor allele in both datasets. In total, 309 and 105 reached $P < 0.05$ and $P < 5 \times 10^{-8}$, respectively, of which all had directional consistency with the primary GWA meta-analysis (**Supplementary Table 3**).

Of the 403 SNPs put forward for replication, 293 represented signals that were not previously identified in a previous meta-analysis of height¹. Previously identified SNPs were classified as having LD $r^2 \geq 0.1$ within 500Kb of the lead SNP. Of the novel SNPs, 262/293 reached $P < 0.05$ (89%) and 64/293 reached $P < 5 \times 10^{-8}$ (22%).

1.4 Validation of inflated statistics using linear mixed models in 59,380 individuals

1.4.1 Accounting for structure within individual studies through linear mixed models

We used 15 studies ($N=59,380$) of which 12 contributed to the main GWAS meta-analysis ($N=35,962$). Each study performed additional association analyses by utilising linear mixed models (LMM) as implemented in EMMAX¹⁴. Briefly, for each LMM analysis, a genomic relationship matrix (GRM) was generated using directly genotyped SNPs available in each study to estimate the genetic similarity between all pairs of individuals. Males and females (and cases and controls where relevant) from the same study were combined to maximise the detection of genetic structure within each cohort. The matrix was then incorporated into the LMM for association testing of the imputed genotypes. The summary results from each study were then combined into an inverse-variance fixed-effects meta-analysis and inflation factors calculated based on SNPs with $N > 25,000$ that passed imputation QC criteria (above). Three LMM analyses were performed: an “all” meta-analysis (GRM-ALL), a “leave-one-out” (GRM-LOO) meta-analysis, and an “odd/even” meta-analysis (GRM-O/E).

1.4.2 GRM-ALL analysis

Each study performed an association analysis whereby the genomic relationship matrix generated utilised all SNPs across the genome, effectively correcting for any genetic similarity amongst individuals genome-wide. As a quality check we compared the directional consistency of the 180 loci previously reported¹ for each study relative to sample size. The mean λ_{GC} value was 1.00 (**Supplementary Table 4**). After performing a meta-analysis, the overall λ_{GC} value was 1.20.

1.4.3 GRM-LOO (Leave One Out) analysis

Each study (with the exception of the Women's Genome Health Study (see below)) performed a leave-one-out analysis whereby each chromosome was tested while adjusting for a genomic relationship matrix generated from directly genotyped SNPs from the other 21 chromosomes. As for the GWA and Metabochip analyses, we performed a quality check by comparing the directional consistency of the 180 loci previously reported for each study relative to sample size. The mean λ_{GC} value was 1.03 (**Supplementary Table 4**). The overall λ_{GC} value from meta-analysis was 1.21.

1.4.4 GRM-O/E (Odd/Even) analysis (Women's Genome Health Study only)

The sample size of 21,354 in the Women's Genome Health Study (WGHS) precluded the creation of 22 individual GRMs due to computation constraints, which consequently excluded WGHS from the GRM-LOO analysis. Therefore, two GRMs were created using GCTA¹¹ that utilised variants on the odd and even chromosomes. The two genomic relationship matrices were then reformatted for EMMAX with R. SNPs on odd chromosomes were tested while correcting for the GRM calculated from the even chromosomes, and SNPs on even chromosomes were tested while correcting for GRM calculated from the odd chromosomes.

The λ_{GC} value obtained from GRM-O/E was 1.07. The λ_{GC} value based on the GWAS summary data was 1.13. These lambdas were based on SNPs with N=21,354 (all SNPs in the data available).

1.4.5 Comparison of λ_{GC} inflation factors in GWA-based meta-analysis

A fixed-effects inverse-variance meta-analysis of the "standard" GWAS summary data from the 15 studies (including three studies that were not part of the primary GWA meta-analysis) was performed to calculate λ_{GC} values corresponding to no genomic correction (1.24) and a single genomic correction (1.20). These λ_{GC} values were then used to determine whether the λ_{GC} from the linear mixed models were more consistent with adjustments through single- or double-genomic control.

Finally, we combined all 15 EMMAX-based studies together (N=59,380) and meta-analyzed GRM-ALL results and GRM-LOO + GRM-O/E to compare the λ_{GC} values obtained (1.20 and 1.29, respectively) to those obtained when meta-analyzing the GWA study data (single λ_{GC} = 1.23) (**Supplementary Table 4**)

1.4.6 Assessing the potential effect of duplicate or cryptically related samples

To access the potential effect of any duplicate or cryptically related samples present between the studies, we calculated λ_{GC} in the overall meta-analysis after duplicating a study file and eliminating another study file with similar sample size (thus creating an entire set of duplicate samples larger than is likely to be present in the actual data). Meta-analyses with duplicate sample sets ranging from 450 to 3,800 individuals resulted in λ_{GC} ranging from 1.98 to 2.02, suggesting that duplicate samples or cryptically related samples, if present, would have a minimal impact on the overall inflation factor.

1.5 Assessing stratification and winner's curse using family-based and non-family based studies

1.5.1 Family-based analyses for validation of inflated statistics using family transmission tests in a subset of 25,849 individuals

Within family analysis was performed in seven studies (Amish, Framingham, TWINGENE, QIMR, SardinIA, Family Health Study and ERF) to assess confounding effects of stratification. We selected 416 genome-wide significant SNPs representing multiple loci ($r^2 < 0.1$ and $> 1\text{Mb}$ distance) by performing a single GC corrected meta-analysis that excluded these studies. Pure transmission-based tests for each of these SNPs were performed using the QFAM option in PLINK⁷ with 100K permutations, except in ERF and SARDiNIA studies where QTDT¹⁵ was used. QC filtering of SNPs for low imputation quality scores was followed by inverse variance method of meta-analysis of the summary statistics. Of 416 genome-wide significant SNPs, 371 (89%) had a consistent direction of effect after the meta-analysis of family based studies.

1.5.2 Estimation of winner's curse and stratification

We partitioned the effects of overestimates of effect sizes by looking at the deficit of replication compared with what would be expected in independent samples, both in family-based samples (where a deficit of replicating signals reflects overestimation of effect size due to both winner's curse and stratification) and in a random set of non-family-based samples (where a deficit of replicating signals reflects overestimation of effect size due to winner's curse alone).

To calculate the effective sample size of a study, we first estimated the linear relationship of the inverse of the median standard error to square root of sample size in the GIANT data (excluding the family-based studies, whose effective sample size is usually much lower than the actual sample size). We then used this linear relationship to determine the equivalent effective sample size for each study by measuring the median standard error for that study⁸. We generated new meta-analyses, after excluding a set of studies, either the family-based studies or in turn each of three sets of randomly selected, non-family-based studies. Each set of non-family-based studies was chosen to have a combined effective sample size similar to that of the family-based studies (**Supplementary Table 5**). For each new meta-analysis, we only applied the study specific single GC correction and estimated effect sizes and association results for each SNP. Apparently independent genome-wide significant SNPs were selected in the same way as for the overall meta-analysis ($r^2 < 0.1$ and $> 1\text{Mb}$ distance).

For these genome-wide significant SNPs, we then calculated the expected number of directionally consistent replications at various P -value thresholds in the excluded studies, by using the SNP effect sizes from the meta-analyses described above, the allele frequencies, and the combined effective sample size of the excluded studies. We then calculated the deficit in directionally consistent replicating signals at each P -value threshold as (number of SNPs expected to achieve the P -value threshold) minus (number of replications actually observed at that threshold).

The deficit in replication could arise from true positive signals whose effect sizes have been inflated by stratification or by the winner's curse (inflation of the effect size estimate in the meta-analysis used to identify the genome-wide significant SNPs). The deficit could also be due to completely false positive signals from stratification. To account for the winner's curse, we calculated the average deficit at each P -value threshold across the three sets of random non-family-based cohorts. We subtracted this average deficit from the deficit observed for the family-based cohorts to obtain an estimate of the remaining deficit attributable to stratification, arising from inflation of effect sizes of true associations and/or completely false positive associations.

To estimate the upper bound for completely false positive signals, we assumed that all of the deficit in replication was due to these false positives (If some of the deficit is attributable to inflation of effect sizes for true associations, it follows that fewer false positive signals would be needed to account for the deficit in replication.). We note that the observed deficit actually

underestimates the number of false positive associations slightly due to the expected rate of chance replication. Specifically, if there are k false positive associations, then we will observe $k/2\alpha$ replications by chance, where α is the P -value threshold. As such, the actual deficit will be $k(1-0.5\alpha)$ instead of k . To account for these chance replications we therefore multiplied the deficit by the inverse of $(1-0.5\alpha)$, which is $2/(2-\alpha)$. For directional consistency, this correction factor is 2; for $p < 0.05$, the correction factor is ~ 1.03 . We also adjusted the deficit to account for slight differences in the number of genome-wide significant SNPs in the meta-analyses excluding different sets of cohorts. (**Supplementary Table 5**).

We also examined the directional consistency of the SNPs available in the 7 family-based studies and observed an overall concordance of 89% after meta-analyzing the seven studies (**Supplementary Table 6**).

2. Biological analyses

2.1 Enrichment of associated SNPs in ENCODE regions, eQTLs and non-synonymous variants

2.1.1 Non-synonymous variants in strong LD with height SNPs

For all 697 SNPs associated with height (method described in Section 1.2), we calculated the pair-wise linkage disequilibrium with the non-synonymous variants detected in the 1000 Genomes Phase 1 release. 100 SNPs out of 697 were in strong LD ($r^2 \geq 0.8$) with at least one missense, nonsense, inframe deletion or inframe insertion. In total we identified 231 1000 Genomes Project data proxies, including 225 missense (including 3 initiator codon variants), 3 nonsense, 2 inframe-deletion and 1 inframe-insertion (**Supplementary Table 7**). We used the SNPinfo tool¹⁶ (<http://snpinfo.nih.gov/snpinfo>) to annotate the predicted functional consequences of the identified non-synonymous proxies.

To test whether height-associated SNPs were enriched for non-synonymous variants in strong LD, we first pruned the 697 SNPs based on LD (cut-off $r^2 > 0.1$), which left 628 SNPs. Using these SNPs we then constructed 10,000 random sets of 628 SNPs each that were matched based on allele frequency (± 5 percentage points), nearby number of genes (using LD cut-off $r^2 > 0.5$; $\pm 10\%$ of seed SNP gene neighbor count), and gene proximity (± 20 kb). Among the 10,000 matched random sets, we observed an average of 78 (range 0 to 114) non-synonymous variants tagged by the matched SNPs compared to 95 with the pruned set of 628 height SNPs. The observed distribution resulted in an empirical enrichment P -value < 0.02 .

2.1.2 Association with other traits

The full NHGRI GWAS catalogue (<http://www.genome.gov/26525384>; accessed on 13th January 2013) was downloaded and all SNPs associated with diseases and traits other than anthropometric (height, BMI, Waist-Hip-Ratio and obesity) at genome-wide significance level ($P < 5 \times 10^{-8}$) were extracted. We next identified all SNPs in strong LD ($r^2 \geq 0.8$) with the 697 height-associated SNPs. Thirty-six of the 697 height SNPs tagged 45 NHGRI GWAS catalogue associations, of which 9 were the height SNP itself (**Supplementary Table 10**). Two height SNPs rs7980687 and rs8756 (two proxies at $r^2 > 0.8$) were associated with childhood head circumference and adult brain structure, where the height increasing allele was also the trait increasing allele. Next we applied the same enrichment assessment framework (as described in section 2.1.1 above) to estimate if the height SNPs tend to show increased overlap with trait associated loci. Among the 10,000 random sets, we observed an average of 13 (range 0 to 26) trait associations from the NHGRI GWAS catalogue tagged ($r^2 \geq 0.8$) by the matched SNPs compared to 33 with the pruned set of 628 height SNPs. The observed distribution resulted in an empirical enrichment P -value < 0.0001 .

2.1.3 Enrichment in ENCODE annotated regions

The two main goals of this analysis were (1) to quantify whether the set of height SNPs identified were enriched for functional genomic elements as catalogued by the ENCODE Project¹⁷ or the Roadmap Epigenomics Project (www.roadmapepigenomics.org), and (2) to use these functional annotations to prioritize functional variants. We downloaded all histone ENCODE tracks (with peak coordinates) available in Summer 2012 for the tier 1 (GM12878, K562, H1-hESC) and tier 2 (HUVEC, HepG2) ENCODE cell lines. We added to this list different epigenomic marks generated in osteoblasts and chondrocytes from bone marrow-derived mesenchymal stem cells by the Roadmap Epigenomics Project. Our algorithm to combine genetic and epigenetic data includes four steps. **STEP 1:** We used SNPs from the 1000 Genomes Project CEU dataset to generate 1,000 sets of variants that are matched with the height “seed” variants based on allele frequency, gene proximity and linkage disequilibrium (LD). Note only 672/697 associated SNPs could be found (and therefore matched) in the 1000 Genomes dataset. **STEP 2:** For each variant in the seed and matched sets, we retrieve all other variants in LD using data from the 1000 Genomes Project CEU datasets and the PLINK software ($r^2 \geq 0.8$). **STEP 3:** We annotated all variants and their proxies in each of the 1,001 sets for overlap with ENCODE/Roadmap Epigenomics features of interest. **STEP 4:** We calculated the statistical significance of the enrichment for each track by counting the number of matched sets with equal or more SNP-peak overlaps than in the seed set of variants. Results from the enrichment analysis are presented in (**Supplementary Table 11**). In all cases, there was a strong enrichment ($P < 0.001$) of peaks within these tracks for the height loci when compared to matched set of markers (**Supplementary Table 11**). These results are consistent with the previously reported enrichment of expression quantitative trait loci (eQTL) among the height loci that was observed in lymphoblastoid cell lines⁶ and suggest that many of these loci will impact phenotype through the regulation of gene expression.

2.1.4 eQTL analysis in peripheral blood

We used *cis*-eQTL data from a total of 2,360 unrelated individuals obtained from three datasets with gene expression data measured from whole peripheral blood (1,240 individuals from Fehrmann-HT12v3, 229 individuals from Fehrmann-H8v2¹⁸ and 891 individuals from the EGCUT study¹⁹). The gene expression data were obtained from total RNA in whole blood samples. Genotype data were first filtered for allele frequency ($MAF > 0.01$), HWE ($P < 1 \times 10^{-6}$), and SNP call rate (95%) before imputation using the Phase II CEU HapMap reference panel and imputation dosage values were used as genotypes during the analysis.

To avoid hybridization artifacts, we harmonized the data by aligning the gene expression probes to the human genome build 18 (Ensembl build 54) using BLAT, SOAPAlign v2, and BWA and excluding any probe that mapped to multiple genomic locations or contained more than two mismatches. Each expression dataset was normalized as follows: (1) quintile normalized, (2) \log_2 transformed, and (3) standardized to have a mean of zero and variance equal to one. We used the software MixupMapper²⁰ to identify and remove sample mix-ups. To correct for possible population structure, we residualized the gene expression data on the first four multi-dimensional scaling components obtained from the genotypic data. We residualized the resulting variable on 40 PCs, derived from the variance-covariance matrix of the genotypic data, that did not show any significant evidence of association with the genotypes (and might therefore have a biological interpretation). A more detailed overview of the method has been published elsewhere²¹.

After normalization of the gene expression data, we correlated genotype dosages of the height-associated SNPs with gene expression values using Spearman's rank correlation. To detect *cis*-eQTLs, we assessed only those combinations of SNPs and probes where the

distance between SNP and the midpoint of the probe was smaller than 1 megabase (Mb). Individual datasets were meta-analyzed using a Z-score method, weighted for the sample size of each dataset. We then permuted the sample labels and repeated this analysis 100 times, in order to obtain the *P*-value distribution used to control the FDR at 5%. Since SNPs can be highly correlated due to LD, *cis*-eQTL effects are often caused by SNPs in high LD with the query SNP. In order to determine whether the height-associated SNPs have independent *cis*-eQTL effects with respect to other SNPs in their locus, we performed a conditional analysis. Using the procedure described above, we first determined which SNPs show the strongest *cis*-eQTL (eSNP) effect for each of the probes associated with the 697 height-associated SNPs. Then, we adjusted the gene expression data for these effects using linear regression, and repeated the *cis*-eQTL analysis on the height SNPs (and vice-versa). This analysis allowed us to identify height-associated variants that were also the best *cis*-eQTL SNP.

We observed a significant *cis*-effect for 295 out of 697 height SNPs, of which 21 were identified as the best eQTL SNP for a given probe (**Supplementary Table 8**). A further 54 SNPs showed residual independent effect on expression after conditioning on the best eQTL SNP in each locus that in combination affected the expression levels for 56 genes. To test if the height-associated SNPs are enriched for SNPs associated with *cis* gene expression, we performed an eQTL lookup with the 10,000 random SNP-sets described in section 2.1.1. On average we observed 272 (range 0 to 321) significant (FDR<0.05) *cis*-effects, of which on average 16 (range 0 to 33) were identified as best eQTL SNPs. The respective counts for the pruned set of 628 height SNPs were 281 and 24. The observed distribution gives an empirical enrichment *P*-value of 0.59 for *cis*-eQTL effects and *P*-value of 0.03 for best eQTL SNP.

2.1.5 Biological enrichment analysis: OMIM genes

Using the Online Mendelian Inheritance in Man (OMIM) (<http://www.omim.org>) database we updated a previously used list¹ of genes that are either 1) linked to human monogenic conditions or syndromes characterized by abnormal skeletal growth, 2) known to regulate growth hormone levels or 3) involved in growth plate physiology. This annotation resulted in 266 “OMIM genes” (**Supplementary Table 9**). We then assessed if a listed OMIM gene was the nearest to the identified height-associated SNP. We observed 39 instances (~14.7%) whereby the OMIM gene was the closest coding gene (**Supplementary Table 9**). When performed a similar search in each locus (see 2.2.2 for details) we observed 73 OMIM genes. We assessed the significance of this result by generating 10,000 matching sets for the 628 height SNPs (see Section 2.1.1) and annotated the nearest coding gene. In the 10,000 random sets the median count of OMIM genes was 54 (range 26 to 88), which was lower than the 73 OMIM genes observed among the 628 observed SNPs (empirical enrichment *P*-value 0.013).

2.2. Enrichment of genes in associated loci in reconstituted gene sets

2.2.1 Data-driven Enrichment-Prioritized Integration for Complex Traits (DEPICT) method – gene prioritization (T.H.P *et al.*, unpublished data). Prioritization of genes within a given associated locus was accomplished by (1) correlating a given gene’s predicted functions (henceforth referred to as co-functionality vector, representing the gene’s membership across all reconstituted gene sets) with the co-functionality vector of genes from all other associated loci, (2) summarizing the given gene’s overall pair-wise relatedness as Z-score, (3) adjusting the given gene’s Z-score by the number of genes in the given locus, and (4) re-computing steps #1-#3 1,000 times based on random loci that were matched by gene density and using the 1,000 null scores to adjust the observed Z-score from step 3 for potential gene length bias and potential biases in the gene expression data, and (5) converting the Z-score to a *P*-value. Please refer to the Supplementary Note section 2.2.4

for a description of the empirical false discovery rate estimation, and to Supplementary Note section 2.2.5 for information on the construction of the gene evidence table used to highlight genes in the main text of this paper.

2.2.2 DEPICT method – reconstituted gene set enrichment. For a given reconstituted gene set, DEPICT quantified enrichment by (1) summing the co-functionality Z-scores of all genes within each associated locus and then computing the sum across all loci, (2) repeating step #1 1,000 times based on random loci that were matched by gene density and using the 1,000 null Z-scores to adjust the observed Z-score by subtracting their mean and dividing with their standard deviation (3) converting the adjusted Z-score to a *P*-value. Please refer to the Supplementary Note section 2.2.4 for a description of the empirical false discovery rate estimation to Supplementary Note section 2.2.6 for information on the gene set pruning and visualization. Supplementary Table 12 lists of pruned gene sets with $P < 1 \times 10^{-11}$, for the complete list of significant gene sets please refer to the GIANT website (see URLs).

2.2.3 DEPICT method – enrichment of tissue and cell type annotations. We used DEPICT to assess whether genes within associated loci were specifically expressed in any of the 209 tissue and cell type annotations present in the gene expression data used in DEPICT. The expression data was based on the available Affymetrix HGU133a2.0 platform human sample microarrays. For each microarray the tissue of origin was determined by text-mining the Gene Expression Omnibus database microarray sample description (SOFT files) for tissue-related Medical Subject Heading (MeSH) terms. We limited this analysis to terms starting with A2-9, A10-11, A14-15, and A17. Individual microarray samples were allowed to map to several tissue or cell type annotations, and we restricted the analysis to tissue annotations comprising at least 10 microarrays. When mapping microarrays to a given MeSH term, we included all microarrays annotated to terms that according to the MeSH ontology were children of the given term. Consequently several of the tissue and cell type annotations were not independent from each other. The expression matrix comprised 19,997 genes and 37,427 samples (the number of samples left after QC of the 43,278 Affymetrix HGU133a2.0 platform human microarray samples) spanning 209 MeSH terms. To down-weight genes that were highly expressed across the majority of tissue or cell type annotations (and hence non-informative in the analysis), we $N(0,1)$ standardized each gene's expression values across all tissues. Hereafter we averaged the expression of microarray samples that were annotated with the same MeSH term. The enrichment procedure and calculation of false discovery rates was conceptually similar to the gene set enrichment approach except that the processed expression matrix (genes x tissue annotations) was substituted for the co-functionality matrix (genes x gene sets) for scoring.

We previously conducted a principal component analysis on the probe correlation matrices of 77,840 microarrays samples from four Affymetrix platforms, resulting in the identification of a total of 2,206 robustly estimated principal components (377 for Human Genome U133A, 777 for Human Genome U133 Plus 2.0, 677 for Mouse Genome 430 2.0 and 375 for Rat Genome 230 2.0; See Cvejic *et al.*²² for more information.) Furthermore, we showed that genes in predefined gene sets may enrich for high or low loading genes on these principal components. In order to construct the DEPICT cofunctionality matrix, which defined the reconstituted gene sets (and genes' predicted functional annotations), we first ascertained 14,461 predefined gene sets (see below) for enrichment on each of the 2,206 principal components (using a *t* test). To ensure normality, the *P*-values were converted into 'enrichment' Z-scores, which for each pre-defined gene sets resulted in a Z-score vector across principal components. We subsequently assessed all individual genes, and correlated the 14,461 enrichment Z-score vectors with the 2,206 PC eigenvector coefficients that we had per gene. Significant positive correlation indicated that a gene was likely to operate in that pathway. To avoid circularity in cases where the gene was part of the given predefined gene set, we left out the gene from the gene set, recomputed the gene set's

enrichment Z-score vector along all components and then correlated the gene with the gene set' Z-score vector. This correlation matrix of genes by gene sets – referred to as co-functionality matrix in the main text – comprised 19,997 genes and 14,461 gene sets and quantified each gene's likelihood to be part of a given gene set. A visual description of the concept behind this method can be found at www.genenetwork.nl/genenetwork (click on "Method"), and it was recently used to identify *SMIM1* as the gene that is responsible for the VEL-blood group²². Using this procedure we reconstituted existing various pathways and gene sets: 737 Reactome pathways²³, 5,083 Gene Ontology terms²⁴, 184 KEGG pathways²⁵, and 2,473 phenotypic gene sets (based on 211,882 gene-phenotype pairs from the Mouse Genetics Initiative²⁶) and 5,984 molecular pathways (based on 169,810 high-confidence experimentally-derived protein-protein interactions²⁷).

2.2.4 DEPICT method – false-discovery rate estimation

To estimate empirical false-discovery rates (FDR) that further controlled for effects of factors such as gene size and gene set composition, we repeated the DEPICT analysis 50 times. Each repeated randomized run was based on 566 input regions (See Online Methods) that were drawn from the pool of random regions matching for gene density. FDRs for the best prioritized gene at each region were estimated by first ordering the regions by the *P*-value (increasingly) of each regions' best-prioritized gene (henceforth referred to as 'tier one genes'). The same was done for each of the 50 repeated runs. The FDR for the region with the best tier one gene was estimated by counting the number of regions across all 50 randomized runs that contained a tier one gene with a *P*-value equal to or smaller than the observed regions' tier one gene's *P*-value. This count was divided by the number of randomized runs ($n=50$) multiplied by region-rank, where region-rank was the rank of the observed region (i.e. 1). Similarly, the FDR for the second best prioritized region and its tier one gene was estimated counting the number of regions across the 50 randomized runs that comprised a tier one gene with a *P*-value equal to or less than the tier one gene of the second best region. The FDR was then calculated by dividing this count by the 50×2 . Using this procedure, an FDR was estimated for the tier one gene in each region. To enable prioritization of multiple genes at a given region, we estimated FDRs for genes that were not tier one genes, but nevertheless exhibited low *P*-values (henceforth referred to as 'tier two genes'). We only considered genes that had *P*-values smaller than the *P*-value of least well prioritized tier one gene, which still had $FDR < 0.05$. The tier 'two' genes' FDRs were estimated by ranking all genes in all regions by their *P*-value and assigning the tier two genes the FDR of the next tier one gene in that list. To estimate the FDR for a given gene set we relied on the same pool of randomized regions as described in the Online Methods for DEPICT. First, we counted the number of gene sets across all 50 randomized runs that had a *P*-value equal to or smaller than the observed *P*-value of the given gene set. We then divided this count by the number of repetitions ($n=50$) multiplied by gene set-rank, where gene set-rank was the rank of the given gene set (among all gene sets ordered by increasing *P*-values).

2.2.5 Construction of DEPICT-based gene evidence table

Supplementary Table 16 includes all genes within any of the 566 DEPICT regions that were significantly prioritized ($FDR < 0.05$). Table columns either contain region specific information (such as genes that have missense variants in LD with or are nearest to the lead SNP), or qualitative/quantitative indications for a given genes' connection with height (such as DEPICT *P*-values). Unless otherwise stated, 'No' denotes that there was no evidence, while 'Yes' means that there was evidence. The 'OMIM human stature gene' column is based on the list of manually curated list of human stature genes²⁸ described in Section 2.1.5 and outlined in **Supplementary Table 9**. The 'Nominally significant in GRAIL' column reports whether a given gene was nominally significant in the GRAIL method (**Supplementary Table 13**) (GRAIL was run with the 697 SNPs as input, and no gene length correction was applied). The 'Associated with rodent growth plate differential tissue expression' column indicates whether the given gene has been reported to be differentially

expressed between growth plate cartilage and soft tissues (lung, kidney, heart) in male mice²⁸. The 'Associated with rodent temporal and spatial growth plate differential expression' indicates whether the given gene has been reported to be spatially regulated across different growth plate zones (resting vs. proliferative or proliferative vs. hypertrophic) or temporally regulated between 3-week and 12-week proliferative zones in male rats²⁸. The 'Mouse Genome Informatics database evidence' column denotes whether the given gene leads to a 'limbs/digits/tail' or skeleton phenotype (phenotype IDs MP:0005371, MP:0005390) upon knockout in mice. Knockout data was downloaded from the Mouse Genome Project database²⁶. The 'Within prioritized gene set' column lists whether the given gene was part of a reconstituted gene set that was significantly (FDR<0.05) prioritized in the DEPICT analysis. Genes were considered to be part of a given reconstituted gene set if their Z-score for the particular gene set was above 4.75 (comparable to $P < 10^{-6}$). The 'Blood eQTL in blood' column lists whether the given gene has been reported to be transcriptionally regulated in whole blood by one of the associated SNPs (or a SNP in linkage disequilibrium, $r^2 > 0.7$, with any of the associated SNPs). The expression quantitative trait locus (eQTL) data originated from a large-scale eQTL meta-analysis²¹. The '1000 Genomes Project nsVariant' column listed whether a gene had a missense variant (**Supplementary Table 7**), that was in linkage disequilibrium with any of the associated SNPs (or a SNP in high linkage disequilibrium, $r^2 > 0.7$) with any of the associated SNPs). The missense variants data originated from the 1000 Genomes Project Phase 1 data²⁹ (Download date 11/20/2012).

2.2.6 DEPICT method – overlap of reconstituted gene sets and visualization

To assess overlap of significant results from the DEPICT gene set enrichment analysis, we first computed the pairwise overlap of all gene sets with false-discovery rates less than 0.05. Hereafter, network diagrams were used to visualize pathway similarity by representing gene sets as nodes and gene set to gene set similarity as edges. All gene sets with a similarity above 0.1 were connected by an edge, gene sets with similarities above 0.25 were merged into single meta nodes, and edges were scaled according degree of similarity (ranging from 0.1 to 0.25). Similarity was calculated based on the *Jaccard* index, which is commonly used to assess the similarity between sets. The Cytoscape software^{30,31} was used to construct the network figure.

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The LifeLines Cohort Study

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