Supplementary Table 1. Genetic loci reported associated with body mass index (BMI), waist-hip-ratio adjusted for BMI (WHRadjBMI), visceral adipose tissue (VAT), body fat percentage or distribution tails of BMI and WHRadjBMI. The column source denotes the article, where the information in the column "description" was accessed. Gene names are those reported in original publication denoted in "source", and should not be regarded as causal genes.

SNP	Chr	Pos (GRCh38.p7)	Locus	Trait	Source	Description
rs977747	1	47219005	TAL1	BMI	Locke	**TAL1**: Transcription factor critical for development . Plays an important role in hemopoietic differentiation, and it has been implicated in hemopoietic malignancies. Serves as a regulator of erythroid differentiation. **Other notable genes: CMPK1**:Encodes an enzyme required for cellular nucleic acid biosynthesis that catalyzes the transfer of a phosphate group from ATP to CMP, UMP, or dCMP, to form the corresponding diphosphate nucleotide. **STIL:** regulator of mitotic entry and cell survival, associated with acute lymphoblastic leukemia.
rs657452	1	49124175	AGBL4	BMI	Locke	**AGBL4:** May play a role in the processing of tubulin and belongs to the peptidase M14 family.
rs11583200	1	50094148	ELAVL4	BMI	Locke	**ELAVL4:** Knockout mice displayed higher insulin levels in pancreatic islets, while ELAVL4-overexpressing mice exhibited lower insulin levels in islets and in plasma. RNA binding protein in neurons.
rs3101336	1	72285502	NEGR1	BMI, BMI tails	Locke	**NEGR1:** Upregulated during brain development, expressed on neurites

						of primary hippocampal neurons.
rs12566985	1	74536509	FPGT-TNNI3K	BMI	Locke	**FPGT-TNNI3K:** This locus represents naturally occurring read-through transcription from the neighboring fucose-1-phosphate guanylyltransferase (FPGT) and TNNI3 interacting kinase (TNNI3K) genes. TNNI3K - cardiac specific MAPK, directly interacts with cardiac troponin I, promotes cardiomyogenesis.
rs12401738	1	77981077	FUBP1- USP33	BMI	Locke	**FUBP1:** ssDNA binding protein that activates the far upstream element, functiosn as an ATP-dependent DNA helicase. **USP33:**This gene encodes a deubiquinating enzyme important in a variety of processes, including Slit-dependent cell migration and beta-2 adrenergic receptor signaling. The protein is negatively regulated through ubiquitination by von Hippel-Lindau tumor protein (VHL). USP33 regulates centrosome biogenesis via deubiquitination of the centriolar protein CP110. USP33 interacts with CP110 and localizes to centrioles primarily in S and G2/M phases, the periods during which centrioles duplicate and elongate. USP33 potently and specifically deubiquitinates CP110, but not other cyclin-F substrates. USP33 activity antagonizes SCF(cyclin F)-mediated ubiquitination and promotes the

						generation of supernumerary centriolar foci, whereas ablation of USP33 destabilizes CP110 and thereby inhibits centrosome amplification and mitotic defects. **Other notable genes:** AK5: Cyclic AMP dependent kinase regulator, ATP-AMP transphosphorylase. Expressed exclusively in brain. **GIPC2** might play important roles in human gastric cancer through modulation of growth factor signaling or cell adhesion.
rs11165643	1	96458541	PTBP2	BMI	Locke	**PTBP2:** Neuron development
rs17024393	1	109612066	GNAT2- AMPD2	BMI	Locke	**GNAT2:** Alpha subunit of G-protein transducin in cones. **AMPD2:** Encodes a protein that plays an important role in purine metabolism by catalyzing the conversion of AMP to IMP.Mutations associated with color blindness. **Other notable genes: GNAI3:** Implicated in adenylyl cyclase inhibition
rs2645294	1	119031964	TBX15-WARS2	WHRadjBMI	Heid	T-box 15 (**TBX15**) is a transcription factor that has been reported to be up-regulated in subcutaneous fat compared to intra-abdominal adipocytes, with expression levels closely correlated with the pattern of fat distribution (WHR) as well as overall obesity (BMI). TBX15 may be involved in embryonic development, pattern specification, adipocyte

						development, and specific adipose depot development underlying variation in fat distribution.. Further, TBX15 is implicated in Cousin syndrome, an autosomal recessive disorder characterized by congenital dwarfism, facial dysmorphism, and skeletal anomalies, phenotypes largely supported by animal models. **WARS2** is a mitochondrial tryptophanyl tRNA synthetase.
rs905938	1	155018913	DCST2	WHRadjBMI	Shungin	**DCST2** encodes dendritic cell-specific transmembrane protein domain containing 2, a multimembrane spanning protein that contains a domain similar to that found in dendritic cells. DC-STAMP proteins have been implicated in skewing hematopoietic differentiation of bone marrow cells toward the myeloid lineage, and in cell fusion during osteoclastogenesis and giant cell formation. A nearby gene is **ZBTB7B**, zinc finger and BTB domain containing 7B, also known as ThPOK, which encodes a zinc finger transcription factor that is critical to CD4+ T cell development in CD4/CD8 lineage commitment, and suppresses CD8- lineage gene expression. ZBTB7B has been shown to function as a transcriptional repressor of fibronectin and alpha1 collagen genes.

rs10919388	1	170403362	GORAB	WHRadjBMI	Shungin	**GORAB** encodes a member of the golgin family, and is a coiled-coil protein localized to the Golgi apparatus. This protein family may play a role in Rab6-regulated membrane-tethering events.
rs714515	1	172383850	DNM3-PIGC	WHRadjBMI	Heid	Dynamin 3 (**DNM3**) is a member of the dynamin family of enzymes that are important for interactions between the cell membrane and actin cytoskeleton. Dominant negative mutations in transfected dynamin enzymes promote GLUT6 and GLUT8 glucose transporters to the cell surface in cultured rat adipocytes in vitro. Phosphatidylinositol glycan anchor biosynthesis, class C (**PIGC**) encodes a subunit of the enzyme that transfers N-acetylglucosamine to phosphatidylinositol, the first step of glycosylphosphatidylinositol (GPI) lipid anchor biosynthesis. GPI anchors many proteins to the cell membrane. Another nearby gene is Fas ligand (**FASLG**) interacts with FAS to triggers apoptosis in some cell types. Additional nearby genes include **C1orf105** and **C1orf9**.
rs543874	1	177920345	SEC16B	BMI, BF%, BMI tails	Locke	**SEC16B**: The SEC16 protein is a peripheral endoplasmic reticulum (ER) membrane protein. SEC16 is part of the COPII coat vesicle proteins that help to transport proteins from the ER to the Golgi complex.

rs2820292	1	201815159	NAV1	BMI	Locke	**NAV1:** Netrin induced directional migration and axon guidance. **Other notable genes: IPO9:** mediates nuclear import of core histones into the nuclei of permeabilized cells. **LMOD1:** Actin binding protein associated with contractile apparatus of smooth muscles.Increased expression linked with Graves disease, an auto-immune related eye disease and thyroid associated-opthalmopathy
rs2820443	1	219580167	LYPLAL1	WHRadjBMI, VAT/SAT	Heid	**LYPLAL1** encodes the lysophospholipase-like 1 protein, which is thought to act as a triglyceride lipase and is reported to be up-regulated in subcutaneous adipose tissue of obese subjects. Other notable genes: Solute carrier family 30, member 10 (**SLC30A10**) belongs to a family of membrane transporters involved in intracellular zinc homeostasis and is expressed in brain and liver. Family member **SLC30A8** is a type 2 diabetes risk-locus
rs13021737	2	632348	TMEM18	BMI, BF%, BMI tails	Locke	**TMEM18**: human obesity gene, found in the majority of all brain sites, including the hypothalamus. Nuclear membrane protein binds DNA to supress expression.

rs10182181	2	24927427	ADCY3- POMC- NCOA1	BMI	Locke	**ADCY3:** Mediates odorant detection via modulation of intracellular cAMP concentration, mice lacking ADCY3 exhibit obesity that is apparently caused by low locomotor activity, hyperphagia, and leptin insensitivity. **NCOA1:**Encodes a member of the p160/steroid receptor coactivator (SRC) family that is a transcriptional coactivator for steroid and nuclear hormone receptors. POMC:Expressed in pituitary;Neurons in the arcuate nucleus of the hypothalamus are crucial for normal energy homeostasis. **CENPO:**This gene encodes a component of the interphase centromere complex. The encoded protein is localized to the centromere throughout the cell cycle and is required for bipolar spindle assembly, chromosome segregation and checkpoint signaling during mitosis. **DNAJC27:** Cerebellar granule neurons
rs11126666	2	26705943	KCNK3	BMI	Locke	**KCNK3:** expressed in many neurons likely to be cholinergic, serotonergic, or noradrenergic.
rs1016287	2	59078490	LINC01122	BMI	Locke	**LINC01122:** uncharacterized locus. **Other notable genes: FANCL**: Encodes an ubiquitin ligase protein that mediates ubiquitination of FANCD2, a key step in the DNA damage pathway. Involved in Fanconi anemia. Necessary for primordial

						germ cell proliferation, associated with acute lung injury in mice.
rs11688816	2	62825913	EHBP1	BMI	Locke	**EHBP1:** Mutation in EHBP1 have been shown to cause a rare form of prostate cancer. EHBP1 is important for the early stages of receptor-mediated endocytosis. With EHDS2, links clathrin-coated vesicles to the actin cytoskeleton. Associated with prostate cancer and suggested role in neuroblastoma. Loss inhibits endocytosis. EHBP1 disrupts insulin-regulated GLUT4 recycling in cultured adipocytes
rs1385167	2	65973514	MEIS1	WHRadjBMI	Shungin	The lead WHRadjBMI-associated SNP is located ~500 kb from **MEIS1**, which encodes a transcription factor that is a member of the three-amino-acid loop extension family of homeobox-containing proteins. Meis1 is essential for hematopoiesis and vascular patterning in the mouse embryoand regulates vascular development in zebrafish. Dysregulation of MEIS1 expression has been linked to a variety of leukemias. The lead SNP is also <400 kb from **miR4778**.

rs1659258	2	88360069	THNSL2	VAT	Fox	The lead SNP for VAT in women, rs1659258, is located in an intergenic region upstream from ***THNSL2*** and ***FABP1***. However, rs1659258 is not in linkage disequilibrium (LD) with any coding SNPs within 88,200-88,700 kb. In addition, the correlations between rs1659258 with coding SNPs in ***FOXI3***, ***C2orf51***, ***THNSL2***, ***EIF2AK3***, ***FABP1***, and ***SMYD1*** genes are low (r2<0.15). Finally, there is no evidence that 2p11-p12, where rs1659258 is located, has been previously implicated in association with copy number variation in adipose-related human disease. Nonetheless, we explored the potential biology in this region. Fatty acid binding protein is produced in the liver and is involved with fatty acid metabolism. Free fatty acid flux has previously been shown to be more strongly associated with visceral as compared to subcutaneous fat. In addition, women have been shown to have a faster rate of non-oxidative free fatty acid disposal as compared to men, but without concomitant worsened metabolic risk factor profiles. While *FABP1* represents an exciting potential candidate gene, rs1659258 resides in a neighboring linkage disequilibrium block that does not contain any genes. ***THNSL2*** is just downstream of

						FABP1 and our lead SNP demonstrates nominal gene expression to *THNSL2*, which is part of the threonine synthase family. A recent analysis of RNA expression in 225 healthy Pima Indian skeletal muscle biopsies showed a bimodal (ie two discrete clusters) expression of *THNSL2*, thought to occur due to cis-acting polymorphisms .
rs2121279	2	142285716	LRP1B	BMI	Locke	**LRP1B:** Membrane-bound receptor protein found deleted or silenced in many cancers. LRP1B is a receptor gene that helps to regulate endocytosis of many proteins.
rs1460676	2	163711179	FIGN	BMI	Locke	**FIGN:** ATPases associated molecular chaperones facilitates a variety of cellular functions
rs10195252	2	164656581	GRB14-COBLL1	WHRadjBMI, WHRadjBMI tails, BF%	Heid	Growth factor receptor-bound protein 14 (**GRB14**) is a member of a family of SH2-containing adaptors and binds directly to the insulin receptor. Grb14-deficient mice exhibit enhanced body weight, mainly explained by increased lean mass on normal diet, improved glucose homeostasis despite lower circulating insulin levels, and enhanced insulin signaling in liver and skeletal muscle. Grb14 expression is increased in adipose tissue of insulin-resistant animal models and type 2 diabetic

						human patients,suggesting that Grb14 may modulate insulin sensitivity. Cordon-bleu protein-like 1 (**COBLL1**) may be involved in neural tube formation. The WHR signal is distinct from a locus previously associated with smoking initiation and current smoking (rs4423615, D'=0.01, r<.01, HapMap CEU).
rs1528435	2	180686235	UBE2E3	BMI	Locke	**UBE2E3:** Enzyme that is thought to accept ubiquitin from the E1 complex and catalyse its covalent attachment to other proteins.
rs1569135	2	187250671	CALCRL	WHRadjBMI	Shungi n	**CALCRL** encodes calcitonin receptor-like protein receptor, involved in G-protein coupled receptor-like signaling. Calcitonin receptor-like receptor, CRLR, along with receptor activity-modifying protein-2, RAMP2, is a receptor for adrenomedullin. Adrenomedullin and LRL/RAMP2 levels were increased in epididymal, mesenteric, and retroperitoneal adipose tissue in rats fed a high-fat diet compared to rats fed a normal diet. CRLR mRNA levels were decreased in epicardial white adipose tissue compared to subcutaneous white adipose tissue from human biopsies. A nearby gene, **TFPI**, encodes a protease inhibitor that regulates the tissue factor (TF)-dependent pathway of blood coagulation. The encoded protein is

						predominantly found in the vascular endothelium and plasma in both free forms and in complexes with plasma lipoproteins.
rs17203016	2	207390794	CREB1- KLF7	BMI	Locke	**CREB1**: Encodes a transcription factor that binds as a homodimer to cAMP-responsive elements and induces transcription of genes in response to hormonal stimulation of the cAMP pathway. **KLF7:** protein may contribute to the progression of type 2 diabetes by inhibiting insulin expression and secretion in pancreatic beta-cells and by deregulating adipocytokine secretion in adipocytes.
rs7599312	2	212548507	ERBB4	BMI	Locke	**ERBB4:** binds to and is activated by neuregulins inducing mitigenesis and differentiation. Tyrosine kinase activated by neuregulins to induce mitogenesis and differentiation. Associated with breast cancer. Expression in hippocampus and cortex
rs492400	2	218485029	PLCD4- CYP27A1- USP37- TTLL4- STK36- ZNF142- RQCD1	BMI	Locke	**CYP27A1:** Degradation of cholesterol to bile acids; Mutations associated with lipid storage disease; involved in Vit D3 metabolism **PLCD4:** Hydrolyzes phosphatidylinositol 4,5-bisphosphate into two intracellular second messengers, inositol 1,4,5-trisphosphate and diacylglycerol

						RQCD1:Transcription factor, but true function unknown. **STK36**: defects lead to congenital hydrocephalus **TTLL4:** Tubulin tyrosine ligase-like **USP37:** removes polyubiquitin from cyclin A, a protein that controls cell cycle progression ZNF142:C2H2-type zinc finger protein
rs2176040	2	226228086	LOC646736- IRS1	BMI, BF%	Locke	**IRS1**: Mutations in this gene are associated with type II diabetes and susceptibility to insulin resistance. **LOC646736:** uncharacterized locus
rs17819328	3	12447843	PPARG	WHRadjBMI	Randall	The protein product encoded by **PPARG** is PPAR - gamma which is a regulator of adipocyte differentiation. Additionally, PPAR - gamma has been implicated in the pathology of numerous diseases including obesity, diabetes, atherosclerosis and cancer. Interestingly, previous studies of the Pro12Ala polymorphism in the PPAR gene have demonstrated genotype – by - sex interaction with BMI, fatty acid concentrations during the first 24h after birth w ere related to PPARG expression in female but not in male lambs, and female 12Ala mutation carriers had greater risk of developing abdominal obesity than female non - carriers while male 12Ala mutation carriers had no significant increase in risk.

rs6804842	3	25064946	RARB	BMI	Locke	**RARB:** regulation of brain functions, and retinoic acid signaling defects may contribute to pathologies such as Parkinson's disease and schizophrenia. Mutated RARB mouse show locomotor defects RARB:Encodes retinoic acid receptor beta, which binds to retinoic acid and mediates cellular signaling in embryonic morphogenesis, cell growth and differentiation. RARB:Primary role in the cell is limiting the growth of many cell types by regulating gene expression. (e.g. through the regulation of IGFBP3).
rs2276824	3	52603470	PBRM1	WHRadjBMI	Heid	Previously named "**NISCH-STAB**" Nischarin is encoded by NISCH and interacts with insulin receptor substrate 4, which is phosphorylated by the insulin receptor tyrosine kinase upon receptor stimulation in kidney cellsbut has also been reported to play a redundant role in insulin receptor signaling. Other nearby genes include troponin C type 1 (**TNNC1**), semaphoring 3G (**SEMA3G**), which is highly expressed in adipocytes, stablin 1 (**STAB1**), which is a scavenger receptor that plays a role in intracellular trafficking, 5'-nucleotidase domain containing 2 (**NT5DC2**), PHD finger protein 7 (**PHF7**), BRCA1 associated protein-1 (**BAP1**), which helps regulate cell

						growth and proliferation, hypothetical protein (LOC440957), polybromo 1 (**PBRM1**), glycerate kinase (GLYCTK), and dynein axonemal heavy chain 1 (DNAH1). Mdhc7 34 null male mice (homologous to DNAH1) are infertile due to decreased sperm motility.
rs2365389	3	61250788	FHIT	BMI	Locke	**FHIT:** Overlaps fragile site FRA3B. Encodes a hydrolase involved in DNA replication and stress response. Tumor suppressor FHIT:Encodes a diadenosine 5',5'''-P1,P3-triphosphate (AP3A) hydrolase involved in purine metabolism and the regulation of DNA replication and signaling stress response.
rs2371767	3	64732582	ADAMTS9	WHRadjBMI	Heid	SNP rs6795735 is located upstream of zinc-dependent protease **ADAMTS9**. It is a member of the ADAMTS family, a group of genes encoding metalloproteases that lack transmembrane domains and are secreted into the extracellular matrix. ADAMTS9 is associated with type 2 diabetes, possibly mediated through decreased insulin sensitivity of peripheral tissues. The index SNP is also located in noncoding RNA BC040632 and within 400 bp of the start of microRNA-548a-2.

rs3849570	3	81742961	GBE1	BMI	Locke	Mutations in **GBE1** lead to glycogen storage disease IV characterized by accumulation of a poorly branched form of glycogen known as polyglucosan. polyglucosan in liver is a degradable source of glucose and readily depleted by fasting. Deficiency of glycogen branching enzyme and different mutations in the GBE1 gene, located on chromosome 3, is causative of Glycogen Storage Disease type IV (GSD-IV), a rare autosomal recessive disorder of the glycogen synthesis, characterized by the accumulation of polyglucosan, in almost all tissues, especially the liver or the neuromuscular system. GBE1:Mutations in this gene are associated with glycogen storage disease IV . Common missense mutation identified in APBD in Ashkenazi patients. APBD is characterized by the accumulation of insoluble glucose polymers within the central and peripheral nervous systems.
rs13078960	3	85758440	CADM2	BMI	Locke	**CADM2**: Homophilic and heterophilic cell adhesion molecule at the synapse leading to cell aggregation.
rs10804591	3	129615390	PLXND1	WHRadjBMI	Shungin	**PLXND1**, plexin D1 PLXND1 encodes plexin D1 protein, a co-receptor for semaphorin proteins. Plxnd1 is expressed in cell from the central nervous system and vascular

						endothelium in mouse embryos. Plexin D1 plays a role in vascular patterning; plxnD1-deficient zebrafish embryos show defects in segmental artery development such as premature and ectopic sprouting and improper blood vessel branching. Semaphorin-plexinD1 signaling antagonizes the proangiogenic activity of vascular endothelial growth factor, VEGF.
rs16851483	3	141556594	RASA2	BMI	Locke	**RASA2:** Regulation of MAPKinase activation in neuronal cells. **Other notable genes: ZBTB38:** Transcriptional activator that binds to methylated DNA. Inhibition linked to apoptosis. Associated with height.
rs17451107	3	157079820	LEKR1	WHRadjBMI	Shungin	**LEKR1**, leucine, glutamate and lysine rich 1 protein LEKR1 encodes leucine, glutamate and lysine rich 1 protein with unknown function. The lead WHRadjBMIassociated SNP is also located near CCNL1, encoding cyclin L1, and two uncharacterized noncoding RNAs, LINC00880 and LINC00881. Also nearby, **TIPARP** encodes a poly(ADP-ribose) polymerase superfamily member, which catalyzes the transfer of multiple ADP-ribose groups from nicotinamideadenine dinucleotide (NAD) onto protein targets, and VEPH1 encodes ventricular zone expressed PH domain-containing 1

rs1516725	3	186106215	ETV5	BMI, BMI tails	Locke	**ETV5:** Expressed in hypothalamus, critical regulator of energy balance. ETV5 knockout mice are infertile but also have reduced body weight compared to wild-type controls, suggesting a role in energy balance.
rs10938397	4	45180510	GNPDA2- GABRG1	BMI, BMI tails	Locke	**GABRG1:** major inhibitory neurotransmitter in the vertebrate brain, alcohol dependence. **GNPDA2** is down-regulated in the hypothalamus of HFD rats compared to chow-fed rats. Same variant associated with increased intramyocellular lipids, but not after bonferoni correction
rs3805389	4	55616583	NMU	WHRadjBMI	Shungin	**NMU**, neuromedin U NMU encodes neuromedin U, a highly conserved neuropeptide. NMU is found at highest levels in the gastrointestinal tract and pituitary, and has been implicated in the regulation of smooth muscle contraction, blood pressure and local blood flow, ion transport in the gut, stress responses, cancer, gastric acid secretion, and feeding behavior. Nmu knockout mice are hyperphagic and obese. Rare coding variants in NMU have been found to be associated with obesity
rs17001654	4	76208415	NUP54- SCARB	BMI	Locke	**NUP54:** creates distinct nuclear and cytoplasmic compartments in eukaryotic cells and regulates the flow of macromolecules between nucleus and cytoplasm. **SCARB** is

						involved in the endocytosis of bacteria, including bacterial GroEL protein.
rs9991328	4	88791970	FAM13A	WHRadjBMI	Shungin	**FAM13A**, family with sequence similarity 13, member A FAM13A has a putative role in signal transduction, however is poorly described. SNPs in this gene region were found to be associated with chronic obstructive pulmonary disease and lung function. Other nearby genes include **HERC3, NAP1L5, PIGY** (phosphatidylinositol-glycan biosynthesis class Y protein), and TIGD2.
rs13107325	4	102267552	SLC39A8	BMI	Locke	SLC39A8:The encoded protein functions in the cellular import of zinc at the onset of inflammation. It is also thought to be the primary transporter of the toxic cation cadmium, which is found in cigarette smoke.
rs303084	4	123145793	SPATA5-FGF2	WHRadjBMI	Shungin	**SPATA5**, spermatogenesis associated 5 – **FGF2**, fibroblast growth factor 2 SPATA5 belongs to the AAA ATPase family and AFG2 subfamily, and may be involved in mitochondrial transformation during spermatogenesis. SNPs at SPATA5 have been associated with alopecia areata. Other nearby genes include **FGF2, NUDT6, and SPRY1**. FGF2 enhanced vascularization for human adipose tissue engineering. NUDT6 (nudix-type motif 6) is an antisense gene to FGF2 that showed

						associations with fat deposition related traits in pigs. Conditional Spry1 (sprouty homolog 1) expression in mouse adipose tissue protected against high-fat diet-induced obesity, bone loss, and metabolic dysfunction
rs11727676	4	144737912	HHIP	BMI	Locke	**HHIP**: Encodes a highly conserved, vertebrate-specific inhibitor of hedgehog signaling. Hedgehog proteins are important in embryonic development.
rs9687846	5	56566067	MAP3K1	WHRadjBMI	Shungin	**MAP3K1**, mitogen-activated protein kinase kinase kinase 1, E3 ubiquitin protein ligase The lead SNP is located within the intron of an uncharacterized transcript AC022431. Located 250 kb away, MAP3K1, also known as MEKK1, encodes a protein in the MAPK group of serine/threonine protein kinases. The protein contains a PHD plant homeodomain that exhibits E3 ubiquitin ligase activity toward ERK1/2. MAP3K1 also activates the JNK signaling pathway and plays a role in apoptosisand wound healing. Along with IL-1beta, MAP3K1 inhibited basal and membrane depolarization and cAMP-induced transcription of the insulin gene in a hamster beta cell line

rs2112347	5	75719417	POC5- HMGCR- COL4A3BP	BMI	Locke	**COL4A3BP:** Involved in ceramide intracellular transport. This gene encodes a kinase that specifically phosphorylates the N-terminal region of the non-collagenous domain of the alpha 3 chain of type IV collagen, known as the Goodpasture antigen. Goodpasture disease is the result of an autoimmune response directed at this antigen. Involved in ceramide intracellular transport **HMGCR**:Encodes HMG-CoA reductase which is the rate-limiting enzyme for cholesterol synthesis.Rate-limiting enzyme in cholesterol synthesis. Negatively regulated by internalization of LDL by LDLR. **POC5:** involved in centrosome biogenesis, associated with BMI
rs1045241	5	119393591	TNFAIP8-HSD17B4	WHRadjBMI	Randall	The protein encoded by **HSD17B4** is a bifunctional enzyme that is involved in the peroxisomal beta - oxidation pathway for fatty acids. Mutations in this gene are known to cause DBP deficiency, an autosomal - recessive disorder of peroxisomal fatty acid beta - oxidation that is generally fatal within the first two years of life . Expression levels of HSD17B4 have been associated with prostate cancer severity , and it is also a significant independ ent predictor of poor patient outcome

rs7715256	5	154158333	GALNT10	BMI	Locke	**GALNT10:** These proteins transfer GalNAc from UDP-GalNAc to either serine or threonine residues of polypeptide acceptors. Obesity in AfricanAmericans.
rs7705502	5	173893812	CPEB4	WHRadjBMI	Heid	Cytoplasmic polyadenylation element binding protein **CPEB4** promotes polyadenylation-induced translation. CPEB nucleates a complex of factors that regulate polyadenylation elongation through a deadenylating enzyme and mediate many processes including germ-cell development, cell division and cellular senescence and synaptic plasticity. Other nearby protein-coding genes include **C5orf47 and HMP19**
rs6556301	5	177100576	FGFR4	WHRadjBMI	Shungi n	**FGFR4,** fibroblast growth factor receptor 4 FGFR4 is a member of the receptor tyrosine kinase family. FGFR4 is expressed mainly in lung, kidney, pancreas, spleen and developing muscle. FGFR4-deficient mice on a normal diet displayed increased mass of white adipose tissue, hyperlipidemia, glucose intolerance, insulin resistance and hypercholesterolemia
rs1294410	6	6738519	LY86	WHRadjBMI, WHRadjBMI tails	Heid	The closest known protein-coding gene is lymphocyte antigen 86 (**LY86**), which plays a role in recognition of lipopolysaccharide via the Tolllike receptor pathway when bound as a heterodimer with RP105. It has been

						associated with asthma and has been suggested to play a role along with RP105 in autoimmune diseases. The lead SNP (rs1294421) is closely correlated (r^2=0.9845) with a 2,832 bp CNV at the LY86 locus, CNVR2760.1. The closest transcripts to this CNV are noncoding RNA BC039678 (12 kb centromeric) and LY86 (87 kb centromeric).
rs7759742	6	32413959	BTNL2	WHRadjBMI	Shungin	**BTNL2**, butyrophilin-like 2 (MHC class II associated) Located 30 kb from the HLA cluster, BTNL2 is an MHC class II gene-linked butyrophilin family member that inhibits T-cell activation. Variants in BTNL2 are associated with inflammatory diseases. Other nearby genes include HLA-DRA, HLA-DRB5, HLA-DRB1, HLA-DRB6, HLA-DRB1, HLA-DQA1, HLADQB1. These HLA genes belong to the HLA class II proteins, which are expressed in antigen presenting cells, such as B lymphocytes, macrophages, and dendritic cells
rs1776897	6	34227234	HMGA1	WHRadjBMI	Shungin	**HMGA1**, high mobility group AT-hook 1 HMGA1 encodes a protein that binds to the minor groove of stretches of A-T-rich DNA. HMGA1 is a downstream nuclear target of the insulin receptor signaling pathway, and Hmga1 knockout mice showed decreased insulin receptor expression, impaired insulin signaling

						and reduced insulin secretion.
rs205262	6	34595387	C6orf106- SNRPC	BMI	Locke	**SNRPC** encodes one of the specific protein components of the U1 small nuclear ribonucleoprotein (snRNP) particle required for the formation of the spliceosome UHRF1BP1:May act as a negative regulator of cell growth. This locus has been associated with systemic lupus erythematosus susceptibility**.C6orf106:**Uncharacterized protein. **Other notable genes: ANKS1A:** Locus associated with human height. Anks proteins enhance EphA receptors which are involved in intracellular signaling pathways regulating bone development, angiogenesis, axon guidance, cell adhesion and migration. ANKS1A:Positive regulation of ephrin receptor signalling pathway
rs2033529	6	40380914	TDRG1- LRFN	BMI	Locke	**LRFN2:** Synaptic adhesion molecule expressed only in brain, spleen, and testis. Promotes neurite outgrowth and targeting of NMDA receptors and PSD95 to dentritic puncta. Promotes neurite outgrowth in hippocampal neurons **TDRG1:** Expressed mainly in post-puberty testis. Expression in seminoma and teratoma suggest possible role as tumor suppressor.

rs1358980	6	43796814	VEGFA	WHRadjBMI	Heid	Multiple variants and mutations in vascular endothelial growth factor A (**VEGFA**) are risk factors for diabetic retinopathy. Variants in VEGFA have been nominally associated with T2D.VEGFA is proposed as a key 35 mediator of adipogenesis and angiogenesis, is highly expressed in adipose tissue, and has increased expression during adipocyte differentiation. VEGFA serum concentrations are elevated in overweight and obese patients compared with lean subjectsand decrease after weight loss following bariatric surgery, behaving similarly to other hormones related to adipose mass, such as leptin and insulin
rs2207139	6	50877777	TFAP2B	BMI, BMI tails	Locke	**TFAP2B:** Encodes a transcription factor involved in embryonic development. Mutations in this gene result in autosomal dominant Char syndrome.
rs9400239	6	108656460	FOXO3- HSS0029640	BMI	Locke	**FOXO3:** Transcriptional activator with roles in cell cycle control and apoptosis. Nerve growth factor receptor signaling pathway; inactivated by insulin via a PI3K/AKT dependent pathway
rs9374842	6	119864519	LOC285762	BMI	Locke	**LOC285762:**Uncharacterized locus. **Other notable genes: MAN1A1:** exerts an inhibitory effect on T-cell activation. **MCM9:** invovled in initiation of eukaryotic genome

						replication
rs1936805	6	127130971	RSPO3	WHRadjBMI, WHRadjBMI tails	Heid	R-spondin 3 homolog **(RSPO3**) encodes a secreted protein that regulates beta-catenin signaling. RSPO3 promotes angiogenesis and vascular development. Rspo3 knockout mice die due to defects in placental development, and Rspo3 is required for Vegf expression and endothelial cell proliferation. Rspo3 has also been shown to be an oncogene in mouse mammary epithelial cells. rs9491696 is also located within 160kb of ring finger 146 (RNF146) and enoyl Coenzyme A hydratase domain containing 1 (ECHDC1), and near to previously identified breast cancer susceptibility variant rs2180341. ECHDC1 encodes for a protein that shares the enoyl-Coenzyme A hydratase domain with trifunctional protein (HADHA), which is an enzyme involved in fatty acid oxidationthat is expressed highly in adipocyte
rs13201877	6	137354404	IFNGR1- OLIG3	BMI	Locke	**IFNGR1:** A genetic variation in IFNGR1 is associated with susceptibility to Helicobacter pylori infection. Defects in IFNGR1 are also a cause of mendelian susceptibility to mycobacterial diseases IL22RA2:functions as an IL22 antagonist, and may be important in

						the regulation of inflammatory response. **OLIG3:** Necessary for development of dorsal spinal cord
rs13191362	6	162612318	PARK2	BMI	Locke	**PARK2** is expressed in the neuronal processes and cell bodies of neurons. Regulates the function and stability of excitatory glutamatergic synapses. Functions within a multiprotein E3 ubiquitin ligase complex. Mutations can cause Parkinson disease and autosomal recessive juvenile Parkinson disease. PARK2 encodes a protein likely involved in calcium-dependent exocytosis in neurites, possibly involving the release of dopamine. **Other notable genes: PACRG** in astrocytes throughout the brain and in pigmented noradrenergic neurons, Lewy bodies, Co-regulated with PARK2 and is found in Lewy bodies of Parkinson's patients. Binds Pael receptor to prevent cell death.
rs10245353	7	25818994	NFE2L3	WHRadjBMI	Heid	The index SNP rs1055144 is more than 300 kb from the nearest known protein-coding gene, the transcription factor nuclear factor (erythroid-derived 2)-like 3 (**NFE2L3**). rs1055144 is located within the exon of spliced ESTs (e.g. BX116657) and <20 kb from a set of unspliced ESTs (e.g. AA553656, corresponding to Contig27623_RC). rs1055144 is also upstream of hypothetical protein **LOC646588,** and >100 kb from

						microRNA-148a, which can repress the DNA methyltransferase DNMT3B.
rs1534696	7	26357619	SNX10	WHRadjBMI	Shungin	**SNX10**, sorting nexin 10 SNX10 encodes a nexin family protein involved in intracellular trafficking. SNX10 has been shown to cause osteopetrosis, a rare disorder resulting from osteoclast dysfunction, and to regulate ciliogenesisand endosome
rs7801581	7	27184152	HOXA11	WHRadjBMI	Shungin	**HOXA11**, homeobox A11 There are 12 HOXA genes at this locus, as well as several antisense transcripts. HOX genes encode conserved transcription factors containing a homeodomain that regulate body and axis development and organogenesis. HOXA11 is necessary for female fertility and regulates embryonic uterine and endometrium development. HOXA11 mutations were found in individuals affected with the blood disease amegakaryocytic thrombocytopenia and the skeletal defect radio-ulnar synostosis

rs1167827	7	75533848	HIP1- PMS2L- PMS2P- WBSCR1	BMI	Locke	**HIP1:** Membrane-associated protein in endocytosis. Loss of interaction with huntingtin protein may lead to defect in membrane-cytoskeleton integrity in brain. HIP1 is involved in the intracellular trafficking of many proteins, most notably, huntingtin. HIP1 is also involved in cellular apoptosis. HIP1:Double knockout mice for HIP1 result in growth failure and dwarfism that can be lessened by the introduction of human HIP1. Neurodevelopmental; Regulates AMPA receptor trafficking in the central nervous system through its function in clathrin-mediated endocytosis. **PMS2L3:** member of a family of genes that share a high degree of identity with PMS2. **PMS2P5:** postmeiotic segregation increased 2 pseudogene 5 POM121C:Component of the nuclear pore complex. **POM121C:**integral membrane components of the nuclear pore complex WBSCR16:encodes an RCC1-like G-exchanging factor. It is deleted in Williams syndrome, a multisystem developmental disorder caused by the deletion of contiguous genes at 7q11.23
rs2245368	7	76978826	PMS2L11	BMI	Locke	**PMS2L11:** pseudogene with similarity to PMS2, associated with BMI

rs9641123	7	93568420	CALCR- hsa-miR-65	BMI	Locke	**CALCR:** High levels are expressed by normal human T and B lymphocytes from tonsils and peripheral blood in relation to their activation status. GPCR, involved in maintaining calcium homeostasis and in regulating osteoclast-mediated bone resorption
rs6465468	7	95540202	ASB4	BMI	Locke	**ASB4:** Asb-4 is a key regulatory protein in the central nervous system, involved in the control of feeding behavior and metabolic rate.
rs7830933	8	23745811	NKX2-6	WHRadjBMI	Shungin	**NKX2-6,** NK2 homeobox 6 NKX2-6 encodes a homeobox-containing protein that is a homolog of Drosophila tinman. At early stages of mouse embryogenesis, NKX2-6 is expressed in the pharyngeal endoderm, developing gut endoderm, cardiac progenitors, and heart. Nearby **NKX3-1** is also a homeobox gene that is involved in prostate epithelium development during embryogenesisand is androgen-regulated. STC1 encodes a secreted, homodimeric glycoprotein that is expressed in a wide variety of tissues and is upregulated by VEGFD. STC1 may play a role in the regulation of renal and intestinal calcium and phosphate transport, cell metabolism, and angiogenesis.

rs12679556	8	71601993	MSC	WHRadjBMI	Shungin	**MSC**, musculin MSC encodes a basic helix-loop-helix transcription factor expressed in developing skeletal muscleand mouse embryonic ectoderm. EYA1 encodes eyes absent homolog1, a protein phosphatase and coactivator for the transcription factor SIX1, which regulates skeletal muscle fiber-type and development. Mutations in EYA1 cause Branchio-oto-renal syndrome and Branchiootic syndrome, which are characterized by hearing loss, branchial arch defects and renal abnormalities. EYA protein phosphatase activity promotes angiogenesis
rs17405819	8	75894349	HNF4G	BMI	Locke	**HNF4G:** A nuclear receptor that serves as a transcriptional activator. HNF4-gamma knockout (HNF4-gamma(-/-)) mice had lowered energy expenditure and locomotor activity during night time that resulted in a higher body weight despite having reduced intake of food and water.
rs16907751	8	80463222	ZBTB10	BMI	Locke	**ZBTB10**: zinc finger and BTB domain binding
rs2033732	8	84167474	RALYL	BMI	Locke	**RALYL:** RNA binding protein
rs4740619	9	15634328	C9orf93	BMI	Locke	**C9orf93:** Coiled coil domain containg protein. **PSIP1**: Transcriptional co-activator, can also coordinate pre-mRNA splicing and prevent apoptosis in photoreceptor cells. Neuro epithelial stem cell differentiation and neurogenesis

rs10968576	9	28414341	LINGO2	BMI	Locke	LINGO2:Expressed exclusively in the neuronal system.
rs10991437	9	104973639	ABCA1	WHRadjBMI	Shungin	ABCA1, ATP-binding cassette, sub-family A (ABC1), member 1 This gene encodes an ATP-binding cassette transporter. Mutations in ABCA1 have been found to be associated with Tangier's disease and familial high-density lipoprotein deficiency. Adipose tissue abundantly expresses ABCA1, and adipose tissue ABCA1-dependent cholesterol efflux and nascent HDL particle formation contribute to systemic HDL biogenesis
rs6477694	9	109170062	EPB41L4B- C9orf	BMI	Locke	**C9orf4:** ferric-chelate reductase 1-like. **EPB41L4B:** Structural constituent of the cytoskeleton
rs1928295	9	117616205	TLR4	BMI	Locke	**TLR4:** Innate immune system participates in the regulation of energy balance and insulin resistance in response to changes in the nutritional environment. :TLR4 cooperates with LY96 and CD14 to mediate the innate immune response to bacterial lipopolysaccharide (LPS).
rs10733682	9	126698635	LMX1B	BMI	Locke	**LMX1B:**Transcription factor essential for the normal development of dorsal limb structures, kidneys, eyes and neurons. Mutations associated with nail-patella syndrome. Essential for the differentiation and survival of the central serotonergic neurons (which are involved in regulation of mood,

						appetite and sleep) during embryonic development LMX1B:Transcription factor involved in dorsal/ventral patterning, limb and eye development. Mutations cause nail-patella syndrome
rs7899106	10	85651147	GRID1	BMI	Locke	**GRID1:** Receptor involved in synaptic transmission by L-glutamate neurotransmitter
rs17094222	10	100635683	HIF1AN	BMI	Locke	**HIF1AN** is essential for tumor growth through suppression of the p53-p21 axis. **Other notable genes: NDUFB8:** This gene encodes an enzyme subunit of the NADH Dehydrogenase, the first enzyme of the mitochondrial electron transport chain. **PAX2:** Formation of midbrain-hindbrain boundary, controls cerebellum and midbrain development. DNA-binding transcription factor, expressed in ears and eyes. Induces kidney development, and is induced by tamoxifen and estrogen. Member of Paired box (Pax) genes and homeobox genes. Pax genes are a family of tissue specific transcription factors that are important during early tissue development. PAX2 is believed to be important for development of the urogenital tract, the eyes, ears, kidneys, and the CNS. In adulthood, PAX2 is important for cellular maintenance under cellular stress. SEC31B: There is nothing known of

						the function of this gene. The GO prediction is based upon the homology of this gene to a similar endocytotic gene in rats. Beta-catenin - TCF pathway through switching intracellular WNT signaling pathways. **WNT8B:** Expression restricted to developing brain. Secreted signaling protein expressed in the developing brain for cell fate and embryonic patterning WNT8B:Member of the Wnt-signaling pathway. Little is known about WNT8B specifically. May play an important role in the development and differentiation of certain forebrain structures, notably the hippocampus.
rs7917772	10	102727686	SFXN2	WHRadjBMI	Shungin	**SFXN2,** sideroflexin 2 SFXN2 encodes a mitochondrial transmembrane protein that may facilitate transport of pyridoxine or enzyme cofactors involved in heme synthesis into the mitochondria. The gene is widely expressed, and is expressed at particularly high levels in adult kidney and liver. Sfxn2 was found upregulated in pancreatic islets from streptozotocin-induced diabetic rats compared to normal rats
rs11191560	10	103109281	NT5C2- CYP17A- SFXN	BMI	Locke	**CYP17A1:** encodes a key biosynthesis enzyme of estrogen, which is critical in regulating adipogenesis and adipocyte development in humans. Candidate gene for obesity

						susceptibility in Caucasians. **NT5C2:** Encodes one of seven 5'-nucleotidases that catalyze the dephosphorylation of nucleoside monophosphates and help regulate and balance ribo- and deoxyribonucleotide levels for RNA and DNA synthesis. **SFXN2:** Sideroflexin (no specific information)
rs7903146	10	112998590	TCF7L2	BMI	Locke	**TCF7L2:** WNT signaling pathway effector. High mobility group(HMG) box-containing transcription factor that plays a role in Wnt signaling. This is the strongest T2D-associated SNP in GWA meta-analyses. Heterozygous knockout mice have 10-15% lower body weight compared to wt littermates.
rs4256980	11	8652392	TRIM66- TUB	BMI	Locke	**TRIM66** plays a role in heterochromatin-mediated gene silencing during spermatogenesis, associated with age at menarche. **TUB:** The mouse homolog 'tubby' gene (tub) causes maturity-onset obesity, insulin resistance, retinal degeneration, and neurosensory hearing loss. Central nervous system-high expr in hypothalamus, obesity resulting from defects in neuroendocrine control of satiety or metabolism**. SNORA3:** guides site-specific rRNA modifications SNORA45:guides site-specific rRNA modifications. **ST5:** CNS development.

						This protein preferentially binds to the SH3 domain of c-Abl kinase, and acts as a regulator of MAPK1/ERK2 kinase, which may contribute to its ability to reduce the tumorigenic phenotype in cells. Involved in a signaling transduction pathway leading to activation of MAPK1/ERK2
rs11030104	11	27662970	BDNF	BMI, BMI tails	Locke	**BDNF:** Hypothalamic regulation of energy homeostasis BDNF:Major regulator of synaptic transmission and plasticity at adult synapses. Downstream components in the MC4R-mediated control of energy balance. Self-amplifying BDNF actions ensure stable elevation of local cAMP/protein kinase A activity that is critical for axon differentiation and growth
rs2176598	11	43842728	HSD17B12	BMI	Locke	**HSD17B12:** Encodes 17beta-hydroxysteroid dehydrogenase that converts estrone into estradiol and is involved in fatty acid elongation.

rs3817334	11	47629441	MTCH2- C1QTNF4- SPI1- CELF1	BMI	Locke	**MTCH2:** Induces mitochondrial depolarization. Found in the mitochondrial inner membrane. **NDUFS3:** This gene encodes one of the iron-sulfur protein (IP) components of mitochondrial NADH ubiquinone oxidoreductase (complex I) **NR1H3:** NR1 family members are key regulators of macrophage function, controlling transcriptional programs involved in lipid homeostasis and inflammation. A nuclear receptor that regulates lipid homeostasis and inflammation and may play a role in pancreatic beta cell proliferation. NR1 family members are key regulators of macrophage function, controlling transcriptional programs involved in lipid homeostasis and inflammation. Codes for the Liver X receptor alpha protein. The liver X receptors are key regulators of macrophage function, controlling transcription involved in lipid homeostasis and inflammation. **SPI1:** transcription factor which is a crucial regulator of hematopoiesis, and its expression is altered in various leukemic processes. **AGBL2** interacts with RRARES1 to regulate the tubulin tyrosination cycle in which AGBL2 acts as a α-tubulin tyrosine carboxypeptidase. Along with RARRES1, AGBL2 also regulates

						proliferation and differentiation of tubulin in adipose-derived mesenchymal stem cells. Increased amounts of detyrosinated α-tubulin create a more stable microtubule. **C1QTNF4**: potential cytokine that stimulates the STAT3 and NF-κB pathways and promotes cell survival in human cancer cells CELF1:may play a role in myotonic dystrophy type 1 (DM1) via interactions with the dystrophia myotonica-protein kinase (DMPK) gene. **FAM180B:** Membrane protein **MADD:** Interacts with TNFalpha receptor 1 to activate MAPK. Genetic variants for proinsulin to insulin conversion. Expression of MADD activates ERK2 signaling. Loss of MADD induces apoptosis in cell lines.
rs11231693	11	64095140	MACROD1-VEGFB	WHRadjBMI	Shungin	**MACROD1,** MACRO domain containing 1, **VEGFB,** vascular endothelial growth factor B Macrodomains are known to bind ADP-ribose derivatives. Also known as LRP16, MACROD1 was found to play a role in estrogen signaling by interacting with estrogen receptor alpha and enhancing the receptor's transcriptional activity. It has also been found to bind to the androgen receptor via its macro domain and amplifies the transactivation of androgen receptor in response to

						androgen. LRP16 regulated insulin content and glucose-stimulated insulin secretion in MIN6 cells, and overexpression of this gene protected MIN6 cells from fatty acid-induced apoptosis. Diabetic db/db Vegfb knockout mice had ectopic lipid deposition, increased muscle glucose uptake and maintained normoglycemia, and treatment of db/db mice with a VEGF-B antibody enhanced glucose tolerance, preserved pancreatic islet architecture, improved β -cell function and improved dyslipidemia. The index SNP is located ~6 kb from FLRT1, fibronectin leucine rich transmembrane protein 1, involved in cell adhesion and fibroblast growth factor mediated signaling.
rs12286929	11	115151684	CADM1	BMI	Locke	**CADM1:** Cadm1 can specifically suppress metastasis without affecting primary tumor growth. Synapse assembly and axon growth
rs10842707	12	26318431	ITPR2-SSPN	WHRadjBMI	Heid	**SSPN** encodes sarcospan, a member of the dystrophin-glycoprotein complex, which links the subsarcolemmal cytoskeleton and the extracellular matrix of muscle cells. **ITPR2** is inositol 1,4,5-triphosphate receptor, 36 type 2, an intracellular calcium release channel. Mice lacking both Itpr2 and Itpr3 exhibited impaired calcium signaling, secretion

						defects, hypoglycemia and lean body type.
rs7138803	12	49853685	BCDIN3D- FAIM2	BMI	Locke	**BCDIN3D** depletion leads to lower pre-miR-145 and concomitantly increased mature miR-145 levels in breast cancer cells, which suppresses their tumorigenic phenotypes FAIM2:Role in cerebellar development by affecting cerebellar size, internal granular layer (IGL) thickness, and Purkinje cell (PC) development
rs7138803	12	49853685	LOC144233	BMI tails	Berndt	Unknown function
rs1443512	12	53948900	HOXC13	WHRadjBMI	Heid	The index SNP is located between **HOXC13** and **HOXC12** of the HOXC gene cluster. This cluster encodes homeobox transcription factors important for the spatial distribution of cells during embryonic development. rs1443512 is also near to noncoding RNA HOTAIR which has been shown to repress transcription of the HOXD gene cluster in trans.
rs11057405	12	122297350	CLIP1	BMI	Locke	**CLIP1** is a regulator of microtubule dynamics. Axon formation and neuronal polarization. CLIP1 is necessary for cell spreading and phagocytosis of active macrophages in mice. **Other notable genes: DIABLO:** This gene encodes an inhibitor of apoptosis protein (IAP)-binding protein. The encoded mitochondrial protein enters the

						cytosol when cells undergo apoptosis, and it moderates the caspase inhibition of IAPs. **KNTC1:** Essential component of the mitotic checkpoint, which prevents cells from prematurely exiting mitosis. Required for the assembly of the dynein-dynactin and MAD1-MAD2 complexes onto kinetochores. **VPS33A** is part of the class C VPS proteins that form a molecular complex necessary for the joining of endosomes to the lysosomes.
rs4765219	12	123955563	CCDC92	WHRadjBMI	Shungin	**CCDC92,** coiled-coil domain containing 92 protein The closest genes to the index variant are not obvious candidate genes. CCDC92 encodes a protein with unknown function that was found to be upregulated in human B lymphoblastoid cells treated with a polychlorinated biphenyl pollutant. DNAH10 encodes dynein, axonemal, heavy chain 10, which may a play a role in cilia or flagella. ZNF664 encodes zinc finger protein 664; coding variants in ZNF664 have been implicated in myopia
rs12016871	13	27443645	MTIF3- GTF3A	BMI	Locke	**GTF3A:** Transcription factor. **MTIF3:** This gene encodes a translation initiation factor that is involved in mitochondrial protein synthesis.
rs12429545	13	53528071	OLFM4	BMI	Locke	**OLFM4**: Antiapoptotic factor that has a role in tumorigenesis (pancreatic,

						cervical and myeloid leukemia). Associated with childhood obesity. Anti-apoptotic factor induces cell differentiation through retinoic acid. Can decrease innate immunity to H. pylori, a risk factor for obesity
rs9540493	13	65631572	MIR548X2- PCDH9	BMI	Locke	**MIR548X2**: miRNA s are noncoding RNA s involved in post-translational regulation of gene expression. **PCDH9:** cadherin-related neuronal receptor that localizes to synaptic junctions
rs1441264	13	79006784	MIR548A2	BMI	Locke	**MIR548A2:** miRNA s are noncoding RNA s involved in post-translational regulation of gene expression.
rs693839	13	80384153	SPRY2	BF%	Lu	While little is known about **SPRY2**, the Spry1 homolog in mice has been implicated in adipose tissue differentiation. Drosophila experiments pinpoint the SPRY2 (or sty) as the potential causal gene within the locus; that is, knockdown flies for sty have significantly lower whole-body TG levels than wild-type flies.
rs10132280	14	25458973	STXBP6	BMI	Locke	**STXBP6**: participate in neuronal neuromediator release, SNARE complex formation. STXBP6 helps to regulate the exocytosis of catecholamine, GH1 protein, norepinephrine.
rs12885454	14	29267632	PRKD1	BMI	Locke	**FOXG1**:Survival of post mitotic neurons

rs11847697	14	30045906	PRKD1	BMI	Locke	**PRKD1**: Crucial to support growth of tumor cells PRKD1:serine/threonine kinase that regulates membrane receptors, implicated in skeletal muscle remodeling
rs7141420	14	79433111	NRXN3	BMI	Locke	**NRXN3:** Drive synapse formation in vitro and control the differentiation of nascent synapses into either excitatory or inhibitory fully mature nerve terminals.
rs8042543	15	31416060	KLF13	WHRadjBMI	Shungin	**KLF13**, Kruppel-like factor 13 KLF13 encodes Kruppel-like factor 13, which belongs to the Sp1-like family of transcription factors that contain 3 C-terminal zinc finger DNA-binding domains, and bind to GC-rich sequences. KLF13 is a regulator of heart development, and was also found to bind and repress the low density lipoprotein receptor promoter. A nearby gene, OTUD7A, belongs to a deubiquitinating enzyme subfamily characterized by an ovarian tumor (OTU) domain. This gene encodes a protease that cleaves ubiquitin linkages.
rs3736485	15	51456413	SCG3- DMXL	BMI	Locke	**DMXL2** regulates Ca(2+)-dependent exocytosis of neurotransmitter, concentrated on synaptic vesicles at synapses. DMXL2 is necessary for the exocytosis of neurotransmitters through the synaptic vesicles. It is abundantly expressed in the brain. Little of the exact function of this

						gene is known. DMXL2 is required for Notch signaling in mammals. **GLDN:** Adhesion molecule important for the formation of the nodes of Ranvier. Rat models of Guillain-Barre syndrome have auto-antibodies to GLDN. **GLDN:** Expressed by myelinating Schwann cells, required for the formation of peripheral nodes of Ranvier SCG3:Hypothalamus expression, binds to POMC; Neuroendocrine function; potential regulator of food intake. A neuroendocrine secretory protein that forms a granule-like structure together with appetite-related neuropeptides SCG3:binds to POMC;may be a potential regulator of food intake based on its capacity to accumulate appetite-related hormones into secretory granules.
rs8030605	15	56212400	RFX7	WHRadjBMI	Shungin	**RFX7,** regulatory factor X, 7 RFX7 encodes a member of the regulatory factor X family of transcription factors. It is a winged-helix transcription factor and contains a well-conserved RFX DNA binding domain. It has high ubiquitous expression, particularly in brain. TEX9, encoding testis-expressed sequence 9, is poorly described. Another nearby gene, NEDD4, encodes neural precursor cell expressed, developmentally down-regulated 4,

						an E3 ligase. Overexpression of Nedd4 suppressed BMP-induced osteoblast transdifferentiation process of mouse premyoblast C2C12 cells, and NEDD4 was also found to be an important modulator of phospho-Smad1 in both BMP-2 and TGF-β1 action
rs1440372	15	66740813	SMAD6	WHRadjBMI	Shungin	**SMAD6,** SMAD family member 6 SMAD6 belongs to the SMAD family of proteins, which are related to Drosophila 'mother's against decapentaplegic' and C elegans Sma. SMAD proteins are signal transducers of the TGF-β superfamily and are involved in cell growth, morphogenesis, development and immune responses. SMAD6 inhibits the Bone morphogenetic protein/Smad1 signaling pathway. 3T3-F442A mouse pre-adipocytes overexpressing Smad6 show increased TGF-β signaling and decreased adipocyte differentiation.
rs16951275	15	67784830	MAP2K5- LBXCOR1	BMI	Locke	**LBXCOR1** is expressed broadly in neuronal precursors, show expression restricted to large granule cell layer interneurons in the adult. Suppresses mainly the BMP signaling pathway, and has been implicated in restless legs syndrome and obesity. MAP2K5:The signal cascade mediated by this MAP kinase is involved in growth factor stimulated cell

						proliferation and muscle cell differentiation. **MAP2K5:** signal cascade mediated by MAP2K5 is involved in growth factor stimulated cell proliferation and muscle cell differentiation
rs7164727	15	72801650	LOC100287559- BBS	BMI	Locke	**BBS4** localizes to cellular structures associated with motile cilia. Mediate LepR signaling in the hypothalamus and BBS4(-/-) mice are resistant to the action of leptin to reduce body weight and food intake regardless of serum leptin levels and obesity. One of 7 BBS proteins that form a complex for ciliogenesis. Mutations cause Bardet-Biedl syndrome of which obesity is a common feature. Required for leptin receptor (LepR) signaling in the hypothalamus, Obesity, decreased POMC expression. **LOC100287559:** ADPGK antisense **RNA1ADPGK:** This enzymye catalyzes the ADP-dependent phosphorylation of glucose to glucose-6-phosphate and may play a role in glycolysis.
rs758747	16	3577357	NLRC3	BMI	Locke	**NLRC3:** Modulates T-cell activation through negative regulation of NFKB transcription factor activity. SLX4:required for DNA interstrand crosslink repair in response to replication fork failure. encodes a structure-specific endonuclease subunit that is a critical scaffold element for the assembly of a

						multiprotein complex involved in DNA replication and repair. **Other notable genes:** CLUAP1: Highly expressed in numerous cancers.
rs12446632	16	19924067	GPRC5B- IQCK	BMI	Locke	**GPRC5B:** Strongly expressed in hippocampus and cerebellum. Specific function is unknown, but may play a role in neurite outgrowth through interaction in the Wnt pathway. GPCR containing a signal peptide and 7 transmembrane helices. **IQCK:** IQ domain (uninterrupted seven-turn α-helix that is distinctly amphiphilic) conatining protein
rs2650492	16	28322090	SBK1- APOB	BMI	Locke	**APOBR:** Apolipoprotein B48 receptor is a macrophage receptor that binds to the apolipoprotein B48 of dietary triglyceride (TG)-rich lipoproteins. In case of elevated plasma triglyceride, the apolipoprotein B48 receptor may contribute to foam cell formation, endothelial dysfunction, and atherothrombogenesis. **SBK1**: Human SBK1 is dysregulated in multiple cancers and promotes survival of ovary cancer SK-OV-3 cells.
rs3888190	16	28878165	SH2B1- APOB48R- ATXN2L- SBK1- SULT1A2- TUFM	BMI, BF%	Locke	**SH2B1:** Substrate for JAK2, involved in Growth Hormone signaling. Adapter protein for members of the tyrosine kinase receptor family. Disrupted in patients with a syndrome is characterized by obesity and developmental delay. **ATP2A1:**

						part of (Ca2+) pump in the sarcoplasmic or endoplasmic reticula of muscle cells, mutations cause forms of Brody disease. **ATXN2L**:This protein is a member of the spinocerebellar ataxia (SCAs) family, which is associated with a complex group of neurodegenerative disorders. **SULT1A2:** Encode phenol sulfotransferases with thermostable enzyme activity. SULT1 enzymes catalyze the sulfate conjugation of many endogenous and exogenous compounds, including estrogens and their oxidative metabolites and thyroid hormones. TUFM: protein translation in mitochondria. Mutations in this gene have been associated with combined oxidative phosphorylation deficiency resulting in lactic acidosis and fatal encephalopathy.

rs4787491	16	30004016	MAPK3- KCTD13- INO80E-TAOK2-YPEL3- DOC2A-FAM57B	BMI	Locke	**MAPK3:** The protein encoded by this gene is a member of the MAP kinase family. MAP kinases, also known as extracellular signal-regulated kinases (ERKs), act in a signaling cascade that regulates various cellular processes such as proliferation, differentiation, and cell cycle progression in response to a variety of extracellular signals. This kinase is activated by upstream kinases, resulting in its translocation to the nucleus where it phosphorylates nuclear targets. **TAOK2:** Serine/threonine-protein kinase involved in different processes such as membrane blebbing and apoptotic bodies formation DNA damage response and MAPK14/p38 MAPK stress-activated MAPK cascade. Phosphorylates itself, MBP, activated MAPK8, MAP2K3, MAP2K6 and tubulins. Activates the MAPK14/p38 MAPK signaling pathway through the specific activation and phosphorylation of the upstream MAP2K3 and MAP2K6 kinases. Serine/threonine-protein kinase involved in different processes such as membrane blebbing and apoptotic bodies formation DNA damage response and MAPK14/p38 MAPK stress-activated MAPK cascade. Phosphorylates itself, MBP, activated MAPK8, MAP2K3, MAP2K6 and

						tubulins. Activates the MAPK14/p38 MAPK signaling pathway through the specific activation and phosphorylation of the upstream MAP2K3 and MAP2K6 kinases. In response to DNA damage, involved in the G2/M transition DNA damage checkpoint by activating the p38/MAPK14 stress-activated MAPK cascade, probably by mediating phosphorylation of upstream MAP2K3 and MAP2K6 kinases. **YPEL3:** growth suppressive protein downregulated by estrogen in estrogen receptor positive breast cancer cell lines. ALDOA**:** The protein encoded by this gene, Aldolase A (fructose-bisphosphate aldolase), is a glycolytic enzyme that catalyzes the reversible conversion of fructose-1,6-bisphosphate to glyceraldehyde 3-phosphate and dihydroxyacetone phosphate. Aldolase A is found in the developing embryo and is produced in even greater amounts in adult muscle. Defects in ALDOA are the cause of glycogen storage disease type 12. It may lead to myopathy with exercise intolerance and rhabdomyolysis BOLA2:Cell proliferation and cell-cell regulation BOLA2B:Cell proliferation and cell-cell regulation DOC2A:DOC2A is mainly expressed in brain and is suggested to

						be involved in Ca(2+)-dependent neurotransmitter release. **FAM57B:** peroxisome proliferator-activated receptor γ (PPARγ)-responsive transmembrane gene that is related to obesity. FAM57B overexpression inhibited adipogenesis, and siRNA-mediated knockdown promoted adipocyte differentiation. INO80E:part of the chromatin remodeling complex INO80, activates transcription KCTD13:KCTD13 is a major driver for the neurodevelopmental phenotypes associated with the 16p11.2 CNV

rs9925964	16	31118574	KAT8-ZNF64- VKORC- ZNF66- STX1-FBXL1	BMI	Locke	**FBXL19:** This gene encodes a member of the Skp1-Cullin-F-box family of E3 ubiquitin ligases. The encoded protein binds to the transmembrane receptor ST2L and regulates its ubiquination and degradation. This protein has been linked to the regulation of pulmonary inflammation and psoriasis. **KAT8:** acetylates nucleosomal histone H4, and is critical for DNA double-strand break repair **STX1B:** directly implicated in the process of calcium-dependent synaptic transmission in rat brain. The expression of this protein is transiently induced by long-term potentiation of synaptic responses in the rat hippocampus. The protein may play an important role in the excitatory pathway of synaptic transmission, which is known to be implicated in several neurologic diseases. STX4:Functions in plasma membrane t-SNARE that mediates docking of transport vesicles. Necessary for the translocation of SLC2A4 from intracellular vesicles to the plasma membrane. Together with STXB3 and VAMP2, may also play a role in docking/fusion of intracellular GLUT4-containing vesicles with the cell surface in adipocytes (By similarity). May also play a role in docking of synaptic vesicles at

						presynaptic active zones VKORC1:mutations in this gene can be associated with deficiencies in vitamin-K-dependent clotting factors and warfarin resistance. **ZNF646**: Zinc finger protein, transcription regulation. **ZNF668** is a nucleolar protein that physically interacts with and regulates p53 and its negative regulator MDM2. BCKDK: regulate pre-mRNA alternative splicing, may play a role in myotonic dystrophy type 1 BCKDK:Codes for an enzyme that plays a major role in the BCKDH complex, which controls the breakdown and synthesis of the branched-chain amino acids, leucine, isoleucine, and valine
rs2080454	16	49028679	CBLN1	BMI	Locke	CBLN1:bi-functional ligand bridging pre-synaptic β-neurexins on granule cells to post-synaptic Grid2 on Purkinje neurons.
rs1558902	16	53769662	FTO	BMI, SAT, BF%, BMI tails	Locke	FTO:Expression hypothalamus, functionally involved in energy homeostasis FTO:iron and 2-oxoglutarate-dependent oxygenase, strong association with BMI, obesity risk, and type 2 diabetes
rs2925979	16	81501185	CMIP	WHRadjBMI	Shungin	**CMIP**, c-MAF inducing protein This gene encodes C-maf inducing protein, which interacts with phosphatidylinositol 3-kinase complex and plays a role in ERK signaling. CMIP is expressed in

						peripheral blood mononuclear cells, kidney, fetal liver, and adult brain and liver. A nearby gene, PLCG2, encodes phospholipase C, gamma 2 (phosphatidylinositol-specific), which hydrolyzes phosphatidyl inositol 4,5-biphosphate (PIP) to inositol- 1,4,5-triphosphate (IP), resulting in an increase in intracellular calcium levels
rs9914578	17	2101842	SMG6- N29617	BMI	Locke	**SMG6**: associates with active telomerase andparticipates in nonsense-mediated mRNA decay SRR:Synthesizes D-Serine from L-serine that activates NMDA receptors thus implying a role in neurotransmission. **SRR:**Expressed in pyramidal neurons in the cerebral cortex and the hippocampus
rs1000940	17	5379957	RABEP1	BMI	Locke	**RABEP1** is involved in the development and fusion of the early endosomes, and plays a major role in the intracellular trafficking and recycling. Localizes to early endosomes to induce Rab5 to activate endocytic pathway. Selectively cleaved during apoptosis RPAIN: and transports replication protein A into nuclear bodies. Part of the **MIS12** complex which is required for normal chromosome alignment and segregation and for kinetochore formation during mitosis **NUP88:** Nuclear pore complex component. Associated with cancers. NUP88:The

						protein encoded by NUP88 is a member of the nucleoporin gene family of molecules that act as gateways for macromolecules (proteins and nucleic acids) into and out of the cellular nucleus.
rs4646404	17	17516885	PEMT	WHRadjBMI	Shungin	**PEMT,** phosphatidylethanolamine N-methyltransferase This gene encodes a liver enzyme that converts phosphatidylethanolamine to the phospholipid phosphatidylcholine by methylation in the liver. The protein localizes to the endoplasmic reticulum and mitochondria-associated membranes. Pemt knockout mice on a high fat diet show adipocyte hypotrophy. Pemt mRNA and protein increase upon adipocyte differentiation in 3T3-L1 cells.
rs9906944	17	49014058	IGF2BP1	BF%	Lu	SNPs in ***IGF2BP1***, in linkage disequilibrium (LD) with rs9906944, have been previously implicated with primary tooth development in infancy. Consistently, the BF% increasing allele of *IGF2BP1* (rs9906944) showed association with a later eruption of the first tooth and reduced number of teeth at 1 year. Even though this suggests a role in maturation, we found no evidence for association with pre-pubertal height or pubertal growth and timing or age at menarche. Although this locus harbours a number of genes, data in

						rodents suggest that *IGF2BP1* might be a potential candidate gene driving the associations observed here, as Igf2bp1 knockout mice demonstrate fetal and postnatal growth retardation
rs8066985	17	70457204	KCNJ2	WHRadjBMI	Shungin	**KCNJ2,** potassium inwardly-rectifying channel, subfamily J, member 2 Inwardly rectifying K+ channels control the resting K+ conductance and stabilize the resting potential in many cells. KCNJ2 was upregulated during myoblast differentiation into skeletal muscleand was expressed in smooth muscleand cardiomyocytes
rs12940622	17	80641771	RPTOR	BMI	Locke	**RPTOR:** Regulatory protein of mTOR (mammalian target of rapamycin) inolved in cell growth in response to nutrient and insulin levels. Substrate for MAPK, regulating cell growth in response to nutrient and insulin levels
rs1808579	18	23524924	NPC1- C18orf8	BMI	Locke	**C18orf8:** Colon cancer associated protein. **NPC1** encodes a protein responsible for maintaining intracellular cholesterol homeostasis though binding and transport of both simple fatty acids and cholesterol to and from the endosome/lysosome compartment. NPC1 is active in late endosome transport and fusion. Mutations in this gene have been shown to cause Niemann-Pick C1 (NPC1) disease, which is characterized by accumulation of cholesterol in the

						lysosomes. NPC1:Transports low-density lipoproteins to late endosomal/lysosomal compartments where they are hydrolized and released as free cholesterol. NPC1:Defects in this gene cause autosomal recessive neurodegenerative disorder RIOK3:alters cytoskeletal architecture and promotes pancreatic ductal cell migration and invasion
rs7239883	18	42567706	LOC284260- RIT2	BMI	Locke	**LOC284260:** long intergenic non-protein coding RNA 907. **RIT2** is part of the RAS superfamily of proteins that assist in regulating many biological processes through signal transduction. RIT2 has been shown to increase the outgrowth of neurites and is most highly expressed in the brain, but found throughout the body. RIT2 is important for the intracellular transport of dopamine. RIT2:selective integration of MAP kinase signaling leading to neuronal development RIT2:essential contribution to nerve growth factor (NGF)-mediated neuronal differentiation
rs7243357	18	59216087	GRP	BMI	Locke	**GRP:** Activates MAPK/ERK1/2 GRP:found in axons in the mediobasal hypothalamus and may also be released from the gut to signal the brain

rs6567160	18	60161902	MC4R	BMI, BF%, BMI tails	Locke	**MC4R:** Encodes a membrane-bound receptor from the melanocortin receptor family. Defects in this gene are a cause of autosomal dominant obesity. A membrane-bound receptor that affects appetite regulation, energy intake, energy expenditure, and satiety. G-protein coupled receptor, Missense mutant unable to generate cyclic AMP in response to ligand. Expressed in the hypothalamus. Main receptor for POMC-derived peptides (alpha-MSH) to regulate appetite. Critical for nicotine-induced appetite suppression. Autosomal dominant mutations in childhood and adult obesity.
rs12454712	18	63178651	BCL2	WHRadjBMI	Shungin	**BCL2,** B-cell CLL/lymphoma 2 B-cell CLL/lymphoma 2 encodes an anti-apoptotic protein that binds the BH3 domain of pro-apoptotic factors and regulates permeabilization of the outer mitochondrial membrane, a critical step in apoptosis. Bcl2 was upregulated and apoptosis was reduced in rat pancreatic beta-cells treated with leptin
rs12608504	19	18278325	JUND	WHRadjBMI	Shungin	**JUND,** jun D proto-oncogene JUND is a component of the Activating protein 1 transcription factor; AP-1 is a dimeric transcription factor with basic leucine zipper domains. JunD dimerizes with DeltaFosB and binds to

						the IL-11 gene promoter. Suppression of osteoblast differentiation by aging involved decreased JunD binding to the IL-11 promoter and reduced IL-11 transcription. IL-11 inhibits the accumulation of adipose in human long-term bone marrow culture stromal layers. Other nearby genes include KIAA1683, LSM4 PIK3R2, PDE4C, and miR3188.
rs17724992	19	18344015	GDF15- PGPEP	BMI	Locke	**GDF15:** Tissue regeneration and maintenance. Encodes a bone morphogenetic protein that regulates tissue differentiation and maintenance; the protein has been associated with CVD and cancer. **PGPEP1:** catalyzes the hydrolysis of N-terminal pyroglutamyl residues from oligopeptides and proteins
rs757318	19	18709498	CRTC1	BF%	Lu	**CRTC1** is primarily expressed in the brain, and it may affect leptin anorexic effect in the hypothalamus. CRTC knockout mice demonstrated hyperphagia, increased white adipose tissue and infertility.
rs4081724	19	33334040	CEBPA	WHRadjBMI	Shungin	**CEBPA,** CCAAT/enhancer binding protein alpha C/EBP alpha is a basic leucine zipper transcription factor that is highly expressed in liver and adipose tissue, and is required for differentiation of white adipose tissue. C/ebp alpha knockout mice have defects in gluconeogenesis, are hypoglycemic, and die shortly after

						birth. Additionally, C/EBP alpha also binds to the leptin promoter, a gene that plays an important role in body weight homeostasis. Other nearby genes include C/EBPG, encoding C/EBP gamma, which forms heterodimers with C/EBP beta, and PEPD, encoding peptidase D
rs29941	19	33818627	KCTD15	BMI	Locke	**KCTD15:** Kctd15 inhibits neural crest formation by attenuating the output of the canonical Wnt pathway. Inhibits neural crest formation. Linked to Wnt pathway through WNT8.
rs2075650	19	44892362	TOMM40- APOE- APOC1	BMI, BF%	Locke	**APOC1:** Inhibits both hepatic lipase and lipoprotein lipase in vitro, and is a physiological inhibitor of CETP through modiication of properties of circulating HDL. **APOE:** Involved in the clearance of chylomicron and VLDL remnants. A ligand for receptors that bind, internalize, and catabolize lipoprotein particles. Aβ amyloidosis. **PVRL2:** Encodes a membrane glycoprotein that serves as an entry for certain mutant strains of herpes simplex virus and pseudorabies virus. PVRL2:Present in adherens junctions. Entry point for viruses. **TOMM40:** Channel-forming protein essential for import of protein precursors into mitochondria. Important for mitochondrial organization, found in the mitochondrial membrane and part of the TOM (translocase of the

						mitochondrial membrane complex). Brain and cognitive changes suggestive of Alzheimer's Disease (late onset) may be associated with TOMM40 polymorphism. Channel forming subunit of the mitochondrial membrane. Immunodepletion causes loss of peptide import activity into mitochondria.
rs2287019	19	45698914	QPCTL- GIPR	BMI	Locke	**GIPR:** Variety of brain functions such as neuro-modulation, neurogenesis, brain development. A receptor for gastric inhibitory polypeptide, which stimulates insulin release in the presence of elevated glucose. GPCR for gastric inhibitory polypeptide (GIP), pathogenesis of diabetes. **QPCTL:** Locus associated with BMI and increased insulinogenic index
rs3810291	19	47065746	ZC3H4	BMI	Locke	**NPAS1:** Central nervous system development. **TMEM160:** Mitochondrial membrane protein. Interacts with HIV-1 in cell culture ZC3H4:contains nucleic acid binding zinc finger domain
rs979012	20	6642727	BMP2	WHRadjBMI	Shungi n	**BMP2,** bone morphogenetic protein 2 BMP2 belongs to the transforming growth factor beta (TGF-β) superfamily of genes. BMPs signal through transmembrane serine/threonine kinase receptors and stimulate Smad, MAPK and Akt signaling pathways. High levels of BMP2 induce chondrogenesis or

						osteogenesis, while low levels of BMP2 promote adipogenesis. BMP2 stimulates commitment of C3H10T1/2 pluripotent stem cells into adipocytes. BMP2, along with IGF-1, induces differentiation of adipose-derived mesenchymal stem cells into cartilage cells
rs224333	20	35436182	GDF5	WHRadjBMI	Shungin	**GDF5**, growth differentiation factor 5 GDF5 is a member of the bone morphogenetic protein BMP family and the transforming growth factor-beta superfamily. GDF5 promoted osteogenic differentiation of rat fat-derived stromal cells and may promote angiogenic activity of stromal cells by increasing vascular endothelial growth factor gene expression in vitro. GDF5 also induced chondrogenesis in rat adipose-derived stem cells. Human mesenchymal stem cells that overexpressed GDF5 displayed osteogenic differentiation. UQCC is a nearby gene, which encodes ubiquinol-cytochrome c reductase complex chaperone, a ZIC-binding protein repressed by basic fibroblast growth factor
rs6090583	20	46930192	EYA2	WHRadjBMI	Shungin	**EYA2**, eyes absent homolog 2. This gene encodes a member of the eyes absent, EYA, family of proteins. EYA2 is a transcriptional coactivator and protein phosphatase. Eya2 acts

						synergistically with both Dach2 and Six1 to regulate myogenic differentiation and development. Eya2 also prevents adverse cardiac remodeling under pressure overload. Nearby, **SLC2A10** encodes solute carrier family 2 (facilitated glucose transporter) member 10.
rs6091540	20	52471323	ZFP64	BMI	Locke	**ZFP64**: mediates mesenchymal cell differentiation by modulating Notch signaling
rs2836754	21	38919816	ETS2	BMI	Locke	**ETS2:** Transcription factor in stem cell development, senescence, and cell death. Can regulate p16 expression to prevent senescence. MAP2K1 inhibition blocks insulin-stimulated phosphorylation of ETS2. PSMG1:Heterodimerizes with PAC2 to aid formation of 20S proteasome. Increased expression in papillary thyroid carcinomas. PSMG1:Deficient gene cause Malformation of cerebrum and cerebellum
rs2294239	22	29053489	ZNRF3	WHRadjBMI	Heid	The index SNP is located within the 3' untranslated region of **ZNRF3**, which encodes zinc and ring finger 3. **KREMEN1** is a kringledomain containing transmembrane protein that functions as a receptor for DKK1, forms a complex with low-density lipoprotein receptor-related protein 6 (LRP6), and regulates Wnt/beta-catenin signaling. KREMEN1 also interacts with R-Spondin 1 (RSPO1).

						Double null Kremen1/Kremen2 mice show increased bone volume compared to wild type. Nearby genes also include C22orf31
rs3761445	22	38199404	PLA2G6-PICK1	BF%	Lu	The BF% increasing allele of the **PLA2G6** locus is associated with lower insulin and TG levels and reduced T2D risk, particularly in men. PLA2G6 is the nearest gene and encodes a calcium-independent phospholipase A2 involved in the hydrolysis of phospholipids. However, this locus harbours a number of other genes that would make plausible candidates for driving the cross-phenotype associations, including **PICK1**, which is membrane sculpting BAR domain protein. PICK1-deficient mice and flies display marked growth retardation, which at least in mice, might be due to impaired storage and secretion of growth hormone from the pituitary and possibly insulin from the pancreas. PICK1-deficient mice, despite their smaller size, demonstrate increased body fat and reduced lean mass, reduced TG levels and impaired insulin secretion, which was compensated by increased insulin sensitivity.