Web Table 1. Candidate genes and SNPs

SNP ID	Gene Name	Chromosome[a]	Position[a]	RefSNP Alleles[b]	SNP information	MAF[c] (CEU)	Selection Criteria
rs12255372	*TCF7L2*	10	114808902	G/T	Intron	0.21	diabetes-associated
rs7903146	*TCF7L2*	10	114758349	C/T	Intron	0.22	diabetes-associated
rs290487	*TCF7L2*	10	114909731	C/T	Intron	0.27	diabetes-associated
rs10885390	*TCF7L2*	10	114640797	T/A	Intergenic	0.24	diabetes-associated
rs3814573	*TCF7L2*	10	114898093	C/T	Intron	0.40	diabetes-associated
rs660339	*UCP2*	11	73689104	G/A	Missense	0.43	Ala55Val
rs1044498	*ENPP1*	6	132172368	A/C	Missense	0.31	K121Q (Lys173Glu) insulin resistance
rs9939609	*FTO*	16	53820527	T/A	Intron	0.38	obesity, BMI
rs8050136	*FTO*	16	53816275	C/A	Intron	0.37	obesity, BMI
rs1421085	*FTO*	16	53800954	T/C	Intron	0.26	obesity, BMI
rs17817449	*FTO*	16	53813367	T/G	Intron	0.35	obesity, BMI
rs4994	*ADRB3*	8	37823798	T/C	Missense	0.10	Trp64Arg, BMI, overweight, obesity and weight gain
rs1801282	*PPARG*	3	12393125	C/G	Intron	0.06	obesity-associated
rs8192678	*PPARGC1A*	4	23815662	G/A	Missense	0.30	Gly482Ser; obesity/metabolic abnormalities
rs3736265	*PPARGC1A*	4	23814707	G/A G/T	Missense	0.11	Thr612Met Thr612Lys obesity/metabolic abnormalities
rs11760956	*LEP*	7	127891087	G/A	Intron	0.29	tagSNP
rs12706831	*LEP*	7	127887068	T/G	Intron	0.46	tagSNP
rs3828942	*LEP*	7	127894305	G/A	Intron	0.45	tagSNP
rs2071045	*LEP*	7	127892980	T/C	Intron	0.26	tagSNP
rs2167270	*LEP*	7	127881349	G/A	5'utr	0.35	tagSNP
rs11924032	*SLC2A2*	3	170735099	G/A	Intron	0.31	tagSNP
rs6785233	*SLC2A2*	3	170756985	T/G	Intergenic	0.19	tagSNP
rs5400	*SLC2A2*	3	170732300	C/T	Missense	0.21	Thr110Ile

[a] Genomic Build 37.1; Group term GRCh37
[b] RefSNP alleles: reference allele/risk allele(minor allele)
[c] MAF (Minor Allele Frequency) source: 1000 Genomes project

Web Appendix. NBDPS Sample Quality Assurance and Laboratory Proficiency (Approved July 2011)

Pre-Characterized Samples DNA Polymorphism Discovery Resource (PDR)

To ensure genotyping proficiency of laboratories genotyping NBDPS buccal-derived DNA specimens independent of source material, the laboratories annually genotype a standard SNP set on a subset of the Coriell Institute for Medical Research's Polymorphism Discovery Resource (PDR) DNA samples (1). The SNP set is determined by the Genetic Analysis Working Group of the NBDPS and includes SNPs assayed by multiple labs and at least one SNP with publicly available genotypes on Coriell's PDR samples. Standard 96 well plates that include 86 PDR samples, 4 replicates, 2 negative controls, and 4 empty wells for internal laboratory genotyping controls are plated by Coriell and sent directly to each laboratory. Plate formats and content are changed yearly. Laboratories are blinded to sample type and report genotyping results centrally to the Centers for Disease Control and Prevention for analysis. Since implementation of annual external quality assessment, call rates of 97-100% have been reported across NBDPS genotyping labs. Concordance rates of 99-100% have been reported between NBDPS labs and between NBDPS labs and the publicly available Coriell PDR genotypes. Due to higher costs, laboratories using higher throughput genotyping methods do not complete this annual assessment; instead, they complete an assessment using blood-buccal trio samples.

Blood-Buccal Trios

To assure data quality across laboratories genotyping NBDPS buccal-derived DNA specimens, 36 blinded specimens ascertained through the University of Washington under an

IRB-approved protocol and 2 DNA-negative controls are sent to each laboratory annually. The 36 specimens are comprised of paired cytobrush buccal and whole blood derived (gold standard) DNAs from 6 parent-offspring trios. The NBDPS Central Laboratory extracted buccal-derived DNAs using phenol chloroform, whole blood-derived DNAs using Gentra Puregene, quantified DNAs using RNaseP real-time quantitative PCR, and verified Mendelian inheritance using a microsatellite panel. Specimens were genotyped at each NBDPS genotyping laboratory for the standard SNP panel. Due to higher costs, laboratories using higher throughput genotyping methods complete this assessment using 6 blinded specimens from 1 parent-offspring trio prior to initiating each project. Since implementation of external quality assessment, call rates of 92-100% have been reported across NBDPS genotyping labs. Concordance rates between paired blood-buccal samples were 100% pre and post WGA. Inter-laboratory concordance rates for variants assayed in common were 100%. Additionally, SNP genotyping was consistent with Mendelian inheritance.

REFERENCES

1. Collins FS, Brooks LD, Chakravarti A. A DNA polymorphism discovery resource for research on human genetic variation. *Genome Res*. 1998;8(12):1229-1231.