The American Journal of Human Genetics, Volume *94*

Supplemental Data

Variant Association Tools for Quality Control and Analysis of Large-Scale Sequence and Genotyping Array Data

Gao T. Wang, Bo Peng and Suzanne M. Leal

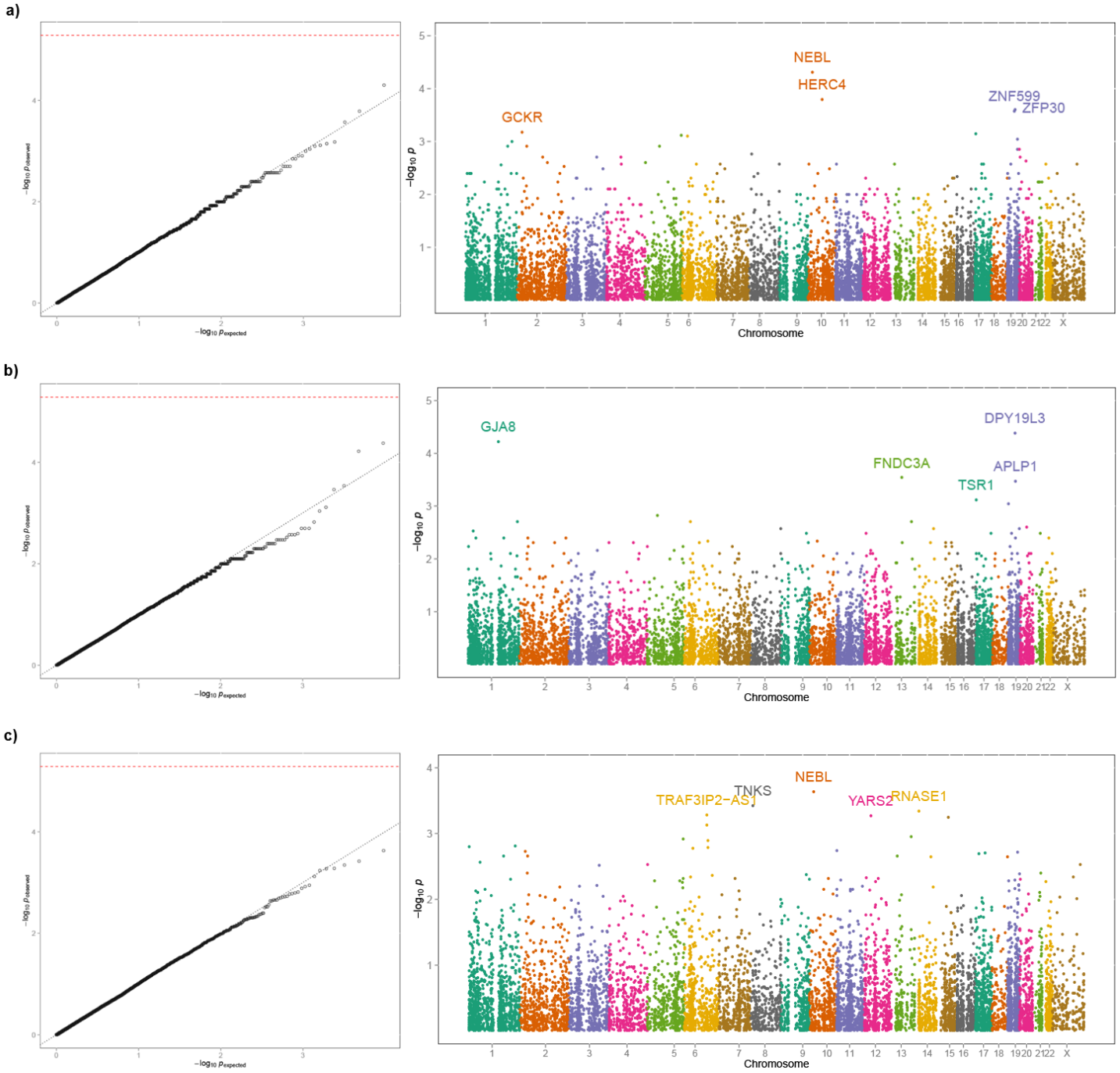

Figure S1: Rare variant association analysis for simulated BMI phenotype using the BRV method. Analysis of whole exome association was performed for the quantile normal transformed BMI phenotype using the BRV method. For panels a to c results are represented by p-values at $-\log_{10}$ scale and displayed in quantile-quantile (QQ) (left) and Manhattan (right) plots. Analysis was performed for Europeans (panel a), Asians (panel b) and Meta-analysis of European and Asian results (panel c) using sample-size based method.

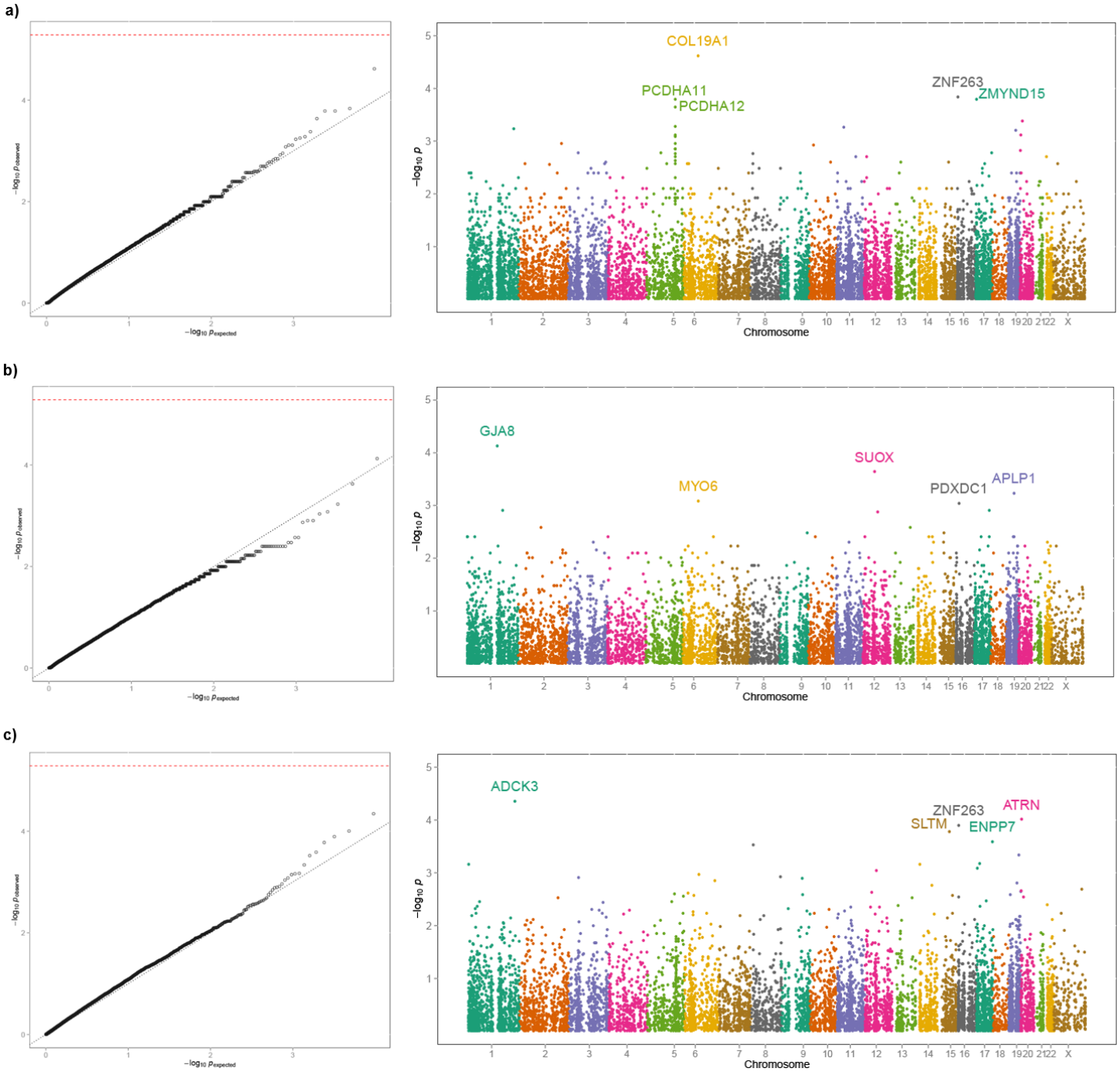

Figure S2: Rare variant association analysis for simulated BMI phenotype using the VT method. Analysis of whole exome association was performed for the quantile normal transformed BMI phenotype using the VT method. For panels a to c results are represented by p-values at $-\log_{10}$ scale and displayed in quantile-quantile (QQ) (left) and Manhattan (right) plots. Analysis was performed for Europeans (panel a), Asians (panel b) and Meta-analysis of European and Asian results (panel c) using sample-size based method.

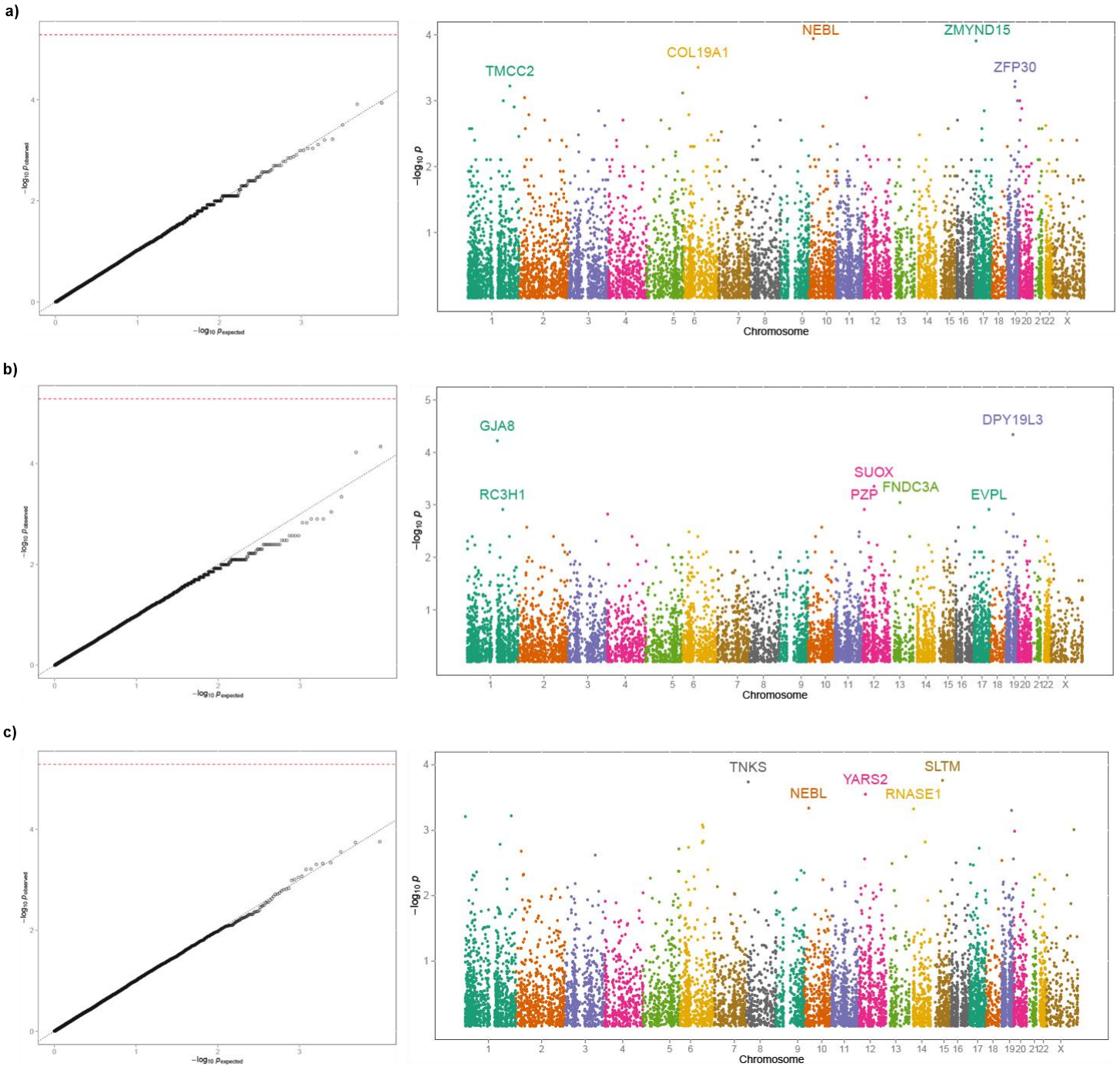

Figure S3: Rare variant association analysis for simulated BMI phenotype using the WSS method. Analysis of whole exome association was performed for the quantile normal transformed BMI phenotype using the WSS method. For panels a to c results are represented by p-values at $-\log_{10}$ scale and displayed in quantile-quantile (QQ) (left) and Manhattan (right) plots. Analysis was performed for Europeans (panel a), Asians (panel b) and Meta-analysis of European and Asian results (panel c) using sample-size based method.

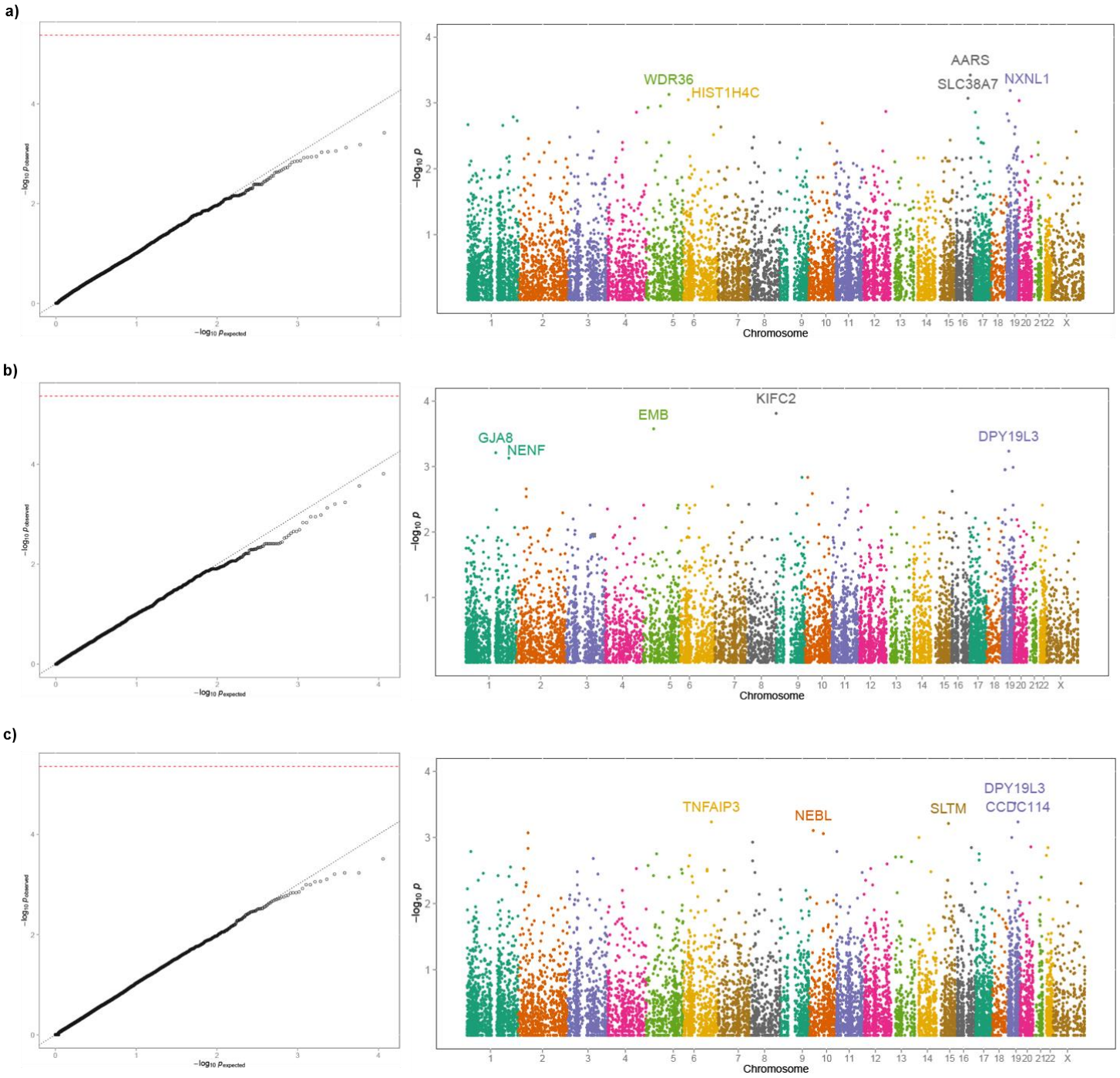

Figure S4: Rare variant association analysis for simulated BMI phenotype using the SKAT method. Analysis of whole exome association was performed for the quantile normal transformed BMI phenotype using the SKAT method. For panels a to c results are represented by p-values at $-\log_{10}$ scale and displayed in quantile-quantile (QQ) (left) and Manhattan (right) plots. Analysis was performed for Europeans (panel a), Asians (panel b) and Meta-analysis of European and Asian results (panel c) using metaSKAT method.

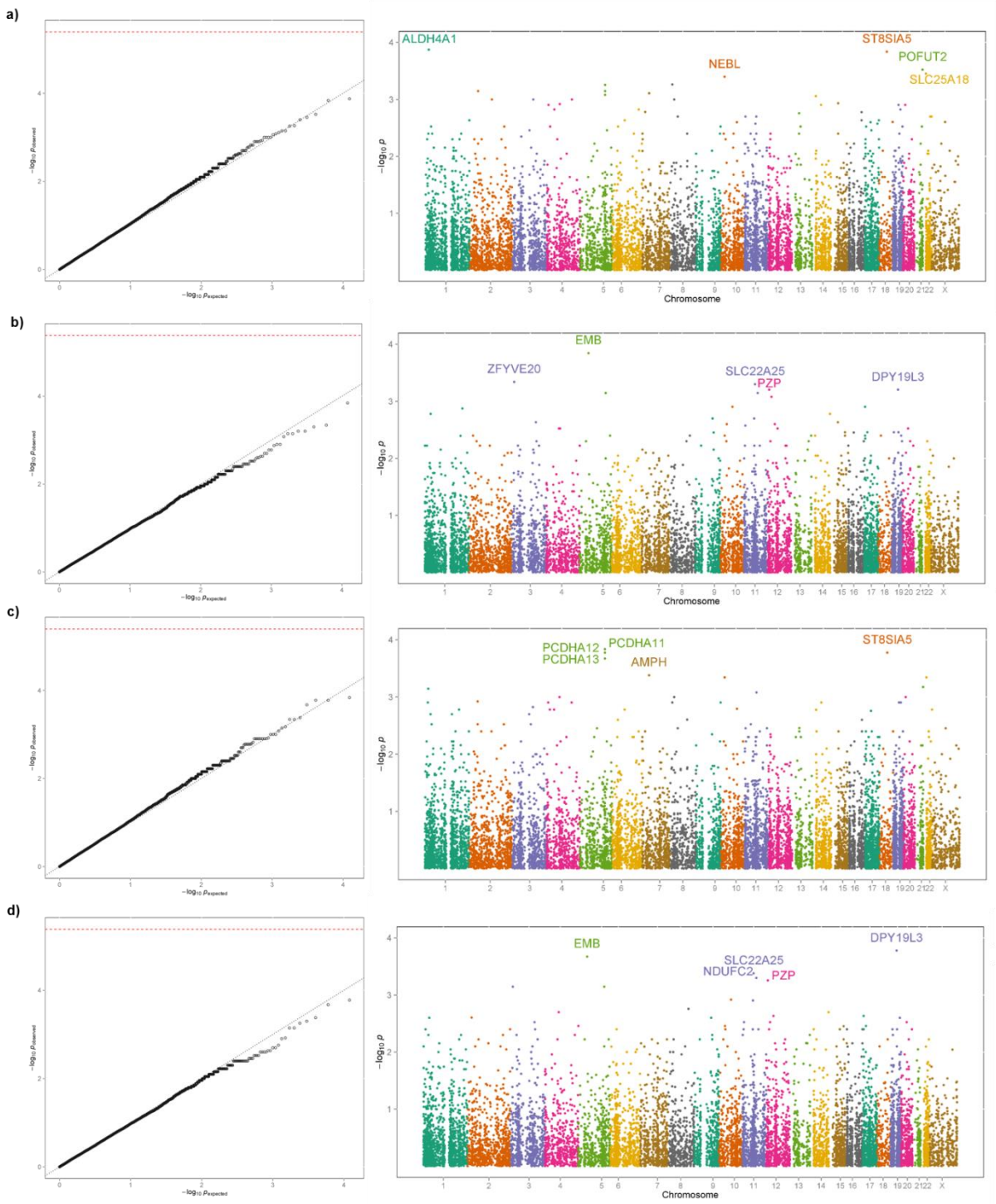

Figure S5: Rare variant association analysis for simulated BMI phenotype using *VAT stacking*. Analysis of whole exome association was performed for the quantile normal transformed BMI phenotype using KBAC and KBAC stacked on the VT algorithm, which allows for performing the KBAC method maximizing the test over allele frequencies. For KBAC variants with a MAF <1% were analyzed while for KBAC stacked on the VT algorithm variants with a MAF <5% were analyzed. Results are shown for KBAC analyses for Europeans (panel a) and Asians (panel b), as well as KBAC stacked on the VT algorithm for Europeans (panel c) and Asians (panel d). For panels a to d results are represented by p-values at $-\log_{10}$ scale and displayed in quantile-quantile (QQ) (left) and Manhattan (right) plots.

Table S1: The 1000 Genomes samples

Population Code	Population Description	Sample size
CHB	Han Chinese in Beijing, China	97
CHS	Southern Han Chinese	100
JPT	Japanese in Tokyo, Japan	89
CEU	Utah Residents with Northern and Western European ancestry	85
TSI	Toscani in Italia	98
FIN	Finnish in Finland	93
GBR	British in England and Scotland	89
IBS	Iberian population in Spain	14
YRI	Yoruba in Ibadan, Nigera	88
LWK	Luhya in Webuye, Kenya	97
ASW	Americans of African Ancestry in SW USA	61
MXL	Mexican Ancestry from Los Angeles USA	66
PUR	Puerto Ricans from Puerto Rico	55
CLM	Colombians from Medellin, Colombia	60