

Supplemental information

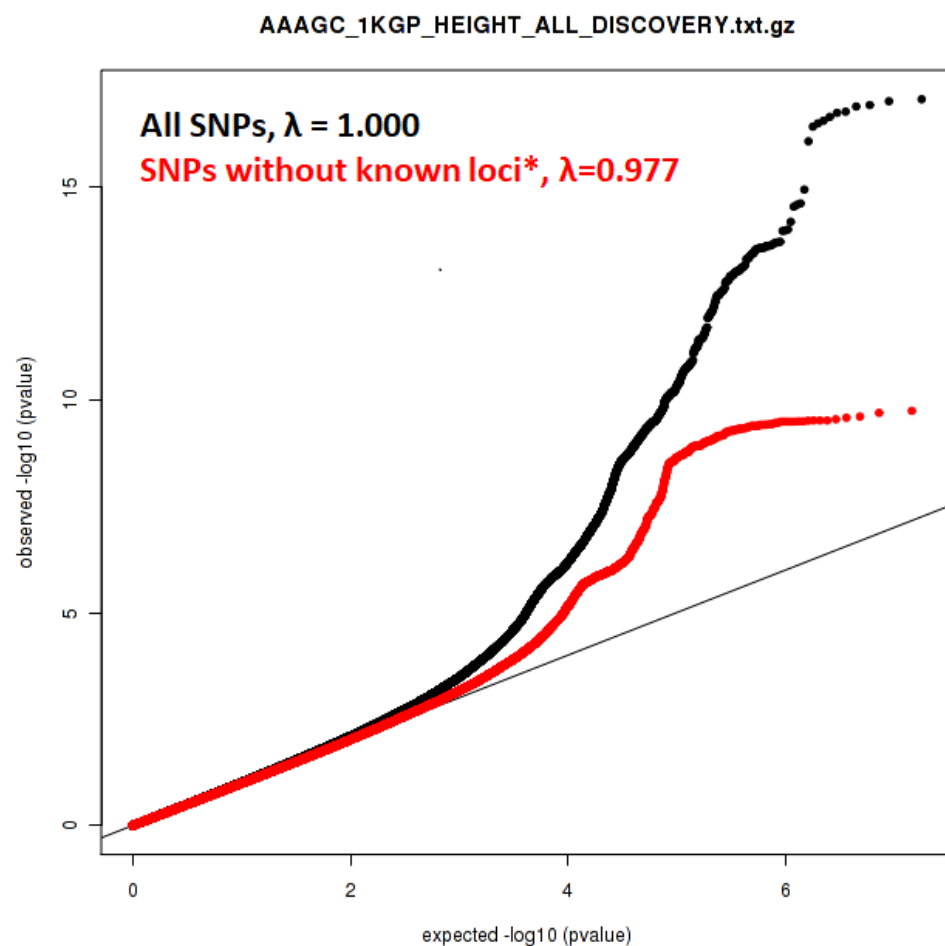
Discovery and fine-mapping of height loci

via high-density imputation of GWASs

in individuals of African ancestry

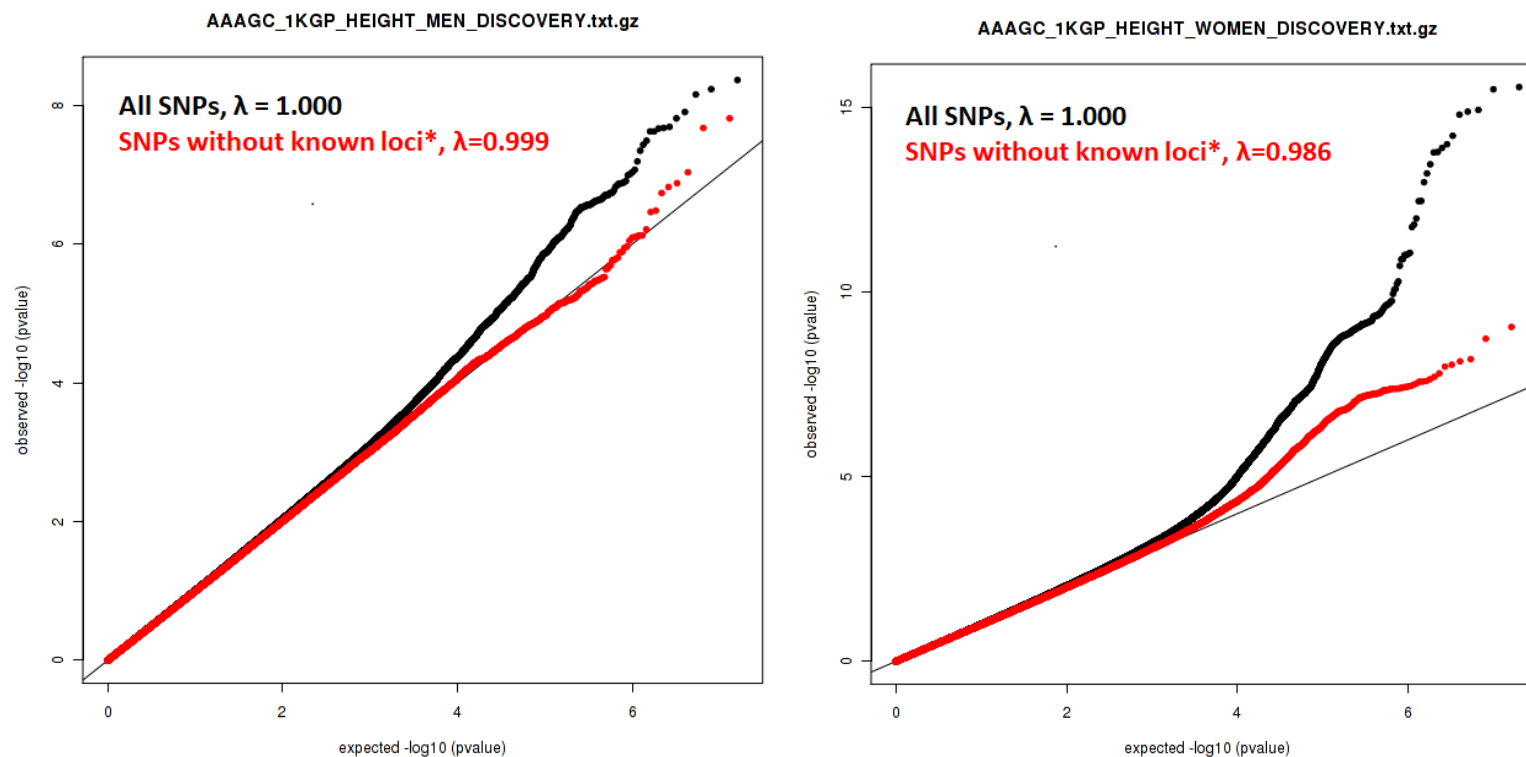
Mariaelisa Graff, Anne E. Justice, Kristin L. Young, Eirini Marouli, Xinruo Zhang, Rebecca S. Fine, Elise Lim, Victoria Buchanan, Kristin Rand, Mary F. Feitosa, Mary K. Wojczynski, Lisa R. Yanek, Yaming Shao, Rebecca Rohde, Adebawale A. Adeyemo, Melinda C. Aldrich, Matthew A. Allison, Christine B. Ambrosone, Stefan Ambs, Christopher Amos, Donna K. Arnett, Larry Atwood, Elisa V. Bandera, Traci Bartz, Diane M. Becker, Sonja I. Berndt, Leslie Bernstein, Lawrence F. Bielak, William J. Blot, Erwin P. Bottinger, Donald W. Bowden, Jonathan P. Bradfield, Jennifer A. Brody, Ulrich Broeckel, Gregory Burke, Brian E. Cade, Qiuyin Cai, Neil Caporaso, Chris Carlson, John Carpten, Graham Casey, Stephen J. Chanock, Guanjie Chen, Minhui Chen, Yii-Der I. Chen, Wei-Min Chen, Alessandra Chesi, Charleston W.K. Chiang, Lisa Chu, Gerry A. Coetzee, David V. Conti, Richard S. Cooper, Mary Cushman, Ellen Demerath, Sandra L. Deming, Latchezar Dimitrov, Jingzhong Ding, W. Ryan Diver, Qing Duan, Michele K. Evans, Adeyinka G. Falusi, Jessica D. Faul, Myriam Fornage, Caroline Fox, Barry I. Freedman, Melissa Garcia, Elizabeth M. Gillanders, Phyllis Goodman, Omri Gottesman, Struan F.A. Grant, Xiuqing Guo, Hakon Hakonarson, Talin Haritunians, Tamara B. Harris, Curtis C. Harris, Brian E. Henderson, Anselm Hennis, Dena G. Hernandez, Joel N. Hirschhorn, Lorna Haughton McNeill, Timothy D. Howard, Barbara Howard, Ann W. Hsing, Yu-Han H. Hsu, Jennifer J. Hu, Chad D. Huff, Dezheng Huo, Sue A. Ingles, Marguerite R. Irvin, Esther M. John, Karen C. Johnson, Joanne M. Jordan, Edmond K. Kabagambe, Sun J. Kang, Sharon L. Kardia, Brendan J. Keating, Rick A. Kittles, Eric A. Klein, Suzanne Kolb, Laurence N. Kolonel, Charles Kooperberg, Lewis Kuller, Abdullah Kutlar, Leslie Lange, Carl D. Langefeld, Loic Le Marchand, Hampton Leonard, Guillaume Lettre, Albert M. Levin, Yun Li, Jin Li, Yongmei Liu, Youfang Liu, Simin Liu, Kurt Lohman, Vaneet Lotay, Yingchang Lu, William Maixner, JoAnn E. Manson, Barbara McKnight, Yan Meng, Keri L. Monda, Kris Monroe, Jason H. Moore, Thomas H. Mosley, Poorva Mudgal, Adam B. Murphy, Rajiv Nadukuru, Mike A. Nalls, Katherine L. Nathanson, Uma Nayak, Amidou N'Diaye, Barbara Nemesure, Christine Neslund-Dudas, Marian L. Neuhouser, Sarah Nyante, Heather Ochs-Balcom, Temidayo O. Ogundiran, Adesola Ogunniyi, Oladosu Ojengbede, Hayrettin Okut, Olufunmilayo I. Olopade, Andrew Olshan, Badri Padhukasahasram, Julie Palmer, Cameron D. Palmer, Nicholette D. Palmer, George Papanicolaou, Sanjay R. Patel, Curtis A. Pettaway, Patricia A. Peyser, Michael F. Press, D.C. Rao, Laura J. Rasmussen-Torvik, Susan Redline, Alex P. Reiner, Suhan K. Rhie, Jorge L. Rodriguez-Gil, Charles N. Rotimi, Jerome I. Rotter, Edward A. Ruiz-Narvaez, Benjamin A. Rybicki, Babatunde Salako, Michele M. Sale, Maureen Sanderson, Eric Schadt, Pamela J. Schreiner, Claudia Schurmann, Ann G. Schwartz, Daniel A. Shriver, Lisa B. Signorello, Andrew B.

Figure S1. Quantile-quantile plot of 1000 genomes phase 1 imputed Stage 1 results and their associations to adult BMI in men and women of African ancestry using all variants and only variants outside of known GWAS loci.



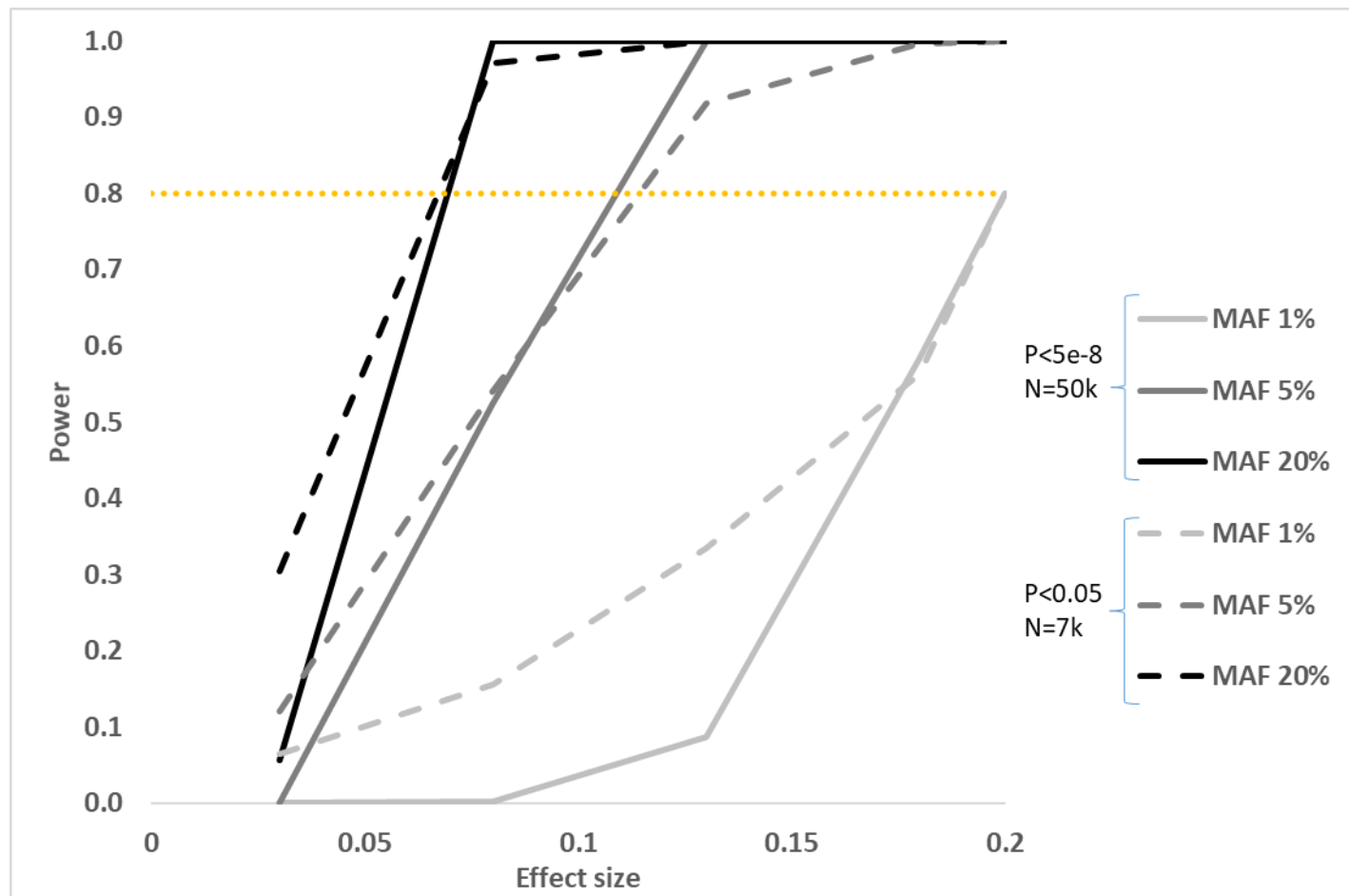
*Known loci defined by +/-500kb around all lead SNPs reported in the literature.

Figure S2. Quantile-quantile plot of 1000 genomes phase 1 imputed Stage 1 results and their associations to adult Height in women only and men only of African ancestry using all variants and only variants outside of known GWAS loci.



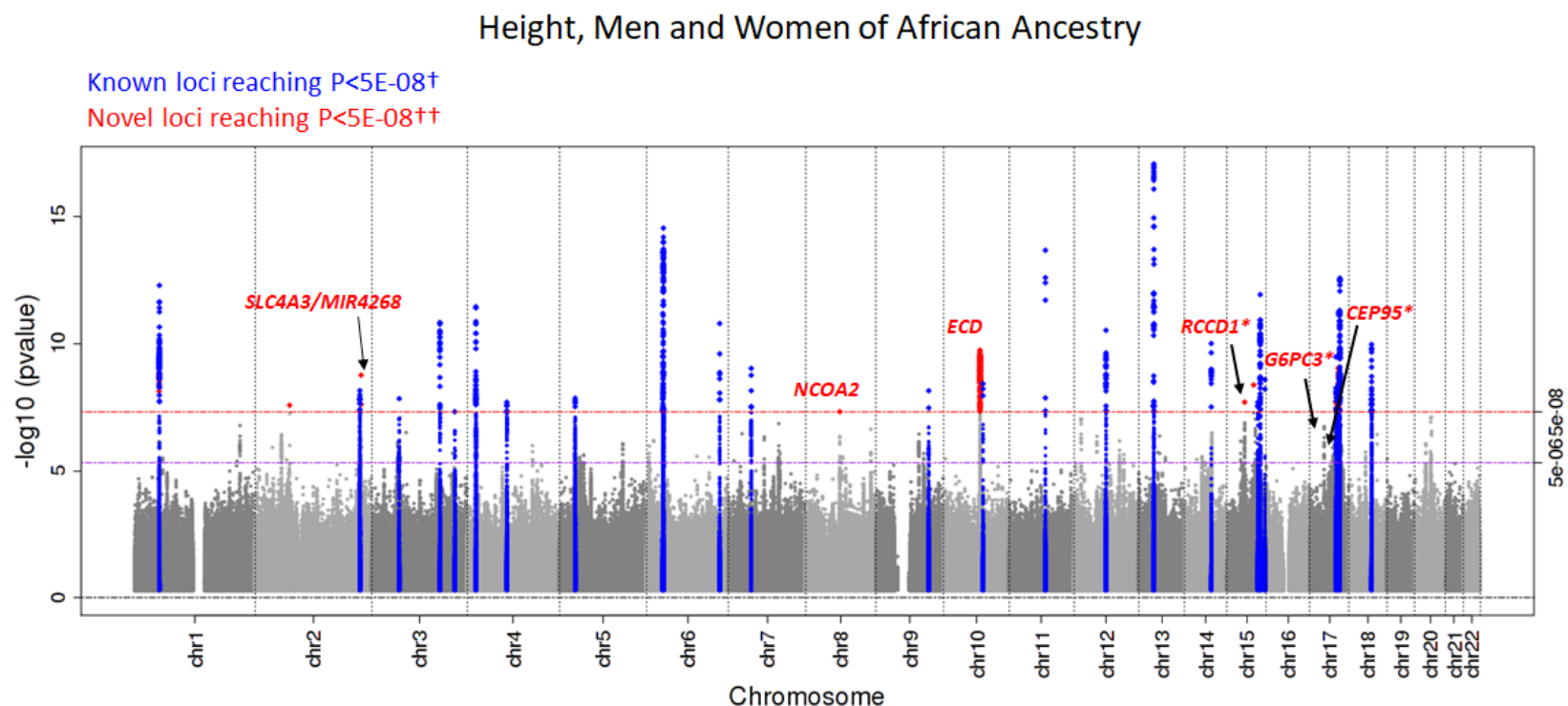
*Known loci defined by $\pm 500\text{kb}$ around all lead SNPs reported in the literature.

Figure S3 Power analyses for Stage 1 plus Stage 2 sample and for validation in children.*



* The solid lines represent power for the shown minor allele frequencies (MAF) in a sample size of 50,000 and genome-wide significance, representative of our Stage 1 plus Stage 2 African ancestry sample. The dashed lines represent power for the shown minor allele frequencies (MAF) in a sample size of 7,000 and nominal significance that we used for our validation in children of African ancestry.

Figure S4. Manhattan plot of 1000 genomes phase 1 imputed Stage 1 results and their associations to adult Height in men and women of African ancestry.

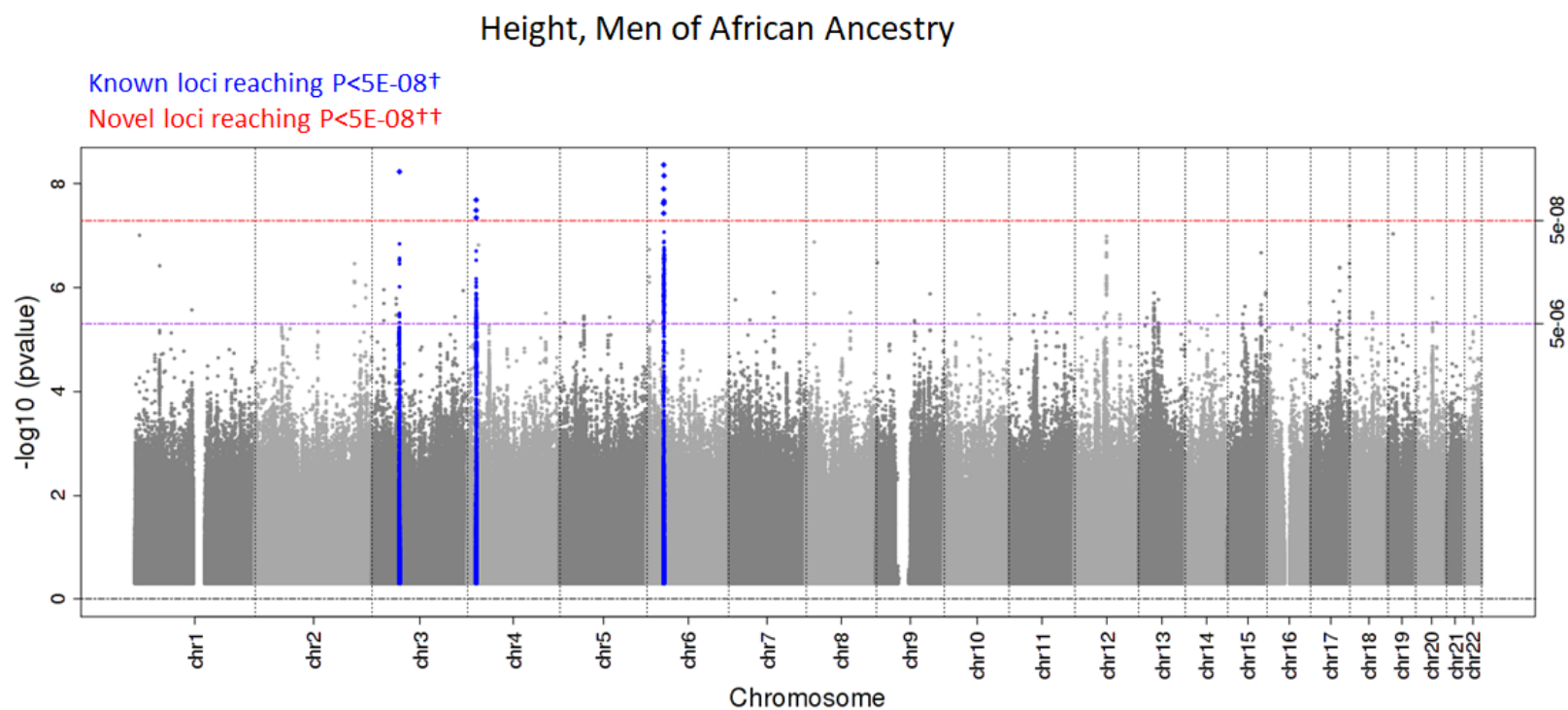


† Known loci with $P < 5 \times 10^{-8}$ in the Stage 1 results in men and women of African ancestry are shown with blue vertical lines.

†† Novel loci with $P < 5 \times 10^{-8}$ in the Stage 1 results in men and women of African ancestry are shown with red vertical lines; novel loci with $P < 5 \times 10^{-8}$ in Stage 1+2 results in men and women of African ancestry and results of men and women of European ancestry results from GIANT are indicated with the loci names.

**RCCD1* (chromosome 15, position 91,500,296), *G6PC3* (chromosome 17, position 42,148,205), *CEP95* (chromosome 17, position 62,534,459) were genome-wide significant after combining the African ancestry Stages 1+2 with men and women of European ancestry results from GIANT.

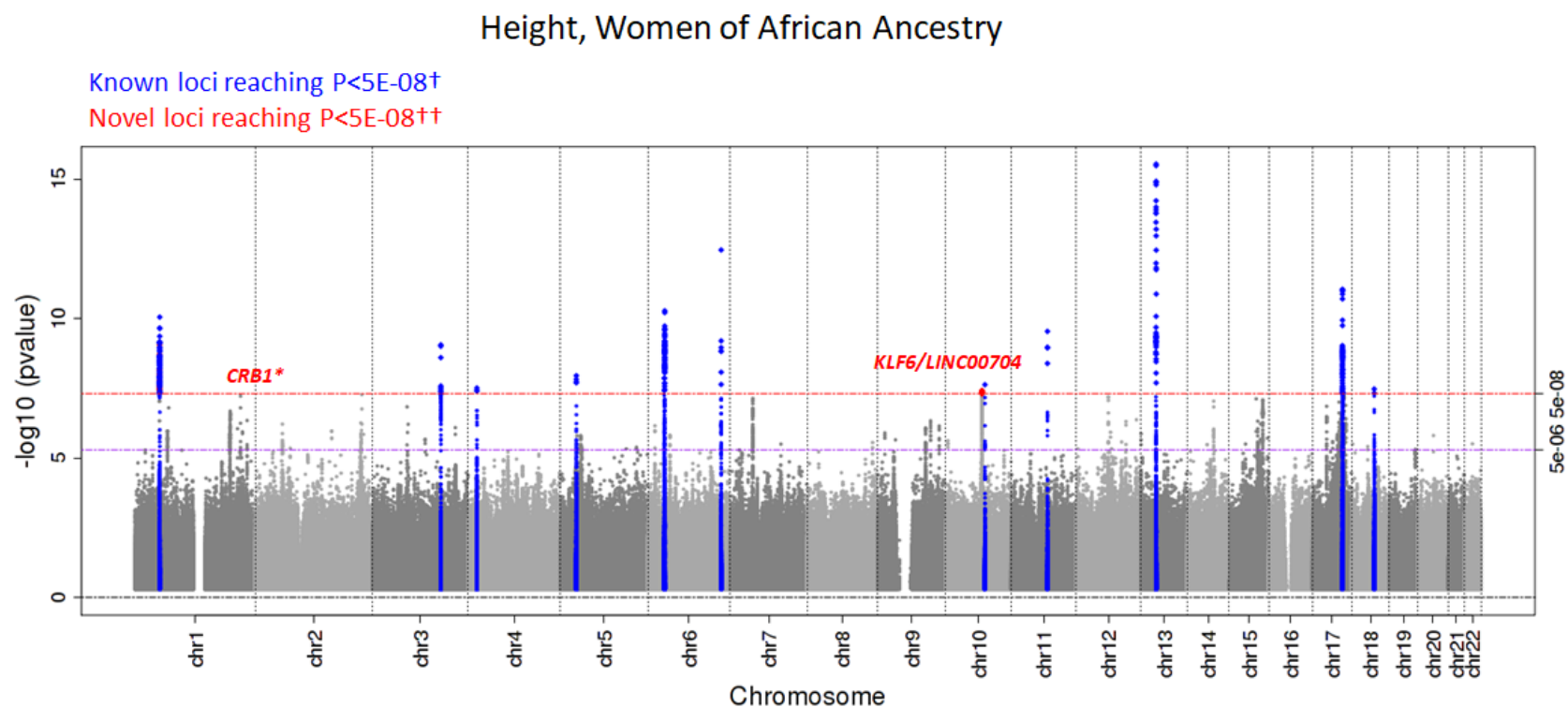
Figure S5. Manhattan plot of 1000 genomes phase 1 imputed Stage 1 results and their associations to adult Height in men of African ancestry.



† Known loci with $P < 5 \times 10^{-08}$ in the Stage 1 results in men and women of African ancestry are shown with blue vertical lines.

$^{++}$ There were no novel loci with $P < 5 \times 10^{-08}$ in men of African ancestry.

Figure S6. Manhattan plot of 1000 genomes phase 1 imputed Stage 1 results and their associations to adult Height in women of African ancestry.



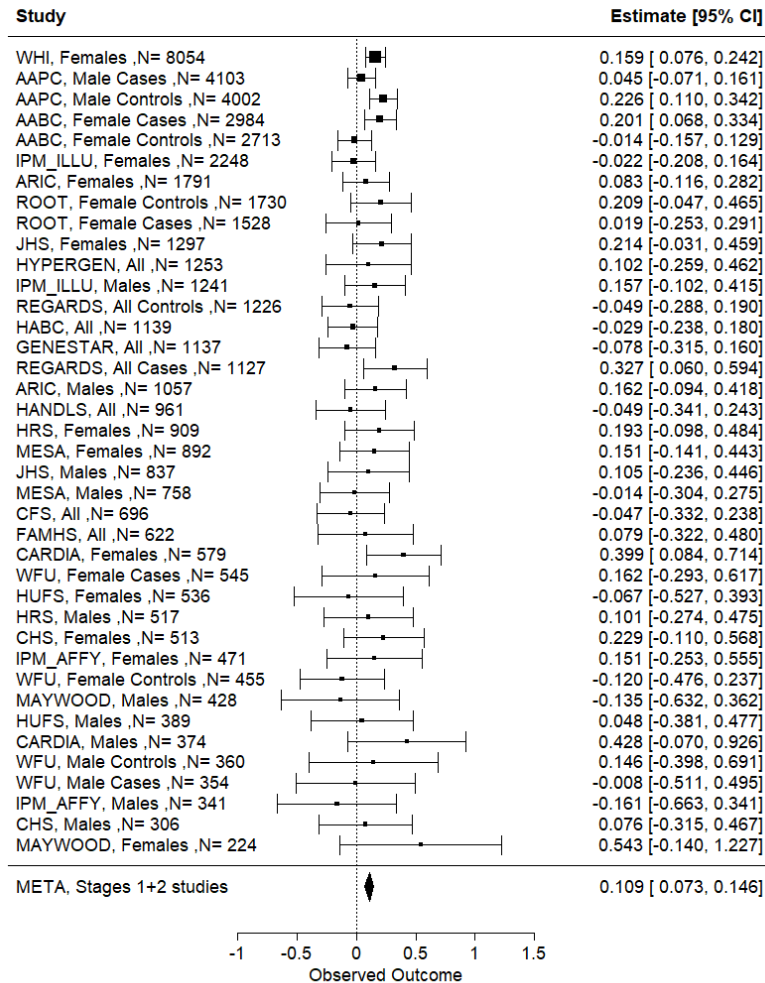
† Known loci with $P < 5 \times 10^{-8}$ in the Stage 1 results in men and women of African ancestry are shown with blue vertical lines.

†† Novel loci with $P < 5 \times 10^{-8}$ in the Stage 1 results in men and women of African ancestry are shown with red vertical lines; novel loci with $P < 5 \times 10^{-8}$ in Stage 1+2 results in men and women of African ancestry.

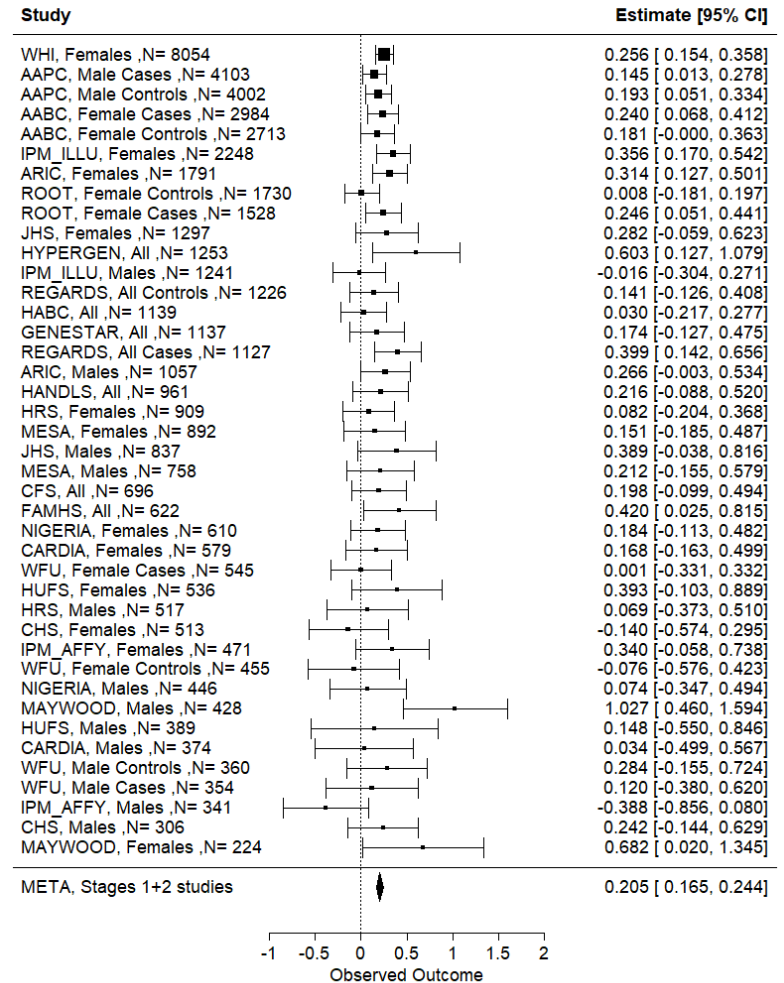
* *CRB1* (chromosome 1, position 197,274,118) was genome-wide significant after combining Stage 1+2 results in women of African ancestry.

Figure S7. Forest plots of variants with effect sizes >0.1 SD in the meta-analyses of stages 1 and 2 for sex-combined results (4 variants: panels A, B, C, D) and for women only results (1 variant: panel E). (Plots are sorted from studies with the largest to the smallest sample size).

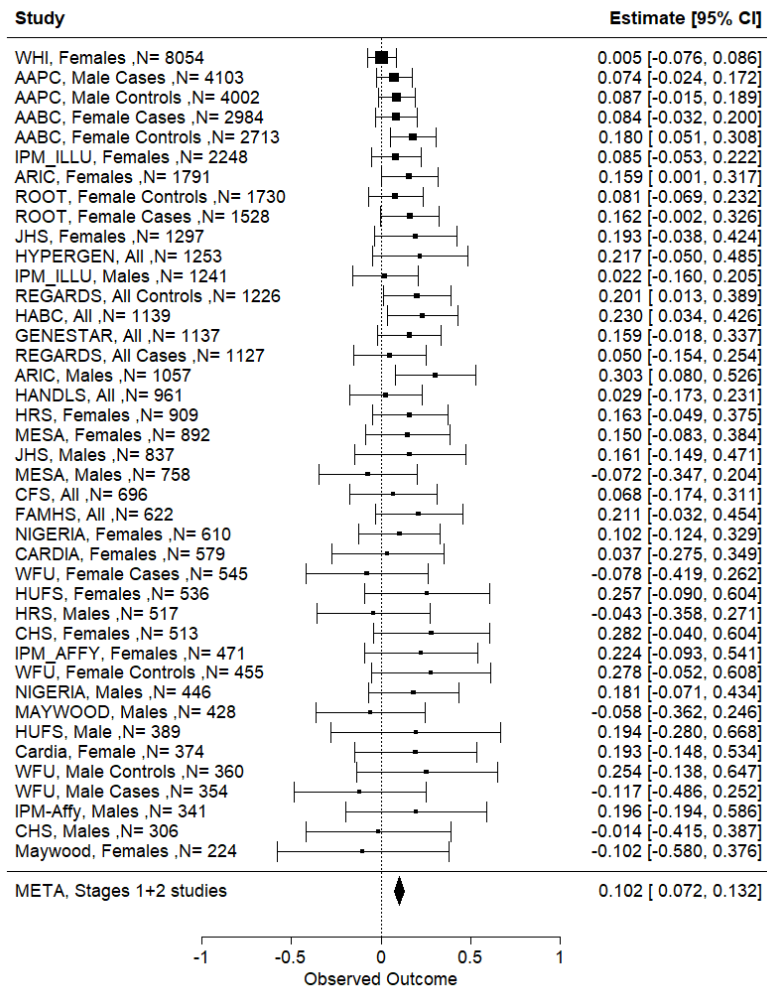
A. Sex-combined:rs112226333,chr5:31525207,Effect/Other alleles=T/G



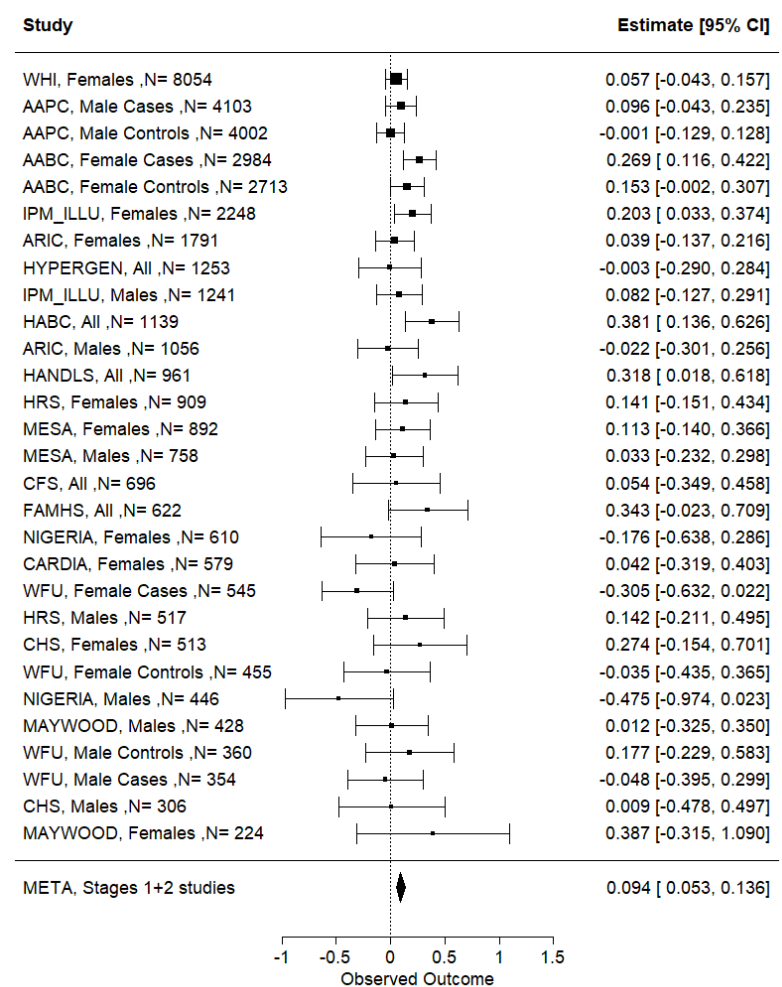
B. Sex-combined:rs75823898,chr13:50669173,Effect/Other alleles=A/C



C.Sex-combined:rs3917155,chr14:76444685,Effect/Other alleles=G/C



D.Sex-combined:rs113229779,chr17:45398018,Effect/Other alleles=T/C



E.Women only:rs672769,chr1:197274118,Effect/Other alleles=T/C

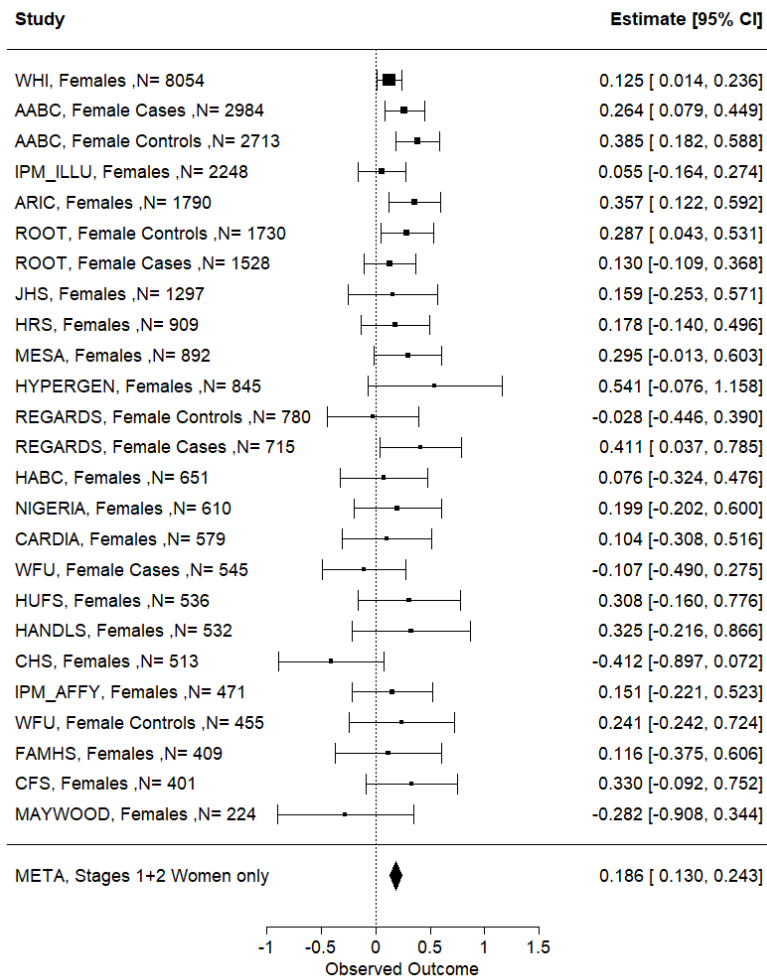


Figure S8. Regression slope for lead variants from effect estimates in children by pubertal status and the effect estimates of stage 1 + stage 2 analyses.

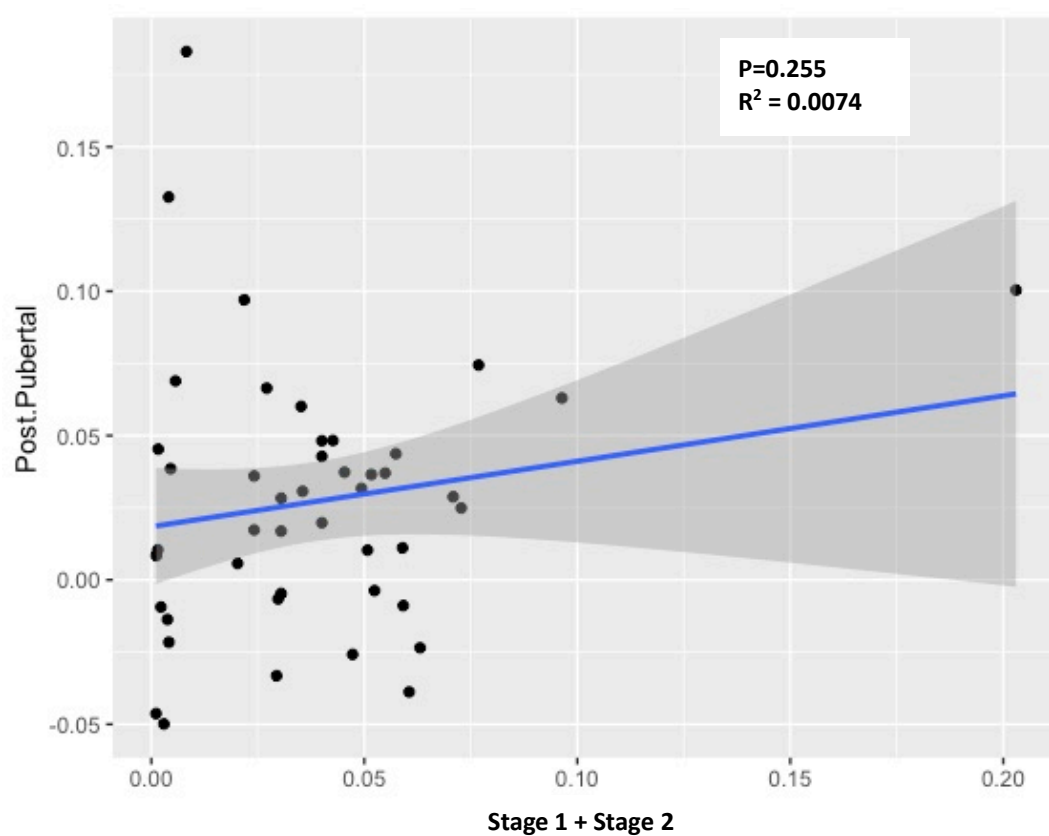
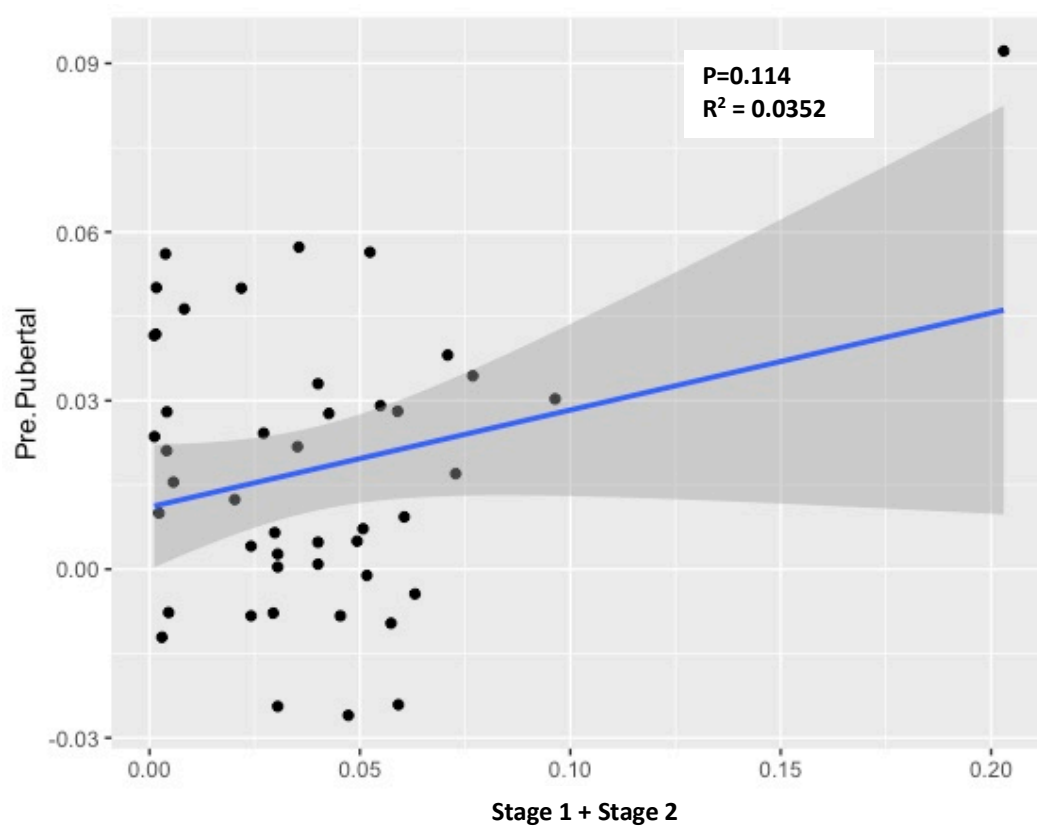


Figure S9. Correlation of effect sizes (A) and effect allele frequencies (B) in European and African ancestry studies in SNP transferability analyses.

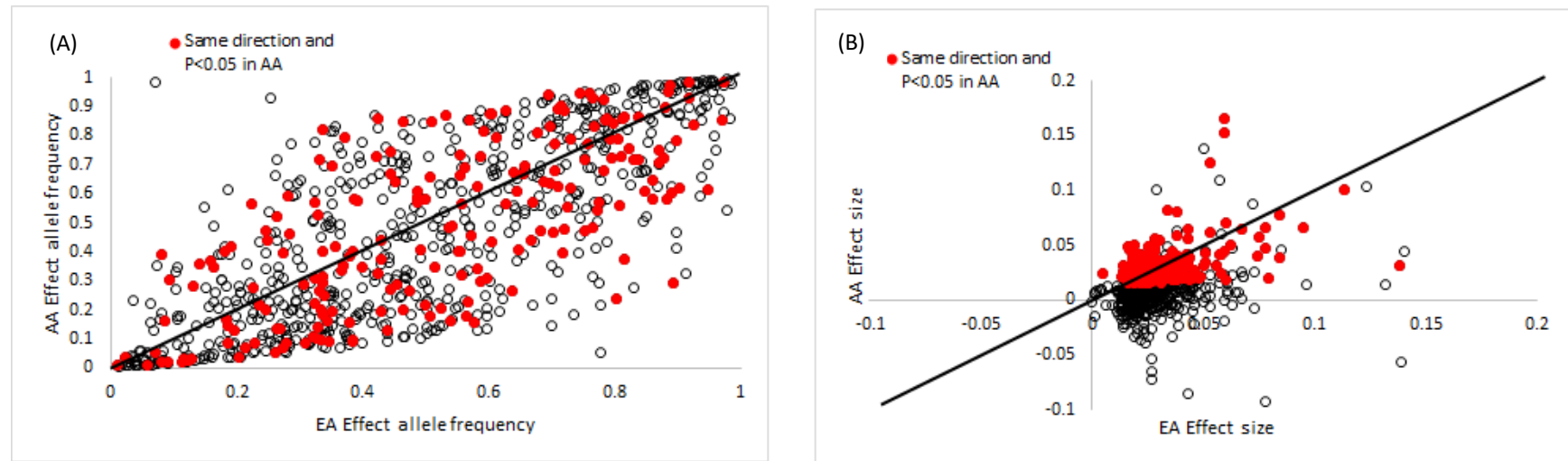


Figure S10. PC1-PC5 reflecting geographical or population structure in Europe, with a focus on PC1 and PC2 (small inset). PC1 and PC2 correspond to the North-South and Southeast-Southwest axes of variation, respectively. Specifically, British in England and Scotland (GBR-orange), Utah Residents with Northern and Western European Ancestry (CEU-blue), Iberian Population in Spain (IBS-green), Toscani in Italia (TSI-red).

