The American Journal of Human Genetics Volume 87

Supplemental Data

A Versatile Gene-Based Test

for Genome-wide Association Studies

Jimmy Z. Liu, Allan F. Mcrae, Dale R. Nyholt, Sarah E. Medland, Naomi R. Wray, Kevin M. Brown, AMFS Investigators, Nicholas K. Hayward, Grant W. Montgomery, Peter M. Visscher, Nicholas G. Martin, and Stuart Macgregor

Supplemental Acknowledgements

The Australian Twin Registry is supported by an Australian National Health and Medical Research Council (NHMRC) Enabling Grant (2004-2009). Genotyping for part of the Australian sample was funded by an NHMRC Medical Genomics Grant. Genotyping for the remainder was performed by the National Institutes of Health (NIH) Center for Inherited Disease Research (CIDR) as part of an NIH/ National Eye Institute (NEI) grant 1RO1EY018246, and we are grateful to Dr Camilla Day and staff. We thank Scott Gordon, Anjali K Henders, Brian McEvoy, Margaret J. Wright, Megan J. Campbell and Anthony Caracella for their assistance in processing the Australian genotyping data.

We acknowledge the contributions of the following individuals for their role in funding, collection and processing the melanoma pooling data: David W. Craig, Zhen Zhen Zhao, Kelly Iyadurai, Anjali Henders, Nils Homer, Megan Campbell, Mitchell Stark, Shane Thomas, Helen Schmid, Elizabeth A Holland, Elizabeth M. Gillanders, David L. Duffy, Judith A. Maskiell, Jodie Jetann, Megan Ferguson, Dietrich A. Stephan, Anne E. Cust, David Whiteman, Adele Green, Håkan Olsson, Susana Puig, Paola Ghiorzo, Johan Hansson, Florence Demenais, Alisa M. Goldstein, Nelleke A. Gruis, David E. Elder, Julia Newton Bishop, Jeffrey M. Trent, Matthew Huentelman, Szabolcs Szelinger, Chantelle Agha-Hamilton, Amanda Baxter, Monica de Nooyer, Isabel Gardner, Dixie Statham, Barbara Haddon, Jane Palmer, Belinda Castellano, Lisa Bardsley, David Smyth, and Harry Beeby. The melanoma sample collection was supported by the National Institutes of Health/National Cancer Institute (CA88363, CA83115), the National Health and Medical Research Council of Australia (NHMRC) (380385, 389892, 496675, 402761), the Cancer Councils NSW, Victoria and Queensland, the Cancer Institute New South Wales, the Melanoma Research Foundation (MRF) and a charitable contribution from F. Najafi.

Figure S1. Comparison of type-I error rates from PLINK set-based test and VEGAS for 1000 permutations on 9 genes. For each gene, height in 3,611 unrelated individuals was permuted 1000 times. The PLINK set-based test (with parameters --set-p 1 --set-r2 1 --maf 0.01 --mperm 1000) and VEGAS was performed for each permutation. The straight diagonal lines indicate a 1-1 relationship.



Figure S2. Comparison of the –log₁₀ p-values from the PLINK set-based test and VEGAS on a GWAS of height in 3,611 individuals using (A) HapMap CEU and (B)

HapMap YRI as the reference population. The PLINK set-based test was performed on 413 genes on chromosome 22 with 10^4 permutations (circles) in addition to 7 genes on other chromosomes, selected on the basis of having the smallest p-values from using VEGAS, at 10^6 permutations (triangles). The p-values from VEGAS were obtained by running 10^3 to 10^6 multivariate normal simulations per gene. The straight diagonal lines indicate a 1-1 relationship.

