

Appendix A

Lists of top *SIS* candidate genes

Top 7% of positively selected protein coding genes and their *SSI* values are given in the following pages.

CHR	- chromosome
GENE NAME	- associated gene name
SIS	- Selection Support Index

Appendix B

Lists of top *FineMAV* candidates

Top 100 *FineMAV* hits in Africans (AFR), East Asians (EAS), Europeans (EUR), Eurasians (EAS+EUR) and non-admixed Native Americans (AMR) are given in the following pages.

SNP	– Single Nucleotide Polymorphism ID
CHR	– chromosome
POS	– genomic position
DER_ALLELE	– derived allele
DAF	– population specific Derived Allele Frequency
DAF_GLOB	– Global Derived Allele Frequency (average across populations)
DAP	– Derived Allele Purity
CADD	– Combined Annotation-Dependent Depletion of derived allele
FineMAV	– population specific Fine-Mapping of Adaptive Variation
CONSEQUENCE	– most severe consequence according to ENSEMBL (NC stands for non-coding; RNA stands for different types of non-coding RNA including lincRNA, snRNA and miRNA)
GENE	– associated gene name

Appendix C

List of primary phenotyping tests

The list below specifies a standard set of phenotyping tests that were applied to all mouse strains generated in this study. Provided here information was derived from Wellcome Trust Sanger Institute Mouse Pipelines internal website (mouse.internal.sanger.ac.uk).

Homozygous viability at P14

Recessive Lethal Study

Homozygous Fertility

General Observations

Weight Curves

Neurological Assessment

Grip Strength

Dysmorphology

Indirect Calorimetry

Glucose Tolerance (ip)

Auditory Brainstem Response

Body Composition (DEXA)

X-ray Imaging

Eye Morphology

Plasma Chemistry

Insulin

Haematology Terminal

Micronuclei

PBL Terminal

Heart Weight

Brain Histopathology

Eye Histopathology

Salmonella Challenge

Citrobacter Challenge

Cytotoxic T Cell Function

Spleen Immunophenotyping

Mesenteric Lymph Node

Bone Marrow

Anti-nuclear Antibody Assay

Epidermal Immune Composition

DSS Challenge

Influenza Challenge

Trichuris Challenge

OBCD Bone

Appendix D

List of publications

A Selective Sweep on a Deleterious Mutation in *CPT1A* in Arctic Populations

American Journal of Human Genetics 2014

Clemente, F. J., Cardona, A., Inchley, C. E., Peter, B. M., Jacobs, G., Pagani, L., Lawson, D. J., Antao, T., Vicente, M., Mitt, M., DeGiorgio, M., Faltyskova, Z., Xue, Y., Ayub, Q., **Szpak, M.**, Magi, R., Eriksson, A., Manica, A., Raghavan, M., Rasmussen, M., Rasmussen, S., Willerslev, E., Vidal-Puig, A., Tyler-Smith, C., Villems, R., Nielsen, R., Metspalu, M., Malyarchuk, B., Derenko, M., Kivisild, T.

Mountain gorilla genomes reveal the impact of long-term population decline and inbreeding

Science 2015

Xue, Y., Prado-Martinez, J., Sudmant, P. H., Narasimhan, V., Ayub, Q., **Szpak, M.**, Frandsen, P., Chen, Y., Yngvadottir, B., Cooper, D. N., de Manuel, M., Hernandez-Rodriguez, J., Lobon, I., Siegismund, H. R., Pagani, L., Quail, M. A., Hvilsom, C., Mudakikwa, A., Eichler, E. E., Cranfield, M. R., Marques-Bonet, T., Tyler-Smith, C., Scally, A.

Prioritizing Candidate Genetic Variants Driving Local Adaptations in Human Populations

Under review

Szpak, M., Mezzavilla, M., Ayub, Q., Chen, Y., Xue, Y., Tyler-Smith, C.

Whole genome sequencing coupled to imputation discovers genetic signals for anthropometric traits

Under review

Tachmazidou, I., Süveges, D., Min, J. L., Ritchie, G. R., Steinberg, J., Walter, K., Iotchkova, V., Schwartzenuber, J., Huang, J., Memari, Y., McCarthy, S., Crawford, A. A., Bombieri, C., Cocca, M., Farmaki, A. E., Gaunt, T. R., Jousilahti, P., Kooijman, M. N., Lehne, B., Malerba, G., Männistö, S., Matchan, A., Medina-Gomez, C., Metrustry, S. J., Nag, A., Ntalla, I., Paternoster, L., Rayner, N. W., Sala, C., Scott, W. R., Shihab, H. A., Southam, L., St Pourcain, B., Traglia, M., Trajanoska, K., Zaza, G., Zhang, W., Artigas, M. S., Bansal, N., Benn, M., Chen, Z., Danecek, P., Lin, W. Y., Locke, A., Luan, J., Manning, A. K., Mulas, A., Sidore, C., Tybjaerg-Hansen, A., Varbo, A., Zoledziwska, M., Finan, C., Hatzikotoulas, K., Hendricks, A. E., Kemp, J. P., Moayyeri, A., Panoutsopoulou, K., **Szpak, M.**, Wilson, S. G., Boehnke, M., Cucca, F., Di Angelantonio, E., Langenberg, C., Lindgren, C., McCarthy, M. I., Morris, A. P., Nordestgaard, B. G., Scott, R. A., Tobin, M. D., Wareham, N. J., SpiroMeta consortium, GoT2D consortium, Burton, P., Chambers, J. C., Davey Smith, G., Dedoussis, G., Felix, J. F., Franco, O. H., Gambaro, G., Gasparini, P., Hammond, C. J., Hofman, A., Jaddoe, V. W., Kleber, M., Kooner, J. S., Perola, M., Relton, C., Ring, S. M., Rivadeneira, F., Salomaa, V., Spector, T. D., Stegle, O., Toniolo, D., Uitterlinden, A. G., arcOGEN consortium, Understanding Society Scientific Group, UK10K consortium, Barroso, I., Perry, J. R., Walker, B. R., Butterworth, A. S., Xue, Y., Durbin, R., Small, K. S., Soranzo, N., Timpson, N. J., Zeggini, E.

