



Universiteit
Leiden
The Netherlands

Algorithms and analysis of human disease genomics

Inouye, M.

Citation

Inouye, M. (2010, April 20). *Algorithms and analysis of human disease genomics*. Retrieved from <https://hdl.handle.net/1887/15277>

Version: Corrected Publisher's Version

[Licence agreement concerning inclusion of doctoral thesis in the Institutional Repository of the University of Leiden](#)

License: <https://hdl.handle.net/1887/15277>

Note: To cite this publication please use the final published version (if applicable).

LIST OF PUBLICATIONS

* indicates joint first authors

1. Jallow M, Teo YY, Small KS, Rockett KA, Deloukas P, Clark TG, Kivinen K, Bojang KA, Conway DJ, Pinder M, Sirugo G, Sisay-Joof F, Usen S, Auburn S, Bumpstead SJ, Campino S, Coffey A, Dunham A, Fry AE, Green A, Gwilliam R, Hunt SE, **Inouye M**, Jeffreys AE, Mendy A, Palotie A, Potter S, Ragoussis J, Rogers J, Rowlands K, Somaskantharajah E, Whittaker P, Widden C, Donnelly P, Howie B, Marchini J, Morris A, Sanjoaquin M, Achidi EA, Agbenyega T, Allen A, Amodu O, Corran P, Djimde A, Dolo A, Doumbo OK, Drakeley C, Dunstan S, Evans J, Farrar J, Fernando D, Hien TT, Horstmann RD, Ibrahim M, Karunaweera N, Kokwaro G, Koram KA, Lemnge M, Makani J, Marsh K, Michon P, Modiano D, Molyneux ME, Mueller I, Parker M, Pesu N, Plowe CV, Puijalon O, Reeder J, Reyburn H, Riley EM, Sakuntabhai A, Singhasivanon P, Sirima S, Tall A, Taylor TE, Thera M, Troye-Bloemberg M, Williams TN, Wilson M, Kwiatkowski DP; Wellcome Trust Case Control Consortium; Malaria Genomic Epidemiology Network. (2009) Genome-wide and fine-resolution association analysis of malaria in West Africa. *Nature Genetics* 41, 657 – 665.
2. Soranzo N, Rivadeneira F, Chinappan-Horsley U, Malkina I, Richards JB, Hammond N, Stolk L, Nicia A, **Inouye M**, Hofman A, Stephens J, Wheeler E, Arp P, Gwilliam R, Jhamai PM, Potter S, Chaney A, Ghori MJ, Ravindrarajah R, Ermakov S, Estrada K, Pols HA, Williams FM, McArdle WL, van Meurs JB, Loos RJ, Dermitzakis ET, Ahmadi KR, Hart DJ, Ouwehand WH, Wareham NJ, Barroso I, Sandhu MS, Strachan DP, Livshits G, Spector TD, Uitterlinden AG, Deloukas P. (2009) Meta-analysis of genome-wide scans for human adult stature identifies novel loci and associations with measures of skeletal frame size. *PLoS Genet*. 5(4):e1000445.
3. Prokopenko I, Langenberg C, Florez JC, Saxena R, Soranzo N, Thorleifsson G, Loos RJ, Manning AK, Jackson AU, Aulchenko Y, Potter SC, Erdos MR, Sanna S, Hottenga JJ, Wheeler E, Kaakinen M, Lyssenko V, Chen WM, Ahmadi K, Beckmann JS, Bergman RN, Bochud M, Bonnycastle LL, Buchanan TA, Cao A, Cervino A, Coin L, Collins FS, Crisponi L, de Geus EJ, Dehghan A, Deloukas P, Doney AS, Elliott P, Freimer N, Gateva V, Herder C, Hofman A, Hughes TE, Hunt S, Illig T, **Inouye M**, Isomaa B, Johnson T, Kong A, Krestyaninova M, Kuusisto J, Laakso M, Lim N, Lindblad U, Lindgren CM, McCann OT, Mohlke KL, Morris AD, Naitza S, Orrù M, Palmer CN, Pouta A, Randall J, Rathmann W, Saramies J, Scheet P, Scott LJ, Scuteri A, Sharp S, Sijbrands E, Smit JH, Song K, Steinhorsdottir V, Stringham HM, Tuomi T, Tuomilehto J, Uitterlinden AG, Voight BF, Waterworth D, Wichmann HE, Willemse G, Witteman JC, Yuan X, Zhao JH, Zeggini E, Schlessinger D, Sandhu M, Boomsma DI, Uda M, Spector TD, Penninx BW, Altshuler D, Vollenweider P, Jarvelin MR, Lakatta E, Waeber G, Fox CS, Peltonen L, Groop LC, Mooser V, Cupples LA, Thorsteinsdottir U, Boehnke M, Barroso I, Van Duijn C, Dupuis J, Watanabe RM, Stefansson K, McCarthy MI, Wareham NJ, Meigs JB, Abecasis GR. (2009) Variants in MTNR1B influence fasting glucose levels. *Nat Genet*. 41(1):77-81.
4. Wang J, Wang W, Li R, Li Y, Tian G, Goodman L, Fan W, Zhang J, Li J, Zhang J, Guo Y, Feng B, Li H, Lu Y, Fang X, Liang H, Du Z, Li D, Zhao Y, Hu Y, Yang Z, Zheng H, Hellmann I, **Inouye M**, Pool J, Yi X, Zhao J, Duan J, Zhou Y, Qin J, Ma L, Li G, Yang Z, Zhang G, Yang B, Yu C, Liang F, Li W, Li S, Li D, Ni P, Ruan J, Li Q, Zhu H, Liu D, Lu Z, Li N, Guo G, Zhang J, Ye J, Fang L, Hao Q, Chen Q, Liang Y, Su Y, San A, Ping C, Yang S, Chen F, Li L, Zhou K, Zheng H, Ren Y, Yang L, Gao Y, Yang G, Li Z, Feng X, Kristiansen K, Wong GK, Nielsen R, Durbin R, Bolund L, Zhang X, Li S, Yang H, Wang J. (2008) The diploid genome sequence of an Asian individual. *Nature*. 456(7218):60-5.
5. Richards JB*, Rivadeneira F*, **Inouye M***, Pastinen TM, Soranzo N, Wilson SG, Andrew T, Falchi M, Gwilliam R, Ahmadi KR, Valdes AM, Arp P, Whittaker P, Verlaan DJ, Jhamai M, Kumanduri V, Moorhouse M, van Meurs JB, Hofman A, Pols HA, Hart D, Zhai G, Kato BS, Mullin BH, Zhang F, Deloukas P, Uitterlinden AG, Spector TD. (2008) Bone mineral density, osteoporosis, and osteoporotic fractures: a genome-wide association study. *Lancet*. 371(9623):1505-12.
6. Loos RJ, Lindgren CM, Li S, Wheeler E, Zhao JH, Prokopenko I, **Inouye M**, Freathy RM, Attwood AP, Beckmann JS, Berndt SI; Prostate, Lung, Colorectal, and Ovarian (PLCO) Cancer Screening Trial,

Jacobs KB, Chanock SJ, Hayes RB, Bergmann S, Bennett AJ, Bingham SA, Bochud M, Brown M, Cauchi S, Connell JM, Cooper C, Smith GD, Day I, Dina C, De S, Dermitzakis ET, Doney AS, Elliott KS, Elliott P, Evans DM, Sadaf Farooqi I, Froguel P, Ghori J, Groves CJ, Gwilliam R, Hadley D, Hall AS, Hattersley AT, Hebebrand J, Heid IM; KORA, Lamina C, Gieger C, Illig T, Meitinger T, Wichmann HE, Herrera B, Hinney A, Hunt SE, Jarvelin MR, Johnson T, Jolley JD, Karpe F, Keniry A, Khaw KT, Luben RN, Mangino M, Marchini J, McArdle WL, McGinnis R, Meyre D, Munroe PB, Morris AD, Ness AR, Neville MJ, Nica AC, Ong KK, O'Rahilly S, Owen KR, Palmer CN, Papadakis K, Potter S, Pouta A, Qi L; Nurses' Health Study, Randall JC, Rayner NW, Ring SM, Sandhu MS, Scherag A, Sims MA, Song K, Soranzo N, Speliotes EK; Diabetes Genetics Initiative, Syddall HE, Teichmann SA, Timpson NJ, Tobias JH, Uda M; SardinIA Study, Vogel CI, Wallace C, Waterworth DM, Weedon MN; Wellcome Trust Case Control Consortium, Willer CJ; FUSION, Wright, Yuan X, Zeggini E, Hirschhorn JN, Strachan DP, Ouwehand WH, Caulfield MJ, Samani NJ, Frayling TM, Vollenweider P, Waeber G, Mooser V, Deloukas P, McCarthy MI, Wareham NJ, Barroso I, Jacobs KB, Chanock SJ, Hayes RB, Lamina C, Gieger C, Illig T, Meitinger T, Wichmann HE, Kraft P, Hankinson SE, Hunter DJ, Hu FB, Lyon HN, Voight BF, Ridderstrale M, Groop L, Scheet P, Sanna S, Abecasis GR, Alba G, Nagaraja R, Schlessinger D, Jackson AU, Tuomilehto J, Collins FS, Boehnke M, Mohlke KL. (2008) Common variants near MC4R are associated with fat mass, weight and risk of obesity. *Nat Genet.* 40(6):768-75.

7. Fisher SA, Tremelling M, Anderson CA, Gwilliam R, Bumpstead S, Prescott NJ, Nimmo ER, Massey D, Berzuini C, Johnson C, Barrett JC, Cummings FR, Drummond H, Lees CW, Onnie CM, Hanson CE, Blaszczyk K, **Inouye M**, Ewels P, Ravindrarajah R, Keniry A, Hunt S, Carter M, Watkins N, Ouwehand W, Lewis CM, Cardon L; Wellcome Trust Case Control Consortium, Lobo A, Forbes A, Sanderson J, Jewell DP, Mansfield JC, Deloukas P, Mathew CG, Parkes M, Satsangi J. (2008) Genetic determinants of ulcerative colitis include the ECM1 locus and five loci implicated in Crohn's disease. *Nat Genet.* 40(6):710-2.
8. Weedon MN, Lango H, Lindgren CM, Wallace C, Evans DM, Mangino M, Freathy RM, Perry JR, Stevens S, Hall AS, Samani NJ, Shields B, Prokopenko I, Farrall M, Dominicak A; Diabetes Genetics Initiative; Wellcome Trust Case Control Consortium, Johnson T, Bergmann S, Beckmann JS, Vollenweider P, Waterworth DM, Mooser V, Palmer CN, Morris AD, Ouwehand WH; Cambridge GEM Consortium, Zhao JH, Li S, Loos RJ, Barroso I, Deloukas P, Sandhu MS, Wheeler E, Soranzo N, **Inouye M**, Wareham NJ, Caulfield M, Munroe PB, Hattersley AT, McCarthy MI, Frayling TM. (2008) Genome-wide association analysis identifies 20 loci that influence adult height. *Nat Genet.* 40(5):575-83.
9. Teo YY*, **Inouye M***, Small KS, Fry AE, Potter SC, Dunstan SJ, Seielstad M, Barroso I, Wareham NJ, Rockett KA, Kwiatkowski DP, Deloukas P. (2008) Whole genome-amplified DNA: insights and imputation. *Nat Methods.* 5(4):279-80.
10. Sandhu MS, Waterworth DM, Debenham SL, Wheeler E, Papadakis K, Zhao JH, Song K, Yuan X, Johnson T, Ashford S, **Inouye M**, Luben R, Sims M, Hadley D, McArdle W, Barter P, Kesäniemi YA, Mahley RW, McPherson R, Grundy SM; Wellcome Trust Case Control Consortium, Bingham SA, Khaw KT, Loos RJ, Waeber G, Barroso I, Strachan DP, Deloukas P, Vollenweider P, Wareham NJ, Mooser V. (2008) LDL-cholesterol concentrations: a genome-wide association study. *Lancet.* 371(9611):483-91.
11. Wellcome Trust Case Control Consortium and Australo-Anglo-American Spondylitis Consortium. (2007) Association scan of 14,500 nonsynonymous SNPs in four diseases identifies autoimmunity variants. *Nat Genet.* 39(11):1329-37.
12. Teo YY*, **Inouye M***, Small KS, Gwilliam R, Deloukas P, Kwiatkowski DP, Clark TG. (2007) A genotype calling algorithm for the Illumina BeadArray platform. *Bioinformatics.* 23(20):2741-6.
13. van Heel DA, Franke L, Hunt KA, Gwilliam R, Zhernakova A, **Inouye M**, Wapenaar MC, Barnardo MC, Bethel G, Holmes GK, Feighery C, Jewell D, Kelleher D, Kumar P, Travis S, Walters JR, Sanders DS, Howdle P, Swift J, Playford RJ, McLaren WM, Mearin ML, Mulder CJ, McManus R, McGinnis R, Cardon LR, Deloukas P, Wijmenga C. (2007) A genome-wide association study for celiac disease identifies risk variants in the region harboring IL2 and IL21. *Nat Genet.* 39(7):827-9.

14. Wellcome Trust Case Control Consortium. (2007) Genome-wide association study of 14,000 cases of seven common diseases and 3,000 shared controls. *Nature*. 447:661-78.

15. Ngan SC, **Inouye MT**, Samudrala R. (2006) A knowledge-based scoring function based on residue triplets for protein structure prediction. *Protein Eng Des Sel*. 19(5):187-93.

CURRICULUM VITAE

Michael Inouye was born on 23 March 1981 in Livingston, New Jersey, USA. He graduated in 1999 from Newport Senior High School (Bellevue, Washington, USA) and, later that year, enrolled at the University of Washington (Seattle, Washington, USA). As a freshman at the University of Washington (UW) studying toward degrees in biochemistry and economics, his interest in bioinformatics was piqued by a summer internship at ZymoGenetics, Inc. Much of his time was spent mining the sequence data still being produced by the Human Genome Sequencing Consortium, analysis which consequently led to a long and fruitful Mary Gates fellowship with Prof. Ram Samudrala in the Computational Genomics Group at the UW Microbiology department. The fellowship was undertaken concurrent to his university studies. With Prof. Samudrala, his first peer-reviewed scientific work was published: an algorithm to differentiate biologically native and non-native protein structures. He was also awarded the Erling J. Ordal prize for best undergraduate research in the department for work on adaptive evolution and sequence-based gene finding. In 2004, he attained his undergraduate degrees and, in 2005, he completed a Masters degree in biochemistry from the University of California at Los Angeles, specializing in protein crystallography and synthetic biochemistry.

He returned to bioinformatics research in late 2005 by moving to the Wellcome Trust Sanger Institute (Cambridge, United Kingdom) where, during the next 4 years, he worked in the groups of Dr. Panos Deloukas and Prof. Leena Peltonen. Throughout this time, his research was primarily in genome-wide association studies (GWAS), leading to many high-profile publications identifying genetic variants underlying bone mineral density and osteoporosis, celiac disease, obesity, and several other complex diseases and phenotypes. However, his main expertise was in the analysis and algorithms used to identify these genetic variants. This included genotype calling algorithms and the statistical, end-point assessment of laboratory protocols (like the whole-genome amplification of DNA). In 2008, he began his PhD studies by splitting his research between the groups of Prof. Peltonen and Prof. Van Ommen in the Department of Human Genetics at Leiden University Medical Center (the Netherlands). In addition to GWAS, his studies during this time were in both clinical genome sequencing and transcriptomics. In particular, he became interested in the

statistical approaches used to dissect biological networks. Using these approaches, he uncovered a novel co-expression network linking inflammation and lipid metabolism, potentially providing a molecular explanation for the role the immune system plays in atherosclerosis.

Currently, he is applying genomics approaches to problems in immunology including the generation of a transcriptional roadmap for the differentiation of the B lymphoblast into an antibody secreting cell.

