# Masahiro Kanai, Ph.D.

# Curriculum Vitae

☑ mkanai@broadinstitute.org

Apr. 2016-Mar. 2017

mkanai.github.io

mkanai

masakanai

## **Education**

**Harvard University** Boston, USA

Ph.D. in Biomedical Informatics Aug. 2017-May 2022

Bioinformatics and Integrative Genomics Ph.D. Program, Harvard Medical School

**Keio University** Yokohama, Japan

Apr. 2013-Mar. 2017 B.S. in Bioinformatics, Department of Biosciences and Informatics

**Research Experience** 

Massachusetts General Hospital & Broad Institute of MIT and Harvard Boston, USA

Research Fellow, Center for Computational and Integrative Biology Aug. 2022-present

Advisor: Dr. Ramnik Xavier

Research themes:

Functional characterization of causal genetic variants

• Integration of multi-omics data for variant Interpretation

Massachusetts General Hospital & Broad Institute of MIT and Harvard

Boston, USA Graduate Student, Analytic and Translational Genetics Unit Jan. 2018-May 2022

**Advisors**: Drs. Mark Daly & Hilary Finucane

Ph.D. thesis: Fine-mapping complex traits in large-scale biobanks across diverse populations

Osaka University Graduate School of Medicine

Osaka, Japan Visiting Fellow, Department of Statistical Genetics July 2022-present

Research Student, Department of Statistical Genetics Apr. 2017-June 2022

Advisor: Dr. Yukinori Okada

Research themes:

Genetic analysis of complex human diseases and traits in the Japanese population

Efficient visualization of high-dimentional multi-omics data

**RIKEN Center for Integrative Medical Sciences** 

Yokohama, Japan May 2015-Jul. 2019 Research Assistant, Laboratory for Statistical Analysis

Advisors: Drs. Yoichiro Kamatani & Yukinori Okada

Research themes:

Genome-wide association study (GWAS) of ~200,000 individuals in the BioBank Japan Project

o Interpretation of GWAS polygenic signals using epigenomic data

Population genetics analysis of 1,037 Japanese whole-genome sequences

**Keio University** Yokohama, Japan

Undergraduate Researcher, Laboratory for Bioinformatics

Advisor: Dr. Yasubumi Sakakibara

Bachelor thesis: Integrative multi-omics analysis of renal cell carcinoma

#### **Tokyo Medical and Dental University**

Tokyo, Japan

Technical Assistant, Department of Human Genetics and Disease Diversity

Apr. 2014-Mar. 2016

Advisors: Drs. Yukinori Okada & Toshihiro Tanaka

#### Research themes:

- Empirical estimation of genome-wide significance thresholds based on GWAS simulations
- HLA imputation analysis using the Japanese-specific reference panel

### **Certification**

# The Certification for Bioinformatics Engineers

certificated by the Japanese Society of Bioinformatics

2015

# **Awards and Fellowships**

Reviewers' Choice Abstract Award	Los Angeles, USA
The American Society of Human Genetics 2022 Annual Meeting	2022
Reviewers' Choice Abstract Award	USA
The American Society of Human Genetics 2021 Virtual Meeting	2021
Charles J. Epstein Trainee Awards for Excellence in Human Genetics Research	USA
Finalist (Predoctoral), The American Society of Human Genetics 2020 Virtual Meeting	2020
Reviewers' Choice Abstract Award	Houston, USA
The American Society of Human Genetics 2019 Annual Meeting	2019
Masason Foundation Fellowship	Tokyo, Japan
Research grant for cross-population analysis of complex human diseases and traits.	2018-2023
, , ,	
Nakajima Foundation Fellowship	Tokyo, Japan
Nakajima Foundation Fellowship	Tokyo, Japan
Nakajima Foundation Fellowship Predoctoral fellowship which covers up to ~\$50,000/year for tuition and stipend.	<b>Tokyo</b> , Japan 2017-2022
Nakajima Foundation Fellowship Predoctoral fellowship which covers up to ~\$50,000/year for tuition and stipend. The 3rd place, Worldwide Finals	Tokyo, Japan 2017-2022 New York, USA

#### **Publications**

#### Selected Publications and Preprints.

(\* = co-first)

- 1. **Kanai, M.**, Elzur, R., Zhou, W., Global Biobank Meta-analysis Initiative, Daly, M. J. & Finucane, H. K. Meta-analysis fine-mapping is often miscalibrated at single-variant resolution. *Cell Genomics* 2, 100210 (2022).
- 2. \*Weissbrod, O., \***Kanai, M.**, \*Shi, H., Gazal, S., Peyrot, W. J., ..., Okada, Y., The Biobank Japan Project, Martin, A. R., Finucane, H. K. & Price, A. L. Leveraging fine-mapping and multipopulation training data to improve cross-population polygenic risk scores. *Nature Genetics* **54**, 450–458 (2022).
- 3. **Kanai, M.**, Ulirsch, J. C., Karjalainen, J., Kurki, M., Karczewski, K. J., ..., Tewhey, R., Sabeti, P. C., Okada, Y., Daly, M. J. & Finucane, H. K. Insights from complex trait fine-mapping across diverse populations. *medRxiv* (2021).

- 4. \*Sakaue, S., \*Kanai, M., Tanigawa, Y., Karjalainen, J., Kurki, M., ..., Palotie, A., Rivas, M. A., Daly, M. J., Matsuda, K. & Okada, Y. A cross-population atlas of genetic associations for 220 human phenotypes. *Nature Genetics* 53, 1415–1424 (2021).
- 5. The COVID-19 Host Genetics Initiative. Mapping the human genetic architecture of COVID-19. *Nature* **600**, 472–477 (2021).
- \*Sakaue, S., \*Kanai, M., Karjalainen, J., Akiyama, M., Kurki, M., ..., Matsuda, K., Murakami, Y., Daly, M. J., Kamatani, Y. & Okada, Y. Trans-biobank analysis with 676,000 individuals elucidates the association of polygenic risk scores of complex traits with human lifespan. *Nature Medicine* 26, 542–548 (2020).
- 7. Martin, A. R., **Kanai, M.**, Kamatani, Y., Okada, Y., Neale, B. M. & Daly, M. J. Clinical use of current polygenic risk scores may exacerbate health disparities. *Nature Genetics* **51**, 584–591 (2019).
- 8. **Kanai, M.**, Akiyama, M., Takahashi, A., Matoba, N., Momozawa, Y., ..., Hirata, M., Matsuda, K., Kubo, M., Okada, Y. & Kamatani, Y. Genetic analysis of quantitative traits in the Japanese population links cell types to complex human diseases. *Nature Genetics* **50**, 390–400 (2018).

# Other Preprints

- 9. Sakaue, S., Weinand, K., Dey, K. K., Jagadeesh, K., **Kanai, M.**, ..., McDavid, A., Donlin, L., Wei, K., Price, A. & Raychaudhuri, S. Tissue-specific enhancer-gene maps from multimodal single-cell data identify causal disease alleles. *medRxiv* (2022).
- 10. Cui, R., Elzur, R. A., **Kanai, M.**, Ulirsch, J. C., Weissbrod, O., Daly, M. J., Neale, B. M., Fan, Z. & Finucane, H. K. Improving fine-mapping by modeling infinitesimal effects. *bioRxiv* (2022).
- 11. Chen, S., Francioli, L., Goodrich, J., Collins, R., **Kanai, M.**, ..., Daly, M., Tiao, G., Neale, B., MacArthur, D. & Karczewski, K. A genome-wide mutational constraint map quantified from variation in 76,156 human genomes. *bioRxiv* (2022).
- 12. Carey, C. E., Shafee, R., Elliott, A., Palmer, D. S., Compitello, J., **Kanai, M.**, ..., Davey Smith, G., Wedow, R., Neale, B. M., Walters, R. K. & Robinson, E. B. Principled distillation of multidimensional UK Biobank data reveals insights into the correlated human phenome. *medRxiv* (2022).
- 13. Meng, X., Navoly, G., Giannakopoulou, O., Levey, D., Koller, D., ..., **Kanai, M.**, ..., Dunn, E. C., Stein, M. B., Gelernter, J., Lewis, C. & Kuchenbaecker, K. Multi-ancestry GWAS of major depression aids locus discovery, fine-mapping, gene prioritisation, and causal inference. *bioRxiv* (2022).
- 14. International League Against Epilepsy Consortium on Complex Epilepsies. Genome-wide metaanalysis of over 29,000 people with epilepsy reveals 26 loci and subtype-specific genetic architecture. *medRxiv* (2022).
- 15. Kurki, M. I., Karjalainen, J., Palta, P., Sipilä, T. P., Kristiansson, K., ..., **Kanai, M.**, ..., Plenge, R., McCarthy, M., Runz, H., Daly, M. J. & Palotie, A. FinnGen: Unique genetic insights from combining isolated population and national health register data. *medRxiv* (2022).
- 16. Lo Faro, V., Bhattacharya, A., Zhou, W., Zhou, D., Wang, Y., ..., **Kanai, M.**, ..., Gamazon, E. R., Jansonius, N. M., Joos, K., Cox, N. J. & Hirbo, J. Genome-wide association meta-analysis identifies novel ancestry-specific primary open-angle glaucoma loci and shared biology with vascular mechanisms and cell proliferation. *medRxiv* (2021).
- 17. Kanoni, S., Graham, S. E., Wang, Y., Surakka, I., Ramdas, S., ..., **Kanai, M.**, ..., Natarajan, P., Deloukas, P., Willer, C. J., Assimes, T. L. & Peloso, G. M. Implicating genes, pleiotropy and sexual dimorphism at blood lipid loci through multi-ancestry meta-analysis. *medRxiv* (2021).
- 18. Tsuo, K., Zhou, W., Wang, Y., **Kanai, M.**, Namba, S., ..., Morisaki, T., Neale, B., Global Biobank Meta-analysis Initiative, Daly, M. J. & Martin, A. Multi-ancestry meta-analysis of asthma identifies novel associations and highlights the value of increased power and diversity. *medRxiv* (2021).

- 19. Wang, Y., Namba, S., Lopera-Maya, E. A., Kerminen, S., Tsuo, K., ..., **Kanai, M.**, ..., Cox, N. J., Surakka, I., Okada, Y., Martin, A. R. & Hirbo, J. Global biobank analyses provide lessons for computing polygenic risk scores across diverse cohorts. *medRxiv* (2021).
- 20. Palmer, D. S., Zhou, W., Abbott, L., Baya, N., Churchhouse, C., ..., Poterba, T., King, D., **Kanai, M.**, Bloemendal, A. & Neale, B. M. Analysis of genetic dominance in the UK Biobank. *bioRxiv* (2021).
- 21. Koskela, J. T., Happola, P., Liu, A., FinnGen, Partanen, J., ..., **Kanai, M.**, ..., Ganna, A., Palotie, A., Laitinen, T., Myllarniemi, M. & Daly, M. J. Genetic variant in SPDL1 reveals novel mechanism linking pulmonary fibrosis risk and cancer protection. *medRxiv* (2021).
- 22. Turley, P., Martin, A. R., Goldman, G., Li, H., **Kanai, M.**, ..., Meyer, M. N., Cesarini, D., Daly, M., Benjamin, D. J. & Neale, B. M. Multi-Ancestry Meta-Analysis yields novel genetic discoveries and ancestry-specific associations. *bioRxiv* (2021).
- 23. Rämö, J. T., Kiiskinen, T., Karjalainen, J., Krebs, K., Kurki, M., ..., **Kanai, M.**, ..., Ripatti, S., Milani, L., Mäkitie, A., Daly, M. J. & Palotie, A. Genome-wide Screen of Otosclerosis in Population Biobanks: 18 Loci and Shared Heritability with Skeletal Structure. *medRxiv* (2020).
- 24. Weeks, E. M., Ulirsch, J. C., Cheng, N. Y., Trippe, B. L., Fine, R. S., ..., **Kanai, M.**, ..., Ardlie, K. G., Hirschhorn, J. N., Lander, E. S., Engreitz, J. M. & Finucane, H. K. Leveraging polygenic enrichments of gene features to predict genes underlying complex traits and diseases. *medRxiv* (2020).

# Other Publications

- 25. Zhou, W., **Kanai, M.**, Wu, K.-H. H., Rasheed, H., Tsuo, K., ..., Zöllner, S., Martin, A. R., Willer, C. J., Daly, M. J. & Neale, B. M. Global Biobank Meta-analysis Initiative: Powering genetic discovery across human disease. *Cell Genomics* 2, 100192 (2022).
- 26. Bhattacharya, A., Hirbo, J. B., Zhou, D., Zhou, W., Zheng, J., **Kanai, M.**, The Global Biobank Meta-analysis Initiative, Pasaniuc, B., Gamazon, E. R. & Cox, N. J. Best practices for multi-ancestry, meta-analytic transcriptome-wide association studies: Lessons from the Global Biobank Meta-analysis Initiative. *Cell Genomics* **2**, 100180 (2022).
- 27. Yengo, L., Vedantam, S., Marouli, E., Sidorenko, J., Bartell, E., ..., **Kanai, M.**, ..., Frayling, T. M., Okada, Y., Wood, A. R., Visscher, P. M. & Hirschhorn, J. N. A saturated map of common genetic variants associated with human height. *Nature* **610**, 704–712 (2022).
- 28. Wang, Q. S., Edahiro, R., Namkoong, H., Hasegawa, T., Shirai, Y., ..., **Kanai, M.**, ..., Miyano, S., Ogawa, S., Kanai, T., Fukunaga, K. & Okada, Y. The whole blood transcriptional regulation landscape in 465 COVID-19 infected samples from Japan COVID-19 Task Force. *Nature Communications* **13**, 4830 (2022).
- 29. Ruotsalainen, S. E., Surakka, I., Mars, N., Karjalainen, J., Kurki, M., **Kanai, M.**, ..., Okada, Y., Palotie, A., Widen, E., Daly, M. J. & Ripatti, S. Inframe insertion and splice site variants in MFGE8 associate with protection against coronary atherosclerosis. *Communications Biology* **5**, 802 (2022).
- 30. Namkoong, H., Edahiro, R., Takano, T., Nishihara, H., Shirai, Y., ..., **Kanai, M.**, ..., Miyano, S., Ogawa, S., Kanai, T., Fukunaga, K. & Okada, Y. DOCK2 is involved in the host genetics and biology of severe COVID-19. *Nature* 609, 754–760 (2022).
- 31. Ramdas, S., Judd, J., Graham, S. E., Kanoni, S., Wang, Y., ..., **Kanai, M.**, ..., Peloso, G., Assimes, T. L., Willer, C. J., Zhu, X. & Brown, C. D. A multi-layer functional genomic analysis to understand noncoding genetic variation in lipids. *The American Journal of Human Genetics* **109**, 1366–1387 (2022).
- 32. The COVID-19 Host Genetics Initiative. A first update on mapping the human genetic architecture of COVID-19. *Nature* **608**, E1–E10 (2022).
- 33. Winkler, T. W., Rasheed, H., Teumer, A., Gorski, M., Rowan, B. X., ..., **Kanai, M.**, ..., Hung, A. M., Kronenberg, F., Köttgen, A., Pattaro, C. & Heid, I. M. Differential and shared genetic effects on kidney function between diabetic and non-diabetic individuals. *Communications Biology* **5**, 580 (2022).

- 34. Wang, Y., Tsuo, K., **Kanai, M.**, Neale, B. M. & Martin, A. R. Challenges and Opportunities for Developing More Generalizable Polygenic Risk Scores. *Annual Review of Biomedical Data Science* **5**, 293–320 (2022).
- 35. Mars, N., Kerminen, S., Feng, Y.-C. A., **Kanai, M.**, Läll, K., ..., Pirinen, M., Palotie, A., Ganna, A., Martin, A. R. & Ripatti, S. Genome-wide risk prediction of common diseases across ancestries in one million people. *Cell Genomics* 2, 100118 (2022).
- 36. Zheng, J., Zhang, Y., Rasheed, H., Walker, V., Sugawara, Y., ..., **Kanai, M.**, ..., Barbour, S., Yu, C., Åsvold, B. O., Zhang, H. & Gaunt, T. R. Trans-ethnic Mendelian-randomization study reveals causal relationships between cardiometabolic factors and chronic kidney disease. *International Journal of Epidemiology* **50**, 1995–2010 (2022).
- 37. Graham, S. E., Clarke, S. L., Wu, K.-H. H., Kanoni, S., Zajac, G. J. M., ..., **Kanai, M.**, ..., Morris, A. P., Assimes, T. L., Deloukas, P., Sun, Y. V. & Willer, C. J. The power of genetic diversity in genome-wide association studies of lipids. *Nature* 600, 675–679 (2021).
- 38. Polygenic Risk Score Task Force of the International Common Disease Alliance. Responsible use of polygenic risk scores in the clinic: potential benefits, risks and gaps. *Nature Medicine* **27**, 1876–1884 (2021).
- 39. Luo, Y., **Kanai, M.**, Choi, W., Li, X., Sakaue, S., ..., Esko, T., Okada, Y., Han, B., McLaren, P. J. & Raychaudhuri, S. A high-resolution HLA reference panel capturing global population diversity enables multi-ancestry fine-mapping in HIV host response. *Nature Genetics* **53**, 1504–1516 (2021).
- 40. Griesemer, D., Xue, J. R., Reilly, S. K., Ulirsch, J. C., Kukreja, K., ..., **Kanai, M.**, ..., Montgomery, S. B., Finucane, H. K., Novina, C. D., Tewhey, R. & Sabeti, P. C. Genome-wide functional screen of 3'UTR variants uncovers causal variants for human disease and evolution. *Cell* **184**, 5247–5260.e19 (2021).
- 41. Reilly, S. K., Gosai, S. J., Guiterrez, A., Ulirsch, J. C., **Kanai, M.**, ..., Butler, G. B., Gladden-Young, A., Finucane, H. K., Sabeti, P. C. & Tewhey, R. Direct characterization of cis-regulatory elements and functional dissection of complex genetic associations using HCR-FlowFISH. *Nature Genetics* 53, 1166–1176 (2021).
- 42. Wang, Q. S., Kelley, D. R., Ulirsch, J., **Kanai, M.**, Sadhuka, S., ..., Okada, Y., Aguet, F., Ardlie, K. G., MacArthur, D. G. & Finucane, H. K. Leveraging supervised learning for functionally informed fine-mapping of cis-eQTLs identifies an additional 20,913 putative causal eQTLs. *Nature Communications* 12, 1–11 (2021).
- 43. Nakatochi, M., Toyoda, Y., **Kanai, M.**, Nakayama, A., Kawamura, Y., ..., Yokota, M., Wakai, K., Okada, Y., Matsuo, H. & Japan Uric Acid Genomics Consortium (Japan Urate). An X chromosome-wide meta-analysis based on Japanese cohorts revealed that non-autosomal variations are associated with serum urate. *Rheumatology* **60**, 4430–4432 (2021).
- 44. Pirastu, N., Cordioli, M., Nandakumar, P., Mignogna, G., Abdellaoui, A., ..., **Kanai, M.**, ..., Neale, B. M., Walters, R. K., Nivard, M. G., Perry, J. R. B. & Ganna, A. Genetic analyses identify widespread sex-differential participation bias. *Nature Genetics* **53**, 663–671 (2021).
- 45. Shi, H., Gazal, S., **Kanai, M.**, Koch, E. M., Schoech, A. P., ..., Huang, H., Okada, Y., Raychaudhuri, S., Sunyaev, S. R. & Price, A. L. Population-specific causal disease effect sizes in functionally important regions impacted by selection. *Nature Communications* **12**, 1098 (2021).
- 46. Atkinson, E. G., Maihofer, A. X., **Kanai, M.**, Martin, A. R., Karczewski, K. J., ..., Finucane, H. K., Koenen, K. C., Nievergelt, C. M., Daly, M. J. & Neale, B. M. Tractor uses local ancestry to enable the inclusion of admixed individuals in GWAS and to boost power. *Nature Genetics* **53**, 195–204 (2021).
- 47. Chen, M.-H., Raffield, L. M., Mousas, A., Sakaue, S., Huffman, J. E., ..., **Kanai, M.**, ..., Soranzo, N., Johnson, A. D., Reiner, A. P., Auer, P. L. & Lettre, G. Trans-ethnic and Ancestry-Specific Blood-Cell Genetics in 746,667 Individuals from 5 Global Populations. *Cell* 182, 1198–1213.e14 (2020).

- 48. Vuckovic, D., Bao, E. L., Akbari, P., Lareau, C. A., Mousas, A., ..., **Kanai, M.**, ..., Butterworth, A. S., Ouwehand, W. H., Lettre, G., Sankaran, V. G. & Soranzo, N. The Polygenic and Monogenic Basis of Blood Traits and Diseases. *Cell* 182, 1214–1231.e11 (2020).
- 49. Shirai, Y., Honda, S., Ikari, K., **Kanai, M.**, Takeda, Y., ..., Morisaki, T., Tanaka, E., Kumanogoh, A., Harigai, M. & Okada, Y. Association of the RPA3-UMAD1 locus with interstitial lung diseases complicated with rheumatoid arthritis in Japanese. *Annals of the Rheumatic Diseases* **79**, 1305–1309 (2020).
- 50. Ishigaki, K., Akiyama, M., **Kanai, M.**, Takahashi, A., Kawakami, E., ..., Inazawa, J., Yamauchi, T., Kadowaki, T., Kubo, M. & Kamatani, Y. Large-scale genome-wide association study in a Japanese population identifies novel susceptibility loci across different diseases. *Nature Genetics* **52**, 669–679 (2020).
- 51. Sakaue, S., Hirata, J., **Kanai, M.**, Suzuki, K., Akiyama, M., ..., Hirata, M., Matsuda, K., Murakami, Y., Kamatani, Y. & Okada, Y. Dimensionality reduction reveals fine-scale structure in the Japanese population with consequences for polygenic risk prediction. *Nature Communications* **11**, 1569 (2020).
- 52. Ray, J. P., de Boer, C. G., Fulco, C. P., Lareau, C. A., **Kanai, M.**, ..., Issner, R., Finucane, H. K., Lander, E. S., Regev, A. & Hacohen, N. Prioritizing disease and trait causal variants at the TNFAIP3 locus using functional and genomic features. *Nature Communications* **11**, 1237 (2020).
- 53. Matoba, N., Akiyama, M., Ishigaki, K., **Kanai, M.**, Takahashi, A., ..., Matsuda, K., Murakami, Y., Kubo, M., Kamatani, Y. & Okada, Y. GWAS of 165,084 Japanese individuals identified nine loci associated with dietary habits. *Nature Human Behaviour* 4, 308–316 (2020).
- 54. Clark, D. W., Okada, Y., Moore, K. H. S., Mason, D., Pirastu, N., ..., **Kanai, M.**, ..., Helgason, A., Stefansson, K., Joshi, P. K., Kubo, M. & Wilson, J. F. Associations of autozygosity with a broad range of human phenotypes. *Nature Communications* **10**, 4957 (2019).
- 55. Leu, C., Stevelink, R., Smith, A. W., Goleva, S. B., **Kanai, M.**, ..., Najm, I. M., Palotie, A., Daly, M. J., Busch, R. M. & Lal, D. Polygenic burden in focal and generalized epilepsies. *Brain* 142, 3473–3481 (2019).
- Tin, A., Marten, J., Halperin Kuhns, V. L., Li, Y., Wuttke, M., ..., Kanai, M., ..., Teumer, A., Pattaro, C., Woodward, O. M., Vitart, V. & Köttgen, A. Target genes, variants, tissues and transcriptional pathways influencing human serum urate levels. *Nature Genetics* 51, 1459–1474 (2019).
- 57. Akiyama, M., Ishigaki, K., Sakaue, S., Momozawa, Y., Horikoshi, M., ..., **Kanai, M.**, ..., Yamamoto, M., Okada, Y., Murakami, Y., Kubo, M. & Kamatani, Y. Characterizing rare and low-frequency height-associated variants in the Japanese population. *Nature Communications* **10**, 4393 (2019).
- 58. Wuttke, M., Li, Y., Li, M., Sieber, K. B., Feitosa, M. F., ..., **Kanai, M.**, ..., Heid, I. M., Scholz, M., Teumer, A., Köttgen, A. & Pattaro, C. A catalog of genetic loci associated with kidney function from analyses of a million individuals. *Nature Genetics* **51**, 957–972 (2019).
- 59. \*Nakatochi, M., \***Kanai, M.**, \*Nakayama, A., \*Hishida, A., \*Kawamura, Y., ..., Shinomiya, N., Yokota, M., Wakai, K., Okada, Y. & Matsuo, H. Genome-wide meta-analysis identifies multiple novel loci associated with serum uric acid levels in Japanese individuals. *Communications Biology* 2, 115 (2019).
- 60. Matoba, N., Akiyama, M., Ishigaki, K., **Kanai, M.**, Takahashi, A., ..., Hirata, M., Matsuda, K., Kubo, M., Okada, Y. & Kamatani, Y. GWAS of smoking behaviour in 165,436 Japanese people reveals seven new loci and shared genetic architecture. *Nature Human Behaviour* 3, 471–477 (2019).
- 61. Suzuki, K., Akiyama, M., Ishigaki, K., **Kanai, M.**, Hosoe, J., ..., Kubo, M., Kamatani, Y., Horikoshi, M., Yamauchi, T. & Kadowaki, T. Identification of 28 new susceptibility loci for type 2 diabetes in the Japanese population. *Nature Genetics* **51**, 379–386 (2019).
- 62. Hirata, J., Hosomichi, K., Sakaue, S., **Kanai, M.**, Nakaoka, H., ..., Momozawa, Y., Inoue, I., Kubo, M., Kamatani, Y. & Okada, Y. Genetic and phenotypic landscape of the major histocompatibilty complex region in the Japanese population. *Nature Genetics* **51**, 470–480 (2019).

- 63. **Kanai, M.**, Maeda, Y. & Okada, Y. Grimon: graphical interface to visualize multi-omics networks. *Bioinformatics* **34**, 3934–3936 (2018).
- 64. Horikoshi, M., Day, F. R., Akiyama, M., Hirata, M., Kamatani, Y., ..., **Kanai, M.**, ..., Ojeda, S. R., Lomniczi, A., Kubo, M., Ong, K. K. & Perry, J. R. B. Elucidating the genetic architecture of reproductive ageing in the Japanese population. *Nature Communications* **9**, 1977 (2018).
- 65. Okada, Y., Momozawa, Y., Sakaue, S., **Kanai, M.**, Ishigaki, K., ..., Matsuda, K., Yamamoto, K., Kubo, M., Hirose, N. & Kamatani, Y. Deep whole-genome sequencing reveals recent selection signatures linked to evolution and disease risk of Japanese. *Nature Communications* **9**, 1631 (2018).
- Malik, R., Chauhan, G., Traylor, M., Sargurupremraj, M., Okada, Y., ..., Kanai, M., ..., Markus, H. S., Howson, J. M. M., Kamatani, Y., Debette, S. & Dichgans, M. Multiancestry genome-wide association study of 520,000 subjects identifies 32 loci associated with stroke and stroke subtypes. *Nature Genetics* 50, 524–537 (2018).
- 67. Hirata, J., Hirota, T., Ozeki, T., **Kanai, M.**, Sudo, T., ..., Sato, S., Mushiroda, T., Saeki, H., Tamari, M. & Okada, Y. Variants at HLA-A, HLA-C, and HLA-DQB1 confer risk of psoriasis vulgaris in Japanese. *Journal of Investigative Dermatology* **138**, 542–548 (2018).
- 68. Akiyama, M., Okada, Y., **Kanai, M.**, Takahashi, A., Momozawa, Y., ..., Minegishi, N., Tsugane, S., Yamamoto, M., Kubo, M. & Kamatani, Y. Genome-wide association study identifies 112 new loci for body mass index in the Japanese population. *Nature Genetics* 49, 1458–1467 (2017).
- Sudo, T., Okada, Y., Ozaki, K., Urayama, K., Kanai, M., Kobayashi, H., Gokyu, M., Izumi, Y. & Tanaka, T. Association of NOD2 Mutations with Aggressive Periodontitis. *Journal of Dental Research* 96, 1100–1105 (2017).
- 70. Okada, Y., Suzuki, A., Ikari, K., Terao, C., Kochi, Y., ..., **Kanai, M.**, ..., Raychaudhuri, S., Matsuda, F., Yamanaka, H., Kamatani, Y. & Yamamoto, K. Contribution of a Non-classical HLA Gene, HLA-DOA, to the Risk of Rheumatoid Arthritis. *The American Journal of Human Genetics* **99**, 366–374 (2016).
- 71. **Kanai, M.**, Tanaka, T. & Okada, Y. Empirical estimation of genome-wide significance thresholds based on the 1000 Genomes Project data set. *Journal of Human Genetics* **61**, 861–866 (2016).
- 72. Okada, Y., Muramatsu, T., Suita, N., **Kanai, M.**, Kawakami, E., lotchkova, V., Soranzo, N., Inazawa, J. & Tanaka, T. Significant impact of miRNA-target gene networks on genetics of human complex traits. *Scientific Reports* 6, 22223 (2016).
- 73. Okada, Y., Momozawa, Y., Ashikawa, K., **Kanai, M.**, Matsuda, K., Kamatani, Y., Takahashi, A. & Kubo, M. Construction of a population-specific HLA imputation reference panel and its application to Graves' disease risk in Japanese. *Nature Genetics* **47**, 798–802 (2015).

# Reviews (in Japanese).

- 1. **Kanai, M.** Genetic analysis of quantitative traits with integration of multi-omics data. *The Medical Frontline (SAISHIN IGAKU)* **74**, 220–226 (2019).
- 2. **Kanai, M.**, Okada, Y. & Kamatani, Y. Genetics of clinical measurements in the Japanese population: large-scale genome-wide association study of 58 quantitative traits. *Clinical Immunology & Allergology* **71**, 78–85 (2019).
- 3. **Kanai, M.**, Okada, Y. & Kamatani, Y. Large-scale genome-wide association study in the Japanese population elucidates genetic backgrounds of 58 quantitative traits. *Life Science First Author's Review* (2018).
- 4. **Kanai, M.** & Okada, Y. Identification of genetic loci associated with chronic kidney disease and applications for drug development. *Kidney and Metabolic Bone Diseases* **31**, 19–26 (2018).
- 5. **Kanai, M.** & Okada, Y. HLA imputation: construction of a population-specific reference panel in Japanese. *Journal of Clinical and Experimental Medicine (IGAKU NO AYUMI)* **257**, 939–940 (2016).

## **Presentations**

## International Conference.....

1. **Kanai, M.**, Ulirsch, J. C., Karjalainen, J., Kurki, M., Ishigaki, K., Kamatani, Y., Reilly, S., Sabeti, P., Tewhey, R., FinnGen, Okada, Y., Daly, M. J. & Finucane, H. K. Insights into fine-mapping causal variants of complex traits from diverse populations. *The 70th Annual Meeting of the American Society of Human Genetics, Plenary Session* (2020).

- 2. **Kanai, M.**, Ulirsch, J. C., McCaw, Z. R., Albors, C., Fan, Z., Ishigaki, K., Karjalainen, J., Kurki, M., FinnGen, Kamatani, Y., Okada, Y., Daly, M. J. & Finucane, H. K. Cross-population fine-mapping of 50 complex traits and diseases in 675,000 individuals across three global biobanks. *The 69th Annual Meeting of the American Society of Human Genetics, Poster Session* (2019).
- 3. **Kanai, M.**, Akiyama, M., Okada, Y., Ikeda, M., Iwata, N., Kubo, M. & Kamatani, Y. Trans-ethnic comparison of partitioned heritability reveals shared cell-type specific enrichment between East Asian and European genome-wide association studies. *The 66th Annual Meeting of the American Society of Human Genetics, Poster Session* (2016).
- Kanai, M., Tanaka, T. & Okada, Y. Empirical estimation of genome-wide significance thresholds based on the 1000 Genomes Project. The 13th International Congress of Human Genetics, Oral Session (2016).
- 5. **Kanai, M.**, Okada, Y., Muramatsu, T., Suita, N., Kawakami, E., lotchkova, V., Soranzo, N., Inazawa, J. & Tanaka, T. Significant impact of miRNA-target gene networks on genetics of human complex traits. *The 13th International Congress of Human Genetics, Oral Session* (2016).

#### Domestic Conference (in Japan).....

- Kanai, M., Tanaka, T. & Okada, Y. Empirical estimation of a genome-wide significance threshold based on the 1000 Genomes Project. The 60th Annual Meeting of the Japan Society of Human Genetics, Oral Session (2015).
- 2. **Kanai, M.**, Yamane, K., Higuchi, C., Tanaka, T. & Okada, Y. Performance evaluation of PLINK v.1.90: a next version of a tool set for genome-wide association study. *The 59th Annual Meeting of the Japan Society of Human Genetics, Poster Session* (2014).

Last updated: November 5, 2022.