

# Masahiro Kanai, Ph.D.

## Curriculum Vitae

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### Education

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#### Harvard University

Ph.D. in Biomedical Informatics

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

**Boston, USA**

Aug. 2017–May 2022

#### Keio University

B.S. in Bioinformatics, Department of Biosciences and Informatics

**Yokohama, Japan**

Apr. 2013–Mar. 2017

### Research Experience

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#### Massachusetts General Hospital & Broad Institute of MIT and Harvard

Research Fellow, Center for Computational and Integrative Biology

**Boston, USA**

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

Graduate Student, Analytic and Translational Genetics Unit

**Boston, USA**

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

Visiting Fellow, Department of Statistical Genetics

Research Student, Department of Statistical Genetics

**Osaka, Japan**

July 2022–present

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-dimensional multi-omics data

#### RIKEN Center for Integrative Medical Sciences

Research Assistant, Laboratory for Statistical Analysis

**Yokohama, Japan**

May 2015–Jul. 2019

**Advisors:** Drs. Yoichiro Kamatani & Yukinori Okada

##### Research themes:

- Genome-wide association study (GWAS) of ~200,000 individuals in the BioBank Japan Project
- Interpretation of GWAS polygenic signals using epigenomic data
- Population genetics analysis of 1,037 Japanese whole-genome sequences

#### Keio University

Undergraduate Researcher, Laboratory for Bioinformatics

**Yokohama, Japan**

Apr. 2016–Mar. 2017

**Advisor:** Dr. Yasubumi Sakakibara

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

**Tokyo Medical and Dental University**

Technical Assistant, Department of Human Genetics and Disease Diversity

**Tokyo, Japan**

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**

The American Society of Human Genetics 2022 Annual Meeting

**Los Angeles, USA**

2022

**Reviewers' Choice Abstract Award**

The American Society of Human Genetics 2021 Virtual Meeting

**USA**

2021

**Charles J. Epstein Trainee Awards for Excellence in Human Genetics Research**

Finalist (Predoctoral), The American Society of Human Genetics 2020 Virtual Meeting

**USA**

2020

**Reviewers' Choice Abstract Award**

The American Society of Human Genetics 2019 Annual Meeting

**Houston, USA**

2019

**Masason Foundation Fellowship**

Research grant for cross-population analysis of complex human diseases and traits.

**Tokyo, Japan**

2018–2023

**Nakajima Foundation Fellowship**

Predoctoral fellowship which covers up to ~\$50,000/year for tuition and stipend.

**Tokyo, Japan**

2017–2022

**The 3rd place, Worldwide Finals**

Microsoft Imagine Cup 2011 Windows 7 Touch Challenge

**New York, USA**

2011

**The 1st place, National Finals & Worldwide Finalist**

Microsoft Imagine Cup 2010 Software Design Competition

**Japan & Poland**

2010

**Publications****Selected Publications and Preprints**

(\* = co-first)

1. \*Karczewski, K. J., \*Gupta, R., \***Kanai, M.**, Lu, W., Tsuo, K., ..., Finucane, H. K., Daly, M. J., Neale, B. M., Atkinson, E. G. & Martin, A. R. Pan-UK Biobank GWAS improves discovery, analysis of genetic architecture, and resolution into ancestry-enriched effects. *medRxiv* (2024).
2. **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).
3. \*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).

4. **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).
5. \*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).
6. The COVID-19 Host Genetics Initiative. Mapping the human genetic architecture of COVID-19. *Nature* **600**, 472–477 (2021).
7. \*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).
8. 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).
9. **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

10. Zhou, W., Cuomo, A., Xue, A., **Kanai, M.**, Chau, G., ..., Xavier, R. J., MacArthur, D. G., Powell, J. E., Daly, M. J. & Neale, B. M. Efficient and accurate mixed model association tool for single-cell eQTL analysis. *medRxiv* (2024).
11. Rossen, J., Shi, H., Strober, B. J., Zhang, M. J., **Kanai, M.**, McCaw, Z. R., Liang, L., Weissbrod, O. & Price, A. L. MultiSuSiE improves multi-ancestry fine-mapping in All of Us whole-genome sequencing data. *medRxiv* (2024).
12. Siraj, L., Castro, R. I., Dewey, H., Kales, S., Nguyen, T. T. L., **Kanai, M.**, ..., Sabeti, P. C., Finucane, H. K., Reilly, S. K., Ulirsch, J. C. & Tewhey, R. Functional dissection of complex and molecular trait variants at single nucleotide resolution. *bioRxiv* (2024).
13. The Psychiatric Genomics Consortium Major Depressive Disorder Working Group. Genome-wide study of major depression in 685,808 diverse individuals identifies 697 independent associations, infers causal neuronal subtypes and biological targets for novel pharmacotherapies. *medRxiv* (2024).
14. Pozarickij, A., Gan, W., Lin, K., Clarke, R., Fairhurst-Hunter, Z., ..., **Kanai, M.**, ..., Collins, R., Li, L., Chen, Z., Millwood, I. Y. & Walters, R. G. Causal relevance of different blood pressure traits on risk of cardiovascular diseases: GWAS and Mendelian randomisation in 100,000 Chinese adults. *medRxiv* (2023).
15. 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).
16. 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).

#### Other Publications

17. Carey, C. E., Shafee, R., Wedow, R., Elliott, A., Palmer, D. S., ..., **Kanai, M.**, ..., King, D., Davey Smith, G., Neale, B. M., Walters, R. K. & Robinson, E. B. Principled distillation of UK Biobank phenotype data reveals underlying structure in human variation. *Nature Human Behaviour* (2024).

18. Trsan, T., Peng, V., Krishna, C., Ohara, T. E., Beatty, W. L., ..., **Kanai, M.**, ..., Jaiswal, A., Stappenbeck, T. S., Daly, M. J., Xavier, R. J. & Colonna, M. The centrosomal protein FGFR1OP controls myosin function in murine intestinal epithelial cells. *Developmental Cell* (2024).
19. Jermy, B., Läll, K., Wolford, B. N., Wang, Y., Zguro, K., ..., **Kanai, M.**, ..., Palotie, A., Heyne, H., Mars, N., Ganna, A. & Ripatti, S. A unified framework for estimating country-specific cumulative incidence for 18 diseases stratified by polygenic risk. *Nature Communications* **15**, 5007 (2024).
20. Sakaue, S., Weinand, K., Isaac, S., Dey, K. K., Jagadeesh, K., **Kanai, M.**, ..., McDavid, A., Donlin, L. T., Wei, K., Price, A. L. & Raychaudhuri, S. Tissue-specific enhancer–gene maps from multimodal single-cell data identify causal disease alleles. *Nature Genetics* **56**, 615–626 (2024).
21. De Vincentis, A., Tavaglione, F., Namba, S., **Kanai, M.**, Okada, Y., ..., Pedone, C., Antonelli Incalzi, R., Valenti, L., Romeo, S. & Vespasiani-Gentilucci, U. Poor accuracy and sustainability of the first-step FIB4 EASL pathway for stratifying steatotic liver disease risk in the general population. *Alimentary Pharmacology & Therapeutics* **59**, 1402–1412 (2024).
22. Lo Faro, V., Bhattacharya, A., Zhou, W., Zhou, D., Wang, Y., ..., **Kanai, M.**, ..., Gamazon, E. R., Janssonius, N. M., Joos, K., Cox, N. J. & Hirbo, J. Novel ancestry-specific primary open-angle glaucoma loci and shared biology with vascular mechanisms and cell proliferation. *Cell Reports Medicine* **5**, 101430 (2024).
23. Scholz, M., Horn, K., Pott, J., Wuttke, M., Kühnapfel, A., ..., **Kanai, M.**, ..., Franceschini, N., Parsa, A., Köttgen, A., Schlosser, P. & Pattaro, C. X-chromosome and kidney function: evidence from a multi-trait genetic analysis of 908,697 individuals reveals sex-specific and sex-differential findings in genes regulated by androgen response elements. *Nature Communications* **15**, 586 (2024).
24. Meng, X., Navoly, G., Giannakopoulou, O., Levey, D. F., Koller, D., ..., **Kanai, M.**, ..., Dunn, E. C., Stein, M. B., Gelernter, J., Lewis, C. M. & Kuchenbaecker, K. Multi-ancestry genome-wide association study of major depression aids locus discovery, fine mapping, gene prioritization and causal inference. *Nature Genetics* **56**, 222–233 (2024).
25. Chen, S., Francioli, L. C., Goodrich, J. K., Collins, R. L., **Kanai, M.**, ..., Daly, M. J., Tiao, G., Neale, B. M., MacArthur, D. G. & Karczewski, K. J. A genomic mutational constraint map using variation in 76,156 human genomes. *Nature* **625**, 92–100 (2024).
26. 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. *Nature Genetics* **56**, 162–169 (2024).
27. Kerimov, N., Tambets, R., Hayhurst, J. D., Rahu, I., Kolberg, P., ..., **Kanai, M.**, ..., Finucane, H., Peterson, H., Mosaku, A., Parkinson, H. & Alasoo, K. eQTL Catalogue 2023: New datasets, X chromosome QTLs, and improved detection and visualisation of transcript-level QTLs. *PLoS Genetics* **19**, e1010932 (2023).
28. Wang, Y., **Kanai, M.**, Tan, T., Kamariza, M., Tsuo, K., ..., BioBank Japan Project, Huang, H., Turley, P., Atkinson, E. G. & Martin, A. R. Polygenic prediction across populations is influenced by ancestry, genetic architecture, and methodology. *Cell Genomics* **3**, 100408 (2023).
29. The COVID-19 Host Genetics Initiative. A second update on mapping the human genetic architecture of COVID-19. *Nature* **621**, E7–E26 (2023).
30. International League Against Epilepsy Consortium on Complex Epilepsies. GWAS meta-analysis of over 29,000 people with epilepsy identifies 26 risk loci and subtype-specific genetic architecture. *Nature Genetics* **55**, 1471–1482 (2023).
31. Gupta, R., **Kanai, M.**, Durham, T. J., Tsuo, K., McCoy, J. G., ..., Chinnery, P. F., Karczewski, K. J., Calvo, S. E., Neale, B. M. & Mootha, V. K. Nuclear genetic control of mtDNA copy number and heteroplasmy in humans. *Nature* **620**, 839–848 (2023).

32. **Kanai, M.** Leveraging fine-scale population structures for precision healthcare. *Nature Medicine* **29**, 1611–1612 (2023).
33. 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. *Nature Genetics* **55**, 1267–1276 (2023).
34. Palmer, D. S., Zhou, W., Abbott, L., Wigdor, E. M., Baya, N., ..., Poterba, T., King, D., **Kanai, M.**, Bloemendal, A. & Neale, B. M. Analysis of genetic dominance in the UK Biobank. *Science* **379**, 1341–1348 (2023).
35. Ogawa, K., Tsoi, L. C., Tanaka, H., **Kanai, M.**, Stuart, P. E., Nair, R. P., Tanaka, Y., Mochizuki, H., Elder, J. T. & Okada, Y. A cross-trait genetic correlation study identified eight diseases and traits associated with psoriasis. *Journal of Investigative Dermatology* **143**, 1813–1816.e2 (2023).
36. Rämö, J. T., Kiiskinen, T., Seist, R., Krebs, K., **Kanai, M.**, ..., Milani, L., Stankovic, K. M., Mäkitie, A., Daly, M. J. & Palotie, A. Genome-wide screen of otosclerosis in population biobanks: 27 loci and shared associations with skeletal structure. *Nature Communications* **14**, 157 (2023).
37. 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 provides genetic insights from a well-phenotyped isolated population. *Nature* **613**, 508–518 (2023).
38. Wang, Y., Namba, S., Lopera, E., Kerminen, S., Tsuo, K., ..., **Kanai, M.**, ..., Cox, N. J., Surakka, I., Okada, Y., Martin, A. R. & Hirbo, J. Global Biobank analyses provide lessons for developing polygenic risk scores across diverse cohorts. *Cell Genomics* **3**, 100241 (2023).
39. 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. *Genome Biology* **23**, 268 (2022).
40. Tsuo, K., Zhou, W., Wang, Y., **Kanai, M.**, Namba, S., ..., Okada, Y., Neale, B. M., Global Biobank Meta-analysis Initiative, Daly, M. J. & Martin, A. R. Multi-ancestry meta-analysis of asthma identifies novel associations and highlights the value of increased power and diversity. *Cell Genomics* **2**, 100212 (2022).
41. 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).
42. 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).
43. 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).
44. 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).
45. 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 MFG8 associate with protection against coronary atherosclerosis. *Communications Biology* **5**, 802 (2022).



46. 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).
47. 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).
48. The COVID-19 Host Genetics Initiative. A first update on mapping the human genetic architecture of COVID-19. *Nature* **608**, E1–E10 (2022).
49. 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).
50. 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).
51. 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).
52. 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).
53. 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).
54. 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).
55. 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).
56. 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).
57. 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).
58. 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**, 3394 (2021).
59. 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).

60. 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).
61. 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).
62. 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).
63. 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).
64. 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).
65. 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).
66. 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).
67. 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).
68. 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).
69. 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).
70. 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).
71. 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).
72. 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).
73. 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).
74. 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).

75. \*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).
76. 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).
77. 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).
78. 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 histocompatibility complex region in the Japanese population. *Nature Genetics* **51**, 470–480 (2019).
79. **Kanai, M.**, Maeda, Y. & Okada, Y. Grimon: graphical interface to visualize multi-omics networks. *Bioinformatics* **34**, 3934–3936 (2018).
80. 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).
81. 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).
82. 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).
83. 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).
84. 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).
85. 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).
86. 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).
87. **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).
88. Okada, Y., Muramatsu, T., Suita, N., **Kanai, M.**, Kawakami, E., Iotchkova, 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).
89. 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 & Allergy* **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., Albers, 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).
4. **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., Iotchkova, 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).....

1. **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: July 7, 2024.