

Masahiro Kanai, Ph.D.

Curriculum Vitae

✉ mkanai@broadinstitute.org
🌐 mkanai.github.io
🔗 mkanai
🐦 masakanai

Education

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

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 Faculty, 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**Innovators Under 35 Japan**

MIT Technology Review

Tokyo, Japan

2024

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. Liu, K., Rasmussen, A., Han, W., Gong, Q., Bohley, S., ..., **Kanai, M.**, ..., Ashenberg, O., Daly, M. J., Chen, F., Graham, D. B. & Xavier, R. J. Sensitive Transcriptomics and Genotyping reveals function of genetic variants in immunity. *bioRxiv* (2025).
11. Finucane, H. K., Parsa, S., Guez, J., **Kanai, M.**, Satterstrom, F. K., Nkambule, L. L., Daly, M. J., Seed, C. & Karczewski, K. J. Variant scoring performance across selection regimes depends on variant-to-gene and gene-to-disease components. *bioRxiv* (2024).
12. Jacobs, H., Gorissen, B., Guez, J., **Kanai, M.**, Finucane, H., Karczewski, K. & Burge, C. Widespread naturally variable human exons aid genetic interpretation. *bioRxiv* (2024).
13. 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).
14. 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).
15. 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).
16. 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).
17. 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.....

18. Sonehara, K., Uwamino, Y., Saiki, R., Takeshita, M., Namba, S., ..., **Kanai, M.**, ..., Murata, M., Matsushita, H., Ogawa, S., Okada, Y. & Namkoong, H. Germline variants and mosaic chromosomal alterations affect COVID-19 vaccine immunogenicity. *Cell Genomics* (2025).

19. Adams, M. J., Streit, F., Meng, X., Awasthi, S., Adey, B. N., ..., **Kanai, M.**, ..., Kuchenbaecker, K., Wray, N. R., Ripke, S., Lewis, C. M. & McIntosh, A. M. Trans-ancestry genome-wide study of depression identifies 697 associations implicating cell types and pharmacotherapies. *Cell* (2025).
20. Pozarickij, A., Gan, W., Lin, K., Clarke, R., Fairhurst-Hunter, Z., ..., **Kanai, M.**, ..., Li, L., Chen, Z., Millwood, I. Y., Walters, R. G. & China Kadoorie Biobank Collaborative Group. Causal relevance of different blood pressure traits on risk of cardiovascular diseases: GWAS and Mendelian randomisation in 100,000 Chinese adults. *Nature Communications* **15**, 6265 (2024).
21. 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* **8**, 1599–1615 (2024).
22. 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* **59**, 2460–2476 (2024).
23. 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).
24. 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).
25. 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).
26. 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. Novel ancestry-specific primary open-angle glaucoma loci and shared biology with vascular mechanisms and cell proliferation. *Cell Reports Medicine* **5**, 101430 (2024).
27. 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).
28. 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).
29. 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).
30. 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).
31. 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).

32. 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).
33. The COVID-19 Host Genetics Initiative. A second update on mapping the human genetic architecture of COVID-19. *Nature* **621**, E7–E26 (2023).
34. 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).
35. 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).
36. **Kanai, M.** Leveraging fine-scale population structures for precision healthcare. *Nature Medicine* **29**, 1611–1612 (2023).
37. 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).
38. 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).
39. 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).
40. 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).
41. 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).
42. 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).
43. 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).
44. 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).
45. 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).
46. 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).

47. 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).
48. 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).
49. 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 MFE8 associate with protection against coronary atherosclerosis. *Communications Biology* **5**, 802 (2022).
50. 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).
51. 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).
52. The COVID-19 Host Genetics Initiative. A first update on mapping the human genetic architecture of COVID-19. *Nature* **608**, E1–E10 (2022).
53. 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).
54. 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).
55. 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).
56. 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).
57. 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).
58. 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).
59. 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).
60. 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).

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

75. 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).
76. 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).
77. 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).
78. 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).
79. *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).
80. 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).
81. 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).
82. 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).
83. **Kanai, M.**, Maeda, Y. & Okada, Y. Grimon: graphical interface to visualize multi-omics networks. *Bioinformatics* **34**, 3934–3936 (2018).
84. 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).
85. 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).
86. 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. Multi-ancestry genome-wide association study of 520,000 subjects identifies 32 loci associated with stroke and stroke subtypes. *Nature Genetics* **50**, 524–537 (2018).
87. 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).
88. 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).
89. 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).

90. 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).
91. **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).
92. 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).
93. 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).

Last updated: March 15, 2025.