Masahiro Kanai, Ph.D.

Curriculum Vitae

mkanai@broadinstitute.org
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**
B.S. in Bioinformatics, Department of Biosciences and Informatics — *Apr. 2013–Mar. 2017*

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**
Research Assistant, Laboratory for Statistical Analysis — *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 — **Yokohama, Japan**
Undergraduate Researcher, Laboratory for Bioinformatics — *Apr. 2016–Mar. 2017*
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**
Predoctoral fellowship which covers up to ~$50,000/year for tuition and stipend. *2017–2022*

The 3rd place, Worldwide Finals **New York, USA**
Microsoft Imagine Cup 2011 Windows 7 Touch Challenge *2011*

The 1st place, National Finals & Worldwide Finalist **Japan & Poland**
Microsoft Imagine Cup 2010 Software Design Competition *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. 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. ***medRxiv*** (2023).
11. 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).
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. 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).
14. 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

15. 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*** (2024).
16. 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*** (2024).
17. 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***, 101430 (2024).

18. 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).
19. 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).
20. 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).
21. 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).
22. 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).
23. 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).
24. The COVID-19 Host Genetics Initiative. A second update on mapping the human genetic architecture of COVID-19. ***Nature*** **621,** E7–E26 (2023).
25. 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).
26. 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).
27. **Kanai, M.** Leveraging fine-scale population structures for precision healthcare. ***Nature Medicine*** **29,** 1611–1612 (2023).
28. 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).
29. 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).
30. 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).
31. 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).
32. 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).

33. 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).

34. 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).

35. 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).

36. 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).

37. 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).

38. 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).

39. 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).

40. 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).

41. 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).

42. 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).

43. The COVID-19 Host Genetics Initiative. A first update on mapping the human genetic architecture of COVID-19. ***Nature*** **608,** E1–E10 (2022).

44. 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).

45. 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).

46. 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).

47. 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).

48. 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).

49. 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).

50. 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).

51. 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).

52. 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).

53. 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).

54. 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).

55. 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).

56. 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).

57. 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).

58. 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).

59. 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).

60. 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).

61. 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).
62. 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).
63. 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).
64. 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).
65. 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).
66. 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).
67. 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).
68. 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).
69. 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).
70. *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).
71. 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).
72. 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).
73. 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).
74. **Kanai, M.**, Maeda, Y. & Okada, Y. Grimon: graphical interface to visualize multi-omics networks. ***Bioinformatics*** **34,** 3934–3936 (2018).
75. 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).

76. 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).
77. 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).
78. 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).
79. 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).
80. 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).
81. 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).
82. **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).
83. 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).
84. 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).
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: April 9, 2024.