

# 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 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. Reeve, M., **Kanai, M.**, Graham, D., Karjalainen, J., Luo, S., ..., Kero, J., Xavier, R., Daly, M., Ripatti, S. & FinnGen. Autoimmune hypothyroidism GWAS reveals independent autoimmune and thyroid-specific contributions and an inverse relation with cancer risk. *Research Square* (2024).
14. 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).
15. 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).
16. 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).
17. 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).
18. 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.....

19. 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* **5**, 100783 (2025).
20. 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* **188**, 640–652.e9 (2025).
21. 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).
22. 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).
23. 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).
24. 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).
25. 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).
26. 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).
27. 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).
28. 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).
29. 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).
30. 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).
31. 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).

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



47. 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).
48. 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).
49. 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).
50. 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).
51. 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).
52. 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).
53. The COVID-19 Host Genetics Initiative. A first update on mapping the human genetic architecture of COVID-19. *Nature* **608**, E1–E10 (2022).
54. 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).
55. 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).
56. 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).
57. 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).
58. 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).
59. 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).
60. 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).

61. 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).
62. Reilly, S. K., Gosai, S. J., Guterrez, 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).
63. 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).
64. 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).
65. 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).
66. 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).
67. 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).
68. 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).
69. 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).
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Last updated: March 27, 2025.