

# 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. \*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).
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. *medRxiv* (2022).
3. **Kanai, M.**, Ulirsch, J. C., Karjalainen, J., Kurki, M., Karczewski, K. J., ..., Tewhey, R., Sabeti, P. C., Okada, Y., Daly, M. J. & Finucane, H. K. Insights from complex trait fine-mapping across diverse populations. *medRxiv* (2021).
4. \*Sakaue, S., \***Kanai, M.**, Tanigawa, Y., Karjalainen, J., Kurki, M., ..., Palotie, A., Rivas, M. A., Daly, M. J., Matsuda, K. & Okada, Y. A cross-population atlas of genetic associations for 220 human phenotypes. *Nature Genetics* **53**, 1415–1424 (2021).

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

### Other Preprints.....

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

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

### Other Publications.....

25. Zhou, W., **Kanai, M.**, Wu, K.-H. H., Rasheed, H., Tsuo, K., ..., Zöllner, S., Martin, A. R., Willer, C. J., Daly, M. J. & Neale, B. M. Global Biobank Meta-analysis Initiative: Powering genetic discovery across human disease. *Cell Genomics* **2**, 100192 (2022).
26. Bhattacharya, A., Hirbo, J. B., Zhou, D., Zhou, W., Zheng, J., **Kanai, M.**, The Global Biobank Meta-analysis Initiative, Pasaniuc, B., Gamazon, E. R. & Cox, N. J. Best practices for multi-ancestry, meta-analytic transcriptome-wide association studies: Lessons from the Global Biobank Meta-analysis Initiative. *Cell Genomics* **2**, 100180 (2022).
27. Yengo, L., Vedantam, S., Marouli, E., Sidorenko, J., Bartell, E., ..., **Kanai, M.**, ..., Frayling, T. M., Okada, Y., Wood, A. R., Visscher, P. M. & Hirschhorn, J. N. A saturated map of common genetic variants associated with human height. *Nature* **610**, 704–712 (2022).
28. Wang, Q. S., Edahiro, R., Namkoong, H., Hasegawa, T., Shirai, Y., ..., **Kanai, M.**, ..., Miyano, S., Ogawa, S., Kanai, T., Fukunaga, K. & Okada, Y. The whole blood transcriptional regulation landscape in 465 COVID-19 infected samples from Japan COVID-19 Task Force. *Nature Communications* **13**, 4830 (2022).
29. Ruotsalainen, S. E., Surakka, I., Mars, N., Karjalainen, J., Kurki, M., **Kanai, M.**, ..., Okada, Y., Palotie, A., Widen, E., Daly, M. J. & Ripatti, S. Inframe insertion and splice site variants in MFGE8 associate with protection against coronary atherosclerosis. *Communications Biology* **5**, 802 (2022).
30. Namkoong, H., Edahiro, R., Takano, T., Nishihara, H., Shirai, Y., ..., **Kanai, M.**, ..., Miyano, S., Ogawa, S., Kanai, T., Fukunaga, K. & Okada, Y. DOK2 is involved in the host genetics and biology of severe COVID-19. *Nature* **609**, 754–760 (2022).
31. Ramdas, S., Judd, J., Graham, S. E., Kanoni, S., Wang, Y., ..., **Kanai, M.**, ..., Peloso, G., Assimes, T. L., Willer, C. J., Zhu, X. & Brown, C. D. A multi-layer functional genomic analysis to understand noncoding genetic variation in lipids. *The American Journal of Human Genetics* **109**, 1366–1387 (2022).
32. The COVID-19 Host Genetics Initiative. A first update on mapping the human genetic architecture of COVID-19. *Nature* **608**, E1–E10 (2022).
33. Winkler, T. W., Rasheed, H., Teumer, A., Gorski, M., Rowan, B. X., ..., **Kanai, M.**, ..., Hung, A. M., Kronenberg, F., Köttgen, A., Pattaro, C. & Heid, I. M. Differential and shared genetic effects on kidney function between diabetic and non-diabetic individuals. *Communications Biology* **5**, 580 (2022).
34. Wang, Y., Tsuo, K., **Kanai, M.**, Neale, B. M. & Martin, A. R. Challenges and Opportunities for Developing More Generalizable Polygenic Risk Scores. *Annual Review of Biomedical Data Science* **5**, 293–320 (2022).

35. Mars, N., Kerminen, S., Feng, Y.-C. A., **Kanai, M.**, Läll, K., ..., Pirinen, M., Palotie, A., Ganna, A., Martin, A. R. & Ripatti, S. Genome-wide risk prediction of common diseases across ancestries in one million people. *Cell Genomics* **2**, 100118 (2022).
36. Zheng, J., Zhang, Y., Rasheed, H., Walker, V., Sugawara, Y., ..., **Kanai, M.**, ..., Barbour, S., Yu, C., Åsvold, B. O., Zhang, H. & Gaunt, T. R. Trans-ethnic Mendelian-randomization study reveals causal relationships between cardiometabolic factors and chronic kidney disease. *International Journal of Epidemiology* **50**, 1995–2010 (2022).
37. Graham, S. E., Clarke, S. L., Wu, K.-H. H., Kanoni, S., Zajac, G. J. M., ..., **Kanai, M.**, ..., Morris, A. P., Assimes, T. L., Deloukas, P., Sun, Y. V. & Willer, C. J. The power of genetic diversity in genome-wide association studies of lipids. *Nature* **600**, 675–679 (2021).
38. Polygenic Risk Score Task Force of the International Common Disease Alliance. Responsible use of polygenic risk scores in the clinic: potential benefits, risks and gaps. *Nature Medicine* **27**, 1876–1884 (2021).
39. Luo, Y., **Kanai, M.**, Choi, W., Li, X., Sakaue, S., ..., Esko, T., Okada, Y., Han, B., McLaren, P. J. & Raychaudhuri, S. A high-resolution HLA reference panel capturing global population diversity enables multi-ancestry fine-mapping in HIV host response. *Nature Genetics* **53**, 1504–1516 (2021).
40. Griesemer, D., Xue, J. R., Reilly, S. K., Ulirsch, J. C., Kukreja, K., ..., **Kanai, M.**, ..., Montgomery, S. B., Finucane, H. K., Novina, C. D., Tewhey, R. & Sabeti, P. C. Genome-wide functional screen of 3'UTR variants uncovers causal variants for human disease and evolution. *Cell* **184**, 5247–5260.e19 (2021).
41. Reilly, S. K., Gosai, S. J., Guiterrez, A., Ulirsch, J. C., **Kanai, M.**, ..., Butler, G. B., Gladden-Young, A., Finucane, H. K., Sabeti, P. C. & Tewhey, R. Direct characterization of cis-regulatory elements and functional dissection of complex genetic associations using HCR-FlowFISH. *Nature Genetics* **53**, 1166–1176 (2021).
42. Wang, Q. S., Kelley, D. R., Ulirsch, J., **Kanai, M.**, Sadhuka, S., ..., Okada, Y., Aguet, F., Ardlie, K. G., MacArthur, D. G. & Finucane, H. K. Leveraging supervised learning for functionally informed fine-mapping of cis-eQTLs identifies an additional 20,913 putative causal eQTLs. *Nature Communications* **12**, 1–11 (2021).
43. Nakatochi, M., Toyoda, Y., **Kanai, M.**, Nakayama, A., Kawamura, Y., ..., Yokota, M., Wakai, K., Okada, Y., Matsuo, H. & Japan Uric Acid Genomics Consortium (Japan Urate). An X chromosome-wide meta-analysis based on Japanese cohorts revealed that non-autosomal variations are associated with serum urate. *Rheumatology* **60**, 4430–4432 (2021).
44. Pirastu, N., Cordioli, M., Nandakumar, P., Mignogna, G., Abdellaoui, A., ..., **Kanai, M.**, ..., Neale, B. M., Walters, R. K., Nivard, M. G., Perry, J. R. B. & Ganna, A. Genetic analyses identify widespread sex-differential participation bias. *Nature Genetics* **53**, 663–671 (2021).
45. Shi, H., Gazal, S., **Kanai, M.**, Koch, E. M., Schoech, A. P., ..., Huang, H., Okada, Y., Raychaudhuri, S., Sunyaev, S. R. & Price, A. L. Population-specific causal disease effect sizes in functionally important regions impacted by selection. *Nature Communications* **12**, 1098 (2021).
46. Atkinson, E. G., Maihofer, A. X., **Kanai, M.**, Martin, A. R., Karczewski, K. J., ..., Finucane, H. K., Koenen, K. C., Nievergelt, C. M., Daly, M. J. & Neale, B. M. Tractor uses local ancestry to enable the inclusion of admixed individuals in GWAS and to boost power. *Nature Genetics* **53**, 195–204 (2021).
47. Chen, M.-H., Raffield, L. M., Mousas, A., Sakaue, S., Huffman, J. E., ..., **Kanai, M.**, ..., Soranzo, N., Johnson, A. D., Reiner, A. P., Auer, P. L. & Lettre, G. Trans-ethnic and Ancestry-Specific Blood-Cell Genetics in 746,667 Individuals from 5 Global Populations. *Cell* **182**, 1198–1213.e14 (2020).
48. Vuckovic, D., Bao, E. L., Akbari, P., Lareau, C. A., Mousas, A., ..., **Kanai, M.**, ..., Butterworth, A. S., Ouwehand, W. H., Lettre, G., Sankaran, V. G. & Soranzo, N. The Polygenic and Monogenic Basis of Blood Traits and Diseases. *Cell* **182**, 1214–1231.e11 (2020).



49. Shirai, Y., Honda, S., Ikari, K., **Kanai, M.**, Takeda, Y., ..., Morisaki, T., Tanaka, E., Kumanogoh, A., Harigai, M. & Okada, Y. Association of the RPA3-UMAD1 locus with interstitial lung diseases complicated with rheumatoid arthritis in Japanese. *Annals of the Rheumatic Diseases* **79**, 1305–1309 (2020).
50. Ishigaki, K., Akiyama, M., **Kanai, M.**, Takahashi, A., Kawakami, E., ..., Inazawa, J., Yamauchi, T., Kadowaki, T., Kubo, M. & Kamatani, Y. Large-scale genome-wide association study in a Japanese population identifies novel susceptibility loci across different diseases. *Nature Genetics* **52**, 669–679 (2020).
51. Sakaue, S., Hirata, J., **Kanai, M.**, Suzuki, K., Akiyama, M., ..., Hirata, M., Matsuda, K., Murakami, Y., Kamatani, Y. & Okada, Y. Dimensionality reduction reveals fine-scale structure in the Japanese population with consequences for polygenic risk prediction. *Nature Communications* **11**, 1569 (2020).
52. Ray, J. P., de Boer, C. G., Fulco, C. P., Lareau, C. A., **Kanai, M.**, ..., Issner, R., Finucane, H. K., Lander, E. S., Regev, A. & Hacohen, N. Prioritizing disease and trait causal variants at the TNFAIP3 locus using functional and genomic features. *Nature Communications* **11**, 1237 (2020).
53. Matoba, N., Akiyama, M., Ishigaki, K., **Kanai, M.**, Takahashi, A., ..., Matsuda, K., Murakami, Y., Kubo, M., Kamatani, Y. & Okada, Y. GWAS of 165,084 Japanese individuals identified nine loci associated with dietary habits. *Nature Human Behaviour* **4**, 308–316 (2020).
54. Clark, D. W., Okada, Y., Moore, K. H. S., Mason, D., Pirastu, N., ..., **Kanai, M.**, ..., Helgason, A., Stefansson, K., Joshi, P. K., Kubo, M. & Wilson, J. F. Associations of autozygosity with a broad range of human phenotypes. *Nature Communications* **10**, 4957 (2019).
55. Leu, C., Stevelink, R., Smith, A. W., Goleva, S. B., **Kanai, M.**, ..., Najm, I. M., Palotie, A., Daly, M. J., Busch, R. M. & Lal, D. Polygenic burden in focal and generalized epilepsies. *Brain* **142**, 3473–3481 (2019).
56. Tin, A., Marten, J., Halperin Kuhns, V. L., Li, Y., Wuttke, M., ..., **Kanai, M.**, ..., Teumer, A., Pattaro, C., Woodward, O. M., Vitart, V. & Köttgen, A. Target genes, variants, tissues and transcriptional pathways influencing human serum urate levels. *Nature Genetics* **51**, 1459–1474 (2019).
57. Akiyama, M., Ishigaki, K., Sakaue, S., Momozawa, Y., Horikoshi, M., ..., **Kanai, M.**, ..., Yamamoto, M., Okada, Y., Murakami, Y., Kubo, M. & Kamatani, Y. Characterizing rare and low-frequency height-associated variants in the Japanese population. *Nature Communications* **10**, 4393 (2019).
58. Wuttke, M., Li, Y., Li, M., Sieber, K. B., Feitosa, M. F., ..., **Kanai, M.**, ..., Heid, I. M., Scholz, M., Teumer, A., Köttgen, A. & Pattaro, C. A catalog of genetic loci associated with kidney function from analyses of a million individuals. *Nature Genetics* **51**, 957–972 (2019).
59. \*Nakatochi, M., **\*Kanai, M.**, \*Nakayama, A., \*Hishida, A., \*Kawamura, Y., ..., Shinomiya, N., Yokota, M., Wakai, K., Okada, Y. & Matsuo, H. Genome-wide meta-analysis identifies multiple novel loci associated with serum uric acid levels in Japanese individuals. *Communications Biology* **2**, 115 (2019).
60. Matoba, N., Akiyama, M., Ishigaki, K., **Kanai, M.**, Takahashi, A., ..., Hirata, M., Matsuda, K., Kubo, M., Okada, Y. & Kamatani, Y. GWAS of smoking behaviour in 165,436 Japanese people reveals seven new loci and shared genetic architecture. *Nature Human Behaviour* **3**, 471–477 (2019).
61. Suzuki, K., Akiyama, M., Ishigaki, K., **Kanai, M.**, Hosoe, J., ..., Kubo, M., Kamatani, Y., Horikoshi, M., Yamauchi, T. & Kadowaki, T. Identification of 28 new susceptibility loci for type 2 diabetes in the Japanese population. *Nature Genetics* **51**, 379–386 (2019).
62. Hirata, J., Hosomichi, K., Sakaue, S., **Kanai, M.**, Nakaoka, H., ..., Momozawa, Y., Inoue, I., Kubo, M., Kamatani, Y. & Okada, Y. Genetic and phenotypic landscape of the major histocompatibility complex region in the Japanese population. *Nature Genetics* **51**, 470–480 (2019).
63. **Kanai, M.**, Maeda, Y. & Okada, Y. Grimon: graphical interface to visualize multi-omics networks. *Bioinformatics* **34**, 3934–3936 (2018).

64. Horikoshi, M., Day, F. R., Akiyama, M., Hirata, M., Kamatani, Y., ..., **Kanai, M.**, ..., Ojeda, S. R., Lomniczi, A., Kubo, M., Ong, K. K. & Perry, J. R. B. Elucidating the genetic architecture of reproductive ageing in the Japanese population. *Nature Communications* **9**, 1977 (2018).
65. Okada, Y., Momozawa, Y., Sakaue, S., **Kanai, M.**, Ishigaki, K., ..., Matsuda, K., Yamamoto, K., Kubo, M., Hirose, N. & Kamatani, Y. Deep whole-genome sequencing reveals recent selection signatures linked to evolution and disease risk of Japanese. *Nature Communications* **9**, 1631 (2018).
66. 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).
67. Hirata, J., Hirota, T., Ozeki, T., **Kanai, M.**, Sudo, T., ..., Sato, S., Mushiroda, T., Saeki, H., Tamari, M. & Okada, Y. Variants at HLA-A, HLA-C, and HLA-DQB1 confer risk of psoriasis vulgaris in Japanese. *Journal of Investigative Dermatology* **138**, 542–548 (2018).
68. Akiyama, M., Okada, Y., **Kanai, M.**, Takahashi, A., Momozawa, Y., ..., Minegishi, N., Tsugane, S., Yamamoto, M., Kubo, M. & Kamatani, Y. Genome-wide association study identifies 112 new loci for body mass index in the Japanese population. *Nature Genetics* **49**, 1458–1467 (2017).
69. Sudo, T., Okada, Y., Ozaki, K., Urayama, K., **Kanai, M.**, Kobayashi, H., Gokyu, M., Izumi, Y. & Tanaka, T. Association of NOD2 Mutations with Aggressive Periodontitis. *Journal of Dental Research* **96**, 1100–1105 (2017).
70. Okada, Y., Suzuki, A., Ikari, K., Terao, C., Kochi, Y., ..., **Kanai, M.**, ..., Raychaudhuri, S., Matsuda, F., Yamanaka, H., Kamatani, Y. & Yamamoto, K. Contribution of a Non-classical HLA Gene, HLA-DOA, to the Risk of Rheumatoid Arthritis. *The American Journal of Human Genetics* **99**, 366–374 (2016).
71. **Kanai, M.**, Tanaka, T. & Okada, Y. Empirical estimation of genome-wide significance thresholds based on the 1000 Genomes Project data set. *Journal of Human Genetics* **61**, 861–866 (2016).
72. Okada, Y., Muramatsu, T., Suita, N., **Kanai, M.**, Kawakami, E., 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).
73. Okada, Y., Momozawa, Y., Ashikawa, K., **Kanai, M.**, Matsuda, K., Kamatani, Y., Takahashi, A. & Kubo, M. Construction of a population-specific HLA imputation reference panel and its application to Graves' disease risk in Japanese. *Nature Genetics* **47**, 798–802 (2015).

#### Reviews (in Japanese).....

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