# Masahiro Kanai

*Curriculum Vitae*

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## Education

**Harvard University** — **Boston, USA**
Bioinformatics and Integrative Genomics PhD Program, Harvard Medical School — *Aug. 2017–present*

**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**
*Graduate Student*, Analytic and Translational Genetics Unit — *Jan. 2018–present*
**Advisors**: Drs. Mark Daly & Hilary Finucane
**Research theme**: Cross-population analysis of complex human diseases and traits.

**Osaka University Graduate School of Medicine** — **Osaka, Japan**
*Research Student*, Department of Statistical Genetics — *Apr. 2017–present*
**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

**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. *2018–2021+*

**Nakajima Foundation Fellowship** **Tokyo, Japan**
Predoctoral fellowship which covers up to ~$50,000/year for tuition and stipend. *2017–2021+*

**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. *Weissbrod, O., ***Kanai, M.***, *Shi, H., Gazal, S., Peyrot, W., Khera, A., Okada, Y., The Biobank Japan Project, Martin, A., Finucane, H. & Price, A. L. Leveraging fine-mapping and non-European training data to improve trans-ethnic polygenic risk scores. ***medRxiv*** (2021).
2. *Sakaue, S., ***Kanai, M.***, Tanigawa, Y., Karjalainen, J., Kurki, M., Koshiba, S., Narita, A., Konuma, T., Yamamoto, K., Akiyama, M., Ishigaki, K., Suzuki, A., Suzuki, K., Obara, W., Yamaji, K., Takahashi, K., Asai, S., Takahashi, Y., Suzuki, T., Sinozaki, N., Yamaguchi, H., Minami, S., Murayama, S., Yoshimori, K., Nagayama, S., Obata, D., Higashiyama, M., Masumoto, A., Koretsune, Y., FinnGen, Ito, K., Terao, C., Yamauchi, T., Komuro, I., Kadowaki, T., Tamiya, G., Yamamoto, M., Nakamura, Y., Kubo, M., Murakami, Y., Yamamoto, K., Kamatani, Y., Palotie, A., Rivas, M. A., Daly, M., Matsuda, K. & Okada, Y. A global atlas of genetic associations of 220 deep phenotypes. ***medRxiv*** (2020).
3. *Sakaue, S., ***Kanai, M.***, Karjalainen, J., Akiyama, M., Kurki, M., Matoba, N., Takahashi, A., Hirata, M., Kubo, 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).
4. 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).
5. **Kanai, M.**, Akiyama, M., Takahashi, A., Matoba, N., Momozawa, Y., Ikeda, M., Iwata, N., Ikegawa, S., 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 Publications and Preprints

6. Koskela, J. T., Happola, P., Liu, A., Partanen, J., Genovese, G., Artomov, M., Myllymaki, M. N. M., **Kanai, M.**, Zhou, W., Karjalainen, J., Palviainen, T., Ronkainen, J., Sebert, S., Tukiainen, T., Palta, P., Kaprio, J., Kurki, M., Ganna, A., Palotie, A., Laitinen, T., Myllarniemi, M., Daly, M. J. & FinnGen. Genetic variant in SPDL1 reveals novel mechanism linking pulmonary fibrosis risk and cancer protection. ***medRxiv*** (2021).
7. Nakatochi, M., Toyoda, Y., **Kanai, M.**, Nakayama, A., Kawamura, Y., Hishida, A., Mikami, H., Matsuo, K., Takezaki, T., Momozawa, Y., Biobank Japan Project, Kamatani, Y., Ichihara, S., Shinomiya, N., 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*** (2021).

8. Turley, P., Martin, A. R., Goldman, G., Li, H., **Kanai, M.**, Walters, R. K., Jala, J. B., Lin, K., Millwood, I. Y., Carey, C. E., Palmer, D. S., Zacher, M., Atkinson, E. G., Chen, Z., Li, L., Akiyama, M., Okada, Y., Kamatani, Y., Walters, R. G., Callier, S., Laibson, D., 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).
9. Pirastu, N., Cordioli, M., Nandakumar, P., Mignogna, G., Abdellaoui, A., Hollis, B., **Kanai, M.**, Rajagopal, V. M., Della Briotta Parolo, P., Baya, N., Carey, C. E., Karjalainen, J., Als, T. D., Van der Zee, M. D., Day, F. R., Ong, K. K., Morisaki, T., de Geus, E., Bellocco, R., Okada, Y., Børglum, A. D., Joshi, P., Auton, A., Hinds, D., 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***, 1–9 (2021).
10. The COVID-19 Host Genetics Initiative. Mapping the human genetic architecture of COVID-19 by worldwide meta-analysis. ***medRxiv*** (2021).
11. Shi, H., Gazal, S., **Kanai, M.**, Koch, E. M., Schoech, A. P., Siewert, K. M., Kim, S. S., Luo, Y., Amariuta, T., 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).
12. Atkinson, E. G., Maihofer, A. X., **Kanai, M.**, Martin, A. R., Karczewski, K. J., Santoro, M. L., Ulirsch, J. C., Kamatani, Y., Okada, Y., 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).
13. Griesemer, D., Xue, J. R., Reilly, S. K., Ulirsch, J. C., Kukreja, K., Davis, J., **Kanai, M.**, Yang, D. K., Montgomery, S. B., Novina, C. D., Tewhey, R. & Sabeti, P. C. Genome-wide functional screen of 3'UTR variants uncovers causal variants for human disease and evolution. ***bioRxiv*** (2021).
14. Rämö, J. T., Kiiskinen, T., Karjalainen, J., Krebs, K., Kurki, M., Havulinna, A. S., Hämäläinen, E., Häppölä, P., Hautakangas, H., FinnGen, Karczewski, K. J., **Kanai, M.**, Mägi, R., Palta, P., Esko, T., Metspalu, A., Pirinen, 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).
15. Wang, Q. S., Kelley, D. R., Ulirsch, J., **Kanai, M.**, Sadhuka, S., Cui, R., Albors, C., Cheng, N., Okada, Y., The Biobank Japan Project, 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. ***bioRxiv*** (2020).
16. Weeks, E. M., Ulirsch, J. C., Cheng, N. Y., Trippe, B. L., Fine, R. S., Miao, J., Patwardhan, T. A., **Kanai, M.**, Nasser, J., Fulco, C. P., Tashman, K. C., Aguet, F., Li, T., Ordovas-Montanes, J., Smillie, C. S., Biton, M., Shalek, A. K., Ananthakrishnan, A. N., Xavier, R. J., Regev, A., Gupta, R. M., Lage, K., 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).
17. Zheng, J., Zhang, Y., Rasheed, H., Walker, V., Sugawara, Y., Li, J., Leng, Y., Elsworth, B., Wootton, R. E., Fang, S., Yang, Q., Burgess, S., Haycock, P., Borges, M. C., Cho, Y., Carnegie, R., Howell, A., Robinson, J., Thomas, L., Brumpton, B., Hveem, K., Hallan, S., Franceschini, N., Morris, A., Kottgen, A., Pattaro, C., Wuttke, M., Yamamoto, M., Kashihara, N., Akiyama, M., **Kanai, M.**, Matsuda, K., Kamatani, Y., Okada, Y., Xu, M., Bi, Y., Ning, G., Davey Smith, G., Barbour, S., Yu, C., Asvold, B. O., Zhang, H. & Gaunt, T. Trans-ethnic Mendelian randomization study reveals causal relationships between cardio-metabolic factors and chronic kidney disease. ***medRxiv*** (2020).

18. Chen, M.-H., Raffield, L. M., Mousas, A., Sakaue, S., Huffman, J. E., Moscati, A., Trivedi, B., Jiang, T., Akbari, P., Vuckovic, D., Bao, E. L., Zhong, X., Manansala, R., Laplante, V., Chen, M., Lo, K. S., Qian, H., Lareau, C. A., Beaudoin, M., Hunt, K. A., Akiyama, M., Bartz, T. M., Ben-Shlomo, Y., Beswick, A., Bork-Jensen, J., Bottinger, E. P., Brody, J. A., van Rooij, F. J. A., Chitrala, K., Cho, K., Choquet, H., Correa, A., Danesh, J., Di Angelantonio, E., Dimou, N., Ding, J., Elliott, P., Esko, T., Evans, M. K., Floyd, J. S., Broer, L., Grarup, N., Guo, M. H., Greinacher, A., Haessler, J., Hansen, T., Howson, J. M. M., Huang, Q. Q., Huang, W., Jorgenson, E., Kacprowski, T., Kähönen, M., Kamatani, Y., **Kanai, M.**, Karthikeyan, S., Koskeridis, F., Lange, L. A., Lehtimäki, T., Lerch, M. M., Linneberg, A., Liu, Y., Lyytikäinen, L.-P., Manichaikul, A., Martin, H. C., Matsuda, K., Mohlke, K. L., Mononen, N., Murakami, Y., Nadkarni, G. N., Nauck, M., Nikus, K., Ouwehand, W. H., Pankratz, N., Pedersen, O., Preuss, M., Psaty, B. M., Raitakari, O. T., Roberts, D. J., Rich, S. S., Rodriguez, B. A. T., Rosen, J. D., Rotter, J. I., Schubert, P., Spracklen, C. N., Surendran, P., Tang, H., Tardif, J.-C., Trembath, R. C., Ghanbari, M., Völker, U., Völzke, H., Watkins, N. A., Zonderman, A. B., Wilson, P. W. F., Li, Y., Butterworth, A. S., Gauchat, J.-F., Chiang, C. W. K., Li, B., Loos, R. J. F., Astle, W. J., Evangelou, E., van Heel, D. A., Sankaran, V. G., Okada, Y., 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).

19. Vuckovic, D., Bao, E. L., Akbari, P., Lareau, C. A., Mousas, A., Jiang, T., Chen, M.-H., Raffield, L. M., Tardaguila, M., Huffman, J. E., Ritchie, S. C., Megy, K., Ponstingl, H., Penkett, C. J., Albers, P. K., Wigdor, E. M., Sakaue, S., Moscati, A., Manansala, R., Lo, K. S., Qian, H., Akiyama, M., Bartz, T. M., Ben-Shlomo, Y., Beswick, A., Bork-Jensen, J., Bottinger, E. P., Brody, J. A., van Rooij, F. J. A., Chitrala, K. N., Wilson, P. W. F., Choquet, H., Danesh, J., Di Angelantonio, E., Dimou, N., Ding, J., Elliott, P., Esko, T., Evans, M. K., Felix, S. B., Floyd, J. S., Broer, L., Grarup, N., Guo, M. H., Guo, Q., Greinacher, A., Haessler, J., Hansen, T., Howson, J. M. M., Huang, W., Jorgenson, E., Kacprowski, T., Kähönen, M., Kamatani, Y., **Kanai, M.**, Karthikeyan, S., Koskeridis, F., Lange, L. A., Lehtimäki, T., Linneberg, A., Liu, Y., Lyytikäinen, L.-P., Manichaikul, A., Matsuda, K., Mohlke, K. L., Mononen, N., Murakami, Y., Nadkarni, G. N., Nikus, K., Pankratz, N., Pedersen, O., Preuss, M., Psaty, B. M., Raitakari, O. T., Rich, S. S., Rodriguez, B. A. T., Rosen, J. D., Rotter, J. I., Schubert, P., Spracklen, C. N., Surendran, P., Tang, H., Tardif, J.-C., Ghanbari, M., Völker, U., Völzke, H., Watkins, N. A., Weiss, S., Cai, N., Kundu, K., Watt, S. B., Walter, K., Zonderman, A. B., Cho, K., Li, Y., Loos, R. J. F., Knight, J. C., Georges, M., Stegle, O., Evangelou, E., Okada, Y., Roberts, D. J., Inouye, M., Johnson, A. D., Auer, P. L., Astle, W. J., Reiner, A. P., 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).

20. Shirai, Y., Honda, S., Ikari, K., **Kanai, M.**, Takeda, Y., Kamatani, 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).

21. Luo, Y., **Kanai, M.**, Choi, W., Li, X., Yamamoto, K., Ogawa, K., Gutierrez-Arcelus, M., Gregersen, P. K., Stuart, P. E., Elder, J. T., Fellay, J., Carrington, M., Haas, D. W., Guo, X., Palmer, N. D., Chen, Y.-D. I., Rotter, J. I., Taylor, K. D., Rich, S., Correa, A., Wilson, J. G., Kathiresan, S., Cho, M., Metspalu, A., Esko, T., Okada, Y., Han, B., NHLBI Trans-Omics for Precision Medicine (TOPMed) Consortium, Mclaren, P. J. & Raychaudhuri, S. A high-resolution HLA reference panel capturing global population diversity enables multi-ethnic fine-mapping in HIV host response. ***medRxiv*** (2020).

22. Ishigaki, K., Akiyama, M., **Kanai, M.**, Takahashi, A., Kawakami, E., Sugishita, H., Sakaue, S., Matoba, N., Low, S.-K., Okada, Y., Terao, C., Amariuta, T., Gazal, S., Kochi, Y., Horikoshi, M., Suzuki, K., Ito, K., Koyama, S., Ozaki, K., Niida, S., Sakata, Y., Sakata, Y., Kohno, T., Shiraishi, K., Momozawa, Y., Hirata, M., Matsuda, K., Ikeda, M., Iwata, N., Ikegawa, S., Kou, I., Tanaka, T., Nakagawa, H., Suzuki, A., Hirota, T., Tamari, M., Chayama, K., Miki, D., Mori, M., Nagayama, S., Daigo, Y., Miki, Y., Katagiri, T., Ogawa, O., Obara, W., Ito, H., Yoshida, T., Imoto, I., Takahashi, T., Tanikawa, C., Suzuki, T., Sinozaki, N., Minami, S., Yamaguchi, H., Asai, S., Takahashi, Y., Yamaji, K., Takahashi, K., Fujioka, T., Takata, R., Yanai, H., Masumoto, A., Koretsune, Y., Kutsumi, H., Higashiyama, M.,

Murayama, S., Minegishi, N., Suzuki, K., Tanno, K., Shimizu, A., Yamaji, T., Iwasaki, M., Sawada, N., Uemura, H., Tanaka, K., Naito, M., Sasaki, M., Wakai, K., Tsugane, S., Yamamoto, M., Yamamoto, K., Murakami, Y., Nakamura, Y., Raychaudhuri, S., 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).

23. Reilly, S. K., Gosai, S. J., Guiterrez, A., Ulirsch, J. C., **Kanai, M.**, Berenzy, D., Kales, S., Butler, G. B., Gladden-Young, A., Finucane, H. K., Sabeti, P. C. & Tewhey, R. HCR-FlowFISH: A flexible CRISPR screening method to identify cis-regulatory elements and their target genes. ***bioRxiv*** (2020).
24. Sakaue, S., Hirata, J., **Kanai, M.**, Suzuki, K., Akiyama, M., Lai Too, C., Arayssi, T., Hammoudeh, M., Al Emadi, S., Masri, B. K., Halabi, H., Badsha, H., Uthman, I. W., Saxena, R., Padyukov, L., 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).
25. Ray, J. P., de Boer, C. G., Fulco, C. P., Lareau, C. A., **Kanai, M.**, Ulirsch, J. C., Tewhey, R., Ludwig, L. S., Reilly, S. K., Bergman, D. T., Engreitz, J. 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).
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## Presentations

### International Conference

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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).
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