

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: Trans-ethnic 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-dimensional 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), <i>The American Society of Human Genetics 2020 Virtual Meeting</i>	2020
Reviewers' Choice Abstract Award	Houston, USA
<i>The American Society of Human Genetics 2019 Annual Meeting</i>	2019
Masason Foundation Fellowship	Tokyo, Japan
Research grant for trans-ethnic analysis of complex human diseases and traits.	2018–2021+
Nakajima Foundation Fellowship	Tokyo, Japan
Predocotrual fellowship which covers up to ~\$50,000/year for tuition and stipend.	2017–2021+
The 3rd place, Worldwide Finals	New York, USA
<i>Microsoft Imagine Cup 2011 Windows 7 Touch Challenge</i>	2011
The 1st place, National Finals & Worldwide Finalist	Japan & Poland
<i>Microsoft Imagine Cup 2010 Software Design Competition</i>	2010

Publications

Selected Publications and Preprints

(* = co-first)

1. *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).
2. 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).
3. **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

4. 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).
5. *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., Gen, F., 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).
6. 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).

7. 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).
8. 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., Kashiwara, 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).
9. 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).
10. 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).
11. 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).

12. 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).
13. 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).
14. 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: A framework allowing for improved inclusion of admixed individuals in large-scale association studies. *bioRxiv* (2020).
15. Reilly, S. K., Gosai, S. J., Guterres, 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).
16. 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).
17. 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., Karjalainen, J., Als, T. D., van der Zee, M. D., Day, F. R., Ong, K. K., 23andMe Research Team, iPSYCH Consortium, Morisaki, T., de Geus, E., Bellocco, R., Okada, Y., Borglum, A., Joshi, P. K., Auton, A., Hinds, D. K., Neale, B., Walters, R. K., Nivard, M. G., Perry, J. & Ganna, A. Genetic analyses identify widespread sex-differential participation bias. *bioRxiv* (2020).
18. 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).
19. Matoba, N., Akiyama, M., Ishigaki, K., **Kanai, M.**, Takahashi, A., Momozawa, Y., Ikegawa, S., Ikeda, M., Iwata, N., Hirata, M., 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).
20. Clark, D. W., Okada, Y., Moore, K. H. S., Mason, D., Pirastu, N., Gandin, I., Mattsson, H., Barnes, C. L. K., Lin, K., Zhao, J. H., Deelen, P., Rohde, R., Schurmann, C., Guo, X., Giulianini, F., Zhang, W., Medina-Gomez, C., Karlsson, R., Bao, Y., Bartz, T. M., Baumbach, C., Biino, G., Bixley, M. J., Brumat, M., Chai, J.-F., Corre, T., Cousminer, D. L., Dekker, A. M., Eccles, D. A., van Eijk, K. R., Fuchsberger, C., Gao, H., Germain, M., Gordon, S. D., de Haan, H. G., Harris, S. E., Hofer, E., Huerta-Chagoya, A., Igartua, C., Jansen, I. E., Jia, Y., Kacprowski, T., Karlsson, T., Kleber, M. E., Li, S. A.,

Li-Gao, R., Mahajan, A., Matsuda, K., Meidtner, K., Meng, W., Montasser, M. E., van der Most, P. J., Munz, M., Nutile, T., Palviainen, T., Prasad, G., Prasad, R. B., Priyanka, T. D. S., Rizzi, F., Salvi, E., Sapkota, B. R., Shriner, D., Skotte, L., Smart, M. C., Smith, A. V., van der Spek, A., Spracklen, C. N., Strawbridge, R. J., Tajuddin, S. M., Trompet, S., Turman, C., Verweij, N., Viberti, C., Wang, L., Warren, H. R., Wootton, R. E., Yanek, L. R., Yao, J., Yousri, N. A., Zhao, W., Adeyemo, A. A., Afaq, S., Aguilar-Salinas, C. A., Akiyama, M., Albert, M. L., Allison, M. A., Alver, M., Aung, T., Azizi, F., Bentley, A. R., Boeing, H., Boerwinkle, E., Borja, J. B., de Borst, G. J., Bottinger, E. P., Broer, L., Campbell, H., Chanock, S., Chee, M.-L., Chen, G., Chen, Y.-D. I., Chen, Z., Chiu, Y.-F., Cocca, M., Collins, F. S., Concas, M. P., Corley, J., Cugliari, G., van Dam, R. M., Damulina, A., Daneshpour, M. S., Day, F. R., Delgado, G. E., Dhana, K., Doney, A. S. F., Dörr, M., Doumatey, A. P., Dzimiri, N., Ebenesersdóttir, S. S., Elliott, J., Elliott, P., Ewert, R., Felix, J. F., Fischer, K., Freedman, B. I., Grotto, G., Goel, A., Gögele, M., Goodarzi, M. O., Graff, M., Granot-HersHKovitz, E., Grodstein, F., Guarrera, S., Gudbjartsson, D. F., Guity, K., Gunnarsson, B., Guo, Y., Hagenaars, S. P., Haiman, C. A., Halevy, A., Harris, T. B., Hedayati, M., van Heel, D. A., Hirata, M., Höfer, I., Hsiung, C. A., Huang, J., Hung, Y.-J., Ikram, M. A., Jagadeesan, A., Jousilahti, P., Kamatani, Y., **Kanai, M.**, Kerrison, N. D., Kessler, T., Khaw, K.-T., Khor, C. C., de Kleijn, D. P. V., Koh, W.-P., Kolcic, I., Kraft, P., Krämer, B. K., Kutalik, Z., Kuusisto, J., Langenberg, C., Launer, L. J., Lawlor, D. A., Lee, I.-T., Lee, W.-J., Lerch, M. M., Li, L., Liu, J., Loh, M., London, S. J., Loomis, S., Lu, Y., Luan, J., Mägi, R., Manichaikul, A. W., Manunta, P., Måsson, G., Matoba, N., Mei, X. W., Meisinger, C., Meitinger, T., Mezzavilla, M., Milani, L., Millwood, I. Y., Momozawa, Y., Moore, A., Morange, P.-E., Moreno-Macias, H., Mori, T. A., Morrison, A. C., Muka, T., Murakami, Y., Murray, A. D., de Mutsert, R., Mychaleckyj, J. C., Nalls, M. A., Nauck, M., Neville, M. J., Nolte, I. M., Ong, K. K., Orozco, L., Padmanabhan, S., Pålsson, G., Pankow, J. S., Pattaro, C., Pattie, A., Polasek, O., Poulter, N., Pramstaller, P. P., Quintana-Murci, L., Rääkkönen, K., Ralhan, S., Rao, D. C., van Rhee, W., Rich, S. S., Ridker, P. M., Rietveld, C. A., Robino, A., van Rooij, F. J. A., Ruggiero, D., Saba, Y., Sabanayagam, C., Sabater-Lleal, M., Sala, C. F., Salomaa, V., Sandow, K., Schmidt, H., Scott, L. J., Scott, W. R., Sedaghati-Khayat, B., Sennblad, B., van Setten, J., Sever, P. J., Sheu, W. H.-H., Shi, Y., Shrestha, S., Shukla, S. R., Sigurdsson, J. K., Sikka, T. T., Singh, J. R., Smith, B. H., Stančáková, A., Stanton, A., Starr, J. 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