

# Masahiro Kanai

## Curriculum Vitae

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## Education

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**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

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**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

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**The Certification for Bioinformatics Engineers**  
certificated by the Japanese Society of Bioinformatics

2015

## Awards and Fellowships

<b>Masason Foundation Fellowship</b>	<b>Tokyo, Japan</b>
Research grant for trans-ethnic analysis of complex human diseases and traits.	2018–
<b>Nakajima Foundation Fellowship</b>	<b>Tokyo, Japan</b>
Predoctoral fellowship which covers up to ~\$50,000/year for tuition and stipend.	2017–2021+
<b>The 3rd place, Worldwide Finals</b>	<b>New York, USA</b>
Microsoft Imagine Cup 2011 Windows 7 Touch Challenge	2011
<b>The 1st place, National Finals &amp; Worldwide Finalist</b>	<b>Japan &amp; Poland</b>
Microsoft Imagine Cup 2010 Software Design Competition	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* (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. 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).
5. 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).
6. 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., Cho, K., 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., Greinacher, A., Haessler, J., Hansen, T., Howson, J., 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., 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., VA Million Veteran Program, Cai, N., Kundu, K., Watt, S., Walter, K., Zonderman, A. B., Wilson, P. W. F., 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. *medRxiv* (2020).
7. Chen, M.-H., Raffield, L. M., Mousas, A., Sakaue, S., Huffman, J. E., Jiang, T., Akbari, P., Vuckovic, D., Bao, E. L., Moscati, A., Zhong, X., Manansala, R., Laplante, V., Chen, M., Lo, K. S., Qian, H., Lareau, C. A., Beaudoin, M., 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, 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., 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., Suren-dran, P., Tang, H., Tardif, J.-C., Ghanbari, M., Völker, U., Völzke, H., Watkins, N. A., Zonderman, A. B., VA Million Veteran Program, Wilson, P. W. F., Li, Y., Butterworth, A. S., Gauchat, J.-F., Chi-ang, C. W. K., Li, B., Loos, R. J. F., Astle, W. J., Evangelou, E., 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. *bioRxiv* (2020).
  8. 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).
  9. 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-Macías, 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., Pálsson, 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. M., Stefansdottir, L., Straker, L., Sulem, P., Sveinbjornsson, G., Swertz, M. A., Taylor, A. M., Taylor, K. D., Terzikhan, N., Tham, Y.-C., Thorleifsson, G., Thorsteinsdottir, U., Tillander, A., Tracy, R. P., Tusié-Luna, T., Tzoulaki, I., Vaccargiu, S., Vangipurapu, J., Veldink, J. H., Vitart, V., Völker, U., Vuoksimaa, E., Wakil, S. M., Waldenberger, M., Wander, G. S., Wang, Y. X., Wareham, N. J., Wild, S., Yajnik, C. S., Yuan, J.-M., Zeng, L., Zhang, L., Zhou, J., Amin, N., Asselbergs, F. W., Bakker, S. J. L., Becker, D. M., Lehne, B., Bennett, D. A., van den Berg, L. H., Berndt, S. I., Bharadwaj, D., Bielak, L. F., Bochud, M., Boehnke, M., Bouchard, C., Bradfield, J. P., Brody, J. A., Campbell, A., Carmi, S., Caulfield, M. J., Cesarini, D., Chambers, J. C., Chandak, G. R., Cheng, C.-Y., Ciullo, M., Cornelis, M., Cusi, D., Smith, G. D., Deary, I. J., Dorajoo, R., van Duijn, C. M., Ellinghaus, D., Erdmann, J., Eriksson, J. G., Evangelou, E., Evans, M. K., Faul, J. D., Feenstra, B., Feitosa, M., Foisy, S., Franke, A., Friedlander, Y., Gasparini, P., Gieger, C., Gonzalez, C., Goyette, P., Grant, S. F. A., Griffiths, L. R., Groop, L., Gudnason, V., Gyllenstein, U., Hakonarson, H., Hamsten, A., van der Harst, P., Heng, C.-K., Hicks, A. A., Hochner, H., Huikuri, H., Hunt, S. C., Jaddoe, V. W. V., De Jager, P. L., Johannesson, M., Johansson, Å., Jonas, J. B., Jukema, J. W., Juntila, J., Kaprio, J., Kardia, S. L. R., Karpe, F., Kumari, M., Laakso, M., van der Laan, S. W., Lahti, J., Laudes, M., Lea, R. A., Lieb, W., Lumley, T., Martin, N. G., März, W., Matullo, G., McCarthy, M. I., Medland, S. E., Merriman, T. R., Metspalu, A., Meyer, B. F., Mohlke, K. L., Montgomery, G. W., Mook-Kanamori, D., Munroe, P. B., North, K. E., Nyholt, D. R., O'Connell, J. R., Ober, C., Oldehinkel, A. J., Palmas, W., Palmer, C., Pasterkamp, G. G., Patin, E., Pennell, C. E., Perusse, L., Peyser, P. A., Pirastu, M., Polderman, T. J. C., Porteous, D. J., Posthuma, D., Psaty, B. M., Rioux, J. D., Rivadeneira, F., Rotimi, C., Rotter, J. I., Rudan, I., Den Ruijter, H. M., Sanghera, D. K., Sattar, N., Schmidt, R., Schulze, M. B., Schunkert, H., Scott, R. A., Shuldiner, A. R., Sim, X., Small, N., Smith, J. A., Sotoodehnia, N., Tai, E.-S., Teumer, A., Timpson, N. J., Toniolo, D., Tregouet, D.-A., Tuomi, T., Vollenweider, P., Wang, C. A., Weir, D. R., Whitfield, J. B., Wijmenga, C., Wong, T.-Y., Wright, J., Yang, J., Yu, L., Zemel, B. S., Zonderman, A. B., Perola, M., Magnusson, P. K. E., Uitterlinden, A. G., Kooner, J. S., Chasman, D. I., Loos, R. J. F., Franceschini, N., Franke, L., Haley, C. S., Hayward, C., Walters, R. G., Perry, J. R. B., Esko, T., 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).
10. Shi, H., Gazal, S., **Kanai, M.**, Koch, E. M., Schoech, A. P., Kim, S. S., Luo, Y., Amariuta, T., Okada, Y., Raychaudhuri, S., Sunyaev, S. R. & Price, A. L. Population-specific causal disease effect sizes in functionally important regions impacted by selection. *bioRxiv* (2019).
  11. Leu, C., Stevelink, R., Smith, A. W., Goleva, S. B., **Kanai, M.**, Ferguson, L., Campbell, C., Kamatani, Y., Okada, Y., Sisodiya, S. M., Cavalleri, G. L., Koeleman, B. P. C., Lerche, H., Jehi, L., Davis, L. K., 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).
  12. Ishigaki, K., Akiyama, M., **Kanai, M.**, Takahashi, A., Kawakami, E., Sakaue, S., Matoba, N., Low, S.-k., Okada, Y., Terao, C., Amariuta, T., Gazal, S., Kochi, Y., Horikoshi, M., Suzuki, K., Ito, 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., Ogawa, O., Obara, W., Ito, H., Yoshida, T., Imoto, I., 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., Minegishi, N., Suzuki, K., Tanno, K., Shimizu, A., Yamaji, T., Sawada, N., Uemura, H., Tanaka, K., Naito, M., Sasaki, M., Tsugane, S., Yamamoto, M., Yamamoto, K., Murakami, Y., Raychaudhuri, S., Inazawa, J., Yamauchi, T., Kadowaki, T., Kubo, M. & Kamatani, Y. Large scale genome-wide association study in a Japanese population identified 45 novel susceptibility loci for 22 diseases. *bioRxiv* (2019).
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## Presentations

### International Conference

1. **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).
2. **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).
3. **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: March 24, 2020.