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-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

Masason Foundation Fellowship **Tokyo, Japan**
Research grant for trans-ethnic analysis of complex human diseases and traits. *2018–*

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. *Sakaue, S., ****Kanai, M.**, Karjalainen, J., Akiyama, M., Kurki, M., Matoba, N., Takahashi, A., Hirata, M., Kubo, M., Matsuda, K., Murakami, Y., FinnGen, 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. ***bioRxiv*** (2019).
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. 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).
5. 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., Surendran, 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., Chiang, 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).

6. 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).
7. 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).
8. 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).
9. 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*** (2020).
10. 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., Girotto, 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., Másson, 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., Räikkönen, K., Ralhan, S., Rao, D. C., van Rheenen, 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., Gyllensten, 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., Junttila, 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).

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. Tin, A., Marten, J., Halperin Kuhns, V. L., Li, Y., Wuttke, M., Kirsten, H., Sieber, K. B., Qiu, C., Gorski, M., Yu, Z., Giri, A., Sveinbjornsson, G., Li, M., Chu, A. Y., Hoppmann, A., O'Connor, L. J., Prins, B., Nutile, T., Noce, D., Akiyama, M., Cocca, M., Ghasemi, S., van der Most, P. J., Horn, K., Xu, Y., Fuchsberger, C., Sedaghat, S., Afaq, S., Amin, N., Ärnlöv, J., Bakker, S. J. L., Bansal, N., Baptista, D., Bergmann, S., Biggs, M. L., Biino, G., Boerwinkle, E., Bottinger, E. P., Boutin, T. S., Brumat, M., Burkhardt, R., Campana, E., Campbell, A., Campbell, H., Carroll, R. J., Catamo, E., Chambers, J. C., Ciullo, M., Concas, M. P., Coresh, J., Corre, T., Cusi, D., Felicita, S. C., de Borst, M. H., De Grandi, A., de Mutsert, R., de Vries, A. P. J., Delgado, G., Demirkan, A., Devuyst, O., Dittrich, K., Eckardt, K.-U., Ehret, G., Endlich, K., Evans, M. K., Gansevoort, R. T., Gasparini, P., Giedraitis, V.,

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Presentations

International Conference

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