

Masahiro Kanai

Curriculum Vitae

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Education

Harvard University Bioinformatics and Integrative Genomics PhD Program, Harvard Medical School	Boston, USA Aug. 2017–present
Keio University B.S. in Bioinformatics, Department of Biosciences and Informatics	Yokohama, Japan Apr. 2013–Mar. 2017

Research Experience

Massachusetts General Hospital & Broad Institute of MIT and Harvard Graduate Student, Analytic and Translational Genetics Unit	Boston, USA Jan. 2018–present
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Advisors: Drs. Mark Daly & Hilary Finucane

Research theme: Trans-ethnic analysis of complex human diseases and traits.

RIKEN Center for Integrative Medical Sciences Research Assistant, Laboratory for Statistical Analysis	Yokohama, Japan May 2015–present
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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
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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
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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
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Awards and Fellowships

Nakajima Foundation Fellowship Predoctoral fellowship which covers up to ~\$50,000/year for tuition and stipend.	Tokyo, Japan 2017–2019+
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The 3rd place, Worldwide Finals Microsoft Imagine Cup 2011 Windows 7 Touch Challenge	New York, USA 2011
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The 1st place, National Finals & Worldwide Finalist Microsoft Imagine Cup 2010 Software Design Competition	Japan & Poland 2010
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Publications

(* = co-first)

1. Wuttke, M., Li, Y., Li, M., Sieber, K. B., Feitosa, M. F., Gorski, M., Tin, A., Wang, L., Chu, A. Y., Hoppmann, A., Kirsten, H., Giri, A., Chai, J.-F., Sveinbjornsson, G., Tayo, B. O., Nutile, T., Fuchsberger, C., Marten, J., Cocca, M., Ghasemi, S., Xu, Y., Horn, K., Noce, D., van der Most, P. J., Sedaghat, S., Yu, Z., Akiyama, M., Afaq, S., Ahluwalia, T. S., Almgren, P., Amin, N., Ärnlöv, J., Bakker, S. J. L., Bansal, N., Baptista, D., Bergmann, S., Biggs, M. L., Biino, G., Boehnke, M., Boerwinkle, E., Boissel, M., Bottinger, E. P., Boutin, T. S., Brenner, H., Brumat, M., Burkhardt, R., Butterworth, A. S., Campana, E., Campbell, A., Campbell, H., Canouil, M., Carroll, R. J., Catamo, E., Chambers, J. C., Chee, M.-L., Chee, M.-L., Chen, X., Cheng, C.-Y., Cheng, Y., Christensen, K., Cifkova, R., Ciullo, M., Concas, M. P., Cook, J. P., Coresh, J., Corre, T., Sala, C. F., Cusi, D., Danesh, J., Daw, E. W., de Borst, M. H., De Grandi, A., de Mutsert, R., de Vries, A. P. J., Degenhardt, F., Delgado, G., Demirkan, A., Di Angelantonio, E., Dittrich, K., Divers, J., Dorajoo, R., Eckardt, K.-U., Ehret, G., Elliott, P., Endlich, K., Evans, M. K., Felix, J. F., Foo, V. H. X., Franco, O. H., Franke, A., Freedman, B. I., Freitag-Wolf, S., Friedlander, Y., Froguel, P., Gansevoort, R. T., Gao, H., Gasparini, P., Gaziano, J. M., Giedraitis, V., Gieger, C., Girotto, G., Giulianini, F., Gögele, M., Gordon, S. D., Gudbjartsson, D. F., Gudnason, V., Haller, T., Hamet, P., Harris, T. B., Hartman, C. A., Hayward, C., Hellwege, J. N., Heng, C.-K., Hicks, A. A., Hofer, E., Huang, W., Hutri-Kähönen, N., Hwang, S.-J., Ikram, M. A., Indridason, O. S., Ingelsson, E., Ising, M., Jaddoe, V. W. V., Jakobsdottir, J., Jonas, J. B., Joshi, P. K., Josyula, N. S., Jung, B., Kähönen, M., Kamatani, Y., Kammerer, C. M., **Kanai, M.**, Kastarinen, M., Kerr, S. M., Khor, C.-C., Kiess, W., Kleber, M. E., Koenig, W., Kooner, J. S., Körner, A., Kovacs, P., Kraja, A. T., Krajcoviechova, A., Kramer, H., Krämer, B. K., Kronenberg, F., Kubo, M., Kühnel, B., Kuokkanen, M., Kuusisto, J., La Bianca, M., Laakso, M., Lange, L. A., Langefeld, C. D., Lee, J. J.-M., Lehne, B., Lehtimäki, T., Lieb, W., Lim, S.-C., Lind, L., Lindgren, C. M., Liu, J., Liu, J., Loeffler, M., Loos, R. J. F., Lucae, S., Lukas, M. A., Lyytikäinen, L.-P., Mägi, R., Magnusson, P. K. E., Mahajan, A., Martin, N. G., Martins, J., März, W., Mascalzoni, D., Matsuda, K., Meisinger, C., Meitinger, T., Melander, O., Metspalu, A., Mikaelssdottir, E. K., Milanese, Y., Miliku, K., Mishra, P. P., Mohlke, K. L., Mononen, N., Montgomery, G. W., Mook-Kanamori, D. O., Mychaleckyj, J. C., Nadkarni, G. N., Nalls, M. A., Nauck, M., Nikus, K., Ning, B., Nolte, I. M., Noordam, R., O'Connell, J., O'Donoghue, M. L., Olafsson, I., Oldehinkel, A. J., Orho-Melander, M., Ouwehand, W. H., Padmanabhan, S., Palmer, N. D., Palsson, R., Penninx, B. W. J. H., Perls, T., Perola, M., Pirastu, M., Pirastu, N., Pistis, G., Podgornaia, A. I., Polasek, O., Ponte, B., Porteous, D. J., Poulain, T., Pramstaller, P. P., Preuss, M. H., Prins, B. P., Province, M. A., Rabelink, T. J., Raffield, L. M., Raitakari, O. T., Reilly, D. F., Rettig, R., Rheinberger, M., Rice, K. M., Ridker, P. M., Rivadeneira, F., Rizzi, F., Roberts, D. J., Robino, A., Rossing, P., Rudan, I., Rueedi, R., Ruggiero, D., Ryan, K. A., Saba, Y., Sabanayagam, C., Salomaa, V., Salvi, E., Saum, K.-U., Schmidt, H., Schmidt, R., Schöttker, B., Schulz, C.-A., Schupf, N., Shaffer, C. M., Shi, Y., Smith, A. V., Smith, B. H., Soranzo, N., Spracklen, C. N., Strauch, K., Stringham, H. M., Stumvoll, M., Svensson, P. O., Szymczak, S., Tai, E.-S., Tajuddin, S. M., Tan, N. Y. Q., Taylor, K. D., Teren, A., Tham, Y.-C., Thiery, J., Thio, C. H. L., Thomsen, H., Thorleifsson, G., Toniolo, D., Tönjes, A., Tremblay, J., Tzoulaki, I., Uitterlinden, A. G., Vaccargiu, S., van Dam, R. M., van der Harst, P., van Duijn, C. M., Velez Edward, D. R., Verweij, N., Vogelesang, S., Völker, U., Vollenweider, P., Waeber, G., Waldenberger, M., Wallentin, L., Wang, Y. X., Wang, C., Waterworth, D. M., Bin Wei, W., White, H., Whitfield, J. B., Wild, S. H., Wilson, J. F., Wojczynski, M. K., Wong, C., Wong, T.-Y., Xu, L., Yang, Q., Yasuda, M., Yerges-Armstrong, L. M., Zhang, W., Zonderman, A. B., Rotter, J. I., Bochud, M., Psaty, B. M., Vitart, V., Wilson, J. G., Dehghan, A., Parsa, A., Chasman, D. I., Ho, K., Morris, A. P., Devuyst, O., Akilesh, S., Pendergrass, S. A., Sim, X., Böger, C. A., Okada, Y., Edwards, T. L., Snieder, H., Stefansson, K., Hung, A. 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).
2. *Nakatochi, M., ***Kanai, M.**, *Nakayama, A., *Hishida, A., *Kawamura, Y., Ichihara, S., Akiyama, M., Ikezaki, H., Furusyo, N., Shimizu, S., Yamamoto, K., Hirata, M., Okada, R., Kawai, S., Kawaguchi, M., Nishida, Y., Shimanoe, C., Ibusuki, R., Takezaki, T., Nakajima, M., Takao, M., Ozaki, E., Matsui, D., Nishiyama, T., Suzuki, S., Takashima, N., Kita, Y., Endoh, K., Kuriki, K., Uemura, H., Arisawa, K., Oze, I., Matsuo, K., Nakamura, Y., Mikami, H., Tamura, T., Nakashima, H., Nakamura, T., Kato, N., Matsuda,

- K., Murakami, Y., Matsubara, T., Naito, M., Kubo, M., Kamatani, 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).
3. 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).
 4. Matoba, N., Akiyama, M., Ishigaki, K., **Kanai, M.**, Takahashi, A., Momozawa, Y., Ikegawa, S., Ikeda, M., Iwata, N., 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).
 5. Suzuki, K., Akiyama, M., Ishigaki, K., **Kanai, M.**, Hosoe, J., Shojima, N., Hozawa, A., Kadota, A., Kuriki, K., Naito, M., Tanno, K., Ishigaki, Y., Hirata, M., Matsuda, K., Iwata, N., Ikeda, M., Sawada, N., Yamaji, T., Iwasaki, M., Ikegawa, S., Maeda, S., Murakami, Y., Wakai, K., Tsugane, S., Sasaki, M., Yamamoto, M., Okada, Y., 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).
 6. Hirata, J., Hosomichi, K., Sakaue, S., **Kanai, M.**, Nakaoka, H., Ishigaki, K., Suzuki, K., Akiyama, M., Kishikawa, T., Ogawa, K., Masuda, T., Yamamoto, K., Hirata, M., Matsuda, K., 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).
 7. **Kanai, M.**, Maeda, Y. & Okada, Y. Grimon: graphical interface to visualize multi-omics networks. *Bioinformatics* **34**, 3934–3936 (2018).
 8. Horikoshi, M., Day, F. R., Akiyama, M., Hirata, M., Kamatani, Y., Matsuda, K., Ishigaki, K., **Kanai, M.**, Wright, H., Toro, C. A., 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).
 9. Okada, Y., Momozawa, Y., Sakaue, S., **Kanai, M.**, Ishigaki, K., Akiyama, M., Kishikawa, T., Arai, Y., Sasaki, T., Kosaki, K., Suematsu, M., 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).
 10. Malik, R., Chauhan, G., Traylor, M., Sargurupremraj, M., Okada, Y., Mishra, A., Rutten-Jacobs, L., Giese, A.-K., van der Laan, S. W., Gretarsdottir, S., Anderson, C. D., Chong, M., Adams, H. H. H., Ago, T., Almgren, P., Amouyel, P., Ay, H., Bartz, T. M., Benavente, O. R., Bevan, S., Boncoraglio, G. B., Brown, R. D., Butterworth, A. S., Carrera, C., Carty, C. L., Chasman, D. I., Chen, W.-M., Cole, J. W., Correa, A., Cotlarciuc, I., Cruchaga, C., Danesh, J., de Bakker, P. I. W., DeStefano, A. L., den Hoed, M., Duan, Q., Engelter, S. T., Falcone, G. J., Gottesman, R. F., Grewal, R. P., Gudnason, V., Gustafsson, S., Haessler, J., Harris, T. B., Hassan, A., Havulinna, A. S., Heckbert, S. R., Holliday, E. G., Howard, G., Hsu, F.-C., Hyacinth, H. I., Ikram, M. A., Ingelsson, E., Irvin, M. R., Jian, X., Jiménez-Conde, J., Johnson, J. A., Jukema, J. W., **Kanai, M.**, Keene, K. L., Kissela, B. M., Kleindorfer, D. O., Kooperberg, C., Kubo, M., Lange, L. A., Langefeld, C. D., Langenberg, C., Launer, L. J., Lee, J.-M., Lemmens, R., Leys, D., Lewis, C. M., Lin, W.-Y., Lindgren, A. G., Lorentzen, E., Magnusson, P. K., Maguire, J., Manichaikul, A., McArdle, P. F., Meschia, J. F., Mitchell, B. D., Mosley, T. H., Nalls, M. A., Ninomiya, T., O'Donnell, M. J., Psaty, B. M., Pulit, S. L., Rannikmäe, K., Reiner, A. P., Rexrode, K. M., Rice, K., Rich, S. S., Ridker, P. M., Rost, N. S., Rothwell, P. M., Rotter, J. I., Rundek, T., Sacco, R. L., Sakaue, S., Sale, M. M., Salomaa, V., Sapkota, B. R., Schmidt, R., Schmidt, C. O., Schminke, U., Sharma, P., Slowik, A., Sudlow, C. L. M., Tanislav, C., Tatlisumak, T., Taylor, K. D., Thijs, V. N. S., Thorleifsson, G., Thorsteinsdottir, U., Tiedt, S., Trompet, S., Tzourio, C., van Duijn, C. M., Walters, M., Wareham, N. J., Wassertheil-Smoller, S., Wilson, J. G., Wiggins, K. L., Yang, Q., Yusuf, S., Bis, J. C., Pastinen, T., Ruusalepp, A., Schadt, E. E., Koplev, S., Björkegren, J. L. M., Codoni, V., Civelek, M., Smith, N. L., Trégouët, D. A., Christophersen, I. E., Roselli, C., Lubitz, S. A., Ellinor, P. T., Tai, E. S., Kooner, J. S., Kato, N., He, J., van der Harst, P., Elliott, P., Chambers, J. C., Takeuchi, F., Johnson, A. D., Sanghera, D. K., Melander, O., Jern, C., Strbian, D., Fernandez-Cadenas, I., Longstreth, W. T., Rolfs, A., Hata, J., Woo, D., Rosand, J., Pare, G., Hopewell,

- J. C., Saleheen, D., Stefansson, K., Worrall, B. B., Kittner, S. J., Seshadri, S., Fornage, M., Markus, H. S., Howson, J. M. M., Kamatani, Y., DeBette, S. & Dichgans, M. Multiancestry genome-wide association study of 520,000 subjects identifies 32 loci associated with stroke and stroke subtypes. *Nature Genetics* **50**, 524–537 (2018).
11. **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).
 12. Hirata, J., Hirota, T., Ozeki, T., **Kanai, M.**, Sudo, T., Tanaka, T., Hizawa, N., Nakagawa, H., 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).
 13. Akiyama, M., Okada, Y., **Kanai, M.**, Takahashi, A., Momozawa, Y., Ikeda, M., Iwata, N., Ikegawa, S., Hirata, M., Matsuda, K., Iwasaki, M., Yamaji, T., Sawada, N., Hachiya, T., Tanno, K., Shimizu, A., Hozawa, A., 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).
 14. 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).
 15. Okada, Y., Suzuki, A., Ikari, K., Terao, C., Kochi, Y., Ohmura, K., Higasa, K., Akiyama, M., Ashikawa, K., **Kanai, M.**, Hirata, J., Suita, N., Teo, Y.-Y., Xu, H., Bae, S.-C., Takahashi, A., Momozawa, Y., Matsuda, K., Momohara, S., Taniguchi, A., Yamada, R., Mimori, T., Kubo, M., Brown, M. A., 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).
 16. **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).
 17. 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).
 18. 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)

1. **Kanai, M.** Genetic analysis of quantitative traits with integration of multi-omics data. *The Medical Frontline (SAISHIN IGAKU)* **74**, 220–226 (2019).
2. **Kanai, M.**, Okada, Y. & Kamatani, Y. Genetics of clinical measurements in the Japanese population: large-scale genome-wide association study of 58 quantitative traits. *Clinical Immunology & Allergy* **71**, 78–85 (2019).
3. **Kanai, M.**, Okada, Y. & Kamatani, Y. Large-scale genome-wide association study in the Japanese population elucidates genetic backgrounds of 58 quantitative traits. *Life Science First Author's Review*. doi:10.7875/first.author.2018.025 (2018).
4. **Kanai, M.** & Okada, Y. Identification of genetic loci associated with chronic kidney disease and applications for drug development. *Kidney and Metabolic Bone Diseases* **31**, 19–26 (2018).
5. **Kanai, M.** & Okada, Y. HLA imputation: construction of a population-specific reference panel in Japanese. *Journal of Clinical and Experimental Medicine (IGAKU NO AYUMI)* **257**, 939–940 (2016).

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: May 31, 2019.