

# 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

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

**Advisor:** Drs. Yoichiro Kamatani & Yukinori Okada

### Research theme:

- 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

**Advisor:** Drs. Yukinori Okada & Toshihiro Tanaka

### Research theme:

- 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

**The Nakajima Foundation Fellowship** **Tokyo, Japan**  
Predoctoral fellowship which covers up to ~\$50,000/year for tuition and stipend. 2017–2019+

**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

## Technical Skills

**Programming Language:** C/C++, C#, Python, R, Java, Matlab, Unix shell, sed/awk

**Analysis:** GWAS, Genotype Imputation, HLA Imputation, Polygenic analysis using GCTA and LDSC

## Publications

1. 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).
2. 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).
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).
4. 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).
5. 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).
6. 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).
7. 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).

8. **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).
9. 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).
10. 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.**, 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).
2. **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).
3. **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. Suita, N., **Kanai, M.**, Higuchi, C., Tanaka, T. & Okada, Y. Comprehensive pathway analysis for 23 large-scale genome-wide association studies. *The 60th Annual Meeting of the Japan Society of Human Genetics, Poster Session* (2015).
3. **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: April 24, 2018.