Masahiro Kanai

Curriculum Vitae

mkanai@broadinstitute.org
mkanai@g.harvard.edu
mkanai.github.io
mkanai

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: Cross-population 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

Charles J. Epstein Trainee Awards for Excellence in Human Genetics Research **USA**
Finalist (Predoctoral), *The American Society of Human Genetics 2020 Virtual Meeting* *2020*

Reviewers' Choice Abstract Award **Houston, USA**
The American Society of Human Genetics 2019 Annual Meeting *2019*

Masason Foundation Fellowship **Tokyo, Japan**
Research grant for cross-population analysis of complex human diseases and traits. *2018–2022+*

Nakajima Foundation Fellowship **Tokyo, Japan**
Predoctoral fellowship which covers up to ~$50,000/year for tuition and stipend. *2017–2022*

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. The COVID-19 Host Genetics Initiative. Mapping the human genetic architecture of COVID-19. ***Nature*** (2021).
2. *Weissbrod, O., ****Kanai, M.***, *Shi, H., Gazal, S., Peyrot, W., Khera, A., Okada, Y., The Biobank Japan Project, Martin, A., Finucane, H. & Price, A. L. Leveraging fine-mapping and non-European training data to improve trans-ethnic polygenic risk scores. ***medRxiv*** (2021).
3. *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., FinnGen, 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).
4. *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).
5. 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).
6. **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

7. Palmer, D. S., Zhou, W., Abbott, L., Baya, N., Churchhouse, C., Seed, C., Poterba, T., King, D., **Kanai, M.**, Bloemendal, A. & Neale, B. M. Analysis of genetic dominance in the UK Biobank. ***bioRxiv*** (2021).
8. Reilly, S. K., Gosai, S. J., Guiterrez, A., Ulirsch, J. C., **Kanai, M.**, Berenzy, D., Kales, S., Butler, G. B., Gladden-Young, A., Finucane, H. K., Sabeti, P. C. & Tewhey, R. Direct characterization of cis-regulatory elements and functional dissection of complex genetic associations using HCR–FlowFISH. ***Nature Genetics*** **53,** 1166–1176 (2021).

9. Ruotsalainen, S. E., Surakka, I., Mars, N., Karjalainen, J., Kurki, M., **Kanai, M.**, Mishra, P. P., Mishra, B. H., Sinisalo, J., Palta, P., Lehtimaki, T., Raitakari, O., Estonian Biobank research team, Milani, L., The Biobank Japan Project, Okada, Y., FinnGen, Palotie, A., Widen, E., Daly, M. J. & Ripatti, S. Loss-of-function of MFGE8 and protection against coronary atherosclerosis. ***medRxiv*** (2021).
10. Wang, Q. S., Kelley, D. R., Ulirsch, J., **Kanai, M.**, Sadhuka, S., Cui, R., Albors, C., Cheng, N., Okada, Y., 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. ***Nature Communications*** **12,** 1–11 (2021).
11. Namkoong, H., Edahiro, R., Fukunaga, K., Shirai, Y., Sonehara, K., Tanaka, H., Lee, H., Hasegawa, T., **Kanai, M.**, Naito, T., Yamamoto, K., Saiki, R., Hyugaji, T., Shimizu, E., Katayama, K., Takahashi, K., Harada, N., Naito, T., Hiki, M., Matsushita, Y., Takagi, H., Aoki, R., Nakamura, A., Harada, S., Sasano, H., Kabata, H., Masaki, K., Kamata, H., Ikemura, S., Chubachi, S., Okamori, S., Terai, H., Morita, A., Asakura, T., Sasaki, J., Morisaki, H., Uwamino, Y., Nanki, K., Mikami, Y., Uchida, S., Uno, S., Ishihara, R., Matsubara, Y., Nishimura, T., Ogawa, T., Ishiguro, T., Isono, T., Shibata, S., Matsui, Y., Hosoda, C., Takano, K., Nishida, T., Kobayashi, Y., Takaku, Y., Takayanagi, N., Ueda, S., Tada, A., Miyawaki, M., Yamamoto, M., Yoshida, E., Hayashi, R., Nagasaka, T., Arai, S., Kaneko, Y., Sasaki, K., Tagaya, E., Kawana, M., Arimura, K., Takahashi, K., Anzai, T., Ito, S., Endo, A., Uchimura, Y., Miyazaki, Y., Honda, T., Tateishi, T., Tohda, S., Ichimura, N., Sonobe, K., Sassa, C., Nakajima, J., Nakano, Y., Nakajima, Y., Anan, R., Arai, R., Kurihara, Y., Harada, Y., Nishio, K., Ueda, T., Azuma, M., Saito, R., Sado, T., Miyazaki, Y., Sato, R., Haruta, Y., Nagasaki, T., Yasui, Y., Hasegawa, Y., Mutoh, Y., Sato, T., Takei, R., Hagimoto, S., Noguchi, Y., Yamano, Y., Sasano, H., Ota, S., Nakamori, Y., Yoshiya, K., Saito, F., Yoshihara, T., Wada, D., Iwamura, H., Kanayama, S., Maruyama, S., Yoshiyama, T., Ohta, K., Kokuto, H., Ogata, H., Tanaka, Y., Arakawa, K., Shimoda, M., Osawa, T., Tateno, H., Hase, I., Yoshida, S., Suzuki, S., Kawada, M., Horinouchi, H., Saito, F., Mitamura, K., Hagihara, M., Ochi, J., Uchida, T., Baba, R., Arai, D., Ogura, T., Takahashi, H., Hagiwara, S., Nagao, G., Konishi, S., Nakachi, I., Murakami, K., Yamada, M., Sugiura, H., Sano, H., Matsumoto, S., Kimura, N., Ono, Y., Baba, H., Suzuki, Y., Nakayama, S., Masuzawa, K., Namba, S., Suzuki, K., Hizawa, N., Shiroyama, T., Miyawaki, S., Kawamura, Y., Nakayama, A., Matsuo, H., Maeda, Y., Nii, T., Noda, Y., Niitsu, T., Adachi, Y., Enomoto, T., Amiya, S., Hara, R., Kishikawa, T., Yamada, S., Kawabata, S., Kijima, N., Takagaki, M., Sasa, N., Ueno, Y., Suzuki, M., Takemoto, N., Eguchi, H., Fukusumi, T., Imai, T., Fukushima, M., Kishima, H., Inohara, H., Tomono, K., Kato, K., Takahashi, M., Matsuda, F., Hirata, H., Takeda, Y., Koh, H., Manabe, T., Funatsu, Y., Ito, F., Fukui, T., Shinozuka, K., Kohashi, S., Miyazaki, M., Shoko, T., Kojima, M., Adachi, T., Ishikawa, M., Takahashi, K., Inoue, T., Hirano, T., Kobayashi, K., Takaoka, H., Watanabe, K., Miyazawa, N., Kimura, Y., Sado, R., Sugimoto, H., Kamiya, A., Kuwahara, N., Fujiwara, A., Matsunaga, T., Sato, Y., Okada, T., Hirai, Y., Kawashima, H., Narita, A., Niwa, K., Sekikawa, Y., Nishi, K., Nishitsuji, M., Tani, M., Suzuki, J., Nakatsumi, H., Ogura, T., Kitamura, H., Hagiwara, E., Murohashi, K., Okabayashi, H., Mochimaru, T., Nukaga, S., Satomi, R., Oyamada, Y., Mori, N., Baba, T., Fukui, Y., Odate, M., Mashimo, S., Makino, Y., Yagi, K., Hashiguchi, M., Kagyo, J., Shiomi, T., Fuke, S., Saito, H., Tsuchida, T., Fujitani, S., Takita, M., Morikawa, D., Yoshida, T., Izumo, T., Inomata, M., Kuse, N., Awano, N., Tone, M., Ito, A., Nakamura, Y., Hoshino, K., Maruyama, J., Ishikura, H., Takata, T., Odani, T., Amishima, M., Hattori, T., Shichinohe, Y., Kagaya, T., Kita, T., Ohta, K., Sakagami, S., Koshida, K., Hayashi, K., Shimizu, T., Kozu, Y., Hiranuma, H., Gon, Y., Izumi, N., Nagata, K., Ueda, K., Taki, R., Hanada, S., Kawamura, K., Ichikado, K., Nishiyama, K., Muranaka, H., Nakamura, K., Hashimoto, N., Wakahara, K., Koji, S., Omote, N., Ando, A., Kodama, N., Kaneyama, Y., Maeda, S., Kuraki, T., Matsumoto, T., Yokote, K., Nakada, T.-A., Abe, R., Oshima, T., Shimada, T., Harada, M., Takahashi, T., Ono, H., Sakurai, T., Shibusawa, T., Kimizuka, Y., Kawana, A., Sano, T., Watanabe, C., Suematsu, R., Sageshima, H., Yoshifuji, A., Ito, K., Takahashi, S., Ishioka, K., Nakamura, M., Masuda, M., Wakabayashi, A., Watanabe, H., Ueda, S., Nishikawa, M., Chihara, Y., Takeuchi, M., Onoi, K., Shinozuka, J., Sueyoshi, A., Nagasaki, Y., Okamoto, M., Ishihara, S., Shimo, M., Tokunaga, Y., Kusaka, Y., Ohba, T., Isogai, S., Ogawa, A., Inoue, T., Fukuyama, S., Eriguchi, Y., Yonekawa, A., Kan-o, K., Matsumoto, K., Kanaoka, K., Ihara, S., Komuta, K., Inoue, Y., Chiba, S., Yamagata, K., Hiramatsu, Y., Kai, H., Asano, K., Oguma, T., Ito, Y., Hashimoto, S., Yamasaki, M., Kasamatsu, Y., Komase, Y., Hida, N., Tsuburai, T., Oyama, B., Takada, M., Kanda, H., Kitagawa, Y., Fukuta, T.,

Miyake, T., Yoshida, S., Ogura, S., Abe, S., Kono, Y., Togashi, Y., Takoi, H., Kikuchi, R., Ogawa, S., Ogata, T., Ishihara, S., Kanehiro, A., Ozaki, S., Fuchimo, Y., Wada, S., Fujimoto, N., Nishiyama, K., Terashima, M., Beppu, S., Yoshida, K., Narumoto, O., Nagai, H., Ooshima, N., Motegi, M., Umeda, A., Miyagawa, K., Shimada, H., Endo, M., Ohira, Y., Watanabe, M., Inoue, S., Igarashi, A., Sato, M., Sagara, H., Tanaka, A., Ohta, S., Kimura, T., Shibata, Y., Tanino, Y., Nikaido, T., Minemura, H., Sato, Y., Yamada, Y., Hashino, T., Shinoki, M., Iwagoe, H., Takahashi, H., Fujii, K., Kishi, H., Kanai., M., Imamura, T., Yamashita, T., Yatomi, M., Maeno, T., Hayashi, S., Takahashi, M., Kuramochi, M., Kamimaki, I., Tominaga, Y., Ishii, T., Utsugi, M., Ono, A., Tanaka, T., Kashiwada, T., Fujita, K., Saito, Y., Seike, M., Omae, Y., Nannya, Y., Ueno, T., Takano, T., Katayama, K., Ai, M., Kumanogoh, A., Sato, T., Hasegawa, N., Tokunaga, K., Ishii, M., Koike, R., Kitagawa, Y., Kimura, A., Imoto, S., Miyano, S., Ogawa, S., Kanai, T. & Okada, Y. Japan COVID-19 Task Force: a nation-wide consortium to elucidate host genetics of COVID-19 pandemic in Japan. ***medRxiv*** (2021).

12. Koskela, J. T., Happola, P., Liu, A., Partanen, J., Genovese, G., Artomov, M., Myllymaki, M. N. M., **Kanai, M.**, Zhou, W., Karjalainen, J., Palviainen, T., Ronkainen, J., Sebert, S., Tukiainen, T., Palta, P., Kaprio, J., Kurki, M., Ganna, A., Palotie, A., Laitinen, T., Myllarniemi, M., Daly, M. J. & FinnGen. Genetic variant in SPDL1 reveals novel mechanism linking pulmonary fibrosis risk and cancer protection. ***medRxiv*** (2021).
13. Nakatochi, M., Toyoda, Y., **Kanai, M.**, Nakayama, A., Kawamura, Y., Hishida, A., Mikami, H., Matsuo, K., Takezaki, T., Momozawa, Y., Biobank Japan Project, Kamatani, Y., Ichihara, S., Shinomiya, N., Yokota, M., Wakai, K., Okada, Y., Matsuo, H. & Japan Uric Acid Genomics Consortium (Japan Urate). An X chromosome-wide meta-analysis based on Japanese cohorts revealed that non-autosomal variations are associated with serum urate. ***Rheumatology*** (2021).
14. Turley, P., Martin, A. R., Goldman, G., Li, H., **Kanai, M.**, Walters, R. K., Jala, J. B., Lin, K., Millwood, I. Y., Carey, C. E., Palmer, D. S., Zacher, M., Atkinson, E. G., Chen, Z., Li, L., Akiyama, M., Okada, Y., Kamatani, Y., Walters, R. G., Callier, S., Laibson, D., Meyer, M. N., Cesarini, D., Daly, M., Benjamin, D. J. & Neale, B. M. Multi-Ancestry Meta-Analysis yields novel genetic discoveries and ancestry-specific associations. ***bioRxiv*** (2021).
15. 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. E., Karjalainen, J., Als, T. D., Van der Zee, M. D., Day, F. R., Ong, K. K., Morisaki, T., de Geus, E., Bellocco, R., Okada, Y., Børglum, A. D., Joshi, P., Auton, A., Hinds, D., Neale, B. M., Walters, R. K., Nivard, M. G., Perry, J. R. B. & Ganna, A. Genetic analyses identify widespread sex-differential participation bias. ***Nature Genetics*** **53,** 663–671 (2021).
16. Shi, H., Gazal, S., **Kanai, M.**, Koch, E. M., Schoech, A. P., Siewert, K. M., Kim, S. S., Luo, Y., Amariuta, T., Huang, H., Okada, Y., Raychaudhuri, S., Sunyaev, S. R. & Price, A. L. Population-specific causal disease effect sizes in functionally important regions impacted by selection. ***Nature Communications*** **12,** 1098 (2021).
17. 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 uses local ancestry to enable the inclusion of admixed individuals in GWAS and to boost power. ***Nature Genetics*** **53,** 195–204 (2021).
18. Griesemer, D., Xue, J. R., Reilly, S. K., Ulirsch, J. C., Kukreja, K., Davis, J., **Kanai, M.**, Yang, D. K., Montgomery, S. B., Novina, C. D., Tewhey, R. & Sabeti, P. C. Genome-wide functional screen of 3'UTR variants uncovers causal variants for human disease and evolution. ***bioRxiv*** (2021).
19. 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).

20. 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).

21. 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., Kashihara, 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).

22. 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).

23. 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).

24. 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).

25. 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).

26. 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).

27. 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).

28. 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).

29. 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).

30. 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).

31. 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).

32. 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., Gieger, C., Girotto, G., Gögele, M., Gordon, S. D., Gudbjartsson, D. F., Gudnason, V., Haller, T., Hamet, P., Harris, T. B., Hayward, C., Hicks, A. A., Hofer, E., Holm, H., Huang, W., Hutri-Kähönen, N., Hwang, S.-J., Ikram, M. A., Lewis, R. M., Ingelsson, E., Jakobsdottir, J., Jonsdottir, I., Jonsson, H., Joshi, P. K., Josyula, N. S., Jung, B., Kähönen, M., Kamatani, Y., **Kanai, M.**, Kerr, S. M., Kiess, W., Kleber, M. E., Koenig, W., Kooner, J. S., Körner, A., Kovacs, P., Krämer, B. K., Kronenberg, F., Kubo, M., Kühnel, B., La Bianca, M., Lange, L. A., Lehne, B., Lehtimäki, T., Liu, J., Loeffler, M., Loos, R. J. F., Lyytikäinen, L.-P., Magi, R., Mahajan, A., Martin, N. G., März, W., Mascalzoni, D., Matsuda, K., Meisinger, C., Meitinger, T., Metspalu, A., Milaneschi, Y., O'Donnell, C. J., Wilson, O. D., Gaziano, J. M., Mishra, P. P., Mohlke, K. L., Mononen, N., Montgomery, G. W., Mook-Kanamori, D. O., Müller-Nurasyid, M., Nadkarni, G. N., Nalls, M. A., Nauck, M., Nikus, K., Ning, B., Nolte, I. M., Noordam, R., O'Connell, J. R., Olafsson, I., Padmanabhan, S., Penninx, B. W. J. H., Perls, T., Peters, A., Pirastu, M., Pirastu, N., Pistis, G., Polasek, O., Ponte, B., Porteous, D. J., Poulain, T., Preuss, M. H., Rabelink, T. J., Raffield, L. M., Raitakari, O. T., Rettig, R., Rheinberger, M., Rice, K. M., Rizzi, F., Robino, A., Rudan, I., Krajcoviechova, A., Cifkova, R., Rueedi, R., Ruggiero, D., Ryan, K. A., Saba, Y., Salvi, E., Schmidt, H., Schmidt, R., Shaffer, C. M., Smith, A. V., Smith, B. H., Spracklen, C. N., Strauch, K., Stumvoll, M., Sulem, P., Tajuddin, S. M., Teren, A., Thiery, J., Thio, C. H. L., Thorsteinsdottir, U., Toniolo, D., Tönjes, A., Tremblay, J., Uitterlinden, A. G., Vaccargiu, S., van der Harst, P., van Duijn, C. M., Verweij, N., Völker, U., Vollenweider, P., Waeber, G., Waldenberger, M., Whitfield, J. B., Wild, S. H., Wilson, J. F., Yang, Q., Zhang, W., Zonderman, A. B., Bochud, M., Wilson, J. G., Pendergrass, S. A., Ho, K., Parsa, A., Pramstaller, P. P., Psaty, B. M., Böger, C. A., Snieder, H., Butterworth, A. S., Okada, Y., Edwards, T. L., Stefansson, K., Susztak, K., Scholz, M., Heid, I. M., Hung, A. M., Teumer, A., Pattaro, C., Woodward, O. M., Vitart, V. & Köttgen, A. Target genes, variants, tissues and transcriptional pathways influencing human serum urate levels. ***Nature Genetics*** **51,** 1459–1474 (2019).

33. Akiyama, M., Ishigaki, K., Sakaue, S., Momozawa, Y., Horikoshi, M., Hirata, M., Matsuda, K., Ikegawa, S., Takahashi, A., **Kanai, M.**, Suzuki, S., Matsui, D., Naito, M., Yamaji, T., Iwasaki, M., Sawada, N., Tanno, K., Sasaki, M., Hozawa, A., Minegishi, N., Wakai, K., Tsugane, S., Shimizu, A., Yamamoto, M., Okada, Y., Murakami, Y., Kubo, M. & Kamatani, Y. Characterizing rare and low-frequency height-associated variants in the Japanese population. ***Nature Communications*** **10,** 4393 (2019).

34. 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., Mikaelsdottir, E. K., Milaneschi, Y., Miliku, K., Mishra, P. P., MohIke, 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., Vogelezang, 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).

35. *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).

36. 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).

37. 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).

38. 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 histocompatibilty complex region in the Japanese population. ***Nature Genetics*** **51,** 470–480 (2019).

39. **Kanai, M.**, Maeda, Y. & Okada, Y. Grimon: graphical interface to visualize multi-omics networks. ***Bioinformatics*** **34,** 3934–3936 (2018).

40. 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).

41. 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).

42. 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).

43. 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).

44. 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).

45. 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).

46. 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).

47. **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).
48. 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).
49. 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 & Allergology*** **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*** (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.**, Ulirsch, J. C., Karjalainen, J., Kurki, M., Ishigaki, K., Kamatani, Y., Reilly, S., Sabeti, P., Tewhey, R., FinnGen, Okada, Y., Daly, M. J. & Finucane, H. K. Insights into fine-mapping causal variants of complex traits from diverse populations. ***The 70th Annual Meeting of the American Society of Human Genetics, Plenary Session*** (2020).
2. **Kanai, M.**, Ulirsch, J. C., McCaw, Z. R., Albors, C., Fan, Z., Ishigaki, K., Karjalainen, J., Kurki, M., FinnGen, Kamatani, Y., Okada, Y., Daly, M. J. & Finucane, H. K. Cross-population fine-mapping of 50 complex traits and diseases in 675,000 individuals across three global biobanks. ***The 69th Annual Meeting of the American Society of Human Genetics, Poster Session*** (2019).
3. **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).
4. **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).
5. **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: August 29, 2021.