Masahiro Kanai

Curriculum Vitae

☑ mkanai@broadinstitute.org

☑ mkanai@g.harvard.edu

mkanai.github.io

nkanai 🕜

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:

o Genetic analysis of complex human diseases and traits in the Japanese population.

o 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:

o Genome-wide association study (GWAS) of ~200,000 individuals in the BioBank Japan Project.

o Interpretation of GWAS polygenic signals using epigenomic data.

o 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.

o 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. 2017-2021 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. 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. 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* (2021).

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).

- 9. 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).
- 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).

- 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).
- 12. 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).
- 13. 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).
- 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).
- 15. 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).
- 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).
- 17. 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).
- 18. 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. Genomewide Screen of Otosclerosis in Population Biobanks: 18 Loci and Shared Heritability with Skeletal Structure. *medRxiv* (2020).

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).

- 20. 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).
- 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).
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