

Masahiro Kanai

Curriculum Vitae

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Education

Harvard University **Boston, USA**
Bioinformatics and Integrative Genomics PhD Program, Harvard Medical School Aug. 2017–present

Keio University **Yokohama, Japan**
B.S. in Bioinformatics, Department of Biosciences and Informatics Apr. 2013–Mar. 2017

Research Experience

Massachusetts General Hospital & Broad Institute of MIT and Harvard **Boston, USA**
Graduate Student, Analytic and Translational Genetics Unit Jan. 2018–present

Advisors: Drs. Mark Daly & Hilary Finucane

Research theme: 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-dimensional 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

Reviewers' Choice Abstract Award	USA
<i>The American Society of Human Genetics 2021 Virtual Meeting</i>	2021
Charles J. Epstein Trainee Awards for Excellence in Human Genetics Research	USA
Finalist (Predoctoral), <i>The American Society of Human Genetics 2020 Virtual Meeting</i>	2020
Reviewers' Choice Abstract Award	Houston, USA
<i>The American Society of Human Genetics 2019 Annual Meeting</i>	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
<i>Microsoft Imagine Cup 2011 Windows 7 Touch Challenge</i>	2011
The 1st place, National Finals & Worldwide Finalist	Japan & Poland
<i>Microsoft Imagine Cup 2010 Software Design Competition</i>	2010

Publications

Selected Publications and Preprints

(* = co-first)

1. **Kanai, M.**, Ulirsch, J. C., Karjalainen, J., Kurki, M., Karczewski, K. J., Fauman, E. B., Wang, Q. S., Jacobs, H., Aguet, F., Ardlie, K. G., Kerimov, N., Alasoo, K., Benner, C., Ishigaki, K., Sakaue, S., Reilly, S., The BioBank Japan Project, FinnGen, Kamatani, Y., Matsuda, K., Palotie, A., Neale, B. M., Tewhey, R., Sabeti, P. C., Okada, Y., Daly, M. J. & Finucane, H. K. Insights from complex trait fine-mapping across diverse populations. *medRxiv* (2021).
2. *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., Shinozaki, N., Yamaguchi, H., Minami, S., Murayama, S., Yoshimori, K., Nagayama, S., Obata, D., Higashiyama, M., Masumoto, A., Koretsune, Y., 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. J., Matsuda, K. & Okada, Y. A cross-population atlas of genetic associations for 220 human phenotypes. *Nature Genetics* **53**, 1415–1424 (2021).
3. The COVID-19 Host Genetics Initiative. Mapping the human genetic architecture of COVID-19. *Nature* (2021).
4. *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).
5. *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).
6. 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).
7. **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.....

8. Global Biobank Meta-analysis Initiative. Global Biobank Meta-analysis Initiative: powering genetic discovery across human diseases. *medRxiv* (2021).
9. Wang, Y., Namba, S., Lopera-Maya, E. A., Kerminen, S., Tsuo, K., Lall, K., **Kanai, M.**, Zhou, W., Wu, K.-H. H., Fave, M.-J., Bhatta, L., Awadalla, P., Deelen, P., Lo Faro, V., Magi, R., Murakami, Y., Brumpton, B. M., Sanna, S., Uzunovic, J., Gamazon, E. R., Cox, N. J., Surakka, I., Okada, Y., Martin, A. R. & Hirbo, J. Global biobank analyses provide lessons for computing polygenic risk scores across diverse cohorts. *medRxiv* (2021).
10. Polygenic Risk Score Task Force of the International Common Disease Alliance. Responsible use of polygenic risk scores in the clinic: potential benefits, risks and gaps. *Nature Medicine* (2021).
11. The COVID-19 Host Genetics Initiative. Mapping the human genetic architecture of COVID-19: an update. *medRxiv* (2021).
12. Luo, Y., **Kanai, M.**, Choi, W., Li, X., Sakaue, S., Yamamoto, K., Ogawa, K., Gutierrez-Arcelus, M., Gregersen, P. K., Stuart, P. E., Elder, J. T., Forer, L., Schönherr, S., Fuchsberger, C., Smith, A. V., Fellay, J., Carrington, M., Haas, D. W., Guo, X., Palmer, N. D., Chen, Y.-D. I., Rotter, J. I., Taylor, K. D., Rich, S. S., Correa, A., Wilson, J. G., Kathiresan, S., Cho, M. H., Metspalu, A., Esko, T., Okada, Y., Han, B., McLaren, P. J. & Raychaudhuri, S. A high-resolution HLA reference panel capturing global population diversity enables multi-ancestry fine-mapping in HIV host response. *Nature Genetics* **53**, 1504–1516 (2021).
13. Griesemer, D., Xue, J. R., Reilly, S. K., Ulirsch, J. C., Kukreja, K., Davis, J. R., **Kanai, M.**, Yang, D. K., Butts, J. C., Guney, M. H., Luban, J., Montgomery, S. B., Finucane, H. K., Novina, C. D., Tewhey, R. & Sabeti, P. C. Genome-wide functional screen of 3'UTR variants uncovers causal variants for human disease and evolution. *Cell* **184**, 5247–5260.e19 (2021).
14. 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).
15. 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).
16. Ruotsalainen, S. E., Surakka, I., Mars, N., Karjalainen, J., Kurki, M., **Kanai, M.**, Mishra, P. P., Mishra, B. H., Sinisalo, J., Palta, P., Lehtimäki, 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).
17. Wang, Q. S., Kelley, D. R., Ulirsch, J., **Kanai, M.**, Sadhuka, S., Cui, R., Albers, 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).
18. 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).
19. 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., Seibert, 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).
 20. 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* **60**, 4430–4432 (2021).
21. 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).
 22. 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., Børghlum, 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).
 23. 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).
 24. 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).
 25. 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).
 26. 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).
 27. 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).
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