Saori Sakaue, MD, PhD

Date Prepared: 04/02/2025

Office Address: 3720 15th Ave NE, Foege S130, Seattle, WA 98105

Email: sakaue@uw.edu

Personal Statement:

I am a physician-scientist with a strong background in statistical genetics, functional genomics and clinical medicine (rheumatology). I study how genetic variations affect our disease susceptibility. I develop statistical genetics tools that combine genetics and novel high-dimensional experimental genomic data. I will use these tools to bridge the current gap from disease risk alleles to causal disease mechanisms, diagnosis, and data-driven treatment strategy.

Education:

| 2011 | MD | Medicine | Faculty of Medicine, The University of Tokyo, |
|------|-----|------------------------|--|
| | | | Tokyo, Japan |
| 2020 | PhD | Statistical Genetics / | Graduate School of Medicine, The University of |
| | | Medical Sciences | Tokyo, Tokyo, Japan |

Academic and Clinical Appointments:

| | • • | | |
|-----------------|--|--|--|
| 04/2011-03/2016 | Resident and Clinical Fellow | Internal Medicine and Rheumatology | The University of Tokyo Hospital |
| 04/2020-09/2020 | Assistant Professor | Department of Statistical Genetics | Osaka University, Japan |
| 09/2020-03/2024 | Postdoctoral Research Fellow | Division of Genetics (Dr. Soumya Raychaudhuri) | Brigham & Women's Hospital, Harvard Medical School and Broad Institute of MIT and Harvard |
| 04/2024-03/2025 | Instructor | Division of Genetics (Dr. Soumya Raychaudhuri) | Brigham & Women's Hospital, Harvard Medical School and Broad Institute of MIT and Harvard |
| 04/2025-present | Assistant Professor (Principal Investigator) | Department of Genome Sciences | University of Washington School of Medicine |

Professional Societies:

2018- American Society of Human Genetics

Honors and Prizes:

| 2010 | Otsubo Tetsumon Fellowship Award | The University of Tokyo | Academic achievements |
|------|---|---|-----------------------|
| 2017 | Finalist of Meeting Award | the 62th Annual Meeting of the Japan Society of Human Genetics | Research |
| 2018 | Semifinalist | American Society of Human Genetics/Charles J. Epstein Trainee Award for Excellence in Human Genetics Research | Research |
| 2019 | Finalist | American Society of Human Genetics/Charles J. Epstein Trainee Award for Excellence in Human Genetics Research | Research |
| 2020 | The President's Award | The University of Tokyo | Academic achievements |
| 2020 | L'Oréal-UNESCO For W | omen in Science International Award | Academic achievements |
| 2020 | Young Investigator's Award | The Japan Society of Human Genetics | Academic achievements |
| 2022 | Young Scientists' Prize | Ministry of Education, Culture, and Technology, Japan | Academic achievements |
| 2022 | Poster Prize (3rd) | Impact of Genomic Variation on Function (IGVF) Annual Meeting | Research |
| 2022 | Semifinalist | American Society of Human Genetics/Charles J. Epstein Trainee Award for Excellence in Human Genetics Research | Research |
| 2023 | Stellar Abstract Award | 17th Annual Program in Quantitative Genomics Conference | Research |
| 2023 | Eric S. Lander Prize in Scientific Excellence | The Broad Institute | Academic achievements |

Publications: (Selected publications are highlighted in blue)

- Sakaue S, Accelerating Medicines Partnership®: RA/SLE Network, Soumya Raychaudhuri. Early and late RNA eQTL are driven by different genetic mechanisms. bioRxiv. 2025 doi: 10.1101/2025.02.24.639351 [Preprint]
- 2. Rumker L, **Sakaue S**, Reshef Y, Kang JB, Yazar S, Alquicira-Hernandez J, Valencia C, Lagattuta KA, Mah-Som A, Nathan A, Powell JE, Loh PR, Raychaudhuri S. *Nat Genet*. 2024 Oct;56(10):2068-2077. doi: 10.1038/s41588-024-01909-1
- 3. **IGVF Consortium**. Deciphering the impact of genomic variation on function. *Nature*. 2024 Sep;633(8028):47-57. doi: 10.1038/s41586-024-07510-0.
- 4. Krishna C, Chiou J, **Sakaue S**, Kang JB, Christensen SM, Lee I, Aksit MA, Kim HI, von Schack D, Raychaudhuri S, Ziemek D, Hu X. The influence of HLA genetic variation on plasma protein expression. *Nat Commun*. 2024 Jul 31;15(1):6469. doi: 10.1038/s41467-024-50583-8.
- 5. Dunlap G, Wagner A, Meednu N, Wang R, Zhang F, Ekabe JC, Jonsson AH, Wei K, Sakaue S, Nathan A; Accelerating Medicines Partnership Program: Rheumatoid Arthritis and Systemic Lupus Erythematosus (AMP RA/SLE) Network; Bykerk VP, Donlin LT, Goodman SM, Firestein GS, Boyle DL, Holers VM, Moreland LW, Tabechian D, Pitzalis C, Filer A, Raychaudhuri S, Brenner MB, Thakar J, McDavid A, Rao DA, Anolik JH. Clonal associations between lymphocyte subsets and functional states in rheumatoid arthritis synovium. *Nat Commun.* 2024 Jun 11;15(1):4991. doi: 10.1038/s41467-024-49186-0.
- 6. Weinand K, **Sakaue S**, Nathan A, Jonsson AH, Zhang F, Watts GFM, Al Suqri M, Zhu Z; Accelerating Medicines Partnership Program: Rheumatoid Arthritis and Systemic Lupus

- Erythematosus (AMP RA/SLE) Network; Rao DA, Anolik JH, Brenner MB, Donlin LT, Wei K, Raychaudhuri S. The chromatin landscape of pathogenic transcriptional cell states in rheumatoid arthritis. *Nat Commun.* 2024 May 31;15(1):4650. doi: 10.1038/s41467-024-48620-7.
- 7. **Sakaue S**. SCENT defines non-coding disease mechanisms using single-cell multi-omics. *Nat Rev Genet*. 2024 Sep;25(9):597. doi: 10.1038/s41576-024-00747-5.
- 8. **Sakaue S**, Weinand K, Dey KK, Jagadeesh K, Kanai VO, Watts GF, Zhu Z, Network AM, Brenner MB, McDavid A, Donlin LT, Wei K, Price AL, Raychaudhuri S. Tissue-specific enhancer-gene maps from multimodal single-cell data identify causal disease alleles. *Nat Genet*. 2024 Apr:56(4):615-626. doi: 10.1038/s41588-024-01682-1.
- 9. Kang JB, Shen AZ, Gurajala S, Nathan A, Rumker L, Aguiar VRC, Valencia C, Lagattuta KA, Zhang F, Jonsson AH, Yazar S, Alquicira-Hernandez J, Khalili H, Ananthakrishnan AN, Jagadeesh K, Dey K; Accelerating Medicines Partnership Program: Rheumatoid Arthritis and Systemic Lupus Erythematosus (AMP RA/SLE) Network; Daly MJ, Xavier RJ, Donlin LT, Anolik JH, Powell JE, Rao DA, Brenner MB, Gutierrez-Arcelus M, Luo Y, Sakaue S, Raychaudhuri S. Mapping the dynamic genetic regulatory architecture of HLA genes at single-cell resolution. Nat Genet. 2023 Nov 30. doi: 10.1038/s41588-023-01586-6.
- 10. Gupta A, Weinand K, Nathan A, Sakaue S, Zhang MJ; Accelerating Medicines Partnership RA/SLE Program and Network; Donlin L, Wei K, Price AL, Amariuta T, Raychaudhuri S. Dynamic regulatory elements in single-cell multimodal data implicate key immune cell states enriched for autoimmune disease heritability. *Nat Genet*. 2023 Nov 30. doi: 10.1038/s41588-023-01577-7.
- **11. Sakaue S**, Gurajala S, Curtis M, Luo Y, Choi W, Ishigaki K, Kang JB, Rumker L, Deutsch AJ, Schönherr S, Forer L, LeFaive J, Fuchsberger C, Han B, Lenz TL, Bakker PI, Smith AV, Raychaudhuri S. A statistical genetics guide to identifying HLA alleles driving complex disease. *Nat Protoc.* 2023 Jul 26. doi: 10.1038/s41596-023-00853-4.
- 12. Ishigaki K*, Sakaue S*, Terao C*, Luo Y, Sonehara K, Yamaguchi K, Amariuta T, Too CL, Laufer VA, Scott IC, Viatte S, Takahashi M, Ohmura K, Murasawa A, Hashimoto M, Ito H, Hammoudeh M, Emadi SA, Masri BK, Halabi H, Badsha H, Uthman IW, Wu X, Lin L, Li T, Plant D, Barton A, Orozco G, Verstappen SMM, Bowes J, MacGregor AJ, Honda S, Koido M, Tomizuka K, Kamatani Y, Tanaka H, Tanaka E, Suzuki A, Maeda Y, Yamamoto K, Miyawaki S, Xie G, Zhang J, Amos Cl, Keystone E, Wolbink G, van der Horst-Bruinsma I, Cui J, Liao KP, Carroll RJ, Lee HS, Bang SY, Siminovitch KA, de Vries N, Alfredsson L, Rantapää-Dahlqvist S, Karlson EW, Bae SC, Kimberly RP, Edberg JC, Mariette X, Huizinga T, Dieudé P, Schneider M, Kerick M, Denny JC; BioBank Japan Project; Matsuda K, Matsuo K, Mimori T, Matsuda F, Fujio K, Tanaka Y, Kumanogoh A, Traylor M, Lewis CM, Eyre S, Xu H, Saxena R, Arayssi T, Kochi Y, Ikari K, Harigai M, Gregersen PK, Yamamoto K, Louis Bridges S Jr, Padyukov L, Martin J, Klareskog L, Okada Y, Raychaudhuri S. Trans-ancestry genome-wide association study identifies novel genetic mechanisms in rheumatoid arthritis. Nat Genet. 2022 Nov;54(11):1640-1651. doi: 10.1038/s41588-022-01213-w.
- 13. Yengo L, Vedantam S, Marouli E, Sidorenko J, Bartell E, **Sakaue S**, et al. (more than 500 authors) A saturated map of common genetic variants associated with human height. *Nature*. 2022 Oct;610(7933):704-712. doi: 10.1038/s41586-022-05275-y.
- 14. Zhang M, Hou K, Dey K, **Sakaue S**, Jagadeesh K, Weinand K, Taychameekiatchai A, Rao P, Pisco A, Zhou J, Wang B, Gandal M, Raychaudhuri S, Pasaniuc B, and Price A. Polygenic enrichment distinguishes disease associations of individual cells in single-cell RNA-seq data. *Nat Genet*. 2022 Oct;54(10):1572-1580. doi: 10.1038/s41588-022-01167-z.
- 15. Sakaue S, Hosomichi K, Hirata J, Nakaoka H, Yamazaki K, Yawata M, Naito T, Yawata N, Umeno, Takaaki Kawaguchi, Toshiyuki Matsui, Satoshi Motoya, Yasuo Suzuki, Hidetoshi Inoko, Atsushi Tajima, Takayuki Morisaki, Koichi Matsuda, Yoichiro Kamatani, Kazuhiko Yamamoto, Ituro Inoue, Yukinori Okada. Decoding the diversity of killer immunoglobulin-like receptor by deep target sequencing and a high-resolution imputation method. *Cell Genomics* 2(3) 100101-100101 (2022).

- 16. Sonehara K*, Sakaue S*, Maeda Y, Hirata J, Kishikawa T, Yamamoto K, Matsuoka H, Yoshimura M, Nii T, Ohshima S, Kumanogoh A, Okada Y. Genetic architecture of microRNA expression and its link to complex diseases in the Japanese population. *Hum Mol Genet*. 2022 Jun 4;31(11):1806-1820.
- 17. Graham SE, Clarke SL, Wu KH, ... **Sakaue S**, et al. (more than 400 authors) The power of genetic diversity in genome-wide association studies of lipids. *Nature*. 2021 Dec;600(7890):675-679.
- Sakaue S, Kanai M, Tanigawa Y, Karjalainen J, Kurki M, Yamamoto K, Konuma T, FinnGen, Biobank Japan, Kamatani Y, Palotie A, Rivas M, Daly MJ, Matsuda K, Okada Y. A crosspopulation atlas of genetic associations for 220 human phenotypes. *Nat Genet.* 2021 Oct;53(10):1415-1424
- 19. Luo Y, Kanai M, Choi W, Li X, **Sakaue S**, Yamamoto K, Ogawa K, Gutierrez-Arcelus M, Gregersen PK, Stuart PE, Elder JT, Forer L, Schönherr S, Fuchsberger C, Smith AV, Fellay J, Carrington M, Haas DW, Guo X, Palmer ND, Chen YI, Rotter JI, Taylor KD, Rich SS, Correa A, Wilson JG, Kathiresan S, Cho MH, Metspalu A, Esko T, Okada Y, Han B; NHLBI Trans-Omics for Precision Medicine (TOPMed) Consortium, McLaren PJ, Raychaudhuri S. A high-resolution HLA reference panel capturing global population diversity enables multi-ancestry fine-mapping in HIV host response. *Nat Genet.* 2021 Oct;53(10):1504-1516.
- 20. Sakaue S, Yamaguchi E, Inoue Y, Takahashi M, Hirata J, Suzuki K, Ito S, Arai T, Hirose M, Tanino Y, Nikaido T, Ichiwata T, Ohkouchi S, Hirano T, Takada T, Miyawaki S, Dofuku S, Maeda Y, Nii T, Kishikawa T, Ogawa K, Masuda T, Yamamoto K, Sonehara K, Tazawa R, Morimoto K, Takaki M, Konno S, Suzuki M, Tomii K, Nakagawa A, Handa T, Tanizawa K, Ishii H, Ishida M, Kato T, Takeda N, Yokomura K, Matsui T, Watanabe M, Inoue H, Imaizumi K, Goto Y, Kida H, Fujisawa T, Suda T, Yamada T, Satake Y, Ibata H, Hizawa N, Mochizuki H, Kumanogoh A, Matsuda F, Nakata K, Hirota T, Tamari M, Okada Y. Genetic determinants of risk in autoimmune pulmonary alveolar proteinosis. *Nat Commun.* 2021 Feb 15;12(1):1032.
- 21. Lin BM, Grinde KE, Brody JA, Breeze CE, Raffield LM, Mychaleckyj JC, Thornton TA, Perry JA, Baier LJ, de Las Fuentes L, Guo X, Heavner BD, Hanson RL, Hung YJ, Qian H, Hsiung CA, Hwang SJ, Irvin MR, Jain D, Kelly TN, Kobes S, Lange L, Lash JP, Li Y, Liu X, Mi X, Musani SK, Papanicolaou GJ, Parsa A, Reiner AP, Salimi S, Sheu WH, Shuldiner AR, Taylor KD, Smith AV, Smith JA, Tin A, Vaidya D, Wallace RB, Yamamoto K, Sakaue S, Matsuda K, Kamatani Y, Momozawa Y, Yanek LR, Young BA, Zhao W, Okada Y, Abecasis G, Psaty BM, Arnett DK, Boerwinkle E, Cai J, Yii-Der Chen I, Correa A, Cupples LA, He J, Kardia SL, Kooperberg C, Mathias RA, Mitchell BD, Nickerson DA, Turner ST, Vasan RS, Rotter JI, Levy D, Kramer HJ, Köttgen A, Nhlbi Trans-Omics For Precision Medicine TOPMed Consortium, TOPMed Kidney Working Group, Rich SS, Lin DY, Browning SR, Franceschini N. Whole genome sequence analyses of eGFR in 23,732 people representing multiple ancestries in the NHLBI trans-omics for precision medicine (TOPMed) consortium. *EBioMedicine*. 2021 Jan;63:103157.
- 22. Sargurupremraj M, Suzuki H, Jian X, Sarnowski C, Evans TE, Bis JC, Eiriksdottir G, Sakaue S, Terzikhan N, Habes M, Zhao W, Armstrong NJ, Hofer E, Yanek LR, Hagenaars SP, Kumar RB, van den Akker EB, McWhirter RE, Trompet S, Mishra A, Saba Y, Satizabal CL, Beaudet G, Petit L, Tsuchida A, Zago L, Schilling S, Sigurdsson S, Gottesman RF, Lewis CE, Aggarwal NT, Lopez OL, Smith JA, Valdés Hernández MC, van der Grond J, Wright MJ, Knol MJ, Dörr M, Thomson RJ, Bordes C, Le Grand Q, Duperron MG, Smith AV, Knopman DS, Schreiner PJ, Evans DA, Rotter JI, Beiser AS, Maniega SM, Beekman M, Trollor J, Stott DJ, Vernooij MW, Wittfeld K, Niessen WJ, Soumaré A, Boerwinkle E, Sidney S, Turner ST, Davies G, Thalamuthu A, Völker U, van Buchem MA, Bryan RN, Dupuis J, Bastin ME, Ames D, Teumer A, Amouyel P, Kwok JB, Bülow R, Deary IJ, Schofield PR, Brodaty H, Jiang J, Tabara Y, Setoh K, Miyamoto S, Yoshida K, Nagata M, Kamatani Y, Matsuda F, Psaty BM, Bennett DA, De Jager PL, Mosley TH, Sachdev PS, Schmidt R, Warren HR, Evangelou E, Trégouët DA; International Network against Thrombosis (INVENT) Consortium; International Headache Genomics Consortium (IHGC), Ikram MA, Wen W, DeCarli C, Srikanth VK, Jukema JW, Slagboom EP, Kardia SLR, Okada Y, Mazoyer B, Wardlaw

- JM, Nyquist PA, Mather KA, Grabe HJ, Schmidt H, Van Duijn CM, Gudnason V, Longstreth WT Jr, Launer LJ, Lathrop M, Seshadri S, Tzourio C, Adams HH, Matthews PM, Fornage M, Debette S. Cerebral small vessel disease genomics and its implications across the lifespan. *Nat Commun.* 2020 Dec 8;11(1):6285.
- 23. Masuda T, Ito H, Hirata J, **Sakaue S**, Ueda Y, Kimura T, Takeuchi F, Murakami Y, Matsuda K, Matsuo K, Okada Y. Fine Mapping of the Major Histocompatibility Complex Region and Association of the HLA-B*52:01 Allele With Cervical Cancer in Japanese Women. *JAMA Netw Open.* 2020 Oct 1;3(10):e2023248.
- 24. Vuckovic D, Bao EL, Akbari P, Lareau CA, Mousas A, Jiang T, Chen MH, Raffield LM, Tardaguila M, Huffman JE, Ritchie SC, Megy K, Ponstingl H, Penkett CJ, Albers PK, Wigdor EM, Sakaue S, Moscati A. Manansala R. Lo KS, Qian H, Akiyama M, Bartz TM, Ben-Shlomo Y, Beswick A, Bork-Jensen J, Bottinger EP, Brody JA, van Rooij FJA, Chitrala KN, Wilson PWF, Choquet H, Danesh J, Di Angelantonio E, Dimou N, Ding J, Elliott P, Esko T, Evans MK, Felix SB, Floyd JS, Broer L, Grarup N, Guo MH, Guo Q, Greinacher A, Haessler J, Hansen T, Howson JMM, Huang W, Jorgenson E, Kacprowski T, Kähönen M, Kamatani Y, Kanai M, Karthikeyan S, Koskeridis F, Lange LA, Lehtimäki T, Linneberg A, Liu Y, Lyytikäinen LP, Manichaikul A, Matsuda K, Mohlke KL, Mononen N, Murakami Y, Nadkarni GN, Nikus K, Pankratz N, Pedersen O, Preuss M, Psaty BM. Raitakari OT, Rich SS, Rodriguez BAT, Rosen JD, Rotter JI, Schubert P, Spracklen CN, Surendran P, Tang H, Tardif JC, Ghanbari M, Völker U, Völzke H, Watkins NA, Weiss S; VA Million Veteran Program, Cai N, Kundu K, Watt SB, Walter K, Zonderman AB, Cho K, Li Y, Loos RJF, Knight JC, Georges M, Stegle O, Evangelou E, Okada Y, Roberts DJ, Inouye M, Johnson AD, Auer PL, Astle WJ, Reiner AP, Butterworth AS, Ouwehand WH, Lettre G, Sankaran VG, Soranzo N. The Polygenic and Monogenic Basis of Blood Traits and Diseases. Cell. 2020 Sep. 3;182(5):1214-1231.e11.
- 25. Chen MH, Raffield LM, Mousas A, Sakaue S, Huffman JE, Moscati A, Trivedi B, Jiang T, Akbari P, Vuckovic D. Bao EL, Zhong X, Manansala R, Laplante V, Chen M, Lo KS, Qian H, Lareau CA, Beaudoin M, Hunt KA, Akiyama M, Bartz TM, Ben-Shlomo Y, Beswick A, Bork-Jensen J, Bottinger EP, Brody JA, van Rooij FJA, Chitrala K, Cho K, Choquet H, Correa A, Danesh J, Di Angelantonio E, Dimou N, Ding J, Elliott P, Esko T, Evans MK, Floyd JS, Broer L, Grarup N, Guo MH, Greinacher A, Haessler J, Hansen T, Howson JMM, Huang QQ, Huang W, Jorgenson E. Kacprowski T, Kähönen M, Kamatani Y, Kanai M, Karthikeyan S, Koskeridis F, Lange LA, Lehtimäki T, Lerch MM, Linneberg A, Liu Y, Lyytikäinen LP, Manichaikul A, Martin HC, Matsuda K, Mohlke KL, Mononen N, Murakami Y, Nadkarni GN, Nauck M, Nikus K, Ouwehand WH, Pankratz N, Pedersen O, Preuss M, Psaty BM, Raitakari OT, Roberts DJ, Rich SS, Rodriguez BAT, Rosen JD, Rotter JI, Schubert P, Spracklen CN, Surendran P, Tang H, Tardif JC, Trembath RC, Ghanbari M, Völker U, Völzke H, Watkins NA, Zonderman AB; VA Million Veteran Program, Wilson PWF, Li Y, Butterworth AS, Gauchat JF, Chiang CWK, Li B, Loos RJF, Astle WJ, Evangelou E, van Heel DA, Sankaran VG, Okada Y, Soranzo N, Johnson AD, Reiner AP, Auer PL, Lettre G. Trans-ethnic and Ancestry-Specific Blood-Cell Genetics in 746,667 Individuals from 5 Global Populations. Cell. 2020 Sep 3;182(5):1198-1213.e14.
- 26. Ishigaki K, Akiyama M, Kanai M, Takahashi A, Kawakami E, Sugishita H, **Sakaue S**, Matoba N, Low SK, 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. *Nat Genet.* 2020 Jul;52(7):669-679.
- 27. Matsunaga H, Ito K, Akiyama M, Takahashi A, Koyama S, Nomura S, Ieki H, Ozaki K, Onouchi Y, **Sakaue S**, Suna S, Ogishima S, Yamamoto M, Hozawa A, Satoh M, Sasaki M, Yamaji T, Sawada N, Iwasaki M, Tsugane S, Tanaka K, Arisawa K, Ikezaki H, Takashima N, Naito M, Wakai K, Tanaka H, Sakata Y, Morita H, Sakata Y, Matsuda K, Murakami Y, Akazawa H, Kubo M, Kamatani Y, Komuro I. Transethnic Meta-Analysis of Genome-Wide Association Studies Identifies Three New Loci and Characterizes Population-Specific Differences for Coronary Artery Disease. *Circ Genom Precis Med.* 2020 Jun;13(3):e002670.
- 28. Sakaue S, Kanai M, Karjalainen J, Akiyama M, Kurki M, Matoba N, Takahashi A, Hirata M, Kubo M, Matsuda K, Murakami Y, FinnGen, Daly MJ, Kamatani Y, Okada Y. Trans-biobank analysis with 676,000 individuals elucidates the association of polygenic risk scores of complex traits with human lifespan. *Nat Med.* 2020 Apr;26(4):542-548.
- 29. **Sakaue S**, Hirata J, Kanai M, Suzuki K, Akiyama M, Too CL, Arayssi T, Hammoudeh M, Emadi SA, Masri BK, Halabi H, Badsha H, Uthman IW, Saxena R, Padyukov L, Hirata M, Matsuda K, Murakami Y, Kamatani Y, Okada Y. Dimensionality reduction enlightens the finest-scale genetic, evolutional, and phenotypic structure within the Japanese population. *Nat Commun.* 2020 Mar 26;11(1):1569.
- 30. Yamamoto K, **Sakaue S**, Matsuda K, Murakami Y, Kamatani Y, Ozono K, Momozawa Y, Okada Y. Genetic and phenotypic landscape of the mitochondrial genome in the Japanese population. *Commun Biol.* 2020 Mar 5;3(1):104.
- 31. Yasumizu Y*, **Sakaue S***, Konuma T, Suzuki K, Matsuda K, Murakami Y, Kubo M, Palamara PF, Kamatani Y, Okada Y. Genome-wide natural selection signatures are linked to genetic risk of modern phenotypes in the Japanese population. *Mol Biol Evol.* 2020 Jan 20. pii: msaa005.
- 32. 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. *Nat Commun.* 2019 Sep 27;10(1):4393.
- 33. **Sakaue S**, Akiyama M, Hirata M, Matsuda K, Murakami Y, Kubo M, Kamatani Y, Okada Y. Functional variants in ADH1B and ALDH2 are non-additively associated with all-cause mortality in Japanese population. *Eur J Hum Genet*. 2020 Mar;28(3):378-382.
- 34. Ogawa K, Okuno T, Hosomichi K, Hosokawa A, Hirata J, Suzuki K, **Sakaue S**, Kinoshita M, Asano Y, Miyamoto K, Inoue I, Kusunoki S, Okada Y, Mochizuki H. Next-generation sequencing identifies contribution of both class I and II HLA genes on susceptibility of multiple sclerosis in Japanese. *J Neuroinflammation*. 2019 Aug 5;16(1):162.
- 35. Shungin D, Haworth S, Divaris K, Agler CS, Kamatani Y, Keun Lee M, Grinde K, Hindy G, Alaraudanjoki V, Pesonen P, Teumer A, Holtfreter B, **Sakaue S**, Hirata J, Yu YH, Ridker PM, Giulianini F, Chasman DI, Magnusson PKE, Sudo T, Okada Y, Völker U, Kocher T, Anttonen V, Laitala ML, Orho-Melander M, Sofer T, Shaffer JR, Vieira A, Marazita ML, Kubo M, Furuichi Y, North KE, Offenbacher S, Ingelsson E, Franks PW, Timpson NJ, Johansson I. Genome-wide analysis of dental caries and periodontitis combining clinical and self-reported data. *Nat Commun.* 2019 Jun 24;10(1):2773.
- 36. **Sakaue S** and Okada Y. GREP: genome for REPositioning drugs. *Bioinformatics*. 2019 Oct 1;35(19):3821-3823.
- 37. Hirata J, Hosomichi K, **Sakaue S**, Kanai M, Nakaoka H, Ishigaki K, Suzuki K, Akiyama M, Kishikawa T, Ogawa K, Masuda T, Yamamoto K, Hirata M, Matsuda K, Momozawa Y, Inoue I, Kubo M, Kamatani Y, Okada Y. Genetic and phenotypic landscape of the major histocompatibility complex region in the Japanese population. *Nat Genet*. 2019 Mar;51(3):470-480.
- 38. **Sakaue S**, Hirata J, Maeda Y, Kawakami E, Nii T, Kishikawa T, Ishigaki K, Terao C, Suzuki K, Akiyama M, Suita N, Masuda T, Ogawa K, Yamamoto K, Saeki Y, Matsushita M, Yoshimura M,

- Matsuoka H, Ikari K, Taniguchi A, Yamanaka H, Kawaji H, Lassmann T, Itoh M, Yoshitomi H, Ito H, Ohmura K, R Forrest AR, Hayashizaki Y, Carninci P, Kumanogoh A, Kamatani Y, de Hoon M, Yamamoto K, Okada Y. Integration of genetics and miRNA-target gene network identified disease biology implicated in tissue specificity. *Nucleic Acids Res.* 2018 Dec 14;46(22):11898-11909.
- 39. 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 wholegenome sequencing reveals recent selection signatures linked to evolution and disease risk of Japanese. *Nat Commun.* 2018 Apr 24;9(1):1631.
- 40. Malik R, Chauhan G, Traylor M, Sargurupremraj M, Okada Y, Mishra A, Rutten-Jacobs L, Giese AK, van der Laan SW, Gretarsdottir S, Anderson CD, Chong M, Adams HHH, Ago T, Almgren P, Amouyel P, Ay H, Bartz TM, Benavente OR, Bevan S, Boncoraglio GB, Brown RD Jr, Butterworth AS, Carrera C, Carty CL, Chasman DI, Chen WM, Cole JW, Correa A, Cotlarciuc I, Cruchaga C, Danesh J, de Bakker PIW, DeStefano AL, den Hoed M, Duan Q, Engelter ST, Falcone GJ, Gottesman RF, Grewal RP, Gudnason V, Gustafsson S, Haessler J, Harris TB, Hassan A, Havulinna AS, Heckbert SR, Holliday EG, Howard G, Hsu FC, Hyacinth HI, Ikram MA, Ingelsson E, Irvin MR, Jian X, Jiménez-Conde J, Johnson JA, Jukema JW, Kanai M, Keene KL, Kissela BM, Kleindorfer DO, Kooperberg C, Kubo M, Lange LA, Langefeld CD, Langenberg C, Launer LJ, Lee JM, Lemmens R, Leys D, Lewis CM, Lin WY, Lindgren AG, Lorentzen E, Magnusson PK, Maguire J, Manichaikul A, McArdle PF, Meschia JF, Mitchell BD, Mosley TH, Nalls MA, Ninomiya T, O'Donnell MJ, Psaty BM, Pulit SL, Rannikmäe K, Reiner AP, Rexrode KM, Rice K, Rich SS, Ridker PM, Rost NS, Rothwell PM, Rotter JI, Rundek T, Sacco RL, Sakaue S, Sale MM, Salomaa V. Sapkota BR, Schmidt R, Schmidt CO, Schminke U, Sharma P, Slowik A, Sudlow CLM, Tanislav C, Tatlisumak T, Taylor KD, Thijs VNS, Thorleifsson G, Thorsteinsdottir U, Tiedt S, Trompet S, Tzourio C, van Duijn CM, Walters M, Wareham NJ, Wassertheil-Smoller S, Wilson JG, Wiggins KL, Yang Q, Yusuf S; AFGen Consortium; Cohorts for Heart and Aging Research in Genomic Epidemiology (CHARGE) Consortium; International Genomics of Blood Pressure (iGEN-BP) Consortium; INVENT Consortium; STARNET, Bis JC, Pastinen T, Ruusalepp A, Schadt EE, Koplev S, Björkegren JLM, Codoni V, Civelek M, Smith NL, Trégouët DA, Christophersen IE, Roselli C, Lubitz SA, Ellinor PT, Tai ES, Kooner JS, Kato N, He J, van der Harst P, Elliott P, Chambers JC, Takeuchi F, Johnson AD; BioBank Japan Cooperative Hospital Group; COMPASS Consortium; EPIC-CVD Consortium; EPIC-InterAct Consortium; International Stroke Genetics Consortium (ISGC); METASTROKE Consortium; Neurology Working Group of the CHARGE Consortium; NINDS Stroke Genetics Network (SiGN); UK Young Lacunar DNA Study; MEGASTROKE Consortium, Sanghera DK, Melander O, Jern C, Strbian D, Fernandez-Cadenas I, Longstreth WT Jr, Rolfs A, Hata J, Woo D, Rosand J, Pare G, Hopewell JC, Saleheen D, Stefansson K, Worrall BB, Kittner SJ, Seshadri S, Fornage M, Markus HS, Howson JMM, Kamatani Y, Debette S, Dichgans M. Multiancestry genome-wide association study of 520,000 subjects identifies 32 loci associated with stroke and stroke subtypes. Nat Genet. 2018 Apr;50(4):524-537.
- 41. **Sakaue S** and Okada Y. Human genetics contributes to the understanding of disease pathophysiology and drug discovery. *J Orthop Sci.* 2017 Nov;22(6):977-981.
- 42. Okada Y, Kishikawa T, **Sakaue S**, Hirata J. Future Directions of Genomics Research in Rheumatic Diseases. *Rheum Dis Clin North Am.* 2017 Aug;43(3):481-487.
- 43. **Sakaue S** and Hagino N. IMAGES IN CLINICAL MEDICINE. Takayasu's Arteritis. *N Engl J Med.* 2016 Aug 18;375(7):675.
- 44. **Sakaue S**, Sumitomo S, Fujio K, Yamamoto K. Unilateral proptosis in a woman with asthma. *BMJ Case Rep.* 2015 Feb 12;2015.
- 45. **Sakaue S**, Sumitomo S, Kubo K, Fujio K, Yamamoto K. Tocilizumab-induced leucocytoclastic vasculitis in a patient with rheumatoid arthritis. *Rheumatology*. 2014 Aug;53(8):1529-30.

^{*} Equal contribution

Thesis:

Sakaue S, Yamamoto K, Okada Y, Fujio K. Elucidation of novel disease pathogenesis through integrative analysis of genome-wide association studies and tissue-specific expression profile of miRNAs [dissertation]. The University of Tokyo; 2020.

Invited Presentations:

| 2019/2 | Integration of GWAS summary statistics and miRNA-target gene network with tissue- |
|---------|--|
| | specific miRNA expression profile identified novel pathogenesis of complex human |
| 0000440 | traits implicated in tissue specificity. Osaka University, Osaka, Japan |
| 2020/10 | East Asian genomic data elucidates how the population-specific natural selection |
| | shaped phenotypic variation and survival outcome of modern humans. American |
| 2022/3 | Society of Human Genetics 2020 (Invited Session, Virtual conference) Cross-population GWAS for M2M2M. International Common Disease Alliance |
| 2022/0 | Scientific Plenary (Virtual conference) |
| 2022/3 | Decoding the diversity of killer immunoglobulin-like receptors by deep sequencing and |
| | a high-resolution imputation method. National Institute of Genetics, Japan |
| 2022/5 | Creating a statistical method to utilize genetics knowledge for improving healthcare. Osaka University, Osaka, Japan |
| 2022/7 | Creating a statistical method to utilize genetics knowledge for improving healthcare. |
| | Keio University, Tokyo, Japan |
| 2022/8 | A cell-type specific enhancer-gene map from single cell multimodal RNA/ATAC-seq |
| | can pinpoint causal variation and genes in complex traits. Impact of Genomic Variation |
| | on Function (IGVF) Scientific Seminar Series (virtual), US |
| 2022/8 | Creating a statistical method to utilize genetics knowledge for improving healthcare |
| 0000/0 | The University of Tokyo, Tokyo, Japan |
| 2022/9 | Defining causal variants and genes for eQTL and GWAS by non-parametric analysis of |
| | single-cell multimodal data. Cell Circuits and Epigenetics Meeting, Broad Institute, Boston, US |
| 2022/10 | Accurate enhancer gene maps from single cell multimodal data enable fine-mapping of |
| 2022/10 | human disease loci. GRIP Meeting, Harvard Medical School, Boston, US |
| 2022/10 | Accurate enhancer gene maps from single cell multimodal data enable fine-mapping of |
| | human disease loci. Impact of Genomic Variation on Function (IGVF) Phenotypic |
| | working group, US |
| 2022/11 | HLA imputation in biobank-scale data. American Society of Human Genetics Genetics |
| | and Genomics Digital Forum (Virtual) |
| 2022/12 | A scalable approach to use single-cell multimodal data to define disease causal |
| 0000440 | variants and links them to target genes. Rush Alzheimer's Disease Center (Virtual |
| 2022/12 | A scalable approach to use single-cell multimodal data to define disease causal |
| | variants and links them to target genes. Icahn School of Medicine at Mount Sinai |
| 2023/01 | (Virtual) |
| 2023/01 | A scalable approach to use single-cell multimodal data to define disease causal variants and links them to target genes. Channing Division of Network Medicine |
| | Genomics Seminar, Boston, US. |
| 2023/02 | Single-cell mutimodal ATAC/RNA-seq provides accurate enhancer-links for fine- |
| | mapping disease causal variants and genes. Medical and Population Genetics |
| | Seminar Series, Boston, US |
| | |

| 2023/02 | A scalable approach to use single-cell multimodal data to define disease causal variants and links them to target genes. Program in Quantitative Genomics Working |
|---------|---|
| | Group Series, Boston, US |
| 2023/02 | Functional fine-mapping of GWAS using single-cell multimodal ATAC/RNA-seq data points to disease causal alleles and genes. RA Genetics Consortium meeting (Virtual) |
| 2023/03 | A statistical genetics guide to identifying HLA alleles driving complex diseases. Medical and Population Genetics Primer, Boston, US. |
| 2023/10 | SCENT defines enhancer-gene maps from single-cell multimodal data that link causal alleles to disease genes. Cell Circuits and Epigenetics Meeting, Broad Institute, Boston, US |
| 2023/11 | HLA Imputation workshop. American Society of Human Genetics, Los Angeles, CA. |
| 2023/11 | Using single cell data to define genetic mechanisms of autoimmunity. International Union of Immunological Societies 2023 meeting, Cape Town, South Africa |
| 2024/05 | Integrating genetics and single-cell genomics to define causal mechanisms of autoimmune diseases. Department of Pharmaceutical Sciences Seminar Series, University of Tokyo, Tokyo, Japan |
| 2024/10 | Tissue-specific enhancer-gene maps from multimodal single-cell data identify causal disease alleles. Variant Effects Seminar Series (Virtual) |
| 2024/11 | Distinct eQTL causal variants between cellular and nucleus RNA-seq. Program in Quantitative Genomics Working Group Series, Boston, US |
| 2025/02 | Missing regulatory function in disease alleles: Early and late RNA eQTLs are driven by different genetic mechanisms. Medical and Population Genetics Seminar Series, Boston, US |

Abstract Oral Presentations

| 2018/10 | Integration of GWAS summary statistics and miRNA-target gene network with tissue- specific miRNA expression profile identified novel pathogenesis of complex human traits implicated in tissue specificity. the 62th Annual Meeting of the Japan Society of Human Genetics, Kobe, Japan. |
|---------|---|
| 2018/10 | Integration of GWAS summary statistics and miRNA-target gene network with tissue- specific miRNA expression profile identified novel pathogenesis of complex human traits implicated in tissue specificity. |
| 2019/04 | American Society of Human Genetics, San Diego, CA. |
| | the 62th Annual General Assembly and Scientific Meeting of the Japan College of Rheumatology, Yokohama, Japan. |
| 2019/10 | Trans-ethnic mega-biobank polygenic risk score analysis involving 676,000 individuals identified blood pressure and obesity as causal drivers affecting human longevity. American Society of Human Genetics, Houston, Tx. |
| 2019/10 | Machine-learning based deconvolution of biobank-driven GWAS data with 170,000 individuals enlightens the finest-scale genetic, evolutional, and polygenic risk score divergence within Japanese population. American Society of Human Genetics, Houston, Tx. |
| 2022/10 | A cell-type-specific enhancer-gene map built from multimodal assay of RNA and ATAC-seq in 160,000 single cells pinpoints causal variants and genes in human diseases. American Society of Human Genetics, Los Angeles, CA. |
| | go.og, |

Funding and Grant:

Osamu Hayaishi Memorial Scholarship (2023)

Postdoctoral fellowship from Uehara Memorial Foundation (2022)

The Kanae Foundation for the Promotion of Medical Science (2021)

The Mochida Memorial Foundation for Medical and Pharmaceutical Research (2020)

The Astellas Foundation for Research on Metabolic Disorders (2020)

Current Licensure and Certification:

2010 USMLE Step 1 (Score: 99 percentile), 2CS (PASS), 2CK (99 percentile)

2011 Japanese Medical License

2019-present Japanese Board of Rheumatology

Service:

- Served as Ad Hoc Reviewer for *Nature, Nature Genetics, Cell Genomics, American Journal of Human Genetics, Nature Communications, Genome Biology, Cell reports, Arthritis & Rheumatology, Bioinformatics, Communications Biology, PLOS Genetics, Journal of Human Genetics, and others*

 Served as an Instructor at the American Society of Human Genetics annual meeting workshop in 2022 and in 2023 for lecturing about HLA imputation to the participants.