

Saori Sakaue, MD, PhD**Date Prepared:** 12/1/2023**Office Address:** Harvard New Research Building
77 Avenue Louis Pasteur, 250D, Boston, MA02115**Email:** ssakaue@broadinstitute.org**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	Genetics / Medicine (Prof. Kazuhiko Yamamoto)	Graduate School of Medicine, The University of 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-present	Postdoctoral Research Fellow	Division of Genetics (Dr. Soumya Raychaudhuri)	Brigham & Women's Hospital, Harvard Medical School and Broad Institute of MIT and Harvard

Professional Societies:

2018- American Society of Human Genetics

Honors and Prizes:

2010	Otsubo Tetsumon Fellowship Award	The University of Tokyo	Academic achievements
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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 Women 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)

1. **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. *medRxiv*, 2022. *Accepted in Nature Genetics* (2023)
2. 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.
3. 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.
4. **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.
5. 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 CI, 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.
6. 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.
 7. 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.
 8. **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).
 9. 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.
 10. 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.
 11. **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 cross-population atlas of genetic associations for 220 human phenotypes. *Nat Genet.* 2021 Oct;53(10):1415-1424
 12. 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.
 13. **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.
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15. 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.
 16. 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.
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 18. 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.
19. 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.
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 22. **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, evolutionary, and phenotypic structure within the Japanese population. *Nat Commun*. 2020 Mar 26;11(1):1569.
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26. **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.
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37. **Sakaue S**, Sumitomo S, Fujio K, Yamamoto K. Unilateral proptosis in a woman with asthma. *BMJ Case Rep.* 2015 Feb 12;2015.
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* 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:

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|---------|---|
| 2019/2 | 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. 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 Society of Human Genetics 2020 (Invited Session, Virtual conference) |
| 2022/3 | Cross-population GWAS for M2M2M. International Common Disease Alliance 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 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 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 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 (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 multimodal 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	Using single cell data to define genetic mechanisms of autoimmunity. International Union of Immunological Societies 2023 meeting, Cape Town, South Africa

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.

2019/10	American Society of Human Genetics, Houston, Tx. Machine-learning based deconvolution of biobank-driven GWAS data with 170,000 individuals enlightens the finest-scale genetic, evolutionary, and polygenic risk score divergence within Japanese population.
2022/10	American Society of Human Genetics, Houston, Tx. 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.

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Osamu Hayaishi Memorial Scholarship (2023)

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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 Step1 (Score: 99 percentile), 2CS (PASS), 2CK (99 percentile)
2011-	Japanese Medical License
2019-2024	Japanese Board of Rheumatology

Service:

Served as Ad Hoc Reviewer for *Nature*, *Nature Genetics*, *Cell Genomics*, *Nature Communications*, *Arthritis & Rheumatology*, *Communications Biology*, *Journal of Human Genetics*, and others