

Saori Sakaue

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Education

The University of Tokyo, Japan
Faculty of Medicine (MD)

April 2005– March 2011

The University of Tokyo, Japan
Graduate School of Medicine (PhD candidate)

April 2016 – March 2020

Department of Statistical Genetics, Osaka University
Co-affiliation

October 2016 – Current

Laboratory for Statistical Analysis, RIKEN
Co-affiliation

April 2017 – Current

Advisors:

- Dr. Yukinori Okada - Professor, Department of Statistical Genetics, Osaka University Graduate School of Medicine, Japan.
- Dr. Yoichiro Kamatani - Team Leader, Laboratory for Statistical Analysis, RIKEN Center for Integrative Medical Sciences, Japan.
- Dr. Kazuhiko Yamamoto - Formerly Professor, The University of Tokyo, Japan. Currently Deputy Director, RIKEN Center for Integrative Medical Sciences, Japan.

Research Experiences

- Biobank-scale genome-wide association studies and collaboration with international consortiums.
- Integration of functional genomics data with genome-wide association studies, and interpretation of novel biological insights.
- Population genetics and its intersect with human complex diseases.
- Genetics-driven drug repositioning analysis.
- Analysis for complex genomic regions such as HLA regions.
- Polygenic risk score analysis and its novel insight into healthcare.

Technical Skills

- Programming; python, R, bash scripting, perl.
- Multi-trait genome-wide association studies and post GWAS analysis.
- Genome-wide imputation and implementation of novel algorithm.
- RNAseq, scRNAseq, and miRNAseq analysis
- Genotype calling of whole genome/exome sequencing data.
- Development of user-friendly statistical genetics softwares and scripts.

Certification

- Japanese Medical Licence.
- Board-certified Rheumatologist.
- USMLE step1, step2 CK, step2 CS

Honors and Achievements

- 2020 Investigator Award of Japanese Society of Human Genetics.
- 2020 L'Oréal-UNESCO For Women in Science International Awards.
- 2020 The President's Award of The University of Tokyo.
- 2019 American Society of Human Genetics/Charles J. Epstein Trainee Award for Excellence in Human Genetics Research – **Finalist**.
- 2018 American Society of Human Genetics/Charles J. Epstein Trainee Award for Excellence in Human Genetics Research – **Semifinalist**.
- 2018 Excellent Abstract Award at the 62th Annual General Assembly and Scientific Meeting of the Japan College of Rheumatology.
- 2017 Finalist of Meeting Award at the 62th Annual Meeting of the Japan Society of Human Genetics.
- 2010 Otsubo Tetsumon Fellowship Award Recipient.

Selected Publications

- **Sakaue S***, Kanai M*, Karjalainen J, et al. 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.
- **Sakaue S**, Hirata J, Kanai M, et al. Dimensionality reduction reveals fine-scale structure in the Japanese population with consequences for polygenic risk prediction. **Nat Commun.** 2020 Mar 26;11(1):1569.
- **Sakaue S**, Hirata J, Maeda Y et al. 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.

Other Publications

- Chen MH, Raffield LM, Mousas A, **Sakaue S**, et al. Trans-ethnic and ancestry-specific blood-cell genetics in 746,667 individuals from 5 global populations. **Cell** 2020 In Press.
- Vuckovic D, Bao EL, Akbari P, ..., **Sakaue S**,. The Polygenic and Monogenic Basis of Blood Traits and Diseases. **Cell** 2020 In Press.
- Ishigaki K, Akiyama M, Kanai M, ..., **Sakaue S**, et al. 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.
- Matsunaga H, Ito K, Akiyama M, ..., **Sakaue S**, et al. 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.
- Yamamoto K, **Sakaue S**, Matsuda K, et al. Genetic and phenotypic landscape of the mitochondrial genome in the Japanese population. **Commun Biol.** 2020 Mar 5;3(1):104.
- Yasumizu Y*, **Sakaue S***, Konuma T, et al. Genome-wide natural selection signatures are linked to genetic risk of modern phenotypes in the Japanese population. **Mol Biol Evol.** 2020 May 1;37(5):1306-1316.
- Akiyama M, Ishigaki K, **Sakaue S** et al. Characterizing rare and low-frequency height-associated variants in the Japanese population. **Nat Commun.** 2019 Sep 27;10(1):4393.
- **Sakaue S**, Akiyama M, Hirata M et al. Functional variants in ADH1B and ALDH2 are non-additively associated with all-cause mortality in Japanese population. **Eur J Hum Genet.** 2019 doi: 10.1038/s41431-019-0518-y.
- Ogawa K, Okuno T, Hosomichi K,..., **Sakaue S**, et al. 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.
- Shungin D, Haworth S, Divaris K,..., **Sakaue S**, et al. Genome-wide analysis of dental caries and periodontitis combining clinical and self-reported data. **Nat Commun.** 2019 Jun 24;10(1):2773.
- **Sakaue S** and Okada Y. GREP: genome for REPositioning drugs. **Bioinformatics.** 2019 Oct 1;35(19):3821-3823.
- Hirata J, Hosomichi K, **Sakaue S** et al. Genetic and phenotypic landscape of the major histocompatibility complex region in the Japanese population. **Nat Genet.** 2019 Mar;51(3):470-480.
- Okada Y, Momozawa Y, **Sakaue S** et al. Deep whole-genome sequencing reveals recent selection signatures linked to evolution and disease risk of Japanese. **Nat Commun.** 2018 Apr 24;9(1):1631.
- Malik R, Chauhan G, Traylor M,..., **Sakaue S**, et al. Multi-ancestry 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.
- **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.
- 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.
- **Sakaue S** and Hagino N. IMAGES IN CLINICAL MEDICINE. Takayasu's Arteritis. **N Engl J Med.** 2016 Aug 18;375(7):675.
- **Sakaue S**, Sumitomo S, Fujio K, Yamamoto K. Unilateral proptosis in a woman with asthma. **BMJ Case Rep.** 2015 Feb 12;2015.
- **Sakaue S**, Sumitomo S, Kubo K et al. Tocilizumab-induced leucocytoclastic vasculitis in a patient with rheumatoid arthritis. **Rheumatology.** 2014 Aug;53(8):1529-30.

Presentations

- 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. PLATFORM talk (Oral Presentation) at Houston, Oct 2019.

- 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.

American Society of Human Genetics. PLATFORM talk (Oral Presentation) at Houston as a surrogate presenter, Oct 2019.

- 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.

American Society of Human Genetics. PLATFORM talk (Oral Presentation) at San Diego, Oct 2018.

Collaborative Research Experiences

- 2017- **GIANT consortium** (https://portals.broadinstitute.org/collaboration/giant/index.php/GIANT_consortium)
- 2017- **GLGC consortium** (<http://lipidgenetics.org>)
- 2017- **Blood Cell consortium** (<http://www.mhi-humangenetics.org/en/resources>)
- 2016 **MEGASTROKE consortium** (<http://www.megastroke.org>)