

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.
- Muti-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

- 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.

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 In Press.
- **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 In Press.
- Yamamoto K, **Sakaue S**, Matsuda K, et al. Genetic and phenotypic landscape of the mitochondrial genome in the Japanese population. **Commun Biol.** 2020 In Press.
- 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 Jan 20.
- 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.
- **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.
- 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. 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.
- **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>)

