**GWAS summary statistics of recurrent pregnancy loss in Japanese**

General information:

* **Sample size**: 1,728 cases vs. 24,315 controls
* **Genotyping array**: Infinium Asian Screening Array
* **Sample QC**: We excluded individuals with low genotyping call rates (call rate < 98%). We included samples of the estimated Japanese ancestry using PCA.
* **Variant QC**: We excluded variants with (1) genotyping call rate < 99%, (2) minor allele count < 5, (3) P-value for Hardy–Weinberg equilibrium < 1.0 × 10−10, and (4) > 5% allele frequency difference compared with the imputation reference panel or the allele frequency panel of Tohoku Medical Megabank Project.
* **Phasing and imputation**: SHAPEIT4 and Minimac4
* **Imputation reference**: in-house Japanese-specific reference panel composed of *n* = 4,561 whole-genome sequence (WGS) data
* **Post-imputation QC**: We excluded imputed variants with *Rsq* < 0.7 and minor allele frequency < 0.5%.
* **Association test**: SAIGE software was used with top five principal components as covariates.

Uploaded file

| **File name** | **Descriptions** |
| --- | --- |
| GWASsummary\_RPL\_Japanese\_SoneharaNatCommun2024.txt.gz | Results for 8,717,430 autosomal and X chromosome variants |

Columns

| **#** | **column name** | **Descriptions** |
| --- | --- | --- |
| 1 | CHR | chromosome |
| 2 | POS | position (hg19) |
| 3 | MarkerID | marker name (CHR:POS:REF:ALT) |
| 4 | Allele1 | REF allele |
| 5 | Allele2 | ALT allele (This allele is the effect allele.) |
| 6 | AC\_Allele2 | allele count of Allele2 (ALT) |
| 7 | AF\_Allele2 | allele frequency of Allele2 (ALT) |
| 8 | imputationInfo | RSQ value in imputation |
| 9 | BETA | effect size of Allele2 |
| 10 | SE | standard error of BETA |
| 11 | Tstat | score statistic of Allele2 |
| 12 | var | estimated variance of score statistic |
| 13 | p.value | *P* value with SPA (suddle point approximation) applied |
| 14 | p.value.NA | *P* value when SPA is not applied |
| 15 | Is.SPA | whether SPA is converged or not |
| 16 | AF\_case | allele frequency of Allele2 in cases |
| 17 | AF\_ctrl | allele frequency of Allele2 in controls |
| 18 | N\_case | sample size in cases |
| 19 | N\_ctrl | sample size in controls |
| 20 | N\_case\_hom | sample size with Allele 2 homozygous genotypes in cases |
| 21 | N\_case\_het | sample size with heterozygous genotypes in cases |
| 22 | N\_ctrl\_hom | sample size with Allele2 homozygous genotypes in controls |
| 23 | N\_ctrl\_het | sample size with heterozygous genotypes in controls |

Reference

If you use the summary statistics, please cite the following paper;
Sonehara K and Yano Y *et al*. Common and rare genetic variants predisposing females to unexplained recurrent pregnancy loss. *Nat Commun* 2024.