**GWAS summary statistics of exome data for paroxysmal atrial fibrillation in Japanese**

General information:

* **Sample size**: 2,348 samples
* **Exome Sequencing**: Exome capture was performed with a SureSelect Human All Exon V6 Kit (Agilent) after library production using the SureSelectXT Kit (Agilent).
* **Sample QC**: Samples with call rates less than 97% and where the imputed genetic sex did not match the reported sex were excluded. Additionally, pairs of closely related individuals were calculated using Hail, and samples that were more than second-order related were filtered so that the kinship coefficient met the recommended value of 0.088. Outliers identified based on the insertion/deletion allele ratio, quantity, and heterozygosity/homozygosity call ratio were excluded.
* **Variant QC**: We excluded variants with (1) genotype quality >= 20; (2) depth >=10; (3) allele balance greater than 0.1 for homozygous reference calls, less than 0.9 for homozygous alternative calls, and either below 0.25 or above 0.75 for heterozygous calls; (4) variant call rate >= 0.97; and (5) Hardy‒Weinberg equilibrium *P*-values >1 × 10−8.
* **Association test**: We conducted gene-based association tests using the burden test, SKAT, and SKAT-O as implemented in the R package SKAT.

Uploaded file

| **File name** | **Descriptions** |
| --- | --- |
| GWASsummary\_PAF\_Japanese\_Exome.tsv | Results of the burden test, SKAT, and SKATO for a Japanese group with PAF |
| Genome-wide\_significant\_PAF.tsv | Details of the variants that showed significant differences in the results of the associated analysis. (p < 0.05/16379) |
| Suggestive\_association\_PAF.tsv | Details of the variants that showed suggestive association in the results of the associated analysis. (p < 5e-5) |

Columns

|  |  |  |
| --- | --- | --- |
| **#** | **column name** | **Description** |
| 1 | Chr | Chromosome number |
| 2 | Pos | Position of the variant on the chromosome (GRCh38) |
| 3 | Rs | dbSNP reference SNP ID |
| 4 | Ref | Reference allele |
| 5 | Alt | Alternative allele |
| 6 | Effect | Predicted effect of the variant |
| 7 | Case | Number of samples with paroxysmal atrial fibrillation |
| 8 | Control | Number of samples without paroxysmal atrial fibrillation. |
| 9 | Maf | Minor Allele Frequency  |
| 10 | Maf (EAS) | Minor Allele Frequency in the East Asian population |
| 11 | CADD |  Combined Annotation Dependent Depletion (CADD) score, a measure of deleteriousness. |
| 12 | SIFT | Sorting Intolerant From Tolerant (SIFT) score, predicts the impact of amino acid substitutions. |
| 13 | LRT | Likelihood Ratio Test (LRT) score, used for functional prediction. |
| 14 | Polyphen2-HDIV | PolyPhen-2 score (High-Divergence model), predicts damaging mutations. |
| 15 | Polyphen2-HVAR | PolyPhen-2 score (High-Variability model), used for clinical relevance predictions. |
| 16 | Mutation Taster | MutationTaster score, predicts variant pathogenicity. |
| 17 | OR (95%Cl) | Odds Ratio with 95% Confidence Interval. |