**Summary statistics of the genome-wide meta-analysis of neuromyelitis optica spectrum disorder in Japanese**

**General information**

|  |  |
| --- | --- |
| **Sample size** | 240 cases and 50,578 controls (163 cases and 40,908 controls in the discovery cohort, and 77 cases and 9,670 controls in the replication cohort) |
| **Genotyping array**: | Infinium Asian Screening Array |
| **Sample QC**: | We excluded samples with low genotyping call rates (call rate < 98%) or potential sex chromosome aneuploidy. We included only the individuals of the estimated East Asian ancestry using the principal component (PC) analysis, and then further restricted to those in Japanese Hondo (the main island of Japan) clusters. |
| **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 and in-house reference panel. |
| **Phasing and imputation**: | SHAPEIT4 and Minimac4 |
| **Imputation reference**: | In-house Japanese-specific reference panel composed of *n* = 11,754 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 age and the top four PCs as covariates. |
| **Meta-analysis:** | METASOFT software was used for the inverse variance fixed-effects meta-analysis. |

**Uploaded files**

|  |  |
| --- | --- |
| File name | Descriptions |
| NMOSD\_meta.txt.gz | Results for 8,894,915 autosomal and X chromosome variants |

| **#** | **column name** | **Descriptions** |
| --- | --- | --- |
| 1 | CHR | Chromosome |
| 2 | POS | Position (hg19) |
| 3 | Allele1 | REF allele |
| 4 | Allele2 | ALT allele (This allele is the effect allele.) |
| 5 | PVALUE\_FE | *P* value for the effect size |
| 6 | BETA\_FE | Effect size of Allele2 |
| 7 | STD\_FE | Standard error of the effect size |

**Columns**