

# Analysis Summary: Parkinsons Disease (Population Controls)

## Phenotype Description

Cases answered "Yes" or reported a diagnosis of Parkinson's disease to at least one of the questions below or joined 23andMe as part of Parkinson's Disease Research Initiative and provided no survey responses (that is, if they didn't provide any information to the contrary, we assumed they truly have PD):

- (PD) Background Survey ("Has a doctor ever diagnosed you with Parkinson's disease?" Answer = Yes)
- Your Medical History ("Have you ever been diagnosed by a doctor with Parkinson's disease"? Answer = Yes)
- Research Snippets ("Has a doctor ever diagnosed you with Parkinson's disease?" Answer = Yes)
- Your Health Profile ("Have you ever been diagnosed or treated for any of the following conditions?" Answer = Parkinson's disease)
- Roots into the Future: Health History ("Has a doctor ever told you that you have any of these conditions?" Answer = Parkinson's disease)
- (PD) Baseline Survey ("Has a doctor ever diagnosed you with Parkinson's disease?" Answer = Yes)

Cases were further filtered by removing respondents 1) who were discordant between the first two questions above (that is, removing those who said "No" to the PD Background Survey question but said "Yes" to the Your Medical History question); 2) whose diagnosis changed because their symptoms disappeared or the cause of the symptoms is unknown (according to the questions below);

- (PD) Background Survey ("Is your diagnosis still Parkinson's disease or has your diagnosis changed?" Answer = My diagnosis changed)
- (PD) Background Survey ("Which of the following best describes how your diagnosis changed?" Answer = I no longer have the symptoms that led to the Parkinson's diagnosis, I still have the symptoms but the cause is unknown, My diagnosis changed)

or 3) whose diagnosis changed to another diagnosis that could be confounded with PD based on the following questions:

- (PD) Background Survey ("Which of the following have you been diagnosed with? Please check all that apply." Answers = Progressive Supranuclear Palsy (PSP); Multiple system atrophy (MSA), Shy-Drager syndrome, or striatonigral degeneration; Spinal cerebellar ataxia (SCA), cerebellar degeneration, olivopontocerebellar atrophy (OPCA); Cortical basal ganglionic degeneration, Parkinsonism due to medications, drug-induced parkinsonism)
- (PD) Background Survey ("Which of the following have you been diagnosed with? Please check all that apply." Answers = Frontotemporal dementia (FTD); Picks disease)
- (PD) Background Survey ("Which of the following have you been diagnosed with? Please check all that apply." Answers = Dystonic tremor; Rubral tremor; Primary writing tremor; Cerebellar tremor; Multiple sclerosis (MS))

Because PD is rare, we did not require controls to affirmatively deny a diagnosis of PD. Instead, population controls were used with the following groups excluded:

- Anyone defined as a case by the first set of criteria described above
- Anyone who was diagnosed with "Another form of parkinsonism (other than Parkinson's disease)", "Dementia, cognitive impairment, or senility", "A tremor disorder") according to this question:
  - (PD) Background Survey ("Has a doctor ever diagnosed you with any of the following? Please check all that apply.)
- Anyone reporting a diagnosis of Alzheimer's disease by answering "Yes" to the following question:
  - Your Medical History ("Have you ever been diagnosed by a doctor with Alzheimer's disease?")
- Anyone reporting a diagnosis of memory loss by answering "Yes" to the following question:
  - Your Medical History ("Have you ever been diagnosed by a doctor with memory loss?")
- Anyone with three or more early symptoms of Parkinson's as defined by the following questions below:
  - (PD) Background Survey ("Do you have trembling or shaking of any part of your body?" Yes is + 1)
  - (PD) Background Survey ("Have you noticed or have people told you that your speech or voice has become softer than before?" Yes is + 1)
  - (PD) Background Survey ("When walking, do you swing your arms less than you used to?" Yes is + 1)
  - (PD) Background Survey ("When standing or walking do you stoop or bend forward more than you used to?" Yes is + 1)
  - (PD) Background Survey ("Do you have trouble with your balance or fall sometimes?" Yes is + 1)
  - (PD) Background Survey ("Have you noticed any of the following changes in your handwriting? Please check all that apply." Answers = Handwriting became slower, Handwriting became smaller, Handwriting became shakier (each answer is + 1))
  - (PD) Background Survey ("Have you noticed any of the following changes in the way you walk? Please check all that apply." Answers = Dragging on or both feet, Feet shuffling, Walking more slowly, Taking smaller steps than before, Steps becoming faster and faster, Feet getting stuck as if glued to the floor (each answer is + 1))

## Phenotype Statistics

The following table shows demographics of unrelated, European individuals included in the GWAS.

| Phenotype      | Group   | Total  | M      | F      | (0,30] | (30,45] | (45,60] | (60,Inf] |
|----------------|---------|--------|--------|--------|--------|---------|---------|----------|
| PD_all_pop_con | case    | 9619   | 5875   | 3744   | 33     | 330     | 1959    | 7297     |
|                | control | 324522 | 174938 | 149584 | 44241  | 93985   | 87425   | 98871    |

The following table shows the phenotypic distribution across 23andMe genotyping platforms for individuals included in the GWAS.

| Phenotype      | Group   | Total  | v1/v2 | v3     | v4    |
|----------------|---------|--------|-------|--------|-------|
| PD_all_pop_con | case    | 9619   | 2947  | 6300   | 372   |
|                | control | 324522 | 21205 | 233307 | 70010 |

## Null Model with Covariates

The following table shows results of fitting a model for the trait based on just the covariates. Principal coordinates have been standardized, so these effect sizes are in units of standard deviations.

|      | Estimate | Std. Error | z value | Pr(> z )              | LRT     | Pr(>Chi)              |
|------|----------|------------|---------|-----------------------|---------|-----------------------|
| age  | 0.0673   | 0.000731   | 92.1    | 0.0                   | 10129.9 | 0.0                   |
| sexF | -0.3477  | 0.021686   | -16.0   | $7.4 \times 10^{-58}$ | 261.9   | $6.6 \times 10^{-59}$ |
| pc.0 | -0.0111  | 0.009583   | -1.2    | 0.25                  | 1.3     | 0.25                  |
| pc.1 | -0.1161  | 0.012860   | -9.0    | $1.8 \times 10^{-19}$ | 88.4    | $5.5 \times 10^{-21}$ |
| pc.2 | -0.0647  | 0.011090   | -5.8    | $5.5 \times 10^{-9}$  | 35.2    | $3.0 \times 10^{-9}$  |
| pc.3 | 0.0442   | 0.010795   | 4.1     | $4.3 \times 10^{-5}$  | 16.7    | $4.4 \times 10^{-5}$  |
| pc.4 | 0.0609   | 0.012376   | 4.9     | $8.6 \times 10^{-7}$  | 26.2    | $3.1 \times 10^{-7}$  |

## SNP-level QC information

The following table shows results for QC filters on the genotyped data:

|                            | failed | passed  |
|----------------------------|--------|---------|
| no filters                 | 0      | 1030430 |
| not V1/V2-only, chrM, chrY | 33033  | 997397  |
| parent-offspring test      | 2129   | 995294  |
| MAF > 0%                   | 3203   | 992310  |
| HWE > 1e-20                | 48225  | 945039  |
| gt.rate > 90%              | 30775  | 925423  |
| batch effects              | 71627  | 889734  |

The following table shows results for QC filters on the imputed dosage data:

|                    | failed  | passed   |
|--------------------|---------|----------|
| no filters         | 0       | 13733809 |
| MAF > 0%           | 0       | 13733809 |
| imputation quality | 0       | 13733809 |
| batch effects      | 1123116 | 12610693 |

The following table shows results for QC filters on the merged association test results:

|                    | passed   | total    |
|--------------------|----------|----------|
| imputed only       | 11813480 | 11813480 |
| both passed        | 797198   | 12610678 |
| genotyped only     | 92536    | 12703214 |
| no test result     | -1446    | 12701768 |
| failed to converge | -263549  | 12438219 |

## Genetic Association Tests

We performed logistic regression assuming an additive model for allelic effects, using the model:

$$PD\_all\_pop\_con \sim age + sex + pc.0 + pc.1 + pc.2 + pc.3 + pc.4 + genotype$$

This genome-wide association analysis includes data from 9619 cases and 324522 controls of European ancestry, filtered to remove close relatives.

The results in this report have been adjusted for a genomic control inflation factor  $\lambda=1.131$ . The equivalent inflation factor for 1000 cases and 1000 controls  $\lambda_{1000}=1.007$ , and for 10000,  $\lambda_{10000}=1.070$ .

## Manhattan Plot



# Quality Statistics for Index SNPs

| assay.name  | is.v2 | is.v3 | is.v4 | gt.rate | hw.p.value            | p.date                | freq.b | avg.rsqr | min.rsqr | p.batch              | dose.b | qc.mask |
|-------------|-------|-------|-------|---------|-----------------------|-----------------------|--------|----------|----------|----------------------|--------|---------|
| rs356182    | FALSE | FALSE | FALSE |         |                       |                       |        | 0.8767   | 0.8611   | 0.20                 | 0.3572 | v2v3v4  |
| rs28903073  | TRUE  | TRUE  | TRUE  | 0.9998  | 5.4×10 <sup>-5</sup>  | 2.3×10 <sup>-10</sup> | 0.9956 | 0.9748   | 0.9575   | 0.00053              | 0.9956 | v2v3v4  |
| rs365825    | FALSE | FALSE | FALSE |         |                       |                       |        | 0.9407   | 0.8998   | 1.5×10 <sup>-6</sup> | 0.2265 | v2v3v4  |
| rs144847051 | FALSE | FALSE | FALSE |         |                       |                       |        | 0.7436   | 0.6980   | 0.0047               | 0.9926 | v2v3v4  |
| rs17425622  | FALSE | FALSE | FALSE |         |                       |                       |        | 0.8535   | 0.7666   | 0.22                 | 0.8528 | v2v3v4  |
| rs2230288   | FALSE | TRUE  | TRUE  | 0.9823  | 0.012                 | 0.0                   | 0.0036 | 0.9545   | 0.8988   | 0.027                | 0.0128 | v2v3v4  |
| rs13016703  | FALSE | FALSE | TRUE  | 0.9999  | 0.28                  | 0.19                  | 0.8747 | 0.9958   | 0.9751   | 0.037                | 0.8733 | v2v3v4  |
| rs35541465  | FALSE | FALSE | FALSE |         |                       |                       |        | 0.9445   | 0.8958   | 7.1×10 <sup>-6</sup> | 0.7323 | v2v3v4  |
| rs11158026  | FALSE | FALSE | TRUE  | 1.0000  | 0.92                  | 0.64                  | 0.3317 | 0.9714   | 0.9591   | 0.20                 | 0.3303 | v2v3v4  |
| rs9858038   | FALSE | FALSE | FALSE |         |                       |                       |        | 0.9839   | 0.9695   | 0.87                 | 0.8029 | v2v3v4  |
| rs10256359  | FALSE | FALSE | FALSE |         |                       |                       |        | 0.9648   | 0.8961   | 0.35                 | 0.3695 | v2v3v4  |
| rs823118    | FALSE | FALSE | FALSE |         |                       |                       |        | 0.9979   | 0.9954   | 0.61                 | 0.5585 | v2v3v4  |
| rs188789342 | FALSE | FALSE | FALSE |         |                       |                       |        | 0.8154   | 0.7608   | 0.17                 | 0.0176 | v2v3v4  |
| rs6812193   | TRUE  | TRUE  | TRUE  | 1.0000  | 0.018                 | 0.014                 | 0.3677 | 1.0000   | 0.9999   | 0.21                 | 0.3677 | v2v3v4  |
| rs4266290   | FALSE | FALSE | FALSE |         |                       |                       |        | 0.9966   | 0.9924   | 0.69                 | 0.7474 | v2v3v4  |
| rs4130047   | TRUE  | TRUE  | TRUE  | 1.0000  | 0.54                  | 0.15                  | 0.6854 | 0.9999   | 0.9995   | 0.30                 | 0.6854 | v2v3v4  |
| rs4713118   | FALSE | TRUE  | FALSE | 0.9995  | 4.3×10 <sup>-29</sup> | 0.85                  | 0.2517 | 0.9960   | 0.9857   | 0.00011              | 0.2527 | v2v3v4  |
| rs11060180  | TRUE  | TRUE  | TRUE  | 0.9997  | 0.47                  | 0.38                  | 0.4498 | 0.9990   | 0.9971   | 0.69                 | 0.4489 | v2v3v4  |
| rs687432    | FALSE | FALSE | FALSE |         |                       |                       |        | 0.9756   | 0.9138   | 0.056                | 0.7042 | v2v3v4  |
| rs148294058 | FALSE | FALSE | FALSE |         |                       |                       |        | 0.5194   | 0.4686   | 0.094                | 0.9830 | v2v3v4  |
| rs2209440   | TRUE  | TRUE  | TRUE  | 0.9990  | 0.29                  | 0.22                  | 0.5236 | 0.9979   | 0.9927   | 0.33                 | 0.5244 | v2v3v4  |
| rs11343     | TRUE  | TRUE  | TRUE  | 0.9999  | 0.29                  | 2.4×10 <sup>-5</sup>  | 0.4432 | 0.9982   | 0.9938   | 0.00024              | 0.4435 | v2v3v4  |
| rs162227    | FALSE | FALSE | FALSE |         |                       |                       |        | 0.9206   | 0.8771   | 4.2×10 <sup>-5</sup> | 0.8965 | v2v3v4  |
| rs144074972 | FALSE | FALSE | FALSE |         |                       |                       |        | 0.9007   | 0.7877   | 0.81                 | 0.9993 | v2v3v4  |
| rs601999    | FALSE | FALSE | FALSE |         |                       |                       |        | 0.9862   | 0.9781   | 0.11                 | 0.3272 | v2v3v4  |

# SNP Statistics in the GWAS Sample

| assay.name  | AA.0   | AB.0   | BB.0   | im.num.0 | dose.b.0 | AA.1 | AB.1 | BB.1 | im.num.1 | dose.b.1 |
|-------------|--------|--------|--------|----------|----------|------|------|------|----------|----------|
| rs356182    |        |        |        | 324522   | 0.3570   |      |      |      | 9619     | 0.4127   |
| rs28903073  | 7      | 2616   | 321831 | 324522   | 0.9960   | 4    | 237  | 9374 | 9619     | 0.9872   |
| rs365825    |        |        |        | 324522   | 0.2271   |      |      |      | 9619     | 0.1925   |
| rs144847051 |        |        |        | 324522   | 0.9928   |      |      |      | 9619     | 0.9862   |
| rs17425622  |        |        |        | 324522   | 0.8521   |      |      |      | 9619     | 0.8678   |
| rs2230288   | 295942 | 2056   | 0      | 324522   | 0.0128   | 6407 | 22   | 0    | 9619     | 0.0210   |
| rs13016703  | 1087   | 15357  | 53557  | 324522   | 0.8743   | 9    | 93   | 270  | 9619     | 0.8541   |
| rs35541465  |        |        |        | 324522   | 0.7355   |      |      |      | 9619     | 0.7084   |
| rs11158026  | 31249  | 31042  | 7718   | 324522   | 0.3313   | 166  | 175  | 31   | 9619     | 0.3036   |
| rs9858038   |        |        |        | 324522   | 0.8032   |      |      |      | 9619     | 0.8271   |
| rs10256359  |        |        |        | 324522   | 0.3700   |      |      |      | 9619     | 0.3422   |
| rs823118    |        |        |        | 324522   | 0.5569   |      |      |      | 9619     | 0.5836   |
| rs188789342 |        |        |        | 324522   | 0.0176   |      |      |      | 9619     | 0.0244   |
| rs6812193   | 129661 | 150504 | 44346  | 324522   | 0.3686   | 4130 | 4351 | 1136 | 9619     | 0.3445   |
| rs4266290   |        |        |        | 324522   | 0.7471   |      |      |      | 9619     | 0.7660   |
| rs4130047   | 31948  | 140038 | 152532 | 324522   | 0.6858   | 1103 | 4276 | 4240 | 9619     | 0.6630   |
| rs4713118   | 131746 | 85842  | 15592  | 324522   | 0.2528   | 3349 | 2441 | 505  | 9619     | 0.2738   |
| rs11060180  | 97845  | 160924 | 65659  | 324522   | 0.4504   | 3124 | 4797 | 1695 | 9619     | 0.4258   |
| rs687432    |        |        |        | 324522   | 0.7042   |      |      |      | 9619     | 0.7239   |
| rs148294058 |        |        |        | 324522   | 0.9833   |      |      |      | 9619     | 0.9785   |
| rs2209440   | 73044  | 162130 | 88997  | 324522   | 0.5252   | 2397 | 4767 | 2450 | 9619     | 0.5030   |
| rs11343     | 101021 | 159837 | 63634  | 324522   | 0.4426   | 2791 | 4703 | 2125 | 9619     | 0.4655   |
| rs162227    |        |        |        | 324522   | 0.8978   |      |      |      | 9619     | 0.8855   |
| rs144074972 |        |        |        | 324522   | 0.9994   |      |      |      | 9619     | 1.0000   |
| rs601999    |        |        |        | 324522   | 0.3280   |      |      |      | 9619     | 0.3466   |

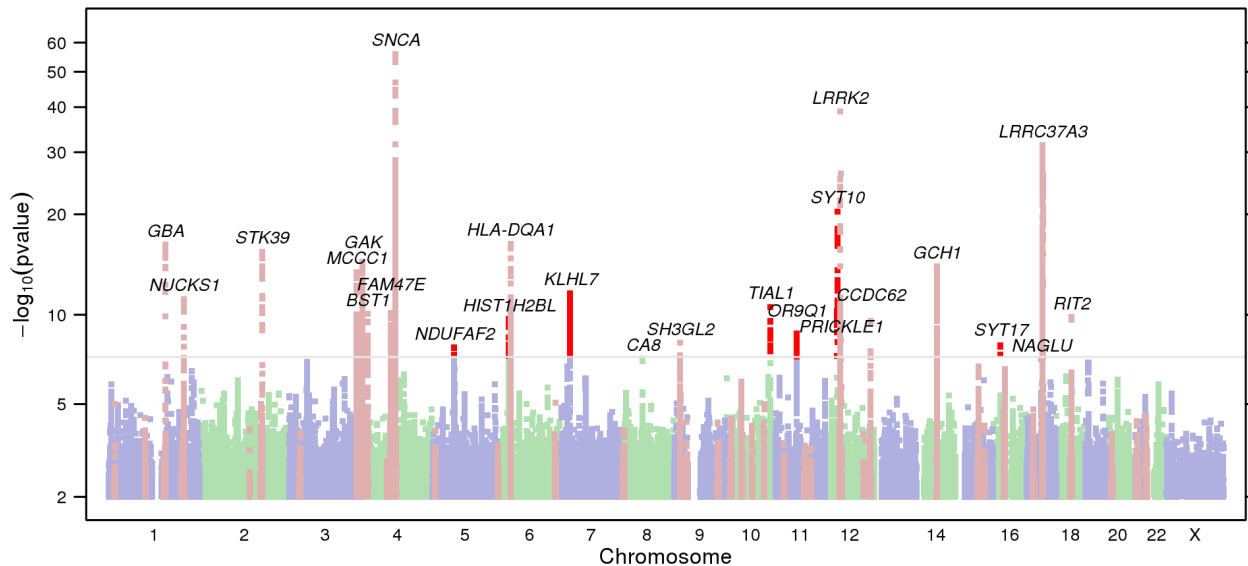
# Annotations from NHGRI GWAS Catalog

The following table shows, for each index SNP, all entries in the NHGRI GWAS Catalog that are within 500kb and in at least moderate linkage disequilibrium ( $r^2 > 0.5$ ).

| region   | position  | our.name   | our.pval              | dist    | rsqr  | assay.name | pvalue                 | pubmed.id | trait                                     | genes               |
|----------|-----------|------------|-----------------------|---------|-------|------------|------------------------|-----------|---|---------------------|
| 4q22.1   | 90626111  | rs356182   | 1.2×10 <sup>-56</sup> | 11490   | 0.758 | rs356219   | 6.0×10 <sup>-65</sup>  | 22438815  | Parkinson's disease                       | SNCA                |
| 4q22.1   | 90626111  | rs356182   | 1.2×10 <sup>-56</sup> | 11490   | 0.758 | rs356219   | 2.0×10 <sup>-47</sup>  | 21292315  | Parkinson's disease                       | SNCA                |
| 4q22.1   | 90626111  | rs356182   | 1.2×10 <sup>-56</sup> | 15229   | 0.520 | rs356220   | 3.0×10 <sup>-11</sup>  | 24511991  | Parkinson's disease                       | SNCA                |
| 4q22.1   | 90626111  | rs356182   | 1.2×10 <sup>-56</sup> | 15229   | 0.520 | rs356220   | 8.0×10 <sup>-35</sup>  | 22451204  | Parkinson's disease                       | SNCA                |
| 4q22.1   | 90626111  | rs356182   | 1.2×10 <sup>-56</sup> | 15229   | 0.520 | rs356220   | 2.0×10 <sup>-19</sup>  | 21738487  | Parkinson's disease                       | SNCA                |
| 4q22.1   | 90626111  | rs356182   | 1.2×10 <sup>-56</sup> | 15229   | 0.520 | rs356220   | 3.0×10 <sup>-8</sup>   | 21084426  | Parkinson's disease                       | SNCA                |
| 4q22.1   | 90626111  | rs356182   | 1.2×10 <sup>-56</sup> | 15229   | 0.520 | rs356220   | 9.0×10 <sup>-16</sup>  | 21044948  | Parkinson's disease                       | SNCA                |
| 4q22.1   | 90626111  | rs356182   | 1.2×10 <sup>-56</sup> | 15229   | 0.520 | rs356220   | 3.0×10 <sup>-11</sup>  | 20711177  | Parkinson's disease                       | SNCA                |
| 17q21.31 | 43705601  | rs365825   | 4.2×10 <sup>-32</sup> | -192160 | 0.707 | rs11012    | 6.0×10 <sup>-8</sup>   | 20070850  | Parkinson's disease                       | PLEKHM1, MAPT, IMP5 |
| 17q21.31 | 43705601  | rs365825   | 4.2×10 <sup>-32</sup> | -189199 | 0.707 | rs17631303 | 1.0×10 <sup>-8</sup>   | 23544013  | Ovarian cancer in BRCA1 mutation carriers |                     |
| 17q21.31 | 43705601  | rs365825   | 4.2×10 <sup>-32</sup> | 9249    | 0.993 | rs2942168  | 1.0×10 <sup>-28</sup>  | 21292315  | Parkinson's disease                       | MAPT                |
| 17q21.31 | 43705601  | rs365825   | 4.2×10 <sup>-32</sup> | 13542   | 1.000 | rs393152   | 2.0×10 <sup>-16</sup>  | 19915575  | Parkinson's disease                       | MAPT, C17orf69, ... |
| 17q21.31 | 43705601  | rs365825   | 4.2×10 <sup>-32</sup> | 218082  | 0.993 | rs12185268 | 3.0×10 <sup>-14</sup>  | 21738487  | Parkinson's disease                       | MAPT                |
| 17q21.31 | 43705601  | rs365825   | 4.2×10 <sup>-32</sup> | 218618  | 0.985 | rs12373124 | 5.0×10 <sup>-10</sup>  | 22693459  | Male-pattern baldness                     | Intergenic          |
| 17q21.31 | 43705601  | rs365825   | 4.2×10 <sup>-32</sup> | 219696  | 0.803 | rs17690703 | 6.0×10 <sup>-9</sup>   | 24429156  | Idiopathic pulmonary fibrosis             | SPPL2C, CRHR1, ...  |
| 17q21.31 | 43705601  | rs365825   | 4.2×10 <sup>-32</sup> | 272226  | 0.993 | rs1864325  | 5.0×10 <sup>-11</sup>  | 22504420  | Bone mineral density                      | MAPT                |
| 17q21.31 | 43705601  | rs365825   | 4.2×10 <sup>-32</sup> | 351166  | 0.993 | rs1981997  | 9.0×10 <sup>-14</sup>  | 23583980  | Interstitial lung disease                 | MAPT                |
| 17q21.31 | 43705601  | rs365825   | 4.2×10 <sup>-32</sup> | 375463  | 0.985 | rs8070723  | 2.0×10 <sup>-118</sup> | 21685912  | Progressive supranuclear palsy            | MAPT                |
| 17q21.31 | 43705601  | rs365825   | 4.2×10 <sup>-32</sup> | 375463  | 0.985 | rs8070723  | 7.0×10 <sup>-12</sup>  | 21044948  | Parkinson's disease                       | MAPT                |
| 17q21.31 | 43705601  | rs365825   | 4.2×10 <sup>-32</sup> | 481656  | 0.964 | rs9303525  | 8.0×10 <sup>-15</sup>  | 22504418  | Intracranial volume                       | MAPT, GRN, ...      |
| 17q21.31 | 43705601  | rs365825   | 4.2×10 <sup>-32</sup> | 481891  | 0.993 | rs17577094 | 8.0×10 <sup>-9</sup>   | 24842889  | Parkinson's disease                       | MAPT                |
| 1q22     | 155206167 | rs2230288  | 4.7×10 <sup>-17</sup> | -98000  | 0.889 | rs12726330 | 5.0×10 <sup>-8</sup>   | 22451204  | Parkinson's disease                       | GBA                 |
| 2q24.3   | 169155595 | rs13016703 | 2.7×10 <sup>-16</sup> | -38570  | 0.697 | rs2102808  | 4.0×10 <sup>-10</sup>  | 21292315  | Parkinson's disease                       | STK39               |
| 2q24.3   | 169155595 | rs13016703 | 2.7×10 <sup>-16</sup> | -12560  | 1.000 | rs9917256  | 3.0×10 <sup>-6</sup>   | 22451204  | Parkinson's disease                       | Intergenic          |
| 4p16.3   | 873286    | rs35541465 | 2.8×10 <sup>-15</sup> | 65801   | 0.677 | rs6599388  | 4.0×10 <sup>-12</sup>  | 21292315  | Parkinson's disease                       | GAK                 |
| 3q27.1   | 182730291 | rs9858038  | 3.0×10 <sup>-14</sup> | 24918   | 0.708 | rs2270968  | 1.0×10 <sup>-16</sup>  | 24816252  | Blood metabolite levels                   | MCCC1               |
| 3q27.1   | 182730291 | rs9858038  | 3.0×10 <sup>-14</sup> | 29782   | 1.000 | rs10513789 | 3.0×10 <sup>-10</sup>  | 21738487  | Parkinson's disease                       | MCCC1, LAMP3        |
| 3q27.1   | 182730291 | rs9858038  | 3.0×10 <sup>-14</sup> | 90984   | 0.594 | rs11711441 | 8.0×10 <sup>-12</sup>  | 21292315  | Parkinson's disease                       | MCCC1, LAMP3        |
| 1q32.1   | 205723572 | rs823118   | 6.3×10 <sup>-12</sup> | -33765  | 0.570 | rs823094   | 2.0×10 <sup>-7</sup>   | 23449627  | Pubertal anthropometrics                  | NUCKS1,RAB7L1       |

|         |           |           |                       |        |       |            |                       |          |                           |            |
|---------|-----------|-----------|-----------------------|--------|-------|------------|-----------------------|----------|---------------------------|------------|
| 1q32.1  | 205723572 | rs823118  | $6.3 \times 10^{-12}$ | -4040  | 0.995 | rs823114   | $3.0 \times 10^{-6}$  | 24842889 | Parkinson's disease       | PARK16     |
| 4q21.1  | 77198986  | rs6812193 | $7.3 \times 10^{-11}$ | 0      | 1.000 | rs6812193  | $8.0 \times 10^{-10}$ | 21738487 | Parkinson's disease       | SCARB2     |
| 4q21.1  | 77198986  | rs6812193 | $7.3 \times 10^{-11}$ | 0      | 1.000 | rs6812193  | $4.0 \times 10^{-7}$  | 19915575 | Parkinson's disease       | STBD1      |
| 4p15.32 | 15737118  | rs4266290 | $8.4 \times 10^{-11}$ | 819    | 0.583 | rs4538475  | $3.0 \times 10^{-9}$  | 19915576 | Parkinson's disease       | BST1       |
| 18q12.3 | 40678235  | rs4130047 | $1.5 \times 10^{-10}$ | -4855  | 1.000 | rs12456492 | $2.0 \times 10^{-10}$ | 22451204 | Parkinson's disease       | RIT2       |
| 18q12.3 | 40678235  | rs4130047 | $1.5 \times 10^{-10}$ | 0      | 1.000 | rs4130047  | $2.0 \times 10^{-7}$  | 21738487 | Parkinson's disease       | RIT2, SYT4 |
| 6p22.1  | 27676794  | rs4713118 | $2.0 \times 10^{-10}$ | 344026 | 0.502 | rs172166   | $5.0 \times 10^{-6}$  | 23247143 | Cardiac Troponin-T levels | OR2B7P     |

## Replication of GWAS Catalog Results



The following table shows, for each GWAS Catalog result for similar traits, our association test result for our best available proxy (distance < 100kb,  $r^2 > 0.8$ ).

| region  | position  | our.name   | our.pval              | dist | rsqr  | assay.name | pvalue                | pubmed.id | trait                          | genes  |
|---------|-----------|------------|-----------------------|------|-------|------------|-----------------------|-----------|--------------------------------|--|
| 4p16.3  | 852313    | rs1564282  | $1.3 \times 10^{-9}$  | 0    | 1.000 | rs1564282  | $7.0 \times 10^{-7}$  | 18985386  | Parkinson's disease (familial) | GAK, DGKQ  |
| 1p36.13 | 19139517  | rs12063142 | 0.24                  | 0    | 1.000 | rs12063142 | $5.0 \times 10^{-7}$  | 20070850  | Parkinson's disease            | TAS1R2   |
| 1p21.2  | 101880005 | rs2338971  | 0.23                  | 0    | 1.000 | rs2338971  | $5.0 \times 10^{-10}$ | 24511991  | Parkinson's disease            | HUSEYO   |
| 1q21.3  | 154855055 | rs1630500  | 0.0086                | 0    | 1.000 | rs1630500  | $2.0 \times 10^{-8}$  | 24842889  | Parkinson's disease            | GBA  |
| 1q22    | 155108167 | rs12726330 | $4.9 \times 10^{-16}$ | 0    | 1.000 | rs12726330 | $5.0 \times 10^{-8}$  | 22451204  | Parkinson's disease            | GBA  |
| 1q22    | 156030037 | rs34372695 | $1.3 \times 10^{-5}$  | 0    | 1.000 | rs34372695 | $4.0 \times 10^{-12}$ | 21292315  | Parkinson's disease            | SYT11  |
| 1q32.1  | 205713378 | rs823128   | $7.7 \times 10^{-6}$  | 0    | 1.000 | rs823128   | $7.0 \times 10^{-8}$  | 19915575  | Parkinson's disease            | PARK16, NUCKS1                                   |
| 1q32.1  | 205719532 | rs823114   | $6.6 \times 10^{-12}$ | 0    | 1.000 | rs823114   | $3.0 \times 10^{-6}$  | 24842889  | Parkinson's disease            | PARK16   |
| 1q32.1  | 205752665 | rs947211   | $6.8 \times 10^{-6}$  | 0    | 1.000 | rs947211   | $2.0 \times 10^{-12}$ | 19915576  | Parkinson's disease            | PARK16, SLC45A3, NUCKS1, RAB7L1, SLC41A1, PM20D1 |
| 1q32.1  | 205764640 | rs823156   | $2.5 \times 10^{-6}$  | 0    | 1.000 | rs823156   | $1.0 \times 10^{-7}$  | 21738487  | Parkinson's disease            | SLC41A1  |
| 2q21.3  | 135539967 | rs6430538  |                       | 0    | 1.000 | rs6430538  | $2.0 \times 10^{-7}$  | 22451204  | Parkinson's disease            | Intergenic                                       |
| 2q24.3  | 169117025 | rs2102808  | $6.5 \times 10^{-16}$ | 0    | 1.000 | rs2102808  | $4.0 \times 10^{-10}$ | 21292315  | Parkinson's disease            | STK39  |
| 2q24.3  | 169143035 | rs9917256  | $8.0 \times 10^{-15}$ | 0    | 1.000 | rs9917256  | $3.0 \times 10^{-6}$  | 22451204  | Parkinson's disease            | Intergenic                                       |
| 3p24.1  | 28705764  | rs7617877  | 0.0095                | 0    | 1.000 | rs7617877  | $3.0 \times 10^{-6}$  | 21044948  | Parkinson's disease            |  |
| 3q27.1  | 182760073 | rs10513789 | $1.1 \times 10^{-13}$ | 0    | 1.000 | rs10513789 | $3.0 \times 10^{-10}$ | 21738487  | Parkinson's disease            | MCCC1, LAMP3                                     |
| 3q27.1  | 182821275 | rs11711441 | $3.3 \times 10^{-9}$  | 0    | 1.000 | rs11711441 | $8.0 \times 10^{-12}$ | 21292315  | Parkinson's disease            | MCCC1, LAMP3                                     |
| 4p16.3  | 858332    | rs11248051 | $2.4 \times 10^{-9}$  | 0    | 1.000 | rs11248051 | $3.0 \times 10^{-9}$  | 20711177  | Parkinson's disease            | GAK  |
| 4p16.3  | 939087    | rs6599388  |                       | 0    | 1.000 | rs6599388  | $4.0 \times 10^{-12}$ | 21292315  | Parkinson's disease            | GAK  |
| 4p16.3  | 939113    | rs6599389  | $8.7 \times 10^{-9}$  | 0    | 1.000 | rs6599389  | $4.0 \times 10^{-8}$  | 21738487  | Parkinson's disease            | GAK  |
| 4p16.3  | 964359    | rs11248060 | $2.3 \times 10^{-11}$ | 0    | 1.000 | rs11248060 | $3.0 \times 10^{-9}$  | 22451204  | Parkinson's disease            | DGKQ   |
| 4p16.3  | 964359    | rs11248060 | $2.3 \times 10^{-11}$ | 0    | 1.000 | rs11248060 | $3.0 \times 10^{-12}$ | 22438815  | Parkinson's disease            | GAK, DGKQ  |
| 4p16.3  | 964359    | rs11248060 | $2.3 \times 10^{-11}$ | 0    | 1.000 | rs11248060 | $2.0 \times 10^{-6}$  | 20070850  | Parkinson's disease            | DGKQ   |
| 4p15.32 | 15737101  | rs11724635 | $2.3 \times 10^{-7}$  | 0    | 1.000 | rs11724635 | $1.0 \times 10^{-16}$ | 21292315  | Parkinson's disease            | BST1   |
| 4p15.32 | 15737348  | rs4698412  | $2.5 \times 10^{-7}$  | 0    | 1.000 | rs4698412  | $3.0 \times 10^{-7}$  | 22451204  | Parkinson's disease            | BST1   |
| 4p15.32 | 15737348  | rs4698412  | $2.5 \times 10^{-7}$  | 0    | 1.000 | rs4698412  | $2.0 \times 10^{-6}$  | 21084426  | Parkinson's disease            | BST1   |
| 4p15.32 | 15737937  | rs4538475  | $2.1 \times 10^{-6}$  | 0    | 1.000 | rs4538475  | $3.0 \times 10^{-9}$  | 19915576  | Parkinson's disease            | BST1   |
| 4q13.2  | 68447249  | rs2242330  | 0.70                  | 0    | 1.000 | rs2242330  | $2.0 \times 10^{-6}$  | 17052657  | Parkinson's disease            | BRDG1  |
| 4q21.1  | 77198986  | rs6812193  | $7.3 \times 10^{-11}$ | 0    | 1.000 | rs6812193  | $8.0 \times 10^{-10}$ | 21738487  | Parkinson's disease            | SCARB2   |
| 4q21.1  | 77198986  | rs6812193  | $7.3 \times 10^{-11}$ | 0    | 1.000 | rs6812193  | $4.0 \times 10^{-7}$  | 19915575  | Parkinson's disease            | STBD1  |
| 4q22.1  | 90637601  | rs356219   | $2.2 \times 10^{-56}$ | 0    | 1.000 | rs356219   | $6.0 \times 10^{-65}$ | 22438815  | Parkinson's disease            | SNCA   |
| 4q22.1  | 90637601  | rs356219   | $2.2 \times 10^{-56}$ | 0    | 1.000 | rs356219   | $2.0 \times 10^{-47}$ | 21292315  | Parkinson's disease            | SNCA   |
| 4q22.1  | 90639515  | rs11931074 | $2.1 \times 10^{-21}$ | 0    | 1.000 | rs11931074 | $7.0 \times 10^{-17}$ | 19915576  | Parkinson's disease            | SNCA   |
| 4q22.1  | 90641340  | rs356220   | $7.6 \times 10^{-54}$ | 0    | 1.000 | rs356220   | $3.0 \times 10^{-11}$ | 24511991  | Parkinson's disease            | SNCA   |
| 4q22.1  | 90641340  | rs356220   | $7.6 \times 10^{-54}$ | 0    | 1.000 | rs356220   | $8.0 \times 10^{-35}$ | 22451204  | Parkinson's disease            | SNCA   |
| 4q22.1  | 90641340  | rs356220   | $7.6 \times 10^{-54}$ | 0    | 1.000 | rs356220   | $2.0 \times 10^{-19}$ | 21738487  | Parkinson's disease            | SNCA   |
| 4q22.1  | 90641340  | rs356220   | $7.6 \times 10^{-54}$ | 0    | 1.000 | rs356220   | $3.0 \times 10^{-8}$  | 21084426  | Parkinson's disease            | SNCA   |
| 4q22.1  | 90641340  | rs356220   | $7.6 \times 10^{-54}$ | 0    | 1.000 | rs356220   | $9.0 \times 10^{-16}$ | 21044948  | Parkinson's disease            | SNCA   |
| 4q22.1  | 90641340  | rs356220   | $7.6 \times 10^{-54}$ | 0    | 1.000 | rs356220   | $3.0 \times 10^{-11}$ | 20711177  | Parkinson's disease            | SNCA   |
| 4q22.1  | 90678541  | rs2736990  | $9.9 \times 10^{-44}$ | 0    | 1.000 | rs2736990  | $7.0 \times 10^{-8}$  | 20070850  | Parkinson's disease            | SNCA   |
| 4q22.1  | 90678541  | rs2736990  | $9.9 \times 10^{-44}$ | 0    | 1.000 | rs2736990  | $2.0 \times 10^{-16}$ | 19915575  | Parkinson's disease            | SNCA   |
| 4q22.1  | 90780902  | rs6532194  | 0.014                 | 0    | 1.000 | rs6532194  | $5.0 \times 10^{-11}$ | 22438815  | Parkinson's disease            | SNCA   |
| 4q22.1  | 90797301  | rs6532197  | $1.8 \times 10^{-7}$  | 0    | 1.000 | rs6532197  | $1.0 \times 10^{-7}$  | 19915575  | Parkinson's disease            | MMRN1  |
| 5p15.31 | 9332281   | rs7702187  | 0.11                  | 0    | 1.000 | rs7702187  | $8.0 \times 10^{-6}$  | 16252231  | Parkinson's disease            | SEMA5A   |
| 5q35.3  | 179794208 | rs10464059 | 0.66                  | 0    | 1.000 | rs10464059 | $3.0 \times 10^{-6}$  | 20070850  | Parkinson's disease            |  |
| 6p21.32 | 32387809  | rs2395163  | $1.6 \times 10^{-10}$ | 0    | 1.000 | rs2395163  | $3.0 \times 10^{-11}$ | 22451204  | Parkinson's disease            | LOC642072  |
| 6p21.32 | 32409530  | rs3129882  | 0.00032               | 0    | 1.000 | rs3129882  | $5.0 \times 10^{-10}$ | 24511991  | Parkinson's disease            | HLA  |

|          |           |            |                       |   |       |            |                       |          |                     |                                     |
|----------|-----------|------------|-----------------------|---|-------|------------|-----------------------|----------|---------------------|-------------------------------------|
| 6p21.32  | 32409530  | rs3129882  | 0.00032               | 0 | 1.000 | rs3129882  | 2.0×10 <sup>-10</sup> | 20711177 | Parkinson's disease | HLA-DRA                             |
| 6q25.2   | 154726988 | rs2275336  | 0.43                  | 0 | 1.000 | rs2275336  | 9.0×10 <sup>-7</sup>  | 22451204 | Parkinson's disease | CNKSRR3                             |
| 8p23.1   | 11698747  | rs1296028  | 0.0048                | 0 | 1.000 | rs1296028  | 1.0×10 <sup>-6</sup>  | 22451204 | Parkinson's disease | Intergenic                          |
| 9p22.2   | 17731921  | rs1536076  | 2.3×10 <sup>-7</sup>  | 0 | 1.000 | rs1536076  | 5.0×10 <sup>-7</sup>  | 22451204 | Parkinson's disease | SH3GL2                              |
| 9p13.3   | 35269819  | rs10121009 | 0.56                  | 0 | 1.000 | rs10121009 | 3.0×10 <sup>-6</sup>  | 21812969 | Parkinson's disease | UNC13B                              |
| 9q33.1   | 122059709 | rs4837628  | 0.57                  | 0 | 1.000 | rs4837628  | 1.0×10 <sup>-6</sup>  | 20070850 | Parkinson's disease | DBC1                                |
| 10p13    | 15561543  | rs7077361  | 0.0083                | 0 | 1.000 | rs7077361  | 5.0×10 <sup>-6</sup>  | 19915575 | Parkinson's disease | ITGA8                               |
| 10q11.21 | 45161109  | rs1480597  | 0.62                  | 0 | 1.000 | rs1480597  | 2.0×10 <sup>-6</sup>  | 17052657 | Parkinson's disease | Intergenic                          |
| 10q22.1  | 71580120  | rs17497526 | 0.26                  | 0 | 1.000 | rs17497526 | 6.0×10 <sup>-7</sup>  | 23793441 | Parkinson's disease | COL13A1                             |
| 10q24.32 | 104591393 | rs17115100 | 0.98                  | 0 | 1.000 | rs17115100 | 7.0×10 <sup>-8</sup>  | 22451204 | Parkinson's disease | CYP17A1, C10orf32, CNM2, SFXN2      |
| 11p14.3  | 22114093  | rs11026412 | 0.34                  | 0 | 1.000 | rs11026412 | 3.0×10 <sup>-6</sup>  | 22451204 | Parkinson's disease | Intergenic                          |
| 11q14.1  | 84417846  | rs10501570 | 0.91                  | 0 | 1.000 | rs10501570 | 7.0×10 <sup>-6</sup>  | 17052657 | Parkinson's disease | DLG2                                |
| 11q21    | 96108428  | rs7118648  | 0.78                  | 0 | 1.000 | rs7118648  | 2.0×10 <sup>-7</sup>  | 23793441 | Parkinson's disease | CCDC82                              |
| 12q12    | 40352996  | rs10877840 | 0.76                  | 0 | 1.000 | rs10877840 | 1.0×10 <sup>-6</sup>  | 22451204 | Parkinson's disease | SLC2A13                             |
| 12q12    | 40428561  | rs1994090  | 0.27                  | 0 | 1.000 | rs1994090  | 3.0×10 <sup>-8</sup>  | 19915575 | Parkinson's disease | LRRK2                               |
| 12q12    | 40620808  | rs1491942  | 7.9×10 <sup>-8</sup>  | 0 | 1.000 | rs1491942  | 6.0×10 <sup>-15</sup> | 22438815 | Parkinson's disease | LRRK2                               |
| 12q12    | 40620808  | rs1491942  | 7.9×10 <sup>-8</sup>  | 0 | 1.000 | rs1491942  | 6.0×10 <sup>-14</sup> | 21292315 | Parkinson's disease | LRRK2                               |
| 12q12    | 40734202  | rs34637584 |                       | 0 | 1.000 | rs34637584 | 2.0×10 <sup>-28</sup> | 21738487 | Parkinson's disease | LRRK2                               |
| 12q12    | 40757328  | rs34778348 |                       | 0 | 1.000 | rs34778348 | 3.0×10 <sup>-21</sup> | 22438815 | Parkinson's disease | LRRK2                               |
| 12q12    | 41365640  | rs1442190  | 2.5×10 <sup>-9</sup>  | 0 | 1.000 | rs1442190  | 2.0×10 <sup>-27</sup> | 24842889 | Parkinson's disease | LRRK2                               |
| 12q23.3  | 106949987 | rs4964469  | 0.86                  | 0 | 1.000 | rs4964469  | 2.0×10 <sup>-7</sup>  | 21084426 | Parkinson's disease | Intergenic                          |
| 12q24.31 | 123296294 | rs12817488 | 3.8×10 <sup>-10</sup> | 0 | 1.000 | rs12817488 | 3.0×10 <sup>-13</sup> | 21292315 | Parkinson's disease | CCDC62, HIP1R                       |
| 14q22.2  | 54290830  | rs12431733 | 0.043                 | 0 | 1.000 | rs12431733 | 3.0×10 <sup>-6</sup>  | 19915575 | Parkinson's disease | BMP4                                |
| 15q22.2  | 62001132  | rs10519131 | 1.2×10 <sup>-5</sup>  | 0 | 1.000 | rs10519131 | 1.0×10 <sup>-6</sup>  | 22451204 | Parkinson's disease | Intergenic                          |
| 15q25.1  | 81626725  | rs3935740  | 0.92                  | 0 | 1.000 | rs3935740  | 2.0×10 <sup>-7</sup>  | 23793441 | Parkinson's disease | TMC3                                |
| 16p11.2  | 31095171  | rs11865038 | 4.2×10 <sup>-5</sup>  | 0 | 1.000 | rs11865038 | 4.0×10 <sup>-7</sup>  | 22451204 | Parkinson's disease | POL3S                               |
| 17p11.2  | 17715101  | rs11868035 | 1.6×10 <sup>-5</sup>  | 0 | 1.000 | rs11868035 | 6.0×10 <sup>-8</sup>  | 21738487 | Parkinson's disease | SREBF1, RAI1                        |
| 17q21.31 | 43513441  | rs11012    | 1.9×10 <sup>-28</sup> | 0 | 1.000 | rs11012    | 6.0×10 <sup>-8</sup>  | 20070850 | Parkinson's disease | PLEKHM1, MAPT, IMP5                 |
| 17q21.31 | 43714850  | rs2942168  | 4.0×10 <sup>-30</sup> | 0 | 1.000 | rs2942168  | 1.0×10 <sup>-28</sup> | 21292315 | Parkinson's disease | MAPT                                |
| 17q21.31 | 43719143  | rs393152   | 4.0×10 <sup>-30</sup> | 0 | 1.000 | rs393152   | 2.0×10 <sup>-16</sup> | 19915575 | Parkinson's disease | MAPT, C17orf69, KIAA1267, LOC644246 |
| 17q21.31 | 43923683  | rs12185268 | 9.9×10 <sup>-31</sup> | 0 | 1.000 | rs12185268 | 3.0×10 <sup>-14</sup> | 21738487 | Parkinson's disease | MAPT                                |
| 17q21.31 | 44081064  | rs8070723  | 7.0×10 <sup>-30</sup> | 0 | 1.000 | rs8070723  | 7.0×10 <sup>-12</sup> | 21044948 | Parkinson's disease | MAPT                                |
| 17q21.31 | 44187492  | rs17577094 | 5.2×10 <sup>-30</sup> | 0 | 1.000 | rs17577094 | 8.0×10 <sup>-9</sup>  | 24842889 | Parkinson's disease | MAPT                                |
| 17q21.31 | 44788310  | rs183211   | 3.5×10 <sup>-21</sup> | 0 | 1.000 | rs183211   | 3.0×10 <sup>-7</sup>  | 21812969 | Parkinson's disease | NSF                                 |
| 17q21.31 | 44828931  | rs199533   | 4.4×10 <sup>-26</sup> | 0 | 1.000 | rs199533   | 1.0×10 <sup>-6</sup>  | 20711177 | Parkinson's disease | MAPT                                |
| 17q21.31 | 44828931  | rs199533   | 4.4×10 <sup>-26</sup> | 0 | 1.000 | rs199533   | 1.0×10 <sup>-14</sup> | 19915575 | Parkinson's disease | NSF                                 |
| 17q21.31 | 44856641  | rs199515   | 3.3×10 <sup>-25</sup> | 0 | 1.000 | rs199515   | 3.0×10 <sup>-17</sup> | 22451204 | Parkinson's disease | WNT3                                |
| 17q21.31 | 44859144  | rs415430   | 4.3×10 <sup>-25</sup> | 0 | 1.000 | rs415430   | 7.0×10 <sup>-7</sup>  | 21812969 | Parkinson's disease | WNT3                                |
| 17q21.31 | 44856603  | rs199498   | 2.8×10 <sup>-23</sup> | 0 | 1.000 | rs199498   | 8.0×10 <sup>-7</sup>  | 24511991 | Parkinson's disease | MAPT                                |
| 18q12.3  | 40673380  | rs12456492 | 2.3×10 <sup>-10</sup> | 0 | 1.000 | rs12456492 | 2.0×10 <sup>-10</sup> | 22451204 | Parkinson's disease | RIT2                                |
| 18q12.3  | 40678235  | rs4130047  | 1.5×10 <sup>-10</sup> | 0 | 1.000 | rs4130047  | 2.0×10 <sup>-7</sup>  | 21738487 | Parkinson's disease | RIT2, SYT4                          |
| 20p12.1  | 13296912  | rs1223271  | 0.83                  | 0 | 1.000 | rs1223271  | 5.0×10 <sup>-6</sup>  | 19915575 | Parkinson's disease | C20orf82                            |
| 21q21.1  | 16914905  | rs2823357  | 0.50                  | 0 | 1.000 | rs2823357  | 6.0×10 <sup>-7</sup>  | 21738487 | Parkinson's disease | USP25                               |
| 21q22.3  | 43286352  | rs2839398  | 0.36                  | 0 | 1.000 | rs2839398  | 5.0×10 <sup>-6</sup>  | 22451204 | Parkinson's disease | PRDM15                              |

## Nearby Nonsynonymous SNPs

| region   | position  | our.name   | our.pval              | dist    | rsqr  | assay.name  | gene     | aa.chg |
|----------|-----------|------------|-----------------------|---------|-------|-------------|----------|--------|
| 17q21.31 | 43705601  | rs365825   | 4.2×10 <sup>-32</sup> | -198304 | 0.594 | rs12949256  | ARHGAP27 | A117T  |
| 17q21.31 | 43705601  | rs365825   | 4.2×10 <sup>-32</sup> | 217341  | 0.993 | rs62621252  | SPPL2C   | S224P  |
| 17q21.31 | 43705601  | rs365825   | 4.2×10 <sup>-32</sup> | 217665  | 0.993 | rs62054815  | SPPL2C   | A332T  |
| 17q21.31 | 43705601  | rs365825   | 4.2×10 <sup>-32</sup> | 218053  | 0.993 | rs12185233  | SPPL2C   | R461P  |
| 17q21.31 | 43705601  | rs365825   | 4.2×10 <sup>-32</sup> | 218082  | 0.993 | rs12185268  | SPPL2C   | I471V  |
| 17q21.31 | 43705601  | rs365825   | 4.2×10 <sup>-32</sup> | 218472  | 0.993 | rs12373123  | SPPL2C   | S601P  |
| 17q21.31 | 43705601  | rs365825   | 4.2×10 <sup>-32</sup> | 218529  | 0.993 | rs12373139  | SPPL2C   | G620R  |
| 17q21.31 | 43705601  | rs365825   | 4.2×10 <sup>-32</sup> | 218599  | 0.985 | rs12373142  | SPPL2C   | P643R  |
| 17q21.31 | 43705601  | rs365825   | 4.2×10 <sup>-32</sup> | 218618  | 0.985 | rs12373124  | SPPL2C   | H649H  |
| 17q21.31 | 43705601  | rs365825   | 4.2×10 <sup>-32</sup> | 218630  | 0.985 | rs12373140  | SPPL2C   | Q653Q  |
| 17q21.31 | 43705601  | rs365825   | 4.2×10 <sup>-32</sup> | 355174  | 0.993 | rs63750417  | MAPT     | P202L  |
| 17q21.31 | 43705601  | rs365825   | 4.2×10 <sup>-32</sup> | 355422  | 0.993 | rs62063786  | MAPT     | D285N  |
| 17q21.31 | 43705601  | rs365825   | 4.2×10 <sup>-32</sup> | 355435  | 0.993 | rs62063787  | MAPT     | V289A  |
| 17q21.31 | 43705601  | rs365825   | 4.2×10 <sup>-32</sup> | 355677  | 0.993 | rs17651549  | MAPT     | R370W  |
| 17q21.31 | 43705601  | rs365825   | 4.2×10 <sup>-32</sup> | 361799  | 0.993 | rs10445337  | MAPT     | S447P  |
| 17q21.31 | 43705601  | rs365825   | 4.2×10 <sup>-32</sup> | 398810  | 0.942 | rs199706121 | MAPT     | ?1735? |
| 17q21.31 | 43705601  | rs365825   | 4.2×10 <sup>-32</sup> | 402885  | 0.756 | rs200384907 | KANSL1   | ?1225? |
| 17q21.31 | 43705601  | rs365825   | 4.2×10 <sup>-32</sup> | 403305  | 0.993 | rs34579536  | KANSL1   | I1085T |
| 17q21.31 | 43705601  | rs365825   | 4.2×10 <sup>-32</sup> | 404931  | 0.993 | rs36076725  | KANSL1   | F917F  |
| 17q21.31 | 43705601  | rs365825   | 4.2×10 <sup>-32</sup> | 404940  | 0.993 | rs35833914  | KANSL1   | D914D  |
| 17q21.31 | 43705601  | rs365825   | 4.2×10 <sup>-32</sup> | 411518  | 0.993 | rs34043286  | KANSL1   | S718P  |
| 6p21.32  | 32571961  | rs17425622 | 3.8×10 <sup>-17</sup> | 38440   | 0.634 | rs9272785   | HLA-DQA1 | A210T  |
| 1q22     | 155206167 | rs2230288  | 4.7×10 <sup>-17</sup> | 0       | 1.000 | rs2230288   | GBA      | E365K  |
| 14q22.2  | 55348869  | rs11158026 | 8.8×10 <sup>-15</sup> | 57448   | 0.646 | rs199893292 | WDHD1    | ?1761? |
| 3q27.1   | 182730291 | rs9858038  | 3.0×10 <sup>-14</sup> | 24918   | 0.708 | rs2270968   | MCCC1    | H464P  |

## Nearby Expression QTLs

| region   | position | our.name | our.pval              | dist    | rsqr  | assay.name  | eqtl.dist | eqtl.gene | eqtl.pval              | eqtl.rsqr | tissue      | pubmed.id |
|----------|----------|----------|-----------------------|---------|-------|-------------|-----------|-----------|------------------------|-----------|-------------|-----------|
| 17q21.31 | 43705601 | rs365825 | 4.2×10 <sup>-32</sup> | -194166 | 0.720 | rs34465449  | 0         | CRHR1-IT1 | 2.3×10 <sup>-13</sup>  | 0.395     | Whole_Blood | 23715323  |
| 17q21.31 | 43705601 | rs365825 | 4.2×10 <sup>-32</sup> | -192160 | 0.707 | rs11012     | 201708    | MGC57346  | 6.7×10 <sup>-23</sup>  | 0.297     | B-Cell      | 22446964  |
| 17q21.31 | 43705601 | rs365825 | 4.2×10 <sup>-32</sup> | -166164 | 0.563 | rs117169618 | 0         | CRHR1     | 2.1×10 <sup>-11</sup>  | 0.362     | Lung        | 23715323  |
| 17q21.31 | 43705601 | rs365825 | 4.2×10 <sup>-32</sup> | -156075 | 0.722 | rs17631676  | 42490     | LRRC37A4  | 6.0×10 <sup>-208</sup> | 0.475     | Monocyte    | 20502693  |
| 17q21.31 | 43705601 | rs365825 | 4.2×10 <sup>-32</sup> | -156075 | 0.722 | rs17631676  | 165623    | MGC57346  | 1.8×10 <sup>-59</sup>  | 0.613     | Monocyte    | 22446964  |
| 17q21.31 | 43705601 | rs365825 | 4.2×10 <sup>-32</sup> | -156075 | 0.722 | rs17631676  | 378322    | HS.554608 | 0.00057                | 0.042     | B-Cell      | 22446964  |
| 17q21.31 | 43705601 | rs365825 | 4.2×10 <sup>-32</sup> | -156075 | 0.722 | rs17631676  | 170002    | C17ORF69  | 7.7×10 <sup>-5</sup>   | 0.055     | B-Cell      | 22446964  |
| 17q21.31 | 43705601 | rs365825 | 4.2×10 <sup>-32</sup> | -150348 | 0.722 | rs56192752  | 0         | MAPT      | 3.0×10 <sup>-6</sup>   | 0.196     | Lung        | 23715323  |

|          |           |            |                       |         |       |                  |          |              |                        |       |                            |          |
|----------|-----------|------------|-----------------------|---------|-------|------------------|----------|--------------|------------------------|-------|----------------------------|----------|
| 17q21.31 | 43705601  | rs365825   | 4.2×10 <sup>-32</sup> | -132705 | 0.676 | rs5026246        | 0        | CRHR1        | 2.3×10 <sup>-10</sup>  | 0.397 | Nerve_Tibial               | 23715323 |
| 17q21.31 | 43705601  | rs365825   | 4.2×10 <sup>-32</sup> | -131952 | 0.781 | rs188710438      | 0        | CRHR1-IT1    | 6.0×10 <sup>-8</sup>   | 0.333 | Esophagus_Mucosa           | 23715323 |
| 17q21.31 | 43705601  | rs365825   | 4.2×10 <sup>-32</sup> | -51133  | 0.892 | rs149133346      | 0        | DND1P1       | 4.8×10 <sup>-7</sup>   | 0.309 | Esophagus_Muscularis       | 23715323 |
| 17q21.31 | 43705601  | rs365825   | 4.2×10 <sup>-32</sup> | -43581  | 0.697 | rs116954807      | 0        | ARHGAP27     | 6.1×10 <sup>-7</sup>   | 0.238 | Skin_Sun_Exposed_Lower_leg | 23715323 |
| 17q21.31 | 43705601  | rs365825   | 4.2×10 <sup>-32</sup> | -14964  | 0.963 | rs147670174      | 0        | CRHR1-IT1    | 4.8×10 <sup>-28</sup>  | 0.699 | Lung                       | 23715323 |
| 17q21.31 | 43705601  | rs365825   | 4.2×10 <sup>-32</sup> | 10554   | 0.685 | rs112550936      | 0        | RP11-798G7.5 | 4.9×10 <sup>-8</sup>   | 0.336 | Esophagus_Mucosa           | 23715323 |
| 17q21.31 | 43705601  | rs365825   | 4.2×10 <sup>-32</sup> | 13542   | 1.000 | rs393152         | 580720   | NMT1         | 0.00045                | 0.156 | Lymphoblastoid             | 19644074 |
| 17q21.31 | 43705601  | rs365825   | 4.2×10 <sup>-32</sup> | 45997   | 0.978 | chr17:43751598:D | 0        | LRR37A4P     | 4.1×10 <sup>-33</sup>  | 0.799 | Thyroid                    | 23715323 |
| 17q21.31 | 43705601  | rs365825   | 4.2×10 <sup>-32</sup> | 45997   | 0.978 | chr17:43751598:D | 0        | LRR37A4P     | 6.7×10 <sup>-20</sup>  | 0.725 | Heart_Left_Ventricle       | 23715323 |
| 17q21.31 | 43705601  | rs365825   | 4.2×10 <sup>-32</sup> | 147132  | 0.993 | rs113871181      | 0        | LRR37A4P     | 1.1×10 <sup>-32</sup>  | 0.706 | Whole_Blood                | 23715323 |
| 17q21.31 | 43705601  | rs365825   | 4.2×10 <sup>-32</sup> | 150738  | 0.578 | rs117631329      | 446394   | KANSL1       | 5.3×10 <sup>-8</sup>   | 0.077 | Lymphoblastoid             | 24037378 |
| 17q21.31 | 43705601  | rs365825   | 4.2×10 <sup>-32</sup> | 313506  | 0.689 | rs111825734      | 0        | LRR37A       | 6.6×10 <sup>-7</sup>   | 0.318 | Heart_Left_Ventricle       | 23715323 |
| 17q21.31 | 43705601  | rs365825   | 4.2×10 <sup>-32</sup> | 376831  | 0.963 | rs112310745      | 0        | MAPT         | 3.2×10 <sup>-6</sup>   | 0.344 | Artery_Aorta               | 23715323 |
| 17q21.31 | 43705601  | rs365825   | 4.2×10 <sup>-32</sup> | 464476  | 0.725 | chr17:44170077:D | 0        | 669E14.6     | 1.5×10 <sup>-6</sup>   | 0.228 | Thyroid                    | 23715323 |
| 17q21.31 | 43705601  | rs365825   | 4.2×10 <sup>-32</sup> | 491826  | 0.704 | chr17:44197427:I | 0        | LRR37A2      | 3.7×10 <sup>-19</sup>  | 0.634 | Nerve_Tibial               | 23715323 |
| 17q21.31 | 43705601  | rs365825   | 4.2×10 <sup>-32</sup> | 491826  | 0.704 | chr17:44197427:I | 0        | 259G18.2     | 4.1×10 <sup>-33</sup>  | 0.799 | Thyroid                    | 23715323 |
| 17q21.31 | 43705601  | rs365825   | 4.2×10 <sup>-32</sup> | 495757  | 0.971 | chr17:44201358:D | 0        | LRR37A4P     | 3.5×10 <sup>-23</sup>  | 0.851 | Artery_Aorta               | 23715323 |
| 6p21.32  | 32571961  | rs17425622 | 3.8×10 <sup>-17</sup> | -12057  | 0.573 | rs35743245       | 246695   | BPG246D15.9  | 1.2×10 <sup>-6</sup>   | 0.061 | Lymphoblastoid             | 24037378 |
| 6p21.32  | 32571961  | rs17425622 | 3.8×10 <sup>-17</sup> | 1601    | 0.602 | rs602457         | -1018080 | LST1         | 0.00022                | 0.048 | Monocyte                   | 22446964 |
| 6p21.32  | 32571961  | rs17425622 | 3.8×10 <sup>-17</sup> | 8656    | 0.660 | rs3997872        | 116324   | HLA-DRB3     | 2.2×10 <sup>-48</sup>  | 0.136 | Monocyte                   | 20502693 |
| 1q22     | 155206167 | rs2230288  | 4.7×10 <sup>-17</sup> | 231544  | 0.830 | rs35777901       | -326859  | RAG1A1P      | 0.00022                | 0.048 | Monocyte                   | 22446964 |
| 2q24.3   | 169155595 | rs13016703 | 2.7×10 <sup>-16</sup> | 1312    | 0.768 | rs12616285       | -346138  | STK39        | 8.6×10 <sup>-7</sup>   | 0.083 | B-Cell                     | 22446964 |
| 4p16.3   | 873286    | rs35541465 | 2.8×10 <sup>-15</sup> | 65801   | 0.677 | rs6599388        | 0        | IDUA         | 8.6×10 <sup>-8</sup>   | 0.342 | Esophagus_Muscularis       | 23715323 |
| 14q22.2  | 55348869  | rs11158026 | 8.8×10 <sup>-15</sup> | 3994    | 0.826 | rs10136545       | 0        | SAMD4A       | 3.6×10 <sup>-6</sup>   | 0.188 | Whole_Blood                | 23715323 |
| 3q27.1   | 182730291 | rs9858038  | 3.0×10 <sup>-14</sup> | -3194   | 0.600 | rs9841498        | 820265   | PARL         | 0.00031                | 0.046 | Monocyte                   | 22446964 |
| 7p15.3   | 23123877  | rs10256359 | 2.1×10 <sup>-12</sup> | -4560   | 0.988 | rs7776649        | 0        | KLHL7-AS1    | 2.8×10 <sup>-8</sup>   | 0.261 | Muscle_Skeletal            | 23715323 |
| 7p15.3   | 23123877  | rs10256359 | 2.1×10 <sup>-12</sup> | -2573   | 0.915 | rs6953728        | 0        | KLHL7-AS1    | 5.5×10 <sup>-24</sup>  | 0.684 | Adipose_Subcutaneous       | 23715323 |
| 7p15.3   | 23123877  | rs10256359 | 2.1×10 <sup>-12</sup> | 16063   | 0.971 | rs10235786       | 0        | KLHL7-AS1    | 1.6×10 <sup>-13</sup>  | 0.528 | Esophagus_Mucosa           | 23715323 |
| 7p15.3   | 23123877  | rs10256359 | 2.1×10 <sup>-12</sup> | 19236   | 0.971 | rs10233039       | 2209     | KLHL7-AS1    | 6.4×10 <sup>-100</sup> | 0.705 | Lymphoblastoid             | 24037378 |
| 7p15.3   | 23123877  | rs10256359 | 2.1×10 <sup>-12</sup> | 19236   | 0.971 | rs10233039       | 0        | KLHL7-AS1    | 9.3×10 <sup>-23</sup>  | 0.651 | Skin_Sun_Exposed_Lower_leg | 23715323 |
| 7p15.3   | 23123877  | rs10256359 | 2.1×10 <sup>-12</sup> | 19236   | 0.971 | rs10233039       | 0        | KLHL7-AS1    | 2.4×10 <sup>-18</sup>  | 0.534 | Artery_Tibial              | 23715323 |
| 7p15.3   | 23123877  | rs10256359 | 2.1×10 <sup>-12</sup> | 21418   | 0.730 | rs10256996       | 107      | KLHL7        | 4.6×10 <sup>-6</sup>   | 0.016 | Monocyte                   | 20502693 |
| 7p15.3   | 23123877  | rs10256359 | 2.1×10 <sup>-12</sup> | 21418   | 0.730 | rs10256996       | 0        | KLHL7-AS1    | 5.0×10 <sup>-8</sup>   | 0.369 | Heart_Left_Ventricle       | 23715323 |
| 7p15.3   | 23123877  | rs10256359 | 2.1×10 <sup>-12</sup> | 21960   | 0.618 | rs1005786        | 0        | AC005082.12  | 3.9×10 <sup>-8</sup>   | 0.261 | Lung                       | 23715323 |
| 7p15.3   | 23123877  | rs10256359 | 2.1×10 <sup>-12</sup> | 28776   | 0.947 | rs73272048       | 0        | KLHL7-AS1    | 3.7×10 <sup>-17</sup>  | 0.508 | Lung                       | 23715323 |
| 7p15.3   | 23123877  | rs10256359 | 2.1×10 <sup>-12</sup> | 29998   | 0.947 | rs764533         | 0        | NUPL2        | 1.8×10 <sup>-6</sup>   | 0.249 | Nerve_Tibial               | 23715323 |
| 7p15.3   | 23123877  | rs10256359 | 2.1×10 <sup>-12</sup> | 46899   | 0.941 | rs4140959        | 50710    | NUPL2        | 3.7×10 <sup>-44</sup>  | 0.126 | Monocyte                   | 20502693 |
| 7p15.3   | 23123877  | rs10256359 | 2.1×10 <sup>-12</sup> | 50889   | 0.935 | rs6948469        | 0        | NUPL2        | 5.9×10 <sup>-9</sup>   | 0.409 | Heart_Left_Ventricle       | 23715323 |
| 7p15.3   | 23123877  | rs10256359 | 2.1×10 <sup>-12</sup> | 64232   | 0.877 | rs6962213        | 0        | NUPL2        | 3.0×10 <sup>-6</sup>   | 0.196 | Lung                       | 23715323 |
| 7p15.3   | 23123877  | rs10256359 | 2.1×10 <sup>-12</sup> | 70728   | 0.787 | rs955187         | 45526    | NUPL2        | 1.4×10 <sup>-9</sup>   | 0.123 | B-Cell                     | 22446964 |
| 7p15.3   | 23123877  | rs10256359 | 2.1×10 <sup>-12</sup> | 70728   | 0.787 | rs955187         | 45526    | NUPL2        | 6.1×10 <sup>-19</sup>  | 0.246 | Monocyte                   | 22446964 |
| 7p15.3   | 23123877  | rs10256359 | 2.1×10 <sup>-12</sup> | 87475   | 0.883 | rs6964665        | 0        | KLHL7-AS1    | 1.2×10 <sup>-21</sup>  | 0.640 | Thyroid                    | 23715323 |
| 7p15.3   | 23123877  | rs10256359 | 2.1×10 <sup>-12</sup> | 96358   | 0.832 | rs10254544       | 0        | KLHL7-AS1    | 2.1×10 <sup>-9</sup>   | 0.489 | Stomach                    | 23715323 |
| 7p15.3   | 23123877  | rs10256359 | 2.1×10 <sup>-12</sup> | 96573   | 0.832 | rs10242104       | 0        | KLHL7-AS1    | 7.7×10 <sup>-18</sup>  | 0.660 | Esophagus_Muscularis       | 23715323 |
| 7p15.3   | 23123877  | rs10256359 | 2.1×10 <sup>-12</sup> | 97993   | 0.758 | rs7808488        | 0        | AC005082.12  | 4.9×10 <sup>-7</sup>   | 0.294 | Esophagus_Mucosa           | 23715323 |
| 7p15.3   | 23123877  | rs10256359 | 2.1×10 <sup>-12</sup> | 119433  | 0.634 | rs858272         | 9530     | GNPMB        | 2.3×10 <sup>-17</sup>  | 0.412 | Brain_Frontal_Cortex       | 20485568 |
| 7p15.3   | 23123877  | rs10256359 | 2.1×10 <sup>-12</sup> | 126951  | 0.780 | rs6970694        | 0        | KLHL7-AS1    | 2.1×10 <sup>-20</sup>  | 0.659 | Nerve_Tibial               | 23715323 |
| 7p15.3   | 23123877  | rs10256359 | 2.1×10 <sup>-12</sup> | 136711  | 0.670 | rs7807879        | 0        | KLHL7-AS1    | 1.2×10 <sup>-6</sup>   | 0.204 | Whole_Blood                | 23715323 |
| 7p15.3   | 23123877  | rs10256359 | 2.1×10 <sup>-12</sup> | 138558  | 0.629 | rs1624451        | 0        | AC005082.12  | 9.2×10 <sup>-11</sup>  | 0.458 | Esophagus_Muscularis       | 23715323 |
| 7p15.3   | 23123877  | rs10256359 | 2.1×10 <sup>-12</sup> | 148267  | 0.658 | rs10227559       | 0        | NUPL2        | 2.4×10 <sup>-8</sup>   | 0.263 | Muscle_Skeletal            | 23715323 |
| 7p15.3   | 23123877  | rs10256359 | 2.1×10 <sup>-12</sup> | 171023  | 0.625 | rs858273         | 0        | AC005082.12  | 3.0×10 <sup>-10</sup>  | 0.459 | Heart_Left_Ventricle       | 23715323 |
| 7p15.3   | 23123877  | rs10256359 | 2.1×10 <sup>-12</sup> | 172656  | 0.610 | rs199355         | 0        | GNPMB        | 1.5×10 <sup>-42</sup>  | 0.122 | Monocyte                   | 20502693 |
| 7p15.3   | 23123877  | rs10256359 | 2.1×10 <sup>-12</sup> | 172656  | 0.610 | rs199355         | 17487    | GNPMB        | 1.6×10 <sup>-6</sup>   | 0.079 | Monocyte                   | 22446964 |
| 1q32.1   | 205723572 | rs823118   | 6.3×10 <sup>-12</sup> | -33765  | 0.570 | rs823094         | 54781    | RAB7L1       | 6.6×10 <sup>-11</sup>  | 0.109 | Lymphoblastoid             | 24037378 |
| 1q32.1   | 205723572 | rs823118   | 6.3×10 <sup>-12</sup> | -33765  | 0.570 | rs823094         | 0        | SLC41A1      | 3.1×10 <sup>-6</sup>   | 0.259 | Esophagus_Mucosa           | 23715323 |
| 1q32.1   | 205723572 | rs823118   | 6.3×10 <sup>-12</sup> | -9200   | 0.604 | rs823130         | 0        | SLC41A1      | 3.9×10 <sup>-6</sup>   | 0.187 | Whole_Blood                | 23715323 |
| 1q32.1   | 205723572 | rs823118   | 6.3×10 <sup>-12</sup> | -4040   | 0.995 | rs823114         | 32423    | NUCKS1       | 7.1×10 <sup>-5</sup>   | 0.195 | T-cell                     | 19644074 |
| 1q32.1   | 205723572 | rs823118   | 6.3×10 <sup>-12</sup> | -4040   | 0.995 | rs823114         | -32107   | NUCKS1       | 2.3×10 <sup>-16</sup>  | 0.215 | B-Cell                     | 22446964 |
| 1q32.1   | 205723572 | rs823118   | 6.3×10 <sup>-12</sup> | 0       | 1.000 | rs823118         | 0        | NUCKS1       | 1.3×10 <sup>-9</sup>   | 0.337 | Thyroid                    | 23715323 |
| 1q32.1   | 205723572 | rs823118   | 6.3×10 <sup>-12</sup> | 730     | 0.610 | rs823121         | 5005     | NUCKS1       | 1.2×10 <sup>-29</sup>  | 0.086 | Monocyte                   | 20502693 |
| 1q32.1   | 205723572 | rs823118   | 6.3×10 <sup>-12</sup> | 12040   | 0.568 | rs2014202        | 0        | RAB7L1       | 3.1×10 <sup>-8</sup>   | 0.255 | Whole_Blood                | 23715323 |
| 1q32.1   | 205723572 | rs823118   | 6.3×10 <sup>-12</sup> | 12319   | 0.568 | rs7522056        | 2507     | RAB7L1       | 6.1×10 <sup>-8</sup>   | 0.100 | Monocyte                   | 22446964 |
| 1q32.1   | 205723572 | rs823118   | 6.3×10 <sup>-12</sup> | 12319   | 0.568 | rs7522056        | 0        | RAB7L1       | 6.1×10 <sup>-9</sup>   | 0.346 | Nerve_Tibial               | 23715323 |
| 1q32.1   | 205723572 | rs823118   | 6.3×10 <sup>-12</sup> | 12319   | 0.568 | rs7522056        | 0        | RAB7L1       | 1.8×10 <sup>-9</sup>   | 0.327 | Skin_Sun_Exposed_Lower_leg | 23715323 |
| 1q32.1   | 205723572 | rs823118   | 6.3×10 <sup>-12</sup> | 12319   | 0.568 | rs7522056        | 0        | RAB7L1       | 1.4×10 <sup>-6</sup>   | 0.231 | Adipose_Subcutaneous       | 23715323 |
| 1q32.1   | 205723572 | rs823118   | 6.3×10 <sup>-12</sup> | 15694   | 0.989 | rs708723         | -51841   | NUCKS1       | 2.1×10 <sup>-9</sup>   | 0.120 | Monocyte                   | 22446964 |
| 1q32.1   | 205723572 | rs823118   | 6.3×10 <sup>-12</sup> | 34252   | 0.620 | rs1775148        | 0        | RAB7L1       | 7.5×10 <sup>-8</sup>   | 0.430 | Artery_Aorta               | 23715323 |
| 1q32.1   | 205723572 | rs823118   | 6.3×10 <sup>-12</sup> | 34252   | 0.620 | rs1775148        | 0        | RAB7L1       | 1.9×10 <sup>-10</sup>  | 0.333 | Artery_Tibial              | 23715323 |
| 1q32.1   | 205723572 | rs823118   | 6.3×10 <sup>-12</sup> | 35623   | 0.549 | rs1772159        | 0        | PM20D1       | 5.3×10 <sup>-19</sup>  | 0.592 | Adipose_Subcutaneous       | 23715323 |
| 1q32.1   | 205723572 | rs823118   | 6.3×10 <sup>-12</sup> | 38834   | 0.552 | rs823154         | -3940    | SLC41A1      | 3.3×10 <sup>-5</sup>   | 0.060 | Monocyte                   | 22446964 |
| 1q32.1   | 205723572 | rs823118   | 6.3×10 <sup>-12</sup> | 38834   | 0.552 | rs823154         | 0        | NUCKS1       | 1.8×10 <sup>-7</sup>   | 0.399 | Stomach                    | 23715323 |
| 1q32.1   | 205723572 | rs823118   | 6.3×10 <sup>-12</sup> | 44313   | 0.501 | rs708727         | 80776    | NUCKS1       | 0.00027                | 0.167 | Lymphoblastoid             | 19644074 |
| 1q32.1   | 205723572 | rs823118   | 6.3×10 <sup>-12</sup> | 44313   | 0.501 | rs708727         | 29269    | PM20D1       | 0.00019                | 0.176 | T-cell                     | 19644074 |
| 1q32.1   | 205723572 | rs823118   | 6.3×10 <sup>-12</sup> | 44313   | 0.501 | rs708727         | -196686  | MFS4         | 0.00027                | 0.046 | Monocyte                   | 22446964 |
| 1q32.1   | 205723572 | rs823118   | 6.3×10 <sup>-12</sup> | 44313   | 0.501 | rs708727         | 0        | PM20D1       | 1.4×10 <sup>-11</sup>  | 0.367 | Artery_Tibial              | 23715323 |
| 1q32.1   | 205723572 | rs823118   | 6.3×10 <sup>-12</sup> | 51267   | 0.504 | rs823074         | 0        | PM20D1       | 1.1×10 <sup>-17</sup>  | 0.679 | Heart_Left_Ventricle       | 23715323 |
| 1q32.1   | 205723572 | rs823118   | 6.3×10 <sup>-12</sup> | 51267   | 0.504 | rs823074         | 0        | PM20D1       | 1.0×10 <sup>-15</sup>  | 0.475 | Lung                       | 23715323 |
| 1q3      |           |            |                       |         |       |                  |          |              |                        |       |                            |          |

|         |          |           |                       |         |       |            |  |          |         |                      |       |                            |          |
|---------|----------|-----------|-----------------------|---------|-------|------------|--|----------|---------|----------------------|-------|----------------------------|----------|
| 6p22.1  | 27676794 | rs4713118 |                       | 243000  | 0.608 | rs200997   |  | 480698   | ZNF323  |                      | 0.214 | Brain Temporal Cortex      | 20485568 |
| 6p22.1  | 27676794 | rs4713118 | $2.0 \times 10^{-10}$ | 303426  | 0.588 | rs149970   |  | 254616   | ZNF187  | $4.4 \times 10^{-6}$ | 0.017 | Monocyte                   | 20502693 |
| 6p22.1  | 27676794 | rs4713118 | $2.0 \times 10^{-10}$ | 314213  | 0.533 | rs149979   |  | 0        | ZKSCAN8 | $8.6 \times 10^{-6}$ | 0.178 | Muscle_Skeletal            | 23715323 |
| 6p22.1  | 27676794 | rs4713118 | $2.0 \times 10^{-10}$ | 356380  | 0.508 | rs9295755  |  | 0        | OR2B8P  | $1.3 \times 10^{-7}$ | 0.262 | Skin_Sun_Exposed_Lower_leg | 23715323 |
| 11q12.1 | 57694260 | rs687432  | $2.0 \times 10^{-9}$  | 36792   | 0.968 | rs7102929  |  | -2151688 | OR5L1   | $2.5 \times 10^{-5}$ | 0.063 | Monocyte                   | 22446964 |
| 11q12.1 | 57694260 | rs687432  | $2.0 \times 10^{-9}$  | 39828   | 0.815 | rs12804093 |  | -414908  | UBE2L6  | 0.00040              | 0.044 | B-Cell                     | 22446964 |
| 16p12.3 | 19279464 | rs11343   | $1.0 \times 10^{-8}$  | -652    | 0.774 | rs12445464 |  | 258      | SYT17   | $9.5 \times 10^{-9}$ | 0.025 | Monocyte                   | 20502693 |
| 16p12.3 | 19279464 | rs11343   | $1.0 \times 10^{-8}$  | -84     | 1.000 | rs1051547  |  | -1121    | SYT17   | $1.3 \times 10^{-5}$ | 0.066 | B-Cell                     | 22446964 |
| 5q12.1  | 60260543 | rs162227  | $1.4 \times 10^{-8}$  | -171107 | 0.733 | rs921898   |  | 0        | ELOVL7  | $3.8 \times 10^{-6}$ | 0.236 | Nerve Tibial               | 23715323 |

## Nearby Clinical Variants

| source  | region | our.name  | our.pval              | dist  | rsqr  | assay.name | gene  | phenotype                                     | accession                   |
|---------|--------|-----------|-----------------------|-------|-------|------------|-------|---|-----------------------------|
| clinvar | 1q22   | rs2230288 | $4.7 \times 10^{-17}$ | 0     | 1.000 | rs2230288  | GBA   | Parkinson's disease                           | SNOMED CT49049000           |
| clinvar | 1q22   | rs2230288 | $4.7 \times 10^{-17}$ | 0     | 1.000 | rs2230288  | GBA   | Lewy body dementia                            | Office of Rare Diseases3243 |
| clinvar | 1q22   | rs2230288 | $4.7 \times 10^{-17}$ | 0     | 1.000 | rs2230288  | GBA   | Gaucher disease, perinatal lethal             | NCBI curation               |
| clinvar | 1q22   | rs2230288 | $4.7 \times 10^{-17}$ | 0     | 1.000 | rs2230288  | GBA   | Gaucher's disease, type 1                     | SNOMED CT62201009           |
| clinvar | 1q22   | rs2230288 | $4.7 \times 10^{-17}$ | 0     | 1.000 | rs2230288  | GBA   | Subacute neuronopathic Gaucher's disease      | SNOMED CT5963005            |
| clinvar | 1q22   | rs2230288 | $4.7 \times 10^{-17}$ | 0     | 1.000 | rs2230288  | GBA   | Acute neuronopathic Gaucher's disease         | SNOMED CT12246008           |
| clinvar | 1q22   | rs2230288 | $4.7 \times 10^{-17}$ | 0     | 1.000 | rs2230288  | GBA   | Gaucher disease type 3C                       | NCBI curation               |
| omim    | 1q22   | rs2230288 | $4.7 \times 10^{-17}$ | 0     | 1.000 | rs2230288  | GBA   |   |                             |
| clinvar | 3q27.1 | rs9858038 | $3.0 \times 10^{-14}$ | 24918 | 0.708 | rs2270968  | MCCC1 | 3 Methylcrotonyl-CoA carboxylase 1 deficiency | Office of Rare Diseases5665 |

## Shared Associations with Other 23andMe Phenotypes

| region  | position  | our.name    | our.pval              | dist   | rsqr  | assay.name  | phenotype                          | pvalue                | gene.context            | match |
|---------|-----------|-------------|-----------------------|--------|-------|-------------|------------------------------------|-----------------------|-------------------------|-------|
| 1p36.23 | 8445360   | rs3765971   | $1.2 \times 10^{-6}$  | 14887  | 0.872 | rs138050288 | any_animal_allergy                 | $1.6 \times 10^{-10}$ | [RERE]                  |       |
| 1p36.23 | 8445360   | rs3765971   | $1.2 \times 10^{-6}$  | 14887  | 0.872 | rs138050288 | any_plant_allergy                  | $5.6 \times 10^{-12}$ | [RERE]                  |       |
| 1p36.23 | 8445360   | rs3765971   | $1.2 \times 10^{-6}$  | 14887  | 0.872 | rs138050288 | dust_mites_allergy                 | $1.2 \times 10^{-8}$  | [RERE]                  |       |
| 1p36.11 | 25806112  | rs35065124  | $1.4 \times 10^{-5}$  | -75195 | 0.527 | rs642255    | hair_curl                          | $5.9 \times 10^{-7}$  | [RHCE]                  | TRUE  |
| 1p36.11 | 25736369  | rs111941366 | $1.1 \times 10^{-5}$  | 0      | 1.000 | rs111941366 | iqb.blood_donation_frequency       | $2.1 \times 10^{-20}$ | [RHCE]                  | TRUE  |
| 1p36.11 | 25736369  | rs111941366 | $1.1 \times 10^{-5}$  | 17656  | 0.941 | rs909832    | iqb.bruise_easily                  | $5.7 \times 10^{-14}$ | RHCE-[]-TMEM57          | FALSE |
| 1p36.11 | 25736369  | rs111941366 | $1.1 \times 10^{-5}$  | 89580  | 0.566 | rs200391466 | high_cholesterol_broad             | $2.8 \times 10^{-7}$  | [TMEM57]                |       |
| 3p21.31 | 48327563  | rs34618864  | $7.3 \times 10^{-5}$  | 19392  | 0.787 | rs34644650  | blood_pressure_meds                | $4.0 \times 10^{-9}$  | NME6-[]-SPINK8          |       |
| 3p21.31 | 49013383  | rs6768433   | $2.6 \times 10^{-7}$  | -28876 | 0.831 | rs200143221 | iqb.carry_a_tune                   | $4.5 \times 10^{-8}$  | [ARIH2]                 |       |
| 3p21.31 | 49013383  | rs6768433   | $2.6 \times 10^{-7}$  | -28876 | 0.831 | rs200143221 | motionsick_combined                | $3.8 \times 10^{-8}$  | [ARIH2]                 |       |
| 3p21.1  | 53094194  | rs11712910  | $4.4 \times 10^{-5}$  | -59150 | 0.874 | rs2581792   | hemorrhoids                        | $6.4 \times 10^{-9}$  | [SFBMT1]                | FALSE |
| 3q13.32 | 118291124 | rs6438460   | $1.6 \times 10^{-5}$  | 37631  | 0.509 | rs6799936   | motionsick_combined                | $5.6 \times 10^{-9}$  | []-IGSF11               | FALSE |
| 3q25.1  | 151111173 | rs12498042  | $7.1 \times 10^{-7}$  | 19168  | 0.653 | rs10935845  | iqb.sound_sleeper                  | $3.6 \times 10^{-7}$  | [MED12L]                | TRUE  |
| 3q27.1  | 182811490 | rs3772722   | $1.6 \times 10^{-9}$  | 38074  | 0.546 | rs3772715   | iqb.fruit_juice_frequency          | $6.6 \times 10^{-7}$  | [LAMP3]                 | FALSE |
| 4p16.3  | 976598    | rs17783233  | $3.3 \times 10^{-12}$ | -24651 | 0.534 | rs34311866  | pd_family_history                  | $7.1 \times 10^{-7}$  | [TMEM175]               | TRUE  |
| 4p16.3  | 1841330   | rs115564335 | $3.3 \times 10^{-5}$  | -47050 | 0.642 | rs10012764  | linebacker                         | $3.6 \times 10^{-7}$  | TACC3-[]-JGFR3          | TRUE  |
| 4p15.31 | 17853921  | rs16895888  | $1.3 \times 10^{-5}$  | 20168  | 0.784 | rs7671110   | height                             | $2.1 \times 10^{-54}$ | NCAPG-[]-LCORL          | TRUE  |
| 4p15.31 | 17853921  | rs16895888  | $1.3 \times 10^{-5}$  | 77397  | 0.775 | rs13146142  | shoe_size_normalized               | $1.9 \times 10^{-17}$ | [LCORL]                 | TRUE  |
| 4p15.31 | 17940197  | rs71603391  | $6.4 \times 10^{-5}$  | 89639  | 0.829 | rs112093994 | weight                             | $6.3 \times 10^{-10}$ | LCORL-[]                |       |
| 4q21.1  | 77336203  | rs13104972  | $2.8 \times 10^{-7}$  | 17827  | 0.568 | rs59136511  | gallstones_or_gall_bladder_removal | $1.6 \times 10^{-7}$  | CCDC158-[]-SHROOM3      | TRUE  |
| 4q22.1  | 90626111  | rs356182    | $1.2 \times 10^{-56}$ | 15229  | 0.520 | rs356220    | any_neurological                   | $5.4 \times 10^{-12}$ | GPRIN3-[]-[]-SNCA       | TRUE  |
| 4q22.1  | 90626111  | rs356182    | $1.2 \times 10^{-56}$ | 15229  | 0.520 | rs356220    | pd_family_history                  | $4.6 \times 10^{-9}$  | GPRIN3-[]-[]-SNCA       | TRUE  |
| 4q22.1  | 90711770  | rs2737029   | $2.4 \times 10^{-38}$ | 32478  | 0.508 | rs2619344   | updrs_constipation                 | $9.0 \times 10^{-7}$  | [SNCA]                  | FALSE |
| 4q22.1  | 90781538  | rs10025824  | $1.7 \times 10^{-5}$  | 34485  | 0.576 | rs1442140   | panic                              | $6.0 \times 10^{-7}$  | SNCA-[]-MMRN1           | FALSE |
| 4q24    | 103188709 | rs13107325  | $8.2 \times 10^{-7}$  | -41821 | 0.820 | rs35225200  | iqb.rotator_cuff_tear              | $4.3 \times 10^{-7}$  | BANK1-[]-[]-SLC39A8     | FALSE |
| 4q24    | 103188709 | rs13107325  | $8.2 \times 10^{-7}$  | 0      | 1.000 | rs13107325  | iqb.excess_hair                    | $3.8 \times 10^{-8}$  | [SLC39A8]               | FALSE |
| 4q24    | 103188709 | rs13107325  | $8.2 \times 10^{-7}$  | 0      | 1.000 | rs13107325  | any_animal_allergy                 | $2.3 \times 10^{-7}$  | [SLC39A8]               | FALSE |
| 4q24    | 103188709 | rs13107325  | $8.2 \times 10^{-7}$  | 0      | 1.000 | rs13107325  | bmi                                | $7.7 \times 10^{-30}$ | [SLC39A8]               | FALSE |
| 4q24    | 103188709 | rs13107325  | $8.2 \times 10^{-7}$  | 0      | 1.000 | rs13107325  | high_plasma_glucose                | $9.0 \times 10^{-7}$  | [SLC39A8]               | FALSE |
| 4q24    | 103188709 | rs13107325  | $8.2 \times 10^{-7}$  | 0      | 1.000 | rs13107325  | iqb.low_hdl                        | $3.6 \times 10^{-14}$ | [SLC39A8]               | FALSE |
| 4q24    | 103188709 | rs13107325  | $8.2 \times 10^{-7}$  | 0      | 1.000 | rs13107325  | obesity                            | $3.0 \times 10^{-9}$  | [SLC39A8]               | FALSE |
| 4q24    | 103188709 | rs13107325  | $8.2 \times 10^{-7}$  | 0      | 1.000 | rs13107325  | weight                             | $2.3 \times 10^{-17}$ | [SLC39A8]               | FALSE |
| 4q24    | 103188709 | rs13107325  | $8.2 \times 10^{-7}$  | 0      | 1.000 | rs13107325  | height                             | $6.0 \times 10^{-11}$ | [SLC39A8]               | TRUE  |
| 4q24    | 103188709 | rs13107325  | $8.2 \times 10^{-7}$  | 0      | 1.000 | rs13107325  | iqb.arthritis_any                  | $2.2 \times 10^{-7}$  | [SLC39A8]               | FALSE |
| 4q24    | 103188709 | rs13107325  | $8.2 \times 10^{-7}$  | 0      | 1.000 | rs13107325  | osteoarthritis_broad               | $1.3 \times 10^{-8}$  | [SLC39A8]               | FALSE |
| 4q24    | 103188709 | rs13107325  | $8.2 \times 10^{-7}$  | 9373   | 0.931 | rs13135092  | iqb.stretch_marks                  | $3.0 \times 10^{-10}$ | [SLC39A8]               | FALSE |
| 4q24    | 103188709 | rs13107325  | $8.2 \times 10^{-7}$  | 9373   | 0.931 | rs13135092  | stretch_marks_any                  | $4.1 \times 10^{-9}$  | [SLC39A8]               | FALSE |
| 5q14.3  | 87721684  | rs35182117  | $3.7 \times 10^{-6}$  | -75557 | 0.666 | rs71639107  | any_psychiatric                    | $2.1 \times 10^{-9}$  | TMEM161B-[]-[]-MEF2C    |       |
| 5q14.3  | 87721684  | rs35182117  | $3.7 \times 10^{-6}$  | -75557 | 0.666 | rs71639107  | depression                         | $6.1 \times 10^{-9}$  | TMEM161B-[]-[]-MEF2C    |       |
| 5q14.3  | 87721684  | rs35182117  | $3.7 \times 10^{-6}$  | -71805 | 0.641 | rs113828108 | morning_person                     | $2.4 \times 10^{-10}$ | TMEM161B-[]-[]-MEF2C    |       |
| 5q31.1  | 134335383 | rs299366    | $4.2 \times 10^{-5}$  | 37302  | 0.606 | rs31198     | height                             | $3.4 \times 10^{-17}$ | [LOC100996485]          | TRUE  |
| 6p22.1  | 27676794  | rs4713118   | $2.0 \times 10^{-10}$ | 17245  | 0.839 | rs67869107  | any_immune                         | $5.8 \times 10^{-7}$  | ZNF184-[]-[]-HIST1H2BL  |       |
| 6p22.1  | 27850873  | rs494887    | $5.7 \times 10^{-6}$  | -75199 | 0.511 | rs200484    | glasses_any                        | $7.4 \times 10^{-8}$  | [HIST1H2BL]             | TRUE  |
| 6p22.1  | 27690453  | rs9468206   | $9.7 \times 10^{-9}$  | 97609  | 0.869 | rs200502    | urine_control                      | $2.1 \times 10^{-7}$  | [HIST1H2BM-[]-HIST1H4]  | FALSE |
| 6p22.1  | 27968350  | rs78094982  | $6.6 \times 10^{-7}$  | -57135 | 0.715 | rs151313790 | any_neurological                   | $7.3 \times 10^{-7}$  | OR2B2-[]-[]-OR2B6       | TRUE  |
| 6p22.1  | 28595107  | rs11970409  | $4.5 \times 10^{-6}$  | -16821 | 0.651 | rs34388845  | bmi                                | $6.8 \times 10^{-10}$ | SCAND3-[]-[]-TRIM27     | TRUE  |
| 6p21.33 | 31184877  | rs9263960   | $1.2 \times 10^{-5}$  | 43533  | 0.601 | rs1986997   | weight                             | $6.3 \times 10^{-15}$ | HCG27-[]-[]-HLA-C       | TRUE  |
| 6p21.32 | 32398853  | rs377537858 | $2.4 \times 10^{-6}$  | 10203  | 0.596 | rs9268652   | hepatitis_b                        | $3.1 \times 10^{-9}$  | [HLA-DRA]               |       |
| 6p21.32 | 32427748  | rs9268831   | $1.0 \times 10^{-5}$  | -4511  | 0.522 | rs9268803   | iqb.poison_oak                     | $5.3 \times 10^{-8}$  | HLA-DRA-[]-[]-HLA-DRB5  | FALSE |
| 6p21.32 | 32406843  | rs3763323   | $1.8 \times 10^{-5}$  | 22739  | 0.853 | rs9268851   | multiple_sclerosis                 | $5.3 \times 10^{-85}$ | HLA-DRA-[]-[]-HLA-DRB5  | TRUE  |
| 6p21.32 | 32572755  | rs9270936   | $7.3 \times 10^{-6}$  | -6178  | 0.773 | rs4329147   | iqb.childhood_ear_infections       | $9.5 \times 10^{-12}$ | HLA-DRB5-[]-[]-HLA-DQA1 | FALSE |
| 6p21.32 | 32572904  | rs574170    | $2.8 \times 10^{-9}$  | -3742  | 0.549 | rs28752504  | depression                         | $2.8 \times 10^{-7}$  | HLA-DRB5-[]-[]-HLA-DQA1 | FALSE |



|          |           |                   |                       |        |       |                   |                                     |                        |  |       |
|----------|-----------|-------------------|-----------------------|--------|-------|-------------------|-------------------------------------|------------------------|--|-------|
| 6p21.32  | 32571961  | rs17425622        | 3.8×10 <sup>-17</sup> | 1304   | 0.523 | rs601148          | iqb.took_meds_prednisone_or_steroid | 6.7×10 <sup>-8</sup>   | HLA-DRB5--[]--<br>HLA-DQA1<br>HLA-DRB5--[]-- | FALSE |
| 6p21.32  | 32582577  | rs3104415         | 4.2×10 <sup>-6</sup>  | 35     | 0.927 | rs3104414         | rheumatoid_arthritis                | 2.8×10 <sup>-39</sup>  | HLA-DRB5--[]--<br>HLA-DQA1<br>HLA-DRB5--[]-- | FALSE |
| 6p21.32  | 32583603  | rs28752515        | 2.0×10 <sup>-5</sup>  | 716    | 0.608 | rs28383241        | heart_metabolic_disease             | 2.2×10 <sup>-8</sup>   | HLA-DRB5--[]--<br>HLA-DQA1<br>HLA-DRB5--[]-- | FALSE |
| 6p21.32  | 32583813  | rs1846190         | 7.6×10 <sup>-8</sup>  | 3200   | 0.681 | rs557011          | vitiligo                            | 6.2×10 <sup>-26</sup>  | HLA-DRB5--[]--<br>HLA-DQA1<br>HLA-DRB5--[]-- | FALSE |
| 6p21.32  | 32587416  | rs9271386         | 1.6×10 <sup>-5</sup>  | -7     | 0.752 | rs9271385         | juvenile_asthma                     | 3.2×10 <sup>-11</sup>  | HLA-DRB5--[]--<br>HLA-DQA1<br>HLA-DRB5--[]-- | FALSE |
| 6p21.32  | 32591332  | rs9271600         | 2.7×10 <sup>-9</sup>  | -2909  | 0.510 | rs9271455         | any_psychiatric                     | 7.3×10 <sup>-8</sup>   | HLA-DQA1<br>HLA-DRB5--[]--                   | FALSE |
| 6p21.32  | 32591332  | rs9271600         | 2.7×10 <sup>-9</sup>  | -2778  | 0.517 | rs9271465         | iqb.bronchitis_frequency            | 1.6×10 <sup>-15</sup>  | HLA-DQA1<br>HLA-DRB5--[]--                   | FALSE |
| 6p21.32  | 32571961  | rs17425622        | 3.8×10 <sup>-17</sup> | 17454  | 0.658 | rs9271509         | blood_sugar_meds                    | 2.1×10 <sup>-14</sup>  | HLA-DQA1<br>HLA-DRB5--[]--                   | FALSE |
| 6p21.32  | 32591332  | rs9271600         | 2.7×10 <sup>-9</sup>  | 5      | 0.989 | rs9271601         | any_skin_cancer                     | 7.6×10 <sup>-10</sup>  | HLA-DRB5--[]--<br>HLA-DQA1<br>HLA-DRB5--[]-- | TRUE  |
| 6p21.32  | 32587553  | rs9271404         | 3.1×10 <sup>-9</sup>  | 3830   | 0.536 | rs9271602         | milk_allergy                        | 7.0×10 <sup>-7</sup>   | HLA-DQA1<br>HLA-DRB5--[]--                   | FALSE |
| 6p21.32  | 32572755  | rs9270936         | 7.3×10 <sup>-6</sup>  | 19167  | 0.617 | rs4959108         | high_cholesterol_broad              | 8.4×10 <sup>-18</sup>  | HLA-DQA1<br>HLA-DRB5--[]--                   | FALSE |
| 6p21.32  | 32602872  | rs9272245         | 4.7×10 <sup>-5</sup>  | -3801  | 0.703 | rs9272050         | plantar_warts                       | 9.6×10 <sup>-31</sup>  | HLA-DQA1<br>HLA-DRB5--[]--                   | FALSE |
| 6p21.32  | 32602872  | rs9272245         | 4.7×10 <sup>-5</sup>  | -476   | 0.958 | rs9272226         | iqb.plantar_warts                   | 1.8×10 <sup>-40</sup>  | HLA-DQA1<br>HLA-DRB5--[]--                   | FALSE |
| 6p21.32  | 32566149  | rs9270664         | 1.3×10 <sup>-6</sup>  | 37338  | 0.513 | rs3104367         | allergic_asthma                     | 3.4×10 <sup>-13</sup>  | HLA-DQA1<br>HLA-DRB5--[]--                   | FALSE |
| 6p21.32  | 32566149  | rs9270664         | 1.3×10 <sup>-6</sup>  | 37338  | 0.513 | rs3104367         | any_asthma                          | 1.0×10 <sup>-40</sup>  | HLA-DQA1<br>HLA-DRB5--[]--                   | FALSE |
| 6p21.32  | 32566149  | rs9270664         | 1.3×10 <sup>-6</sup>  | 37338  | 0.513 | rs3104367         | iqb.took_meds_asthma_or_lung        | 5.3×10 <sup>-39</sup>  | HLA-DQA1<br>HLA-DRB5--[]--                   | FALSE |
| 6p21.32  | 32571961  | rs17425622        | 3.8×10 <sup>-17</sup> | 31837  | 0.629 | rs1391371         | iqb.acne_as_teenager                | 1.2×10 <sup>-9</sup>   | HLA-DQA1<br>HLA-DQA1--[]HLA-                 | TRUE  |
| 6p21.32  | 32626021  | rs6905775         | 8.7×10 <sup>-6</sup>  | 251    | 0.534 | rs9273363         | t1d                                 | 1.1×10 <sup>-201</sup> | DQB1<br>HLA-DQA1--[]HLA-                     | FALSE |
| 6p21.32  | 32569659  | rs9270810         | 3.6×10 <sup>-7</sup>  | 56833  | 0.530 | rs9273370         | hypothyroidism                      | 4.5×10 <sup>-34</sup>  | DQB1   | FALSE |
| 6p21.32  | 32636757  | rs3134998         | 5.9×10 <sup>-5</sup>  | -8790  | 0.595 | rs1063346         | juvenile_allergic_asthma            | 6.3×10 <sup>-13</sup>  | [HLA-DQB1]                                   | FALSE |
| 6p21.32  | 32663986  | rs6457619         | 4.1×10 <sup>-5</sup>  | -10260 | 0.520 | rs9275170         | acid_reflux_meds                    | 6.8×10 <sup>-10</sup>  | HLA-DQB1--[]--<br>HLA-DQA2<br>HLA-DQB1--[]-- | FALSE |
| 6p21.32  | 32667638  | rs9275348         | 4.2×10 <sup>-9</sup>  | 462    | 0.508 | rs2647045         | any_non_melanoma_skin_cancer        | 4.9×10 <sup>-10</sup>  | HLA-DQA2<br>HLA-DQB1--[]--                   | TRUE  |
| 6p21.32  | 32658933  | rs9275218         | 1.1×10 <sup>-5</sup>  | 14821  | 0.539 | rs28772279        | lightheaded                         | 3.7×10 <sup>-7</sup>   | HLA-DQA2<br>HLA-DQB1--[]--                   | FALSE |
| 6p21.32  | 32587117  | rs28383313        | 9.7×10 <sup>-9</sup>  | 90795  | 0.505 | rs9275563         | iqb.oral_corticosteroid_ever        | 3.5×10 <sup>-7</sup>   | HLA-DQA2<br>HLA-DQB1--[]--                   | FALSE |
| 6p21.32  | 32587117  | rs28383313        | 9.7×10 <sup>-9</sup>  | 90795  | 0.505 | rs9275563         | iqb.pneumonia_frequency             | 1.4×10 <sup>-11</sup>  | HLA-DQA2<br>HLA-DQB1--[]--                   | FALSE |
| 6p21.32  | 32684935  | rs9275642         | 6.5×10 <sup>-5</sup>  | 0      | 1.000 | rs9275642         | basal_cell_carcinoma                | 7.7×10 <sup>-11</sup>  | HLA-DQA2                                     | TRUE  |
| 6q21     | 112240786 | rs12661198        | 9.3×10 <sup>-5</sup>  | -55022 | 0.618 | rs139971631       | mpn_or_v617f                        | 2.0×10 <sup>-7</sup>   | FYI--[]--WISP3                               | FALSE |
| 7p15.3   | 23586607  | rs111254487       | 7.2×10 <sup>-5</sup>  | -92994 | 0.665 | rs112316332       | iqb.math_class                      | 4.4×10 <sup>-7</sup>   | [IGF2BP3]                                    | FALSE |
| 9p22.2   | 16718436  | rs10756788        | 2.9×10 <sup>-6</sup>  | 17727  | 0.926 | rs13284478        | hair_curl                           | 5.0×10 <sup>-8</sup>   | [BNC2]<br>MUC19--[]--                        | TRUE  |
| 12q12    | 40824058  | rs149557347       | 7.7×10 <sup>-7</sup>  | -183   | 0.932 | rs189621487       | iqb.rotator_cuff_tear               | 2.9×10 <sup>-7</sup>   | MUC19  | FALSE |
| 12q12    | 41870804  | rs187043470       | 7.6×10 <sup>-27</sup> | 0      | 1.000 | rs187043470       | any_neurological                    | 8.4×10 <sup>-8</sup>   | [PDZRN4]                                     | TRUE  |
| 12q12    | 41870804  | rs187043470       | 7.6×10 <sup>-27</sup> | 0      | 1.000 | rs187043470       | pd_family_history                   | 6.7×10 <sup>-16</sup>  | [PDZRN4]                                     | TRUE  |
| 16p11.2  | 28620572  | rs750155          | 2.1×10 <sup>-5</sup>  | -81186 | 0.543 | rs189278832       | iqb.stretch_marks                   | 7.3×10 <sup>-11</sup>  | [NPIPL1]                                     | FALSE |
| 16p11.2  | 28620572  | rs750155          | 2.1×10 <sup>-5</sup>  | -81186 | 0.543 | rs189278832       | stretch_marks_any                   | 7.8×10 <sup>-11</sup>  | [NPIPL1]                                     | FALSE |
| 16p11.2  | 28564146  | rs11364090        | 2.1×10 <sup>-5</sup>  | -18559 | 0.558 | rs231976          | mosquito_severe                     | 1.7×10 <sup>-7</sup>   | [NPIPL1]                                     | FALSE |
| 16p11.2  | 28620572  | rs750155          | 2.1×10 <sup>-5</sup>  | -13040 | 0.512 | rs4115668         | any_allergy                         | 5.9×10 <sup>-8</sup>   | [NPIPL1,SULT1A2]                             | TRUE  |
| 16p11.2  | 28620572  | rs750155          | 2.1×10 <sup>-5</sup>  | -13040 | 0.512 | rs4115668         | any_plant_allergy                   | 3.8×10 <sup>-8</sup>   | [NPIPL1,SULT1A2]                             | TRUE  |
| 16p11.2  | 28620572  | rs750155          | 2.1×10 <sup>-5</sup>  | 0      | 1.000 | rs750155          | seasonal_allergies_broad            | 6.1×10 <sup>-8</sup>   | [NPIPL1,SULT1A1]                             | TRUE  |
| 16p11.2  | 28927818  | rs12919058        | 2.3×10 <sup>-5</sup>  | -61902 | 0.675 | rs7187333         | bmi                                 | 2.6×10 <sup>-17</sup>  | [NPIPL1]                                     | FALSE |
| 16p11.2  | 28927818  | rs12919058        | 2.3×10 <sup>-5</sup>  | -56618 | 0.671 | rs199766562       | weight                              | 9.6×10 <sup>-21</sup>  | [NPIPL1]                                     | FALSE |
| 16p11.2  | 28927818  | rs12919058        | 2.3×10 <sup>-5</sup>  | -29025 | 0.760 | rs6565259         | iqb.ingrown_hair_bumps              | 1.4×10 <sup>-7</sup>   | [NPIPL1,ATP2A1]                              | FALSE |
| 16p11.2  | 28927818  | rs12919058        | 2.3×10 <sup>-5</sup>  | -8477  | 0.814 | rs7206214         | shoe_size_normalized                | 5.5×10 <sup>-11</sup>  | [NPIPL1,RABEP2]                              | FALSE |
| 16p11.2  | 30914626  | rs1046276         | 5.6×10 <sup>-6</sup>  | -85954 | 0.602 | rs7192475         | number_of_cavities                  | 6.3×10 <sup>-7</sup>   | ZNF629--[]--BCL7C                            | FALSE |
| 16p11.2  | 30980130  | rs4346218         | 2.6×10 <sup>-7</sup>  | -93487 | 0.599 | rs897984          | iqb.took_meds_prednisone_or_steroid | 2.2×10 <sup>-7</sup>   | ZNF629--[]--BCL7C                            | FALSE |
| 16p11.2  | 31037020  | rs11150604        | 7.1×10 <sup>-6</sup>  | -85454 | 0.795 | rs13336347        | high_blood_pressure                 | 3.4×10 <sup>-8</sup>   | [FBXL19]                                     | FALSE |
| 16p11.2  | 31084394  | rs11281612        | 7.0×10 <sup>-6</sup>  | -90571 | 0.610 | rs11150602        | heart_metabolic_disease             | 6.2×10 <sup>-11</sup>  | [SETD1A]                                     | FALSE |
| 16p11.2  | 31084394  | rs11281612        | 7.0×10 <sup>-6</sup>  | -70215 | 0.837 | rs9926533         | heart_metabolic_disease_burden      | 5.6×10 <sup>-11</sup>  | [STX1B]                                      | FALSE |
| 16p11.2  | 31084394  | rs11281612        | 7.0×10 <sup>-6</sup>  | -36315 | 0.884 | rs10871454        | warfarin_dosage                     | 1.7×10 <sup>-28</sup>  | [STX4]                                       | FALSE |
| 16p11.2  | 31084394  | rs11281612        | 7.0×10 <sup>-6</sup>  | 3953   | 0.879 | rs749671          | warfarin_sensitivity                | 2.3×10 <sup>-9</sup>   | [ZNF646]                                     | FALSE |
| 16q12.2  | 52635000  | rs12922061        | 5.9×10 <sup>-6</sup>  | -54500 | 0.619 | rs3095602         | breast_cancer                       | 1.6×10 <sup>-19</sup>  | [TOX3]                                       | TRUE  |
| 16q12.2  | 52635000  | rs12922061        | 5.9×10 <sup>-6</sup>  | -54500 | 0.619 | rs3095602         | breast_or_ovarian_cancer            | 1.9×10 <sup>-17</sup>  | [TOX3]                                       | TRUE  |
| 16q12.2  | 52644895  | rs3104798         | 2.0×10 <sup>-5</sup>  | -3709  | 0.937 | rs3104793         | iqb.restless_leg_syndrome           | 3.3×10 <sup>-15</sup>  | TOX3--[]--CHD9                               | FALSE |
| 17p11.2  | 17715101  | rs11868035        | 1.6×10 <sup>-5</sup>  | -18346 | 0.936 | rs11649804        | myringotomy                         | 2.9×10 <sup>-7</sup>   | [RAI1]                                       | TRUE  |
| 17p11.2  | 17853454  | rs8077530         | 3.5×10 <sup>-5</sup>  | -91207 | 0.823 | rs11658309        | iqb.strep_throat                    | 4.7×10 <sup>-7</sup>   | [TOM1L2]                                     | TRUE  |
| 17q21.2  | 40698158  | rs601999          | 6.7×10 <sup>-8</sup>  | 0      | 1.000 | rs601999          | body_type                           | 3.5×10 <sup>-7</sup>   | NAGLU-[]HSD17B1<br>PLEKHM1-[]-               | TRUE  |
| 17q21.31 | 43515927  | rs35354512        | 1.7×10 <sup>-28</sup> | 56969  | 0.591 | rs5026246         | personality_assertiveness           | 3.1×10 <sup>-8</sup>   | ARL17A<br>LRRC37A3--[]--                     | TRUE  |
| 17q21.31 | 43705601  | rs365825          | 4.2×10 <sup>-32</sup> | 88227  | 0.985 | rs4479288         | nearsightedness                     | 3.7×10 <sup>-7</sup>   | CRHR1<br>LRRC37A3--[]--                      | TRUE  |
| 17q21.31 | 43807911  | rs192006062       | 5.0×10 <sup>-29</sup> | 26760  | 0.501 | rs71363552        | extraversion_combined               | 9.7×10 <sup>-8</sup>   | CRHR1<br>LRRC37A3--[]--                      | TRUE  |
| 17q21.31 | 43807911  | rs192006062       | 5.0×10 <sup>-29</sup> | 26760  | 0.501 | rs71363552        | iqb.talk_versus_listen              | 5.2×10 <sup>-10</sup>  | CRHR1<br>LRRC37A3--[]--                      | FALSE |
| 17q21.31 | 43849572  | 17:43849572:AT_A  | 4.8×10 <sup>-30</sup> | 0      | 1.000 | 17:43849572:AT_A  | unibrow                             | 2.9×10 <sup>-32</sup>  | CRHR1<br>LRRC37A3--[]--                      | FALSE |
| 17q21.31 | 43924337  | rs12373168        | 3.0×10 <sup>-31</sup> | -73405 | 0.912 | rs113518470       | beighton_hypermobility_qtl          | 4.7×10 <sup>-11</sup>  | CRHR1<br>LRRC37A3--[]--                      | TRUE  |
| 17q21.31 | 43924337  | rs12373168        | 3.0×10 <sup>-31</sup> | -67998 | 0.564 | rs117631329       | any_neurological                    | 8.0×10 <sup>-10</sup>  | CRHR1  | TRUE  |
| 17q21.31 | 43924337  | rs12373168        | 3.0×10 <sup>-31</sup> | 960    | 0.790 | rs17690703        | iqb.forward_splits                  | 1.0×10 <sup>-8</sup>   | SPPL2C[]--MAPT                               | TRUE  |
| 17q21.31 | 43879703  | rs72834575        | 9.1×10 <sup>-5</sup>  | 50997  | 0.879 | rs62054826        | iqb.shaves_legs                     | 3.0×10 <sup>-8</sup>   | SPPL2C-[]--MAPT                              | TRUE  |
| 17q21.31 | 43931855  | 17:43931855:GAGAG | 4.4×10 <sup>-22</sup> | 0      | 1.000 | 17:43931855:GAGAG | iqb.self_rated_attractiveness       | 4.7×10 <sup>-7</sup>   | SPPL2C-[]--MAPT                              | TRUE  |
| 17q21.31 | 44041771  | rs2664006         | 6.0×10 <sup>-11</sup> | -2406  | 0.928 | rs242554          | iqb.nail_biting_frequency           | 2.9×10 <sup>-7</sup>   | [MAPT]                                       | FALSE |
| 17q21.31 | 44083948  | rs8067056         | 4.9×10 <sup>-15</sup> | 0      | 1.000 | rs8067056         | can_lift_left_eyebrow               | 2.3×10 <sup>-10</sup>  | [MAPT]                                       | TRUE  |
| 17q21.31 | 44046934  | rs113925422       | 1.9×10 <sup>-31</sup> | 44952  | 0.907 | rs9891103         | lift_one_eyebrow                    | 6.3×10 <sup>-9</sup>   | [MAPT]                                       | TRUE  |
| 17q21.31 | 44046934  | rs113925422       | 1.9×10 <sup>-31</sup> | 60672  | 0.905 | rs367721874       | iqb.pee_middle_night                | 5.4×10 <sup>-10</sup>  | [KANSL1]                                     | FALSE |

|          |           |             |                       |        |       |             |                           |                       |                          |       |
|----------|-----------|-------------|-----------------------|--------|-------|-------------|---------------------------|-----------------------|--------------------------|-------|
| 17q21.31 | 44191483  | rs11653367  | $2.0 \times 10^{-7}$  | -69904 | 0.582 | rs72836318  | heart_metabolic_disease   | $8.1 \times 10^{-7}$  | [KANSL1]                 | FALSE |
| 17q21.31 | 44191483  | rs11653367  | $2.0 \times 10^{-7}$  | -69904 | 0.582 | rs72836318  | bmi                       | $6.4 \times 10^{-9}$  | [KANSL1]                 | FALSE |
| 17q21.31 | 44191483  | rs11653367  | $2.0 \times 10^{-7}$  | -69904 | 0.582 | rs72836318  | weight                    | $5.1 \times 10^{-8}$  | [KANSL1]                 | FALSE |
| 17q21.31 | 44227393  | rs2696579   | $6.5 \times 10^{-32}$ | -61315 | 0.613 | rs201408539 | male_pattern_meta         | $8.4 \times 10^{-20}$ | [KANSL1]                 |       |
| 17q21.31 | 44227393  | rs2696579   | $6.5 \times 10^{-32}$ | -44410 | 0.547 | rs78381082  | height                    | $9.2 \times 10^{-8}$  | [KANSL1]                 | TRUE  |
| 17q21.31 | 44289832  | rs2696466   | $4.0 \times 10^{-19}$ | -89640 | 0.537 | rs9908639   | iqb.cartwheel             | $3.4 \times 10^{-7}$  | [KANSL1]                 | TRUE  |
| 17q21.31 | 44227393  | rs2696579   | $6.5 \times 10^{-32}$ | 55079  | 0.599 | rs2668658   | iqb.fresh_cucumbers_yummy | $6.8 \times 10^{-9}$  | KANSL1--[ ]--<br>LRRC37A | TRUE  |
| 17q21.31 | 44227393  | rs2696579   | $6.5 \times 10^{-32}$ | 77806  | 0.765 | rs2260227   | bald_image                | $2.8 \times 10^{-13}$ | KANSL1--[ ]--<br>LRRC37A | TRUE  |
| 17q21.31 | 44699851  | rs116956554 | $1.5 \times 10^{-26}$ | -26900 | 0.681 | rs147847247 | nearsightedness           | $3.7 \times 10^{-7}$  | [NSF]                    | TRUE  |
| 17q21.31 | 44699851  | rs116956554 | $1.5 \times 10^{-26}$ | 0      | 1.000 | rs116956554 | hair_loss                 | $5.4 \times 10^{-18}$ | [NSF]                    | TRUE  |
| 17q21.31 | 44812188  | rs199447    | $2.5 \times 10^{-26}$ | -12142 | 0.661 | rs7224296   | community_standing        | $6.0 \times 10^{-8}$  | [NSF]                    | TRUE  |
| 17q21.31 | 44812188  | rs199447    | $2.5 \times 10^{-26}$ | 13868  | 0.992 | rs9896243   | vertex                    | $9.5 \times 10^{-7}$  | [NSF]                    | TRUE  |
| 17q21.31 | 44812188  | rs199447    | $2.5 \times 10^{-26}$ | 36250  | 0.767 | rs199524    | eyecolor_herc2_blue       | $1.1 \times 10^{-10}$ | [WNT3]                   | TRUE  |
| 17q24.2  | 65892280  | rs59640213  | $1.0 \times 10^{-6}$  | 55360  | 0.658 | rs12449442  | weight                    | $4.6 \times 10^{-7}$  | [BPTF]                   | FALSE |
| Xq24     | 119436726 | rs5909698   | $4.4 \times 10^{-5}$  | 2609   | 0.966 | rs5957354   | eyecolor_herc2_blue       | $2.2 \times 10^{-16}$ | [TMEM255A]               | TRUE  |
| Xq24     | 119436726 | rs5909698   | $4.4 \times 10^{-5}$  | 2609   | 0.966 | rs5957354   | eyecolor_simple           | $2.3 \times 10^{-14}$ | [TMEM255A]               | TRUE  |

## Regional Association Plots

