Table S1. FEV­1 associations that overlapped with previously reported loci with 100 Kb flanking region in the GWAS Catalog.

|  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **GWAS Catalog** | | | | |  | **SAGE II** | | | |
| **Flanking region Tag SNP** | **p** | **OR / β** | **Initial sample size** | **PMID** |  | **Variant ID** | **MAF** | **p** | **β** |
| 2.135964308.136164309 rs35337335 | 5.00E-07 | 0.02 | 48,943 European ancestry individuals. | 28166213 |  | chr2:136073416\_G/A | 0.13 | 5.24E-05 | 0.29 |
|  | 4.00E-09 | 0.13 | 19,493 European ancestry individuals with low FEV1 measurements, 9,808 European ancestry individuals with high FEV1 measurements | 26423011 |  | chr2:136073416\_G/A | 0.13 | 5.24E-05 | 0.29 |
|  |  |  |  |  | chr2:136074329\_T/G | 0.14 | 1.52E-05 | 0.30 |
|  |  |  |  |  | chr2:136074329\_T/G | 0.14 | 1.52E-05 | 0.30 |
|  |  |  |  |  | chr2:136083006\_G/T | 0.17 | 4.82E-05 | 0.26 |
|  |  |  |  |  | chr2:136083006\_G/T | 0.17 | 4.82E-05 | 0.26 |
| 3.73713464.73913465 rs586936 | 3.00E-06 | 0.01 | 321,047 European ancestry individuals | 30804560 |  | chr3:73785307\_A/C | 0.06 | 3.77E-05 | 0.40 |
| 5.180071770.180271771 rs79898473 | 3.00E-08 | 0.01 | 321,047 European ancestry individuals | 30804560 |  | chr5:180100641\_T/G | 0.34 | 3.85E-05 | -0.21 |
| 9.98770571.98970572 rs57649467 | 6.00E-08 | 0.01 | 321,047 European ancestry individuals | 30804560 |  | chr9:98775103\_G/C | 0.01 | 4.65E-05 | 0.96 |
| 9.116371778.116571779 rs10983184 | 5.00E-14 | 0.02 | 321,047 European ancestry individuals | 30804560 |  | chr9:116551938\_A/G | 0.34 | 3.29E-05 | 0.22 |
|  |  |  |  |  |  | chr9:116552816\_T/C | 0.33 | 2.25E-05 | 0.22 |
|  |  |  |  |  |  | chr9:116564334\_G/A | 0.34 | 2.65E-05 | 0.22 |
|  |  |  |  |  |  | chr9:116567781\_T/G | 0.31 | 4.28E-05 | -0.22 |
| 16.75277546.75533123 rs11858992 | 5.00E-21 | 0.02 | 321,047 European ancestry individuals | 30804560 |  | chr16:75399722\_C/T | 0.01 | 4.11E-05 | -0.86 |
| rs3743609 | 4.00E-06 | 0.03 | 48,943 European ancestry individuals. | 28166213 |  | chr16:75399722\_C/T | 0.01 | 4.11E-05 | -0.86 |

Flanking region in the format of chromosome, chromosome start and stop in hg38.

Table S2. Conditional analysis identified rs11312747 and rs58475486 as two independent signals (p < 0.05).

|  |  |  |  |
| --- | --- | --- | --- |
|  | β | p | Conditioned variants\* (R2) |
| rs17016065 | 0.30 | 2.79E-06 | None |
| rs17016065 | 0.20 | 2.57E-03 | rs73429450 (0.10) |
| rs58475486 | 0.33 | 2.90E-05 | None |
| rs58475486 | 0.17 | 4.32E-02 | rs73429450 (0.14) |
| rs58475486 | 0.30 | 1.26E-04 | rs17016065 (< 0.01) |
| rs58475486 | 0.19 | 2.70E-02 | rs17016065 (< 0.01), rs73429450 (0.14) |

\*Also corrected for other covariates in the model used in the discovery GWAS.

Table S3. Descriptive characteristics of 251 SAGE II African American children without asthma.

|  |  |
| --- | --- |
|  | **African American** |
| **(N=251)** |
| **Age** |  |
| Mean (SD) | 16.5 (3.74) |
| Median [25%, 75%] | 17.5 [13.94, 19.50] |
| **Sex** |  |
| Male | 106 (42.2%) |
| Female | 145 (57.8%) |
| **Height (m)** |  |
| Mean (SD) | 1.63 (0.130) |
| Median [25%, 75%] | 1.64 [1.56, 1.71] |
| **African ancestry** |  |
| Mean (SD) | 0.798 (0.111) |
| Median [25%, 75%] | 0.820 [0.775, 0.868] |
| **Smoking status** |  |
| Never | 215 (85.7%) |
| Past | 35 (13.9%) |
| Current | 1 (0.4%) |
| **Number of smokers in family** |  |
| 0 | 74 (29.5%) |
| 1 | 36 (14.3%) |
| 2 | 15 (6.0%) |
| 3+ | 4 (1.6%) |
| Missing | 122 (48.6%) |
| **SO2 first year exposure (ppb)** |  |
| Mean (SD) | 1.53 (0.505) |
| Median [25%, 75%] | 1.47 [1.17, 1.77] |
| Missing | 139 (55.4%) |
| **SO2 past year exposure (ppb)** |  |
| Mean (SD) | 0.897 (0.275) |
| Median [25%, 75%] | 0.810 [0.705, 1.08] |
| Missing | 136 (54.2%) |
| **SO2 lifetime exposure (ppb)** |  |
| Mean (SD) | 1.41 (0.214) |
| Median [25%, 75%] | 1.41 [1.33, 1.45] |
| Missing | 136 (54.2%) |
| **TOPMed sequencing center and phase** | |
| phase 3, NW | 251 (100%) |

25% and 75%, 25 and 75 percentiles. SO2 exposure are hourly exposure averaged over the specified time period before spirometry testing as previously described in Neophytou *et al.* 2016. ppb, parts per billion or µg/m3.

Table S4. No association of 45 variants with FEV1.res.rnorm in SAGE II children without asthma.

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
|  | **Discovery (n=867)** | | **Control (n=251)** | |
| **rsID** | **β** | **p** | **β** | **p** |
| rs11835305 | 0.32 | 3.69E-05 | -0.05 | 4.05E-01 |
| rs17015963 | 0.32 | 3.69E-05 | -0.05 | 4.05E-01 |
| rs58475486 | 0.32 | 2.81E-05 | -0.05 | 4.05E-01 |
| rs17015979 | 0.32 | 2.81E-05 | -0.05 | 4.05E-01 |
| rs57692452 | 0.65 | 1.06E-06 | -0.11 | 3.41E-01 |
| rs112585732 | 0.62 | 3.06E-07 | -0.11 | 2.32E-01 |
| rs113837356 | 0.72 | 2.49E-06 | -0.09 | 4.42E-01 |
| rs61441836 | 0.67 | 5.46E-07 | -0.11 | 3.41E-01 |
| rs73438172 | 0.67 | 5.46E-07 | -0.11 | 3.41E-01 |
| rs1044043958 | 0.72 | 2.49E-06 | -0.09 | 4.42E-01 |
| rs73438182 | 0.38 | 9.18E-05 | -0.14 | 1.21E-01 |
| rs73438185 | 0.38 | 9.18E-05 | -0.14 | 1.21E-01 |
| rs73438188 | 0.79 | 4.42E-08 | -0.10 | 4.26E-01 |
| rs73438190 | 0.49 | 1.22E-05 | -0.13 | 2.14E-01 |
| rs73438195 | 0.49 | 1.22E-05 | -0.13 | 2.14E-01 |
| rs111857459 | 0.49 | 1.22E-05 | -0.13 | 2.14E-01 |
| rs144369986 | 0.76 | 9.44E-07 | -0.08 | 5.48E-01 |
| rs73440106 | 0.49 | 1.22E-05 | -0.13 | 2.14E-01 |
| rs73440107 | 0.79 | 4.42E-08 | -0.10 | 4.26E-01 |
| rs111453514 | 0.79 | 4.42E-08 | -0.10 | 4.26E-01 |
| rs73440112 | 0.79 | 4.42E-08 | -0.10 | 4.26E-01 |
| rs73440115 | 0.79 | 4.42E-08 | -0.10 | 4.26E-01 |
| rs11312747 | 0.36 | 8.51E-06 | -0.12 | 9.36E-02 |
| rs73440120 | 0.76 | 9.44E-07 | -0.08 | 5.48E-01 |
| rs111289668 | 0.79 | 4.42E-08 | -0.10 | 4.26E-01 |
| rs73440122 | 0.77 | 1.55E-07 | -0.10 | 4.26E-01 |
| rs73440123 | 0.77 | 1.55E-07 | -0.10 | 4.26E-01 |
| rs17016065 | 0.30 | 2.54E-06 | -0.08 | 1.59E-01 |
| rs17016066 | 0.30 | 2.54E-06 | -0.08 | 1.59E-01 |
| rs147400083 | 0.30 | 2.54E-06 | -0.08 | 1.59E-01 |
| rs866852270 | 0.30 | 2.54E-06 | -0.08 | 1.59E-01 |
| rs141293300 | 0.77 | 1.55E-07 | -0.10 | 4.26E-01 |
| rs1398303 | 0.27 | 1.12E-05 | -0.07 | 2.37E-01 |
| rs61924868 | 0.27 | 1.14E-05 | -0.07 | 2.37E-01 |
| rs73440134 | 0.77 | 1.55E-07 | -0.10 | 4.26E-01 |
| rs73429413 | 0.77 | 1.55E-07 | -0.10 | 4.26E-01 |
| rs73429415 | 0.25 | 4.84E-05 | -0.07 | 2.37E-01 |
| rs112449284 | 0.64 | 3.87E-06 | -0.03 | 7.58E-01 |
| rs111981782 | 0.79 | 4.09E-08 | -0.12 | 3.08E-01 |
| rs150942400 | 0.78 | 4.01E-08 | -0.13 | 2.50E-01 |
| rs147527487 | 0.23 | 8.14E-05 | 0.00 | 9.94E-01 |
| rs111243672 | 0.69 | 3.99E-07 | -0.10 | 3.58E-01 |
| rs73429450 | 0.80 | 9.01E-09 | -0.02 | 8.29E-01 |
| rs758775577 | 0.57 | 1.85E-06 | 0.04 | 6.74E-01 |
| rs142679473 | 0.76 | 4.62E-08 | -0.02 | 8.29E-01 |

Table S5. Region-based lung function association using all or functional common and rare variants in 1 Kb sliding windows.

|  |  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
|  |  | All variants# | | | |  | | Functional variants | | | | |
| ID | Region name | p.rc | nTotal | nRare | nCommon | |  | | p.rc | p.r | nRare | nCommon |
| 1 | 12.89612935.89613935 | \*8.84E-04 | 12 | 9 | 3 | |  | | - | - | - | - |
| 2 | 12.89602435.89603435 | \*9.02E-04 | 11 | 8 | 3 | |  | | - | - | - | - |
| 3 | 12.89007935.89008935 | \*9.04E-04 | 16 | 14 | 2 | |  | | 2.60E-01 | 3.56E-01 | 2 | 0 |
| 4 | 12.89838935.89839935 | \*2.04E-03 | 8 | 6 | 2 | |  | | \*6.38E-04 | 7.95E-02 | 1 | 1 |
| 5 | 12.89794935.89795935 | \*2.31E-03 | 7 | 5 | 2 | |  | | - | - | - | - |
| 6 | 12.89776935.89777935 | \*2.39E-03 | 10 | 6 | 4 | |  | | - | - | - | - |
| 7 | 12.89829935.89830058 | \*2.41E-03 | 2 | 1 | 1 | |  | | - | - | - | - |
| 8 | 12.89829435.89830058 | \*2.47E-03 | 6 | 5 | 1 | |  | | - | - | - | - |
| 9 | 12.88722435.88723435 | \*2.52E-03 | 16 | 13 | 3 | |  | | - | - | - | - |
| 10 | 12.89656435.89657435 | \*2.53E-03 | 13 | 9 | 4 | |  | | - | - | - | - |
| 11 | 12.89634935.89635935 | \*3.27E-03 | 4 | 3 | 1 | |  | | - | - | - | - |
| 12 | 12.88998935.88999935 | \*3.42E-03 | 17 | 10 | 7 | |  | | - | - | - | - |
| 13 | 12.89634435.89635435 | \*3.70E-03 | 7 | 6 | 1 | |  | | - | - | - | - |
| 14 | 12.89606435.89607435 | \*3.90E-03 | 19 | 12 | 7 | |  | | - | - | - | - |
| 15 | 12.88999435.89000435 | \*4.83E-03 | 17 | 9 | 8 | |  | | - | - | - | - |
| 16 | 12.89839435.89840435 | \*5.10E-03 | 9 | 6 | 3 | |  | | \*6.60E-04 | 1.12E-01 | 2 | 1 |
| 17 | 12.89007435.89008435 | \*5.11E-03 | 10 | 5 | 5 | |  | | - | - | - | - |
| 18 | 12.88771935.88772935 | \*5.16E-03 | 17 | 9 | 8 | |  | | - | - | - | - |
| 19 | 12.88425935.88426935 | \*5.23E-03 | 14 | 10 | 4 | |  | | - | - | - | - |
| 20 | 12.89652935.89653935 | \*5.33E-03 | 18 | 14 | 4 | |  | | - | - | - | - |

Bonferroni-corrected and suggestive significance of the all variant analysis were defined as \*\*p < 2.80 x 10-4 and \*p < 5.60 x 10-3 respectively. All regions that passed the suggestive threshold in the all variant analysis are reported under the all variants columns and sorted by their p-values. Additional results from analyses using functional variants only are reported under the functional variants columns. p.rc/p.r, FEV1.res.rnorm association p-value using common+rare/rare variants. Region name is in the format of chr.start.end of the hg38 coordinates of the 1 Kb window. nTotal/nRare/nCommon, number of common+rare/rare/common variants included in the region. Functional variants were defined in the Method Section. -, test was not performed because no functional variants were found in that region.

Table S6. Drop-one analysis in region 16 (12.89839435.89840435).

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
| Dropped variant | p | POS | A1 | MAF | C1 |
| None | 6.60E-04\* | - | - | - | - |
| rs1895710 | **7.74E-02** | 89839693 | A | 0.0294 | 51 |
| rs751095312 | 6.38E-04 | 89840078 | C | 0.0012 | 2 |
| rs990979778 | **1.21E-03** | 89839784 | A | 0.0006 | 1 |

\* Original p-value when no variants were dropped. An increase in p-value indicates the original p-value was driven by the dropped variant while a decrease in p-value indicates a dilution of association signal by the dropped variant.

Table S7. HUGIN association with 45 variants associated with FEV1 at p < 1 x 10-4.

|  |  |  |
| --- | --- | --- |
| SNP | IMR90 (-log10(p-value)/counts/expected) | Lung (-log10(p-value)/counts/expected) |
| rs73429450 | KITLG (17.04/134/57) | POC1B (0.77/4/2) |
| rs150942400 | KITLG (32.96/184/64) | ATP2B1 LINC00936 (0.64/3/1) |
| rs111981782 | KITLG (32.96/184/64) | ATP2B1 LINC00936 (0.64/3/1) |
| rs73438188 | KITLG (73.53/280/74) | POC1B (3.05/8/1) |
| rs73440107 | KITLG (73.53/280/74) | POC1B (3.05/8/1) |
| rs111453514 | KITLG (73.53/280/74) | POC1B (3.05/8/1) |
| rs111243672 | KITLG (32.96/184/64) | ATP2B1 LINC00936 (0.64/3/1) |
| rs73440115 | KITLG (73.53/280/74) | POC1B (3.05/8/1) |
| rs111289668 | KITLG (73.53/280/74) | POC1B (3.05/8/1) |
| rs142679473 | KITLG (17.04/134/57) | POC1B (0.77/4/2) |
| rs141293300 | KITLG (32.96/184/64) | ATP2B1 LINC00936 (0.64/3/1) |
| rs73440123 | KITLG (73.53/280/74) | POC1B (3.05/8/1) |
| rs58475486 | KITLG (38.71/236/87) | POC1B (0.93/4/1) |
| rs73440134 | KITLG (32.96/184/64) | ATP2B1 LINC00936 (0.64/3/1) |
| rs73429413 | KITLG (32.96/184/64) | ATP2B1 LINC00936 (0.64/3/1) |
| rs112585732 | KITLG (38.71/236/87) | POC1B (0.93/4/1) |
| rs73440106 | KITLG (73.53/280/74) | POC1B (3.05/8/1) |
| rs61441836 | KITLG (73.53/280/74) | POC1B (3.05/8/1) |
| rs73438172 | KITLG (73.53/280/74) | POC1B (3.05/8/1) |
| rs144369986 | KITLG (73.53/280/74) | POC1B (3.05/8/1) |
| rs73440120 | KITLG (73.53/280/74) | POC1B (3.05/8/1) |
| rs57692452 | KITLG (38.71/236/87) | POC1B (0.93/4/1) |
| rs758775577 | KITLG (17.04/134/57) | POC1B (0.77/4/2) |
| rs113837356 | KITLG (73.53/280/74) | POC1B (3.05/8/1) |
| rs17016065 | KITLG (73.53/280/74) | POC1B (3.05/8/1) |
| rs17016066 | KITLG (73.53/280/74) | POC1B (3.05/8/1) |
| rs147400083 | KITLG (32.96/184/64) | ATP2B1 LINC00936 (0.64/3/1) |
| rs866852270 | KITLG (32.96/184/64) | ATP2B1 LINC00936 (0.64/3/1) |
| rs112449284 | KITLG (32.96/184/64) | ATP2B1 LINC00936 (0.64/3/1) |
| rs11312747 | KITLG (73.53/280/74) | POC1B (3.05/8/1) |
| rs1398303 | KITLG (32.96/184/64) | ATP2B1 LINC00936 (0.64/3/1) |
| rs61924868 | KITLG (32.96/184/64) | ATP2B1 LINC00936 (0.64/3/1) |
| rs73438190 | KITLG (73.53/280/74) | POC1B (3.05/8/1) |
| rs73438195 | KITLG (73.53/280/74) | POC1B (3.05/8/1) |
| rs111857459 | KITLG (73.53/280/74) | POC1B (3.05/8/1) |
| rs73440112 | KITLG (73.53/280/74) | POC1B (3.05/8/1) |
| rs73440122 | KITLG (73.53/280/74) | POC1B (3.05/8/1) |
| rs17015979 | KITLG (38.71/236/87) | POC1B (0.93/4/1) |
| rs11835305 | KITLG (38.71/236/87) | POC1B (0.93/4/1) |
| rs17015963 | KITLG (38.71/236/87) | POC1B (0.93/4/1) |
| rs73429415 | KITLG (32.96/184/64) | ATP2B1 LINC00936 (0.64/3/1) |
| rs147527487 | KITLG (32.96/184/64) | ATP2B1 LINC00936 (0.64/3/1) |
| rs73438182 | KITLG (73.53/280/74) | POC1B (3.05/8/1) |
| rs73438185 | KITLG (73.53/280/74) | POC1B (3.05/8/1) |

-log10(p-value)/counts/expected: the statistical significance, observed and expected read counts of the long range chromatin interaction. IMR90, human fetal lung fibroblast cell line. Lung, human primary lung tissue.

Table S8. Variants that are eQTLs of *KITLG*.

|  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- |
| rsID | gene | distance | MAF | nominal p | nominal β | nominal SE | nominal threshold |
| rs17016065 | KITLG | 311486 | 0.058108 | 6.41E-06 | 0.296 | 0.064 | 9.49E-05 |
| rs17016066 | KITLG | 311726 | 0.058108 | 6.41E-06 | 0.296 | 0.064 | 9.49E-05 |
| rs1398303 | KITLG | 318523 | 0.062162 | 1.78E-05 | 0.267 | 0.061 | 9.49E-05 |
| rs61924868 | KITLG | 318781 | 0.062162 | 1.78E-05 | 0.267 | 0.061 | 9.49E-05 |
| rs73429415 | KITLG | 326516 | 0.063514 | 3.23E-05 | 0.257 | 0.061 | 9.49E-05 |

eQTL was defined as having a lower nominal p-value than the nominal threshold.

Table S9. Descriptive characteristics of replication cohorts with study subjects with asthma.

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
|  | **UK Biobank** | **SAPPHIRE** | **GCPD-A** | **Puerto Rican** | **Mexican** |
| **Blacks (n=627)** | **(n=3428)** | **(n=1464)** | **(n=1109)** | **(n=649)** |
| **Age** |  |  |  |  |  |
| Mean (SD) | 51.8 (8.0) | 32.8 (13.9) | 11.8 (3.7) | 12.7 (3.3) | 13.0 (3.4) |
| Median [25%, 75%] | 50 [45, 58] | 31.9 [19.8, 45.6] | 11.5 [8.4, 14.9] | 12.1 [10.0, 14.9] | 12.4 [10.0, 15.5] |
| **Sex** |  |  |  |  |  |
| Male | 191 (30.5%) | 1210 (35.3%) | 680 (46%) | 592 (53.4%) | 362 (55.8%) |
| Female | 436 (69.5%) | 2218 (64.7%) | 784 (54%) | 517 (46.6%) | 287 (44.2%) |
| **Height (m)** |  |  |  |  |  |
| Mean (SD) | 1.66 (0.087) | 1.67 (0.1) | 1.48 (0.187) | 1.51 (0.138) | 1.51 (0.141) |
| Median [25%, 75%] | 1.65 [1.60, 1.72] | 1.65 [1.60, 1.73] | 1.49 [1.32, 1.62] | 1.52 [1.42, 1.60] | 1.52 [1.40, 1.62] |
| **Any control medications\* in last 2 weeks** | |  |  |  |  |
| No | - | 3326 (97%) | 73 (5%) | 842 (75.9%) | 402 (61.9%) |
| Yes | - | 102 (3%) | 1391 (95%) | 267 (24.1%) | 247 (38.1%) |
| **ICS in last 2 weeks** |  |  |  |  |  |
| No | - | 696 (20.3%) | 556 (38%) | 150 (13.5%) | 109 (16.8%) |
| Yes | - | 87 (2.5%) | 908 (62%) | 172 (15.5%) | 216 (33.3%) |
| Missing | - | 2645 (77.2%) | 0 | 787 (71.0%) | 324 (49.9%) |
| **LABA in last 2 weeks** |  |  |  |  |  |
| No | - | 721 (21%) | 1246 (85.2%) | 7 (0.6%) | 3 (0.5%) |
| Yes | - | 62 (1.8%) | 218 (14.8%) | 71 (6.4%) | 58 (8.9%) |
| Missing | - | 2645 (77.2%) | 0 | 1031 (93.0%) | 588 (90.6%) |
| **Leukotriene inhibitor in last 2 weeks** | |  |  |  |  |
| No | - | 755 (22%) | 1179 (80.5%) | 111 (10.0%) | 36 (5.5%) |
| Yes | - | 28 (0.8%) | 285 (19.45%) | 152 (13.7%) | 99 (15.3%) |
| Missing | - | 2645 (77.2%) | 0 | 846 (76.3%) | 514 (79.2%) |
| **African ancestry** |  |  |  |  |  |
| Mean (SD) | 0.88 (0.16) | 0.81 (0.11) | 0.67 (0.20) | 0.22 (0.12) | 0.04 (0.03) |
| Median [25%, 75%] | 0.93 [0.78, 1] | 0.83 [0.77, 0.87] | 0.71 [0.61, 0.80] | 0.19 [0.14, 0.27] | 0.04 [0.02, 0.05] |
| **Smoking status** |  |  |  |  |  |
| Never | 416 (66.3%) | 2349 (68.5%) | 1457 (99.5%) | 1087 (98.0%) | 613 (94.5%) |
| Past | 130 (20.7%) | 316 (9.2%) | 1 (0.06%) | 14 (1.3%) | 34 (5.2%) |
| Current | 78 (12.4%) | 762 (22.2%) | 6 (0.4%) | 7 (0.6%) | 2 (0.3%) |
| Missing | 3 (0.5%) | 1 (0.0%) | 0 | 1 (0.1%) |  |
| **Number of smokers in family** |  |  |  |  |  |
| 0 | 499 (79.6%) | 2197 (64.1%) | 965 (65.9%) | 662 (59.7%) | 485 (74.7%) |
| 1 | 59 (9.4%) | 808 (23.6%) | 485 (33.4%)\*\* | 134 (12.1%) | 86 (13.3%) |
| 2 | 15 (2.4%)\* | 263 (7.7%) | 38 (3.4%) | 31 (4.8%) |
| 3+ | 145 (4.2%) | 7 (0.6%) | 1 (0.2%) |
| Missing | 54 (8.6%) | 15 (0.4%) | 14 (0.95%) | 268 (24.2%) | 46 (7.1%) |
| **TOPMed sequencing center and phase** | |  |  |  |  |
| CCDG year 1, NYGC | - | 0 (0%) | 0 (0%) | 588 (53.0%) | 0 (0%) |
| phase 1, CAAPA | - | 0 (0%) | 0 (0%) | 2 (0.2%) | 0 (0%) |
| phase 1, NYGC | - | 0 (0%) | 0 (0%) | 472 (42.6%) | 483 (74.4%) |
| phase 3, NW | - | 3428 (100%) | 1464 (100%) | 47 (4.2%) | 166 (25.6%) |

25% and 75%, 25 and 75 percentiles. \* Number indicated 2+ smokers in family as described in the UK Biobank data field 1259 (http://biobank.ndph.ox.ac.uk/showcase/field.cgi?id=1259). \*\* Number indicated 1+ smokers in family.

Table S10. Replication of association of 45 variants with FEV1 in black UK Biobank subjects and African Americans in SAPPHIRE and GCPD-A.

|  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
|  |  |  |  | **UKB (n=627)** | | | |  | **SAPPHIRE (n=3428)** | | | |  | **GCPD-A (n=1464)** | | | |
| **chr** | **pos (hg38)** | **rsID** | **Alt** | **MAF** | **p** | | **β** |  | **MAF** | **p** | | **β** |  | **MAF** | **p** | | **β** |
| 12 | 88729352 | rs11835305 | T | 0.111 |  | 0.408 | 0.040 |  | 0.102 |  | 0.507 | 0.015 |  | 0.110 |  | 0.692 | -0.014 |
| 12 | 88732007 | rs17015963 | C | 0.111 |  | 0.394 | 0.041 |  | 0.102 |  | 0.507 | 0.015 |  | 0.110 |  | 0.660 | -0.015 |
| 12 | 88749547 | rs58475486^ | T | 0.112 |  | 0.407 | 0.040 |  | 0.103 |  | 0.462 | 0.016 |  | 0.111 |  | 0.575 | -0.019 |
| 12 | 88750296 | rs17015979 | T | 0.112 |  | 0.409 | 0.040 |  | 0.103 |  | 0.462 | 0.016 |  | - |  | - | - |
| 12 | 88765415 | rs57692452 | C | 0.028 | \* | 0.020 | 0.203 |  | 0.027 | \* | 0.020 | -0.096 |  | 0.033 |  | 0.113 | -0.095 |
| 12 | 88765947 | rs112585732 | T | 0.051 |  | 0.097 | 0.114 |  | 0.033 |  | 0.067 | -0.069 |  | 0.043 |  | 0.128 | -0.079 |
| 12 | 88772911 | rs113837356 | T | 0.028 | \*\* | **0.016** | 0.213 |  | 0.021 |  | 0.054 | -0.090 |  | 0.027 |  | 0.050 | -0.128 |
| 12 | 88773595 | rs61441836 | G | 0.029 | \* | 0.019 | 0.204 |  | 0.026 | \* | 0.030 | -0.090 |  | 0.033 |  | 0.079 | -0.105 |
| 12 | 88774420 | rs73438172 | A | 0.029 | \* | 0.019 | 0.204 |  | 0.026 | \* | 0.030 | -0.090 |  | 0.028 |  | 0.107 | -0.104 |
| 12 | 88778604 | rs1044043958 | A | - |  | - | - |  | 0.021 |  | 0.058 | -0.088 |  | 0.028 |  | - | - |
| 12 | 88779036 | rs73438182 | G | 0.061 |  | 0.335 | 0.059 |  | 0.060 | \*\* | 0.012 | -0.070 |  | 0.028 | \* | 0.025 | -0.099 |
| 12 | 88779473 | rs73438185 | A | 0.061 |  | 0.335 | 0.059 |  | 0.060 | \*\* | 0.012 | -0.070 |  | 0.028 | \* | 0.025 | -0.099 |
| 12 | 88783270 | rs73438188 | A | 0.028 | \*\* | **0.016** | 0.208 |  | 0.024 |  | 0.067 | -0.081 |  | 0.028 |  | - | - |
| 12 | 88783315 | rs73438190 | C | 0.050 |  | 0.123 | 0.102 |  | 0.047 | \* | 0.028 | -0.069 |  | 0.028 |  | 0.083 | -0.082 |
| 12 | 88783668 | rs73438195 | A | 0.050 |  | 0.123 | 0.103 |  | 0.047 | \* | 0.028 | -0.069 |  | 0.028 |  | 0.083 | -0.082 |
| 12 | 88785774 | rs111857459 | T | 0.050 |  | 0.123 | 0.102 |  | 0.047 | \* | 0.028 | -0.069 |  | 0.028 |  | 0.083 | -0.082 |
| 12 | 88789109 | rs144369986 | T | - |  | - | - |  | 0.021 |  | 0.052 | -0.091 |  | 0.028 | \* | 0.043 | -0.131 |
| 12 | 88791281 | rs73440106 | G | 0.050 |  | 0.123 | 0.102 |  | 0.047 | \* | 0.028 | -0.069 |  | 0.028 |  | - | - |
| 12 | 88793808 | rs73440107 | A | 0.028 | \*\* | **0.016** | 0.209 |  | 0.024 |  | 0.067 | -0.081 |  | 0.028 |  | 0.118 | -0.096 |
| 12 | 88796826 | rs111453514 | C | 0.028 | \*\* | **0.016** | 0.208 |  | 0.024 |  | 0.067 | -0.081 |  | 0.028 |  | - | - |
| 12 | 88797371 | rs73440112 | T | 0.028 | \*\* | **0.016** | 0.208 |  | 0.024 |  | 0.067 | -0.081 |  | 0.028 |  | 0.118 | -0.096 |
| 12 | 88798068 | rs73440115 | G | 0.028 | \*\* | **0.016** | 0.209 |  | 0.024 |  | 0.066 | -0.081 |  | 0.028 |  | - | - |
| 12 | 88800273 | rs11312747 | A |  |  | - | - |  | 0.100 |  | 0.083 | -0.039 |  | 0.028 |  | 0.380 | -0.030 |
| 12 | 88801090 | rs73440120 | A | 0.027 | \*\* | **0.015** | 0.216 |  | 0.021 |  | 0.052 | -0.091 |  | 0.028 |  | - | - |
| 12 | 88801600 | rs111289668 | G | 0.028 | \*\* | **0.016** | 0.209 |  | 0.024 |  | 0.067 | -0.081 |  | 0.028 |  | 0.118 | -0.096 |
| 12 | 88802121 | rs73440122 | C | 0.028 | \*\* | **0.016** | 0.208 |  | 0.024 |  | 0.084 | -0.076 |  | 0.028 |  | 0.103 | -0.099 |
| 12 | 88803531 | rs73440123 | G | 0.028 | \*\* | **0.016** | 0.209 |  | 0.024 |  | 0.085 | -0.076 |  | 0.028 |  | 0.111 | -0.097 |
| 12 | 88804280 | rs17016065^ | G | 0.186 |  | 0.796 | 0.010 |  | 0.181 | \* | 0.047 | -0.035 |  | 0.028 |  | 0.412 | -0.022 |
| 12 | 88804520 | rs17016066 | A | 0.186 |  | 0.796 | 0.010 |  | 0.181 | \* | 0.047 | -0.035 |  | 0.028 |  | 0.412 | -0.022 |
| 12 | 88807139 | rs147400083 | T | - |  | - | - |  | 0.181 | \* | 0.042 | -0.036 |  | 0.028 |  | - | - |
| 12 | 88807149 | rs866852270 | T | - |  | - | - |  | 0.181 | \* | 0.042 | -0.036 |  | 0.028 |  | 0.412 | -0.022 |
| 12 | 88810932 | rs141293300 | C | - |  | - | - |  | 0.024 |  | 0.094 | -0.074 |  | 0.028 |  | 0.103 | -0.099 |
| 12 | 88811317 | rs1398303 | A | 0.194 |  | 0.738 | 0.012 |  | 0.193 | \* | 0.032 | -0.037 |  | 0.028 |  | 0.409 | -0.022 |
| 12 | 88811575 | rs61924868 | T | 0.194 |  | 0.738 | 0.012 |  | 0.192 | \* | 0.032 | -0.037 |  | 0.028 |  | 0.391 | -0.023 |
| 12 | 88813735 | rs73440134 | T | 0.028 | \*\* | **0.016** | 0.209 |  | 0.023 |  | 0.079 | -0.079 |  | 0.028 |  | 0.103 | -0.099 |
| 12 | 88817859 | rs73429413 | G | 0.028 | \*\* | **0.016** | 0.210 |  | 0.024 |  | 0.094 | -0.074 |  | 0.028 |  | 0.103 | -0.099 |
| 12 | 88819310 | rs73429415 | A | 0.197 |  | 0.574 | 0.020 |  | 0.195 |  | 0.057 | -0.032 |  | 0.028 |  | - | - |
| 12 | 88823778 | rs112449284 | T | 0.035 | \*\* | **0.016** | 0.188 |  | 0.034 |  | 0.160 | -0.052 |  | 0.028 |  | - | - |
| 12 | 88826878 | rs111981782 | C | 0.029 | \*\* | **0.010** | 0.221 |  | 0.027 |  | 0.106 | -0.067 |  | 0.028 |  | 0.250 | -0.069 |
| 12 | 88829116 | rs150942400 | T | 0.029 | \*\* | **0.010** | 0.219 |  | 0.027 |  | 0.125 | -0.064 |  | 0.028 |  | - | - |
| 12 | 88830592 | rs147527487 | C | 0.213 |  | 0.724 | -0.013 |  | 0.210 |  | 0.472 | -0.012 |  | 0.028 |  | 0.451 | -0.020 |
| 12 | 88839615 | rs111243672 | A | 0.031 | \* | 0.025 | 0.189 |  | 0.030 |  | 0.215 | -0.049 |  | 0.028 |  | - | - |
| 12 | 88846435 | rs73429450^ | A | 0.032 | \*\* | **0.005** | 0.232 |  | 0.027 |  | 0.098 | -0.069 |  | 0.028 |  | 0.294 | -0.062 |
| 12 | 88850005 | rs758775577 | C | - |  | - | - |  | 0.040 |  | 0.776 | -0.010 |  | 0.028 |  | 0.375 | -0.043 |
| 12 | 88863271 | rs142679473 | C | - |  | - | - |  | 0.027 |  | 0.082 | -0.072 |  | 0.028 |  | 0.305 | -0.060 |

\* , significant at p < 0.05; \*\*, significant at bonferroni-corrected p < 0.0167. The three independent signals identified in the conditional analyses are marked with ^ near the rsID.

Table S11. Replication association statistics using variants with MAF >= 0.1% in GALA II.

|  |  |  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
|  |  |  | **Discovery (n=867)** | | |  | **Puerto Ricans (n=1109)** | | |  | **Mexicans (n=649)** | | |
| **rsID** | **Pos** | **Alt** | **MAF** | **p** | **β** |  | **MAF** | **p** | **β** |  | **MAF** | **p** | **β** |
| rs11835305 | 88729352 | T | 0.1038 | 3.82E-05 | 0.324 |  | 0.0343 | 0.868 | -0.020 |  | 0.0062 | 0.843 | 0.071 |
| rs17015963 | 88732007 | C | 0.1038 | 3.82E-05 | 0.324 |  | 0.0347 | 0.691 | -0.046 |  | 0.0062 | 0.843 | 0.071 |
| rs58475486^ | 88749547 | T | 0.1050 | 2.90E-05 | 0.327 |  | 0.0343 | 0.868 | -0.020 |  | 0.0054 | 0.621 | -0.189 |
| rs17015979 | 88750296 | T | 0.1050 | 2.90E-05 | 0.327 |  | 0.0343 | 0.868 | -0.020 |  | 0.0054 | 0.621 | -0.189 |
| rs57692452 | 88765415 | C | 0.0335 | 1.16E-06 | 0.660 |  | 0.0126 | 0.865 | 0.033 |  | 0.0008 | - | - |
| rs112585732 | 88765947 | T | 0.0410 | 3.34E-07 | 0.631 |  | 0.0126 | 0.670 | 0.082 |  | 0.0015 | 0.722 | 0.253 |
| rs113837356 | 88772911 | T | 0.0254 | 2.69E-06 | 0.725 |  | 0.0077 | 0.700 | 0.095 |  | 0.0008 | - | - |
| rs61441836 | 88773595 | G | 0.0335 | 5.95E-07 | 0.677 |  | 0.0126 | 0.865 | 0.033 |  | 0.0008 | - | - |
| rs73438172 | 88774420 | A | 0.0335 | 5.95E-07 | 0.677 |  | 0.0126 | 0.865 | 0.033 |  | 0.0008 | - | - |
| rs1044043958 | 88778604 | A | 0.0254 | 2.69E-06 | 0.725 |  | 0.0077 | 0.700 | 0.095 |  | 0.0015 | 0.876 | -0.111 |
| rs73438182 | 88779036 | G | 0.0640 | 8.99E-05 | 0.385 |  | 0.0194 | 0.961 | 0.008 |  | 0.0015 | 0.663 | -0.311 |
| rs73438185 | 88779473 | A | 0.0640 | 8.99E-05 | 0.385 |  | 0.0194 | 0.961 | 0.008 |  | 0.0015 | 0.663 | -0.311 |
| rs73438188 | 88783270 | A | 0.0283 | 4.78E-08 | 0.801 |  | 0.0113 | 0.656 | 0.090 |  | 0.0008 | - | - |
| rs73438190 | 88783315 | C | 0.0479 | 1.25E-05 | 0.492 |  | 0.0180 | 0.793 | 0.043 |  | 0.0015 | 0.663 | -0.311 |
| rs73438195 | 88783668 | A | 0.0479 | 1.25E-05 | 0.492 |  | 0.0180 | 0.793 | 0.043 |  | 0.0015 | 0.663 | -0.311 |
| rs111857459 | 88785774 | T | 0.0479 | 1.25E-05 | 0.492 |  | 0.0180 | 0.793 | 0.043 |  | 0.0015 | 0.663 | -0.311 |
| rs144369986 | 88789109 | T | 0.0248 | 1.01E-06 | 0.764 |  | 0.0077 | 0.700 | 0.095 |  | 0.0008 | - | - |
| rs73440106 | 88791281 | G | 0.0479 | 1.25E-05 | 0.492 |  | 0.0180 | 0.793 | 0.043 |  | 0.0015 | 0.663 | -0.311 |
| rs73440107 | 88793808 | A | 0.0283 | 4.78E-08 | 0.801 |  | 0.0113 | 0.656 | 0.090 |  | 0.0008 | - | - |
| rs111453514 | 88796826 | C | 0.0283 | 4.78E-08 | 0.801 |  | 0.0113 | 0.656 | 0.090 |  | 0.0008 | - | - |
| rs73440112 | 88797371 | T | 0.0283 | 4.78E-08 | 0.801 |  | 0.0113 | 0.656 | 0.090 |  | 0.0008 | - | - |
| rs73440115 | 88798068 | G | 0.0283 | 4.78E-08 | 0.801 |  | 0.0113 | 0.656 | 0.090 |  | 0.0008 | - | - |
| rs11312747 | 88800273 | A | 0.1003 | 8.96E-06 | 0.361 |  | 0.0275 | 0.163 | 0.185 |  | 0.0023 | 0.451 | -0.439 |
| rs73440120 | 88801090 | A | 0.0248 | 1.01E-06 | 0.764 |  | 0.0077 | 0.700 | 0.095 |  | 0.0008 | - | - |
| rs111289668 | 88801600 | G | 0.0283 | 4.78E-08 | 0.801 |  | 0.0113 | 0.656 | 0.090 |  | 0.0008 | - | - |
| rs73440122 | 88802121 | C | 0.0271 | 1.67E-07 | 0.783 |  | 0.0113 | 0.656 | 0.090 |  | 0.0008 | - | - |
| rs73440123 | 88803531 | G | 0.0271 | 1.67E-07 | 0.783 |  | 0.0113 | 0.656 | 0.090 |  | 0.0008 | - | - |
| rs17016065^ | 88804280 | G | 0.1770 | 2.79E-06 | 0.298 |  | 0.0564 | 0.220 | 0.116 |  | 0.0100 | 0.877 | 0.044 |
| rs17016066 | 88804520 | A | 0.1770 | 2.79E-06 | 0.298 |  | 0.0564 | 0.220 | 0.116 |  | 0.0100 | 0.877 | 0.044 |
| rs147400083 | 88807139 | T | 0.1770 | 2.79E-06 | 0.298 |  | 0.0564 | 0.220 | 0.116 |  | 0.0100 | 0.877 | 0.044 |
| rs866852270 | 88807149 | T | 0.1770 | 2.79E-06 | 0.298 |  | 0.0564 | 0.220 | 0.116 |  | 0.0100 | 0.877 | 0.044 |
| rs141293300 | 88810932 | C | 0.0271 | 1.67E-07 | 0.783 |  | 0.0113 | 0.656 | 0.090 |  | 0.0008 | - | - |
| rs1398303 | 88811317 | A | 0.1863 | 1.21E-05 | 0.276 |  | 0.0604 | 0.138 | 0.134 |  | 0.0154 | 0.651 | 0.104 |
| rs61924868 | 88811575 | T | 0.1851 | 1.23E-05 | 0.277 |  | 0.0604 | 0.138 | 0.134 |  | 0.0154 | 0.651 | 0.104 |
| rs73440134 | 88813735 | T | 0.0271 | 1.67E-07 | 0.783 |  | 0.0113 | 0.656 | 0.090 |  | 0.0008 | - | - |
| rs73429413 | 88817859 | G | 0.0271 | 1.67E-07 | 0.783 |  | 0.0113 | 0.656 | 0.090 |  | 0.0008 | - | - |
| rs73429415 | 88819310 | A | 0.1886 | 5.19E-05 | 0.255 |  | 0.0609 | 0.094 | 0.151 |  | 0.0162 | 0.800 | 0.057 |
| rs112449284 | 88823778 | T | 0.0311 | 3.92E-06 | 0.650 |  | 0.0135 | 0.201 | 0.238 |  | 0.0031 | 0.485 | 0.355 |
| rs111981782 | 88826878 | C | 0.0288 | 4.32E-08 | 0.796 |  | 0.0113 | 0.656 | 0.090 |  | 0.0008 | - | - |
| rs150942400 | 88829116 | T | 0.0294 | 4.10E-08 | 0.791 |  | 0.0113 | 0.656 | 0.090 |  | 0.0008 | - | - |
| rs147527487 | 88830592 | C | 0.2053 | 7.65E-05 | 0.232 |  | 0.0663 | 0.673 | -0.035 |  | 0.0069 | 0.897 | -0.043 |
| rs111243672 | 88839615 | A | 0.0323 | 4.28E-07 | 0.698 |  | 0.0131 | 0.827 | -0.042 |  | 0.0023 | 0.810 | -0.139 |
| rs73429450^ | 88846435 | A | 0.0306 | 9.50E-09 | 0.812 |  | 0.0140 | 0.507 | 0.122 |  | 0.0023 | 0.903 | 0.071 |
| rs758775577 | 88850005 | C | 0.0410 | 1.96E-06 | 0.580 |  | 0.0216 | 0.375 | -0.132 |  | 0.0031 | 0.772 | 0.148 |
| rs142679473 | 88863271 | C | 0.0311 | 4.86E-08 | 0.765 |  | 0.0122 | 0.734 | 0.067 |  | 0.0023 | 0.903 | 0.071 |

The three independent signals identified in the conditional analyses are marked with ^ near the rsID.

Table S12. Statistical and functional annotations for the 45 lung function associations.

|  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| M | rsID | Alt | β | p | βnorm | pnorm | ccRE | DNaseI | TFBS | Nasal eQTL | UKBB | SAPPHIRE | GCPD-A | Score |
| 1 | rs11835305 | T | 0.126 | 1.93E-05 | 0.320 | 3.69E-05 | N | N | N | N | N | N | N | 0 |
| 2 | rs17015963 | C | 0.126 | 1.93E-05 | 0.320 | 3.69E-05 | N | N | N | N | N | N | N | 0 |
| 3 | rs58475486\* | T | 0.127 | 1.45E-05 | 0.323 | 2.81E-05 | Y | N | FOXA1 (A549) | N | N | N | N | 2^ |
| 4 | rs17015979 | T | 0.127 | 1.45E-05 | 0.323 | 2.81E-05 | N | N | N | N | N | N | N | 0 |
| 5 | rs57692452 | C | 0.245 | 1.63E-06 | 0.654 | 1.06E-06 | Y | N | N | N | Y | FF | N | 2^ |
| 6 | rs112585732 | T | 0.235 | 4.35E-07 | 0.625 | 3.06E-07 | N | N | N | N | N | N | N | 0 |
| 7 | rs113837356 | T | 0.270 | 3.44E-06 | 0.719 | 2.49E-06 | N | N | N | - | Y | N | N | 1 |
| 8 | rs61441836 | G | 0.252 | 8.19E-07 | 0.671 | 5.46E-07 | N | N | N | N | Y | FF | N | 1 |
| 9 | rs73438172 | A | 0.252 | 8.19E-07 | 0.671 | 5.46E-07 | N | N | N | N | Y | FF | N | 1 |
| 10 | rs1044043958& | A | 0.270 | 3.44E-06 | 0.719 | 2.49E-06 | N | N | N | - | N | N | N | 0 |
| 11 | rs73438182 | G | 0.138 | 1.68E-04 | 0.378 | 9.18E-05 | N | N | N | N | N | FF | FF | 0 |
| 12 | rs73438185 | A | 0.138 | 1.68E-04 | 0.378 | 9.18E-05 | N | N | N | N | N | FF | FF | 0 |
| 13 | rs73438188 | A | 0.297 | 6.97E-08 | 0.792 | 4.42E-08 | N | N | N | N | Y | N | N | 1 |
| 14 | rs73438190 | C | 0.181 | 1.86E-05 | 0.486 | 1.22E-05 | N | N | N | N | N | FF | N | 0 |
| 15 | rs73438195 | A | 0.181 | 1.86E-05 | 0.486 | 1.22E-05 | N | N | N | N | N | FF | N | 0 |
| 16 | rs111857459 | T | 0.181 | 1.86E-05 | 0.486 | 1.22E-05 | N | N | N | N | N | FF | N | 0 |
| 17 | rs144369986& | T | 0.285 | 1.21E-06 | 0.756 | 9.44E-07 | N | N | N | - | N | N | - | 0 |
| 18 | rs73440106 | G | 0.181 | 1.86E-05 | 0.486 | 1.22E-05 | N | N | N | N | N | FF | N | 0 |
| 19 | rs73440107 | A | 0.297 | 6.97E-08 | 0.792 | 4.42E-08 | N | N | N | N | Y | N | N | 1 |
| 20 | rs111453514 | C | 0.297 | 6.97E-08 | 0.792 | 4.42E-08 | N | N | N | N | Y | N | N | 1 |
| 21 | rs73440112 | T | 0.297 | 6.97E-08 | 0.792 | 4.42E-08 | N | N | N | N | Y | N | N | 1 |
| 22 | rs73440115 | G | 0.297 | 6.97E-08 | 0.792 | 4.42E-08 | N | N | N | N | Y | N | N | 1 |
| 23 | rs11312747& | A | 0.133 | 1.43E-05 | 0.357 | 8.51E-06 | N | N | N | - | N | N | N | 0 |
| 24 | rs73440120 | A | 0.285 | 1.21E-06 | 0.756 | 9.44E-07 | N | N | N | - | Y | N | N | 1 |
| 25 | rs111289668 | G | 0.297 | 6.97E-08 | 0.792 | 4.42E-08 | Y | N | N | N | Y | N | N | 2^ |
| 26 | rs73440122 | C | 0.292 | 2.08E-07 | 0.775 | 1.55E-07 | Y | GM12865 | SPI1 (HL-60) | N | Y | N | N | 3^ |
| 27 | rs73440123 | G | 0.292 | 2.08E-07 | 0.775 | 1.55E-07 | N | N | N | N | Y | N | N | 1 |
| 28 | rs17016065\* | G | 0.112 | 3.19E-06 | 0.296 | 2.54E-06 | N | N | N | Y | N | FF | N | 1# |
| 29 | rs17016066 | A | 0.112 | 3.19E-06 | 0.296 | 2.54E-06 | N | N | N | Y | N | FF | N | 1# |
| 30 | rs147400083& | T | 0.112 | 3.19E-06 | 0.296 | 2.54E-06 | N | N | N | - | N | - | N | 0 |
| 31 | rs866852270 | T | 0.112 | 3.19E-06 | 0.296 | 2.54E-06 | N | N | N | - | N | FF | N | 0 |
| 32 | rs141293300& | C | 0.292 | 2.08E-07 | 0.775 | 1.55E-07 | Y | N | N | - | N | N | N | 1 |
| 33 | rs1398303 | A | 0.104 | 1.22E-05 | 0.274 | 1.12E-05 | N | N | N | Y | N | FF | N | 1# |
| 34 | rs61924868 | T | 0.104 | 1.24E-05 | 0.275 | 1.14E-05 | N | N | N | Y | N | FF | N | 1# |
| 35 | rs73440134 | T | 0.292 | 2.08E-07 | 0.775 | 1.55E-07 | N | N | N | N | Y | N | N | 1 |
| 36 | rs73429413 | G | 0.292 | 2.08E-07 | 0.775 | 1.55E-07 | N | N | N | N | Y | N | N | 1 |
| 37 | rs73429415 | A | 0.096 | 5.13E-05 | 0.253 | 4.84E-05 | N | N | N | Y | N | N | N | 1# |
| 38 | rs112449284 | T | 0.242 | 4.64E-06 | 0.640 | 3.87E-06 | N | N | N | N | Y | N | N | 1 |
| 39 | rs111981782 | C | 0.296 | 5.78E-08 | 0.786 | 4.09E-08 | N | N | N | N | Y | N | N | 1 |
| 40 | rs150942400 | T | 0.293 | 6.01E-08 | 0.780 | 4.01E-08 | N | N | N | N | Y | N | N | 1 |
| 41 | rs147527487 | C | 0.086 | 8.49E-05 | 0.226 | 8.14E-05 | N | N | N | N | N | N | N | 0 |
| 42 | rs111243672 | A | 0.258 | 6.41E-07 | 0.690 | 3.99E-07 | N | N | N | N | Y | N | N | 1 |
| 43 | rs73429450\* | A | 0.302 | 1.26E-08 | 0.801 | 9.01E-09 | N | N | N | N | Y | N | N | 1 |
| 44 | rs758775577 | C | 0.217 | 2.22E-06 | 0.574 | 1.85E-06 | N | N | N | N | N | N | N | 0 |
| 45 | rs142679473& | C | 0.285 | 6.30E-08 | 0.756 | 4.62E-08 | N | N | N | - | N | N | N | 0 |

M, marker number that corresponds to those in Figure and Table 2. Score, priority score based on counting the statistical (UKBB, SAPPHIRE and GCPD-A) and functional (ccRE, DNaseI, TFBS, Nasal eQTL) evidence. Candidate variants were prioritized if they had a functional score of greater than 2 (^) or if they are eQTL of KITLG in nasal epithelial cells (#). The three independent signals identified in the conditional analyses are marked with \* near the rsID. Indels were marked with the superscript & near the rsID. β (p) and βnorm(pnorm) are the effect sizes (p-values) of the genetic associations of the alternate allele (alt) with FEV1 and FEV1.res.rnorm respectively. ccRE, candidate cis-regulatory elements derived from ENCODE data and stored in the SCREEN web interface. DNaseI, ENCODE DNase I hypersensitivity clusters derived from assays in 95 cells (UCSC hg38 wgEncodeRegDnaseClustered table) in embryonic, blood, bone marrow or lung-related cell lines. TFBS, ENCODE transcription factor ChIP-Seq clusters representing 340 transcription factors and 129 cell types (UCSC hg38 encRegTfbsClustered table). Nasal eQTL, KITLG eQTLs in nasal epithelial cells. UKBB/SAPPHIRE/GCPD-A, replication association analysis of FEV1 in UK Biobank Blacks and African Americans in SAPPHIRE and GCPD-A (detailed results in Supplementary Table 3). GM12865, a B-lymphoblastoid cell line. HL-60, an acute promyelocytic leukemia cell line. A549, a lung adenocarcinoma cell line. -, not available; FF, flip-flop association defined as replication association (p < 0.05) of the opposite allele instead of the original allele identified in the discovery cohort.