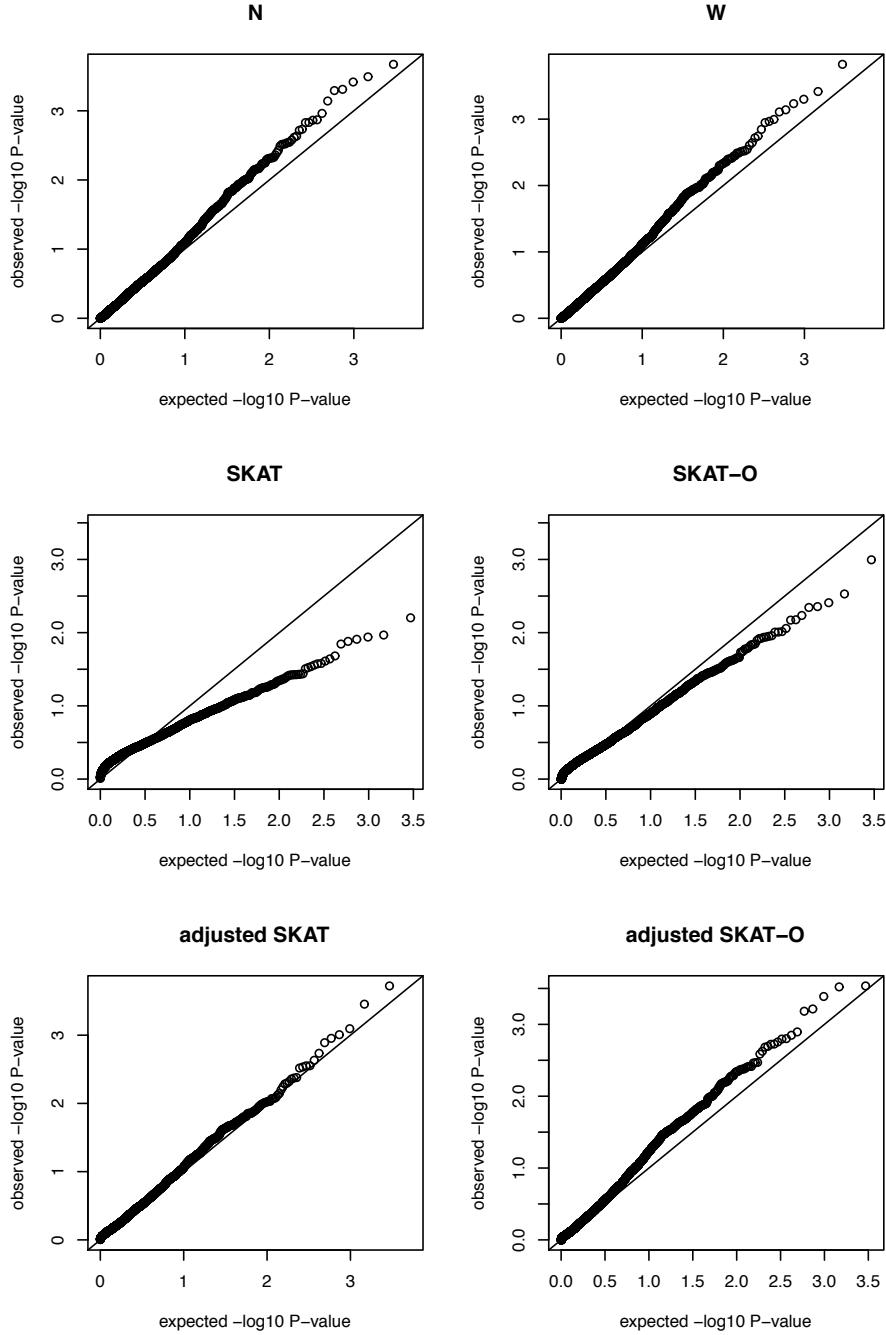


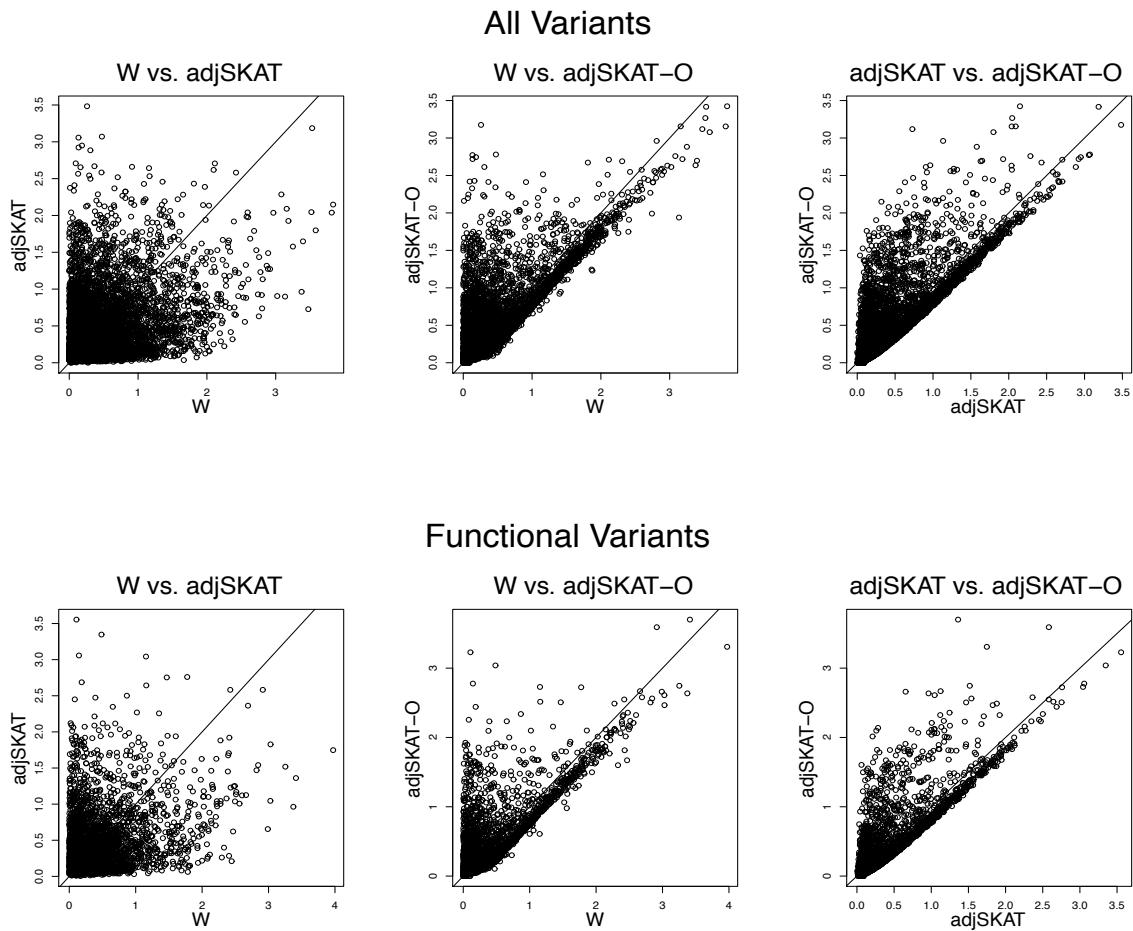
## **SUPPLEMENTAL DATA**

Supplementary Table 1: Observed number of causal variants within randomly selected 3kb regions in the power simulation. Each entry represents the average number of observed causal variants (harmful + protective), observed harmful variants, and observed protective variants in the simulated datasets. “all” represents the number of causal variants observed among all samples. “case” and “control” represent the number of observed causal variants among cases and controls, respectively. Harmful variants increase the chance to be a case ( $\beta > 0$ ) and protective variants reduce the chance to be a case ( $\beta < 0$ ).

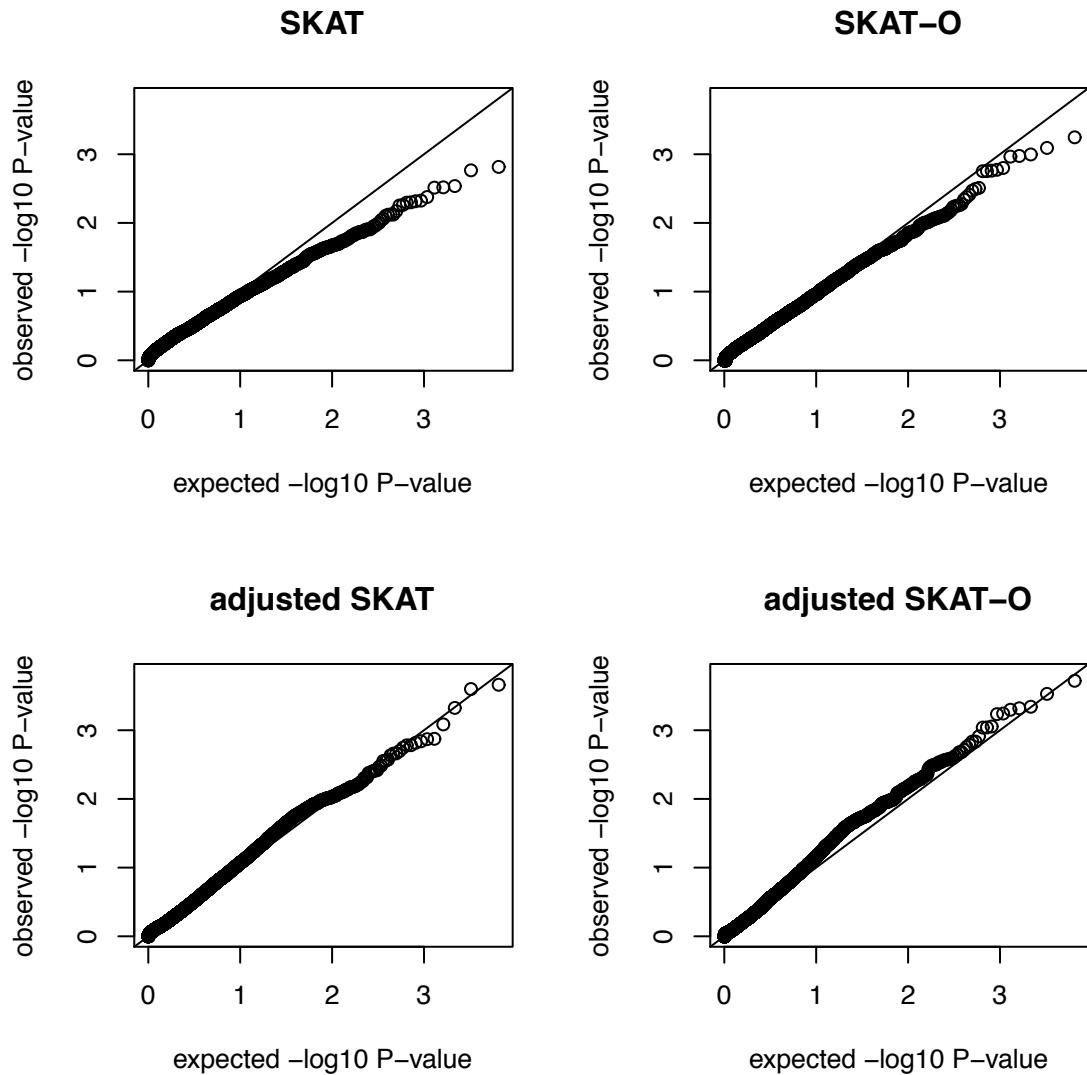
Total sample size	% of protective variants	observed causal			observed harmful			observed protective		
		all	case	control	all	case	control	all	case	control
10% variants were causal (average number of causal variants = 4.9)										
200	0%	3.38	3.38	0.00	3.33	3.33	0.00	0.70	0.70	0.00
	20%	3.02	2.90	0.12	2.89	2.87	0.02	0.72	0.60	0.12
	50%	2.31	1.94	0.37	2.00	1.91	0.09	0.78	0.43	0.36
500	0%	4.39	4.39	0.00	4.37	4.37	0.00	1.16	1.16	0.00
	20%	3.95	3.78	0.17	3.81	3.77	0.05	1.12	0.95	0.17
	50%	3.02	2.43	0.59	2.58	2.42	0.16	1.20	0.63	0.57
1000	0%	4.74	4.74	0.00	4.74	4.74	0.00	1.56	1.56	0.00
	20%	4.33	4.07	0.27	4.12	4.05	0.06	1.62	1.36	0.26
	50%	3.43	2.62	0.81	2.82	2.62	0.20	1.66	0.87	0.80
20% variants were causal (average number of causal variants = 10.2)										
200	0%	5.85	5.85	0.00	5.68	5.68	0.00	1.49	1.49	0.00
	20%	5.33	5.02	0.31	4.94	4.85	0.09	1.62	1.33	0.29
	50%	4.05	3.20	0.84	3.34	3.11	0.23	1.59	0.79	0.80
500	0%	8.15	8.15	0.00	8.01	8.01	0.00	2.49	2.49	0.00
	20%	7.42	6.93	0.48	6.96	6.82	0.14	2.51	2.05	0.46
	50%	5.67	4.34	1.33	4.70	4.27	0.42	2.60	1.32	1.28
1000	0%	9.41	9.41	0.00	9.33	9.33	0.00	3.47	3.47	0.00
	20%	8.55	7.88	0.67	8.03	7.81	0.22	3.58	2.94	0.64
	50%	6.96	5.03	1.94	5.56	4.98	0.58	3.70	1.84	1.86
50% variants were causal (average number of causal variants = 26.3)										
200	0%	10.14	10.14	0.00	9.08	9.08	0.00	4.10	4.10	0.00
	20%	9.23	8.31	0.92	7.84	7.39	0.45	4.23	3.44	0.79
	50%	7.47	5.17	2.30	5.64	4.61	1.04	4.08	2.11	1.97
500	0%	15.45	15.45	0.00	14.27	14.27	0.00	6.75	6.75	0.00
	20%	13.97	12.56	1.41	12.20	11.56	0.64	6.75	5.48	1.26
	50%	11.67	7.82	3.86	8.94	7.20	1.74	6.86	3.39	3.47
1000	0%	19.80	19.80	0.00	18.63	18.63	0.00	9.53	9.53	0.00
	20%	18.23	16.20	2.03	16.13	15.20	0.93	9.60	7.78	1.82
	50%	15.14	10.00	5.14	11.64	9.43	2.21	9.54	4.80	4.73



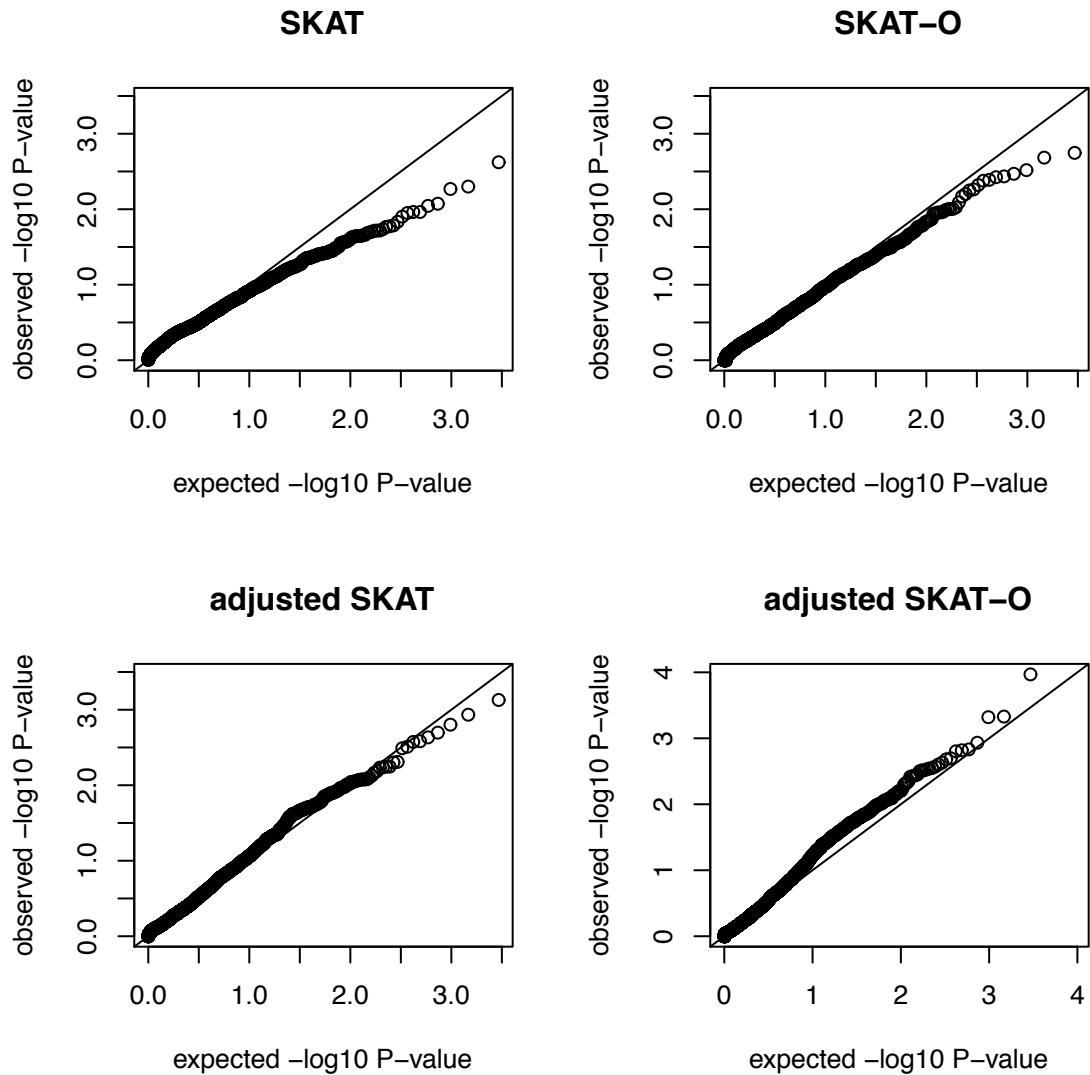
Supplementary Figure 1:  $-\log_{10}$  QQ plots of observed vs. expected p-values for the ALI whole exome sequencing data using the six methods: burden tests (W,N), SKAT, SKAT-O, adjusted SKAT, adjusted SKAT-O. All the variants were used. X-axis represents  $-\log_{10}$  expected p-values, and Y-axis represents  $-\log_{10}$  observed p-values. Total 2,939 genes with at least four rare functional variants were tested for associations with ALI.



Supplementary Figure 2: Scatter plots of  $-\log_{10}$  p-values to compare burden test (W), adjusted SKAT, adjusted SKAT-O. The top panel considers to test all variants, and bottom panel considers to test functional variants.



Supplementary Figure 3:  $-\log_{10}$  QQ plots of observed vs. expected p-values for the ALI whole exome sequencing data with logistic weight ( $w_j = \exp((a_1 - p_j)a_2) / \{1 + \exp((a_1 - p_j)a_2)\}$ ) with  $a_1 = 0.07$  and  $a_2 = 150$ . X-axis represents  $-\log_{10}$  expected p-values, and Y-axis represents  $-\log_{10}$  observed p-values. Total 6,488 genes with at least four rare variants were tested for associations with ALI.



Supplementary Figure 4:  $-\log_{10}$  QQ plots of observed vs. expected p-values for the ALI whole exome sequencing data with logistic weight ( $w_j = \exp((a_1 - p_j)a_2)/\{1 + \exp((a_1 - p_j)a_2)\}$ ) with  $a_1 = 0.07$  and  $a_2 = 150$ . Only functional variants were used for testing the association. X-axis represents  $-\log_{10}$  expected p-values, and Y-axis represents  $-\log_{10}$  observed p-values. Total 2,939 genes with at least four rare functional variants were tested for associations with ALI.

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[http://www.whiscience.org/publications/WHI\\_investigators\\_shortlist.pdf](http://www.whiscience.org/publications/WHI_investigators_shortlist.pdf)

## **NHLBI GO Exome Sequencing Project**

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## **ESP Groups**

<sup>1</sup>Anthropometry Project Team, <sup>2</sup>Blood Count/Hematology Project Team, <sup>3</sup>Blood Pressure Project Team, <sup>4</sup>Data Flow Working Group, <sup>5</sup>Early MI Project Team, <sup>6</sup>ELSI Working Group, <sup>7</sup>Executive Committee, <sup>8</sup>Family Study Project Team, <sup>9</sup>Lipids Project Team, <sup>10</sup>Lung Project Team, <sup>11</sup>Personal Genomics Project Team, <sup>12</sup>Phenotype and Harmonization Working Group, <sup>13</sup>Population Genetics and Statistical Analysis Working Group, <sup>14</sup>Publications and Presentations Working Group, <sup>15</sup>Quantitative Analysis Ad Hoc Task Group, <sup>16</sup>Sequencing and Genotyping Working Group, <sup>17</sup>Steering Committee, <sup>18</sup>Stroke Project Team, <sup>19</sup>Structural Variation Working Group, <sup>20</sup>Subclinical/Quantitative Project Team

## **ESP Cohorts**

<sup>21</sup>Acute Lung Injury (ALI), <sup>22</sup>Atherosclerosis Risk in Communities (ARIC), <sup>23</sup>Cardiovascular Health Study (CHS), <sup>24</sup>Chronic Obstructive Pulmonary Disease (COPDGene), <sup>25</sup>Coronary Artery Risk Development in Young Adults (CARDIA), <sup>26</sup>Cystic Fibrosis (CF), <sup>27</sup>Early Pseudomonas Infection Control (EPIC), <sup>28</sup>Framingham Heart Study (FHS), <sup>29</sup>Jackson Heart Study (JHS), <sup>30</sup>Lung Health Study (LHS), <sup>31</sup>Multi-Ethnic Study of Atherosclerosis (MESA), <sup>32</sup>Pulmonary Arterial Hypertension (PAH), <sup>33</sup>Severe Asthma Research Program (SARP), <sup>34</sup>Women's Health Initiative (WHI)

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