

**Table S12. Summary of RegulomeDB scores of 153 SCARB1 variants.**

| RegulomeDB Score <sup>a</sup> | No of Variants (n) |            |                           |            |                  |            | Total N (%)         |
|-------------------------------|--------------------|------------|---------------------------|------------|------------------|------------|---------------------|
|                               | MAF ≥5% (n = 101)  |            | MAF between 1-5% (n = 27) |            | MAF ≤1% (n = 25) |            |                     |
|                               | Coding             | Non-coding | Coding                    | Non-coding | Coding           | Non-coding |                     |
| 1f                            |                    | 1          |                           |            |                  |            | 1 (0.65)            |
| 2a                            |                    | 1          |                           | 1          |                  | 1          | 3 (1.96)            |
| 2b                            | 1                  | 4          | 1                         | 1          |                  |            | 7 (4.58)            |
| 3a                            | 1                  | 1          |                           |            |                  |            | 2 (1.31)            |
| 3b                            |                    | 1          |                           |            |                  |            | 1 (0.65)            |
| 4                             | 1                  | 18         | 1                         | 10         |                  | 6          | 36 (23.53)          |
| 5                             | 1                  | 40         |                           | 11         | 2                | 13         | 67 (43.79)          |
| 6                             |                    | 12         |                           | 1          |                  | 2          | 15 (9.80)           |
| 7                             |                    | 19         |                           | 1          |                  | 1          | 21 (13.73)          |
| <b>Total N</b>                | <b>4</b>           | <b>97</b>  | <b>2</b>                  | <b>25</b>  | <b>2</b>         | <b>23</b>  | <b>153 (100.00)</b> |

MAF, minor allele frequency; SNP, single nucleotide polymorphism.

A list of 83 variants identified by sequencing is shown in Additional file 3 Table S3.

A list of 68 common HapMap-YRI tagSNPs successfully genotyped in our entire sample is shown in Additional file 7 Table S5.

<sup>a</sup> The RegulomeDB (version 1.0) scoring scheme: **score 1a**, expression quantitative trait loci (eQTL) + transcription factor (TF) binding + matched TF motif + matched DNase Footprint + DNase peak; **score 1b**, eQTL + TF binding + any motif + DNase Footprint + DNase peak; **score 1c**, eQTL + TF binding + matched TF motif + DNase peak; **score 1d**, eQTL + TF binding + any motif + DNase peak; **score 1e**, eQTL + TF binding + matched TF motif; **score 1f**, eQTL + TF binding / DNase peak; **score 2a**, TF binding + matched TF motif + matched DNase Footprint + DNase peak; **score 2b**, TF binding + any motif + DNase Footprint + DNase peak; **score 2c**, TF binding + matched TF motif + DNase peak; **score 3a**, TF binding + any motif + DNase peak; **score 3b**, TF binding + matched TF motif; **score 4**, TF binding + DNase peak; **score 5**, TF binding or DNase peak; **score 6**, others; **score 7**, no data (see <http://regulome.stanford.edu/help>).