**Table 1. Genetic variants within *TOMM40-APOE* region associated with risk of AD.**

|  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- |
| Gene | SNP | Position | MAF (cases/NC) | MAF\*  dbSNP | OR (95% CI) | *P* value |
| *PVRL2*: Intron variant | rs394221 | 45368424 | 0.51/0.38 | 0.45 | 1.7 (1.2~2.4) | 0.001 |
| *TOMM40*:Synonymous, p.Ser66= | **rs772262361**\* | 45394870 | 0.013/0.0 | 0.00004 | - | - |
| *TOMM40*: Intron | rs184017 | 45394969 | 0.34/0.16 | 0.20 | 2.8 (2.0~4.0) | 4.2E-08 |
| *TOMM40*: Intron | rs2075650 | 45395619 | 0.25/0.07 | 0.13 | 4.2 (2.8~6.1) | 1.1E-10 |
| *TOMM40*: Missense, p.Phe113Leu | **rs157581**\* | 45395714 | 0.38/0.23 | 0.23 | 2.1 (1.5~2.9) | 4.4E-05 |
| *TOMM40* : Missense, p.Phe131Leu | **rs11556505**\* | 45396144 | 0.26/0.10 | 0.11 | 3.3 (2.2~4.8) | 2.5E-08 |
| *TOMM40*: Intron | rs157582 | 45396219 | 0.34/0.18 | 0.22 | 2.4 (1.7~3.3) | 3.3E-06 |
| *APOE*: Missense, p.Asn14Lys | **rs440446**\* | 45409167 | 0.56/0.33 | 0.38 | 2.6 (1.9~3.6) | 1.4E-08 |
| *APOE*: Intron | rs769449 | 45412079 | 0.25/0.08 | 0.11 | 3.6 (2.4~5.3) | 2.7E-09 |

\*SNPs marked in bold were missense variants. **rs772262361**was a point mutation.