Association between ARNTL (BMAL1) rs2278749 polymorphism T >C and susceptibility to Alzheimer disease in a Chinese population

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ABSTRACT. In the present study, we examined whether the ARNTL (BMAL1) rs2278749 T/C polymorphism was associated with the susceptibility to Alzheimer disease (AD). This case-control study examined the genotypes of apolipoprotein E (APOE e4) and BMAL1 rs2278749 T/C using restriction fragment length polymorphism and the TaqMan assay, respectively. A total of 296 unrelated AD patients and 423 control subjects were included. Both in the entire sample and in APOE e4 non-carriers, the prevalence of T carriers in BMAL1 rs2278749 T/C in AD patients was significantly higher than that in control subjects (entire sample: χ² = 12.950, P < 0.0001; APOE e4 non-carriers: χ² = 13.094, P < 0.0001). Both in the entire sample and in APOE e4 non-carriers, the prevalence of TT genotypes in AD patients was also significantly higher than that in control subjects (entire sample: χ² = 7.765, P = 0.024; APOE e4 non-carriers: χ² = 13.062, P < 0.0001). However, among APOE e4 carriers, the difference
in the prevalence of T carriers or TT genotypes in the \(BMAL1\) rs2278749 T/C between patients and control subjects presents was not significant (T carriers: \(\chi^2 = 0.078, P = 0.851\) or TT genotypes: \(\chi^2 = 2.576, P = 0.325\)). Among \(APOE e4\) non-carriers, T carriers in the \(BMAL1\) rs2278749 T/C were associated with a high susceptibility to AD, but among \(APOE e4\) carriers, the association between AD and \(BMAL1\) rs2278749 T/C was not significant.

**Key words:** Alzheimer Disease; Apolipoprotein E; \(BMAL1\) rs2278749T/C; Case-control study; Genetic risk factor; Metabolism; Polymorphism; Susceptibility