



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: $\chi^2 = 12.950$, $P < 0.0001$; *APOE e4* non-carriers: $\chi^2 = 13.094$, $P < 0.0001$). Both in the entire sample and in *APOE e4* non-carriers, the prevalence of TT genotypes 2278749 in AD patients was also significantly higher than that in control subjects (entire sample: $\chi^2 = 7.765$, $P = 0.024$; *APOE e4* non-carriers: $\chi^2 = 13.062$, $P < 0.0001$). 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