Estrogen receptor 1 PvuII and XbaI polymorphisms and susceptibility to Alzheimer’s disease: a meta-analysis

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ABSTRACT. The aim of this study was to explore whether estrogen receptor 1 (ESR1) PvuII and XbaI polymorphisms are associated with susceptibility to Alzheimer’s disease (AD). We conducted a meta-analysis of the associations between AD and ESR1 PvuII and XbaI polymorphisms as well as haplotypes of the ESR1 PvuII and XbaI polymorphisms. A total of 1359 patients and 1387 controls from 9 studies on the ESR1 PvuII polymorphism and 1525 patients and 1575 controls from 8 studies on the ESR1 XbaI polymorphism were included in this meta-analysis. Gender-specific meta-analysis showed an association between the ESR1 PP+Pp genotype and AD in males (OR = 0.302, 95%CI = 0.100-0.914, P = 0.034), but not in females. No association was observed between AD and the ESR1 XbaI X allele (OR = 1.114, 95%CI = 0.868-1.429, P = 0.397). However, country-specific meta-analysis identified an association between AD and the ESR1 X allele in Japanese (OR = 1.386, 95%CI = 1.055-1.822, P = 0.019), but not Chinese or Italian populations. Meta-analyses results indicated an association between the PP/XX haplotypes and AD in Chinese
population (OR for PP/XX vs others = 2.758, 95%CI = 1.750-4.346, 
$P = 1.2 \times 10^{-6}$). This meta-analysis showed associations between the $ESR1$ $PvuII$ polymorphism and AD susceptibility in males, between AD risk and the $ESR1$ $XbaI$ polymorphism in the Japanese population, and between the PP/XX haplotype and AD susceptibility in the Chinese population.

**Key words:** Alzheimer’s disease; Estrogen receptor; Polymorphism; Meta-analysis