The cathepsin D (224C/T) polymorphism confers an increased risk to develop Alzheimer's disease in men.

Abstract:
The lysosomal protease cathepsin D is likely involved in beta-amyloidogenesis in Alzheimer's disease (AD). There is evidence for a single nucleotide polymorphism (rs17571) of the cathepsin D gene to be associated with increased AD risk. However, little is known about gender-specific differences. Therefore, we performed a genetic association study focusing on gender-specific differences in 434 participants (219 AD and 215 controls). Screening of the rs17571 shows a significantly higher proportion of T-allele carriers among male Alzheimer patients (28.5%) when compared with male controls (13.8%, p = .013, p(corr) = .039). The odds ratio was 2.48 (95% confidence interval: 1.14-5.58). There was no significant difference in the T-allele distribution in women. Including APOE4 status and age did not have an additional effect on the morbidity risk. Thus, our results support the idea that rs17571 confers an increased risk for AD in men but not in women. Further investigation should substantiate the role of gender for AD risk of rs17571.