A genetic variation of cathepsin D is a major risk factor for Alzheimer's disease

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Author(s) Papassotiropoulos, A; Bagli, M; Kurz, A; Kornhuber, J; Förstl, H; Maier, W; Pauls, J; Lautenschlager, N; Heun, R
Author(s) at UniBasel Papassotiropoulos, Andreas ;
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Cathepsin D (catD) is an intracellular acid protease possibly involved in Alzheimer's disease (AD)-related neurodegeneration through cleavage of amyloid precursor protein into amyloidogenic components. We studied whether an exonic polymorphism of the catD gene (C –>T [Ala –>Val] transition at position 224), which possibly influences pro-catD secretion and intracellular maturation of the enzyme, was associated with the risk for the development of AD in 127 demented patients and 184 controls. The catD*T allele was significantly overrepresented in demented patients (11.8%) compared with nondemented controls (4.9%). Carriers of the catD*T allele had a 3.1-fold increased risk for developing AD than noncarriers. Carriers of the apolipoprotein E (ApoE) epsilon4 allele (ApoE*4) had a 3.9-fold increased risk than noncarriers. The adjusted odds ratio for subjects with the ApoE*4 and the catD*T allele was 19.0 compared with subjects with neither of these two alleles. Our data confirm the results of a recently performed pilot study in an independent sample and suggest that the catD genotype is strongly associated with the risk for AD.

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