


Genes related to iron metabolism and susceptibility to Alzheimer's disease in Basque population

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Abstract

Alzheimer's disease (AD) is the most common dementing disorder and presents with a progressive and irreversible cognitive decline of gradual onset. To date, several reports have involved iron in AD pathophysiology. In this study, we have analysed TFC2 variant and HFE mutations (H63D and C282Y) in 211 AD patients and 167 controls recruited from an area of the Basque Country. Furthermore, we have studied APOE genotype as it is a well-known risk factor for AD. APOE 4 allele was associated with an increased risk of AD and an earlier age at onset, whereas no association was found between TFC2 or HFE C282Y mutation and disease susceptibility. The frequency of H63D mutation was higher in control population (29.9%) than in AD patients (18%), suggesting a protective role of this allele on AD either due to the presence of the mutation itself or through the effect of other related genes in the ancestral haplotype in which it is included.

Keywords

- Alzheimer's disease;
- HFE;
- APOE;
- Transferrin;
- Iron;
- Haemochromatosis