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EXTREME ALLELES AT THE HUMAN CAVEOLIN 1 GENE NOVEL PURINE COMPLEX AND RISK OF ALZHEIMER´S DISEASE

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Caveolin-1 (CAV1) is the principal structural protein of caveolae membranes that are found in most cell types. Aberrant expression and mutation of this gene are associated with a wide range of disorders including neurodegenerative disorders and various cancers. We report a novel purine complex of three polymorphic motifs located at the enhancer region of the gene and risk of late-onset Alzheimer´s disease. Extreme haplotypes with accumulated homozygosity for those haplotypes were observed in the Alzheimer´s cases comparing with the controls ($p < 0.000$). Based on our findings, there is a window of haplotypes and haplotype lengths in the controls. Shorter and longer haplotypes were associated with Alzheimer´s disease in our cases.
This purine complex contains GGAA and GAAA motifs, the consensus binding sites for the Ets and IRF family transcription factors, respectively, and is highly conserved in **distantly-related** non-human primates in respect with location and motif sequence. The effect of the extreme haplotypes in the expression of the gene and the pathophysiology of Alzheimer´s disease remain to be clarified.

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