Further analysis of KIFAP3 gene in ALS patients from Switzerland and Sweden

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A series of studies suggests that susceptibility to ALS may be influenced by variants in multiple genes. While analyses of the 10% of cases of familial origin have identified more than 33 monogenic ALS-causing genetic defects, little is known about genetic factors that influence susceptibility or phenotype in sporadic ALS (SALS). We and others conducted a genome-wide association study (GWAS) in a cohort of 1014 ALS cases from Western Europe, England and the United States, and identified an intronic single nucleotide polymorphism (SNP) rs1541160 in the KIFAP3 gene that was statistically associated with improved survival. We have now completed an additional survival analysis examining the impact of the rs1541160 genotype in a cohort of 264 ALS and progressive bulbar palsy (PBP) cases. In the combined cohort of 264 patients, the CC, CT and TT genotypes for rs1541160 were detected, respectively, in 8.3% (22), 41.7% (110) and 50.0% (132). This study does not show an influence of KIFAP3 variants on survival in the studied Swiss and Swedish cohort. There was a difference in survival between the US and English patients and the patients from the Netherlands. The effect of KIFAP3 variants may be population specific, or the rs1541160 association reported previously may have been a false-positive.

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