INVESTIGATION OF VITAMIN D RECEPTOR (VDR) GENE POLYMORPHISMS IN SPORADIC AMYOTROPHIC LATERAL SCLEROSIS

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BACKGROUND:

There are aberrations in vitamin D–endocrine system in sporadic amyotrophic lateral sclerosis (SALS). Vitamin D deficiency and the rise of the levels of calcium and parathormon were measured in the sera of ALS patients.

Diverse proteins were identified which link vitamin D to the theories of the selective degeneration of motor neurons too, albeit alterations of the VDR gene have not been reported.

OBJECTIVE:

Our aim was to investigate the single nucleotide polymorphisms of VDR gene in SALS patients in Hungary.

METHODS:

75 SALS patients and 97 healthy controls were enrolled to reveal the supposed different proportion of the alleles of the VDR receptor. Restriction fragment length polymorphism was used. For data analysis, SPSS software version 20.0 was utilized.

RESULTS:

Apal SNP was associated with the disease (χ 2 = 11.09; P = 0.004), the A allele of this SNP proved to be significantly associated with the ALS group (χ 2 = 5.352, df = 1, P = 0.021, OR = 0.600, 95% CI = 1.080– 2.569) so it may be an ALS risk factor. None of the investigated alleles influenced the age at disease onset.

CONCLUSIONS:

One allele of the examined polymorphisms of the VDR gene (Apal A allele) seems to be a risk factor in the Hungarian SALS population. However, due to the low number of cases, another study from the Central European region is needed.

To our knowledge, this is the second described VDR SNP investigation in this devastating disease.