



Session P09 - Neurogenetic and Psychiatric Disorders

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P09.017.B / B - Investigation of proven genetic risk factors in the Hungarian amyotrophic lateral sclerosis population

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e-Poster Area

Authors

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Disclosures

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Abstract

Introduction: Amyotrophic lateral sclerosis (ALS) is a fatal neurodegenerative disease which affects both upper and lower motor neurons. ALS has an oligogenic background: several genes and mutations are known to cause ALS, and additionally there are well established genetic risk factors and disease modifying variants for the disease. In this study we investigated 9 independent genetic risk factors of ALS in the Hungarian population. **Patients and methods:** 157 patients of Hungarian origin diagnosed with ALS were recruited for this study. For the analysis of the *ATXN1* and *ATXN2* genes repeat sizing was used. MLPA technique was used to screen for the duplications of *SMN1* gene. We reanalyzed whole exome sequencing data from a previous study then used Sanger sequencing to confirm the presence of the detected variants. **Results:** We could not confirm the previously detected association between the *CYLD* gene and ALS. We observed a non-significant but tendentious relationship between *ATXN2* intermediate-length repeat expansion and the duplication of the *SMN1* gene and ALS. In the *ANXA11*, *PON1*, *PON3* and *GLT8D1* genes we identified 1-1 relevant variants of uncertain significance (VUS), and in the *TIA1* gene 4 variants were detected; all have been identified as ALS-associated genetic variants. A potentially pathogenic variant in the *MFSN8* gene was revealed. **Discussion:** With our results we contribute to the fine mapping of the genetic heterogeneity of ALS. For the development and clinical application of novel therapeutic modalities in ALS it is essential to stratify the patients based on their genetic background. **Funding:** Hungarian Brain Research Program (Grant No. 2017-1.2.1-NKP-2017-00002).