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Motor Neuron Disease

Analysis of C9orf72 repeat expansions in Georgian patients with Amyotrophic lateral sclerosis (ALS)

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Background: Amyotrophic lateral sclerosis (ALS) is a fatal progressive neurodegenerative disorder that affects the upper and lower motor neurons. Several genetic risk factors have been identified in the past decade with a hexanucleotide repeat expansion in the C9orf72 gene being the most significant. However, the presence of C9orf72 repeat expansion has not been examined in the Transcaucasian region, therefore we aimed to analyze its frequency in Georgian patients with ALS.

Methods: We included 64 self-reported Georgian patients with ALS from different parts of the country, fulfilling the Gold Coast criteria. To investigate the presence of an expanded GGGGCC hexanucleotide repeat in the non-coding region of the C9orf72 gene, we performed Repeat-Primed PCR (RP-PCR).

Results: In total, 64 sporadic and two familial ALS cases were identified. Patients were aged 26 to 84 years with a mean age of 58.3 years at disease onset. Bulbar onset was observed in 21.88%, upper limb onset in 34.38%, and lower limb onset in 43.75% of the patients. Frontotemporal dementia (FTD) fulfilling the Strong criteria was diagnosed in seven patients (10.94%). C9orf72 repeat expansion was detected in only one case using RP-PCR; the patient had a family history of dementia.

Conclusions: Our results indicate that C9orf72 hexanucleotide expansion does not belong to the major genetic risk factor of ALS in Georgian patients. Further genetic studies in a bigger study population are needed to reveal the genetic causes of ALS in the Transcaucasian population.

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From Amyotrophic Lateral Sclerosis to Polyneuropathy: A Nerve-Wracking Relationship

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Introduction: Amyotrophic lateral sclerosis(ALS) is a devastating neurodegenerative condition of the motor neurons affecting around 4.5 people in 100,000. Chronic polyneuropathy is quite common in the general population, with a prevalence of up to 7%. Although ALS and axonal polyneuropathy can coexist without a causal relationship, the possibility of overlapping risk factors and shared molecular mechanisms was suggested by recent studies.

Methods: We conducted a cross-sectional study on the electronic medical record database at Colentina Clinical Hospital, a tertiary referral centre for ALS in Romania. We screened the available records that had G12.2 as the main ICD-10 diagnosis (i.e., motor neurone disease), up to December 2024. We included data from patients meeting criteria for ALS for which the result of at least one electroneuromyographic study was available.

Results: Data from 50 patients matched the inclusion criteria: 29 males and 21 females; mean age 59.7 years. Chronic axonal polyneuropathy was documented in 7 patients (14%) – sensory-motor in 6, sensory in 1. Of these, 4 patients had other possible explanations for their polyneuropathy (i.e., diabetes mellitus, toxic/nutritional), while 3 patients had no other possible explanation, 1 with flail arm onset, 1 with flail leg onset, and the third with a monogenic form. None of the patients had demyelinating polyneuropathy.

Conclusions: Three out of the 50 patients included in the study had unexplained chronic axonal polyneuropathy. Chronic axonal polyneuropathy could be part of the ALS disease spectrum. Understanding the link between these pathologies could open new directions in research and treatment.