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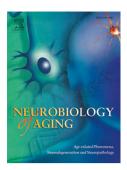
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#### ACCEPTED MANUSCRIPT

C9orf72 hexanucleotide repeat expansions and Ataxin 2 intermediate length repeat expansions in Indian patients with amyotrophic lateral sclerosis

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#### **Abstract**

Repeat expansions in the C9orf72 gene have been recognized as a major contributor to amyotrophic lateral sclerosis (ALS) and frontotemporal dementia (FTD) in the Caucasian population. Intermediate length repeat expansions of CAG (polyQ) repeat in the ATXN2 gene have also been reported to increase the risk of developing ALS in North America and Europe. We screened 131 ALS patients and 127 healthy controls from India for C9orf72 and ATXN2 repeat expansions. We found pathogenic hexanucleotide expansions in 3 of the 127 sporadic ALS patients, in 1 of the 4 familial ALS patients and in none of the healthy controls. In addition, our findings suggest that the 10-bp deletion that masks detection of C9orf72 repeat expansion does not explain the low frequency of this repeat expansion among Indian ALS patients. Intermediate length polyQ expansions (27Qs - 32Qs) in the ATXN2 gene were detected in 6/127 sporadic ALS patients and 2/127 of the healthy controls. Long ATXN2 polyQ repeats ( $\geq 33Qs$ ) were not present in any of the ALS patients or controls. Our findings highlight the need for large-scale multi-center studies on Indian ALS patients to better understand the underlying genetic causes.

#### **Keywords**

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