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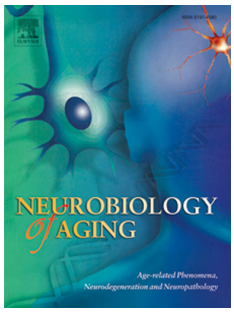

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Priyam Narain, James Gomes, Rohit Bhatia, Inder Singh, Perumal Vivekanandan

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

Priyam Narain[a], James Gomes[a], Rohit Bhatia[b], Inder Singh[b], and Perumal Vivekanandan[a]*

[a]*Kusuma School of Biological Sciences, Indian Institute of Technology Delhi, New Delhi, India*

[b]*All India Institute of Medical Sciences, New Delhi, India*

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 (≥ 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