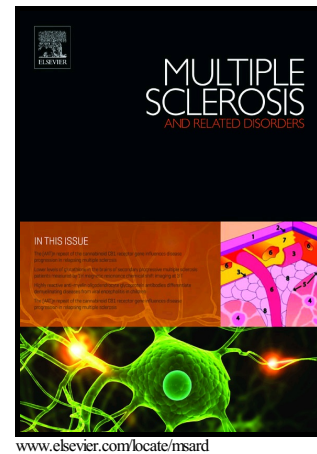


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PII: S2211-0348(17)30143-8
DOI: <http://dx.doi.org/10.1016/j.msard.2017.06.010>
Reference: MSARD589

To appear in: *Multiple Sclerosis and Related Disorders*

Received date: 28 February 2017
Revised date: 11 June 2017
Accepted date: 19 June 2017

Cite this article as: Miguel Oliveira Santos, Inês Caldeira, Marta Gromicho, Ana Pronto-Laborinho and Mamede de Carvalho, Brain white matter demyelinating lesions and amyotrophic lateral sclerosis in a patient with C9orf72 hexanucleotide repeat expansion, *Multiple Sclerosis and Related Disorders* <http://dx.doi.org/10.1016/j.msard.2017.06.010>

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Brain white matter demyelinating lesions and amyotrophic lateral sclerosis in a patient with C9orf72 hexanucleotide repeat expansion.

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Abstract

A hexanucleotide repeat expansion in the *C9orf72* gene is associated with amyotrophic lateral sclerosis (ALS) and frontotemporal lobar degeneration. It has been described before four patients with multiple sclerosis (MS) and C9orf72-ALS. However, C9orf72 positivity is not associated with increased risk of MS. Inflammatory pathways related to NF-κB have been linked to ALS and MS, and appear to be important in C9orf72-ALS patients. A 42-year-old woman presented with progressive bulbar symptoms for 9 months. Neurological examination disclosed spastic dysarthria, atrophic tongue with fasciculations, brisk jaw and limb tendon reflexes, and bilateral Hoffman sign. Electrophysiological assessment confirmed ALS. Brain MRI revealed multiple and bilateral juxtacortical and periventricular

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