ABSTRACT
Introduction: Mutations in the hexanucleotide repeat expansion in the C9orf72 gene are associated with frontotemporal dementia (FTD) and amyotrophic lateral sclerosis (ALS). Diagnosis is challenging due to highly variable penetrance. This case report describes a rare manifestation of C9orf72 associated-FTD that presented as Capgras delusion.

Case: This is a case of a 77-year-old male with no diagnosed medical history who was admitted to an inpatient psychiatric facility due to delusions that his wife had been replaced by an imposter. MRI of the brain revealed symmetric mild to moderate cortical atrophy. Patient had an extensive family history of FTD and ALS prompting genetic testing for the C9orf72 gene, which was abnormal for heterozygous pathogenic mutation of greater than 30 hexanucleotide repeats. Physical examination was significant for florid fasciculations, but no motor weakness or abnormal reflexes, thus meeting the clinical spectrum of motor neuron disease. An evaluation by a memory and aging specialist was consistent with dementia of mild stage. Clinical follow up is ongoing. The patient continues to have intermittent Capgras delusion with limited insight into his condition.

Conclusion: This case highlights the phenotypic diversity of C9orf72 gene.

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