A case of sporadic CJD presented with intestinal pseudo-obstruction: broadening the clinical spectrum of CJD.

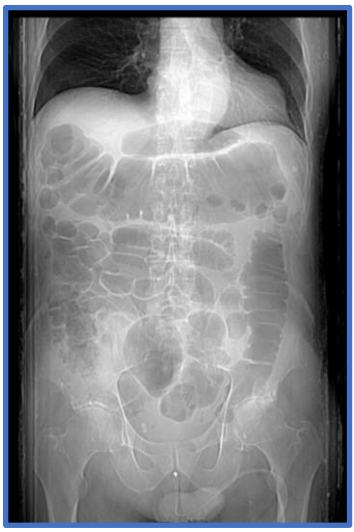
Francesca Vignaroli, V. Badioni, E. Domina, S. Fermi, M.E. Fruguglietti, S. Iurlaro, A. La Malfa, G. Mazza, MG. Pascarella, S.A. Sperber, A. Zilioli, V. Belcastro
Neurology Unit, Ospedale Maggiore di Lodi, Italy

INTRODUCTION

We report a case of sCJD presenting with intestinal pseudo-obstruction. Intestinal involvement in CJD is extremely rare. The main hypothesis is that prion protein, typically accumulating in the central nervous system, may also involve the peripheral nervous system.

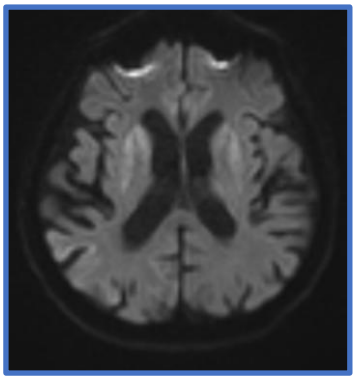
CASE PRESENTATION

A 76-year-old Caucasian man presented with a two-month history of postural instability, constipation and painless abdominal distension. At admission in Department of Neurology, he developed subacute intestinal pseudo-obstruction, followed by rapid neurological deterioration with cognitive decline, extrapyramidal signs, severe postural instability and hallucinations. He underwent comprehensive neurological and neuropsychological evaluations, MRI, CT scan, CSF analysis, EEG, and blood tests.



RESULTS

Investigation	Findings
Neuropsychological assessment	Deficits in praxis, attention, visuospatial functions
MRI (DWI)	Hyperintensities in basal ganglia, frontal, temporal, and parietal cortices
Full-body CT scan	No cancer lesions or obstruction points
EEG	Triphasic wave pattern
CSF analysis	↑ T-tau, ↑ P-tau, 14-3-3 negative, RT-QuIC positive



CONCLUSIONS

This case highlights a possible gastrointestinal involvement in prion disease. Prion protein accumulation has been reported beyond the CNS, including autonomic ganglia and peripheral nerves, but its role remains unclear. Further studies are needed to understand prion dissemination and the clinical impact of peripheral nervous system involvement.

24-28 Ottobre 2025
Padova Congress
55° CONGRESSO SOCIETÀ ITALIANA DI NEUROLOGIA