

A Case of Intranuclear Inclusion Disease in Adult Neurons Presenting with Urinary Retention and Cognitive Impairment

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Abstract

Neuronal intranuclear inclusion disease (NIID) is a rare neurodegenerative disorder characterized by eosinophilic intranuclear inclusions in both central and peripheral nervous systems, manifesting in diverse symptoms including cognitive decline, altered consciousness, limb weakness, seizures, and autonomic dysfunction. We present a case of a 62-year-old male with an 8-year history of cognitive decline and recent urinary retention. Neurological examination revealed comprehensive cognitive decline, bilateral pupil shrinkage, and limb weakness. Brain MRI demonstrated diffuse white matter hyperintensity adjacent to the cortex and characteristic subcortical ribbon sign on DWI, while electrophysiological studies indicated multiple peripheral nerve damages and autonomic dysfunction. Skin biopsy confirmed eosinophilic inclusions in fibroblasts and sweat gland cells, and genetic testing revealed GGC repeat amplification in the 5'UTR of the NOTCH2NLC gene, confirming the diagnosis of NIID. The patient was treated with donepezil for cognition, nutritive nerve agents, and intermittent catheterization due to urinary retention. This case highlights the importance of considering NIID in the differential diagnosis of complex neurodegenerative disorders, especially with characteristic neuroimaging findings and the recent advancement in genetic diagnosis. Although there is no definitive cure, early recognition and multidisciplinary management can optimize patient outcomes.

Keywords

Cognitive Impairment, Diffusion-Weighted Imaging, Neuronal Intranuclear Inclusions Disease, Urinary Retention