Rectal biopsy in the diagnosis of neuronal intranuclear hyaline inclusion disease.

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Neuronal intranuclear hyaline inclusion disease is a neurodegenerative disorder of childhood characterized by eosinophilic intranuclear inclusions and neuronal loss throughout the nervous system. Although most cases have been diagnosed postmortem, rectal biopsy may be diagnostic during life. We identified two patients: an 11-year-old boy (P1) with new-onset bulbar weakness and parkinsonism and a 15-year-old boy (P2) with severe cognitive and motor deterioration of uncertain etiology who presented at 4 years of age with a gait disturbance and motor slowing. Both patients had a history of behavioral problems marked by frequent temper tantrums. Both had nondiagnostic magnetic resonance imaging of the head and metabolic work-ups. Rectal biopsy was diagnostic 1 year (P1) and 11 years (P2) after the initial evaluation. Rectal biopsies should be considered in children presenting with otherwise unexplained multisystem degeneration, particularly in the presence of both upper and lower motoneuron signs accompanied by behavioral problems.

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