

**46th ANNUAL DIAGNOSTIC SLIDE SESSION, 2005  
REFERENCES AND DIAGNOSES**

**MODERATOR: E. Tessa Hedley-Whyte, M.D.**

**EDITOR: Leroy R. Sharer, M.D.**

**Case 2005-3**

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**Diagnosis: Neuronal intranuclear inclusion disease (NIID), juvenile-  
sporadic**

**Comment:** In addition to the inclusions, there was nerve cell loss in the substantia nigra, the dentate nuclei of the cerebellum, the red nucleus, and the anterior horns of the spinal cord. The inclusions were negative on immunocytochemistry (ICC) for measles antigen, tau, and polyglutamine, but they were positive for ubiquitin and SUMO-1 (small ubiquitin-like modifier). On electron microscopy the inclusions were non-membrane bound, consisting of randomly arrayed filaments.

**References:**

Pountney DL, Huang Y, Burns RJ et al: SUMO-1 marks the nuclear inclusions in familial neuronal intranuclear inclusion disease. *Exp Neurol* 2003; 184:436-446.

Takahashi-Fujigasaki J: Neuronal intranuclear hyaline inclusion disease. *Neuropathology* 2003; 23:351-359.

Takahashi J, Tanaka J, Arai K, et al: Recruitment of nonexpanded polyglutamine proteins to intranuclear aggregates in neuronal intranuclear hyaline inclusion disease. *J Neuropathol Exp Neurol* 2001; 60:369-376.

**Note:** Dr. McFadden received the first O.T. Bailey-Helena Riggs Award for best presentation by a trainee at the Diagnostic Slide Session, voted by the Charter Members of the Diagnostic Slide Session and presented after the President's Symposium on Sunday, June 12, 2005. Also, this case has recently been published (*J Neuropathol Exp Neurol* 2005; 64:545-554).