A 15-year-old boy presented with a history of an early-onset spastic paraparesis that progressed toward a severe quadriparesis (video on the Neurology® Web site at www.neurology.org), hypokinesia and bradykinesia, dysphagia, dysarthria, and hypomimia. Delayed motor evoked potentials and corticobulbar tract signal abnormality on brain MRI (figure) suggested corticospinal tract involvement. Cognitive functioning was preserved (Leiter-R IQ 86). ALS2 gene sequencing detected a homozygous c.2992C>T (p.R998X) substitution in exon 18 and confirmed the diagnosis of infantile ascending hereditary spastic paralysis (IAHSP).1

IAHSP may be misdiagnosed as a static encephalopathy because of its slow progression. Children with slowly progressive quadriparesis should be tested for ALS2 gene mutations.2

AUTHOR CONTRIBUTIONS
Mario Mastrangelo, Pia Bernasconi, and Paola De Liso contributed to the design and the conceptualization of the study and to drafting the manuscript. Sara Bertino and Caterina Caputi contributed to recording the video and drafting the manuscript. Vincenzo Leuzzi contributed to revising the manuscript.

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REFERENCES

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Teaching Video NeuroImages: Clinical course of infantile ascending hereditary spastic paralysis
Mario Mastrangelo, Pia Bernasconi, Paola De Liso, et al.

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