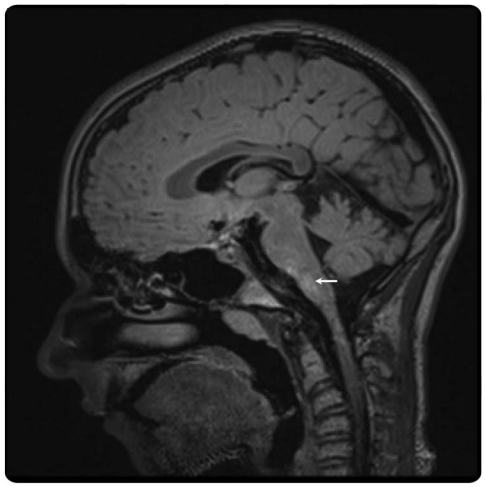
Teaching Video NeuroImages: Clinical course of infantile ascending hereditary spastic paralysis

Mario Mastrangelo, MD, PhD
Pia Bernasconi, PhD
Paola De Liso, MD
Caterina Caputi, MD
Sara Bertino, MD
Vincenzo Leuzzi, MD

Correspondence to Dr. Leuzzi: vincenzo.leuzzi@uniroma1.it

Figure Corticobulbar tract involvement in infantile ascending hereditary spastic paralysis



T1-weighted hyperintensity at the level of the corticobulbar pyramidal decussation (arrow) in a brain MRI at age 15.

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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DISCLOSURE

The authors report no disclosures relevant to the manuscript. Go to Neurology.org for full disclosures.

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Supplemental data at www.neurology.org

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From the Division of Child Neurology (M.M., P.D.L., C.C., S.B., V.L.), Department of Pediatrics, Child Neurology and Psychiatry, "Sapienza" University of Rome; and Neurology IV–Neuromuscular Diseases and Neuroimmunology Unit (P.B.), Neurological Institute Foundation "Carlo Besta," Milan, Italy.

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