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Teaching Video NeuroImages: Sodium channel myotonia can present with stridor

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An 11-month-old girl presented with episodic severe stridor from birth, often resulting in cyanosis. Her parents had noted recurrent unilateral ptosis. Later she developed spasms of her hands during exercise. Psychomotor development was normal. Neurologic examination showed generalized hypertonia. Cerebral MRI excluded a Chiari malformation. Laryngoscopy under general anesthesia showed no abnormalities. We observed retraction of one eye and ptosis, as shown on the video. The recognition of myotonia, confirmed by EMG, led to the diagnosis of a sodium channel myotonia,[1] with severe neonatal episodic laryngospasms.[2] A mutation in the *SCN4A* gene (c.3917G>A, p.Gly1306Glu) confirmed the diagnosis.[2]

AUTHOR CONTRIBUTIONS

Dr. Brandt-Wouters has contributed in concept and design of the manuscript. Dr. Klinkenberg and Dr. Roelfsema have contributed in critical revision of the manuscript for important intellectual content. Dr. Ginjaar has performed the DNA analysis and interpretation. Dr. Faber has contributed in critical revision of the manuscript for important intellectual content. Dr. Nicolai has contributed in concept and design of the manuscript.

STUDY FUNDING

No targeted funding reported.

DISCLOSURE

E. Brandt-Wouters, S. Klinkenberg, V. Roelfsema, I. Ginjaar, and C. Faber report no disclosures relevant to the manuscript. J. Nicolai has received travel grants for congress visits from UCB Pharma and Actelion. Go to Neurology.org for full disclosures.

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Supplemental data at www.neurology.org

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Neurology 2013;80;e108
DOI 10.1212/WNL.0b013e3182840c0b