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.

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Teaching Video NeuroImages: Sodium channel myotonia can present with stridor
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