



Teaching Video *NeuroImages*: Sodium channel myotonia can present with stridor



Evelyn Brandt-Wouters,
MD
Sylvia Klinkenberg, MD
Vincent Roelfsema, MD,
PhD
Ieke B. Ginjaar, PhD
Catharina G. Faber, MD,
PhD
Joost Nicolai, MD, PhD

Correspondence to
Dr. Nicolai:
j.nicolai@mumc.nl

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.

REFERENCES

1. Matthews E, Fialho D, Tan SV, et al; CINCH Investigators. The non-dystrophic myotonias: molecular pathogenesis, diagnosis and treatment. *Brain* 2010;133:9–22.
2. Lion-Francois L, Mignot C, Vicart S, et al. Severe neonatal episodic laryngospasm due to de novo *SCN4A* mutations: a new treatable disorder. *Neurology* 2010;75:641–645.

Neurology®

Teaching Video *NeuroImages*: Sodium channel myotonia can present with stridor

Evelyn Brandt-Wouters, Sylvia Klinkenberg, Vincent Roelfsema, et al.

Neurology 2013;80:e108

DOI 10.1212/WNL.0b013e3182840c0b

This information is current as of March 4, 2013

Updated Information & Services	including high resolution figures, can be found at: http://n.neurology.org/content/80/10/e108.full
Supplementary Material	Supplementary material can be found at: http://n.neurology.org/content/suppl/2013/03/03/80.10.e108.DC1
References	This article cites 2 articles, 1 of which you can access for free at: http://n.neurology.org/content/80/10/e108.full#ref-list-1
Subspecialty Collections	This article, along with others on similar topics, appears in the following collection(s): All Neuromuscular Disease http://n.neurology.org/cgi/collection/all_neuromuscular_disease All Pediatric http://n.neurology.org/cgi/collection/all_pediatric Muscle disease http://n.neurology.org/cgi/collection/muscle_disease Neonatal http://n.neurology.org/cgi/collection/neonatal
Permissions & Licensing	Information about reproducing this article in parts (figures, tables) or in its entirety can be found online at: http://www.neurology.org/about/about_the_journal#permissions
Reprints	Information about ordering reprints can be found online: http://n.neurology.org/subscribers/advertise

Neurology® is the official journal of the American Academy of Neurology. Published continuously since 1951, it is now a weekly with 48 issues per year. Copyright © 2013 American Academy of Neurology. All rights reserved. Print ISSN: 0028-3878. Online ISSN: 1526-632X.

