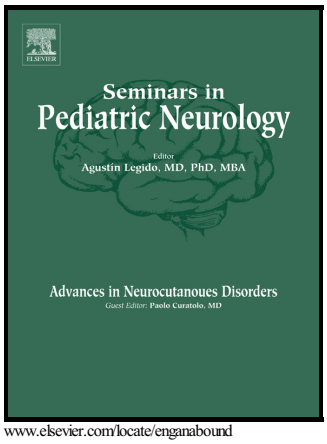

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PII: S1071-9091(17)30022-0
DOI: http://dx.doi.org/10.1016/j.spen.2017.03.006
Reference: YSPEN629

To appear in: *Seminars in Pediatric Neurology*

Cite this article as: Josefine Radke, Mona Dreesmann, Michael Radke, Arpad von Moers, Angela Abicht, Werner Stenzel and Hans H. Goebel, The Curse of Apneic Spells, *Seminars in Pediatric Neurology,* http://dx.doi.org/10.1016/j.spen.2017.03.006

The Curse of Apneic Spells

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Abstract

A 6-year-old girl had reduced fetal movements, numerous apneic spells, muscle hypotonia and developmental motor delay. Her muscle biopsy tissue showed variation in myofiber diameters, small minicores by electron microscopy, and near-uniformity of type I fibers. While no mutations were detected in *RYR1, SMN,* and *SMARD1* genes, the *RAPSN* gene revealed one known mutation, p.Asn88Lys, from the mother, and one novel mutation, p.Cys366Gly, from the father. Life-saving pyridostigmine treatment suppressed her apneic spells and improved her motor development.

Clinical features

During gestation this girl had reduced fetal movements, oligohydramnios, and retarded intrauterine growth. She was delivered by Caesarean section. After delivery she showed a prominent forehead, a deep-seated nose root, a high-arched palate, a tented upper lip, bilateral ptosis, brisk deep tendon reflexes, proximal muscle weakness, and generalized hypotonia.

When she was nine months old, she had had a respiratory arrest during a respiratory infection, requiring assisted ventilation in the intensive care unit. She also had