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**Format:** Abstract**Full text links**Neuromuscul Disord. 2017 Jun 27. pii: S0960-8966(17)30176-1. doi: 10.1016/j.nmd.2017.06.015. [Epub ahead of print]

## Congenital myopathy due to myosin heavy chain 2 mutation presenting as chronic aspiration pneumonia in infancy.

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### Abstract

A 7-week-old infant presented with persistent noisy breathing and aspirations during swallowing. Neurological examination and brain MRI were normal. His 12-year-old brother underwent pneumonectomy at the age of 10 years due to recurrent aspirations leading to severe lung damage. The older brother developed subsequently ophthalmoplegia and nystagmus along with mild weakness of the neck flexors and proximal muscles. Exome analysis revealed homozygosity for a novel truncating mutation p.G800fs27\* in the Myosin Heavy Chain 2 (MYH2) gene in both brothers, while parents and an unaffected sibling were heterozygous. A muscle biopsy from the older brother showed absence of type-2 muscle fibers and predominance of type-1 fibers. The aspirations causing pneumonia likely result from weakness of the laryngeal muscles, normally rich in type-2 fibers. The findings expand the phenotypic spectrum of MYH2 deficiency. MYH2 mutations should be included in the differential diagnosis of infants presenting with recurrent aspirations.

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**KEYWORDS:** Aspiration pneumonia; Congenital myopathy; MYH2; Myosin heavy chain

PMID: 28729039 DOI: [10.1016/j.nmd.2017.06.015](https://doi.org/10.1016/j.nmd.2017.06.015)

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