

PubMed



Format: Abstract

Full text links



Brain Dev. 2016 Mar;38(3):324-30. doi: 10.1016/j.braindev.2015.08.010. Epub 2015 Sep 9.

Respiratory management of patients with Fukuyama congenital muscular dystrophy.

Sato T¹, Murakami T¹, Ishiguro K¹, Shichiji M¹, Saito K¹, Osawa M¹, Nagata S¹, Ishigaki K².

Author information

Abstract

BACKGROUND: Fukuyama congenital muscular dystrophy (FCMD), characterized by intellectual impairment associated with cortical migration defects, is an autosomal recessive disorder caused by mutation in the fukutin gene. It is the second most common type of muscular dystrophy in Japan. Respiratory dysfunction, along with cardiomyopathy, can be life-threatening in patients with advanced-stage FCMD. However, few reports have focused on this issue.

METHODS: We retrospectively studied respiratory dysfunction and therapeutic management in 48 genetically diagnosed FCMD patients (mean age 11.0 years; range 3.6-31.9 years).

RESULTS: Mechanical ventilation was initiated at a median age of 12.1 years in 16 patients, 14 of whom received non-invasive positive pressure ventilation (NPPV) while the other 2 underwent tracheostomy with invasive ventilation (TIV). The two TIV cases had unexpectedly required the initiation of ventilatory support at the ages of 15.7 and 18.0 years, respectively, because of unsuccessful extubation followed by serious respiratory infections, despite rather good respiratory function before these episodes. Patients carrying a compound heterozygous founder mutation or with a severe phenotype tended to need ventilatory support 2-3 years earlier than homozygous patients and those with the typical or mild phenotype. Mechanical insufflation-exsufflation (MI-E) interventions were also employed in six patients with serious dysphagia and were well-tolerated in all cases.

CONCLUSION: For respiratory management, it is important to regularly evaluate respiratory function in FCMD patients over 10 years of age, since intellectual impairment and insomnia often mask the signs of respiratory dysfunction. Most patients, despite poor cooperation due to intellectual impairment, can tolerate NPPV and MI-E provided that a carefully worked-out plan is adopted.

Copyright © 2015 The Japanese Society of Child Neurology. Published by Elsevier B.V. All rights reserved.

KEYWORDS: Fukuyama congenital muscular dystrophy; Mechanical insufflation-exsufflation (MI-E); Mechanical ventilation; Non-invasive positive pressure ventilation; Respiratory management

PMID: 26363734 DOI: [10.1016/j.braindev.2015.08.010](https://doi.org/10.1016/j.braindev.2015.08.010)

[Indexed for MEDLINE]

Publication type, MeSH terms



LinkOut - more resources



PubMed Commons

[PubMed Commons home](#)

0 comments

[How to join PubMed Commons](#)