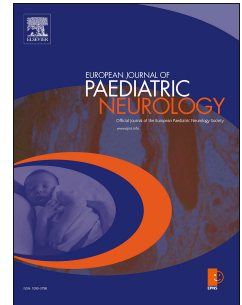


Accepted Manuscript

A novel homozygous MFN2 mutation associated with severe and atypical CMT2 phenotype

Giulia Iapadre, Giovanni Morana, Maria Stella Vari, Francesca Pinto, Paola Lanteri, Alessandra Tessa, Filippo Maria Santorelli, Pasquale Striano, Alberto Verrotti



PII: S1090-3798(17)31883-4

DOI: [10.1016/j.ejpn.2017.12.020](https://doi.org/10.1016/j.ejpn.2017.12.020)

Reference: YEJPN 2355

To appear in: *European Journal of Paediatric Neurology*

Received Date: 13 September 2017

Revised Date: 12 December 2017

Accepted Date: 28 December 2017

Please cite this article as: Iapadre G, Morana G, Vari MS, Pinto F, Lanteri P, Tessa A, Santorelli FM, Striano P, Verrotti A, A novel homozygous MFN2 mutation associated with severe and atypical CMT2 phenotype, *European Journal of Paediatric Neurology* (2018), doi: 10.1016/j.ejpn.2017.12.020.

This is a PDF file of an unedited manuscript that has been accepted for publication. As a service to our customers we are providing this early version of the manuscript. The manuscript will undergo copyediting, typesetting, and review of the resulting proof before it is published in its final form. Please note that during the production process errors may be discovered which could affect the content, and all legal disclaimers that apply to the journal pertain.

A novel homozygous MFN2 mutation associated with severe and atypical CMT2 phenotype

Giulia Iapadre¹, Giovanni Morana², Maria Stella Vari³, Francesca Pinto³, Paola Lanteri⁴, Alessandra Tessa⁵, Filippo Maria Santorelli⁶, Pasquale Striano³, Alberto Verrotti¹

Author Affiliations:

¹Department of Pediatrics, University of L'Aquila, L'Aquila, Italy

²Neuroradiology, Department of Head and Neck, "G. Gaslini" Institute, Genova, Italy

³Pediatric Neurology and Muscular Diseases Unit, Department of Neurosciences, Rehabilitation, Ophtalmology, Genetics, Maternal and Child Health, University of Genoa, "G. Gaslini" Institute, Genova, Italy

⁴Child Neuropsychiatry, Department of Head and Neck, "G. Gaslini" Institute, Genova, Italy

⁵Molecular Medicine and Neurogenetics, IRCCS Stella Maris, Pisa, Italy

⁶Child Neurology, IRCCS Stella Maris, Pisa, Italy

Corresponding author: Alberto Verrotti, Department of Pediatrics, University of L'Aquila, Via Vetoio, 1. Coppito (L'Aquila), Italy. Phone number: +39 0862/368726. E-mail address: alberto.verrottidipianella@univaq.it

Ethical Publication Statement: We confirm that we have read the Journal's position on issues involved in ethical publication and affirm that this report is consistent with those guidelines.

Abbreviations: CMT, Charcot-Marie-Tooth disease; CMT2A, Charcot-Marie-Tooth type 2A; MFN2, mitofusin 2; NCV, nerve conduction velocity; EMG, electromyography; CMAP, compound motor action potential; CTDP1, congenital cataracts facial dysmorphism; CCFDN, congenital cataracts facial dysmorphism and neuropathy; SIL1, Marinesco-Sjögren syndrome.

Download English Version:

<https://daneshyari.com/en/article/8684429>

Download Persian Version:

<https://daneshyari.com/article/8684429>

[Daneshyari.com](https://daneshyari.com)