

# Accepted Manuscript

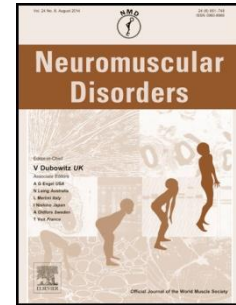
Title: Tubular aggregates in congenital myasthenic syndrome

Author: Amalia Feresiadou, Olivera Casar-Borota, Anca Dragomir, Carola Hedberg Oldfors, Erik Stålberg, Anders Oldfors

PII: S0960-8966(17)30379-6  
DOI: <https://doi.org/10.1016/j.nmd.2017.11.009>  
Reference: NMD 3476

To appear in: *Neuromuscular Disorders*

Received date: 11-4-2017  
Revised date: 15-10-2017  
Accepted date: 19-11-2017



Please cite this article as: Amalia Feresiadou, Olivera Casar-Borota, Anca Dragomir, Carola Hedberg Oldfors, Erik Stålberg, Anders Oldfors, Tubular aggregates in congenital myasthenic syndrome, *Neuromuscular Disorders* (2017), <https://doi.org/10.1016/j.nmd.2017.11.009>.

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.

## Tubular aggregates in congenital myasthenic syndrome

Amalia Feresiadou<sup>a,\*</sup>, Olivera Casar-Borota<sup>b,c</sup>, Anca Dragomir<sup>b,c</sup>, Carola Hedberg Oldfors<sup>d</sup>, Erik Stålberg<sup>e</sup>, Anders Oldfors<sup>d</sup>

<sup>a</sup> Department of Neuroscience, Uppsala University, Uppsala, Sweden

<sup>b</sup> Department of Clinical Pathology, Uppsala University Hospital, Uppsala, Sweden

<sup>c</sup> Department of Immunology, Genetics and Pathology, Uppsala University, Uppsala, Sweden

<sup>d</sup> Department of Pathology and Genetics, Institute of Biomedicine, University of Gothenburg, Gothenburg, Sweden

<sup>e</sup> Institute of Neurosciences, Uppsala University, Department of Clinical Neurophysiology, University Hospital, Uppsala, Sweden

\*Corresponding author. Department of Neuroscience, Uppsala University, Uppsala, Sweden. E-mail adress: [feresiadouamalia@gmail.com](mailto:feresiadouamalia@gmail.com), tel +46186115178, fax+4618502678

**Keywords:** CMS, tubular aggregates, DPAGT 1

A 20-year-old man with history of fluctuating muscle weakness since early childhood was referred to our department. There was no history of ptosis, double vision or dysarthria and no cognitive impairment. A muscle biopsy at the age of 8 years was inconclusive. Electromyography was performed at the age of 7 and 12 years with evidence of myopathy in proximal muscles. Neuromuscular transmission was not tested at that time. A muscle biopsy at age 17 showed myopathic changes, tubular aggregates and autophagic vacuoles. The tubular aggregates demonstrated

Download English Version:

<https://daneshyari.com/en/article/8689850>

Download Persian Version:

<https://daneshyari.com/article/8689850>

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