East African Scholars Journal of Medical Sciences
*Abbreviated Key Title: East African Scholars J Med Sci*
ISSN 2617-4421 (Print) | ISSN 2617-7188 (Online) |
Published By East African Scholars Publisher, Kenya

OPEN ACCESS

DOI: 10.36349/easms.2019.v02i03.013 Volume-2 | Issue-3 | March-2019 |

**Letter To The Editor**

# Motor Fibers Are Affected In MFN2 Carriers

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**Keywords:** HSAN, compound muscle action potential, electromyography.

In a recent article Wu *et al.*,(2018) presented a retrospective study of a 53yo male with pretended hereditary sensory and autonomic neuropathy (HSAN) due to the mutation c.776G>A in the MFN2 gene (Wu, R. *et al.*, 2018). We have the following comments and concerns.

We do not agree with the statement that neuropathy in the presented patient should be classified as HSAN and as a new phenotype (Wu, R. *et al.*, 2018). Among the two motor nerves undergoing nerve conduction studies, the patient had a reduced compound muscle action potential (CMAP) amplitude in the peroneal nerve. Reduction of the CMAP conforms with axonal loss on sural nerve biopsy, suggesting that there was also loss of motor axons. Since only two motor nerves had been tested and affection of other motor nerves thus might have been missed, it is crucial that additional motor nerves are investigated. It is also crucial that motor nerves are repeatedly investigated since there may be progression of the disease and involvement of motor fibers may develop later in the course. Disregarding these considerations, neuropathy in the index patient cannot be classified as HSAN since at least one motor nerve was affected.

An indirect indication for motor involvement could be the documentation of muscle wasting. However, the authors neither provide a description of muscle trophism when describing the neurological exam nor do they provide a figure of the leg muscles. Since muscle wasting is a frequent feature of hereditary neuropathies in general, we should be informed if there was reduction of muscle mass or not.

Another indicator for involvement of the motor nerves is needle-electromyography (EMG) (Kane, N. M., & Oware, A. 2012). However, in the results section no description of a needle-EMG is provided. Did the patient ever undergo needle-EMG and did it show a neurogenic pattern, suggesting affection of the supplying motor nerve, or was the needle-EMG normal?

Concerning the autonomic involvement, it would be interesting to know if there were autonomic disturbances other than anhidrosis of the hands, such as pupillary reflex abnormalities, sicca syndrome, decreased heart rate variability, orthostasis, obstipation, urinary bladder dysfunction, or impotentia. Were tests for autonomic involvement, such as the sympathetic skin response, tilt test, sweating test, SUDOSCAN, or fast Fourier transformation (FFT) of the heart variability on Holter recordings carried out? Which were the results?

Since MFN2 mutations may not only manifest as CMT2A, but also as optic atrophy, multiples lipomatosis, hypoacusis, vocal cord paralysis, abnormal glucose tolerance, spasticity, leucoencephalopathy, mimicking multiple sclerosis (Klein, C. J. *et al.*, 2011), learning problems, and cortical atrophy (Genari, A. B. *et al.*, 2011) (CMT2A plus), the index case and all first degree relatives need to be prospectively investigated not only for neuropathy, but also for CMT2A plus. Additionally, we should be informed about the results of the genetic work-up of all first-degree relatives. Did any of the first-degree relatives also carry the mutation?

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Journal homepage:
http://www.easpublisher.com/easjms/

Article History
Received: 18.02.2019
Accepted: 27.02.2019
Published: 14.03.2019

Overall, this interesting case could be more meaningful if the study design would have been a prospective one, if more than two motor nerves would have been investigated, if follow-up nerve conduction studies would have been carried out, if muscle trophism would have been described, and if first-degree relatives would have been investigated for CMT2A plus and for the MFN2 mutation. .

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