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Letter To The Editor

Mitochondrial disorder due to MTFMT mutations

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We want to thank Dr. La Piana *et al.*, for their enthusiastic reply (La Piana, R. *et al.*, 2017) to our letter (Finsterer, J., & Zarrouk-Mahjoub, S. 2017) concerning their previous article (LaPiana, R. *et al.*, 2017). Contrary to what is pretended by Dr. La Piana *et al.*,(2017) our letter (Finsterer, J., & Zarrouk-Mahjoub, S. 2017) does not all lack substance, the background literature was carefully studied, and there is inconsistency, as documented below.

In the case description the post-chiasmatic white matter lesions are described as "mildly asymmetric" and made responsible for homonymous hemianopsia to the left, whereas in the discussion white matter lesions are described as "bilateral, symmetric". Furthermore, La Piana *et al.*, diagnosed "bilateral visual loss" and later in the case description "left homonymous visual field deficits" (LaPiana, R. *et al.*, 2017), which is contradictory.

In their original article Dr. La Piana *et al.*,. described that the 4th cranial nerve palsy "improved spontaneously over time", suggesting that it did not resolve completely, but in their reply they mention that the 4th cranial nerve palsy "resolved during infancy". Attributing the 4th cranial nerve lesion to the cerebral lesions is misleading since these lesions are described as progressive, whereas the 4th cranial nerve lesion "resolved already during infancy" (LaPiana, R. *et al.*, 2017).

Concerning the visual impairment, we want to add that it is not at all excluded that optic neuropathy contributed to visual impairment. Optic atrophy was first observed 6 months after "bilateral visual loss" at age 18y. However, visual impairment due to affection of the retinal ganglion cells may not necessarily go along with immediate optic nerve atrophy, particularly in the early stages. Visually-evoked potentials may be delayed in pre- and post chiasmatic visual tract lesions. Optic nerve atrophy not necessarily needs to be visible on fundoscopy, particularly if the posterior parts of the nerve are affected. Dr. La Piana *et al.*, (2017) mention in their reply (La Piana, R. *et al.*, 2017) that OCT at age 24y showed "thinning of the peripapillary retinal nerve fiber layer" but we do not know if this was already the case at age 18y.

We do not agree with the statement that "there is no proven treatment for LHON". At least one double-blind, placebo-controlled study showed a beneficial effect in LHON (Klopstock, T. *et al.*, 2011).

Concerning the term "quadruspasticity" it is a matter of definition if generalised hyperreflexia and positive pyramidal signs are already regarded as quadruspasticity or only if muscle tone is increased. We use the term "quadruspasticity" according to the former definition.

Overall, we appreciated the discussion about this interesting case, which hopefully, contributes to a better understanding of some imponderables.

REFERENCES

1. La Piana, R., Weraarpachai, W., Ospina, L.H., Tetreault, M., Majewski, J. Care, R.C. C., Bruce, P. G., Decarie, J.C., Tampieri, D., Brais, B., Shoubridge, E.A. (2017). Reply to the letter by Finsterer and Zarouk-Mahjoub. Neurogenetics, (submitted).

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2. Finsterer, J., & Zarrouk-Mahjoub, S. (2017). Mitochondrial disorder due to MTFMT mutations. Neurogenetics, (submitted).
3. LaPiana, R., Weraarpachai, W., Ospina, L.H., Tetreault, M., Majewski, J. Care4Rare, C.C., Bruce, P. G., Decarie, J.C., Tampieri D, B.B., & Shoubridge, E.A. (2017). Identification and functional characterization of a novel MTFMT mutation associated with selective vulnerability of the visual pathway and a mild neurological phenotype. Neurogenetics, 48-16.
4. Klopstock, T., Yu-Wai-Man, P., Dimitriadis, K., Rouleau, J., Heck, S., Bailie, M., Atawan, A., Chattopadhyay, S., Schubert, M., Garip, A., Kernt, M., Petraki, D., Rummey, C., Leinonen, M., Metz, G., Griffiths, P.G., Meier, T., Chinnery, P.F. (2011). A randomized placebo-controlled trial of idebenone in Leber's hereditary optic neuropathy. Brain, 134, 2677-86.