• Comment •

Retinal nerve fiber and ganglion cell layer thinning in hereditary and acquired mitochondrial optic neuropathies

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Dear Editor,

With interest we read the article by Teng *et al*[1] about a study of the retinal nerve and ganglion cell layers by means of optic coherence tomography (OCT) in 32 patients with a mitochondrial optic neuropathy (MON). Included were 20 patients with hereditary MON [Leber's hereditary optic neuropathy (LHON)], 12 patients with acquired MON [ethambutol-induced optic neuropathy (EION)], and 41 healthy controls. Retinal nerve fiber layer (RNFL) thickness was reduced in the nasal, superior, temporal, and inferior quadrants in LHON patients but only in the temporal quadrant in the EION patients. Thickness of the retinal ganglion cell layer (RGCL) was similarly reduced in LHON and EION patients. We have the following comments and concerns.

Among the 32 patients with MON, 20 patients were diagnosed with hereditary MON (LHON) and 12 patients were diagnosed with secondary, acquired MON[1]. Surprisingly, Table 1 lists among the 12 patients with EION, 3 patients with an *OPA1* mutation and 1 patient with an *NDxx* mutation. Does it mean that 4 patients with EION also carried a mutation causing hereditary MON? Since it was the aim of the study to compare RNFL and RGCL thickness between primary and secondary MON patients, mixing patients with primary and secondary MON in the EION group is confusing and not acceptable.

One patient with human immunodeficiency virus (HIV) was included among the EION group. We should know if he also had tuberculosis and underwent ethambutol treatment or if optic neuropathy in this patient was attributable to other causes including anti-retroviral therapy induced optic neuropathy[2].

Another shortcoming of the study design is that the LHON cohort was small and not coherent. Twelve patients carried the variant m.11778x>x, 5 patients the variant m.14484x>x, and one patient each the variants m.3260x>x, m.14568x>x and m.11696x>x respectively. Unfortunately, the nucleotides being substituted were not provided.

A further shortcoming is that heteroplasmy rates of the detected mtDNA variants was not provided. Phenotypic expression depends at least in part on the amount of mutated mtDNA within a mitochondrion[3]. In this respect it should be mentioned how many of the 20 LHON patients had a positive family history for LHON and in how many patients the mutation occurred sporadically.

Another shortcoming is that tissues other than the optic nerve were not investigated. Since it is well appreciated that LHON is a multisystem mitochondrial disorder (MID) already at onset of the disease or becomes a multisystem disease during the further course[4], it is crucial that these patients undergo prospective investigations for multisystem disease. This is of relevance as involvement of other organs in LHON particularly the brain and the heart may strongly influence the outcome of these patients.

Overall, this interesting study could be more meaningful if more profound genetic information would have been provided, if the family history of LHON patients would have been mentioned, and if the study groups would have been more coherent.

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Conflicts of Interest: Finsterer J, None.

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