Letter to the Editor

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The spectrum of neuro-ophthalmologic involvement in mitochondrial disorders is broad

With interest we read the article by Lock *et al*. about the neuro-ophthalmologic involvement in mitochondrial disorders (MIDs).[1] Neuro-ophthalmologic abnormalities addressed in the review were progressive external ophthalmoplegia (PEO) with or without ptosis, cataract, macula pattern dystrophy, pigmentary retinopathy, optic neuropathy, and retro-chiasmal visual loss.[1] It was concluded that the treatment of ophthalmologic involvement in MIDs is largely supportive and symptomatic, but that this will likely change over the coming years and is already occurring in the realms of family planning.[1] The study is appealing but raises the following comments and concerns.

We do not agree with the notion that cataract is not a prominent feature of MIDs.[1] On the contrary, cataract is a striking feature of MIDs and has been reported in syndromic and nonsyndromic MIDs. Among the syndromic MIDs, cataract has been reported in MELAS, Leigh syndrome,[2] Kearns-Sayre syndrome (KSS),[3] autosomal dominant optic atrophy (ADOA), Wolfram syndrome, Leber's hereditary optic neuropathy (LHON), myoclonic epilepsy with ragged-red fibers syndrome, mitochondrial diabetes and deafness (MIDD), and PEO.[4] Cataract has been also reported in several nonsyndromic MIDS.[5]

Another ophthalmologic manifestation of MIDs not extensively discussed is glaucoma.[4] Glaucoma has been reported in MELAS, KSS, LHON, ADOA, myo-neuro-gastro-intestinal encephalopathy (MNGIE), Wolfram syndrome, pontocerebellar hypoplasia (PCH),[4] and several nonsyndromic MIDs.

A further neuro-ophthalmologic manifestation of MIDs not discussed is migraine. Migraine or migraine-like headache is a common manifestation of various syndromic and nonsyndromic MIDs, such as MELAS, MNGIE, or KSS.[6] Particularly in patients with auravisual phenomena, such as scotoma, can migraine be a dominant feature.

Another neuro-ophthalmologic abnormality not addressed in the review is nystagmus. Since cerebral involvement, including the cerebellum, is common in MIDs, several of the syndromic but also nonsyndromic MIDs present with nystagmus. Among the syndromic MIDs, nystagmus has been reported in Leigh syndrome, Alpers-Huttenlocher disease, LHON, MELAS, and ADOA.[4]

There is no mentioning of choroideal atrophy as has been reported in MELAS, MIDD, Mohr-Tranebjaerg syndrome, and nonsyndromic MIDs.[4] Keratoconus has been particularly reported in nonsyndromic MIDs.[4] Patients with PCH may present with a megaloconrea.[4] Iris atrophy has been found in patients with MELAS.[4] Whether pupillary dysfunction in LHON or nonsyndromic MIDs is due to involvement of the autonomic nervous system or due to involvement of the sphincter pupillae muscles remains speculative.

Overall, the review highlights important neuro-ophthalmologic manifestations of MIDs but does not cover the entire spectrum of neuro-ophthalmologic disease in MIDs. Missing are glaucoma, nystagmus, migraine, choroideal atrophy, megalocornea, pupillary dysfunction, and keratoconus.

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Conflicts of interest

The authors declare that there are no conflicts of interests of this paper.

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