

Response to Newman *et al.*

To the Editor: We thank Dr Newman and colleagues¹ for their careful reading of our article² and for their additional recommendations.

We agree that creating a single set of guidelines covering the multitude of systemic and neurological manifestations of mitochondrial diseases is difficult, and that the recommendations will inherently not apply to every situation or every patient. However, the Delphi methodology we used allowed us to synthesize what were sometimes diverse views, and thereby to present a consensus based on the experience of a panel of mitochondrial disease physicians. This approach has been shown to be most valuable when there is limited objective evidence base, as for mitochondrial disorders.

In regard to the concern of whether a neuro-ophthalmologist is required for routine care of these patients, the writing group considered that such a specialist, if accessible by the patient, would be the ideal physician to monitor the ophthalmological manifestations of the disease. A neuro-ophthalmologist would also be the best person to determine the need for subspecialty ophthalmology referrals. It is important to note that this was only a recommendation, and not considered to be essential for each and every patient.

With regard to the monitoring of intraocular pressure, while it should be an integral part of every general eye exam, some members of the consortium have noted that this portion of the examination is not always performed in some pediatric patients. For this reason, an additional statement was included to recommend this testing be done when necessary.

We agree that lubrication is required to prevent the keratopathy that occurs with corneal exposure in some mitochondrial patients, and not only due to the inappropriate spread of tears. We also agree that it would be wise for affected and unaffected Leber hereditary optic neuropathy (LHON) mitochondrial DNA mutation carriers to avoid cigarette smoking and excessive alcohol use, although the evidence that well-established LHON deteriorates with smoking or excess alcohol is limited. The seventh recommendation was included to alert the clinician to the presence of LHON plus phenotypes, with some authors feeling that a periodic neurologic exam may detect preclinical symptoms, and a follow-up electrocardiogram may detect a pre-excitation syndrome not apparent on the first examination.

It is important to stress that our recommendations were not a universal view held by all of the authors but a consensus statement reflecting the majority. This approach has both strengths and weaknesses. Inevitably this means that the outcome of the review was dependent on the authors who engaged the process. To address this, we made an open

invitation to interested clinician-members of the Mitochondrial Medicine Society to participate. However, most importantly, the consensus describes a “current state of play,” which definitely should be revised as additional objective information comes to hand.

We therefore welcome their additional comments, and look forward to incorporating these suggestions in any future revised version of the criteria.

DISCLOSURE

The authors declare no conflict of interest.

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