The Nerve! Readers Speak

Reader response: Consensus-based care recommendations for adults with myotonic dystrophy type 1

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In the recently published consensus-based care recommendations for adults with myotonic dystrophy type 1 (DM1),¹ visual symptoms caused by retinal dysfunction are not considered despite several studies reporting frequent abnormal electroretinograms (ERGs) in patients with DM1.^{2–4}

Because ERGs provide easily accessible and noninvasive objective information about the electrical activity of cells within the retina in response to light stimulation, they are of considerable interest to assess CNS alterations associated with DM1. Of note, even with no changes in ophthalmoscopy, ERGs in patients with DM1 and asymptomatic parents were shown to be moderately affected like that seen in early retinitis pigmentosa. Importantly, both rod and cones systems can be assessed when testing with scotopic low intensity or dim blue or red stimulation.⁵ Assessment of such changes can be crucial as they are consistent with complaints recorded in patient with DM1 support group including longer adaptation to darkness and struggle in dim-light environment such as movie theaters. Such perturbations are often associated with increased risks of falling and driving difficulties, especially at dusk. Altogether, this suggests that ERG screening should be considered as a potential biomarker for patients with DM1 and warrants the interdisciplinary involvement of neurologists and ophthalmologists.

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We thank Brignol and Fort for their comments on our article.¹ Ophthalmology played a critical role in the discovery of anticipation involving lens opacities in myotonic dystrophy type 1 (DM1) as early as the 1990s. Retinopathy has been recognized in the literature since 1918. We thank you for reminding us that retinopathy is a part of multisystemic phenotype of this disease. Eyes have also shown other ocular impairments in DM1. At present, we do not know the impact of retinopathy on daily life of patients with DM1. The validation of retinopathy in patient-reported outcome measures is confounded by the presence of other ocular

abnormalities in DM1, such as lens opacities. However, we agree that retinopathy could be an objectively measurable biomarker in clinical trials of DM1. Further studies to develop electroretinogram and other retinal functional measures in correlation with the clinical or molecular measures of DM1 need to be considered. Bringing back ophthalmologists to the field of DM is very welcome.

1. Ashizawa T, Gagnon C, Groh WJ, et al. Consensus-based care recommendations for adults with myotonic dystrophy type 1. *Neurol Clin Pract* 2018;8:507–520.

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