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New BEST1 mutations in autosomal recessive bestrophinopathy

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Reply

We thank Boon et al. for their interest in our article (Fung et al., New BEST1 mutations in autosomal recessive bestrophinopathy. Retina 2015;35(4):773–82)¹. Their research has helped to advance our understanding of bestrophinopathies. Most cases of autosomal recessive bestrophinopathy exhibit visual impairment on presentation.^{1,2} This may be multifactorial in etiology and include: cystoid maculopathy, subretinal fluid, vitelliform material or fibrosis, atrophy and choroidal neovascularization. Although Boon et al.² detected progressive photoreceptor dysfunction on full field electroretinogram in some but not all of their patients, a “relatively constant clinical picture with relatively stable visual acuity was seen in all individuals”, consistent with our findings and those of others^{3,4}. This suggests that the electrophysiological progression is not necessarily associated with functional decline. It is interesting that of the five patients described in both our papers who received oral acetazolamide for cystoid maculopathy, only one patient demonstrated a response. Given that the response in this patient was dramatic and rapid, we feel that the lack of response in our 3 patients cannot be ascribed to the shorter follow-up period, as suggested. It is possible that either the cystic maculopathy was too chronic to be reversed or the dosage prescribed (500mg daily) was too low, since the patient who responded in Boon et al.’s² study only did so at a 250mg tid, not a 125mg tid dosage.

References

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