

International Conference on Eye Disorders and Treatment

July 13-15, 2015 Baltimore, USA

Vision-related quality of life improvement after implantation of Boston keratoprosthesis

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The introduction of the type I Boston Keratoprosthesis (KPro) by Dohlman and associates in 1974 was a major development in the management of patients with corneal disease and poor prognosis for traditional penetrating keratoplasty. The Boston keratoprosthesis has benefited from a great increase in popularity in the last several years due to the continuous improvements in its design as well as changes in postoperative management introduced since the FDA approval in 1992. The favorable outcomes reported in most studies of Boston keratoprosthesis are definitely encouraging, but are mostly focusing on the corneal surgeon's definition of success, based on excellent device retention, good visual acuity results, and acceptable complication rates. However, a correlation with patient's performance of daily activities is lacking. Some studies have shown that the perceived benefit of treatment by patients may differ from what the ophthalmologist defines as success and therefore, taking into consideration the subjective perception of patient's own disease and visual function is imperative in the care of these patients. In this prospective study, we utilized the NEI VFQ-25 to determine the impact of Boston KPro implantation on patient-reported visual function. We specifically targeted two questions: Does the quality of life of patients implanted with Boston KPro change significantly over the course of their post-operative treatment, and do patients who have unilateral disease or better vision in the contralateral eye also show improvement in vision related quality of life measures?

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Using stem cells to understand and treat diseases due to mutations in BEST1

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Mutations in BEST1 encoding Bestrophin 1 (Best1) causes the bestrophinopathies, a group of 5 retinal degenerative diseases including Best vitelliform macular dystrophy and autosomal recessive bestrophinopathy (ARB). Best1 is a Ca²⁺ activated anion channel (CAAC) that also regulates Ca²⁺ homeostasis. In the eye Best1 is expressed uniquely in retinal pigment epithelial (RPE) cells. Why over 200 different mutations in Best1 cause 5 distinct diseases is not clear nor is there an obvious path to treatment. To address this we are generating a bank of skin fibroblasts from patients with bestrophinopathies. The fibroblasts are reprogrammed to induced pluripotent stem cells (iPSCs) which are differentiated into retinal pigment epithelial (RPE) cells for study in the laboratory. Among our study participants are a 14 year old female with ARB and her parents. The ARB patient is compound heterozygous for Best1R141H and Best1I366fsX18. Whole cell patch clamp analysis of these Best1 mutants indicates that Best1R141H Cl⁻ currents are impaired but that Best1R141H does not impair wild type currents. Best1I366fsX18 exhibits normal currents which are not impaired when co-expressed with Best1R141H. In iPSC derived RPE cells, neither mutant is mislocalized. These data suggest that neither loss of Best1 CAAC activity nor mislocalization of Best1 play significant roles in the pathogenesis of ARB. We are currently examining the effects of ARB mutations on Ca²⁺ stores and other RPE functions with the goal of using iPSC derived RPE to test the efficacy of gene augmentation, gene repair and small molecule drugs in rescuing the ARB phenotype.

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