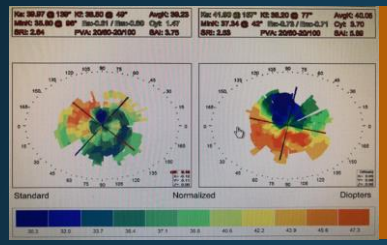

Scleral contact lenses aiding in the treatment of Aniridia

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Background

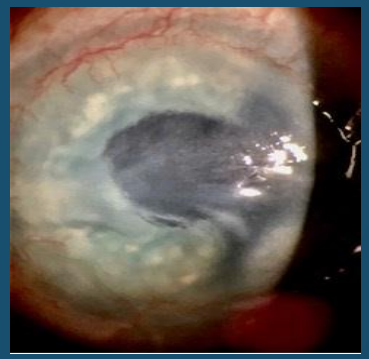
A 46-year-old Caucasian male presented to the University of Virginia Genetic Disorder Clinic, with a primary interest in discussing alternative corneal treatments. The patient has an ocular history of foveal hypoplasia, nystagmus, pseudophakia with bilateral artificial iris implants, and bilateral keratolimbal allografts (KLAL). In 2010, KLAL were failing resulting in the progression of his aniridic keratopathy. His current medications include mycophenolate mofetil and Refresh artificial tears as needed. The patient was previously using topical cyclosporine and difluprednate; however, he was instructed to discontinue these medications in March 2017 due to their ineffectiveness. His mother and both of his sisters also have aniridia.

Discussion

Aniridic keratopathy is thought to stem from limbal stem cell deficiency secondary to PAX6 gene mutations. The PAX6 genetic defect influences the regulation of stem cells, resulting in the impaired function of basal limbal stem cells and basal corneal epithelial cells. The corneal epithelial cells have numerous adhesion mechanisms, such as tight junctions and desmosomes, as well as a large variety of adhesion molecules, such as catenins, integrins, and desmogleine. Research has indicated a link between the PAX6 gene mutation and a reduction in desmogleine, as well as beta- and alpha-catenin. The reduction in these adhesion molecules results in poor epithelial cell adhesion, leading to the biochemical and pathological changes seen in aniridic keratopathy. This results in the corneal surface becoming very fragile, and it weakens the function of the epithelial barrier.

Case Description

Scleral contact lenses have been reported to be effective in managing patients with limbal stem cell deficiency. This can be attributed to their design, which unlike corneal designs completely vaults over both the cornea and the limbus, protecting these structures from any further damage or desiccation. With this design, the limbus and corneal epithelium are continually hydrated due to the saline fluid reservoir, and they are protected from mechanical forces from the eyelids during blinking.
In a case that Schornack reported, a patient with clinically diagnosed limbal stem cell deficiency was not responding to maximal topical and systemic medical therapy. Upon the initiation of treatment with scleral contact lenses, this condition rapidly improved, and the results were maintained even after 18 months of discontinuing lens wear. This suggests that intervention with scleral contact lenses allows for some regenerative potential of limbal stem cells.



Conclusion:

The management of aniridic keratopathy continues to be a challenge for eye care providers; however, the use of scleral contact lenses may provide both ocular surface protection and symptomatic relief for the patient. Scleral contact lenses can be offered as a non-surgical intervention in the management of this condition. However, additional research is needed to further evaluate their efficacy and the mechanism by which they improve limbal stem cell regeneration.

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