An Unusual Form of Avellino Dystrophy after Laser in situ keratomileusis: A Late Onset or Recurrence?

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Abstract

Purpose: To report an unusual manifestation of a corneal deposit of Avellino corneal dystrophy (ACD) after bilateral, simultaneous laser in situ keratomileusis (LASIK).

Methods: A 36-year-old Korean woman who underwent uncomplicated, bilateral LASIK and repeated phototherapeutic keratectomy (PTK) in her left eye due to corneal opacities, showed numerous, fine, white opacities with central corneal stromal haze in the left eye, which was the typical manifestation of the recurrent form of ACD. However, in the right eye, several discrete white opacities were deposited in the central anterior stroma, which was a morphologic feature of the natural course of ACD.

Results: The heterozygous R124H (CGC→CAC) mutation of the BIGH3 gene was found in her genomic DNA extraction.

Conclusion: Further studies should be focused on what is responsible for the differences of the onset period and the shapes of the deposits in patients with ACD.

Keywords: Avellino corneal dystrophy; Laser in situ keratomileusis; Phototherapeutic keratectomy

Introduction

Granular dystrophy type II (Avellino corneal dystrophy, ACD) is an autosomal dominant corneal stromal disease that shares features of both granular and lattice corneal dystrophies. This disorder is caused by a R124 mutation in the TGFBI gene, which is activated by transforming growth factor (TGF)-β [1-4].

Recent studies have shown that laser in situ keratomileusis (LASIK) aggravates corneal deposits in patients with exacerbated ACD and so LASIK should be avoided in these patients [5-8]. All of the exacerbated corneal deposit of ACD after LASIK in the literature showed multiple, fine, extensive opacities in the anterior stroma, and they were mainly concentrated in the LASIK flap interface with or without diffuse central corneal stromal haze. The manifestations of the recurred, or secondary form of ACD is significantly different from the natural-onset, or primary form for the morphological features.

We report here on an unusual manifestation of a corneal deposit of ACD after bilateral, simultaneous LASIK.

Case Report

A 36-year-old Korean woman reported blurred vision of both eyes for 6 years. She underwent uncomplicated, bilateral LASIK that was performed elsewhere 8 years ago in other clinic. Decreased visual acuity occurred 24 months after LASIK surgery, and two additional phototherapeutic keratectomy (PTK) procedures were done in her left eye at the same clinic. Preoperatively, the spherical equivalent manifest refraction was -3.50 diopter in the right eye and -3.00 diopter in the left eye, yielding 20/20 bilateral BSCVA. Slit-lamp examination showed numerous, fine, white opacities with central corneal stromal haze in the left eye, which was the typical manifestation of the recurrent form of ACD after LASIK. However, in the right eye, several discrete white opacities were deposited in the central anterior stroma, which was a morphologic feature of the natural course of ACD. There was no signs of inflammation, edema or thinning, and no other ocular abnormalities were noted. A pedigree analysis and slit-lamp examination of her parents and siblings showed no family history of corneal dystrophy.

After informed consent was obtained, genomic DNA was extracted from the peripheral leukocytes of the patient and the heterozygous R124H (CGC→CAC) mutation of the BIGH3 gene was found.

Twenty four months after the surgery, the patient reported glare and visual discomfort, and especially in the left eye. Her BSCVA was 20/20 in both eyes and the ophthalmologist found a few white granules on the anterior stroma, and this was worse in the left eye. The surgeon performed PTK two times at 2 and 4 years after the previous LASIK in the left eye to remove the corneal deposit. The visual disturbance was improved immediately after the PTK; however, the corneal deposits were exacerbated after a few months. The patient was referred to our clinic for consultation.

On her first visit, the manifest refraction was +1.00 -1.75 x 170 OD and +1.25 -1.00 x 50 OS, yielding 20/20 BSCVA in both eyes. Slit-lamp examination showed numerous, fine, white opacities with central corneal stromal haze in the left eye, which was the typical manifestation of the recurrent form of ACD after LASIK. However, in the right eye, several discrete white opacities were deposited in the central anterior stroma, which was a morphologic feature of the natural course of ACD. There was no signs of inflammation, edema or thinning, and no other ocular abnormalities were noted. A pedigree analysis and slit-lamp examination of her parents and siblings showed no family history of corneal dystrophy.

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is widely accepted that the diagnosis of Avellino dystrophy should be based on the morphological findings. Due to the development of gene analysis, it is now possible to genetically confirm ACD. However, the pathogenesis of ACD is unclear, except for the mutation in codon 124 of the BIGH3 gene (histidine replacing arginine), and this codes for the resultant TGFβ-induced cell adhesion protein keratoepithelin (68 Kda), which is responsible for the disease progression. The mechanism for the worsening of ACD after LASIK remains elusive. TGFβ is a well-known cytokine associated with BIGH3 protein and any insult to the cornea could be related to an increased TGFβ production and the resultant BIGH3 protein deposit. In our patient, a serial PTK induction may have played a role in the worsening of ACD.

The prevailing theory is that the stimulation of the mutated keratoepithelin protein in ACD corneas by LASIK seems to be independent of TFGβ [7]. Generally, the recurrence of ACD after LASIK is more severe than that after PRK or PTK. However, the epithelial basement membrane and Bowman's layer remained intact after LASIK surgery, and there was a minimal increase in TGFβ in the first few months and this became undetectable after only a few months. When we look at our patient from this point of view, we could conclude that the right eye showed the primary form of ACD in the natural course, and only the left eye showed the recurrent form of ACD after PTK. The morphological manifestation of the left eye was similar to the majority of the previously reported cases of recurrence after PTK.

BIGH3 mutation analysis may help to distinguish ACD from granular corneal dystrophy, yet for cases like ours, there is no other tool to differentiate the primary and the secondary forms of ACD. Hence, we should consider every possibility whether it is late onset with a natural course of ACD or it is a laser-induced recurrent form of ACD.

In summary, great care should be taken not to miss even the minimal evidence of ACD before performing LASIK. Although surface ablation procedures such as PTK have been considered effective methods for removing the opacities, the potential for recurrence and exacerbation of these deposits should be considered. Further studies should be focused on what is responsible for the differences of the onset period and the shapes of the deposits in patients with the primary and secondary forms of ACD, even though they share the same mutation in exon 4 (the R124H mutation) in the TFGβ gene.

**References**


