A case report of visual outcome in keratoconus with retinitis pigmentosa

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SUBJECT AREAS
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Abstract

Background: It is uncommon to see retinitis pigmentosa in keratoconus patients. The main difficulty of visual rehabilitation for this is restricted visual field. We presented the treatment and gene screen of visual system homeobox 1 (VSX 1) gene in this case. Case presentation: A 24-year-old man with retinitis pigmentosa presented with progressively blurred vision. Slit lamp examination revealed Vogt’s striae over both eyes, and corneal topography indicated bilateral keratoconus. We had tested 5 exons of VSX 1 gene from him and the did not find mutation on direct sequence. To improve visual acuity, we prescribed keratoconus rigid gas permeable (RGP) contact lens for him with good efficacy. However, lens dislocation occurred occasionally. He could not find dislocated lens easily due to visual field restriction, so he asked for more stable visual aids. Therefore, we instead prescribed scleral lens (SL), which were more stable on the ocular surface and led to more stable vision. Visual acuity was also gained with SL, but the tolerance time for SL was shorter compared to that of keratoconus RGP contact lens. To compare the efficacy of these two lenses, we surveyed life quality using the National Eye Institute Visual Functioning Questionnaire – 25 in three situations: baseline, with keratoconus RGP contact lens, and with SL. Conclusion: The patient used the two lens types according to his needs, and benefited from vision rehabilitation with both keratoconus RGP contact lens and SL. Keywords: keratoconus; retinitis pigmentosa; rigid gas permeable contact lenses; scleral lenses.

Background

Keratoconus is characterized on the anterior corneal surface by a cone-shape protrusion[1, 2], and the characteristic pattern of the corneal architecture is used for evaluating the severity of keratoconus. It is uncommon to see retinitis pigmentosa in keratoconus patients. These two diseases may be coexisted in Leber congenital amaurosis
(LCA), which was diagnosed by clinical features. The manifestation of LCA are various, including poor vision, nystagmus, multi-systemic involvement such as renal, cardiac, skeletal or neurology anomalies[3, 4].

Rigid gas permeable (RGP) contact lens and keratoconus RGP contact lens have been important tools for visual rehabilitation of patients with keratoconus[5], and there are various materials and designs of keratoconus RGP contact lens, e.g., Rose K lenses (Menicon Z material from Menicon CO., Ltd, Nagoya, Japan) and HiClear keratoconus RGP contact lens (Brighten optix company, Corp., Taipei, Taiwan). For severe cases, scleral lenses (SL) offer an alternative strategy [6, 7]. However, to the best of our knowledge, there were seldom reports to compare keratoconus RGP contact lens and SL in same patients with long follow-up time. Herein, we represent a case of severe keratoconus with visual rehabilitation through keratoconus RGP contact lens and SL alternatively under 2-year follow-up.

Case Presentation

A 24-year-old man visited our outpatient department complaining of progressively blurred vision in both eyes. He had been diagnosed of retinitis pigmentosa for many years.

The slit lamp examination for him revealed Vogt's striae, and corneal topography (Oculus Optikgerte GmbH, Wetzlar, Germany) (Fig. 1& Fig.2) indicated bilateral keratoconus. His best corrected visual acuity, with spectacle correction, was 0.5 logMAR to right and 0.7 logMAR to left eye. His keratometric readings, measured with autorefraction (Topcon RM6000, Japan), were 51.60 D/58.30 D at 28° and 57.50 D/70.00 D at 106°. The thinnest corneal thickness observed was 480 mm in the right eye and 378 mm in the left eye. Visual field examination revealed peripheral constriction of bilateral eyes. We had tested 5 exons of visual system homeobox 1 (VSX 1) gene from the subject and did not find mutation on direct sequence. There was no nystagmus, macular coloboma or multi-system anomalies in
this subject either.

To improve visual acuity, we applied keratoconus RGP contact lens (Brighten optix company, Corp., Taipei, Taiwan) for him. The patient gained visual acuity to 0.3 logMAR in each eye, and binocular visual acuity improved to 0.2 logMAR (Table 1).

However, he experienced instability and dislocation with lens, and he could not successfully retrieve it due to restricted visual fields. Moreover, there were complaints of two events of missing lens after dislocation. Thus, the patient was subsequently fitted with scleral lenses (SL) (Brighten optix company, Corp., Taipei, Taiwan). Visual acuity was also improved (Table 1), however, he complained of intolerance with extended use of SL, with an upper limit of tolerance time of about 4 to 5 hours. In contrast, the longest tolerable time for the keratoconus RGP contact lens was 8 to 9 hours. Depending on his needs, he wore both alternately.

Since the condition of the patient was stable, we assess his visual function with National Eye Institute Visual Functioning Questionnaire – 25 (NEI VFQ-25) in Mandarin, which is his native language and was valid in 2010[8] (Table 2). The composite NEI VFQ-25 score were 45.75, 69.46, and 63.38 with baseline, corrected with keratoconus RGP contact lens, and with SL, respectively (Table 2). The score improved both with two types of lens.

Discussion and Conclusions
It is uncommon to see retinitis pigmentosa in keratoconus patients. These two diseases may be coexisted in congenital disorders, for example, LCA[3, 4]. In this subject, the clinical presentation was unlikely to be LCA. The etiology of keratoconus is multigenic[9, 10], and it was reported that VSX 1 gene expressed in the retinal layer[11, 12], so we tested 5 exons of VSX 1 gene from him and the did not find mutation on direct sequence. The role of VSX 1 in the pathogenesis of keratoconus is controversial[13, 14], and may be only accounts for rare cases[15].

Contact lens are the primary form of visual correction for patients with keratoconus[16]. In a large lens trial for keratoconus, it was observed that aspheric RGP was preferred by patients wearing lenses 4-8 hours/day, but Rose K was reported to have wearing time more than 8 hours a day[17]. In our patient, his wearing habit of keratoconus RGP contact lens was 8 to 9 hours in a day.

In a 6-year cohort study of SL[18], the overall failure rate of SL wear was 27%, and mainly be caused of ocular complications such as intolerance or corneal edema. In our patient, intolerance was presented of 4 hours after wearing SL. SL was used for severe keratoconus patients. However, there was no gold-standard for severe keratoconus grading. Koppen[19] reported the cutoff value for maximal keratometry was 70 D, and our patient matched this criteria in one of his eyes.

In our patient, the NEI VFQ-25 showed that both two lenses improved visual function compared with baseline in composite scores, and keratoconus RGP contact lens presented even better than SL in general vision scores (Table 2).

In conclusion, this subject with keratoconus and retinitis pigmentosa, had good visual rehabilitation with keratoconus RGP contact lens and SL, and his visual function also got
improved with both visual aids.

Abbreviations

LCA: Leber congenital amaurosis

RGP: Rigid gas permeable

SL: Scleral lenses

NEI VFQ-25: National Eye Institute Visual Functioning Questionnaire – 25

VSX 1 gene: visual system homeobox 1 gene

Declarations

Ethics approval and consent to participate This study was approved by the ethic committee of Chung Gang memorial hospital (Taoyuan, Taiwan) and was performed in accordance with the tenets of the Declaration of Helsinki for research involving human subjects.
Consent for publication: Informed consent was obtained from the patient for publication. The consent is available for review by the editor of the journal.

Availability of data and material: All data supporting our findings is contained within the manuscript.

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Authors’ contributions

Yun-Wen Chen: contributor of writing manuscript

Ken-Kuo Lin: conception and design of data

Jiahn-Shing Lee: analysis and interpretation of data

Lai-Chu See: analysis and interpretation of data

Wan-Hua Cho: contributor of writing manuscript

Chiun-Ho Hou: manuscript writing, final approval of the manuscript

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Figures

Figure 1
Topography of the right eye indicated keratoconus

Figure 2
Topography of the left eye indicated keratoconus

Supplementary Files

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