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Case Report

Optical Rehabilitation of a Patient with Keratoconus and Nystagmus

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ABSTRACT

Keratoconus is a progressive corneal disease characterized by bilateral yet usually asymmetric thinning of the cornea with an onset typically in teenage years. While it often presents as an isolated condition, keratoconus may also be associated with many systemic and/or ocular diseases, such as connective tissue and chromosomal disorders. Its association with nystagmus has been described in Leber's congenital amaurosis, where patients also exhibit abnormal pupillary responses, early-onset retinal dystrophy, mental developmental delays, and eventual blindness. The case described here, however, was a high-functioning teenager with keratoconus and infantile nystagmus, and oscillopsia on left gaze and a compensatory head turn to the patient's left. The initial distance visual acuities of 20/60 and 20/150 in the right and left eye, respectively improved to 20/25 and 20/40 by the use of corneal rigid gas permeable contact lenses. In addition, the patient's neck strain and overall gait were eased by yoked prism spectacles.

KEYWORDS

Keratoconus; Ectasia; Infantile Nystagmus; Rigid Contact Lens; Yoked Prisms

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INTRODUCTION

Keratoconus is a progressive corneal disease, characterized by bilateral yet usually asymmetric thinning of the cornea with an onset, typically in teenage years [1, 2]. Its prevalence has been reported at 54 per 100 000 in the general population[3]. There has not been a consensus regarding its distribution between the two genders, yet it has a much higher incidence among Asians when compared with Caucasians [3]. Patients most commonly present different extents of decreased vision, the more severe of which is not correctable by spectacles, due to the increase in higher order aberrations, as the disorder progresses. While keratoconus often presents as an isolated condition, it may also be associated with many systemic and/or ocular diseases, such as connective tissue and chromosomal

disorders [4-6]. Therefore, keratoconus is suspected to have a familial association [7].

Infantile nystagmus, on the other hand, has been characterized by involuntary rhythmic eye movements that are noticeable within the first six months of birth [8, 9]. It may be associated with other ocular conditions, such as ocular albinism, achromatopsia, and Leber's congenital amaurosis [10]. Its association with keratoconus has been described in the latter, where patients also exhibit abnormal pupillary responses, early-onset retinal dystrophy, mental developmental delays, and eventual blindness.

The case reported here was a high-functioning teenager with keratoconus and infantile nystagmus, with oscillopsia on left gaze and a compensatory head turn to the patient's left, whose visual acuity and ocular stability significantly improved with optical rehabilitation.

CASE REPORT

History

A 19-year-old white male was referred for a comprehensive eye examination. He was alert and oriented with appropriate affect. His chief complaint was nystagmus with oscillopsia and blurry vision in both eyes, since birth, yet significant progression was noticed within the past year. His mother recounts bilateral subconjunctival hemorrhages at birth with normal gestational history and no caesarean or forceps delivery. Ocular history included a strabismus surgery to both eyes at age 10 in an attempt to dampen nystagmus, yet he reported that it had made the nystagmus worse. He had worn glasses in the past, which was minimally helpful. The medical history was unremarkable and he denied vigorous eye rubbing. He had already completed high school with no delays and enjoyed art and painting. He planned to enroll in college and wished to obtain a driver's license. He had two siblings and his father had nystagmus, yet his family did not include anyone with keratoconus.

Measurements

Entrance tests, Refraction, and Binocularity

On objective examination, his distance vision without correction was 20/60 in the right eye, 20/150 in the left eye, and 20/60 with both eyes. His near visual acuity was 0.4/1.2M in the right eye, 0.4/4.0M in the left eye, and 0.4/1.2M with both eyes. During distance vision testing, he consistently exhibited a head turn towards his left shoulder. He reported oscillopsia when gazing to the left side. His extraocular motilities were full in both eyes, without ophthalmoplegia. There was a horizontal left-beating nystagmus of both eyes, which was greater in amplitude and frequency on left gazes. It had no rotary component and dampened on convergence and right gaze. The pupils were equal, round, and reactive to light and there was no afferent pupillary defect. Peripheral vision test demonstrated full to single as well as simultaneous finger counting in all quadrants of each eye. The red-cap color comparison was equal between the two eyes. Cerebellar skills were normal without ataxia, and cranial nerves II, III, IV, V, VI, and VII were intact and symmetric. There was no response on both global and local stereopsis tests, and he exhibited an intermittent esotropic posture at approximately 12 prism diopters with intermittent left suppression, using the von Graefe technique [11]. Color vision discrimination, using the Farnsworth D15 color vision test, showed correct sequencing in each eye. Retinoscopy showed scissoring reflexes, which were worse in the left eye. Subjective refraction provided minimal improvement in vision in the right eye to 20/50+ and no improvement in the left eye, both of which matched results obtained with a pinhole. Automated keratometry (TONOREF II, NIDEK Co Ltd, Japan) revealed corneal astigmatism greater than 3 diopters in the right eye and greater than 7 diopters in the left eye.

Biomicroscopy and Optical Coherence Tomography (OCT)

On ocular health examination, anterior segment biomicroscopy showed mild corneal thinning inferior to the pupil in both eyes. There was no Fleischer's ring, Vogt's striae, stromal edema, corneal opacities, or Rizzuti's sign in either eye. The examination of lids, lashes, conjunctiva, iris, and anterior chamber showed no signs of allergies or other significant findings. Posterior segment evaluation with dilated pupils showed lens, vitreous, macula, optic nerves, vasculature, and peripheral retina in both eyes to be unremarkable and normal for his age. Optical Coherence Tomography (OCT) scans (Cirrus-HD OCT, Carl Zeiss Meditec AG, Germany) confirmed pathology-free macula (Fig 1).

Corneal Topography

The anterior surface corneal topography of the right eye, obtained by a Scheimpflug camera (Pentacam, OCULUS Optikgeräte GmbH, Germany), showed a simulated minimum keratometry reading of 42.5 diopter (D), at the 42.5-degree meridian and a 3.3 D central toricity. For the left eye, the simulated minimum keratometry reading was 42.3 D at the 109.2-degree meridian and the corneal toricity was 7.2 D. The maximum keratometry readings were 49.7 D and 54.3 D in right and left eyes, respectively. Corneal pachymetry measurements at the thinnest locations were 468 microns and 458 microns for

the right and left eye, respectively (Fig 2 and 3). These results are summarized in Table 1.

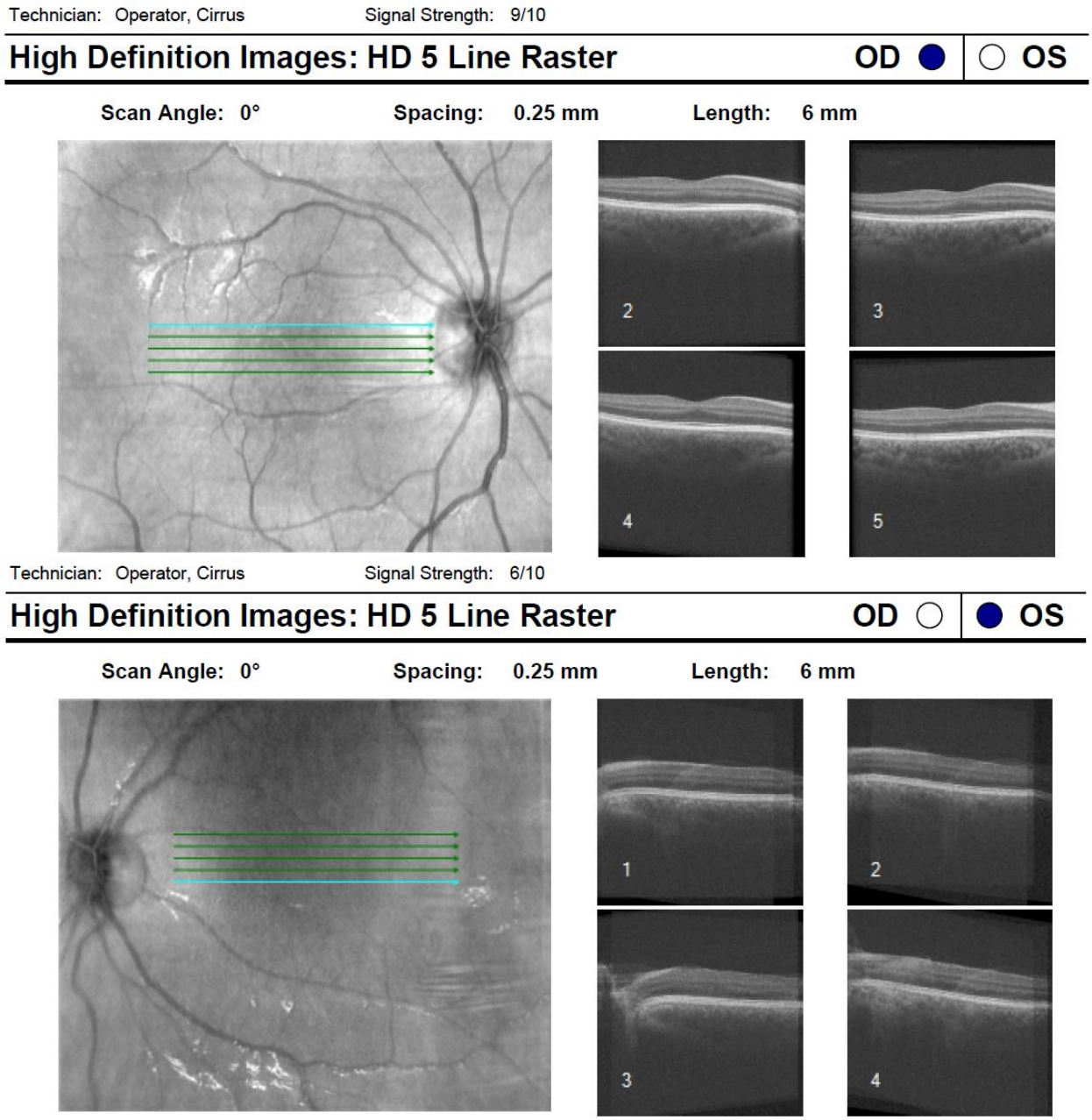

Figure 1: Posterior Segment Optical Coherence Tomography (OCT). Normal Findings of the Right (OD) and Left (OS) Eyes.

Table 1: Summary of corneal topography and pachymetry data

	Right eye	Left eye
Simulated Keratometry – Minimum Curvature	42.5 D	42.3 D
Simulated Keratometry – Maximum Curvature	45.7 D	49.5 D
Axis (Minimum Meridian)	42.5 degree	109.2 degree
K. Max – Maximum Curvature (Front)	49.7 D	54.3 D
Pachymetry (Thinnest)	468 µm	458 µm

D: diopter; µm: micrometer.

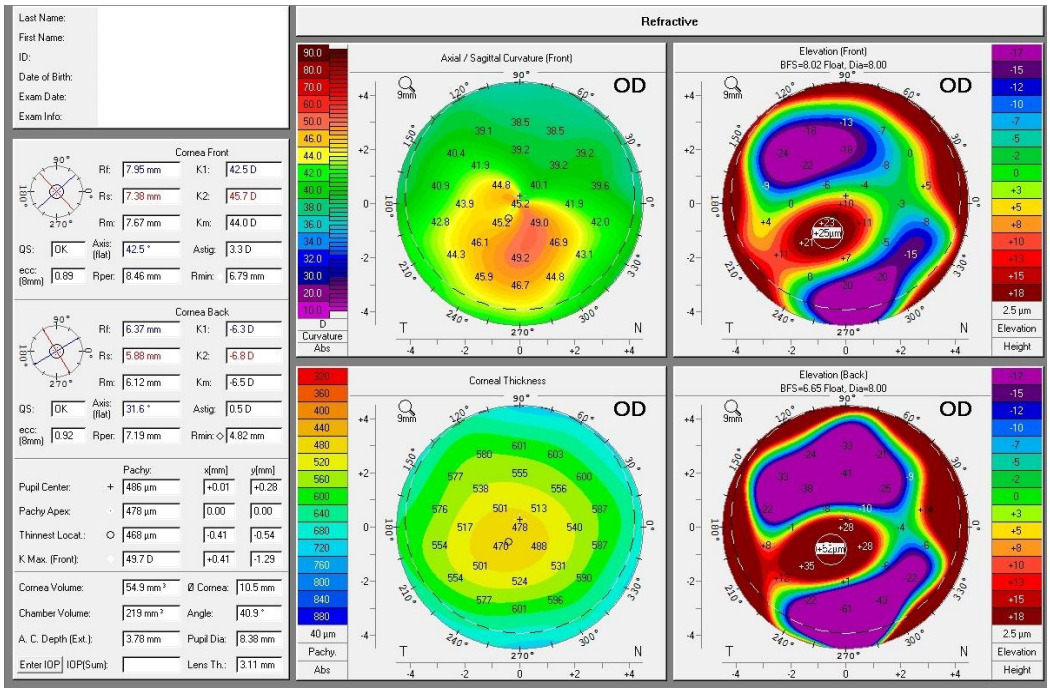

Figure 2: Corneal Topography Findings of the Right Eye.

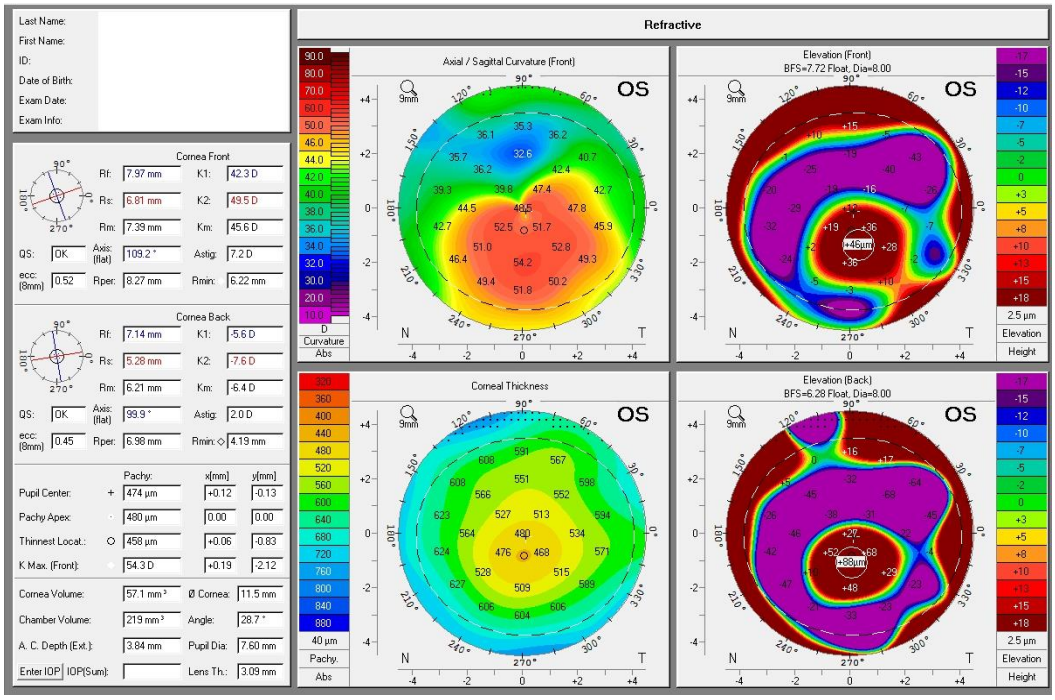

Figure 3: Corneal Topography Findings of the Left Eye.

Diagnoses

All clinical findings were considered, along with a thorough examination of the patient's personal and family history, and patient was diagnosed with bilateral keratoconus and suspected congenital motor nystagmus by linkage analysis (Fig 6), as genetic mapping was not available.

Treatment

Contact Lenses

Rigid gas-permeable lenses have been accepted as the best nonsurgical management tool for vision rehabilitation for patients with moderate to severe irregular astigmatism from keratoconus [12, 13]. In evaluating patient's corneal topography data, the Rose K2 (Blanchard Lab, Manchester NH) corneal rigid gas-permeable contact lens design, which is specifically developed for keratoconus, appears to be an appropriate treatment option. It utilizes a floating optic zone diameter, which varies by base curve radius to match the sagittal depth of the cone. A three-point touch fitting pattern with light feather-touch or slightly greater central touch over the apex of the cone and adequate edge clearance is achieved in vivo in each eye (Fig 4). This fitting pattern assures a protection of the fragile corneal apex while assuring adequate tear exchange beneath the contact lens back surface [14]. The Rose K2 aspheric aberration-control optics allow a three-line improvement in visual acuity in the right eye from 20/60 to 20/25 and a five-line improvement in the left eye from 20/150 to 20/40. The parameters of the final contact lenses that he received are outlined in Table 2.

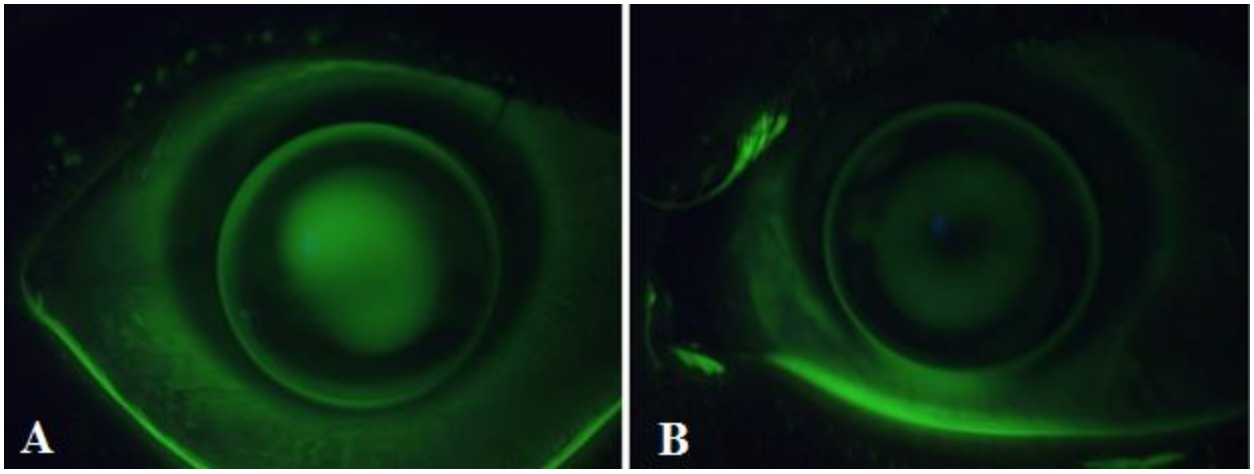

Figure 4: Corneal Rigid Gas-permeable Contact Lens fitting Pattern, using Sodium Fluorescein.

(A) Right Eye: Light Feather Touch Over Apex of Cone ; (B) Left Eye: Slightly Greater Central Touch Over Apex of Cone

Table 2: Summary of Contact Lens Parameters

	Right eye	Left eye
Lens design	Rose K2	Rose K2
Material	Menicon Z	Menicon Z
Dk value (ISO)	163	163
Base curve radius (mm)	7.35 mm	7.03 mm
Overall diameter (mm)	9.1 mm	9.0 mm
Back surface power (D)	-2.00 D	-5.50 D
Center thickness (mm)	0.19 mm	0.16 mm
Edge profile	Standard edge lift	Standard flat +1.00 edge lift

Dk: Oxygen Permeability; ISO: International Standard Organization; mm: Millimeter; D: Diopter.

Spectacles

Spectacles have been prescribed for two purposes. While he denies eye rubbing, a spectacle frame will serve as a physical barrier. In addition, prisms have been suggested to improve vision by dampening the nystagmus [8, 9]. When evaluating his nystagmus, he reported the least amount of oscillopsia in right and inward gazes. Even though previous records of patient's evaluation showed an increased signal strength on Visual Evoked Potential (VEP) with horizontal base-out prisms and no contact lenses, trial frame refraction with contact lens wear revealed the least amount of head turn with yoked prisms (Fig 5):

Right Eye: Plano Sphere Horizontal Prism 6.00 base-in (BI).

Left Eye: Plano Sphere Horizontal Prism 6.00 base-out (BO).

Figure 5: Yoked Prism Spectacles, Prescribed to Compensate for Head Turn.

DISCUSSION

Keratoconus is a progressive, non-inflammatory ectasia of the cornea, resulting in steepening and protrusion of the cornea, which may be noticeable as early as teenage years. It is usually bilateral yet may be asymmetric [8, 9]]. Infantile nystagmus, on the other hand, describes an early onset (before six months old) involuntary yet repetitive eye movement that is often associated with other ocular and systemic conditions [15]. Subtypes of infantile nystagmus include congenital motor nystagmus, sensory defect nystagmus, periodic alternating nystagmus, and latent nystagmus [16]. It seems that he had an autosomal dominant form of congenital motor nystagmus except for his oscillopsia symptom [17]. His father and all siblings (one older brother, one younger sister) had nystagmus, yet none had mal-developed iris, optic nerve, retina, or other ocular conditions, commonly associated with other subtypes of infantile nystagmus. The mother of the three siblings was not affected by any ocular condition other than refractive error and presbyopia. It should be noted that patient was the only member in the family with both keratoconus and nystagmus. Both parents had very low corneal astigmatism, while both siblings had moderate yet regular astigmatism. Fig 6 describes a linkage analysis pedigree by phenotype.

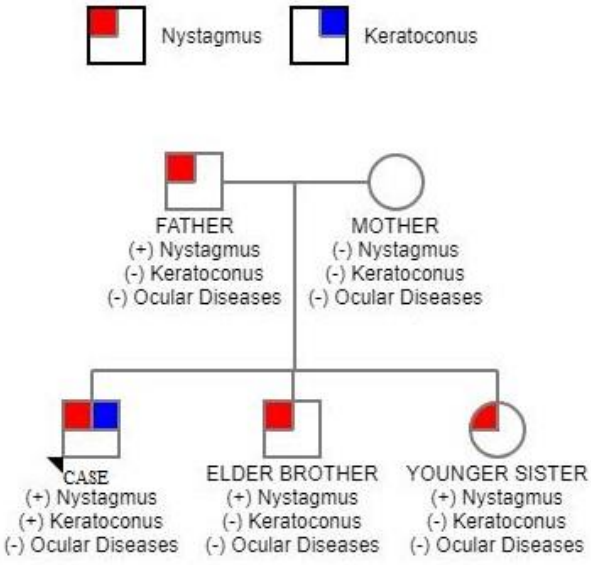

Figure 6: Linkage Analysis by Phenotype

To address his decreased vision due to keratoconus, he had been fitted keratoconus-design corneal rigid gas-permeable contact lenses that allowed him to achieve 20/30+ vision in the right eye and 20/40 vision in the left eye. In addition, spectacles with yoked horizontal prisms were prescribed for wear over the contact lenses, which family members reported to have improved his head posture and overall gait. Finally, patient himself also admitted to experiencing less strain/fatigue with an only part-time utilization of the prism spectacles.

Corneal cross-linking has been discussed yet due to the concern with nystagmus, he was hesitant about undergoing the procedure. This option will be explored with a corneal specialist.

DISCLOSURE

Ethical issues have been completely observed by the authors. All named authors meet the International Committee of Medical Journal Editors (ICMJE) criteria for authorship of this manuscript, take responsibility for the integrity of the work as a whole, and have given final approval for the version to be published. No conflict of interest has been presented.

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