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CASE REPORT ON SCLERAL LENS MANAGEMENT OF CORNEAL IRREGULARITY SECONDARY TO OCULOCUTANEOUS ALBINISM.

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Abstract

The purpose of this case report is to show the benefits of scleral lenses in the management of corneal irregularity and primary corneal ectasia. Scleral lenses are special corneal gas permeable lenses designed with a larger diameter to rest on the conjunctiva and vault over the cornea. This way, the lenses replace the damaged cornea with a smooth, uniform, regular surface that corrects the visual distortions caused by an irregular cornea. Scleral lenses provide optimization of vision with additional benefits of protection and continuous hydration of the ocular surface through the post-lens tear reservoir. In Nigeria, scleral lenses are a new line of treatment that is rapidly becoming a valuable treatment option in the management of irregular astigmatism, corneal irregularity, and ocular surface diseases like keratoconus, keratoglobus, and pellucid marginal degeneration. This is a case of a teenage Nigerian girl with oculocutaneous albinism, nystagmus, and irregular astigmatism.

Key words: Scleral lenses, corneal irregularity, irregular astigmatism, oculocutaneous albinism, ocular surface disease.

Introduction

Scleral contact lenses are a special type of rigid gas-permeable lens with larger diameters designed to vault over the entire corneal surface and rest on the sclera. In recent years, scleral lenses have been growing in popularity for the treatment of ocular surface problems and other forms of corneal ectasia as they significantly improve vision and give added benefits of improved comfort, stability, and continuous lubrication of the ocular surface. The space between the cornea and the back of the scleral

lens acts as a fluid reservoir continuously bathing the cornea, scleral lenses are also utilized for the treatment of dry eye syndrome. According to the World Health Organization (WHO), albinism is a rare genetic disorder that affects approximately 1 in 17,000 to 20,000 people worldwide,¹ it is more prevalent in sub-Saharan Africa, where an estimated 1 in 1,400 people are affected.²

The prevalence of albinism varies widely among different ethnic groups in sub-Saharan Africa, ranging from 1 in 1,000 to 1 in 15,000 individuals,³ a study conducted in Tanzania

1. World Health Organization. (2021). Albinism <https://www.who.int/teams/noncommunicable-diseases/eye-health/albinism>
2. Sturm, R. A., & Frudakis, T. N. (2014). Eye color: portals into pigmentation genes and ancestry. *American Journal of Human Genetics*, 94(5), 809-824.
3. Mchome, Z., Richards, E., Namadingo, H., Ngwira, B., & Chilunga, F. (2017). Disability visibility on the agenda in sub-Saharan Africa: An analysis of the 2013–2015 UNCRPD periodic reports. *African Journal of Disability*, 6, 1-9.

found a prevalence of 1 in 1,400 people affected with albinism.⁴ In Nigeria, its prevalence was found to be 1 in 5,000.⁵ Overall, albinism is a relatively rare disorder worldwide, but it is more common in certain regions, particularly sub-Saharan Africa.

Oculocutaneous albinism is a heterogeneous group of rare autosomal recessive disorders affecting melanin synthesis that is characterized by congenital hypopigmentation of the skin, hair, and eyes, it also reduces pigmentation of the iris and the light-sensitive retina. Oculocutaneous albinism patients usually have vision problems such as reduced visual acuity (blurred distance and near vision), nystagmus, photophobia, iris transillumination, foveal hypoplasia and abnormal decussation of nerve fibers at the optic chiasm.⁶ In the albino group, most common refractive profiles are astigmatism and hypermetropia, the irregularity of the corneal surface of oculocutaneous albinism patients results in irregular astigmatism.⁶

In the case presented here, conventional glasses and contact lenses (soft and hard) could not improve vision significantly. The case presented here discusses how a patient suffering from low vision and decreased vision due to oculocutaneous albinism achieved improved distant and near vision by being fit with scleral contact lenses.

CASE REPORT

A 12-year-old Nigerian albino female presented for examination on 13th September 2018 having

been referred for an optometric consultation for evaluation of blurry distant and near vision even with conventional spectacle and soft contact lenses (on a trial basis). The patient was a habitual wearer of transition-antireflective spectacle lenses and complained that her prescription glasses always reduced in clarity after 3-4 months of getting them. She also complained of difficulty seeing clearly at distance and close ranges even with her spectacle correction. She had other complaints like excessive light sensitivity (photophobia), headaches, and a gritty sensation. The patient reported that she had been using transition lenses which provided mild relief from the sunlight especially as she lives in the tropics in Africa.

OCULAR AND MEDICAL HISTORY

The patient's ocular history was positive for hypermetropia, astigmatism, and evaporative dry eye disease, her medical history was positive for oculocutaneous albinism. A review of systems revealed no other positives, and she reported no known allergies to medications, her social history was negative for alcohol or recreational drug use. She was a high school student ready to find a lasting solution to her vision problems. Her family history was positive for hypertension (father) and hypercholesterolemia (mother), her mood was appropriate, and she was oriented to person, place, and time.

EXAMINATION

Uncorrected visual acuity was 6/60 in the right eye (OD) and 6/60 in the left eye (OS).

4. Kromberg, J. G. R., & Manga, P. (2002). Albinism in Africa. *South African Medical Journal*, 92(6), 377-381.

5. Oladipo, G. S., Kolawole, O. U., & Ogbole, G. I. (2021). Prevalence and clinical profile of albinism in a tertiary hospital in North Central Nigeria. *African Vision and Eye Health*, 80(1), 1-6.

6. Gronskov K, Brondum-Nielsen K, et al. Oculocutaneous albinism. *Orphanet J Rare Dis*. 2007; 2: 43

With her habitual spectacle correction of OD: +3.50/-3.50x180 and OS: +3.50/-3.50X180, corrected entry visual acuity was 6/36 in both eyes (OU), near visual acuity was N18. Her pupils were equal, round, and reactive to light and accommodation, and no afferent pupillary defect was noted. She experienced nystagmus which is a hallmark sign of oculocutaneous albinism, confrontation visual fields were full to finger counting in both eyes.

Slit-lamp bio-microscopy revealed normal external adnexa, lids, lashes, and puncta in both eyes, examination of both eyes showed that the conjunctiva was white and quiet, and the cornea was clear. Tear break-up time (TBUT) was 5 seconds OU, and Schirmer's test result was 5mm in both eyes, this is indicative of evaporative dry eye disorder. One drop of fluorescein was instilled into both eyes and intraocular pressures were measured with Perkins tonometer. The intraocular pressure readings were 18mmHg OD and 19mmHg OS at 12:05 pm. Both anterior chambers were deep and quiet, both irides were flat and intact, lacked melanin pigmentation and so were light brown in color.

The patient's eyes were dilated for fundus examination which revealed clear lenses, normal vitreous, hypopigmented pale appearance of the fundus, distinct optic nerves with a cup-to-disc ratio of both eyes, normal retinal vessels (OU), foveal hypoplasia, and flat intact retinal periphery. Fundus photography was also carried out.

The visual acuity with best spectacle correction was 6/24-1 in both eyes and there was no improvement in vision with over-refraction or pinhole. The patient's emphasis was on improved

vision, however, we preferred the scleral lens option since it showed better improvement for her as regards overall vision and to manage the mild dry eye disorder. The patient was prescribed a Telescope of magnification 3.5X to improve vision, visual acuity improved significantly to 6/18 in both eyes (OU) and she was educated on how to use the low-vision devices, she was also prescribed Refresh/Systane preservative-free artificial tears every 4 hours to relieve dry eye symptoms and instructed to use the dry eye treatment continuously.



Figure 1: Posterior segment photography of patient's right eye.

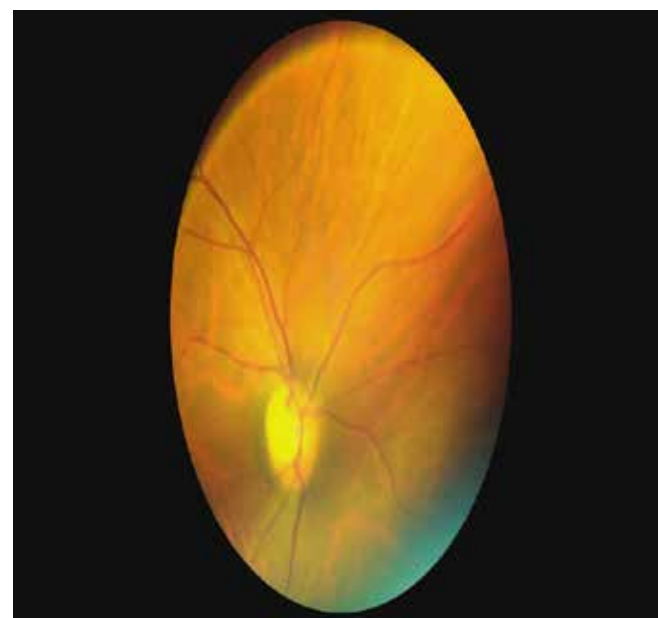


Figure 2: Posterior segment photography of patient's left eye.

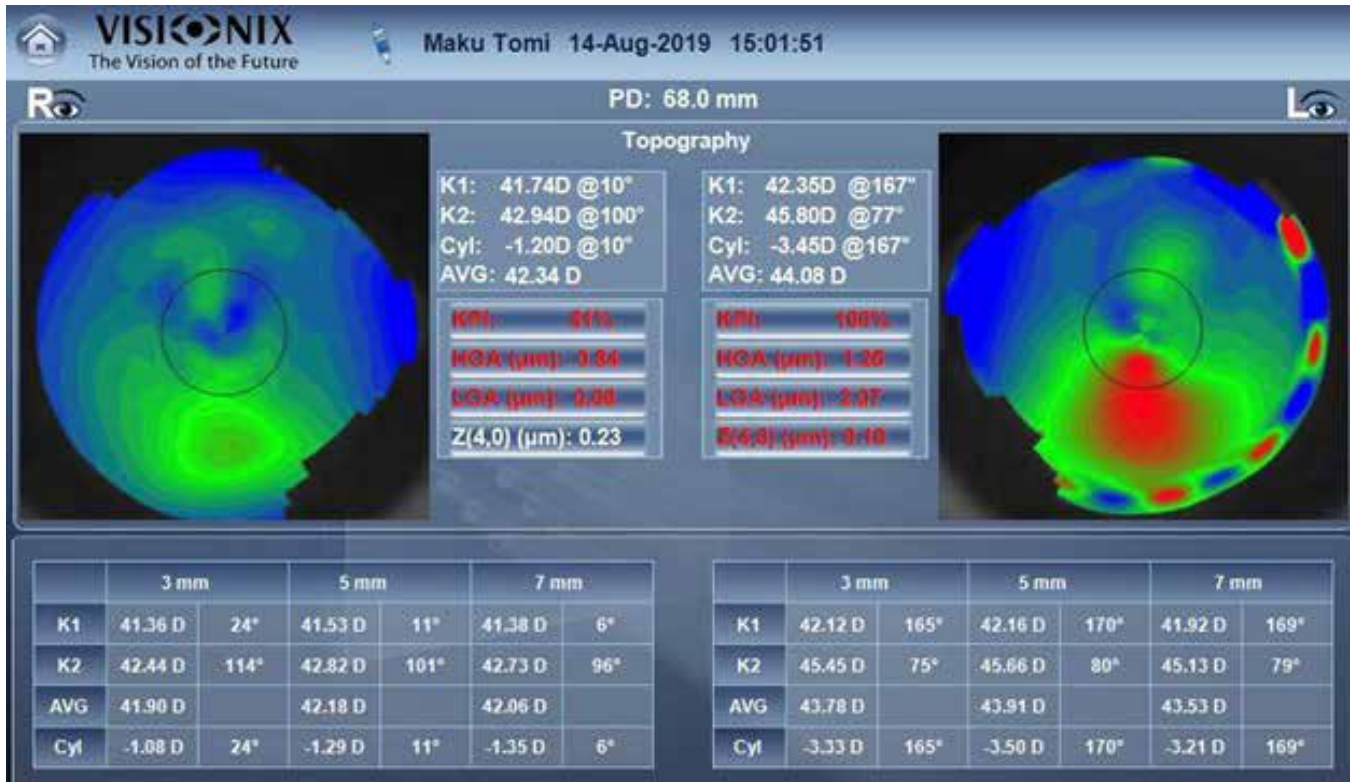


Figure 3: Corneal topography

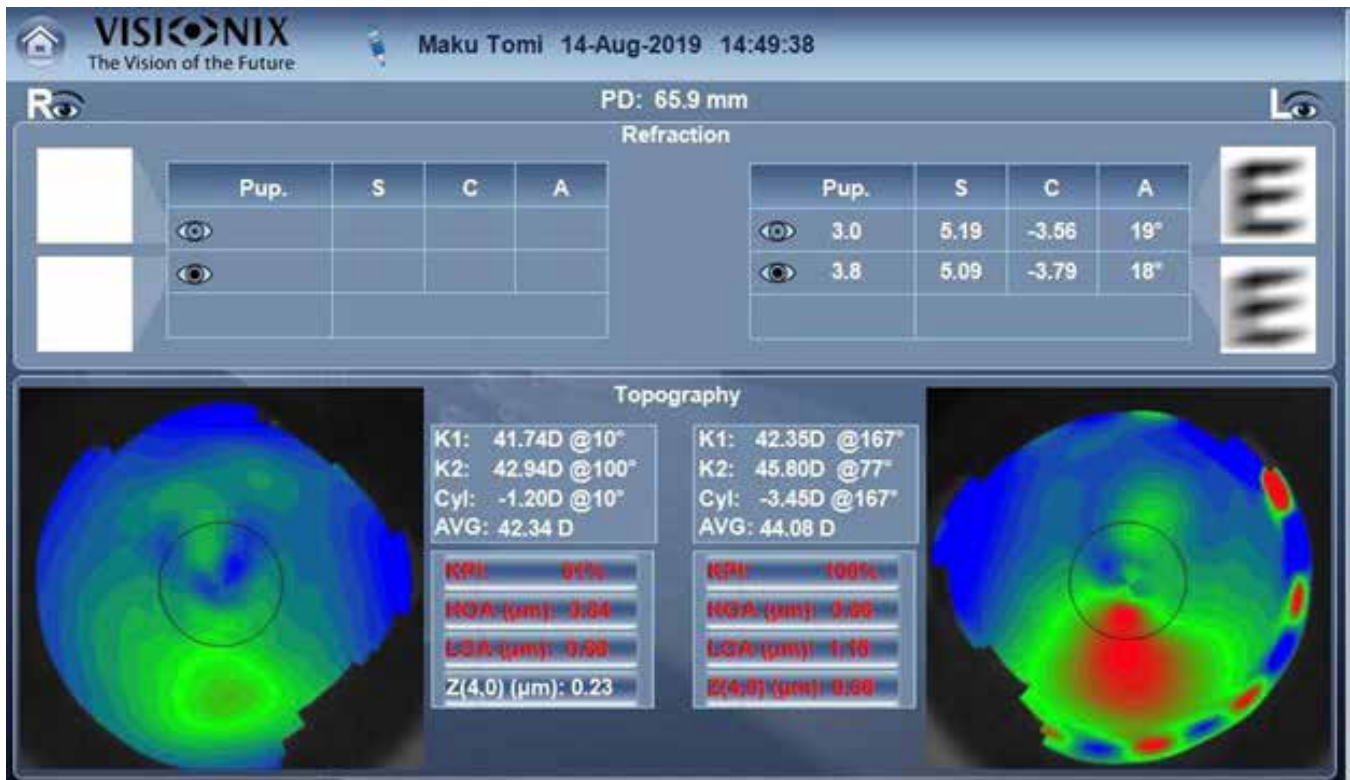


Figure 4: Corneal Topography

FOLLOW-UP-VISIT #2

The patient returned to the clinic on the 12th of July 2019 for a routine yearly examination. She reported that since she started using the artificial tear drops, the gritty sensation and tearing had reduced drastically. The patient reported difficulties in reading and no other changes in vision or health since the gritty sensation and tearing had reduced drastically.

EXAMINATION

The patient's corrected entry visual acuity with a Telescope of magnification 3.5X was 6/18 OU. Slit lamp bio-microscopy revealed normal adnexa, lids, lashes, and puncta in both eyes. Examination of both eyes showed that the conjunctiva was white and quiet, and the cornea was clear, TBUT was 10 seconds OU. The Schirmer's test results were 10mm OU. This was an improvement in the ocular surface dryness. The intraocular pressures were measured to be 15mmHg right eye and 16mmHg left eye.

PLAN

The patient was to continue Systane preservative-free artificial tear drops 4 hourly as previously prescribed. A hand-held magnifier of magnification 2.5X was prescribed to improve her near visual acuity and aid reading as she was a high school student reading lots of small prints on paper. A Scleral contact lens fitting was recommended for both eyes as the scleral lens would likely provide significant improvement in distant and near vision, provide continuous lubrication for the ocular surface, and reduce signs and symptoms of dry eye. The patient's near visual acuity improved from N18 TO N12 which aided her schoolwork.

FOLLOW-UP VISIT #3

She presented for a scleral contact lens fitting both eyes on the 7th of February 2020 and reported that the telescope and hand-held magnifier only helped improve vision slightly but were cumbersome to use and not aesthetically pleasing. The patient was still using Systane preservative-free artificial tears 4 hourly as previously prescribed and did not experience symptoms of dry eye syndrome anymore. She reported no other changes since the last visit.

EXAMINATION

The patient's corrected entry distance visual acuity with a Telescope of magnification 3.5X was 6/18 in both eyes and with a hand-held magnifier of magnification 2.5X was N12. Slit lamp biomicroscopy revealed normal adnexa, lids, lashes, and puncta in both eyes. Examination of both eyes showed that the conjunctiva was white and quiet, and the cornea was clear. The patient's TBUT was 12 seconds in both eyes, and Schirmer's test result was 12mm in both eyes. This was an improvement in the extent of the ocular surface dryness. The intraocular pressures were measured to be 14mmHg right eye and 15mmHg left eye.

We went on to scleral lenses as we sought ways to improve the patient's vision as she developed some skepticism to low vision aids due to the nature of the devices and their acceptance amongst her peers, adaptability issues also arose. A discussion was had with her parents about contact lenses and then scleral lenses which they accepted as better options to low vision aids. The soft contact lenses were tried, along with rigid gas permeable lenses and the results were unremarkable, so we moved on to scleral lenses.

SCLERAL CONTACT LENS FITTING

Corneal/Scleral topography (Figures 5a and 5b) was done for both eyes using the Eaglet Profilometer (Eye Surface Profiler) and the corneal topography using Visionix VX110. Calculation was done with the software to choose a trial lens from the trial lens set.

A trial scleral contact lens was selected from a MAXIM (ACCU LENS) Scleral lens set, for the right eye (OD), a diameter of 15.9 was

selected based on the patient’s horizontal visible iris diameter (HVID) which measured 11.7mm in the right eye. The fitting guide for this lens recommended selecting an initial diameter of 15.9mm for HVIDs in the range of 11.6-12.4mm, the Sagittal depth of the initial trial lens was 4.73. For the left eye (OS), a diameter of 15.9/8.5 was also selected, the sagittal depth of the trial lens was 4.91.

All parameters of the trial lens were as follows:

Table 1: Scleral Trial Lens Parameters

	OD	OS
Manufacturer	ACCU LENS	ACCU LENS
Brand	MAXIM Scleral lens	MAXIM Scleral lens
Diameter	15.9/8.5	15.9/8.5
Base Curve	7.50	7.34
Sagittal Depth (SAG)	4.73	4.91
Prescription (PWR)	-4.00	-5.00

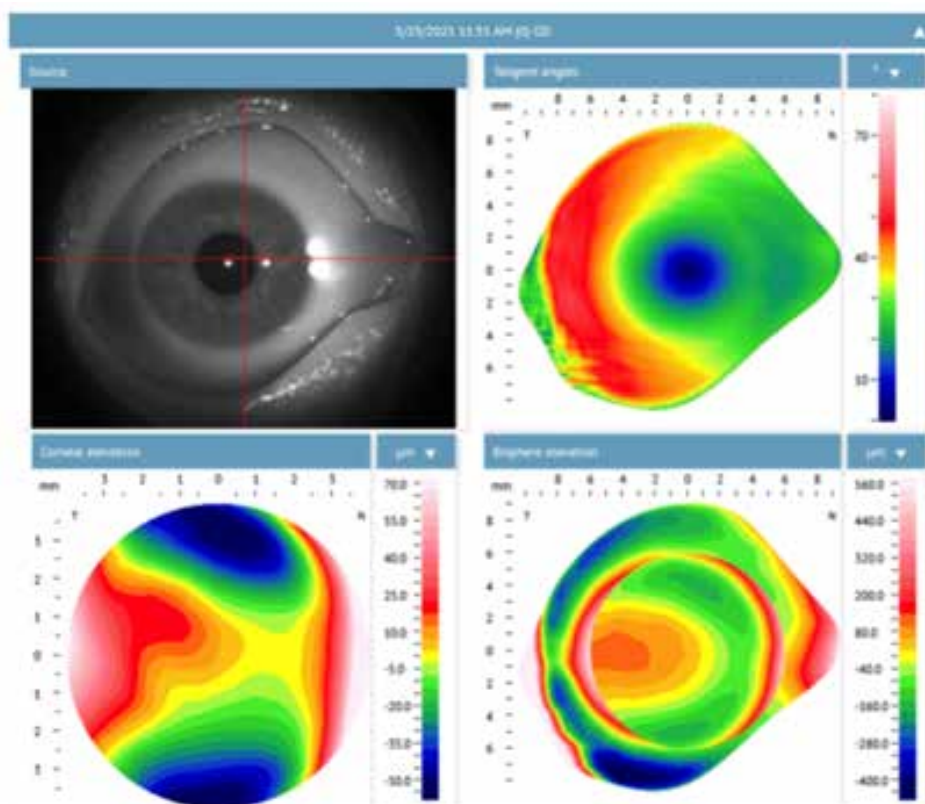


Fig. 5a Corneal/Scleral topography (right eye)

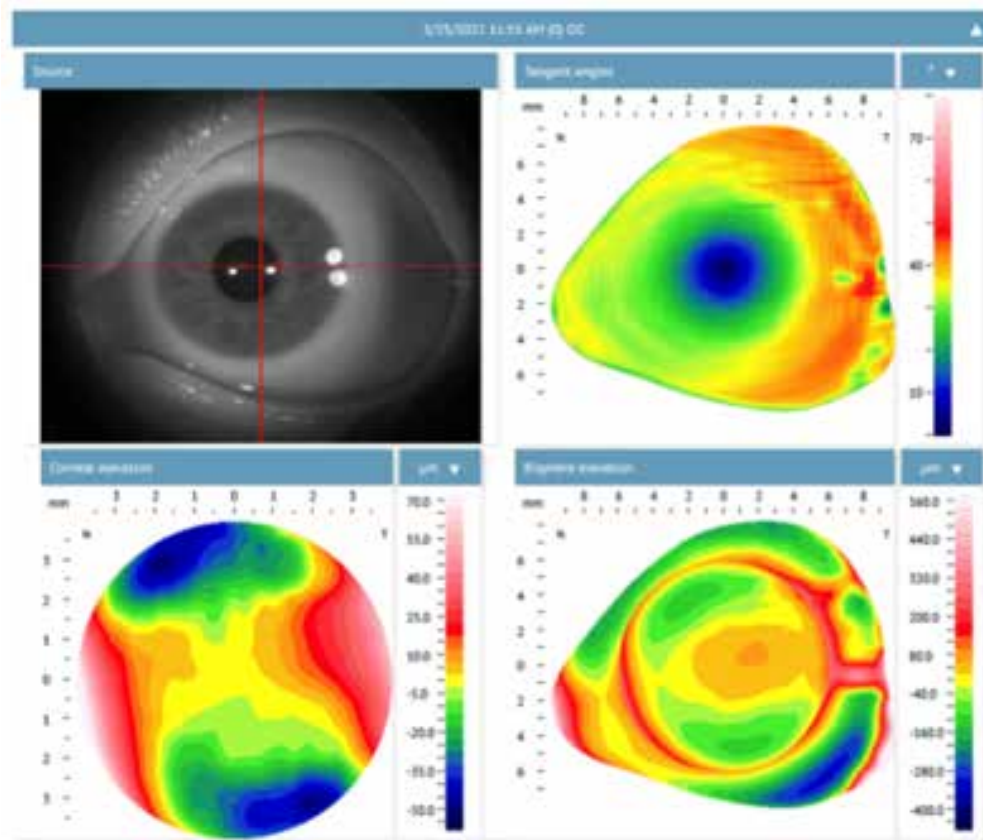


Fig. 5b Corneal/Scleral topography (left eye)

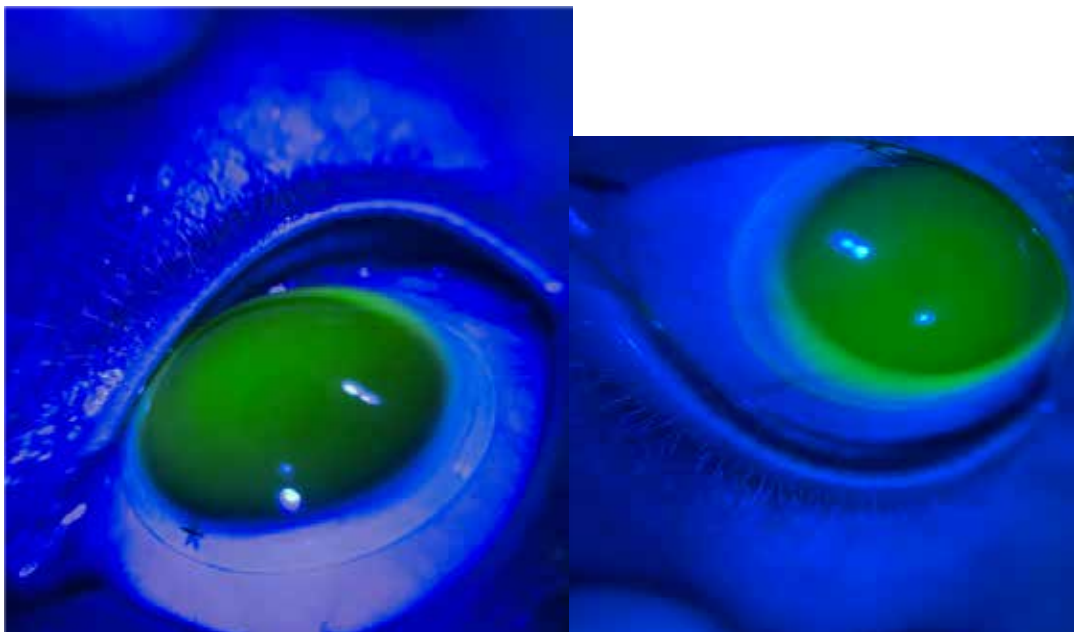


Figure 6 Anterior segment photography of patient's right and left eye with MAXIM scleral lens on the eye.

The central clearance was measured by estimating the thickness of the lens tear reservoir to the scleral lens thickness using the slit lamp, there were about 250 microns of central clearance immediately after insertion (about 20

to 30 minutes) and 30 microns of clearance at the limbus (figure 6).

An over refraction was done, and it showed a much clearer visual acuity and because of this the reorder necessitated a slightly reduced vault

of about 170 to 180 microns of central clearance. There was an improvement of lens centration with a reduced central vault, the limbal coverage remained adequate while the sag was changed because of the difference in visual acuity.

Visual acuity on OD with this trial lens was 6/18. With a spherical over-refraction of +1.50, the patient's visual acuity improved to 6/12.

Visual acuity on OS with this trial lens was 6/24. With a spherical over-refraction of +3.00, the patient's visual acuity improved to 6/18+

Visual acuity with trial lenses and over-refraction OU was 6/12+. Near visual acuity with trial lenses improved from N18 to N10 OU.

Upon slit-lamp evaluation, the external index was in order.

The patient was able to achieve better visual acuity with the MAXIM trial scleral lens than she had previously achieved with her habitual spectacle correction and telescope/hand-held magnifier.

PLAN

Modifications were made to the lens parameters to incorporate the over-refraction and to give a

better, comfortable fit. Based on the previous over-refraction which revealed a much clearer visual acuity, a reorder with a slightly reduced vault of about 170 to 180 microns of central clearance was made. There was a reduced central vault and an improvement of lens centration while the limbal coverage remained adequate, she was asked to continue using Systane preservative-free artificial tears as prescribed.

FOLLOW-UP VISIT #4

The patient presented at the clinic on the 25th of May 2021 for another trial fitting of her MAXIM Scleral lens (OU), she reported no new changes in symptoms since the last visit.

Scleral Contact Lens Evaluation.

The ordered MAXIM scleral lenses were placed on both eyes. Visual acuity measured with the lens was 6/12 in the right eye and 6/18 in the left eye.

Corneal topography was done for both eyes using the Eaglet Profilometer (Eye Surface Profiler) (figure 5a and 5b). Calculation was also done with the software to choose another trial lens from the trial lens set.

Table 2: Parameters of the trial lens were as follows:

	OD	OS
Manufacturer	ACCU LENS	ACCU LENS
Brand	MAXIM Scleral lens	MAXIM Scleral lens
Diameter	15.9/8.5	15.9/8.5
Base Curve	7.85	7.50
Sagittal Depth (SAG)	4.42	4.73
Prescription (PWR)	-2.00	-4.00

For OD, the visual acuity with this trial lens was 6/12. With a spherical over-refraction of +2.50 Sphere, the patient's visual acuity improved to 6/9. For OS, the visual acuity with this trial lens was 6/18, with a spherical over-refraction of +4.00 Sph, the patient's visual acuity improved to 6/12. Visual acuity with trial lenses and over-refraction OU was 6/9+. Near visual acuity with trial lenses improved from N18 to N8 OU.

Upon Slit-lamp evaluation, the patient was able to achieve better visual acuity with the MAXIM trial scleral lens than she had previously achieved with her habitual spectacle correction and telescope/hand-held magnifier.

PLAN

Modifications to the landing zones were to be made to the new lens parameters to incorporate the over-refraction and to give a better, comfortable fit.

FOLLOW-UP VISIT #5

The patient presented to the clinic on the 28th of June 2021 for dispense of her MAXIM scleral lenses (OU).

The final prescription was

OD: Maxim 3D 7.85 +0.50 15.9/8.5 4.42 SAG.
"0" Angular offset – free form

OS: Maxim 3D 7.50 +0.00 15.9 /8.5 4.73 SAG
"0" Angular offset – free form

She reported using Systane preservative free artificial tears once every 4 hours.

Scleral Contact Lens Evaluation

The ordered MAXIM scleral lens was placed on OD, and visual acuity measured with the lens

was 6/9 in OD. The ordered MAXIM scleral lens was placed on the left eye OS. Visual acuity measured with the lens was 6/12 in the left eye OS. The lens was allowed to settle on the patient's eye for 20 minutes and then the fit was evaluated using a slit lamp. The fit of the lens showed good centration and coverage, the scleral landing zone was well aligned with the sclera in all quadrants and showed no areas of compression or impingement upon the conjunctival or conjunctival vessels.

Excellent fit, vision, and comfort were achieved in both eyes OU with the ACCU MAXIM scleral lens. The patient reported an immediate reduction in symptoms of dryness and an improvement in the quality of vision compared to her habitual spectacle correction. She was successful with practicing the insertion and removal of the scleral lens in the clinic after some trials.

PLAN

The patient was thoroughly educated on scleral lens insertion, removal, and cleaning. This included filling the lens with scleral lens preservative-free saline solution (PuriLens/Nutrifill) before insertion, disinfecting the lenses nightly with scleral lens hydrogen peroxide cleaning solution, and using Nutrifill/PuriLens solution if needed for rinsing before insertion. The patient was instructed never to sleep, swim, or shower with the scleral lenses on. She was also instructed to discontinue wear and return to the clinic if significant redness, pain, photophobia, or decreased vision occurred.

The MAXIM scleral lenses were dispensed. She was advised to start at 4 hours per day of wear and gradually increase wear time, but not to

exceed 12 hours of wear time prior to the next visit and was scheduled to return to our clinic in 1 month for follow-up.

DISCUSSION

Oculocutaneous albinism is a heterogeneous group of rare inherited autosomal recessive disorders affecting melanin synthesis, characterized by congenital hypopigmentation of the skin, hair, and eyes.⁶ These conditions are caused by mutations in specific genes that are necessary to produce melanin pigment in specialized cells called melanocytes. Absent or insufficient melanin pigment results in abnormal development of the eyes, resulting in vision abnormalities and light skin that is very susceptible to damage from the sun including skin cancer. Several vision problems occur with this condition, these include nystagmus, iris transillumination, retinal pigment, macular hypoplasia resulting in abnormal foveal development, poor visual acuity, strabismus, photophobia, and reduced depth perception. Visual acuity in individuals ranges from 20/60 to 20/400, usually depending on the amount of pigment present in the eyes.^{4,6}

Treatment options include measures taken to improve vision and to protect the skin from sun damage and an annual eye examination by an Optometrist is required to monitor the visual state of patients with Oculocutaneous

albinism. Treatment for vision problems includes correction of refractive errors with the use of spectacle glasses, soft contact lenses, hard contact lenses, rigid gas permeable lenses, scleral lenses, low vision aids/devices, use of sunglasses or special filter glasses for light sensitivity, and in some cases extraocular muscle surgery to restore alignment and improve head posture that is associated with nystagmus.^{17,18}

According to a study,⁷ scleral lenses can be an effective option for individuals with albinism who experience visual problems due to abnormal development of the visual pathways. The researchers found that scleral lenses improved visual acuity and reduced photophobia in a group of individuals with oculocutaneous albinism.

Another study⁸ also reported positive outcomes for scleral lens use in individuals with oculocutaneous albinism, the researchers found that scleral lenses improved visual acuity and reduced symptoms of dry eye in a group of individuals with oculocutaneous albinism.

After exhausting every optical form of the then-available options for treating this patient in trying to improve her vision considering her age and how important it was for her to function optimally in school, low vision devices were opted for. However, this came with its attending challenges as the young patient complained of mobility issues with the device, and acceptance

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6. Gronskov K, Brondum-Nielsen K, et al. Oculocutaneous albinism. *Orphanet J Rare Dis*. 2007; 2: 43

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especially amongst her peers due to its poor cosmetic appeal. (The patient was not satisfied with the spectacle prescription results. After all the modifications with the glasses, we decided to opt for other tests like the corneal topography, which revealed the corneal irregularities and prompted the specialty lens trials, which proved most effective).

The case described in this report showed that scleral lenses can be a beneficial treatment option to address ocular surface disease, dry eye, and irregular astigmatism. A scleral lens is a large diameter gas permeable contact lens that vaults over the cornea and rests on the anterior scleral surface.¹² The lens is filled with non-preserved saline solution prior to insertion, creating an enclosed liquid reservoir that allows for optical correction of irregular astigmatism without direct contact between the lens and the cornea, the fluid reservoir gives added benefits of improved comfort, stability, and continuous lubrication of the ocular surface. Scleral lenses are also utilized as a treatment for dry eye syndrome, they reduce mechanical stress to the cornea in comparison with corneal gas permeable lenses and are better options for correction of irregular astigmatism.^{12, 13}

In cases of nystagmus, contact lenses are the preferred optical correction, as the refractive correction moves with the eye.¹¹ Additionally, case reports and laboratory-based studies suggest that contact lens wear (with or without a

tint or artificial iris) reduces the amplitude and frequency of the nystagmus in some patients, potentially by providing some tactile or proprioceptive feedback about eye movements.¹⁰

Some complications of scleral lens wear include microbial keratitis and hypoxia-related complications such as corneal edema and corneal neovascularization. The risk for infections can be reduced by thoroughly training the patient in proper contact lens hygiene and lens care, regular follow-up visits to ensure patients are maintaining good habits.^{13,14,16}

In Nigeria, the scleral lens is a new line of treatment for the management of corneal irregularities like keratoconus, ectasia, and high astigmatism. Some challenges being faced in the usage of scleral lenses among eye care practitioners in Nigeria include the cost of instruments needed like the Profilometer which is an ocular surface topographer that helps to increase the accuracy of corneal topography mapping and reduces the number of hours spent on each patient for scleral lens fitting (reduces chair time). Another challenge being faced in Nigeria is the cost of the scleral lenses for the patients.

Scleral lenses helped in resolving the frequent dryness the patient was experiencing and served to alleviate the irregularity the corneal surface experienced causing significant astigmatism most likely because of the nystagmus.¹¹ Also,

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10. Taibbi G, Wang ZI, Dell'Osso LF. Infantile nystagmus syndrome: Broadening the high-foveation-quality field with contact lenses. *Clin Ophthalmol* 2008; 2: 585–589.
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 16. Schomack MM, Pyle J, Patel SV. Scleral lenses in management of ocular surface disease. *Ophthalmol*. 2014; 121(7): 1398-405.

important to note is that the manufacturing laboratory team was very instrumental in designing this fit.

There is a significant amount of aberrations in both corneas likely causing some additional decrease in visual acuity and contrast. This seems to be a keratoconic patient more advanced in the left eye and newly developing in the right eye.

The Keratoconus prediction index (KPI) is calculated by a combination of 8 topographic

indices and uses a linear discriminant function, a value greater than 0.23 is suggestive of keratoconus. It has a sensitivity of 68 % and a specificity of 99 % for keratoconus.¹⁵ The nystagmus made it very hard to get good quality/usable images, but this is what the topography image is showing, so relying on the topography images alone was not satisfactory. The high amount of aberrations is likely because of the nystagmus, the topography also revealed a with the rule astigmatism which is common in young people.

CONCLUSION

The patient described in this case report reported improved visual acuity at distance and at close range. She reported a better ability to do her schoolwork and participate in other activities too and a relief of ocular surface dryness with continuous use of her scleral lenses. In addition to improving vision by correcting irregular astigmatism, the patient's scleral lenses will allow corneal rehabilitation. Scleral lenses are good options for the correction of irregular astigmatism and offer the additional benefit of providing continuous hydration of the ocular surface. Scleral gas permeable contact lenses should be considered in cases of irregular cornea, irregular astigmatism, severe dry eye, and ocular surface diseases. Scleral lenses reduce ocular discomfort and improve quality of life however, it is important for individuals with albinism to work closely with an eye care professional to determine if scleral lenses are appropriate for their specific needs.

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