# Visual rehabilitation of people with oculocutaneous albinism in a tertiary clinical setting in Pakistan

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## Abstract:

**PURPOSE:** All people with oculocutaneous albinism (OCA) have reduced vision. This study aimed to assess the benefits of low vision aids for people with OCA.

**METHODS:** Seventy-seven consecutive people with OCA age 4 years and above examined in a low vision clinic were included in the study. Uncorrected and best-corrected visual acuity (VA), VA with low vision devices, types of low vision aids, and refractive errors data were analyzed.

**RESULTS:** Of the 77 people with OCA, 57% were in the age group between 4 and 15 years and 43% in the age group of 16 years and above. At the time of presentation, the percentages of visually impaired, severe visually impaired, and blind (using WHO low vision criteria) were 52%, 22%, and 25%. Among them, 39% has near VA of 1 M or better. Difference in the means of the spherical equivalent refractive error in the right eyes and left eyes was −0.494 diopters (−01.686, 0.699; 95% confidence interval). VA improved significantly after adequate refractive correction by more than one log MAR lines in 38.6% ($P < 0.01$). With low vision devices, in 85.7% ($n = 66$) participants, VA was enhanced to normal level (6/18 or better) in the better eye while 7.8% still remained in the blind category. Telescopes were prescribed to 61% people for the enhancement of distance VA and hand hold magnifiers were prescribed to 22% people to meet their needs.

**CONCLUSION:** Low vision aids can be successfully used in visual rehabilitation of people with OCA to meet their needs.

**Keywords:**

Blindness, low vision aids, oculocutaneous albinism, Pakistan, visual rehabilitation

## Introduction

Oculocutaneous albinism (OCA) is a group of inherited recessive disorders. People with OCA have either a reduction or a complete lack of melanin biosynthesis resulting in hypopigmentation of the eyes, skin, and hair.[1,2] The prevalence of different types of OCA occurs regardless of race, ethnic backgrounds, or gender and varies considerably worldwide.[3,4] It has been estimated that one in 17,000 people have one of the types of OCA.[1,2] With a complete lack of melanin, OCA 1A is the most severe type. Other types include OCA1B, OCA 2, OCA 3, and OCA 4 which show the presence of melanin to some extent. Although the prevalence of OCA in the general population in Pakistan is not known, a study conducted in a tertiary eye care setting reported that OCA contributed to 6.7% of vision impairment in children with low vision in a tertiary eye care setting in Pakistan.[5]

As a result of reduced or lack of pigment in the retinal pigment epithelium of the eye, people with OCA have abnormalities of visual system development such as foveal hypoplasia, optic nerve hypoplasia, and iris translucency.[6,7] Reduced vision, nystagmus, photophobia, strabismus, and amblyopia are some of the common ocular features in people with OCA.[6,7] People with OCA also have a wider range of refractive errors including high with-the-rule astigmatism.[7,8]

As compare to the normal pigmented retinas, the photoreceptor outer segment of the retina of

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people with albinism does not develop normally.[9] In addition, the lack of pigment in people with OCA also results in the abnormal decussation and misrouting of the optic nerve fibers during the development resulting in poor vision.[10,11] Many of them are classified as being in the category of low vision due to visual dysfunction.[5,7,12] People with low vision can be helped with low vision aids such as spectacles, filters, telescopes, and magnifiers.[5,13-15] The major disability in people with OCA is poor vision. This study aimed to explore the potential benefits of refraction and low vision aids in visual rehabilitation of people with OCA.

## Methods

This was a cross-sectional, descriptive, hospital-based, retrospective study. Record of 77 consecutive people diagnosed with OCA aged 4 years and above and who attended two successive follow-up visits after initial examination were included in the study. These patients were assessed for low in the low vision clinic from June 2017 to June 2018. To avoid confusion of complete and incomplete albinism, only people with OCA were included. These people with OCA were referred for low vision rehabilitation to the low vision clinic in Department of Ophthalmology Hayatabad Medical Complex, Peshawar, from various hospitals in the Khyber Pakhtunkhwa province in Pakistan.

Mostly, low vision clinics are established in tertiary eye care settings in Pakistan. In Khyber Pakhtunkhwa province, low vision clinics are established in two tertiary eye care centers. People with low vision are referred to these clinics from secondary as well as tertiary eye care settings. The low vision clinic where this study was conducted is the most advanced and the first low vision clinic in the public sector hospitals in Pakistan. Therefore, people with OCA who contributed to 6.7% of the overall workload of low vision patients in this low vision clinic are mostly referred to this clinic from all over the province.

The patient notes were reviewed for the detailed history of the patient and their family history. Cases with incomplete record and those who lost for the first two follow-ups were excluded from the study. The standard regimen of examination was as follows:

1. Detailed optometric examination of all participants was conducted by the first author of this study who has more than 15 years' experience in the management of people with low vision. The anterior segment examination was performed using a slit-lamp bio-microscope. Posterior segment examination was performed by direct or indirect ophthalmoscopy after mydriasis. The key findings in the complete ocular examination including visual acuity (VA), refraction, and low vision assessment were noted
2. Presenting distance VA was measured using a Bailey-Lovie Log. Minimum angle of resolution (MAR) (logarithm of the MAR) chart with five optotypes on each line. Log MAR chart consisting of "E," numeric was used for patient who could not read English. VA was tested at 4 m and if necessary, at 3 and 2 m on each eye separately while the patient wore his or her current spectacles (if worn). For those people whose distance VA was very low, one meter or half meter testing distance was used. Presenting distance VA was measured first binocularly then monocularly. Binocular VA in the patients' habitual head position was recorded as person's VA
3. For assessing near VA "Near Reading Card for the partially sighted" by William Feinbloom and "Lea Cards for near VA" were used. Near VA for hypermetropes was tested on presentation as well as with correction for refractive errors. While near VA for myopes was tested without distance correction. Near VA was banded in three groups: 1M (newspaper size), <1M to 3.2M (display materials), and <3.2M.[15] The near VA and eye to chart distance were recorded for each eye. Filters and low vision aids such as telescopes, stand and hand magnifiers, and closed circuit television (CCTV) were used during the low vision assessment
4. Assessment of VA was followed by objective examination of the refractive conditions in each eye. In people aged 14 years and above, dry (undilated) retinoscopy was performed using Heinne retinoscope under diminished room lighting conditions by closing the door and window curtains. Results of dry retinoscopy were refined by subjective refraction and use of Jackson's cross cylinder. Retinoscopy with cycloplegia, using cyclopentolate 1% eye drops three times in each eye was carried out 30 min after instillation of last dose of cyclopentolate 1% eye drops on people with age <14 years. Cycloplegic retinoscopy was followed by a subjective refraction on the follow-up visit after 5 days of the initial visit. VA in each eye was measured with best possible correction and binocular VA was recorded as person's VA.

Ethics approval was obtained from the Research and Ethics Committee of the Hayatabad Medical Complex Peshawar.

Statistical Package for the Social Sciences software version 19 (IBM Corp, Armonk, NY, USA) was used for the analysis, and the data were analyzed in frequencies and cross tabulation.

## Results

Of this cohort of 77 people with OCA, 44 (57%) were in the age group of 4 to <16 years and 33 (43%) were aged 16 years and above. The mean age was 18 years. Among them, 47 (61%) were male and 30 (39%) were female.

Only one patient had VA 6/18 in the better eye at the time of presentation and 57 (74%) had moderate-to-severe vision impairment (presenting VA in the better eye <6/18 but at least 3/60). With adequate refraction, 5 (6.5%) people achieved VA 6/18 or better in the better eye. VA improved significantly ($P < 0.01$) after adequate refractive correction

by more than one log MAR lines in 38.6% people with OCA. With low vision devices, distance VA was enhanced to 6/18 or better in the better eye in 66 (85.7%) people with OCA while 6 (7.8%) remained in the blind category (VA <3/60 in the better eye). VA person at presentation, best corrected VA, and VA after prescribing low vision devices are given in Table 1.

The mean of spherical equivalent refractive error in right eyes was −0.29 diopters (standard deviation [SD] 3.83 D) while the mean of spherical equivalent refractive errors in left eyes was 0.21 diopters (SD 3.66 D). The SD describes these data is large reflecting the large scatter in the refractive errors data in people with OCA. Overall, the refractive errors among these people with OCA ranged from +10.00 D to −14.00 D. The estimated difference in means of the spherical equivalent refractive error in the right eyes and left eyes was −0.494 diopters (−01.686, 0.699; 95% confidence interval [CI]).

In all participants, the mean of cylindrical refractive error in the right eyes was −1.09 diopters (SD 1.17 D) while the mean of cylindrical refractive errors in the left eyes was 0.21 diopters (SD 1.24 D). The estimated difference in means of the cylindrical refractive error in the right eyes and left eyes was −0.130 diopters (−0.514, 0.254; 95% CI). Mean cylindrical refractive error in right eyes in the age group of 4–15 years and 16 years and above were −1.25 diopters (SD 1.170 diopters) and 0.879 (SD 1.161 diopters), respectively. Mean cylindrical error in the left eyes in the age group of 4–15 years was −1.057 diopters (SD 1.319 diopters) and in age group 16 years and above was −0.833 diopters (SD 1.128 diopters).

With-the-rule astigmatism was more present (42% in the right eye and 40% in the left eye) than against-the-rule astigmatism (9% in the right eye and 8% in the left eye). Thirty-nine percent did not have astigmatism in the right eye and 44% did not have astigmatism in the left eye. Five percent have oblique astigmatism in the right eye and 4% in the left eye.

The percentage of female with OCA in the blind category and severe visual impairment category was higher on presentation as well as after prescribing low vision aids [Table 2].

More children (9%) remained in the blind category than adults (6%) after prescribing low vision aids [Table 3].

On presentation, thirty people (39%) with OCA were able to discern 1M (newspaper print) or better. This number increased to 39 (50.6%) after the correction of refractive errors in hypermetropes. With low vision aids, further 28 people with OCA achieved 1M or better. Near VA person at presentation and near VA after prescribing low vision devices are given in Table 4.

For distance vision, spectacles were prescribed to 67 peoples. After conventional glasses, monocular telescopes ranged from 3X to 8X magnification were the most common visual aid (43%) prescribed to people with OCA. These telescopes are obtained from the Hong Kong Society for the Blind-Vision 2020 Low Vision Resource Center. Although 57 (74%) people with OCA desired for Ocutech Telescope, but it was prescribed to five peoples because the rest could not afford Ocutech Telescope.

Functional and measurable VA is improved with refraction and low vision aides while subjective improvement in quality of vision such as reduction of photophobia, relief in eye-strain, and eye discomfort was noted with filters in 59 people with OCA. Tinted spectacles were prescribed to all participants for outdoor activities, especially in summer and sunlight because summer is very hot and sunny in Pakistan. P-cap or hat with a brim was recommended for 69 people with OCA who complain of photophobia in outdoor activities. Eight people with OCA were already using P-cap.

For enhancing near VA, glasses including prismatic glasses (Fonda glasses) were prescribed to 33 peoples. In addition, stand magnifiers were prescribed to 15 peoples; Hand Hold Magnifier to 17 people; reading cap with Ocutech telescopes were prescribed to three people and CCTV was prescribed to one person with OCA to meet their needs.

## Discussion

All people with OCA have poor vision that restricted their activities of daily living.[16-18] This study demonstrated that functional VA in people with OCA could be improved with adequate refraction and provision of low vision aids. These included both for distance viewing aids such as telescopes and reading aids such as prismatic glasses, hand hold magnifiers, and stand magnifiers. Similar improvement in functional vision with low vision aids in people with OCA has been reported in the literature.[8,19,20]

As identified in the literature,[8,16,18] this study also showed that higher spherical and astigmatic refractive errors and with-the-rule astigmatism are associated with OCA. The distributions of spherical-equivalent and astigmatic refractive errors are broader in our participants with OCA. The similar spread of the refractive error distributions in people with OCA has been indicated in the literature.[18,21]

In this study, we found that the number of people with OCA in the blind category (VA <3/60) on presentation dropped by 52% (from 19 to 10) after refraction indicating significant improvements in distance VA [Table 1]. The findings from this study demonstrated that adequate refraction for the correction of refractive errors is the first step in visual rehabilitation of people with OCA as reported in the literature.[8,16,18,21]

The results of this study showed that half of people with OCA were able to discern 1M (Newspaper print) or better with distance correction only and they could utilize their vision for reading purposes. This shows that distance vision is not a criterion to determine what the person with OCA can read at near. These findings indicated that appropriate refraction for correction of refractive errors, specifically for hypermetropes, helps to improve reading performance in people with OCA.

**Table 1: Presenting visual acuity, visual acuity with refraction and visual acuity with low vision devices (*n*=77)**

| VA Snellen (LogMAR) | VA on presentation, *n* (%) | VA with refraction, *n* (%) | VA with LVDs, *n* (%) |
|---|---|---|---|
| 6/18 (0.54) or better | 1 (1.3) | 5 (6.5) | 66 (85.7) |
| <6/18-6/60 (<0.54-1.0) | 40 (52) | 51 (66.2) | 4 (5.2) |
| <6/60-3/60 (<1.0-1.3) | 17 (22) | 11 (14.3) | 1 (1.3) |
| <3/60 (<1.3) | 19 (24.7) | 10 (13) | 6 (7.8) |
| Total | 77 (100) | 77 (100) | 77 (100) |

VA=Visual acuity; LVDs=Low vision devices, LogMAR=Logarithm of the Minimum Angle of Resolution.

**Table 2: Presenting visual acuity and visual acuity with low vision devices by gender (*n*=77)**

| VA | VA on presentation, *n* (%) | | VA with refraction, *n* (%) | | VA with LVDs, *n* (%) | |
|---|---|---|---|---|---|---|
| | Male | Female | Male | Female | Male | Female |
| 6/18 (0.54) or better | 1 (2.1) | 0 (0) | 4 (8.5) | 1 (3.3) | 44 (93.6) | 22 (73.3) |
| <6/18-6/60 (<0.54-1.0) | 30 (63.8) | 10 (33.3) | 34 (72.3) | 17 (56.7) | 2 (4.3) | 2 (6.7) |
| 6/60-3/60 (<1.0-1.3) | 9 (19.1) | 8 (26.7) | 6 (12.8) | 5 (16.7) | 0 (0) | 1 (3.3) |
| <3/60 (<1.3) | 7 (15) | 12 (40) | 3 (6.4) | 7 (23.3) | 1 (2.1) | 5 (16.7) |
| Total | 47 (100) | 30 (100) | 47 (100) | 30 (100) | 47 (100) | 30 (100) |

VA=Visual acuity; LVDs=Low vision devices

**Table 3: Presenting visual acuity and visual acuity with low vision devices by age (*n*=77)**

| VA | VA on presentation, *n* (%) | | VA on presentation, *n* (%) | | VA on presentation, *n* (%) | |
|---|---|---|---|---|---|---|
| | 4 ≤Age <16 years | Age ≥16 years | 4≤ Age <16 years | Age ≥16 years | 4 ≤Age <16 years | Age ≥16 years |
| 6/18 (0.54) or better | 0 (0.0) | 1 (3.0) | 3 (6.8) | 2 (6.1) | 37 (84.1) | 29 (88.0) |
| <6/18-6/60 (<0.54-1.0) | 20 (45.5) | 20 (60.6) | 27 (61.4) | 24 (72.7) | 3 (6.8) | 1 (3.0) |
| 6/60-3/60 (<1.0-1.3) | 10 (22.7) | 7 (21.2) | 8 (18.2) | 3 (9.1) | 0 (0.0) | 1 (3.0) |
| <3/60 (<1.3) | 14 (31.8) | 5 (15.2) | 6 (13.6) | 4 (12.1) | 4 (9.1) | 2 (6.0) |
| Total | 44 (100) | 33 (100) | 44 (100) | 33 (100) | 44 (100) | 33 (100) |

VA=Visual acuity; LVDs=Low vision devices

**Table 4: Near visual acuity at presentation and with low vision devices (*n*=77)**

| VA | Near VA with distance correction, *n* (%) | Near VA with Near correction, *n* (%) | Near VA with LVDs, *n* (%) |
|---|---|---|---|
| 1M or better | 30 (39.0) | 39 (50.6) | 67 (87.0) |
| <1M-3.2M | 26 (33.8) | 18 (23.4) | 4 (5.2) |
| <3.2M | 21 (27.2) | 20 (26) | 6 (7.8) |
| Total | 77 (100) | 77 (100) | 77 (100) |

VA=Visual acuity; LVDs=Low vision devices

Numerous literature reported that conventional glasses to correct the refractive error and strong reading additions particularly in the form of bifocals are the most acceptable method for improving vision in people with OCA.[16,18,19] This study also showed that people with OCA preferred conventional glasses and strong reading glasses with base-in prism (Fonda glasses) to telescopes and magnifiers for enhancing their vision. Because the conventional glasses or strong reading glasses are less expensive, have larger field of vision, and cosmetically more acceptable. People with OCA who could not get enough advantage from conventional glasses or could not afford Ocutech telescopes accepted monocular telescopes and magnifiers to meet their needs.

Nonoptical low vision aids help people with low vision to deal with everyday task and can be prescribed in combination with optical low vision devices. The findings from our study demonstrated that 77% of people with OCA reported subjective improvement in their functional vision such as reduction of photophobia, relief in eye-strain, and eye discomfort with the use of filters. Similar results have been reported in the literature.[22]

Due to nonavailability of brightness acuity tester, the extinct of vision impairment caused by photophobia or glare was not noted. Another limitation of this study is that surgery was not prescribed for nystagmus. Effects of low vision rehabilitation on quality of life of people with OCA have been planned in another manuscript.

## Conclusion

The findings from this study demonstrate that adequate refraction improves VA of people with OCA. The mean improvement was one log. MAR unit in this study. Low vision aids improved the visual functions of majority of people with OCA. Therefore, early referral of people with OCA to low vision clinic is necessary to monitor the potential enhancement in their visual functions. Awareness among people and eye care professionals is essential for bringing up people with low vision due to any ocular disorder to low vision clinics. Further research is needed to investigate availability, acceptance and utilization of low vision services in the community and effect of low vision services on quality of people with low vision.

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## Conflicts of interest
There are no conflicts of interest.

## References
1. von dem Hagen EA, Houston GC, Hoffmann MB, Morland AB. Pigmentation predicts the shift in the line of decussation in humans with albinism. Eur J Neurosci 2007;25:503-11.
2. von dem Hagen EA, Houston GC, Hoffmann MB, Jeffery G, Morland AB. Retinal abnormalities in human albinism translate into a reduction of grey matter in the occipital cortex. Eur J Neurosci 2005;22:2475-80.
3. Suzuki T, Tomita Y. Recent advances in genetic analyses of oculocutaneous albinism types 2 and 4. J Dermatol Sci 2008;51:1-9.
4. Rooryck C, Roudaut C, Robine E, Müsebeck J, Arveiler B. Oculocutaneous albinism with TYRP1 gene mutations in a Caucasian patient. Pigment Cell Res 2006;19:239-42.
5. Shah M, Khan M, Khan MT, Khan MY, Saeed N. Causes of visual impairment in children with low vision. J Coll Physicians Surg Pak 2011;21:88-92.
6. Hutton SM, Spritz RA. Comprehensive analysis of oculocutaneous albinism among non-Hispanic caucasians shows that OCA1 is the most prevalent OCA type. J Invest Dermatol 2008;128:2442-50.
7. Wildsoet CF, Oswald PJ, Clark S. Albinism: Its implications for refractive development. Invest Ophthalmol Vis Sci 2000;41:1-7.
8. Schulze Schwering M, Kumar N, Bohrmann D, Msukwa G, Kalua K, Kayange P, *et al*. Refractive errors, visual impairment, and the use of low-vision devices in albinism in Malawi. Graefes Arch Clin Exp Ophthalmol 2015;253:655-61.
9. Mohammad S, Gottlob I, Kumar A, Thomas M, Degg C, Sheth V, *et al*. The functional significance of foveal abnormalities in albinism measured using spectral-domain optical coherence tomography. Ophthalmology 2011;118:1645-52.
10. Grønskov K, Ek J, Brondum-Nielsen K. Oculocutaneous albinism. Orphanet J Rare Dis 2007;2:43.
11. Hertle RW, Yang D, Ma L, Gao J, Madhavi G, Roberts TL, *et al*. Visual rehabilitation of patients with oculocutaneous albinism type 1 (OCA): Results in 85 patients. New Front Ophthalmol 2017;3:1-14.
12. Kutzbach BR, Summers CG, Holleschau AM, MacDonald JT. Neurodevelopment in children with albinism. Ophthalmology 2008;115:1805-8, 1808.e1-2.
13. Shah M, Zaman M, Khan MT, Khan MD. Visual rehabilitation of patients with Stargardt's disease. J Coll Physicians Surg Pak 2008;18:294-8.
14. Pollard TL, Simpson JA, Lamoureux EL, Keeffe JE. Barriers to accessing low vision services. Ophthalmic Physiol Opt 2003;23:321-7.
15. Shah M, Khan MD. Causes of low vision amongst the low-vision patients attending the low-vision clinic at Khyber institute of ophthalmic medical sciences (KIOMS), Hayatabad medical complex Peshawar, Pakistan. J Vis Imp Res 2004;6:89-97.
16. Bhari AM, Bajracharya K, Thapa HB, Gautam M, Pandey A, Bassett K. Effectiveness of refractive error correction for people with oculocutaneous albinism in Nepal. Asian J Med Sci 2017;8:30-4.
17. Khanal S, Pokharel A, Kandel H. Visual deficits in Nepalese patients with oculocutaneous albinism. J Optom 2016;9:102-9.
18. Anderson J, Lavoie J, Merrill K, King RA, Summers CG. Efficacy of spectacles in persons with albinism. J AAPOS 2004;8:515-20.
19. Woo SY, Perez AM, Jose RT. Comprehensive low vision rehabilitation care for oculocutaneous albinism: A longitudinal case report. Int Congress Series 2005;1282:337-41.
20. Joshi MR, Yamagata Y, Akura J, Shakya S. The efficacy of low vision devices for students in specialized schools for students who are blind in Kathmandu Valley, Nepal. J Vis Impair Blind 2008;102:430.
21. Sampath V, Bedell HE. Distribution of refractive errors in albinos and persons with idiopathic congenital nystagmus. Optom Vis Sci 2002;79:292-9.
22. Rosenblum YZ, Zak PP, Ostrovsky MA, Smolyaninova IL, Bora EV, Dyadina UV, *et al*. Spectral filters in low-vision correction. Ophthalmic Physiol Opt 2000;20:335-41.